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Volume 02

Anthropology for Civil Services Examination

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Physical Anthropology

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Table of Contents

1.4. HUMAN EVOLUTION AND EMERGENCE OF MAN **7**
A. BIOLOGICAL & CULTURAL FACTORS IN HUMAN EVOLUTION 7
B. THEORIES OF ORGANIC EVOLUTION 11
PRE-DARWINIAN PHASE *12*
THE DARWINIAN PHASE *14*
THEORY OF NATURAL SELECTION (DARWINISM) *14*
POST–DARWINIAN PHASE *17*
C. SYNTHETIC THEORY OF EVOLUTION 17
BASIC CONCEPTS AND TERMS IN EVOLUTIONARY BIOLOGY 23
CONVERGENCE *23*
PARALLELISM *23*
HOMOLOGY AND ANALOGY *24*
ADAPTIVE RADIATION *24*
DOLLO'S LAW *25*
MOSAIC EVOLUTION – THE EVOLUTION OF PARTS AND WHOLES *25*
COPE'S RULE *26*
PRINCIPLE OF COMPETITIVE EXCLUSION OR GAUSE'S RULE *26*

1.5 CHARACTERISTICS OF PRIMATES **26**
PRIMATES EVOLUTIONARY TRENDS 26
PRIMATE TAXONOMY & BEHAVIOR 31
TERTIARY AND QUATERNARY FOSSIL PRIMATES 42
A COMPARATIVE ANATOMY OF MAN AND APES 51
PRIMATE ADAPTATIONS - ARBOREAL AND TERRESTRIAL 54
SKELETAL CHANGES DUE TO BIPEDALISM AND THEIR IMPLICATIONS 58

1.6 PHYLOGENETIC STATUS, CHARACTERISTICS, AND DISTRIBUTION OF HUMAN FOSSIL ANCESTORS **60**
(A) PLIO-PLEISTOCENE HOMINIDS IN SOUTH AND EAST AFRICA - AUSTRALOPITHECINES 61
(B) HOMO ERECTUS 67
HOMO ERECTUS HEIDELBERGENSIS 70
(C) NEANDERTHAL MAN 70
(D) RHODESIAN MAN 76
(E) HOMO SAPIENS 77
CRO-MAGNON MAN *77*
GRIMALDI MAN *78*
CHANCELADE MAN *79*

1.7 THE BIOLOGICAL BASIS OF LIFE **80**
THE CELL 80
DNA STRUCTURE 87
DNA REPLICATION 90
PROTEIN SYNTHESIS 92
GENE MUTATION 99
CHROMOSOMES 100
CELL DIVISION 101

9.1 HUMAN GENETICS – METHODS AND APPLICATIONS **107**
PEDIGREE ANALYSIS 107
THE TWIN METHOD 110

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CO-TWIN METHOD 114
FOSTER CHILD 114
CYTOGENETIC METHOD 115
CHROMOSOMAL AND KARYOTYPE ANALYSIS 115
BIOCHEMICAL METHODS 117
IMMUNOLOGICAL METHOD 118
RECOMBINANT DNA TECHNOLOGY 119

9.2 MENDELIAN GENETICS IN MAN 121
THE MENDELIAN PRINCIPLES 121
SINGLE FACTOR, MULTIFACTOR AND POLYGENIC INHERITANCE IN MAN 126
LETHAL GENE ACTION 127

9.3 CONCEPT OF GENETIC POLYMORPHISM AND SELECTION 129
GENETIC POLYMORPHISM AND SELECTION 130
MENDELIAN POPULATION 130
GENETIC EQUILIBRIUM 133
HARDY - WEINBERG EQUILIBRIUM 133
CAUSES AND CHANGES IN GENE FREQUENCY 134
CONSANGUINEOUS AND NON-CONSANGUINEOUS MATING 138
GENETIC LOAD 139
GENETIC EFFECTS OF CONSANGUINEOUS AND COUSIN MARRIAGES 140

9.4 CHROMOSOMES AND CHROMOSOMAL ABERRATIONS IN MAN 141
NUMERICAL AND STRUCTUAL ABERRATIONS 142
SEX CHROMOSOMAL ABERRATIONS 142
AUTOSOMAL ABERRATIONS 144
GENETIC IMPRINTS IN HUMAN DISEASES 146
GENETIC SCREENING 148
GENETIC COUNSELING 148
HUMAN DNA PROFILING 150
GENE MAPPING & GENOME STUDY 154

9.5 RACE AND RACISM 159
RACE & RACISM 159
BIOLOGICAL BASIS OF MORPHOLOGICAL VARIATION OF NON-METRIC AND METRIC CHARACTERS 161
SEROLOGICAL AND GENETIC CRITERIA 166
RACIAL CRITERIA, RACIAL TRAITS IN RELATION TO HEREDITY AND ENVIRONMENT 171
RACIAL CLASSIFICATION AND DIFFERENTIATION 175

9.6 AGE, SEX AND POPULATION VARIATION 183

9.7 CONCEPTS AND METHODS OF ECOLOGICAL ANTHROPOLOGY 187
BIO-CULTURAL ADAPTATIONS – GENETIC AND NON GENETIC FACTORS 189
MAN'S PHYSIOLOGICAL RESPONSES TO ENVIRONMENTAL STRESSES 192
HOT DESERTS 192
COLD HIGH ALTITUDE CLIMATES 194

9.8 EPIDEMIOLOGICAL ANTHROPOLOGY 197
CONCEPT OF EPIDEMIOLOGICAL ANTHORPOLOGY 197
HEALTH AND DISEASE 197
INFECTIOUS AND NON INFECTIOUS DISEASES 200
INFECTIOUS DISEASES 209
NUTRITIONAL DEFICIENCY RELATED DISEASES 217

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10. CONCEPT OF HUMAN GROWTH AND DEVELOPMENT **225**
CONCEPT OF HUMAN GROWTH AND DEVELOPMENT 225
STAGES OF HUMAN GROWTH 227
PRENATAL DEVELOPMENT *227*
INFANCY *228*
CHILDHOOD *229*
ADOLESCENCE *231*
MATURITY *232*
SENESCENCE *234*
FACTORS AFFECTING GROWTH AND DEVELOPMENT 236
AGEING AND SENESCENCE 239
BIOLOGICAL AND CHRONOLOGICAL LONGEVITY *239*
THEORIES AND OBSERVATIONS *240*
HUMAN PHYSIQUE AND SOMATOTYPES 242
METHODOLOGIES FOR GROWTH STUDIES 245

11 SOCIAL DEMOGRAPHY **247**
11.1 RELEVANCE OF MENARCHE, MENOPAUSE AND OTHER BIOEVENTS TO FERTILITY 247
FERTILITY PATTERNS AND DIFFERENTIALS 248
11.2 DEMOGRAPHIC THEORIES – BIOLOGICAL, SOCIAL AND CULTURAL 250
11.3 BIOLOGICAL AND SOCIO-ECOLOGICAL FACTROS INFLUENCING FECUNDITY, FERTILITY, NATALITY AND MORTALITY 256

12. APPLICATIONS OF ANTHROPOLOGY **273**
ANTHROPOLOGY OF SPORTS 273
NUTRITIONAL ANTHROPOLOGY 274
DESIGNING OF DEFENSE AND OTHER EQUIPMENTS 276
FORENSIC ANTHROPOLOGY, METHODS AND PRINCIPLES OF PERSONAL IDENTIFICATION AND RECONSTRUCTION 278
APPLIED HUMAN GENETICS 282
PATERNITY DIAGNOSIS 282
GENETIC COUNSELING 283
EUGENICS 283
DNA TECHNOLOGY IN DISEASES AND MEDICINE 286

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1.4. HUMAN EVOLUTION AND EMERGENCE OF MAN

A. BIOLOGICAL & CULTURAL FACTORS IN HUMAN EVOLUTION

Hominization Process: Hominization Process is the evolutionary transformation of hominoids into hominids. It is a process that occurred in the hominid line since its divergence from the last common hominoid ancestor shared with any living ape. The term is applied to include all those aspects of structural and behavioral changes that occurred in the hominid line finally leading to the evolution of modern man. These changes essentially constitute the essential biological and cultural factors that contributed to human evolution.

1. Erect Posture & Bipedal Locomotion

Perhaps the most obvious thing about human beings that differentiates them from all other members of the animal kingdom is their upright posture and their associated habit of walking (known technically as bipedal motion). Many of the distinctively human morphological traits are directly attributable to these two facts of human life.

The shape of the vertebral column in mammals is delicately adjusted to support the animal's center of gravity. In a buffalo this point is over the forelegs, and its backbone resembles the profile of a cantilever bridge with the weight carried on a central pillar. In Anthropoidea, the center of gravity is shifted back considerably and the vertebral column is unusually flexible for quadrupeds- features that reflect a tree-dwelling way of life, featuring agile grasping and jumping with strong rear leg thrusting. The stresses put on the backbone by brachiating resulted in its becoming somewhat stiffened, with several lumbar vertebrae becoming thorasic vertebrae. This fact suggests that humans are more closely related to brachiators than non-brachiating primates: quadrupedal monkeys have an average of 9 lumbar vertebrae and 10 thorasic vertebrae; gibbons average 6 and 13 respectively; great apes 4 and 13; and humans 5 and 12.

Characteristics of the human vertebral column that are specific adaptations to an upright posture include: enlarging of the lower vertebrae to absorb the forces of compression; relative constancy in the size of the spines protruding from each vertebra, resulting from a lack of weight-bearing stress points along the spine (as opposed, for instance, to the gorilla, in which the spines of the neck vertebrae are elongated to provide a solid anchor for the heavy neck muscles that must support the forward-jutting head - unnecessary in humans, where the head is nicely balanced on top of the vertebral column); increase in the size and number of bones in the sacrum to take up the transmission of weight through the pelvis and legs (humans average 5-6 sacral bones, great apes around 5, quadrupedal lower primates 2-4); and finally, a sharp backward curving of the spine in the lumbar region (lower back) providing a solid platform to transfer the weight of the body onto the pelvis and giving the human spinal column its distinctive S-shape.

It is observed that these and other human features emerged as our ancestors became increasingly bipedal. True enough, many have mostly teeth and jaw fragments from Ramapithecus, but we think that these creatures spent a great deal of their time on the ground, on the fringes of the forests in the Miocene epoch; and if we remember that primates developed their vision rather than their sense of smell, standing on hind legs might well have been crucial for spotting the abundant meat-eating predators for which Ramapithecus - with no specialized defensive weapons - must have been an attractive and even easy meal. Thus we can speculate that there were strong selective pressures (in the form of carnivores) on Ramapithecus to develop highly cooperative social groups, an efficient communicative code (depending on sounds rather than visual signals that would be difficult to see in the tall grass), and the habit of

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rearing up on the hind legs to scan the countryside for danger. Certainly, these features became the principal adaptive characteristics of subsequent hominid groups.

Australopithecines are already clearly bipedal and erect, although certain human features had not yet completely evolved: the pelvis is not yet broadened and rounded as much as in modern humans (for support of the internal organs as well as the enhancement of the passing of a large-brained infant at birth), although it is already much broader and rounder than in modern great apes; the lumbar curve is not yet fully developed; and the foramen magnum (the hole through which the spinal cord passes out of the skull and into the vertebral column) into the rear of the center of the base of the skull, meaning the head was not yet fully balanced at the top of the vertebral column. On the other hand, their feet were nearly modern; they had arches front-to rear and sideways and a full heel to provide good walking leverage, as we do. It appears that Australopithecines developed persistence hunting - pursuing game until the game was exhausted - as a major survival technique that put a premium on long distance walking. In any event, walking must have been of extreme importance to the Australopithecines because by the time of Homo erectus, the skeletal structure (with the exception of the head) was essentially fully modern, completely adapted to walking. Even the skull shows adaptation to erect posture, with the shifting forward of the foramen magnum closer to the center of the base of the skull.

Thus by the time Homo erectus trekked out across the continents of the old world, virtually all of the uniquely human traits associated with erect posture and bipedal locomotion had evolved.

2. Remodeling of Face & Teeth

When an infant primate is born, it passes out of its mother's womb through the birth canal, through the vagina, and into the realities of social life. The birth canal is surrounded by that forms the lower front of the mother's pelvis. This bone sets a limit on the size that a newborn infant can be at the moment of its birth: if it (or any part of it) is too large it will get stuck, with the result that both the infant and the mother are quite likely to die - which means that, they will not contribute their genes to the ongoing evolutionary process.

It was noted that a major trend of human evolutionary development was a dramatic increase in the size of the brain, which we deduce from the fossil evidence showing a rapid increase in the size of the brain case since the Australopithecines. Since the size of the head as a whole should not keep getting larger beyond limits allowed by the process of birth, there was a strong adaptive pressure to shorten the snout and reduce the size of the face to make room for cranial expansion in the course of human evolution. Fortunately, other processes came into play that made this possible.

But first, what changes took place in the face and teeth among our hominid ancestors? Already with the arrival of Ramapithecus we can see some hominid facial trends. The upper and lower canines interlock only slightly, and there is little or no space (diastema) next to the canines to make a room for such interlocking. Australopithecines continue this trend, although the face and jaw are still massive by modern standards. Heavy ridges of bone arch over the eye sockets, probably affording a measure of protection as well as a shelf against which some of the large jaw muscles could be anchored. Indeed, in robust forms, the sagittal crest running from front to back on the top of the skull provides more space for such attachments. For all practical purposes, Australopithecines display little in the way of foreheads, although the gracile line shows some movement in that direction. Neither Ramapithecus, nor the Australopithecines, nor even Homo erectus have any chin whatever; in fact, many still have a ridge of bone running part-way back which forces the front teeth to protrude outward. All three groupings have very large teeth (in proportion to the whole head) by modern standards.

The face of Homo erectus is by far its most primitive aspect. It is still massively jawed, with a minimal forehead sloping sharply back from heavy bone brow ridges. The skull narrows sharply behind the eye

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sockets, providing a groove along which the large cheek muscles that operate the jaws can run. Its front teeth still "buck" outwards and its nasal cavity, palate, and tongue are long and flat compared to those of modern humans, while its larynx sits much higher than in its throat than ours. And of course, its head hangs slightly forward, somewhat awkwardly balanced on its vertebral column, with a foramen magnum that is still somewhat behind the center of the skull's base. But in virtually every other respect Homo erectus has essentially modern skeletal features.

Even Neanderthal - the earliest member of our own species Homo sapiens - retained a heavy-looking face and large teeth by modern standards, especially, the "classical" Neanderthal of southwestern Europe. However, by this time we can observe the emergence of a real forehead (although some specimens still retain the heavy brow ridging of Homo erectus); the front teeth are rooted vertically, and the lower jaw that supports them juts out beyond them to give them a solid base; and a fully modern chin has developed.

These developments are further elaborated in the transition to fully modern Homo sapiens: the brow ridges diminish; the forehead approaches being vertical; the chin is prominent; the face (with its small vertical teeth) is flat, with a nose that protrudes. The foramen magnum is now exactly at the center of the skull's base, the head balancing nicely on the vertebral column. The nasal cavity and palate are shortened and arched, the tongue thickened and shortened, and the larynx sunken down into the throat, which for the first time joins the mouth pretty much at a right angle rather than obliquely.

Many factors brought about these changes. First, the trend toward upright posture resulted in the foramen magnum being moved forward, tilting the head upright into a vertical position and possibly exerting some "squeeze" against the face. Second, the long, flat mouth, tongue, palate and nasal cavity with a high larynx and the wide angle at which the throat joined the mouth prior to Homo sapiens, drastically limited the number of sounds the vocal apparatus could produce and the speed with which it could produce them. If, the evolution of speech were of crucial importance to the overall process of human evolution, then there would have been selective pressures for molding the face in the form it finally assumed. In fact, there is evidence that the mouth and tongue of Neanderthal are still too flat to produce the modern range of sounds, and also probably lack enough quickness of tongue to produce sounds at a modern rate. Finally, it appears that human teeth only really were free to become small when people gave up using the mouth as a "tool" - a "fifth hand" used for holding, tearing, and even chewing items to soften them (as Eskimos soften skins). This happened in the last 10,000 years with the development of food production, when the agricultural revolution replaced hunting and foraging as the principal means of subsistence for most of the peoples in the world.

3. Expansion & Development of Brain

There is a dramatic increase of the Brian case as we approach specimens of Homo sapiens. This is indeed worth noting, and physical anthropologists have spent a great deal of time studying the rapid increase in brain size in the course of post-australopithecine evolution. Why did it happen?

The reasons are complicated. First, in general, the overall size of our ancestors increased at each evolutionary stage. We concluded that there were strong selective pressures for this increase in size, probably because a larger body size made it easier to hold and use tools, and also increased the amount of muscle available to hunters and foragers on long treks.

But the brain grew larger, proportionately did the body. The most likely reason is that an increase in brain size tremendously increases the possibility - indeed the probability - of an increase in both the number and kinds of connections between the brain cells. It is an increase in the kinds of connections between brain cells that is apparently responsible for the emergence of new kinds of mental operations such as thinking and using language, operations that are fundamental to human existence.

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By carefully studying the contours of the insides of fossil brain cases, scientists have been able to document that the tremendous growth in brain size between the Australopithecines and Homo erectus was accompanied by an increase in size and complexity of the outside surface of the brain called the Cerebral Cortex. This expansion of the cortex is the most recent evolutionary development of the brain, and it is the cortex that is primarily associated with thinking and language use.

Each of the senses is represented in a specific area of the cortex, a part of the outer layer of the brain, where the fibers carrying messages from the surface of the body concentrate according to the sense. Thus, for example, stimulation of the retina in the eye ball results in messages flashing along the optic nerve fibers and flooding into one area of the cortex; messages from the fingers (touch) arrive in another area; taste messages in yet a third area, and so forth. The areas of the brain that are specialized to process these incoming messages are called the nuclear zones of cortex; and each sense has its own.

Around each nuclear zone anatomists have found that specialized bundles of fibers are concentrated into what are called association areas; and the clear differentiation of separated association areas increases as we move up the order of the Primates towards human beings. Now the job that each association area performs is very complicated, but it amounts to modifying the operation of its nuclear zone. For example, damage to the visual association area in the human brain results in the person loosing his or her ability to identify the objects, even though the objects are still seen (the messages are still represented in the visual nuclear zone). It appears, then, that the association areas of the brain are central to the process of recognition. They are where information about the outside world is stored, where incoming stimuli can be matched against stored experience and decisions can be made: upon seeing a Saber-toothed tiger, one can decide to issue a warning to one's neighbors and take to one's heels - or alternatively, face the predator and try to convert into a month of meals. In humans large bundles of nerve fibers connect the association areas of the cortex. This provides us with the ability to pass messages back and forth directly between these association areas, to compare and contrast there different recognitions - in other words to think.

The cortex also houses the nuclear zones for triggering muscle actions (motor functions). An outstanding characteristic of Anthropoidea is that they have evolved direct link ups between the association areas of sight and touch and the association areas around the major motor function centers. These direct link ups result in the ability to "fit" ones bodily movements to what ones sees and holds, while freed from other functions of the brain - such as emotions - that might interfere. Thus, if you think about it for a moment, you will see that these structural features of anthropoid brains amount to being a necessary precondition for using tools. In other words, regardless of why the anthropoids acquired these features, without them regular tool use - a preeminent specialty of our hominid ancestors as they evolved - could not have developed.

4. Culture and Hominization Process

Each stage of hominid organic evolution seems to have been accompanied by the major advances in cultural evolution. Because stone tools are relatively indestructible, much of early cultural evolution is represented by the evolution of tool industries.

The importance of tools as molders of hominid evolution has been recognized for a long time. Upright posture, leaving the hands free to manipulate objects and carry things form long distances, certainly is dynamically connected with the adoption of tool use by early hominids. In order for a tool to be useful, one must have it with him when the moment arises to put it to work. This takes making the tool in advance - planning; walking on two legs to free one's hands to carry the tool; and commitment to using the tool. To see how much of a commitment this represents, try carrying a brick around with you for a whole day. Those parts of the human brain most needed for manipulating tools are very evolved. Aside from the ones mentioned, other highly developed areas include the frontal lobes that organize behavior

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into sequences and the motor association areas that control the fingers and the thumbs. The hand itself is marvelously evolved. It combines the powerful curled - fingered grip with which heavy objects can be moved, with the delicate manipulations possible when small objects are held between the fingers and the thumb (and the ability to fully oppose the thumb to all the fingers is uniquely human).

Thus, much of what we take for granted about man today is the result of natural selection operating on his ancestors, adapting them to an environment he himself has created (or began to create): tools. But culture is more than just tools. One of the most important features of culture is language, which also profoundly influenced and was influenced by human evolution.

Language and evolution:

There are three areas of the brain that are highly evolved in the humans and appear to be crucial for human linguistic ability. One is called Broca's area and is located toward the front of the dominant side of the brain. This area activates, among things, the muscles of the jaw, lips, tongue, and larynx. The second is Wernicke's area, which is found in the temporal lobe of the dominant side of the brain. It is connected to Broca's area by a large bundle of nerve fibers (called the Arcuate Fasciculus) and is the brain site where verbal comprehension takes place. The third area is the Angular Gyrus, situated next to Wernicke's area serving as a link up between parts of the brain that receives stimuli from the sense organs of touch, hearing and sight.

Man could not possibly speak without these brain areas. It is interesting to note that all three are located in the cortex - the "new" brain - which, as already been emphasized is most evolved in humans and appears to have first approached its modern size and complexity in Homo erectus. The fact that all three are located in the cortex allows sensory inputs and verbal representations to be connected with each other without having to go through the "old" brain - especially the limbic system, which activates such very basic responses as aggression, fear, hunger and sexual arousal. Consequently, human beings can think, talk and experience the world without involving these "gut - level" states. Other animals, including our primate relatives, have not developed these brain areas nearly as much as man has. Thus many important aspects of man's brain seem to have evolved as speech specializations; and it can be reasonably supposed that verbal communication was so adaptive for the man's ancestors, that strong selective pressures molded these changes. Certainly, language is a principle cornerstone of human existence.

These, then, are the major themes that characterize human evolution; erect bipedalism, reduction of face, and expansion and development of the brain. All these themes interacted with each other, and all are dynamically connected to the evolution of social groups and culture - the primary mechanisms of human adaptation.

B. THEORIES OF ORGANIC EVOLUTION

(PRE-DARWINIAN, DARWINIAN & POST-DARWINIAN)

Meaning Of Organic Evolution: The English philosopher Herbert Spencer first applied the term "evolution" to denote the historical development of life. Evolution means change. The term "evolution" may be defined in several ways. Thus, we can speak of the geological evolution or evolution of planet earth, evolution of solar systems and the evolution of the automobiles, radios, telephones etc. The changes involved in the rise of human civilization can be called cultural evolution. In the same manner, the term "organic evolution" is applied to the changes that have taken place in the living things, viz., plants and animals. The whole idea of evolution probably started with Charles Darwin, who defined evolution as descent with modification. The word "descent" refers to the process of origin of new species from an ancestral stock. The word "modification" introduces an idea of change that is inherent in evolution. Thus, the term "organic evolution" may be explained in another way, i.e., the origin of new species of animals and plants from ancestors who previously lived on the earth.

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There are two different types of development, viz., ontogeny, and phylogeny. "Ontogeny" is primarily concerned with the history of development of individual organisms. Thus, for example, man begins his life as a single cell, which undergoes a complex process of development and finally culminates in a multi-cellular adult. These ontogenetic changes are of great importance only to embryologists. But, a student of evolution is primarily concerned with the second kind of development, which has been referred to as phylogeny. "Phylogeny" deals with the evolution of a genetically related group of organisms, in contrast to the development of the individual organism.

PRE-DARWINIAN PHASE

The rules of heredity have always intrigued humans for ages. In the fourth century B.C., Theophrastus, a Greek, has written a book on seed germination. Some early Greeks believed that heredity among humans was a question of which sex dominated in the sex act. Linnaeus suggested a two-layered theory, which held that "the outer layer including the vascular system is derived from the father, the inner layer including the nervous system comes from the mother". The theory of pre-formation, dating as far back as Aristotle was based on the belief that "the development of an organism is no more than unfolding of that, which is already present in miniature. Every organism must therefore contain in its reproductive organs an infinite series representing all of its future descendants". Ovists believed the female possessed this future of the series, and the spermists insisted it was the male.

While there were differences of opinion about the role of sex and ignorance of the laws of animal heredity, plants were not generally discussed in these terms. That plants lacked sexuality was the conventional wisdom of the day. Not until the end of the seventeenth century was the presence of sex organs in plants demonstrated in the experiments performed in Germany by Rudolph Camerarius. The discovery of sexual reproduction of plants was a notable achievement because it makes possible an experimental approach to plant hybridization. Once begun by Camerarius, crossing hybrids and observing what regularities might occur among the offspring became one of the most popular methods of investigating the laws of heredity.

Through hybridization experiments, which continue in the 18th and 19th Centuries, people were able to work out the rules of heredity. Though many investigators worked on hybrids, the secrets of hybridization, however, eluded them. What the early experimenters saw were offspring in a confusing array of forms and colors, which they were unable to explain. There conclusion – there probably were no universal laws of heredity! Thus, the matters stood more or less at a standstill as far as the laws of heredity were concerned, until Gregor Johaan Mendel solved the enigma in the 1860s.

THEORY OF INHERITANCE OF ACQUIRED CHARACTERS (LAMARCKISM)

The theory of inheritance of acquired characters states that ***modifications which the organism acquires in adaptation to the environments which it meets during its life time are automatically handed down to its descendants and so become part of hereditary.***

A renowned French naturalist, Lamarck shortly after the occurrence of French revolution, propounded this theory. As a result of his systematic studies he became convinced that species evolve continuously. This idea was in total conflict with the view of the period - fixity of species. As a result Lamarck's views were challenged by most of the biologists of that time, particularly by Georges Cuvier. In 1809, Lamarck published "Philosphic Zoologique", which included his theory explaining the changes that occur in the formation of new types. Although his views on evolutionary mechanism are outmoded now, he still occupies a very important place in the history of evolutionary thought. He was the first evolutionist to conclude that evolution is a general fact covering all forms of life. His evolutionary ideas are discussed below.

Lamarck's Evolutionary Propositions

The Lamarckian theory may be summarized in four propositions:

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1. Living organisms and their component parts tend continually to increase in size.
2. Production of a new organ result from new need and from the new movement which this need starts and maintains.
3. If an organ is used constantly, it will tend to become highly developed, whereas disuse results in degeneration.
4. Modification produced by the above principles during the life time of an individual will be inherited by its offspring.

Thus, Lamarck believed that organic changes seen in animals are a result of the influence of environment on gradual changes of species due to their tendency to become more and more perfect. According to him, when an animal's environment changes, its needs change and this leads to special demands on certain organs. Organs used more extensively would enlarge and become more efficient.

Conversely, an organ or organs no longer used would degenerate and atrophy. He postulated that such changed characteristics (acquired traits) would be transmitted to the offspring.

To explain his theory Lamarck used examples like long neck of giraffes, limblessness in snakes, webbed feet of ducks, blindness of moles, and also dimorphism of submerged and aerial leaves in aquatic plants. He visualized the evolution of the giraffe as follows: An original deer-like animal finding the supply of grass and herbs inadequate, started to feed on the leaves of trees. It needed greater height to reach the higher leaves and in the process of reaching, its neck became longer and longer. In the course of generations the long neck became a more accentuated feature, and our modern giraffe was the result. Likewise, according to Lamarckian evolutionary theory, the webbed foot of an aquatic duck would have developed in the following way: The duck would stretch its toes apart to give more push during swimming. This new characteristic would be inherited, and the subsequent generation of duck would, upon stretching their toes, form a more defined web. Each generation would do the same until the webbed foot seen on ducks today was fully formed. This would then be passed on from generation to generation, essentially unchanged once the perfected state was attained. For plants, Lamarck accepted the theory of his compatriot St. Helaire, who developed the notion that plant form is shaped by the combined effects of the environment.

Significance of Lamarckism

Lamarckian theory was simple and it had some appeal as it provided a way in which changes in organisms could come about. It was the first completely comprehensive theory that was offered. Further more, it was a theory that lent itself to predictions and therefore to testing. Thus, Lamarckian theory enjoyed popular acceptance for about 70 years because it was exemplified by many common examples. Most persons know that exercise results in larger muscles, deep-sea fishes have rudimentary eyes, and carnivorous animals possess claws. The theory also afforded a means of explaining reduced or vestigial structures. Further, through disuse, the eyes of a cave animal might become functionless and might even disappear.

Critical Analysis of Lamarck's Propositions

Lamarck defended his evolutionary theory vigorously until his death. For it, he suffered both social and scientific ostracism. He was criticized for following reasons by the contemporary scientists during his lifetime and afterwards.

1. The first preposition of Lamarck suggests the tendency to increase in size. While the evolutionary trend in a certain groups of organisms may be associated with increase of size, there are many cases, where evolution proceeded not only without any increase in size but rather through a reduction in size. Many plants contradict the Lamarckian principle by showing such a reduction in size during

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their evolution. Many ferns and conifers that became extinct were gigantic trees and the more highly evolved flowering plants are really much smaller in size.

2. The second Lamarckian principle that new organs result from new needs is quite manifestly false. In the case of plants, Lamarck believed that the environment acted directly upon the plant causing the production of such new characters as might adapt the plant to its environment. In the case of animals, he believed that the environment acted through the nervous systems; in other words, the desire of the animal leads to the formation of new structures. In its crudest form this would mean that a man who mused, *"Birds can fly, so why can't I?"* should have sprouted wings and taken to the air.

3. The third Lamarckian principle that organs will develop due to use and degenerate due to disuse may be correct as far as growth of an organ within the lifetime of an individual is concerned. For example, it is a commonly observed fact that if muscles are put to use these would develop. However, this principle is meaningful only when it is studied in relation to following fourth principle.

4. The fourth and final proposition of Lamarck was that the inheritance of characters acquired during the lifetime of the individual. This principle has been tested by many biologists through experiments and has found it entirely incorrect.

Experiments to Disprove Lamarckism:

1. The noted German scientist August Weismann was the first person who made a definite distinction between heritable changes and those which cannot be inherited. In 1890, he performed some experiments to test if characters may disappear due to disuse. This he did by cutting the tails of white mice for more than 20 generations to see if this has any effect on tail length. The measuring of tail length of the offspring of 20 successive generations revealed that on average, the tails were not shorter. It means that acquired character (cut tail in this case) was not inherited.

2. Castle and Philips performed transplantation experiments to show that environment has no effect on heredity. In one of the experiments they transplanted the ovary of a black female guinea pig into the body of white female guinea pig and the recipient female was mated with a white male guinea pig. They found that all the individuals from this pair were black. This shows that the environment does not affect the heredity as has been suggested by Lamarck.

3. Boring of ears and nostrils in Indian women has been continued as the tradition from centuries but their offspring do not show any trace of holes in ears and nostrils. The Chinese bind the feet of their women for many generations, yet this has not resulted in any modification of the feet of present day Chinese women.

THE DARWINIAN PHASE

THEORY OF NATURAL SELECTION (DARWINISM)

In 1858, two Englishmen published jointly in the proceedings of the Linnaean Society papers on a new concept of evolution. This concept, the theory of the natural selection was destined to influence greatly, the thinking of modern man in many different fields and to become the basis of most modern biological concepts of evolution.

One of these men, Alfred Russell Wallace (1823-1913) was a naturalist. In the year 1859, Darwin published a book, "The Origin of Species by Natural Selection" or "The Preservation of Favored Races in the Struggle for Life". This book has been judged by many as the most important book of the nineteenth century, since it contained overwhelming evidence of the fact that evolution has and does occur, and presented a logical theory regarding the major mechanism of evolution and natural selection.

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Darwin's Evolutionary Propositions

"The change in species by the survival of an organismal type exhibiting a natural variation that gives it an adaptive advantage in an environment, thus leading to new environment equilibrium, is evolution by natural selection".

Thus, natural selection is a continuous process of trial and error on a gigantic scale for all of living matter is involved. It includes following elements:

The Universal Occurrence of Variation: Variation is the characteristic of every group of animals and plants and there are many ways in which organisms may differ. (Darwin and Wallace did not understand the cause of variation and assumed it was one of the innate properties of living things).

An Excessive Natural Rate of Multiplication: Every species in the absence of environmental check tends to increase in a geometrical manner. If a population of a given species doubles in one year and if there are no checks on its increase, it will quadruple the next year and so on. Such a great reproductive potential of different species may be easily observed in nature. It has been estimated that a common Atlantic Coast Oyster may shed as many as 80 million eggs in one season. A salmon produces 28,000,000 eggs in a season. A single pair of English sparrows would be the ancestors of over 275 billion individuals in 10 years if they and their descendants could reproduce at their natural rate without any check. Darwin calculated that even a pair of elephants, which are about the slowest breeding animals known, could, in the absence of any checks, have 29 million descendants at the end of 800 years.

Thus, more organisms of each kind are born than can possibly obtain food and survive. Since the number of each species remains fairly constant under natural conditions, it must be assumed that most of the offspring in each generation perishes. If all the offspring of any species remained alive and reproduced, they would soon crowd all other species on the earth.

Struggle for Existence: Since more individuals are born than those that can survive, there is an intra-specific or inter-specific or environmental struggle for survival - a competition for food, mates, and space. This contest may be an active killed or be-killed struggle, or one less immediately apparent but no less real, such as the struggle of plants or animals to survive drought or cold.

Survival of the Fittest: The consequent elimination of the unfit and the survival of only those that is satisfactorily adapted. Some of the variations exhibited by living things make it easier for them to survive while others have handicaps that bring about the elimination of their possessors. This idea of "the survival of the fittest" is the core of the theory of natural selection.

Inheritance of Variations: The surviving individuals will give to the next generation their characters and in this way, the "successful" variations are transmitted to the succeeding generations. The less fit will tend to be eliminated before they have reproduced.

Successive generations in this way tend to become better adapted to their environment. As the environment changes, further adaptations occur. The operation of natural selection over many generations may produce descendants which are quite different from their ancestors, different enough to be separate species. Further more, certain members of a population with one group of variations may become adapted to the environment in one way, while others in a different way, or become adapted to a different environment. In this way two or more species may arise from a single ancestral stock.

Critical Analysis of Darwinism:

With the appearance of Darwin's book in 1859, a veritable storm of controversy broke out. Many people became interested in his presentation; the arguments (for and against) were often heated. The concept of natural selection was opposed primarily on ethical and religious grounds. But the sheer weight of evidence and the logic of Darwin's presentation finally convinced a majority of educated people. The concept of evolution affected many aspects of man's thinking and it will continue to do so. However, the

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idea of "the survival of the fittest" of Darwinism has been erroneously interpreted by many to mean survival only by "tooth and claw", an interpretation that has led in many cases to rationalizations for the attitude of "every man for himself" in social and economic affairs. In fact, the process of evolution is not at all "tooth and claw". Plants have evolved too and there is no bloody competition between them! In all forms, plants and animals alike, a part of the selective process involves the action of inanimate natural factors such as drought, moisture, temperature, etc. Active competition between organisms is a part of the process, but cooperation has in many cases been involved in survival. Natural selection results in the survival of those that are best integrated with the various factors of the environment in which they live.

The greatest weakness of Darwinism is that it did not explain the origin and transmission of variations. Although Darwin criticized many aspects of Lamarck's theory, he did not deny that acquired characteristics can be transmitted. He realized that the nature of inheritance was unknown, but he devised a working hypothesis called hypothesis of pangenesis to explain how acquired variations may be transmitted.

Pangenesis Hypothesis: Darwin's pangenesis hypothesis assumed that all the organs and perhaps all the cells in the body of an animal produced miniatures of themselves. These miniatures called gemmules or pangenes were shed into the blood stream and carried to the sex glands (viz., reproductive organs, the testes, and ovaries) where they were assembled to form sex cells - the eggs or sperms. Later, when a fertilized egg undergoes development, the pangenes present are responsible for the particular features of the new individuals. In this manner, environmental changes would produce modified organs which in turn would produce modified pangenes that would transmit the change to the next generation.

In the year 1875, Galton made pangenesis hypothesis untenable by presenting several experimental proofs. He and others made a series of experiments involving blood transfusions and later transplants of ovaries between black and white varieties of rabbits and chicken. Gametes produced by the transplanted ovaries were consistent with the phenotype of the individual in which the ovary originated and not with the animal currently carrying the ovary. Blood transfusions had no effect on the gametes produced. These experiments readily demonstrated that the pangenesis hypothesis was incorrect.

Certain other demerits of Darwinism are discussed below.

1. Darwinism or natural selection theory does not account for the beginning of organs, which may appear at first as rudiments having as yet no selection value. In other words, it remains concerned with the survival of fittest but not for the arrival of the fittest.

2. Over specialization in certain cases like extinct Irish deer in which huge antlers outweigh the entire skeleton, or the immense spiral tusks of the Jefferson Mammoth, or huge dinosaurs of Mesozoic, all cannot be explained on the basis of continuous variations and natural selection. These organs or body structures should not have reached such a harmful stage if natural selection was operating. However, such case of over specializations have been explained by Darwinism as the basis of discontinuous variations or "sports", which according to him, do not play any role in evolution.

3. Natural selection cannot account for degeneracy. To say an organ is no longer useful and hence disappears is to state the effect and not the cause. If under some changed conditions, a character built up by natural selection becomes a menace, the reversal of selection can accomplish its removal but his theory will not explain this phenomenon.

4. One of the classical objections to natural selection is that new variations would be lost by "dilution" as the individuals possessing them breed with others without them. We now know that although the phenotypic expression of a gene may be altered when it exists in combination with certain other genes, but the gene itself is not altered and is transmitted to succeeding generations.

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5. Darwin indirectly accepted the Lamarckian idea of inheritance of acquired characters in the form of pangenesis hypothesis, which cannot be accepted in the light of knowledge genetics made available later in that century.

6. Lastly, some people have objected to natural selection because it is essentially a materialistic doctrine, depending as it does, purely on the laws of chance.

POST–DARWINIAN PHASE

During later half of 19th century and at the beginning of 20th century many important ideas have been forwarded for explaining the inheritance of variations from one generation to successive generations and also for explaining evolution. Thus, the rediscovery of Mendel's laws of heredity by Correns and Tschermak made it clear that (1) the factors given to the offspring by the parents do not mix but are segregated and (2) if more than one pair of contrasting characters is considered in the same cross, the factors responsible for these are inherited independently. Likewise, mutation theory of evolution of Hugo de Vries (1886-1887) stated that new species arise by sudden changes or steps called mutations rather than by gradual processes. According to him, it was mutations and not selection that should be considered as the primary factor in evolution. Further, the studies of Wagner (1868) suggested the role of geographic or spatial isolation in the formation of every species, race, or tribe of animals or plants on the earth. Even more, certain population geneticists realized that the actual physical struggle between animals for survival or the competition between plants for space, sun and water is much less important as an evolutionary force than Darwin believed. The evolution of any given kind of organism occurs over many generations during which individuals are born and die, but the population has certain continuity. Thus, the unit in evolution is recognized not as the individual but rather a population of individuals.

A population of similar individuals living within a circumscribed area and interbreeding is termed a deme or genetic population. The next larger unit of population in nature is the species, composed of a series of inter-breeding demes. Further, in a population, the relative frequencies of the genes will remain constant from one generation to the next (1) if the population is large, (2) if mating occurs at random, (3) if no mutations occur and (4) if there is no migration of individuals into or out of the population. The operation of the Hardy-Weinberg principle will result in maintaining a given gene frequency in a population. Thus, a population undergoing evolution is one in which the gene pool is changing from generation to generation. The gene pool is the sum total of all the allelic genes in a population and the gene pool of a given population may be changed (1) by mutation, (2) by hybridization, that is the introduction of genes from some outside population or (3) by recombination brought about by crossing over and by the assortment of chromosomes in meiosis may also lead to new combinations of genes and phenotypes with some specific advantage or disadvantage for survival that would be reflected in a change in the gene pool.

All the modern understandings in cytology, genetics, cytogenetics, population genetics, and evolution gave a way for the formulation of a coherent theory called "Modern Synthesis" around 1930s' by S.Wright, H.J.Muller, Th.Dibzhansky, R.B.Goldschmidt, J.S.Huxley, R.A.Fisher, J.B.S. Haldane, Ernst Mayr and G.L.Stebbins.

C. SYNTHETIC THEORY OF EVOLUTION

By the beginning of the 20th Century, the essential foundations for evolutionary theory had already been developed. Darwin and Wallace had articulated the key principle of natural selection forty years earlier and the rediscovery of Mendelian genetics contributed the other major component – a mechanism of inheritance. But these two basic concepts did not join rather quickly into a consistent theory of evolution. For the first thirty years of the 20th Century, geneticists emphasized sharp contrasts within particular characteristics of organisms. As such, evolution was seen as a process of fairly large radical "jumps" and this "mutationist" view came to be seen as an alternative to the "selectionist" tradition. A *synthesis* of these two views was not achieved until the mid 1930s and we owe much of our current view of the evolutionary process to this intellectual development.

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Biologists working on models of evolutionary change in the late 1920s and early 1930s came to realize that genetic and selective processes were not opposing themes, but that a comprehensive explanation of organic evolution required both. Small new changes in the genetic material are in fact the fuel for natural selection. The two major foundations of the biological sciences had thus been brought together in what Julian Huxley termed the "The Modern Synthesis".

From the perspective of modern synthesis, evolution is a two stage process:

1. Production and redistribution of **variation**
2. Natural selection acts on this variation

Darwin saw evolution as the gradual unfolding of new varieties of life from previous forms over long periods of time. This depiction is what most of us think of as "evolution" and it is indeed the end result of the evolutionary process. But these long-term effects can only come about by the accumulation of many small evolutionary changes occurring in every generation. In order to understand how the process of evolution works, we must necessarily study these short term events. Today we study evolutionary changes occurring between generations and are able to demonstrate how evolution works. From such a modern genetic perspective, we define evolution as a ***change in allele frequency from one generation to the next.***

Allelic frequencies are numerical indicators of the genetic makeup of a population, the latter being an interbreeding group of individuals. An inherited trait may be of slightly different form in different individuals. We call the variant genes that underlie these different forms of in inherited trait, alleles. The different expression of inherited traits is the result of genetic variation within a population. The frequencies for combination of genes represent the proportions of a total and hence allele frequencies refer to only the whole groups of individuals i.e. populations. Individuals do not have allelic frequencies; they have the genes or the combination of these genes. Nor can individuals change alleles. From conception onward, the genetic composition of an individual is fixed. Therefore, an individual cannot evolve; only a group of individuals – a population – can evolve over time.

Evolution is an incredibly common phenomenon and may occur between every generation for every group of organisms in the world, including humans. Over a period of time, the relative proportions of alleles in the population will change, some may increase, and some decrease and still others may remain the same. Over a short run of just a few generations, such changes in inherited traits may be only very small, but if further continued and elaborated, the results can and do produce spectacular kinds of adaptation and whole new varieties of life. Whether we are talking about such short-term effects in one population from one generation to the next, which is sometime called **microevolution** or the long-term effects through fossil history, sometimes called **macroevolution**, the basic evolutionary mechanisms are similar.

Now, how do allelic frequencies change? Or to put it in another way, how does evolution occur? The modern theory of evolution isolates general factors that can produce alterations in allele frequencies. As already stated, evolution is a two stage process. Genetic variation must first be produced and distributed before it can be acted upon by natural selection.

Factors that Produce & Redistribute Variations

Mutations: An actual alteration in the genetic material is called mutation. A mutation is a change in the base sequence of DNA. For such changes to have evolutionary significance, they must occur in the sex cells, which are passed between generations. Evolution is a change in the allelic frequencies between generations. If mutations do not occur in sex cells, either the egg or the sperm, they will not be passed to the next generation and no evolutionary change can result. If however a genetic change does occur in the sex cells, this change or mutation can change the allelic frequencies in the subsequent generation.

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Geneticists often distinguish between two kinds of mutations, chromosomal aberrations, which may result in alterations in the amount of position of genetic material and point mutations, which are permanent, heritable changes within a gene. Mutations occur frequently in the nature and have been reported in many organisms. In man mutations cause variation in hair, color, skin pigmentation and several somatic malformations. Usually mutations bring about changes in the genetic structure at the gene level. All those factors which bring about a change above the gene level are termed as chromosomal aberrations. Chromosomal aberrations bring about a change in either the structural aspects of a chromosome or they may bring about a change in the number of the chromosomes present in the organism.

Actually, it would be rare to see evolution occurring by mutation alone. Mutation rates for any given trait are quite low, and thus, their effects would rarely be seen in small populations. In larger populations, mutations might be observed but have little impact on shifting allele frequencies. However, when mutation is coupled with natural selection, evolutionary changes are quite possible.

Mutation is the basic creative force in evolution and in fact is the only way to produce "new" variation. Its key role in the production of variation represents the first stage of evolutionary process. Darwin was not aware of the nature of mutation. Only in the last century, with the spectacular development of molecular biology, have the secrets of this phenomenal force have been revealed to evolutionary biologists.

Migration: Migration is the movement of genes from one population to another. If all individuals in a population do not choose their mates from within the group, significant changes in allele frequencies could occur. If a change in the allelic frequency does take place, evolution will have occurred, this time by migration. Migration works both ways – changes in the genetic frequencies will be affected in both the cases of in-migration and out-migration. In humans, social rules, more than any other factor determine mating patterns, and cultural anthropologists must work closely with physical anthropologists in order to isolate and measure this aspect of evolutionary change. Detailed explanation on this factor can be found in the chapter on Population Genetics..

Genetic Drift: The random factor in evolution is called **Genetic Drift**, and is due primarily to sampling phenomena (i.e., the size of the population). Since evolution occurs in populations, it is directly tied not only to the nature of the initial allele frequencies of the population, but to the size of the group as well. If, in a population of say 100 individuals, two blood type O individuals are killed in an auto accident before completing reproduction, their genes would have been removed from the population. The frequency of the O allele would have been reduced in the next generation, and evolution would have occurred. In this case, with only 100 individuals in the population, the change due to the accident would have altered the O frequency in a noticeable way. If, however, the initial population had been very large (say 10,000 people), then the effect of removing a few individuals will be very small indeed. In fact, in a population of large size, random effects such as traffic accidents would be balanced out by the equal probabilities of such events affecting all the other individuals with different genetic combinations (i.e., different genotypes). As you can see, evolutionary change due to genetic drift is directly and inversely related to population size. To put it simply, the smaller the population, the larger is the effect of genetic drift.

When considering genetic drift, we must remember that the genetic makeup of individuals is in no way related to the chance happenings that affect their lives. When applied to our example, this fact means that the genetic makeup of individuals has absolutely nothing to do with their being involved in automobile accidents. This is what is meant by random event and why this factor is usually called **random genetic drift.** If, for example, a person had died in an auto accident caused by hereditary poor eyesight, such an event would not be genetic drift. If the individual, because of some such hereditary trait, dies early and produces fewer offspring than other individuals, this is not random genetic drift but natural selection.

Recombination: Since in any sexually reproducing species both parents contribute genes to the offspring, the genetic information is inevitably reshuffled every generation. Such recombination does not in itself change allele frequencies (i.e. cause evolution). However, it does produce the whole array of

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genetic combinations, which natural selection can then act upon. In fact, the reshuffling of chromosomes during meiosis can produce literally trillions of gene combinations, making every human being genetically unique.

The phenomenon and the process in which the genetic combination of the offspring is different from the parental combination is called recombination or genetic recombination. Compared to mutation, recombination has got greatest role to produce variability. Mutation and gene flow or migration can produce variability in a population with respect to single gene. Recombination can combine the novel alleles, which at first are likely to be carried out by different individuals in a single genotype. Furthermore, recombination can multiply the number of different gene types in the population. It converts a small initial stock of multiple gene variation into a much greater amount of genotypic variation.

Adaptation is the attribute of the assembly of beneficial genes rather than a single gene. These assemblies are harmonized in the covers of evolution to suit best in the environment and obviously, such assembly is essential for the evolution. Recombination is the only process which can form such assembly. Sexual reproduction is the chief mechanism of recombination in higher eukaryotes which have high chromosome number, have high gene number, and reproduce sexually. Hence these organisms have maximum recombinant forms. This is the reason why no two individuals are alike, which develop from different zygotes (exception being identical twins which develop from same zygote).

The commencement of evolution is from mutation in the form of multiple gene variation. Later, through recombination the assembly of adaptive gene is over and they are competent enough to be selected i.e., attainment of requisite adaptation. Thus, recombination is the mid point in the process of evolution.

Natural Selection Acts on Variation

The evolutionary factors just discussed – mutation, migration, genetic drift and recombination – interact to produce variation and to distribute genes within and between populations. But there is no long-term direction to any of these factors. What then does enable populations to adapt to changing environments? The answer is, of course, natural selection – the process so well elucidated by Darwin more than 125 years ago. Given that there is genetic variation among individuals within a population, some of these variations may influence reproductive success (number of offspring successfully raised). If, as a result of genetic variation, some individuals contribute more offspring to succeeding generations, this is natural selection. In fact, natural selection is defined as ***differential net reproductive success.***

How then do populations adapt? A result of natural selection is a change in allele frequency relative to specific environmental factors. If environment changes the selection pressures change as well. Such a functional shift in allele frequencies is what we mean by **adaptation.** If there are long-term environmental changes in a consistent direction then allele frequencies should also shift gradually each generation. If sustained for many generations, the results may be quite dramatic.

Unit of Selection: Selection acts on individual. It is individuals who reproduce or do not reproduce and who continually attempt to maximize their own reproductive success. Thus, *the individual, not the group, is the unit of selection.* If the total reproductive success of all members of a population continuously falls below replacement value (where more individuals die than are born in a generation) the population will become extinct. Individuals will attempt to maximize their own reproductive success even in the face of such impending extinction.

Overpopulation is the result of individuals maximizing their reproductive success. Even if such behavior means the whole group will perish, no special evolutionary mechanisms exist to keep individuals from reproducing at their greatest capacity. Of course, humans have the potential to manipulate their numbers, and the entire world now faces the problem of controlling an exploding population. Evolution, therefore, has no built-in mechanism to guard against extinction. Indeed, extinction is actually the rule – not the

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exception – in evolution. Of all the species that have ever lived on earth, it is estimated that less than .1% are now currently living; the remaining 99.9% met their almost inevitable evolutionary fate.

The Unit of Evolution: While selection acts on the individual, changes in allele frequency occur between generations for an entire population; that is, an interbreeding group of organisms. The net result of all individuals' reproductive success (natural selection) – in addition to the possible effects of mutation, migration, and genetic drift – will affect the entire population. *The population, then, is the unit of evolution,* and it is the population that changes from generation to generation.

Modes of Selection: Depending upon the environmental change i.e., the rate at which the change occurs like slow change, abrupt change or change in the ecological diversity, the population is thrown to different environments and is hence selected accordingly. This sets particular pattern of selection which is termed as the mode of selection. Basically there are three modes of selection.

1. **Directional Selection:** As environmental pressures change gradually, selection pressures should also shift. If the environmental pressure is directional, selection should also be directional. For example, in the last one million years, there have been long periods of gradual cooling in the earth's climate. One means of coping with cold among mammals has been increase in body size. If we assume animals, such as mammoths, are descendants from smaller ancestors, how did they gradually get bigger? The answer lies in understanding natural selection. Those animals with genotypes for lager overall size perhaps resisted cold better, lived longer, mated more often, cared better for their young, and so forth. In any case, the result was that they reproduced more successfully than other smaller individuals. As the climate continued to grow progressively colder, allele frequencies also continued to shift, and average mammoth size gradually increased.

2. **Stabilizing Selection:** If, on the other hand, environments remain relatively stable, there should be selection for those genotypes already established within an "adaptive plateau". In other words, those phenotypes in the center of the population distribution ("modal" varieties) should have higher reproductive success, and those at either extreme will be selected against, as long as the environment remains stable. If we again choose size as a characteristic, we can note that some varieties of turtles have changed little in millions of years. In any given generation, the environment of turtles has not changed much from that of previous generations. Therefore, the "optimal" phenotype is probably an average-sized turtle, and those much larger or much smaller should be selected against.

3. **Diversifying Selection:** In this type of selection, phenotypic variants at both extremes are favored, and those in the center of the distribution (closer to the mean) are selected against. For example, in the case of baboon size relative to predation pressure, it may be advantageous to be small (less conspicuous) or very large (for active defense). Over time, such selection pressure would act to create a twin-peak (bimodal) phenotypic distribution. In fact, such a process would not have long-term effects in a fully reproductive population, since individuals of all sizes would be mating with each other, thus producing individuals of intermediate body size. In order for the diversifying selection pressures to be maintained, some degree of reproductive isolation between segments of the population would be required. Another possibility is that phenotypic variation can be partitioned and maintained by other genetic differences – sex, for example. In fact, baboons do differ markedly in size, with males averaging about twice the size of females.

Evolution at the Species Level

A species is defined as a group of interbreeding organisms that are reproductively isolated and, therefore, cannot successfully interbreed with other groups (species). The capacity to reproduce is the critical factor in defining species. Theoretically, one can test whether two kinds of organisms are members of different species by observing their reproductive behavior under natural conditions (who mates with whom), and by observing the results (are the offspring fertile?).

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Actually, a species is composed of subunits that are breeding communities, which we have already defined as populations. All members of a species can potentially interbreed, and some degree of interbreeding (migration) is theoretically possible between all populations of that species. The net result of all forces of evolution acting on all populations determines the fate of the species as a whole. If sustained over a long period of time, gradual changes in allele frequencies between member populations can eventually lead to sufficient genetic differences, so that fertile reproduction is no longer possible. We then may recognize a new form of life having arisen from one species "splitting" and producing new species, a process called **speciation.** If on the other hand, total reproductive success is so low that all or even most populations become extinct, the whole species will be doomed and will disappear from the earth forever.

Isolation & Speciation

According to Mayr, the way new species are first produced involves some form of isolation. Picture a single species of some organism composed of several populations distributed over a wide geographic area. Gene exchange (migration) will be limited if a geographic barrier, such as a river, ocean, or mountain range, separates these populations. This extremely important form of isolating mechanism is called ***geographic isolation.***

Now, if one population (A) is separated from another population (B) by a mountain range, individual animals of A will not be able to mate with the animals of form B. As time passes (several generations) genetic differences will accumulate in both populations. If group size is small, we can predict that drift will cause allele frequency changes in both population A and population B. Since drift is random in nature, we would not expect the effects to be the same. Thus, the two populations would begin to diverge.

As long as gene exchange is limited, the populations can only become more divergent with time. Moreover, further genetic difference would be expected if the groups are occupying somewhat different habitats. These additional genetic differences would arise through the process of natural selection. Certain individuals in population A may be most reproductively fit in their own environment, but may be less fit in the environment occupied by population B. Consequently, allele frequencies will change, and the pattern will be dissimilar in two populations.

Again, as the time passes, and perhaps as environments change further, the populations will become still more divergent. With the cumulative effects of drift and natural selection acting over many generations, the result will be two populations which could no longer interbreed fertilely. More than just geographic isolation would now apply; there may, for instance, be behavioral differences interfering with courtship, what we call ***behavioral isolation.*** There may even be sufficient differences in body size and proportions to make copulation physiologically impossible, what we call ***mechanical isolation.*** Using our biological definition of species, we now would recognize two distinct species where initially only one existed.

The pre-requisite for speciation is reproductive isolation. Mechanisms that prevent successful reproduction between members of two or more populations (viz., closely related species) that have descended from the same original population are called isolating mechanisms. The significance of isolating mechanisms (i.e., reproductive isolation) in speciation is first of all recognized by the post-Darwinian evolutionist, Dobzhansky in 1937. Since that time a great amount of literature has accumulated about isolating mechanisms.

Most modern evolutionists such as Mecham (1961), Mayr (1948, 1970), Stebbins (1966, 1971), etc., have classified the reproductive isolating mechanisms into two classes namely pre-mating or pre-zygotic isolating mechanisms and post-mating or post-zygotic isolating mechanisms. Both types of isolating mechanisms differ fundamentally with each other in following manner: Pre-mating or pre-zygotic isolating mechanisms prevent wastage of gametes (germ cells) and so are highly susceptible to improvement by natural selection; Post-mating or post-zygotic isolating mechanisms do not prevent wastage of gametes and their improvement by natural selection is indirect (Mayr, 1970). It is further

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recognized that for these reproductive isolating mechanisms to evolve, the separated populations of an original single group must be separated spatially or geographically or by time.

One of the evident functions of isolating mechanisms is to increase the efficiency of mating. Where other closely related species do not occur, courtship signals can "afford" to be general, non-specific, and variable. Where other related species coexist, however non-specificity of signals may lead to wasteful courtship and delays, even where no heterospecific hybridization occurs. Under these circumstances there will be a selective premium on precision and distinctiveness of signals. Moreover, each species is a delicately integrated genetic system that has been selected through many generations to fit into a definite niche in its environment. Hybridization usually leads to a breakdown of this system and results in the production of disharmonious types. It is the function of isolating mechanisms to prevent such a breakdown and to protect the integrity of the genetic system of species. Any attribute of a species that would favor the production of inferior hybrids is not selected, since it results in wastage of gametes. Such selection maintains the efficiency of the isolating mechanisms and indeed helps to perfect them. Isolating mechanisms are among the most important biological properties of species.

BASIC CONCEPTS AND TERMS IN EVOLUTIONARY BIOLOGY

CONVERGENCE

In some situations, similarities are not the result of being in a line of descent or of common origin, for some organisms may be alike in living habits and appearance, although of different ancestries. Such similar evolutionary development in different forms is termed **Convergence.** Convergence refers to the development of similar characteristics or adaptations in animals that differ in direct ancestry. The hummingbird and the hummingmoth, for example, have converged in their flying habits as a result of their common search for nectar in flowers as a source of food. Convergence ordinarily applies to one or a few characteristics rather than to the overall makeup. Similarities in the retina, the layer of visual cells in the eyes, of some quite different nocturnal animals are an example. Of the two main types of retinal cells (rods and cones), only rods, which are more sensitive to dim light, are present in some deep-sea fish, bats, some lizards and snakes, and probably guinea pigs, whales and some lemurs. All these animals, however, differ markedly from each other in respect to other characteristics less directly related to their adaptation to low light conditions. It is improbable that any instance of evolutionary convergence has been as dramatic and complete as to hide all traces of the diversity of origins. A number of similarities between tarsier and human skulls once were thought by some to demonstrate that the tarsier, not the great apes, was the closest living relative to humans. This is now known to be a convergence caused by the fact that the tarsier and we have both evolved large orbits for large eyes, along with small noses, as less dependence on the sense of smell was the trend.

PARALLELISM

An evolutionary development similar to convergence, but in *related forms* is **parallelism.** Parallelism implies a similarity in biological makeup of the ancestral forms, whereas convergence does not. If the common ancestor of two organisms were not very ancient, and if evolution in the descendant lines followed more or less the same course, the term *parallelism* is used. The term is usually applied to two species of organisms that were similar in origin, and that remained similar as they evolved like having some of the same changes occurring in both of them even after they have separated and evolved into two different species. The Old World and New World Monkeys provide an excellent example of parallelism between groups living today, since they appear to have evolved in parallel from a prosimian ancestor that probably lived at least 35 million years ago.

The reason for parallelism as well as convergence is the same. The organisms, in order to survive in similar environment, must develop similar biological structures. Parallelism, like convergence, is a matter of adaptation under the control of natural selection. The lack of a tail in gibbons, on the one hand, and the great apes and humans, on the other, is probably a case of parallelism, since their common ancestors

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probably had tails that were lost in a parallel fashion in the separate evolutionary lines after they diverged. All the monkeys, however, have tails. The Cercopithecidae, the monkeys most closely related to humans and apes, are quite varied in tail length and those species with similar tail lengths are not the most closely related to each other. The tail is a functionally important member used for balance, and very diverse species of Cercopithecidae (the Colobus monkeys and the vervets, for instance) are both arboreal and have long tails, probably as a parallel evolutionary adaptation to arboreal quadrupedal locomotion. Similarly, the reduction of the tail in the brachiating gibbons and the terrestrial Hominoidea is probably a parallel response to locomotor requirements.

HOMOLOGY AND ANALOGY

Another way of looking at some of the same phenomena places emphasis on the structures rather than on the processes by which they evolved. In evolutionary biology the term **homology** means similarity in origin (implying a common ancestor), **homoplasy** means similarity in appearance but not in origin, and **analogy** means similarity in function but not in origin. Thus the wings of different species of birds are homologous as wings, but there is no homology between the wings of a bird and a bat. Wings of bird and bat, considered as wings are merely analogous, but both evolved from parts of the arm, forearm, and hand of four-limbed, five-fingered vertebrates, and as such are homologous as forelimbs (along with seal's fingers and the human upper extremity). The wings of the hummingbird and the hummingmoth, however, are not homologous by any standard (except perhaps as animal appendages); they are analogous as wings.

Evaluation of structures in terms of homology, analogy, and homoplasy can be valuable in determining whether apparently similar organisms have evolved in a convergent or parallel manner, and prior knowledge of the evolutionary history of particular species aids in identifying homologous and analogous structures. Independent information is also important in making such assessments, but it should be obvious that parallel evolution following divergence of lines tends to result in homologies, while analogies and homoplasy are likely results of convergent evolution.

SERIAL HOMOLOGY: Serial homology is the similarity of structures between one part of an animal with another part of the same animal. In this case, two or more structures of a single individual are compared. The arm and leg of man illustrate an instance of serial homology. The humerus bone of the upper arm corresponds to the femur of the leg (thigh). The radius and ulna of the fore arms correspond to the tibia and fibula of the legs (shin). The wrist bones (carpals) may be comparable with ankle bones (tarsal). The bones of the palm of the hand (metacarpals) correspond to the bones of the foot (metatarsals). Finally, the bones of the fingers as well as toes correspond to each other (phalanges). However, the arms of man are suited for grasping and handling, while the legs are used in locomotion.

ADAPTIVE RADIATION

The evolutionary spread and differentiation of the descendants of one type of animal, of whatever level of classification, is called **adaptive radiation.** Unlike parallelism and convergence, which refer to the ways two species of organisms remain similar or become more similar, adaptive radiation refers to the way a particular species evolves into progressively dissimilar organisms. The descendants of a single species sometimes evolve to take advantage of many different environments and opportunities, and rapid changes in the external environment may cause new forms of animals to develop from a single ancestral form. The evolution of a trait that opens up many new possibilities may also give rise to adaptive radiation.

Adaptive radiation is well exemplified by the history of the mammals. With the geological revolution that marked the end of the Mesozoic era (the age of the reptiles) and the start of the Tertiary, the previously stable climates became more changeable. The dinosaurs did not adapt and so became extinct, while the mammals evolved in many distinct lines. The rodents specialized for gnawing, the carnivores for hunting, the hoofed animals for grazing, the primates and sloths took to the trees; the whales, seals, and sea cows adapted for life in the oceans, and the bat took to the air. Furthermore, each of these mammalian orders

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in turn gave rise to sub lines that colonized new environments by acquiring new modes of life. Many of today's mammals are far different from their primitive common ancestors of the Paleocene epoch.

In addition, various orders and suborders of mammals have undergone further differentiation, branching or "radiating" into types adapted to different habitats. Thus, all the chief groups of primates today include species with contrasting dietary habits. Insect eating, seed eating, leaf eating, and more or less omnivorous genera recurred in different branches of the primates as these branches departed more or less from the ancestral line. This adaptive radiation within the branches is thus accompanied by *parallelism* between the branches and by *convergence* of adaptations towards those of some non-primate lines.

DOLLO'S LAW

Louis Dollo, a Belgian paleontologist, in 1893 has published the theory that evolution is irreversible. There are no examples of present forms that have developed independently and that are exactly like some ancient prototype. There are, however, examples of the re-evolution of types highly similar in some particular respects to ancient forms. For example, the extinct flying reptiles, the pterosaurs, developed wings but eventually died out. However, wings were re-evolved independently in the birds and bats. A change of the natural environment in an exact step-wise reverse order is exceedingly rare; hence, an occasion for reversed natural selection involving a number of factors in a given order would be just as rare. Furthermore, if a previous allele has been entirely eliminated from the gene pool, a reversal of evolution would also require precisely the "correct" reverse mutation just as the environmental conditions also reverse in proper order. For instance, the implication of a number of biologically absurd "horror" movies notwithstanding, it is difficult to imagine that a nuclear holocaust would leave human survivors exactly like some of the fossils we shall discuss in subsequent chapters.

Single steps backward, returning to an ancestral condition, sometimes occur in reversed mutations, as has been well documented in bacteria. However, that such reversals will not recapitulate a whole sequence is evidence for Dollo's principle. Evolution is thus irreversible to the extent that true **atavisms** or throwbacks to earlier forms, do not occur in detail. The cetaceans (whales and dolphins) may have returned to the aquatic environment in which vertebrates first evolved, but they have not once again become fish. They retain air-breathing lungs and numerous other physiological traces of their terrestrial past, and their flippers and streamlined shape are merely **analogous** to similar structures (fins) and shape in fish. The many different evolutionary details thus prevent a complete reversal of evolution.

MOSAIC EVOLUTION – THE EVOLUTION OF PARTS AND WHOLES

According to this principle, the evolution of the species tends to be inconstant and asymmetrical. That is, it may be rapid at one time and slow at another. In rare cases it may even virtually stop altogether. At one time, evolution may affect the limbs, and at another it may affect the jaws. With a change in the food supply or some other alteration in the environment, running or biting ability, for example, would become more important or less important in natural selection. This variability in tempo of evolution of different anatomical structures in the same line (that is, in ancestors and descendants) makes it unwise to draw sweeping conclusions concerning the relationship of two fossil forms on the basis of single characteristics. Instead, it is necessary to follow the evolution of whole functional systems. Since the systems themselves sometimes evolve at different rates, one must also take into account the total morphological pattern of the animal insofar as it is preserved. Although all past animals must have been integrated in the same way as existing ones, the interrelationships between functional systems in a single fossil animal are not always self-evident. Knowing the conditions under which it lived may help interpretation, but often the wisest response to a fragmentary fossil is suspended judgment while one searches for additional material. A few observations or measurements evaluated out of context can lead the unwary anthropologist far astray.

The evolutionary trends of different organ systems like locomotor system, nervous system, respiratory system, and so on, respond separately to environmental conditions. The systems can therefore sometimes

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be studied separately. For example, from studies of comparative anatomy and function it is possible to identify some particularly striking hallmarks of Homo sapiens, such as the bipedal gait and the relatively large and functionally differentiated brain. The evolutionary trends toward these characteristics, as they unfold in the fossil record, are referred to as **bipedalization** and **encephalization**, respectively. The evolution of these traits was distinct and separate, with bipedalism apparently preceding encephalization. Nevertheless, the development of a cultural way of life made both advantageous and, in this sense, both are part of the same process of human evolution. The complex **mosaic** pattern of human evolution led first to the evolution of a manipulative apparatus with hand and eye coordination, then to bipedalization to permit use of the hands even during locomotion, but human encephalization was late and rapid. In each case there is a feedback to the earlier established trend: Encephalization reinforces the bipedalism, and bipedalism permits further evolutionary emphasis on a life niche utilizing exact hand-eye coordination. The evolution of different structures at different rates (but all interacting as parts of an evolving whole) was a source of confusion to evolutionists for many years. It was this sort of confusion, which in part, allowed the Piltdown Hoax to be taken as a "missing link" earlier in the last century. After the recognition that mosaic evolution is the usual pattern, many theoretical and practical problems became easier to resolve, among them the question of missing links.

COPE'S RULE

According to this principle, living organisms have a tendency to increase in their size during the course of organic evolution. This tendency has been found in vertebrates, invertebrates, and plants. To some extent this principle is very similar to Lamarck's proposition that every organism has a tendency to continually increase in size. Though a majority of present species are probably the largest of their respective class, this principle is not universal and has exceptions in both plant and animal kingdoms. Refer to the criticism leveled against the first proposition of Lamarckism for further details.

PRINCIPLE OF COMPETITIVE EXCLUSION OR GAUSE'S RULE

In the normal course of evolution, if two organisms occupy the same trophic level in the ecosystem, these organisms try to reach equilibrium through different strategies. Like for example, two organisms may adaptively radiate to occupy different niches in the same trophic level. However, in some cases, it so happens that the very presence of a competitor results in the domination of other organism which tries to adapt very effectively ultimately resulting in the exclusion of the competitor. Such adaptive strategies adopted by the organism to reduce the other to the point of extinction, called Competitive Exclusion, is not seen in these organisms in the absence of a competitor. The famous Russian biologist Gause has conducted experiments in this context on different species of paramecium and explained how effectively each organism causes or simulates a selection pressure on the other.

1.5 CHARACTERISTICS OF PRIMATES

PRIMATES EVOLUTIONARY TRENDS

The problem of defining and characterizing the primates on the basis of anatomical traits is an old one, which has long confronted students of this mammalian group. Not only do living primates lack the distinguishing bodily specializations of other mammalian orders but they also present an amazing variety of forms and grades of organization. The problem is only compounded when fossil forms are considered. Fortunately, the inability to satisfactorily define the primates has not discouraged people from studying them.

The best and most useful solution to the problem of a definition was devised by one of the foremost primate anatomists, Sir Wilfred E. LeGros Clark. After remarking on the general lack of specialization, he proceeds to characterize the order by a series of evolutionary trends. The beauty of this approach is that it not only permits a more accurate definition of the primates but at the same time conveys a wealth of

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information about primate evolution, organized in a way that relates numerous anatomical details to a few important generalizations.

Clark's ten evolutionary trends are the following...

1. The preservation of a generalized structure of the limbs with a primitive pentadactyly, and the retention of certain elements of the limb skeleton (such as the clavicle) which tend to be reduced or to disappear in some groups of mammals.
2. An enhancement of the free mobility of the digits, especially the thumb and big toe (which are used for grasping purposes).
3. The replacement of sharp compressed claws by flattened nails, associated with the development of highly sensitive tactile pads on the digits.
4. The progressive abbreviation of the snout or muzzle.
5. The elaboration and perfection of the visual apparatus with the development to varying degrees of binocular vision.
6. Reduction of the apparatus of smell.
7. The loss of certain elements of the primitive mammalian dentition, and the preservation of a simple cusp pattern of the molar teeth.
8. Progressive expansion and elaboration of the brain, affecting predominantly the cerebral cortex and its dependencies.
9. Progressive and increasingly efficient development of those gestational processes concerned with the nourishment of the fetus before birth.
10. Prolongation of postnatal life periods.

The first thing to keep in mind is that these are trends rather than characteristics. They are not uniformly present, but are expressed to varying degrees among the members of the order. As a rule, except for the first one, these trends are less well developed in the living prosimians and in fossil forms than in the more advanced, modern Higher Primates.

Though there are ten trends listed, it can be readily seen that they relate to evolution in three principal areas: (a) the limbs and extremities (trends 1-3); (b) the head- eyes, brain and dentition (trends 4-8); and (c) the life cycle-reproduction, growth, and longevity (trends 9 and 10). All of these trends, including number 10, are recognizable to some extent in fossils as well as in living forms. They are all interrelated and adaptive to a mode of life which has been the outstanding factor in primate evolution-the arboreal habitat. Other mammals have taken up arboreal life but none have done it in exactly the same way. Many of man's characteristics are simply refinements of a basic primate pattern that evolved millions of years ago in response to the necessities of arboreal living.

Trend 1 really denotes a lack of change which is in itself remarkable among mammals. The primates have kept the same basic limb structure of their primitive ancestors. There is a single bone in the upper segment of each limb (upper arm and thigh) and paired bones in the lower segment (forearm and leg) with five digit (pentadactyl) extremities. In addition, a clavicle, or collarbone, has been retained in the shoulder girdle. These features contribute to the great mobility of the limbs needed in climbing and grasping. The paired bones in the lower segments of the arm and leg permit some degree of rotation. The five digits are useful in grasping. The collarbone functions to hold the upper limb out to the side of the chest permitting a greater reach and range of movement in a variety of directions. Since the features involved in this trend are skeletal structures, they are observable in fossils where the limb skeleton or parts of it have been preserved.

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Trend 2, having to do with prehension or grasping ability, is essential to the primate adaptation of climbing by grasping. Evolutionary advancement along these lines is indicated by the ability to move the digits independently of one another. The most advanced digits in this respect are the thumb and great toe that are not only independent, but opposable in the majority of primates; that is, they diverge in their axis from the other digits and their tips can thus be brought opposite those of the other digits. Opposability is undeveloped in the tree shrew, is not complete in the prosimian, and has been lost in the human foot that has become a weight bearing rather than a grasping organ. The human condition is atypical of primates, most of whom have grasping hands and feet. Naturally, those species in which digits have been reduced or lost also represent exceptions. The structure and functions of the hand and the implications of it will be discussed more fully under locomotory adaptations.

Trend 3 involves modifications of the tips of the digits that facilitate finding and maintaining a secure grip in the trees. Nails on the backs of the digits and tactile pads on the opposite surface evolved together, the former providing protection and support for the latter. Nails undoubtedly evolved from claws that became flattened out in the process and lost their deep horny layer. Tree shrews have claws on all ten digits. The aye-ayes and the marmosets have claws on all digits except the great toe. The lemurs, lorises, and tarsiers have nails on all except the second (in the tarsiers, the second and third) toe. This is known as a "toilet claw," which is used for scratching. In primates, the palms and soles are naked of hair, and the skin is padded, covered with ridges, and richly supplied with sweat glands. This combination of features allows for a better grip by creating friction and some suction on the gripping surface of the extremities. In addition, the sensitivity of these surfaces is enhanced by numerous nerve endings so that tactile discrimination is greatly increased. Digital pads are not observable in fossils but the flattened shape of the ends of the digital bones can indicate that a form possessed nails rather than claws.

Trends 4 through 8 involving the head and face are all generally attributable to the change in sensory orientation that goes with arboreal life, notably the emphasis on vision and de-emphasis of olfaction, or the sense of smell. This is accompanied by a change in the overall shape of the skull as well as the soft parts. Thus, there is a general tendency for the facial portions of the skull, particularly the nose and snout area, to become relatively smaller, accompanied by an increase in the size of the area that houses the brain. This trend is observable in any ranked series of living or fossil primate skulls. In living primates, accompanying behavioral trends are observable as well. For example, as a general rule, the Higher Primates will use their eyes and hands to do things that a prosimian would use his snout and teeth for, such as investigating objects, grooming, or communicating with other animals.

The importance of good vision, especially depth perception, to animals that climb and leap about in the trees cannot be overemphasized. Here mistakes in judging distances can be fatal. Trend number 5 is the result of several factors, the most important of which are: (a) the enlargement of the visual areas of the brain, (b) alterations in the structure of the retina and the nerves connecting it with the brain so that vision is more acute in each eye and coordination between the two is improved, and (c) the moving forward of the eyes in the head so that the visual areas of the right and left eye overlap, permitting stereoscopic vision. A bony eye socket develops for protection as the eyes move forward. The latter is observable in fossils through the changing position of the eye sockets in the skull and their gradual enclosure from the side and from behind by bone. In living tree shrews, the eyes are located on the sides of the head, while in the Higher Primates they point directly forward. The lemurs represent an intermediate condition.

Trends 4 and 6 are correlated with the general decrease in the importance of the sense of smell. With it goes (a) a decrease in the size of the olfactory areas of the brain, (b) alteration in the structure of the external nose so that in the tarsiers and the Higher Primates the nose is not covered with a naked, moist membrane for the rapid conduction of odors, and (c) a reduction of the bony snout. As a rule, prosimians use olfactory communication to a greater extent than the Higher Primates. This involves the use of specialized scent glands and "marking" behavior which are universal among the Lower Primates, found also in some South American monkeys, but are relatively rare in Old World Higher Primates.

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In the dentition of the primates, evolutionary changes have taken place in both the numbers of teeth that a group possesses and in their form. Like most mammals, primates are both diphyodont, having two sets of teeth (deciduous and permanent), and heterodont, having teeth of different shapes which perform different functions (incisors, canines, premolars, and molars). The number of teeth characteristic of a species can be indicated by a dental formula that expresses the number of each kind of tooth in one quarter of the jaw (such as upper right). Primitive mammals had forty-four teeth and a dental formula of 3-1-4-3, or three incisors, one canine, four premolars, and three molars. Primates have undergone a reduction in the number of teeth, which is probably related to the reduction of the face and jaws in general.

This table shows the dental formulae and numbers of teeth characteristic of living primate groups. Here it can be seen that there is quite a bit of variation among the prosimian groups though all have lost at least one incisor and one premolar from the primitive mammalian formulae. The New World monkeys have as many premolar teeth as prosimians, but show an evolutionary tendency for the reduction of the third molar that is small and occasionally absent in many of the cebid monkeys and have disappeared from the dentition of the marmosets. Among the Higher Primates of the Old World, the dental formula is uniformly 2-1-2-3 (thirty-two teeth). There are, however, differences in the structure and arrangement of the teeth, which allow one to distinguish among the dentitions of the Old World monkeys, apes, and man. Just as they show wide variability in their dental formulae, the living prosimians also have variations in tooth form, to some of which represent specialized or aberrant features. The dental peculiarities of the aye-aye, with its dental formula reflecting the emphasis on its large gnawing upper incisors, have already been mentioned and have been shown to be related to its peculiar diet and way of life. The living tree shrews, lemurs, and lorises share an unusual specialization of the incisor teeth known as a "dental comb." The lower front teeth (incisors and sometimes canines) are slanted forward, and the crowns are narrow and closely spaced, forming an apparatus that looks very much like the teeth of a comb, while the upper incisors are reduced. Observations of captive animals indicate that these teeth are primarily used for grooming the fur and rarely in feeding or fighting.

In the Higher Primates the canine teeth are usually large and sharply pointed and stand out beyond the level of the other teeth. This necessitates some modifications in the adjacent teeth so that the large canines can fit in when the mouth closes. In animals with large canines there is usually a gap or diastema, between the upper second incisor and the upper canine for the large lower canine to fit. The crown of the first lower premolar is sectorial (slanted back with a sharp edge) so that the tip of the upper canine fits against it and is sharpened or honed by the grinding that occurs.

Dental Formulae & Number of Teeth in Various Living Primates

Group	Dental Formula	No. of Permanent Teeth
Lemuridae	2-1-3-3/2-1-3-3	36
Indridae	2-1-2-3/2-0-2-3	30
Lorisoidea	2-1-3-3/2-1-3-3	36
Tarsioidea	2-1-3-3/1-1-3-3	34
Cebidae	2-1-3-3/2-1-3-3	36
Callithricidae	2-1-3-2/2-1-3-2	32
Cercopithecoidea	2-1-2-3/2-1-2-3	32
Hominoidea	2-1-2-3/2-1-2-3	32

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The molar teeth of primates have remained rather simple and unspecialized as far as mammalian molars go. This may be related to the relatively generalized diets that most primates have. The primitive mammalian three-cusped molars have been transformed by the addition of a fourth cusp in the Higher Primates. The structure of the molars in the Cercopithecoidea is highly distinctive with four cusps, two forward and two behind each pair being joined by an enamel ridge. The crown of the tooth is constricted in the middle. This type, known as a bilophodont molar, is a useful distinguishing feature of the fossil and living Old World monkeys and indicates their genetic similarity. Observations of baboons living in East Africa indicate that the value of the bilophodont structure is that the teeth can still function when extremely worn. The modern apes and man usually have either four or five cusped molars with grooves separating the cusps. These grooves may form a *Y* which when associated with five cusps is called a *Y* -5 pattern. The *Y* -5 pattern is found in fossil apes and men and gives evidence of their close phylogenetic relationship.

Teeth are very valuable in the study of phylogeny because they are good genetic indicators and are often preserved in the fossil record. They can also give some clues as to diet and ecology. However, observations from living primates serve to emphasize the fact that all dental evolution is not necessarily related to diet. The dental comb of the prosimians is a good example. The large, sharp canine teeth of the Old World monkeys and apes serve important functions in predator defense and social displays. It is well known that in many human groups teeth are used as tools in such non-dietary activities as tanning hides.

Certainly one of the most significant features of primate evolution is the trend involving expansion and reorganization of the brain and its consequences for behavior. Primates are large-brained animals for their body size. The most important changes have occurred in the cerebral cortex which has progressively expanded in size so that it covers the rest of the brain, and its surface has become folded. While the olfactory area of the cortex has become reduced in the Higher Primates, the visual area and those having to do with coordination of sensory and motor activities, association, and memory have greatly expanded. These changes in the structure of the brain permit (1) more accurate sensory perception, especially visual and tactile, (2) better coordination between sensory stimuli and muscular response, and (3) greater variety of behavioral responses to environmental factors. The first two are obviously advantageous to animals that leap and climb about in the trees. The third is extremely important as an adaptive mechanism, for primates can modify their behavior to meet a wide variety of environmental problems. In a sense, their behavioral flexibility has taken the place of bodily specializations. This trend has been carried farthest in man.

Trends number 9 and 10 are related to primate reproductive patterns that might be said to emphasize quality rather than quantity of offspring. Though mammals have fewer offspring and take better prenatal and postnatal care of them than do reptiles, none have carried this tendency to the extremes seen in the primates. In the placental mammals, the young develop inside the mother's body and take nourishment from her bloodstream by means of a special structure known as the placenta which attaches to the wall of the uterus. After birth the mother continues to nourish her young by means of mammary glands or breasts and to otherwise protect and care for them. Thus, the young continue to be physically dependent on the mother for some time. During this dependency and association with an adult (or adults, if the mother lives in a social group), the young are in a position to learn various forms of behavior which promote their survival. They can also experiment to some extent without their mistakes-being fatal, since there will be an adult around to help bail them out of trouble. Learned behavior is advantageous over inborn, or instinctive behavior because it is more flexible and adaptable to a wide range of circumstances. Therefore, it is not surprising that increased intelligence, advanced brains, and better maternal care go together in the mammals and especially in the primates.

As a rule, female primates give birth to one infant at a time. Only the tree shrew has litters and the marmosets and some lemurs regularly have twins and triplets. The advantage of having a single infant may be ultimately due to arborealism and the difficulty of carrying more than one offspring. Rather than leaving them in a nest, most Higher Primate mothers carry their infants, or, more often, the infant clings to the mother's fur while she climbs, swings, leaps, or walks about. While it is usual for the prosimians to

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have multiple nipples for nursing their young; the Higher Primates typically have a single pair located on the chest. Since only one infant is usually born at a time, its survival is more critical and features that enhance its chances of survival have evolved. One of these is a more efficient type of placenta that permits a closer contact between the fetal and maternal blood supplies. Going from the Lower to the Higher Primates and from monkeys to apes to man, there is a regular and well-marked trend for a lengthening of the period of growth and development and an accompanying delay of maturation. As a result, there is a longer gestation period, an increasing immaturity of the infant at birth, a longer postnatal growth period, and a later attainment of sexual maturity. The extreme expression of this trend is seen in man whose growth period lasts for about twenty years.

Comparative Data on Growth & Development			
Species	Gestation	Age at sexual maturity	Length of growth period
Rhesus Monkey	5 ½ months	2-3 years	7-8 years
Chimpanzee	7 ½ months	8-9 years	11-12 years
Man	9 months	13-15 years	20-21 years

This Table gives some comparative figures for a cercopithecoid monkey (the rhesus macaque), an ape (the chimpanzee), and man. There is also an apparent increase in longevity that goes with the lengthening of the pre-adult phases of life, but this is difficult to document in wild primates.

The fact that the Higher Primates are helpless and immature at birth and take a long time to reach adulthood means that their learning period is greatly expanded. The delayed maturation also affects reproduction. Not only do female primates usually have only one infant at a time, but their reproductive capacity during a lifetime is relatively low because they reach sexual maturity late. Also, because of the length of gestation and unusually long period of nursing, Higher Primate females do not typically have infants in rapid succession. At the most they can have one infant a year and in many it is less often than that. Thus trends number 9 and 10 both ensure that primates have relatively few infants, but invest considerable time and effort in caring for and raising them. The return on this investment must be great; otherwise such a pattern would not have evolved. In summary, the major prevailing tendencies of primate evolution have been to deemphasize the nose, teeth, and face and to emphasize the hands, eyes, and brain. The limb skeleton remains little changed, and the reproductive system becomes more efficient. It can be seen that man is very similar to the other primates in his basic anatomy, and that most of his differences are simply a matter of degree. This makes it very difficult to define man logically.

PRIMATE TAXONOMY & BEHAVIOR

RELATIONSHIP OF PRIMATES WITH OTHER MAMMALS

To define our place in nature, we can start with our widest memberships and then consider our more intimate relationships. R. H. Whittaker (1969) reviewed the evolutionary relations of all organisms and proposed a major revision of the traditional system of classification. He noted that bacteria and other one-celled organisms were variously classified and do not "fit" comfortably into either the plant or the animal kingdom. Following Whittaker, it is now customary to classify the bacteria and similar organisms within a kingdom of their own – the Monera – characterized by unicellular organisms which are usually nourished by absorption (an exception being the blue-green algae, which carry on photosynthesis). Another kingdom, Protista, includes most other one-celled forms and a few colonial organisms.

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The multi-cellular organisms are differentiated on the basis of nutrition into those dependent on absorption, photosynthesis, and ingestion. This new classification allows the animal kingdom to be defined more precisely as multi cellular organisms that ingest their food. Most animals have also evolved a sensory-neuro-motor complex that makes possible perception of food and movement to acquire it. Complementary to the capacity to ingest food are digestive, circulatory and excretory complexes, the regulation of which requires rapid conduction of information along nerves as well as self-regulating chemical feedback through internal secretions. Whittaker added that the logic of this "has led to high levels of structural and functional complexity among animals and ultimately towards complexity of inherited behavior or toward intelligence". The degree of differentiation exceeds that of all other organisms. We humans are thus animals; we ingest food, move and feel.

A feature of the animal kingdom, which all animals use in some generations, and some (including humans) use in all generations, is bisexual reproduction. That is, reproduction is not simply the production of an image of one parent but the recombination of elements inherited form two parents. Bisexual variation provides for great variability. Only half of the chromosomal material of each parent is involved and it is a chance matter which of each parent's paired genes will be involved in the recombination in the offspring.

Radiation: As the single celled organisms have given rise to multi-celled ones with differentiated tissues, Whittaker divides these in turn, by the three ways they transform food into energy. There are

1. The producers (plants with their photosynthesis)
2. The Reducers (fungi) and
3. The Consumers (animals).

Each of these functions is possible in any setting and so, in turn, there has been a tremendous differentiation through the evolution of variants. This evolution can be depicted as branches of a tree and is therefore cladogenetic.

CHORDATES

The major divisions of a kingdom are called **Phyla** (singular: **Phylum**, meaning a clan or tribe). Among the animals, the human species belongs to the phylum of the **Chordates.** These animals are bilaterally symmetrical, that is, the left and right sides are, for the most part, mirror images of one another. Chordates are also distinguished by having, at some time during their life, a flexible rod called a **Notochord** along their backs (from the Greek word Noton: The Back). The notochord is a rod of cartilage which serves to stiffen and support the body. Nearly all extant chordates also have a **Spinal Chord,** but it is the notochord, not the spinal chord, that distinguishes the phylum Chordata.

Of the chordates, the most numerous group shares still other features with humans. This sub-phylum is the **Vertebrates** and includes the fishes, amphibians, reptiles, birds, and mammals. These forms have a spinal cord with a brain at the head end. The cord and the brain form the central nervous system, which coordinates movements and sensations. In addition, vertebrates have a well-developed vertebral column to surround and protect the spinal cord. They also have an internal skeleton of bone or cartilage that surrounds and protects such organs as the brain and sense organs, and enables the limbs to support and move the body. The vertebrates regularly have locomotor appendages. Except for certain fish, these always consist of two pairs of limbs or, as in snakes and whales, some evidence of descent from animals that had them.

MAMMALS

The subdivisions of phyla are called **Classes** by biologists. One of these is the mammals (from mammae meaning the breasts). Animals of this distinct class of vertebrates have breasts and suckle their young with milk. Because mammals nourish their young after birth, they establish social relations between

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mother and offspring. Social relations are characteristic to various degrees, throughout the class and are also present in various other kinds of animals. Sustained social relationships are especially useful to human mammals for the generation-to-generation transmission of culture through learning.

Mammals are "warm blooded" (**endothermic**) animals. This means that their body temperature is internally controlled. Mammals can therefore remain active in cold weather. Hair, which is unique to mammals and serves as an insulator, and sweat glands which aid in cooling, are part of the temperature control mechanism. The body temperature of some mammals changes somewhat, but even the bear during hibernation does not undergo a slowing of body processes to the extent characteristic of fishes or reptiles similarly exposed to the cold. Reptiles are **ectothermic**, i.e., they have temperatures which vary with the outside temperature. Such animals therefore become sluggish in cold weather. In cold environments mammals manage better than reptiles. No crocodiles and few varieties of lizards and snakes are found outside the tropics. The large carnivorous dinosaurs may well have been endothermic.

The mammals share another main feature with the birds. Besides both being warm blooded, in both the young remain dependent for a time on the adults. In addition, birds and mammals share some secondary features, such as efficient body insulation and some systems of communication.

Mammals are also characterized by their *differentiated teeth*. Reptilian teeth are generally fairly simple in shape, usually looking like sharp cones with no important differences from one part of the mouth to another. (Only the specialized venom-injecting grooved fangs of some snakes are the only exception). In most mammals, however, there are four different kinds of teeth which serve various functions. The front teeth, or **incisors**, have sharp edges for cutting or biting, and behind them the pointed and often projecting **canine** teeth serve to grasp prey in some meat-eating mammals or as defensive weapons in others. Further back are the **premolars** (often called **bicuspids** in humans, since ours have two cusps or points) and the **molars** which process the food for swallowing. This may involve grinding, chopping, or shearing in various species. Molar form in mammals is highly variable, with the net result of greater dental adaptability than in reptiles or amphibians in the sense that mammalian species have teeth that are closely related to their particular ecological niche.

The study of ancient mammalian teeth may provide a clue to diet, since tooth form and diet are evidently correlated. Also, similar tooth structure can be informative, since similar structure in spite of the possible range of variation may suggest that two forms share a common ancestor. The mammal like reptiles of 180 million years ago, from which the mammals presumably evolved have been identified largely on the basis of their differentiated teeth.

Another feature of mammals is that they learn by trial and error. This gives them a very wide behavioral repertoire, including of course, human behavior. Other animals sometimes have developed potentiality for learning. Thus, some birds learn details of their song from trial-and-error repetition of what they hear, whereas other species of bird inherit their characteristic songs. Nevertheless, the mammalian capacity for learning seems unsurpassed in other animals, and mammals depend heavily upon learned behavior. The reproductive mechanism in mammals is marked by economy. That is, higher proportions of the young survive to maturity than in other classes. This reaches its maximum in humans, where a high percentage of offspring conceived are born and a majority of those born survive to adulthood. By contrast, most insects and other invertebrates lay thousands upon thousands of eggs that fail to survive to become reproductive adults. There is a trade-off involved in these very different reproductive strategies, however. Mammals invest a great deal of time and energy in each of their relatively few offspring, whereas invertebrates invest little beyond the production of fertilized eggs. Both reproductive approaches are obviously successful in an evolutionary sense. In mammals the reproductive economy is closely related to the importance of learned behavior, which is enhanced and promoted by continued parent-offspring contact.

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SUBCLASSES OF MAMMALS

The class of mammals is divided into three subclasses. These differ from each other primarily in respect to model of reproduction. The first of these subclasses is that of the **Monotremes or Prototheria.** These primitive mammals, in common with reptiles and birds, lay eggs, but they do have functional mammary glands. The duckbilled platypus and the spiny anteater are only two extant mammals belonging to this subclass. The existence of this subclass suggests that mammals acquired the capacity to suckle their young before any of them substituted live bearing for egg bearing. The ultimate reptilian origin of mammals is also suggested by the fact that the platypus is the only living mammal that is poisonous. Spurs on the hind limbs can inject venom comparable to that of the rattlesnake.

The **Marsupials** constitute a second subclass (**Metatheria**) of mammals. This group is somewhat more widespread than the egg-laying mammals and, besides numerous types in Australia, including the kangaroo and the teddy bear koala and also includes the American opossum. The newborn are very immature, however and must find their way to a pouch on the mother's abdomen where they can attach themselves to her nipples. They remain within the pouch until they are adequately developed to re-emerge. The monotremes and marsupials, with the exception of the opossum, have survived only in that part of the world where for geological resources they did not have to compete with more recent and highly evolved mammals. As primitive mammals, they are now threatened with extinction by the introduction of modern placental mammals in their natural habitats.

PLACENTAL MAMMALS

Humans belong to the third subclass, **Eutheria** or the **Placental Mammals.** These develop by the special process in which the egg or ovum is shed from the mother's ovary, fertilized internally, and than implants itself in the wall of the mother's womb. Placental mammals use the physiological mechanisms of the adult mother to remove fetal wastes and supply oxygen and nourishment to the developing embryo, whereas the egg of a bird or reptile must contain enough yolk to nourish the embryo until it is hatched and a device for innocuously storing the waste products. The offspring of a placental mammal is protected within its mother's womb until it has grown to a size larger than is usual in egg-laying animals, at the time they hatch. Other animals, including some snakes and fishes, are viviparous and have the capacity to harbor the eggs within the mother's body until ready to hatch, but only in the eutherian mammals is there a placenta capable of sustaining a long intrauterine development and the birth of a relatively large infant. The primates have well developed placentas.

It would be impossible to study or understand the enormous variety of living things and their evolutionary past without some means of organizing them. Scientists have therefore arranged the primates, like all living things, in hierarchical categories called taxa. The organization of taxa or taxonomy that we use today is based on the system invented in 1758 by Linnaeus. He grouped organisms according to their anatomical features. Organisms that shared most or all of their physical features were placed in the same category. The fewer the number of features any two species share, the more likely they are to be placed in different categories.

Taxonomy in its modern use also reflects evolutionary relationships. The system takes into account to some degree the phylogeny, or evolutionary relationships, of the various species. Orangutans, chimpanzees, gorillas, and humans, for instance, are classified together into a single super-family, reflecting the present belief that they diverged rather recently from a common ancestor.

In this classification of primates, we shall progressively isolate the characteristics that define humans. We have already looked at the generalized characteristics that we share with the other primates. Our distinctive human traits will come into focus as we move through increasingly narrow taxonomic categories to Homo sapiens. Along the way, the major taxa into which humans fall will be distinguished from those into which they do not fall.

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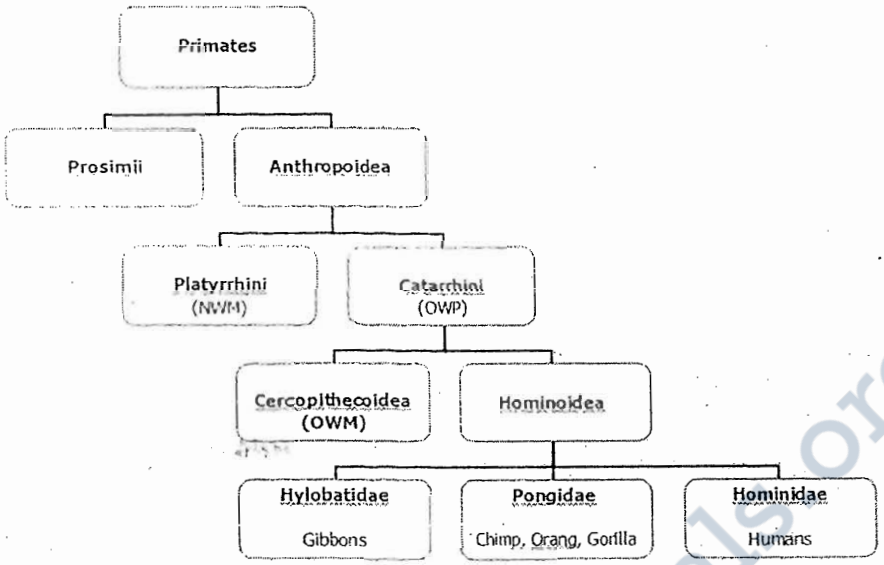
CLASSIFICATION OF ORDER PRIMATES



DISTINGUISHING PROSIMI FROM ANTHROPOIDEA

Prosimians are the least complex of the primates. They are distinguishable on the basis of certain primitive characteristics thought to have been possessed by the earliest primates. Some of these traits are also found among non-primates, especially those in the order insectivora, which includes shrews, moles, and hedgehogs. In both Prosimians and insectivores, the sense of smell, for instance, is very well developed. This trait is less emphasized in more complex primates in favor of sight. The eyes of Prosimians are placed at the sides of the skull. Because there is a limited overlap in the fields of vision, three-dimensional sight is not fully developed in many species.

In addition to their retention of primitive characteristics, prosimian species also have many specialized characteristics. These characteristics are adaptations that have allowed Prosimians to live in new areas and avoid competition with anthropoid populations. Many, for example, have developed specializations for nocturnal or night time, activity. These adaptations (such as huge eyes) have cut down on their competition with diurnal or day-living creatures. Varying patterns of evolution have created prosimians as unlike the tiny frog-like tarsier and the large bear-like indri. The characteristics that distinguish prosimians from the higher primates are:

1. Either of two primitive adaptations to movement in the trees:
   a. Modifications for clinging to vertical limbs and leaping from tree to tree (as in the tarsiers): long hind limbs, strong hind limb muscles, skeletal arrangement to support a vertical posture;
   b. Modifications for very slow climbing on all fours (as in the Loris): short limbs, arms almost as long as the legs, mobile hip and ankle joints for hanging, short tail or none at all.
2. Ability of fingers to act in unison; inability to act separately. This arrangement allows powerful grasping, but not manipulation of objects, as in the anthropoids.
3. Claws on some digits instead of nails. These claws are called "toilet claws" because they are used for cleaning dirt and parasites from fur and skin.

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4. Primitive arrangement of the pads on the fingertips and toes. These pads aid in clinging to vertical surfaces. In more complex primates, the pads are smaller and little more than thickened skin.
5. Eyes set apart at a greater angle than in anthropoids.
6. Adaptations by some prosimians to nocturnal life. These include enlarged eyes and loss of retinal cones from the eye structure, which respond to bright light and allow color vision.
7. Large, mobile ears which can scan for sound.
8. Well-developed sense of smell. Some have olfactory snouts with bare, moist tips, curved nostrils, and a tethered upper lip, attached in such a way as to allow little facial expression. The sense of smell is less emphasized in the anthropoids.
9. Relatively large olfactory lobes in brain. There is less cortical area than in anthropoids, and ratios of brain size to body weight are smaller.
10. Dental specializations: the upper front teeth, or incisors, are very small and the canine teeth (pointed teeth between the incisors and the grinding teeth) are large. The lower incisors and canines project forward in many species to form a dental comb for grooming the fur.

THE PROSIMIANS

There are three major infra-orders in the prosimian suborder.

1. Tarsilformes;
2. Lorisiformes
3. Lemuriformes.

Tarsilformes: The tarsier is the only remaining member of the once large Tarsilformes infra-order. These tiny creatures, which live in Borneo and the Philippines, are equipped with a very long tail and hind limbs for hopping from branch to branch. Their huge eyes and the pads at the ends of their fingers add to their strange frog like appearance. The special pads help them cling to smooth, vertical surfaces. Tarsiers can walk on a vertical sheet of glass. During the day they sleep clinging to a vertical branch. They wake at dusk and spend the evening hopping from tree to tree looking for insects and lizards to eat.

Lorisiformes: Nocturnal primates, Lorisiformes are divided into two distinct types - lorises and galagos - according to their locomotor patterns. Lorises are found in sub-Saharan Africa and in Asia. They are adapted to slow climbing and creeping. This movement is made possible by a very powerful grasp and strong muscles. Lorises can hang by one hind limb while reaching for food. Insects seem to form the bulk of their diet, but they will eat almost anything from flowers to young birds. Galagos, by contrast, leap gracefully through the trees, taking long kangaroo like hops with their strong hind limbs. Their shorter forelimbs are used to grab hold of branches when they land. They can easily leap from one vertical support to another. Galagos range from the size of large mice to the size of large rabbits. Like the lorises, they are omnivores, that is, that eat both plants and animals.

Lemuriformes: The Lemuriformes, which include lemurs, the indrises, and the aye-ayes, are found primarily on the island of Madagascar, off the southeast coast of Africa. They have developed many variations because of the lack of competition and predators. The members of one genus, the mouse lemurs, are smaller than a mouse and they are the tiniest living primates. The largest members of this infra-order are the indrises, which measure over three feet from head to tail. Most lemuriformes are active during the day and at dusk, but some are nocturnal. Most are completely arboreal, different species living at different levels in the same trees. All lemurs are wonderful acrobats. They can leap great distances, sometimes to the smallest of branches, swinging themselves up gracefully at the end of the jump. Their ability to do so is due not only to their musculature but also to unusually well-developed

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binocular vision. Unlike other prosimians, the lemurs avoid animal foods and seem to be highly social. Among other prosimians, only females and infants live together. But the lemurs live in troops of up to 60 members and sleep cuddled in small groups with their tails wrapped around one another.

THE ANTHROPOIDS

The anthropoids are also a diverse suborder. But they are characterized by their own shared set of specialized adaptations. These adaptations mainly allow a more effective use of arboreal environments. Forward-facing eyes permit stereoscopic vision among the anthropoids, and the visual centers of the brain are complex, allowing integration of large amounts of visual data. Depth perception and quick reaction to visual cues are essential to tree-living. Anthropoids also possess color vision, which aids in depth perception and the sighting of brightly colored fruit in trees. Adaptations for grasping and handling objects are crucial to life in trees, as well. Finally, the characteristically high brain-to-body ratio of anthropoids increases their ability to survive by agility and learning. These qualities are more appropriate in the trees than are the specializations of terrestrial animals for attack and defense, such as large horns, tusks, and claws.

DISTINGUISHING PLATYRRHINI FROM CATARRHINI

Within the suborder Anthropoidea, there are two infra-orders: Platyrrhini, the New World monkeys, and Catarrhini, the Old World primates. The most important traits that distinguish these two groups are:

1. Distinctive Locomotory Pattern: New World monkeys usually run along branches on all fours. While feeding at the ends of small branches, they spread their weight between their hands, feet, and tails. Old World primates are more likely to leap from tree to tree, swing by their arms, or walk on the ground.

2. Tails: All New World monkeys have tails, some of which are prehensile. Although some Old World primates have tails, none are prehensile.

3. New World monkeys lack ischial callosities, which are tough pads of skin on the rump that allow the animal to sit on tree limbs for long periods of time.

4. New World monkeys have flat noses with round nostrils and a broad fleshy area between them. Old World forms have nostrils that project and point downward.

5. New World monkeys have a 2.1.3.3. dental pattern (like that of the prosimians). On each side of the jaw there are two incisors, one canine, three premolars, and three molars. Marmosets, an exception, lack the third molar. Old World forms have one less premolar, top and bottom, and hence their dental formula is 2.1.2.3.

THE NEW WORLD MONKEYS:

The New World monkeys live throughout the heavily forested regions of Central and South America. They are divided into two families: the Callithricidae, which include the marmosets and tamarins, and the Cebidae, which contain six subfamilies. Most New World monkeys belong to the family Cebidae.

Most New World monkeys seem to spend their lives in the trees. The woolly titis, for example, lives only in the smallest branches of the highest trees. They do not come down, even for water. Apparently they get enough moisture from the flowers, fruits, and nuts they eat. But the more highly evolved capuchins, which are commonly known as "organ grinder monkeys", some times leave the forests to steal fruit and vegetables from farms. Squirrel monkeys too will sometimes come down out of the trees to catch insects.

Locomotor patterns vary among the New World monkeys. Some walk on all fours, both on the ground and on tree limbs. Others, like the spider monkeys, are best at moving in an upright position. They do so by swinging from their arms, which are longer than their legs. Spider monkeys use their strong, prehensile tails to swing and to grasp limbs as they pick fruit. Reportedly, they can jump as far as 30 feet.

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We know very little about the social behavior of most New World monkeys because they live in thick jungles, high in the trees, where they cannot be observed easily. They have been seen in troops of up to 30 and in family bands.

OLD WORLD PRIMATES:

Within the Old World line, there are two super families - Cercopithecoidea (or Old World monkeys) and Hominoidea (humans, apes, gibbons, orangutans, chimpanzees, and gorillas). The major differences that have evolved between these two groups are:

1. Most species of Old World monkeys have tails; hominoids have no tails.
2. Old World monkeys are generally smaller than hominoids.
3. Old World monkeys have prenatal development of ischial callosities. Most hominoids are not born with these rump pads, although they may grow after birth.
4. Old World monkeys have a shorter life span than the hominoids.
5. Old World monkeys have smaller brain size to body weight ratios as well as less complex brains than the hominoids. The cerebral cortex is especially well developed in hominoids.

The distinctions here are somewhat more finely drawn than the others we have made. This is because both super families are included in the same infra-order, a sign of their similarity of anatomy. They are more closely related than, say, members of suborders or infra-orders.

The superfamily Cercopithecoidea contains a single family, which can be divided into two subfamilies, the Cercopithecinae and the Colobinae. The most numerous genera in the Cercopithecinae are:

1. The arboreal monkeys of the genus Cercopithecus;
2. The mostly arboreal mangabeys, or Cerocebus; and
3. The terrestrial macaques and baboons, Macaca and Papio.

Langurs are the most common genus in the Colobinae subfamily.

Guenons: The cercopithecine monkeys, or guenons, are widely distributed in Africa. Although they live mostly in trees, some species come down to feed on the forest floor, in plantations, in open bush, and in savanna country. They live largely on insects, fruits and vegetation, but will sometimes eat birds, bird eggs, and even small mammals. They seem to live in small groups made up of a dominant male and small harem of females and their children.

Mangabeys: The long-tailed mangabeys that are sometimes seen with cercopithecines spend much of their day on the ground. Though they possess some adaptations to life in the trees, they also have some of the traits of terrestrial primates. Their hands, for instance, are more like those of the terrestrial baboons than those of totally arboreal primates. They live mostly in the swampy forests of Africa.

Baboons and Macaques: Though classed as separate genera, baboons and macaques are closely related. If they mate, they can sometimes produce fertile offspring. But they inhabit different ranges. The baboons live in Africa and the Arabian Peninsula. The macaques range from India east to Japan. They both live mainly on the ground, though they sleep in trees. Both have developed finely controlled hands with precision grips that function like ours.

Since terrestrial animals are much easier to watch than those that move in the treetops, the social behaviors of baboons and macaques have been studied fairly well. Baboons live in highly structured troops of 10 to 100 members. Within the troop a clear dominance hierarchy reduces tensions and provides protection from predators. The threat of predators may be the reason for their extreme sexual

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dimorphism (different physical forms for the two genders). Male and female arboreal monkeys, who face few predators and rely on their agility for fleeing, look pretty much the same, but among the ground-living baboons, the males are specialized for defense on the ground. They may be twice as big as the females, and they can put on impressive threat displays with their huge canine teeth and their great ruffs of fur at the neck.

Langurs: Among these slender, long-tailed, Asiatic monkeys, group organization is not as important to defense. They often feed on the ground, but prefer graceful flight into the trees rather than an organized show of force. Langurs sometimes live alone and sometimes in troops. In troops, the relationship between males and females is not well defined. The status of females is high when they are paired with a male. But when their infants are born the mothers seem to drop out of the status system altogether. When dominant males are found near temples, they form harems (groups of females) and actually chase away other males.

DISTINGUISHING HUMANS FROM THE APES:

The Apes:

Finally, we shall look at the differences that divide us from the Hylobatidae (gibbons), and our closest relatives, the Pongidae (chimpanzees, gorillas, and orangutans). A list of the anatomical traits distinguishing these two groups from hominids is as follows:

1. Limited bipedal walking: Although pongids and hylobatids are better than any other nonhumans at walking bipedally in a semi-erect posture, they do not do so for long periods of time.
2. Short thumbs: As a result, most of pongids and hylobatids have a limited precision grip.
3. Ridges of bone directly above the eyes: Pongids lack a forehead.
4. Teeth specialized for a diet that includes massive quantities of fruit and vegetation: The canines, used to open fruit and strip plants, are large and projecting. To hold their heavy teeth and support the chewing of such bulky food, pongids have massive jaws that jut forward. A bony simian shelf buttresses the inside of the front part of the jaw. Large cheek bones and in males a ridge of bone called a sagittal crest at the top of the skull anchors the strong chewing muscles.
5. Lower brain size-to-body weight ratio than among humans, who have the most complex cortical structure of the hominoids.

GIBBON: It is one of the smallest apes inhabiting South East Asiatic Islands, viz., Java, Sumatra, Borneo, and Philippines. They are also seen in Southern China. There are about 12 varieties of gibbon. The height of the gibbon does not exceed 3 feet and the weight of the adult gibbon ranges from 11 to 15 pounds. There is no sexual dimorphism with regards to the size. The body of these animals is covered with fine wooly hairs, whereas, the hairs of gorilla, chimpanzee and orangutan are coarse. The color of the hair is white, gray and sometimes black or a combination of these three colors.

At times, the gibbon can stand erect. The arms are excessively long in comparison to the length of the leg. It is said that during erect position, the finger tips of the gibbons generally touch the ground. The gibbon can pick up any material from the ground without stooping. Due to their adaptation to arboreal condition, the hands of the gibbons, especially the radius and the ulna (fore arm bones) have elongated enormously. The hand is very long and narrow with a relatively short thumb, which is not perfectly opposable. With the help of the light and slender body, powerful arms provided with long fingers and short thumb, the gibbon can easily travel through the trees with a free swinging and pendulum motion. In length of the leg, as compared to the trunk, the gibbon exceeds all other anthropoids and stands next to man. The foot is elongated and narrow. The great toe will be found far apart from other digits. This wide separation has an advantage in grasping the branches of the tree.

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The head is large. The size and general appearances of the skull resemble those of the cercopithecidae group. The brain case is larger in comparison to the face, which is short and provided with a flat and broad nose and thin lips. The forehead is low, and the large and oval eye sockets are composed of thick bony rims. They have large canines and these interlock at the corners of the mouth. The temporal area of the cranium is rough, caused by the attachment of the temporal muscles. The cranial length of the gibbon is 7.5 cm. The cranial capacity varies from 76-90 CC. It is greatly below than that of the higher apes, but higher than those of the monkeys.

The gibbons are gentle and pleasant, and they live in families, which are composed of the parents and the minor children. They can be kept as pets. Their food consists of fruits, leaves, buds, eggs, different kinds of insects and smaller birds. The gibbons possess ischial callosities which is the characteristic of Old world monkeys. In males, the ischial callosities are widely separated in two halves.

The gibbon family can be divided into two groups - Hylobatidae and the Symphalangus. The former includes gibbons proper and the representative of the second group is the Simiang found only in the Sumatra region. The Simiang is larger in size than the gibbon. They have laryngeal air sacs. According to some scientists, the simiangs form the intermediate group between the gibbons and the giant apes.

ORANGUTAN: The orangutans differ very much from the gibbons in their bulky and heavy bodies. They are found mostly in the islands of Borneo and Sumatra. In erect position, the animal is nearly 5 feet high. The weight of the adult males varies from 165 to 200 pounds. The females are somewhat higher. The body of orangutan is covered with long, coarse hairs, of reddish brown color. No hair is found on the face, ears, palm and the soles. The chest is barrel like. The breasts are laterally situated near the armpits. The arms are long and narrow with elongated fingers. The thumb is small and all the digits have flat nails. The forearm is much longer than the upper arms. The legs are short and the foot is long and narrow. The great toe is opposable and very small.

The head is large with a high and rounded forehead. The brain case is small in relation to the large head. The orbits are elliptical in shape. The average cranial capacity is 416 cc. The nasal bones are small and the bridge is not elevated. The root of the nose is very narrow. The jaw of orangutan is extremely large and projects forward. The jaws are provided with long and tusk like canines which interlock.

The orangutans are arboreal in nature. They live on trees by building nests on the branches. They walk on all fours when they come to the ground. The whole body of orangs is built in such a manner that it can enjoy the arboreal life to a lesser degree than the gibbon, but to a higher degree than the two other apes. The food of the orangutan consists of fruits, different varieties of leaves, insects etc. The orangutan is represented by a single species.

CHIMPANZEE: The body of the chimpanzee is not so well built as in the case of the orangutan and the gorilla. The chimps are found in large numbers in the tropical forests of Africa. There are three species of chimpanzees - the common chimpanzee, the black-faced, bald-headed chimpanzee and the pygmy chimpanzee. The body proportion of chimpanzee shows some tendency towards that of man. The average height of the full grown common chimpanzee is 5 feet. The weight of the adult male and female are 110 and 88 pounds respectively.

The body is covered with loose and coarse hairs of various colors. There are no hairs on the face, hands and feet. The heads of the bald-headed chimpanzees are devoid of hairs. Most species of the chimpanzees have round low vaulted heads. The average cranial capacity is 400 cc. There is a poor development of the supra orbital ridge and it is continuous. The nasal bones are very small, the bridge is not elevated and the tip of the nose is not like that of the man. In chimpanzee, the forward projection of the jaw is clearly seen. The canines are large and projecting but are smaller in comparison to that of the orangutans and the gorilla. The lips are thin. The general appearance of the skull is oval; the facial region is small in proportion to the skull. The orbits are elliptical but not so as we find in the case of the orangutans.

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The hand of the chimpanzee is elongated and narrow. The fingers are long and the thumb is small and opposable. The legs are long and these are larger than those of the orangutan. The foot is long. The hind limbs of this animal are poorly adapted for walking upright. The great toe is opposable and it is not in line with the other toes. The heel is rudimentary and in erect position the hands of the chimpanzee generally reach up to the knee level.

The chimpanzees are experts in climbing and brachiating. They build nests on the trees and sleep there during the night. Their food consists of fruits, different vegetable products, eggs, small birds and sometimes rodents.

GORILLA: Among all the primates, the gorilla is the largest and the stoutest. They are found in equatorial regions of Africa. The gorillas are represented by two species - the Western Coastal or lowland gorilla and the eastern or mountain gorilla. The former inhabit mainly the regions of Cameroons while the latter is found in the region of the Eastern Congo, west of Lake Edward and Lake Kivu.

When the animal stands erect, the hands go below the knee joint. The height ranges from 5 feet to 6 feet in the lowland gorillas and 5 feet 3 and a half inches to 5 feet 11 inches in the mountain gorillas. The weights of the body of adult males range from 350 to 600 pounds.

The body is covered with long and black colored hairs. The face, palms and soles are completely devoid of hair. The massive skull of the gorilla is a note worthy feature. Above the eye openings, an enormous supra orbital goes from left to right side of the skull. The forehead is very low. The general appearance of the skull is more or less oval and there is a prominent sagittal crest. It is weaker in females. The average cranial capacities of the males and females are 550 cc and 450 cc respectively. The big cranium consists of solid bones and there is little space for the brain. The facial portion is larger in proportion to the cranial part. The jaws are large and projected forward and downwards. The upper jaw is prognathus and the lower jaw is massive. The size of the canines is enormous and they interlock. The nasal bones are long, low and narrow; the bridge is slightly elevated. Nostrils are broad and forwardly directed and these are covered by cartilaginous rings mostly like nasal wings but not so developed as we find in man. The lips are thin and chin is completely absent.

The gorilla is more human in different features than in other anthropoid apes. The upper arms are longer than the forearms and therefore it is different from gibbon, orangutan and the chimpanzees. The hand is shorter and broader than in other apes and the thumb is well developed. On the whole, the hand of the gorilla resembles more that of man than any other ape.

The leg is short. The foot of the gorilla also resembles that of man in many respects. The thumb is opposable and it is set apart from the other digits. The heel is more or less well developed. The anatomy of foot of the gorilla speaks that it is less adapted for arboreal prehension. The gorillas spend most of their time on the ground. They climb on the trees also, build their nests and pass the nights there. Sometimes, they can walk erect but their usual gait is obliquely quadrupedal.

Difference between the mountain gorilla and the lowland gorilla: The mountain gorilla differs from the lowland gorilla in the following characteristics - narrow skull, longer trunk, larger face, shorter limbs, shorter and broader hands, body covered by thick and black colored hairs etc.

MODERN MAN: The Homo sapiens have spread and multiplied over the whole world within the past 70,000 years. All the living varieties of mankind stem from a common background. But as they settled in different environmental conditions, they acquired distinctive traits. On the whole, man possesses certain unique characteristics, which have helped him to surpass all other animals and thereby establish a new genus Homo.

The relative lightness of the bones is one of the unique features of the mankind. The skull is a large bony case situated at the anterior of the vertebral column. The skull consists of two parts - the cranium and the face. The former is a large and hollow bony case in which the brain is placed. The brain is complex and

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all its parts are relatively enlarged. In size it is from two and a half to three times large as that of the gorilla. The height of the face is short and it is nearly at a right angle to the base of the skull. In man the snout is vanished and the sense of smell is diminished. But, the power of the sight is increased. Our vision is stereoscopic. The orbit is more or less rectangular. The forehead is bulging and the supra orbital ridges have diminished. The nose is prominent and it is raised out of the plain of the face. The nasal bones are short, wide and fused. The fleshy tip of the nose is made of osteo-cartilaginous frame work. The lips are out-rolled and the mucus membranes are visible. The upper lip is provided with a median furrow and is the unique characteristic of man. The nature of the teeth has undergone a great change because of the change in the diet. They are adapted for dealing with a wide range of food. The canines are diminished in size and they do not extend beyond the level of other teeth. In general, all the teeth have become relatively small and closely held together. The molars have been modified to a great extent for grinding. The dental arcade is parabolic in shape. The chin is well developed. In man, the unique characteristic about the skull is that it is well balanced. The spinal cord enters into the foramen magnum of the skull which is situated centrally at the base of the skull. The average cranial capacity of man ranges from 1300 to 1450 cc.

The adaptability of man is seen greatly in the remarkable nature of the limbs. The arms are shorter than the legs. This has resulted due to the bi-pedal locomotion. It is believed by the scientists that our ape ancestors developed the ground living habit when a vast jungle area became treeless due to a climatic change. Walking erect requires an enlargement of the legs. The ridges for muscle attachments on the bone are much more defined than as we find in the apes. The foot has also changed. The great toe is not opposable. Innermost digit is the most dominant one. The metatarsals are bound together by powerful transverse ligament. The lateral toes are reduced, the fifth becoming a rudiment. Humans possess a well developed heel, and the legs are strictly used for walking.

The pelvis is bowl shaped and the transverse diameter is greater than antero-posterior diameter. The sacrum becomes short and broad. The vertebral column has four curves while in the apes only two of such curves are seen.

Man's body is more or less hairless compared to the other apes. The unique characteristic of man is that there is no tactile hair on his body. The upright position, the bipedal locomotion, complex brain, hairlessness, excellent eye sight, power of speech - all these factors have jointly given man a better place in the animal kingdom and thereby he establishes a new genus Homo sapiens.

TERTIARY AND QUATERNARY FOSSIL PRIMATES

The Geological Time Scale during Cenozoic

EPOCH	APPROXIMATE BEGINNING (MYA)
Pleistocene	1.8
Pliocene	5
Miocene	22.5
Oligocene	37
Eocene	53
Paleocene	65

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EARLIEST PRIMATES

The first radiation of the primate order has its roots in the beginning of the explosive adaptive radiation of placental mammals in general. Therefore, it is not surprising that the earliest suggestions of primates in the fossil record are difficult to discern from early members of other placental mammal groups, particularly the insectivores.

The earliest discovered traces suggesting the beginnings of our order came from the late Cretaceous and early Paleocene in the Dakota area of North America. Known from several teeth and jaw specimens, this fossil form, called **Purgatorius,** exhibits the primate tendency towards a bulbous cusp pattern in the molar teeth compared to the sharper cusps seen in insectivores. Such a dental pattern apparently indicates that early primates were adapting to fruit and leaf-eating diets, or perhaps a diet also including insects exploited in an arboreal niche. Reconstructions indicate that the dental formula is still that of a generalized primitive mammal: 3-1-4-3. All that can be said now of this still fragmentary and mysterious animal, *Purgatorius,* is that if it was a primate, it was quite "primitive", which is not surprising considering its early date and still close ties with primitive placental mammals.

PALEOCENE PRIMATES (65 my – 53 my)

During the Paleocene, the first clearly recognizable primates begin to diversity. In fact, these forms are extremely diverse, with as many as 6 families and numbering more than 25 genera and 75 species. One of the most widely distributed and best represented of these early primates is **Plesiadapis,** found in both North America and Europe. First known from the late Paleocene, the time range of *Plesiadapis* extends up into the early Eocene. This animal, known from several skulls plus several parts of the limb skeleton, has an estimated size range varying widely between that of a squirrel and a house cat. While these animals show definite primate tendencies, such as dependence on vision (though not completely binocular) and flexible wrists and ankles (though still probably with claws), they retain numerous primitive characteristics, such as lack of a postorbital bar, which serves to form a bony division between the orbit and the braincase. In addition, *Plesiadapis* and the majority of other Paleocene primates display unusual (compared to modern primates) specializations of their front teeth, which are large and procumbent (angled forward) and also show other dental oddities.

Given these dental specializations, the possibility that any of the Paleocene forms thus far discovered are direct ancestors of later primates seems unlikely. This poses no insurmountable theoretical problem, for we must keep in mind that our meager discoveries represent only a tiny sample of the already diverse kinds of primates living at this time. The overall anatomical plan of all these early forms could be described as prosimian or even "proto prosimian", for they are certainly more primitive than any living primate. In fact, they are more similar in evolutionary grade to the living tree shrews, which give a fairly good model of some Paleocene primates. We must note, however, that Paleocene forms are highly diverse, numbering more than twice as many as the living prosimians.

EARLY PRIMATE EVOLUTION

The distribution and the eventual fate of early primate forms is understandable only within the context of the environments in which these animals lived. First and foremost, we must remember that 60 mya land masses were not arranged as they are today. The continents have drifted to their present position, carried along on the shifting plates of the earth's surface.

In the late Mesozoic, the huge conglomerate lad mass called Pangea began to break up into northern and southern continents. To the north, North America, Europe, and Asia were joined into Laurasia; to the south, Africa, South America, Australia, India and Antarctica formed Gondwanaland. Throughout the Mesozoic, the two basic land masses continued to move, with Gondwanaland breaking up into the southern continents. In the north, the continents also were separating, but North America and Europe continued to be connected through Greenland and would remain close to each other for several millions of years. North America and Europe remained in close proximity until mid Eocene, i.e., around 45 mya.

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What makes all this geological activity relevant to primate studies is that land-living animals could cross over land bridges, but were effectively cut off by water barriers. Thus, we see species of *Plesiadapis* in both North America and Europe. As far as primates go, then, from their earliest beginnings (65 mya) up until 40 mya they were mostly limited to North America and Europe. With further continental movements, the "New World" and "Old World" became completely separated and thereby influenced the evolutionary histories of primates still living today.

As the continents moved, climatic conditions changed dramatically. In the Mesozoic and into the Paleocene, the continental masses were clustered closer to the equator, and as Laurasia in particular moved north, its climate cooled. Moreover, the fragmenting of the land masses and the consequent altering of marine flow patterns (less exchange between northern and southern seas) caused the climate to cool even further.

Finally, these climatic shifts also heavily affected plant communities. Rather than the primitive, mostly tropical flora characteristic of the Mesozoic (ferns, cycads etc), what we see emerging in the Cenozoic is the rapid radiation of the seed plants (including flowering plants, grasses etc). Indeed, many of these plants are frequently pollinated by insects. As insects thus became more abundant, so did the animals who fed on them – including early primates. The world was never to be the same again. Clearly then, it is extremely important to interpret primate evolution within the context of the earth's changing environments.

EOCENE PRIMATES (53 my – 37 my)

The first primates of modern aspect appear during the Eocene. These can now clearly be called prosimians and closely resemble the Loris/lemur evolutionary grade. Primate diversification accelerates during this epoch, with 4 new families and more than 60 genera represented during this 16 million year span of the Eocene.

The Eocene may be characterized as the heyday of prosimians, who attained their widest geographic distribution and broadest adaptive radiation during this period. Indeed, almost four times as many genera are found in the Eocene than are known for the whole world today (16 living genera, with 10 of these confined to Madagascar).

The most diversified and best-known Eocene primates are members of the lemur-like Adapidae, which includes some 10 genera. The four best known of these are:

Cantius	North America and Europe
Adapis	Europe
Notharctus	North America
Smilodectes	North America

Some of these animals have been known from fossil evidence for a remarkably long time. In fact, the initial discovery of *Adapis* was made in France in 1821 and first described by Cuvier himself.

As mentioned, all these animals are fairly lemur-like in general adaptive level and show distinctive primate tendencies not seen in the Paleocene forms. For example, they all now have a complete postorbital bar, larger, rounder braincases, nails instead of claws, and the eyes are rotated forward, allowing overlapping fields of perception and thus binocular vision. In the limb skeleton, further developments in prehensility are suggested, and some evidence points to the presence of an opposable large toe. In all these respects, we see the typical primate adaptive strategies allowing exploitation of an arboreal environment. Whereas these forms resemble lemurs in overall anatomical plan, they do not show the same specializations seen in contemporary lorises and lemurs, such as development of dental comb – projecting lower incisors and canines forming a structure used for grooming and hence called dental comb.

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The separate evolution of the Malagasy lemurs may date to late Eocene times, but since this island was already isolated by a deep channel from mainland Africa, they apparently reached their island sanctuary by unintentionally floating over drifting debris. The other major group of Eocene prosimians is the family Omomyidae, known from numerous specimens of jaws and teeth. Omomyids are the most widely distributed of known Eocene prosimians, with discoveries in North Africa and Europe, and a few specimens form Asia as well.

The earlier members of the family are somewhat more generalized than the later ones and may form an ancestral basis for all later anthropoids, New World monkeys, Old World Monkeys, Apes and hominids. Additionally, some of the omomyids from the late Eocene of Europe have been suggested as closely related to the tarsier. However, many of the similarities noted are apparently superficial ones, not necessarily indicating any unique relationship. Nevertheless, at least one feature, the position of the olfactory bulb, links these Eocene forms with later haplorhines. ((The suborder that includes lemurs and lorises is termed **Strepsirhini** (moist nosed), while tarsier, monkeys, apes, and humans are included within another suborder, the **Haplorhini** (hairy-nosed)). Also of interest is the fact that, when we enter later epochs, there are forms quite clearly sharing tarsier-like affinities, form the Oligocene of Egypt and from the Miocene in Thailand.

NEW WORLD MONKEYS

The center of action for primate evolution after the close of the Eocene is confined largely to the Old World, for only on the continents of Africa and Eurasia can we trace the evolutionary development of apes and hominids. However, the New World, while geographically separated from the Old World, was not completely devoid of anthropoid stock, for there the ceboids evolved in their own right.

Any discussion of ceboid evolution and its relation to Old World anthropoid developments must consider crucial geological events, particularly continental drift. In the late Eocene and early Oligocene, South America was an island continent separate from North America and Africa.

In such geological context, the introduction of monkeys in South America poses a certain problem. Some authorities have postulated that because the open water distance between South America and Africa was still not very great, monkeys, originating in Africa, could then have rafted to the New World.

In any case, that Old and New World primates share any evolutionary history since at least the early Oligocene (37-35 mya) is unlikely. There is no trace of any ancestor of our lineage anywhere in the New World following this time until fully modern *Homo sapiens* walked into the New World during the late Pleistocene.

While primate fossils abound in the Western Hemisphere during the early Cenozoic, the record is extremely sparse later on. For the entire span of Oligocene to late Pleistocene, we have only a few bits and pieces, a jaw fragment from Bolivia, a nearly complete skull from Texas, several specimens from southern Argentina, and a few other small fragments from Colombia and Jamaica. Together, all the evidence comprises barely a dozen individuals. Thus, tracing the evolutionary heritage of New World monkeys with any degree of certainty is a difficult task.

OLD WORLD ANTHROPOIDS

It is apparent that during this epoch a great deal of evolutionary action was taking place; by the end of Oligocene, Old World Monkeys and hominoids were probably evolving along their separate evolutionary pathways. No doubt, diverse species of anthropoids were adapting to varied ecological niches in Africa and probably Asia and Europe as well. Unfortunately, the fossil record for the entire period is limited to only one locality in Egypt, 60 miles southwest of Cairo. This site, called the *Fayum*, is today an extremely arid region 100 miles inland, but, in Oligocene times, it was located close to the Mediterranean shore and was traversed by meandering streams crisscrossing through areas of tropical rain forest. The extremely rich fossil-bearing beds were laid down during the early Oligocene, between 37 and 31 mya.

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Much of the Fayum fossil material is quite fragmentary. Consequently, evolutionary interpretations are not as unambiguous as we would like. Given the nature of the material, the classification of the fossils into recognized genera and species is somewhat disputed. The leading researcher of the Fayum fossil primates, E..L. Simons of Duke University has recognized seven different genera from the Fayum, six anthropoids and one prosimian. In addition, the new tarsier-like fossil previously mentioned would add yet another form to this impressive tally. Altogether, an estimated 1,000 specimens have been recovered from the remarkably productive Fayum area. The three best known of these genera are discussed below.

Apidium (Two Species): A well known 30 million year old fossil animal from the Fayum is **Apidium**, represented by about 80 jaws or partial dentitions and over 100 postcranial elements. This animal, about the size of a squirrel, had several anthropoid features, but also shows quite unusual aspects in its teeth. *Apidium's* dental formula is 2-1-3-3 which, as you can easily see, reveals an extra premolar not found in any contemporary Old World anthropoid. For that matter, extremely, few fossil anthropoids in the Old World have this extra premolar. Some researchers suggest, therefore, that this genus and its close relatives (together called the parapithecids) may lie near or even before the divergence of Old and New World anthropoids. As noted, *Apidium* is represented by a large array of specimens, both dental and from the limb skeleton. The teeth suggest a diet composed of fruits and perhaps some seeds. In addition, preserved remains of the limbs indicate that this creature was a small arboreal quadruped, adept at leaping and springing.

Propliopithecus (Two Species): Among the first fossils found at the Fayum, an incomplete *Propliopithecus* mandible was recovered in 1907. Unfortunately, since detailed geological and paleontological methods were not yet developed, the precise geological position of this form is not known.

Morphologically, this fossil is a quite generalized Old World anthropoid, displaying a 2-1-2-3 dental formula. In every relevant respect, this early *Propliopithecus* form is quite primitive, not showing particular derived tendencies in any direction. Both species appear small to medium in size (8 to 13 pounds) and were most likely fruit-eaters. To date only this one specimen represents this *Propliopithecus (haeckeli)* species.

The second *Propliopithecus* species is considerably better known, with several new specimens discovered at the Fayum between 1977 and 1979. Geologically, this species (*P. chirobates)* comes from the upper beds and is, therefore, probably later in time than the isolated *haeckeli* specimen. Consequently, it is not surprising that *chirobates* is more derived in several anatomical features. Still, this form is a remarkably primitive Old World anthropoid. Considerably more evolutionary change would be required to transform this animal into anything distinctively recognizable as either an ape or a monkey.

Earlier interpretations of fragmentary remains of this fossil suggested affinities with gibbons. However, more complete recent discoveries have shown that such an evolutionary relationship is not likely.

Aegyptopithecus: The most complete and probably evolutionarily most significant fossil from the Fayum is *Aegyptopithecus* which is known from several well preserved crania of different aged individuals as well as numerous jaw fragments and several limb bones. The largest of the Fayum anthropoids, *Aegyptopithecus* is rougly the size of a modern howler monkey, 13 to 18 pounds. *Aegyptopithecus* is important because, better than any other fossil, it bridges the gap between the Eocene prosimians on the one hand and the Miocene hominoids on the other.

With a dental formula of 2-1-2-3, *Aegyptopithecus* shows the familiar Old World anthropoid pattern. Even more primitive than the teeth of this specimen, is the skull, which is small and resembles the skull of a monkey in some detail. Brain size and relative proportions can be reconstructed from internal casts of the crania thus far discovered. It appears that the brain was somewhat intermediate between that of prosimians and anthropoids. The visual cortex was large compared to prosimians, with concomitant reduction of the olfactory bulbs, but the frontal lobes were not especially expanded. Even considering the relative small size of this animal, the brain – estimated at only 40 cc – was by no means large.

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Evidence from the limb skeleton also revealed nothing particularly distinctive. From analysis of limb proportions and muscle insertions, primatologist John Fleagle has concluded that *Aegyptopithecus* was short-limbed, heavily muscled, slow-moving, arboreal quadruped.

Further detailed study of *Aegyptopithecus* anatomy has even allowed primatologists to speculate about the social behavior of this ancient primate. For instance dental remains from different individuals vary greatly in canine size. This fact implies male/female differences (sexual dimorphism), were apparently quite marked. Comparisons with living primates further suggest that males may have been competing for females and that the mating pattern was probably polygynous.

All in all, *Aegyptopithecus* presents somewhat of a paleontological enigma. In most respects, it is quite primitive as an Old World anthropoid and could thus be potentially an ancestor for *both* Old World monkeys and hominoids. There are some slight yet suggestive clues in the teeth that have led some researchers to already place *Aegyptopithecus* on the hominoid line. Other researchers are not convinced primarily because of its primitive characteristics.

MIOCENE (22.5 my – 5 my) – HOMINOIDS GALORE

If the Eocene was the age of prosimians and the Oligocene the time of great diversity for early anthropoids, the Miocene was certainly the epoch of hominoids.

A great abundance of hominoid fossil material has been found in the Old World from the time period 22-7 my. The remarkable evolutionary success represented by this adaptive radiation is shown in the geographic range already established for hominoids during this period. Miocene hominoid fossils have been discovered in France, Austria, Spain, Czechoslovakia, Greece, Hungary, China, India, Pakistan, Turkey, Saudi Arabia, Egypt, Uganda, and Kenya. While several intriguing discoveries potentially relating to gibbon evolution have been discovered at Miocene sites, the vast majority of this hominoid material relates to large-bodied forms. Unless we state otherwise, all further discussion of Miocene hominoids refers only to large-bodied varieties.

Interpretations of this vast array of fossil material (now including more than 500 individuals, and perhaps as many as 1,000) were greatly complicated for several decades due to inadequate appreciation of the range of biological variation that a single genus or species could represent. As a result, the taxonomic naming of the various fossil finds became a terrible muddle, with close to 30 genera and over 100 species proposed. The biological implications of such taxonomic enthusiasm were unfortunately not seriously considered. In such an atmosphere, it was possible for two genera to be named one with only upper jaws represented, the other with only lower jaws, and each matching the other!

It is not difficult to understand why such confusion arose if we consider that discoveries of these fossils spanned more than 100 years (the earliest find came from France in 1856) and took place on three continents. Not until the early 1960s did scientists systematically study *all* the material, the result being a considerable simplification of the earlier confusion. As a result of this research, E. L. Simons and David Pilbeam "lumped" the vast majority of Miocene forms into only two genera: one presumably quite "pongid like" and the other "hominid like." In just the last few years, however, a tremendous amount of new data has come to light from both new field discoveries and finds in museum collections of previously unrecognized material. Consequently, it is now apparent that the Simons-Pilbeam simplification went too far. Hominoid evolutionary radiation during the Miocene produced a whole array of diverse organisms, many of which have no living descendants (and thus no clear analogy among living higher primates).

As these new discoveries are analyzed, many of the perplexing problems concerning Miocene hominoids should be solved. For the moment, it is possible to make only general interpretive statements regarding Miocene hominoid adaptive patterns.

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PALEOGEOGRAPHY AND MIOCENE HOMINOID EVOLUTION

As they were to early primate evolution, the factors of changing geography and climates (at work as well in the Miocene) are also crucial to interpretations of the later stages of primate evolution. The Oligocene revealed a proliferation of early Old World anthropoid forms from one area in North Africa. In the Early Miocene, the evidence is also restricted to Africa, with fossils coming from rich sites in the eastern part of the continent (Kenya and Uganda). It would thus appear on the basis of current evidence that hominoids originated in Africa and experienced a successful adaptive radiation there before dispersing to other parts of the Old World.

The hominoids would maintain this exclusive African foothold for some time. The earliest of these East African hominoid fossils is more than 20 mya, and later fossil finds extend the time range up to at least 14 mya. For most of this period, East Africa is thought to have been more heavily forested, with much less woodlands and grasslands (savannas) than exist today (Pickford, 1983).

As in the earlier Cenozoic, the shifting of the earth's plates during the Miocene played a vital role in primate evolution. Before about 16 mya, Africa was cut off from Eurasia; consequently, once hominoids had originated there in the Early Miocene, they were isolated. However, around 16 mya the African plate "docked" with Eurasia (Bernor, 1983) through the Arabian Peninsula, a contact that was to revolutionize mammalian faunas of the later Miocene. Many forms, such as proboscideans, giraffoids, and pigs that originated in Africa now migrated into Eurasia). Apparently, among these mid-Miocene intercontinental pioneers were some hominoids. Since they had evolved in the mainly tropical setting of equatorial Africa, most of the earlier hominoids probably remained primarily arboreal. Accordingly, it has been suggested that a relatively continuous forest would have been necessary across the Afro-Arabian-Eurasian land bridge.

Ecological changes were, however, already afoot in Africa. By 16 mya, the environment was getting drier, with less tropical rainforest and, conversely, more open woodland/bush-land and savanna areas emerging. In other words, the environments in East Africa were being transformed more into their contemporary form. With the opportunities thus presented, some African hominoids were almost certainly radiating into these more open niches about 16 to 17 mya. Part of this adaptation probably involved exploitation of different foods and more ground-living than practiced by the arboreal ancestors of these hominoids. Some partly terrestrial, more woodland or mosaic environment adapted hominoids were probably on the scene and fully capable of migrating into Eurasia, even through areas that were not continuously forested.

The environments throughout the Old World were, of course, to alter even more. Later in the Miocene, some of these environmental shifts would further influence hominoid evolution and may have played a part in the origin of our particular evolutionary lineage, the hominids.

EAST AFRICAN FORMS (23-14 mya)

A wealth of early hominoid fossils has come from the deep and rich stratigraphic layers of Kenya and Uganda. These diverse forms are presently classified within at least 2 separate families, including perhaps 9 different genera. Indeed, important and mostly new finds (from the 1980s) have been uncovered, suggesting as many as 3 further hominoid genera in an as-yet-undetermined (and possibly a third) family. In other words, over the 9-million-year period for which we have evidence, as many as 12 different hominoid genera have been sampled from East Africa, with the potential for yet more as the fossil sample accumulates.

Most of the newer material has yet to be described completely, and relationships among specimens are uncertain. The best samples and thus best-known forms are those of the genus *Proconsul* (belonging to the proconsulid family). From this full array of proconsulid remains (mostly dental pieces), considerable variation is apparent. Body size estimates range from that of a small monkey (about 10 lb), to as large as a female gorilla (about 150 lb). Environmental niches were probably also quite varied, for some species

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were apparently confined to dense rainforests, while others potentially exploited more open woodlands. Some researchers have also suggested considerable diversity of locomotory behaviors, including perhaps some forms that were at least partly terrestrial. Indeed, when on the ground, some of these proconsulids may even have occasionally adopted a bipedal stance.

The dentition of all the proconsulid forms is, however, quite uniform, showing the typical Old World anthropoid pattern of 2-1-2-3. Moreover, these forms all display broad upper central incisors and large sexually dimorphic canines. In the molars, the enamel is fairly thick (i.e., high cusps), but the softer dentine below penetrates well into these cusps, so that the enamel wore through fairly quickly with use. We can get some idea of diet from these teeth, which suggest that most forms were probably fruit-eaters.

From those well-preserved pieces of crania (representing currently only one species), brain size estimates are at least as large or larger than contemporary Old World monkeys (although probably not as large as contemporary hominoids; note, however, that *relative* brain size compared to body size is the crucial feature-a tricky estimate indeed for incomplete fossilized fragments). The surface features of the brain do not apparently show the derived characteristics of living large-bodied hominoids, and, in fact, show many primitive hominoid features similar to that seen in gibbon brains.

A full understanding of the evolutionary relationships of the East African hominoids has not yet been attained. Indeed, in many cases, the classification still remains a muddle. For example, one fossil (discovered in the 1950s) from Rusinga Island in Kenya has been renamed and reassigned to different evolutionary groups at least six times. Just as paleoanthropologists begin to think that the situation is becoming better defined, new fossil discoveries muddy the waters still further. Some of this new material may date as early as 18-17 mya, but most of it is in the range 16- 14 mya. Thus, for the most part, these finds are *later* than the proconsulids just discussed. It is not so surprising, then, to find that *Proconsul* is a more primitive hominoid. In fact, many primate evolutionists would place *Proconsul* before the split of small- and large-bodied hominoids. Therefore, while *Proconsul* may *not* actually have been the last common ancestor of gibbons as well as all large-bodied hominoids (including us), something resembling it may well have been.

EUROPEAN FORMS (13-11 mya)

Although they are the first of the Miocene hominoids to have been discovered, the European varieties still remain enigmatic. Very few fossils have been discovered, and what has been found consists almost entirely of jaws and lower dentitions. Among the only features that distinguishes this varied lot of specimens from France, Spain, Austria (and maybe Hungary, too) is that the molar teeth are thinly enameled (i.e., the dentine penetrates far into the cusps). Most researchers would place all these forms into the genus *Dryopithecus.*

Discovery of similar forms from the Rudabanya Mountains in Hungary during the 1970s have complicated matters further. Initially thought to be similar to the thick-enameled varieties from southern and southwestern Asia (see below), the Hungarian fossils are now placed closer to *Dryopithecus* from Western Europe. Nevertheless, many researchers still believe the Rudabanya fossils are probably a distinct genus *(Rudapithecus)* (Kelly and Pilbeam, 1986). It seems unlikely that these *Dryopithecus-group* fossils are related closely to any living hominoid.

SOUTH/SOUTHWEST ASIAN FORMS (? 16- 7 mya)

Three sites from Turkey have yielded fragmentary fossil hominoid remains dating to the early Middle Miocene (16-14 mya). As we noted earlier, following "docking" of the Arabian plate with East Africa about 16 mya, land routes became available for animal migration from Africa into Eurasia. It would thus seem, from these Turkish remains, that hominoids quickly took advantage of this route and reached Eurasia by 16 mya. Most researchers would assign these remains to the genus *Sivapithecus.*

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Far more complete samples of *Sivapithecus* have been recovered from southern Asia, in the Siwalik Hills of India and Pakistan. Most dramatically, over the last 15 years, paleoanthropologists led by David Pilbeam of Harvard University have recovered numerous excellent specimens from the Potwar Plateau of Pakistan. Included in this superb Pakistani collection is a multitude of mandibles (15 in all, some of which are nearly intact), many postcranial remains, and a partial cranium, including most of the face.

Sivapithecus from Turkey and Pakistan was probably a good-sized hominoid, ranging in size from 70-150 lb. It probably inhabited a mostly arboreal niche, and its locomotion was "apelike," at least in the sense that *Sivapithecus* most likely displayed some suspensory abilities. *Sivapithecus* differs morphologically from *Proconsul* or *Dryopithecus* in its dentition and facial anatomy. The front teeth, especially the upper central incisors are often quite large, while the canine is fairly good-sized (low-crowned and robust). There are, however, large discrepancies in canine size among *Sivapithecus* individuals, partly because some species were larger overall, but also because there was considerable variation (sexual dimorphism) within the same species. In diet, like most other hominoids, *Sivapithecus* was probably a fruit- eater.

The first lower premolar is also quite variable in shape. Usually it is fairly sectorial in shape; that is, it shows the shearing surface typical of most catarrhines (consequently, this is probably the primitive condition). The most distinctive aspect of *Sivapithecus* dentition is seen in the back tooth row, where molars are large, flatwearing, and thick-enameled (with dentine not penetrating far into cusps).

The thickness of the enamel cap has played a significant role in recent interpretations of Miocene hominoid evolution. Among living hominoids, relative to body size, humans have by far the thickest enamel caps. Gorillas and chimps have thin enamel, but orangs could be described as moderately thick. Thick, in fact very thick, enamel is also seen in early hominids (in the time period 4-1 mya). As we have seen in *Proconsul* and *Dryopithecus*, their enamel thickness itself varies, but dentine usually penetrates into the cusps, so that the enamel wears through during use.

Probably, the most characteristic anatomical aspects of *Sivapithecus* are seen in the face, especially the area immediately below the nose. Facial remains of *Sivapithecus* from Pakistan and Turkey have concave profiles and projecting incisors (and, overall, remarkably resemble the modern orang). In particular, the partial cranium discovered in 1980 at the Potwar Plateau (Pakistan, *circa* 8 mya) and published two years later (Pilbeam, 1982) bears striking similarities to the orangutan. The published description of this specimen had a tremendous impact on paleoanthropology. The biochemical evidence demonstrates the distinctiveness of the orang from the African apes and humans; here, then, was fossil evidence suggesting some ancient Asian traces of the orang lineage. As a result, the views of biochemists and paleoanthropologists agree more closely.

It must be noted however that, except for the face and jaw, *Sivapithecus* is *not* like an orangutan. In fact, especially in the post-cranium (i.e., all skeletal parts except the head) *Sivapithecus* is distinctively *unlike* an orang, or any other known hominoid, for that matter.

Many earlier fossil-based interpretations of Miocene evolutionary affinities had, of course, to be reevaluated. As we hinted at the beginning of our discussion of Miocene hominoids, in the 1960s E. L. Simons and David Pilbeam suggested a Middle Miocene hominoid as the first hominid. According to this view, this early hominid was *"Ramapithecus"* known at that time mostly from India, with some bits from East Africa.

As a consequence of these new discoveries, the earlier suggestion that *"Ramapithecus"* was a definite hominid was seriously questioned and has now been rejected altogether. One primary advocate of this revised view is David Pilbeam (1977; 1982; 1986), an initial architect of the earlier widely accepted theory. Pilbeam, who has led the highly successful paleoanthropological project at the Potwar Plateau, has been swayed by the new fossils recovered there and elsewhere. These more complete specimens are dentally very similar to what had been called *"Ramapithecus."* Researchers now simply lump *"Ramapithecus"* with *Sivapithecus.*

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In summary, then, the fossil remains of *Sivapithecus* from Turkey and India - Pakistan are the most clearly derived large-bodied hominoids we have from the whole Miocene. While some forms (e.g., *Proconsul)* are seemingly quite primitive and others *(Dryopithecus)* are derived in directions quite unlike any living form, *Sivapithecus* has several derived features of the face, linking it evolutionarily with the orang. The separation of the Asian large-bodied hominoid line from the African stock (leading ultimately to gorillas, chimps, and humans) thus occurred at least 12 mya.

OTHER MIOCENE HOMINOIDS

Pliopithecus: Another interesting but still not well-understood hominoid is *Pliopithecus,* from the Middle and Late Miocene of Europe. Since this is a fairly small hominoid (estimated at 11-16 lb), for several years primatologists suggested *Pliopithecus* was a gibbon ancestor. Moreover, dental features were also thought to mirror gibbon morphology. However, these similarities are superficial at best. In those respects in which *Pliopithecus* resembles contemporary small-bodied hominoids, the features are all primitive for hominoids in general. In fact, for most relevant anatomical details, *Pliopithecus* is a remarkably primitive hominoid (in fact, at least as primitive as *Proconsul).* This is surprising, given its relatively late date and Eurasian distribution (where hominoids are generally more derived than their African cousins). It may be that *Pliopithecus* is a long-surviving descendant of an Early Miocene, very primitive ancestor, one that antedated the radiation of major hominoid lineages.

Greece ("Ouranopithecus," 12-11 mya): From the Ravin de la Pluie near Salonika, Greece, have come several hominoid specimens (mostly mandibles, but also a partial face) discovered in the 1970s. Because the molar teeth have thick enamel, researchers initially grouped these finds with *Sivapithecus.* However, recent analysis of the critical facial anatomy has shown that the Greek finds are not similar to *Sivapithecus* (or the orang), but their molar morphology also makes them unlike *Dryopithecus.* The evolutionary relationships of this Greek hominoid thus still remain a mystery.

Lufeng, Yunnan Province, Southern China (8-7 mya): The recent discoveries from the Lufeng site in southern China have been remarkable, now totaling over 1,000 specimens (including several crania, mostly crushed mandibles, and hundreds of isolated teeth). Since the fossil collection is so large, and since the crania are in need of much restoration, most of the material has yet to be fully described. Therefore, conclusions regarding this most important fossil collection must remain highly tentative. Indeed, there is still argument concerning how many genera are represented among the Lufeng hominoids, with some experts favoring two, while others see only one genus. Ongoing interpretation of the vast dental remains has led some researchers to suggest that *only* one species may be represented. If so, this would be an extremely variable species, most likely reflecting extreme sexual dimorphism. In fact, such a degree of sexual dimorphism (at least dentally) would exceed that seen even in the modern orang (i.e., males more than twice the size of females). Like the Greek fossils discussed in the preceding section, the evolutionary relationships of the Chinese specimens are unclear. They also do not show the shared derived features of the *Sivapithecus-orang* lineage. Determining exactly where they fit thus remains a major challenge for primate evolutionists.

A COMPARATIVE ANATOMY OF MAN AND APES

Darwin's declaration regarding the descent of man from non-human ancestors caused a great deal of controversy among the scientists and the general public of those times. It was a matter of a great degradation when any attempt was made to draw any line of relation between the apes and man. However, in the due course, with newer inventions and discoveries in this line, Darwin's view was accepted by all and thereby the people are forced to embrace their strange relatives.

Apes and men are the members of families Pongidae, Hylobatidae and Hominidae. They belong to the suborder Anthropoidea and the order Primates. The relationship of apes and man will be understood if attempts are made to study the various anatomical features critically.

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The skulls present many contrasting features which should be studied with care. The skull of man is highly developed in the frontal region. The forehead extends almost vertically in the upward direction. The supra-orbital ridge is not so developed and it may be feeble, trace or moderately developed. No sagittal crest is present in the skull. The maxilla and the pre-maxilla have been fused. The foramen magnum is situated at the center of the skull. The head is well balanced and hence the face is perfectly vertical. The line of muscle attachments are less developed on the back side of the skull. In apes, the forehead is less developed and the head slopes backwards. The supra orbital is highly developed and prominent. A sagittal crest is present in the skull. The foramen magnum is seen further backward at the base of the skull. It is for this reason that the ape's face hangs downwards. The line of muscle attachment is seen high up in the backward part of the skull.

The lower jaw of man is small in size in comparison to those of the apes. The muscle responsible for the movement of the lower jaw is weak. In man, there is always a well developed chin. Man's facial portion never projects forward but the prognathism of the lower jaw is sometimes noticed, though very rare. Ape's lower jaw is massive and there is no trace of chin. The muscles for the movement of the lower jaw are strong and well developed. Facial prognathism is very common among the apes.

Teeth in man are smaller in size than those of the apes. The canines do not project forward beyond the level of the other teeth. The chewing motion in man is from side to side and also up and down - generally known as the rotary motion. The dental arch, in the case of man, takes the shape of a parabola, whereas, in apes, it is U-shaped. In apes, the sizes of the teeth are large and the canines project beyond the level of the other teeth. Due to this nature, the canines of the apes interlock when the jaws are closed. This arrangement prevents the lateral movement in chewing. The chewing motion in ape is only up and down. With the shortening of the canines, man has developed a new rotary motion.

The nose in the primates is composed of two parallel bony plates, united by a suture in the middle line. From these nasal bones extend the cartilaginous nose which is divided into two separate chambers - the nostrils - by the septum. In man, the nose is well developed. The root and the bridge of the nose have a slightly marked elevation and the cartilaginous portion is seen well above the surface of the nose. The tip of the nose in man has a thick bulb that overhangs the septum. The nasal wings are strongly developed, but they do not grow under the long axis of the nose to form the lower borders of the nostrils as is found in the case of the gorilla. The nostrils are smaller and they generally point downward. The elevation at the root and bridge in ape's nose is very little or absent. The cartilaginous portion of the nose is very wide, unlike man, and it is little raised on the surface of the face. The tip of the nose is lacking in the apes and this is why the nostrils have become prominent and look like large holes.

The lips of the apes are seen stretched over their bulging jaws and they are loose and protrusive. The ape's lips are thin as the red portion of the lip is scarcely seen when the mouth is closed. The integumental lip has little quantity of fat. In man, the integumental upper lip shows a median furrow which starts from the nasal septum and continues upto the edge of the membranous lip. This median furrow is the characteristic feature of man. The lips vary from thin to very thick.

In apes, the arms are greatly elongated, an adaptation to the habit of hanging and walking in the branches. This type of locomotion by arm of the apes is called brachiation. In apes, the upper arms acts as movable levers and hence the length is short and in man the fore arm is movable lever and has hence shortened.

The femur (thigh bone) of ape is short, thick and curved. In man it is long, slender and elongated. Due to this, the ape walks with a shambling gait. The ridges for muscle development in femur are greatly developed in man than in the apes. The linea aspera - a rough ridge on the back side of the femur is characteristic to man. It has resulted due to the great development of the extensor muscles which play a most important part in the erect posture of man and the bipedal gait. Owing to the great development of the ridge, the cross section of the human femur gives a prismatic form, while in apes it is round and oval.

The foot of man has witnessed a remarkable change due to its new mode of locomotion. The foot supports the weight of the body and helps in standing and walking erect. In apes, the foot is used for both

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locomotion and for the grasping of branches on the trees. The great toe in man is non opposable. It lies in the same line with other digits. The lateral toes are reduced in size and the fifth one is rudimentary. But in the case of the apes, the great toe is opposable and it is not in line with the other digits. The lateral toes are well developed in apes. Unlike the ape, man's foot is arched antero-posteriorly.

As regards to the stature and weights, the apes vary. The gibbon is the smallest among the apes and its average height and weight are three feet and fourteen pounds respectively. The orangutan attains an average height of four feet six inches and the average weight of about 165 pounds. The Chimpanzees and the gorillas are considered as the tallest among the apes. Their average heights are five feet and five and a half feet respectively and their average weights are 100 and 350 pounds respectively. The average height of man is five feet six inches and the average weight is 145 pounds. The vertebral column of man possesses four curves which help in supporting and transmitting the weight of the head and trunk in erect posture. In apes, the vertebral column has two such curves.

In brain development, man shows more advancement than the apes. The brain in man is not only much larger than that of the apes but it is highly developed. The weight of man's brain is three times as heavy as that of the gorilla, the largest ape. The frontal region of human brain is especially developed and the cerebral cortex presents a much more complex convolutionary development than that of the apes. But it should be kept in mind that the structural difference is totally one of degree. The brain case in man is the largest ranging from 1300 to 1450 cc. The average cranial capacities of the gorilla, chimpanzee, orangutan and the gibbon are 549, 400, 416 and 98 cc respectively.

Man is distinguished from the apes mainly by his power of articulating speech. But there are some scientists who have felt that the use of some sort of language among the apes. According to G. L. Garner, the vocal language of the monkeys and apes differs only in its complexity and degree from that of man.

We have seen the differences and the similarities in the characteristics of man and apes. According to Huxley, "whatever system of organs be studied, the comparison of their modification in the ape series leads to one and the same result - that the structural differences which separate man from the gorilla and the chimpanzee are not so great as those which separate the gorilla from the lower apes". He concludes that in the sum total of classificatory differences, the apes are like men. Recent studies about the physiological characteristics of the uterus and placenta of apes revealed a lot of similarities with those of the human beings. The physiological sexual rhythm in the female chimpanzees is similar to that of human females. Even the serological studies point to a close relationship between apes and man. Even many diseases of humans like typhoid, cholera and syphilis can be transmitted to the apes.

ANATOMICAL SIMILARITIES AND DISSIMILARITIES BETWEEN MAN AND APES

Gorilla and Man: In hands, feet, and pelvis and in size of brain, the gorilla shows close relationship with man. But the massive jaws of the gorillas have no resemblance with those of man.

Chimpanzee and Man: The chimpanzee and man relate to each other to a great extent by means of the likeness of the skull and the pigmentation of the body. The chest proportion of chimpanzee is mostly human.

Orangutan and Man: In his high forehead and the same number of pair of ribs, the orang demands a closer relationship with man, than all other apes. On the other hand, the two factors like the shortness and degenerate character of the legs and the adoption of the feet for suspension, separate these two individuals from each other.

Gibbon and Man: The length of legs and the erect gait of the gibbon indicate its closer relationship with man, anatomically. But in other characters like excessive arm lengths, general size, pelvis, hands and feet, length of canine teeth and the size of the brain, the gibbon goes farthest from the man.

It is very difficult to identify the man's closest relative among the four apes. This is especially true when all the physiological, anatomical, morphological, anthropometric and genetic characters are considered.

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Among the four apes, it is seen that each of them develops one or more special features, with which it can demand the closest relationship with man.

PRIMATE ADAPTATIONS - ARBOREAL AND TERRESTRIAL

One of the major contributions of the study of living primates is in the area of functional anatomy. The evolutionary importance of anatomical characters and complexes cannot be fully understood by studying skeletons or dissecting specimens. Studies of structure must be supplemented by observation of the behavior of living primates who are in the process of using their anatomy in various ways.

Two functional complexes that have received great deal of attention in primates are the hand and the locomotor apparatus. The two are not fundamentally different since all primates, except man, use their hands in locomotion to some extent. The reason for emphasizing on locomotion is the fact that man's full time, bipedal, upright locomotion, which has played such a crucial role in his evolution, is unique among the primates and its origins unclear from the fossil record. Thus, primatologists and anthropologists began to look at the locomotor systems of other primates in an attempt to discover what might have led to the human pattern.

LOCOMOTORY ADAPTATIONS

It is probable that the primitive mammalian ancestor of the primates was a small, essentially quadrupedal and arboreal animal who scurried along the tops of branches and climbed by digging its claws into the bark. From this pattern several different forms of locomotion have evolved. There is a naked trend toward upright posture in the primates which overlies the various forms of locomotion and can be considered a primate characteristic. Thus, the upright posture of man is not unique but is simply a part of this general trend.

The essential factors in upright posture are the reorientation of the body with respect to gravity and this necessitates many skeletal, muscular and visceral changes. Among these are the modifications in the proportions and balance of the skull, with the face or muzzle being reduced and the skull being set directly on top of the vertebral column. A good indicator of the degree of uprightness in fossil skulls is the position of the foramen magnum, the large hole in the base of the skull where the spinal cord joins the brain. In quadrupeds like the dog, the foramen magnum is located at the very back of the skull and there is a large muzzle in the front. With uprightness foramen magnum moves forward, and the face is reduced so that the skull nicely balances on centre of its base. Humans require little muscle action to hold their heads erect because of this balance; animals like gorilla with heavy muzzles require heavy neck muscles to hold them up.

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Group	Hand Movement	Thumb	Grasping Patterns
Tupaioidea	Convergence and divergence only	Non-opposable	Use two-handed prehension, whole-hand control
Lemuroidea Lorisoidea Tarsioidea	Convergence and prehension	Pseudo-opposable	Use a single grip pattern for power and precision, whole-hand control
Ceboidea	Convergence and prehension	Pseudo-opposable	Some separation of precision and power grips, but precision grip different from Cercopithecoid-Hominoid type. Little evidence of independent finger control.
Cercopithecoidea	Convergence and prehension	Truly opposable	Use separate power and precision grips. Have separate control of the thumb and, in some species, separate control of the index finger as well.
Hominoidea	Convergence and preshension	Truly opposable	Use separate power and precision grips. Have separate control of all fingers.

With regard to the locomotion, several basic types have been defined and these are presented in the table enclosed. It should be pointed out that locomotor behavior and anatomical structure do not always coincide. For example, the anatomy and skeletal proportions of the chimp and gorilla would lead us to classify them as brachiators, yet neither uses brachiation to any great extent. Behaviorally, they are quadrupeds; but their anatomy and hand posture are unlike those of other quadrupedal primates. Thus, some animals are difficult to classify in any simple manner. Also, it should be remembered that classification is based on the most typical form of locomotion used, and that animals in the natural habitat always use a variety of locomotor pattern.

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A CLASSIFICATION OF PRIMATE LOCOMOTOR PATTERNS

CATEGORY		SUB TYPE	ACTIVITIES	PRIMATE GROUPS
Vertical Clinging and leaping			Leaping using hindlimbs for propulsion and clinging to vertical support	Glagos, some lemurs, indrises, tarsiers
QUADRUPEDALISM	ARBOREAL	Slow climbing	Slow climbing by grasping firmly with hands and feet	Lorises, pottos
		Branch running and walking	Walking and running along tops of branches; climbing and leaping also seen	Tree shrews, some lemurs, aye-ayes, marmosets, some cebid monkeys, guenons, mangabeys, some macaques
		New World semibra-chiation	Swinging beneath branches by arms, legs, and prehensile tail; quadrupedal running and walking also used.	Howler monkey, spider monkey, woolly monkey, woolly spider monkey
		Old World semibra-chiation	Arm-swinging combined with climbing; quadrupedal walking and leaping	Colobus monkeys, langurs, and relatives (Colobinae)
	Terrestrial	Ground Running and walking	Quadrupedal walking and running on soles and undersides of fingers	Baboons, some macaques, patas monkey
		Knuckle-walking	Quadrupedal walking and running on soles and backs of curled fingers	Chimpanzee, gorilla
BRACHIATION		True Brachiation	Arm-swinging, bipedal walking on branches	Gibbon, siamang
		Modified Brachiation	Arm-swinging, with feet used for support, climbing, hanging by feet	Orangutan
Bipedalism			Striding upright on hindlimbs alone	Man

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1. VERTICAL CLINGING AND LEAPING

This type of locomotion is found only among the prosimians. It involves leaping from one vertical support to another, using the hind limbs for propulsion. The trunk is held in an upright position at rest, and the forelimbs are used for clinging to tree trunks and other vertical supports. The prosimians with this type of locomotion are primarily arboreal, but can hop on the hind limbs when placed on the ground.

The main bodily feature associated with this locomotor pattern is the elongation of hind limbs which are used as springs in leaping. This form of locomotion is found in species with widely different ecological niches, from nocturnal insectivores to diurnal vegetarians. It may be a basic primate locomotor adaptation which was achieved very early in the evolutionary history of the order and has been retained in a few prosimian groups today. If so, it may have begun the trend towards an upright posture in the primates.

2. QUADRUPEDALISM

Within this type of locomotory adaptation, two distinct traits can be differentiated - **Arboreal** and **Terrestrial quadrupedalism.** In this general category, both fore and hind limbs are used in locomotion. There are also several sub-types in this category which differ mainly in the degree to which climbing, leaping and swinging are employed in addition to quadrupedal running or walking.

ARBOREAL QUADRUPEDALISM:

Slow Climbing Quadrupedalism: The primates which show this type of locomotory adaptation combine slow climbing and deliberate movement with a powerful grip. The adaptation includes reduction in the index fingers and the thumb, and big toes are splayed out at a wide angle. Such an anatomical change allows them to use their hands and feet like forceps to enclose a branch in a firm grip. For example, Lorises, Pottos.

Branch Running and Walking Quadrupedalism: The primates adapted to this type of locomotion are usually seen walking or running along the tops of branches. Their fore limbs and hind limbs are of equal length though the hind limbs are slightly longer. Since the tree-shrews and marmosets have claws, they hold on to the branches with their claws while the others use their prehensile hands and feet to grasp the branches. Gaps in the trees are crossed by leaping and the tails are used for balance.

Old World Semi-brachiation: Found in the old world monkeys, the animals adapted to this type of locomotion are found sending a good deal of time swinging beneath the branches. The anatomical changes adapted to this type of locomotion are the presence of short, broad trunks, elongated arms, and long fingered hands and all but the wooly and howler monkeys have reduced thumbs. These animals not only swing but also leap.

New World Semi-brachiation: The new world members of this semi-brachiating group have prehensile tails which they also use to suspend themselves. In rest of the characteristics, this group resembles the old-world semi-brachiating primates.

Terrestrial Quadrupedalism:

There are two distinct terrestrial quadrupedal locomotory adaptations - **the ground running and walking** and **knuckle walking.**

Ground Running and Walking: The baboons and patas monkeys run and walk on the ground, on the soles of their feet. Their limbs are of about equal length. They do not have to grasp the surface on which they walk as their arboreal relatives do. The hand proportions, notably the relative lengths of the thumb and index finger, favor a more effective precision grip in this group than in arboreal quadrupeds.

Knuckle Walking Quadrupedalism: This type of locomotion is a secondary adaptation to ground living. The chimpanzee and gorilla have become quadrupedal, probably because of their large body size, but at

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the same time have retained the long arms, long fingers, and short thumbs of their brachiating ancestors. While they walk on all fours, their torso is actually slanted with the head end higher because of their disproportionately long arms. Chimpanzees do climb and brachiate in trees while feeding, but return to the ground and knuckle-walk when traveling. The anatomical changes associated with this type of locomotion are the elongation of pelvis; the hand of femur not forming great angle with the shaft which restricts the movement of leg; habitually bent knee and the whole foot making contact with the surface.

3. BRACHIATION

Brachiation is again of two sub-types - **True brachiation** and **Modified brachiation.**

True Brachiation: Gibbon and Siamang are true brachiators. These are the good models for studying bodily adaptations to these locomotor patterns and for investigating its adaptive value. In brachiation, the body weight is suspended from the arms and hands beneath the branches and progression is accomplished by swinging the body from one handhold to another. The hind limbs are sometimes used for support and occasionally for suspension as well. The gibbon body combines relatively light-body weight with long arms and long fingered hands. Since the hands must function as hooks during brachiation, a long opposable thumb is in the way. All of the brachiators have some means of getting their thumbs out of the way during locomotion. Some, like new world spider monkey or the African Colobus monkey, have thumbs that are absent or reduced to tubercles. Others, like the woolly monkey, use their thumb like another finger to form part of the hook. The gibbon which has a relatively long thumb gets it out of the way by tucking it up against the side of the first finger. Field studies of the gibbon have shown that brachiation is a locomotor adaptation which is primarily useful for feeding at the terminal ends of branches. The animal will hang from the branch with one hand and use his weight to bend the branch toward him while plucking fruit with the other hand.

Modified Brachiation: This type, found in the orangutans, involves arms swinging with feet used for support, climbing and hanging by feet. The orangutans are really cautious climbers than brachiators, and climb using any combination of their four grasping extremities.

4. BIPEDALISM

Found in the human beings, it involves the hind limbs alone supporting the body weight and propel it along the ground. The main bodily modifications involve the elongation of legs and changes in the foot and pelvis. In the foot, the big toe is no longer opposable, but has been brought into line with the other toes, an arch has developed, and the heel has become longer to aid in balance. Thus, the grasping primate foot is lost in man due to the need for the feet to bear the body weight. The pelvis has become short, broad and basin shaped. It must hold the abdominal viscera and provide areas of attachment for the muscles which hold the body upright and move the legs. The reasons why man's ancestors first adopted bipedalism are unknown and speculative. Other primates have come to the ground and abandoned the forests, but none have become bipedal except man. One of the main advantages of this locomotor pattern is that it frees the hand from locomotion, so they can be used in carrying and manipulating objects such as tools.

SKELETAL CHANGES DUE TO BIPEDALISM AND THEIR IMPLICATIONS

The evolution of bipedalism approximately four million years ago has led to significant changes in the anatomy of Homo sapiens. The morphological alterations to the human skeleton that have occurred since the first bipedal hominid include changes in foot bone arrangement and size, hip size and shape, knee size, leg length, and vertebral column shape and orientation. The evolutionary factors that produced these changes have been the subject of several theories.

Foot: The human foot has been redesigned to act as a platform to support the entire weight of the body, rather than acting as a grasping structure, as it did in early hominids. Humans therefore have smaller toes than their bipedal ancestors. This includes a non-opposable hallux (Big Tow), which is relocated in line

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with the other toes. Moreover, humans have a foot arch rather than flat feet. When non-human hominids walk upright, weight is transmitted from the heel, along the outside of the foot, and then through the middle toes. Conversely, a human foot transmits weight from the heel, along the outside of the foot, across the ball of the foot, and finally through the big toe; this transference of weight contributes to energy conservation during locomotion.

Hip: Modern human hip joints are larger than quadrupedal ancestral species to better support the greater amount of body weight passing through them, as well as a shorter, broader shape. This alteration in shape brought the vertebral column closer to the hip joint, providing a stable base for support of the trunk while walking upright. Also, because bipedal walking requires humans to balance on a relatively unstable ball and socket joint the placement of the vertebral column closer to the hip joint allows humans to invest less muscular effort to balance. The shape change of the hip may have led to the decrease in the degree of hip extension, an energy efficient adaptation.

Knee: Human knee joints are enlarged for the same reason as the hip – to better support an increased amount of body weight. The degree of knee extension (the angle between the thigh and shank in a walking cycle) has decreased. The changing pattern of the knee joint angle of humans shows a small extension peak, called the "double knee action," in the midstance phase. Double knee action decreases energy lost by vertical movement of the center of gravity. Humans walk with their knees kept straight and the thighs bent inward so that the knees are almost directly under the body, rather than out to the side, as is the case in ancestral hominids. This type of gait also increases balance.

Limbs: An increase in leg length since the evolution of bipedalism changed how leg muscles functioned in upright gait. In humans, the push in walking comes from the leg muscles acting at the ankle. A longer leg allows the use of the natural swing of the limb so that when walking, humans do not need to use muscle to swing the other leg forward for the next step. As a consequence, as the human forelimbs are not needed for locomotion, they are instead optimized for carrying, holding, and manipulating objects with great precision.

Skull: The human skull is balanced on the vertebral column: The foramen magnum is located inferiorly under the skull which puts much of the weight of the head behind the spine. Furthermore, the flat human face help maintaining the balance on the occipital condyles. Because of this arrangement, the erect position of the head is possible without the prominent supraorbital ridges and the strong muscular attachments found in, for example, apes. As a result, in humans, the muscles of the forehead (the occipitofrontalis) are only used for facial expressions.

Vertebral column: The vertebral column of humans takes a forward bend in the lumbar (lower) region and a backward bend in the thoracic (upper) region. Without the lumbar curve, the vertebral column would always lean forward, a position that requires much more muscular effort for bipedal animals. With a forward bend, humans use less muscular effort to stand and walk upright. Together, the lumbar and thoracic curves bring the body's center of gravity directly over the feet. Also, the degree of body erection (the angle of body incline to a vertical line in a walking cycle) is significantly smaller to conserve energy.

Significance

Even with much anatomical modification, some features of the human skeleton remain poorly adapted to bipedalism, leading to negative implications prevalent in humans today. The lower back and knee joints are plagued by osteological malfunction, with lower back pain among the leading causes of lost working days. These problems resulted because human joints are forced to support more weight in bipedalism, now that the body's full weight is borne on two legs rather than four. In fact, arthritis has been a problem since hominids became bipedal. Scientists have discovered instances of vertebral arthritis in prehistoric hunter-gatherers; arthritis is not merely pathology of modern lifestyles. Physical constraints have made it difficult to modify the joints for further stability while maintaining efficiency of locomotion.

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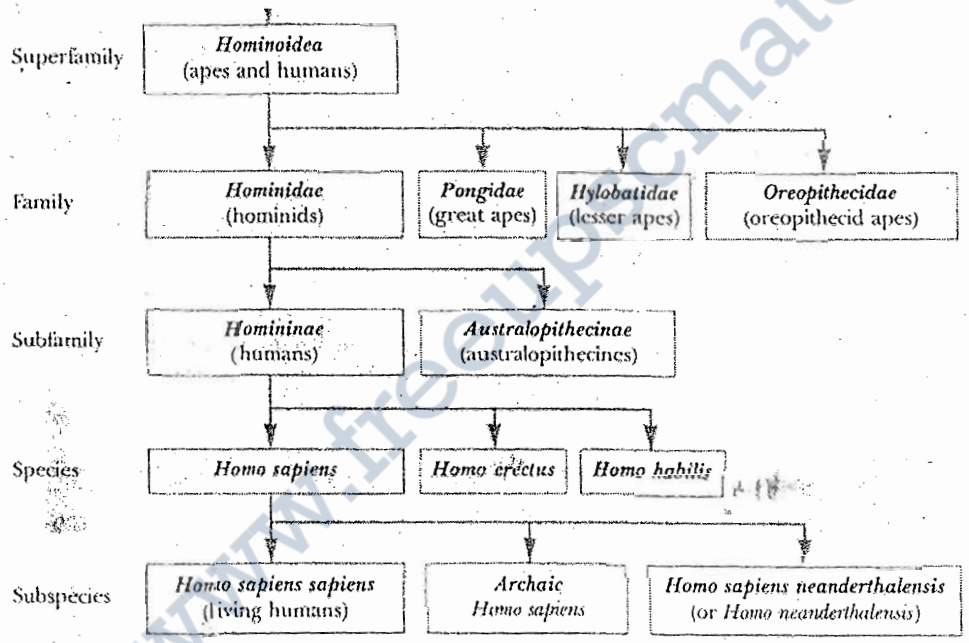
1.6 PHYLOGENETIC STATUS, CHARACTERISTICS, AND DISTRIBUTION OF HUMAN FOSSIL ANCESTORS

FROM MAMMALS TO HUMANS

The geological time scale starts with the formation of the earth, some 4.6 billion years ago. The first and longest span of time was the Precambrian, where forms of life were mainly small, simple, and soft-bodied. The onset of the Cambrian about 570 million years ago marked the rise of shelled animals in the sea. Then followed the "ages" of the fish, Amphibians and Reptiles, culminating in the domination of land by the dinosaurs, from about 200 to 65 million years ago.

Mammals first appeared more than 200 million years ago, but were overshadowed by the reptiles. However, some 65 million years ago all the dinosaurs – as well as other groups of reptiles on land, in the sea and in the air, and certain other animal groups and many plants too – became extinct over a relatively short period. This was only one of several "mass extinctions" that have occurred through geological time. The extinction marked the beginning of the Tertiary period and the Age of Mammals. The Tertiary is divided into epochs, and developments in the primate group can be traced from the fossils they left in the rocks formed during the time.

The first hominids (members of our own family Hominidae) crop up in the fossil record less than 4 million years ago. Mammalian evolution covers only about 4 percent of the Earth's entire history, and humans have been around for only 0.1 percent of the history of our planet.



HUMAN ORIGINS

The quest for our ultimate origins begins with the origin of life itself. The earth is about 4.6 billion years old. Fossil evidence shows that small, simple organisms were living at least 3 billion years ago. A great deal of evidence supports the notion that all present day organisms are related to each other, and these forms as diverse as slime molds and elephants, oak trees and beetles, roses and humans – ultimately arose from a single common ancestor, some 3.5 billion years ago. This means that a single evolutionary tree, or phylogeny, relates all organisms, living and extinct. Biologists and paleontologists assume that life

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evolved from its simple beginnings through a succession of stages, as represented in the fossil record, toward the present day diversity of some 10-30 million species. This does not mean that today's diversity is in some way the destiny or the end-point of evolution; there has been great diversity in the past, and there may be in the future.

RAMAPITHECUS

The last and most important hominid from Miocene is Ramapithecus. It is accepted by many scholars to be the first true hominid. The fossils of Ramapithecus (primarily teeth and jaw) come from two areas: the Siwalik Hills in India and Fort Ternan in Kenya. Other specimens have been discovered from Turkey, Hungary and Greece. The Ramapithecine fossils roughly date back to periods between 14 and 9 million years ago. The ecological setting of Fort Ternan and the Siwalik Hills fossils is that of a forest woodland environment. The Greek fossils, being younger, are that of a drier, savanna like environment.

The hominid features of Ramapithecus include reduced and vertically implanted incisors, and canines, little or no diastema, flattened and thick enameled premolars and molars that appear to be adapted for heavy chewing and processing of heavy food stuffs. Moreover, the placements of chewing muscles indicate an increased chewing pressure brought to bear on the food being eaten. These features, sufficiently different from the earlier Miocene fossils, indicate Ramapithecus direction to hominid line - perhaps the first hominid.

Ramapithecus specimens very strongly suggest the exploitation of a new dietary source - most likely seeds, nuts and grasses - that indicate a shift away from the softer forest fruits and vegetables relied upon by apes. This dietary shift is rather clearly associated with the climatic changes in the later part of Miocene that led to an increase in open grasslands and the decrease in the forest habitat of apes. There is a greater probability that this hominid form apparently was moving into a new ecological niche; it was beginning to exploit a more open ground environment similar to that inhabited by later hominids. Ramapithecus is also the most likely candidate for the ancestry of later hominids because of its presence in an area where the next hominids - the Australopithecines - have been found.

The possible adaptations that Ramapithecus made to open-ground living include an increased degree of hand and finger preparation of food, perhaps more frequent use of tools in such preparation, a tendency towards upright posture and bipedal locomotion for movement with a wide field of vision through the tall grasses on the open plain, possibly longer periods of growth and development, and perhaps, even a more frequent inclusion of meat in the diet. None of these adaptations can be clearly demonstrated because of lack of fossil evidences, but what we do know is that these adaptations were clearly present by the time the next phase of hominid evolution - the australopithecines - had begun. It is highly likely that Ramapithecus had begun to evolve and acquire those hominid features that led to the evolution of australopithecines.

(A) Plio-pleistocene hominids in South and East Africa - Australopithecines

After the youngest Ramapithecus, there is about 4 million years gap; then new hominid forms appear. These fossils show a continuation and elaboration of the basic hominid evolutionary trends. They are at least partially erect, bipedal, with reduced snouts and expanded braincases. They emerged on the rolling savannas of eastern and southern Africa some 5-6 million years ago.

There is a remarkable range of physical variation among these specimens, and this has made it difficult for scholars to discern where they fit in the evolutionary sequence. These variations in body size, facial shape, and brain size were caused by a number of factors, including sexual dimorphism, and localized adaptations to differences in the environment. It now appears that we can classify them into two major, largely contemporaneous groups - the *robust* forms and the *gracile* forms. These suggest two separate and distinct lines of evolution.

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The robust forms do not represent the main line of hominid evolution but are an aberrant sideline. Hominid evolution continued with the gracile or slender forms. To reflect this growing realization, australopithecines are divided into two genera - **Australopithecus**, which includes the slender forms like *A.africanus, A.aferensis, A.ramidus* and *A.anamensis*; and **Paranthropus** - which includes the robust forms - *P.robustus and P.boisei.*

Geographical Distribution

It was in 1925, Dart discovered the first fossil from Taung in South Africa. In 1936, Robert Brown discovered an adult cranium from Sterkfontein. Dart, Brown and Robinson discovered may more fossils at Kromdraii, Swartkrans, Makapansgat etc. In 1960s and 1970s many similar fossils have been discovered from Olduvai George in North Tanzania by Leakey, Hadar in Ethiopia by Johanson and Timothy White. Richard Leakey discovered similar fossil from Omo and Koobi Fora, in 1994, from Aramis, Ethiopia by White and in 1995 from Kanapoi, Kenya by Leaky.

Distribution of the australopithecines outside Africa is doubtful. In Africa, they are scattered in south and east. The distribution of this fossil type is summed up below.

South Africa

1. Taung, Cape province - A.africanus;
2. Sterkfontein, Transvaal - A.africanus;
3. Kromdraai, Transvall - P.robustus;
4. Makapansgat, Transvaal - A.africanus;
5. Swartkrans, Transvall - P.robustus.

East Africa

1. Tanzania - Garusi, Olduvai - P.boisei
2. Kenya - Lake Turkana - P.boisei
3. Ethiopia - Lake Omo - P.robustus
4. Ethiopia - Aramis - A.ramidus
5. Kenya - Kanapoi - A.anamensis.

AUSTRALOPITHECUS

This genus includes the gracile forms and is represented by four important species mentioned earlier. These creatures were roughly four feet tall, and weighed no more than 50 to 100 lbs. as mature forms. They were bipedal and erect and lived in open savanna country. Their bone structure was much less rugged compared to their robust relatives, and they generally lacked the sagital crest - a bony crest running down the middle top of head which served as a platform for the attachment of massive muscles that worked the jaw. Their teeth were fully hominid with small canines and incisors, no canine diastema and thick cheek teeth or the molars. Compared to Paranthropus, their incisors and canines were slightly larger and premolars and molars were slightly smaller.

General Charactersitics

1. A well developed fore-head suggesting the expansion of frontal lobes of brain.
2. Weakly developed supra-orbital ridges.
3. An elevated cranial vault indicating that a major portion of brain lies above the level of face, unlike pongids or apes where it is behind or below the face level.

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4. An estimated cranial capacity of 450-600 cc. Relative to its body size, Australopithecus has a slightly larger brain.
5. Skull probably is well balanced and foramen magnum placed slightly forward. This partially indicates an erect posture.
6. Extremely prognathus (forward jutting) lower jaw and relatively large jaws in general.
7. Hominid dentition and parabolic dental arcade.
8. No sagital crest indicating weak temporal muscles.
9. Hominid post cranial skeleton and Illium of pelvis is broad and short indicating a bipedal locomotion. Probably a better runner than a walker.

These morphological characteristics of the genus Australopithecus, in general, suggest them to be ancestors to modern man.

PARANTHROPUS

These robust adults average close to 5 ft in height and may have weighed as much as 100-150 lbs. They had the limbs of erect and efficient bipeds and were thus well adapted to the savanna grassland environment which they inhabited during the late Pliocene and early Pleistocene. Their cranial capacity approximated 500 cc. They had heavy brow ridges, no vertical forehead and sagital crest. Their teeth were fully hominid, arranged in a parabolic arch with relatively small incisors and canines, and no canine diastema. Their premolars and molars, however, were quite large.

This genus is represented by two species - *P.robustus* and *P.boisei*. *P.robustus* was a massive form from South Africa measuring 3 ft. 11 inches and weighing around 40 - 80 Kgs. They had larger cranial capacity with pronounced supra-orbital ridges and massive zygomatic arch.

The fossil *P.boisei* was from East Africa measuring 4 feet 4 inches tall and 40-80 Kgs in weight. Previously it was referred to as *Zinzanthropus boisei* by the Leakey's. These fossils were also discovered from Tanzania, Kenya and Ethiopia.

General Characteristics

1. Height averaged 4 feet and weight about 120 lbs.
2. Skull larger than Australopithecus.
3. Cranial capacity is around 450 - 600 cc compared to body size. This animal had a lower brain to body size ratio compared to its australopithecine counterpart.
4. Large supra orbital ridges without conspicuous forehead.
5. Cranial vault is not well rounded and expanded.
6. Moderately developed sagital crest.
7. Massive jaws with moderate prognathism.
8. More extensive neck muscles.
9. Forwardly (slightly) placed foramen magnum on the skull base.
10. Parabolic dental arcade with slightly more U-shape. Entire dentition probably designed for extreme crushing and grinding.

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11. Post-cranial skeleton shows a mixture of Gorilloid and Hominid traits.
12. Hand skeleton indicates knuckle-walking.
13. Divergent big toe.

The general characteristics of Paranthropus made Robinson suggest an arboreal adaptation of this fossil. Though the total morphological pattern shows some pongid traits, it is a hominid without any controversy. Probably these creatures adapted to an ecological niche similar to that of Gorillas.

AUSTRALOPITHECUS AND PARANTHROPUS – A COMPARISON

TRAIT	AUSTRALOPITHECUS	PARANTHROPUS
Cranial capacity	450 - 600 cc	450 - 600 cc
Cranial vault	High and expanded	Low placed and not expanded
Sagital crest	~~Present~~ absent	Absent in male
Frontal region	Rounded and well developed	Under developed
Supra-orbital ridges	No development	Well developed
Jaws	Large	Massive
Simian shelf	Absent	Present
Dental Arcade	Parabolic	Less parabolic
Foramen magnum	Hominid like, but not in modern position	Even more forwardly placed
Foot	Non-divergent big toe	Divergent big toe

Phylogenetic Implications

These Pliocene & Pleistocene fossils have shown both hominid and ape-like features. However, the hominid features are overwhelming. The presence of ape-like features can be accounted for by way of common inheritance from a hominoid or pongid ancestor. The Hominid characters however can be accounted for by way of independent acquisition demonstrating and highlighting the fact that these fossils were on a direct line of human evolution and not pongid line. There are many schools of thought prevailing with reference to the course of evolution these australopithecines have taken. The most important of these are discussed below.

According to Donald C. Johnson and Timothy White, the East African fossil *Australopithecus aferensis* split into two branches, an australopithecine line represented by *A.africanus, P.robustus, P.boisei* and a hominid line represented by *Homo habilis, Homo erectus* and *Homo sapiens*. This split was supposed to have happened 3 million years ago. The australopithecine line progressively became robust. This pattern, generally called Two Branched Theory had its variants also. For some, it is *A.africanus* which is the common link between the Australopithecine line and Homo line. For still others, these two branches represent parallel evolution.

According to this two-branch theory, *A.aferensis* gave rise to *A.africanus* 3 mya, of the same height, living up to 2 mya. Next arrived *A. robustus*, which showed marked increase in robustness of body, face, jaws and teeth and it had lived up to 2.3 to 1.8 mya. Finally, the last and the most robust form, *A.boisei*, lived in East Africa from roughly 1.8 to 1 mya.

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The second branch of this model, the Homo line, also shows a shortening of the face but there is a marked decrease in the size of both the cheek teeth and the front teeth. There is a massive increase in the size of brain also. This line begins with a transition from *A.aferensis* to *H .habilis*, the first hominid who made and used tools, and lived in Africa from 2 - 1.5 mya. They had human like teeth and a larger brain than australopithecines (750 cc). However, we can conclude that hominid evolution may not have been so simple, isolated and clear-cut in its operation. Instead, there is a possibility that three or more hominid lineages may have been evolving and interacting with each other.

This two branch theory was widely accepted till the discovery of a new type of hominid skull – *Australopithecus aethiopicus*, in Northern Kenya in 1985 by Alan Walker. This skull is considered to be the most robust form ever discovered. It had massive teeth and ape-like brain. The dating of this specimen indicates that the family of *A. boisei* did not evolve in the last leg of the australopithecine evolution, as indicated by earlier theory, but it originated directly from *A.aferensis*. Thus, the revised theory holds a three line evolutionary sequence, one to *boisei* line, second to *Homo* and third to *africanus, robustus* line having *A.aferensis* as the common ancestor.

The discovery of the youngest Australopithecine - A. *ramidus* has added a new dimension to the three branch theory. It holds that A.aferensis is the common ancestor for *Homo, P.boisei* and *P.robustus* but it itself evolved from *A.ramidus*. With over specialization of diet, competition for food with *H. habilis* and the latter's predation along with *H. erectus* led to the extinction of the australopithecine group.

HOMO HABILIS

In the early 1960s, the Leakey's discovered at Olduvai Gorge, close to and contemporaneous with *Australopithecus boisei*, a form that appeared much closer to modern human beings. Its features were more refined, with somewhat more modern teeth. The average cranial capacity of the specimen proved to be about 640 cc. They concluded that they found the first "real" human beings; and they called these finds *Homo habilis*. To this day, the taxonomic status of *H. habilis* is disputed. Some regard them as more modern australopithecine forms. These forms are considered to be on a direct evolutionary line between the Australopithecines and Homo erectus, the more primitive Paranthropus is best viewed as a side branch on the tree of human evolution.

In 1994, Bernard Wood emphasized that the foramen magnum is further forwardly placed in *H. habilis*. The teeth are narrower than, but not as small as that of, *H. erectus*. The age of the fossil is estimated to be around 2 - 1.5 mya.

General Characteristics

1. The cranial capacity ranges between 650 - 800 cc. The cranial capacity was greater than that of australopithecines and approached that of *H. erectus*. The brain to body size ratio also suggested evolutionary advancement.
2. Unlike australopithecines, the frontal lobes (seats of mental abilities) of brain were well developed. They almost approximated that of modern man. The cranial vault was rounded.
3. Hominid dental structure. The premolars are particularly human like.
4. The mandibles (lower jaw) are comparatively less massive than that of australopithecines. However, the dental arcade was not parabolic.
5. The hind limb morphology clearly approached the human foot.
6. *H. habilis* was a tool maker. The toolkit is referred to as "Olduwan industry". It included cutting tools, scraping tools and tools to make tools. Meat, plants and wood were worked upon. Bones, seeds and nuts were cracked open. There is an evidence of campsites also.

Discovery / habitat / Characteristics

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7. Larger brain involves longer gestation, larger newborns and greater longevity. The brain enlargement was perhaps the cause and the effect of dietary shifts in *H. habilis*.

The distribution of *H. habilis* and *P.boisei* overlap. Both are found in East Africa during 2 to 1 mya. Homo habilis originated from some gracile australopithecines and represents a massive increase in brain size in a short span of time. Some scholars disagree to the view that these are true homo. Their teeth are large relative to body size and their limb proportions show they are closer to australopithecines than homo. Some even suggest placing these fossils in separate genus.

Phylogenetic Implications

There has always been a question haunting the students of Paleoanthropology; *why Homo habilis, not Australopithecus habilis?*

One of the main criteria for inclusion in the genus Homo was brain size. Different authorities had different threshold sizes, ranging from 700-800 cc. Most australopithecines were around 500 cc, while *Homo erectus* was above 1000 cc. What of *H. habilis*?

For some others, a brain size of more than 600 cc would suffice for inclusion in Homo, along with other features indicated by the fossils, such as upright posture and bipedal walking, and a precision grip in the hand. The first *H. habilis* specimen had a brain size estimated at 680 cc.

With the discovery of the Olduwan tool tradition, the discussion centered on tool making. Many authorities were unhappy about tools as admissible evidence in support of this new species. The history of hominid fossil discoveries is littered with names coined in excitement of a new find. Many of these names have not stood the test of time.

In the mid 1960s, many people thought it probable that *Homo erectus* had evolved from some type of australopithecine. Therefore, it would be natural to find fossils representing transitional form - the result of evolution in action. Rather than create a separate species, this new form should be placed either with its predecessors, the australopithecines, or with its successor, *Homo erectus*. According to Le Gros Clark, *Homo habilis* can be easily accommodated within *A.africanus*.

There were plenty of other opinions about where *Homo habilis* fitted into the Hominid evolutionary tree. Leaky long held the view that our modern species, *Homo sapiens* was in fact very ancient and could be a direct descendant of *Homo habilis*. This would push Homo erectus out onto a limb, making it our cousin and not our ancestor.

In 1972, Richard discovered a skull at Koobi Fora which had a cranial capacity of 800 cc. A brain size of this surely meant the creature to be a member of genus Homo?

Till today more than 20 *Homo habilis* individuals have been found. Besides having a bigger brain than australopithecines, *Homo habilis* also had a higher brain-to-body ratio. Compared to apes, the temporal lobes which deal with memory and other mental functions are more developed; so are the parietal lobes which analyze information coming in from senses and the speech processing area is also developed.

The version of our evolutionary tree in which *A. aferensis* gave rise to *H. habilis*, which evolved into *Homo erectus*, which in turn produced *Homo sapiens*, has an appealingly neat straight line feel to it. But standing back from the main habilis time period, about 1.9 - 1.6 mya, brings various complications.

What about *before* the habilines? An alternative to the *aferensis - habilis - erectus - sapiens* view is that "early Homo" was already living more than 3 mya. Gradually the early Homo became *Homo sapiens*. Whether it passed through the habilis stage on the way is an area of lively debate.

What about *after* the habilines? By 1.6 million years ago, another human species had appeared. The taller, long-legged, bigger brained *Homo erectus*. If *habilis* did evolve into *erectus*, could such a great evolutionary change have taken place so quickly? One possibility is punctuation - a rapid burst of change

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in the type of evolutionary process known as **punctuated equilibrium**. Another is that some relatively isolated habiline populations began to evolve into *erectus* at much earlier date, leaving the rest of their kind to continue unchanged, and eventually go extinct about 1.6 million years ago.

The time between 2 and 1.5 million years ago has been called the "**crucial humanizing period**". Recent fossil finds from this time have widened the debate as to how many species of hominids lived in Africa then, and who evolved into what. More fossils may serve to clarify the picture, or further to confuse it.

(B) Homo erectus

In the year 1891, Eugene Dubois discovered a fossil skullcap with few associated teeth and bone, from the solo river region of Java. He provisionally named it *Anthropopithecus erectus*, "upright man-like ape". By 1894, the position regarding this fossil changed. The upright man-like ape had become *Pithecanthropus erectus*, the "upright ape-like man". The discovery of thighbone – femur - revealed a distinctly non-ape like upright stance and gait. This fossil was commonly called as the "**Java Man**".

Homo erectus ranged from Africa to Eurasia from over 1.6 to 0.12 mya. A primitive face characterized this hominid with considerably large brain than any of the australopithecines, approaching the lower limit of brain capacity in modern humans. This hominid made stone tools, and knew the use of fire and was probably the direct ancestor of *Homo sapiens*. It is supposed to have evolved side-by-side Paranthropus and became extinct by 0.12 mya in the wake of competition from more efficient Homo forms.

In the year 1921-29, W. C. Pai discovered several fossil skulls and teeth from Zhoukordian (Choukeutian) near Peking, China. These fossils were studied by Black who assigned the name *Sinanthropus pekinensis* to it. These two earlier finds from Java and China were supplemented by two more finds from these two regions. Because of the similarities of the Javanese and Chinese fossils, the different generic names were done away with. Initially the Chinese fossil was named *Pithecanthropus pekinensis*. Later even its name changed to *Pithecanthropus erectus*.

Dubois' fossils are now regarded as the first discovered specimens of *Homo erectus*, an immediate predecessor of *Homo sapiens* that lived in Africa, Asia and Europe, from 1.6 mya to less than 300,000 years ago. The Java specimens along with those from China were renamed *Homo erectus* in 1950s by evolutionary biologist Ernst Mayr. Thus, *Homo erectus* became the first officially named pre-historic species of human genus Homo.

Homo erectus was about 5ft tall, with a body and limbs that were within the range of variation of modern humans. The only primitive aspect of this ancestor of ours was its head, which exhibited such features as heavy ridges of bone across the eyes, a small fore head that sloped back dramatically, a narrowing of skull behind the eye orbits, and a slight sagital keel (all of which suggest heavy facial musculature), and a cranial capacity ranging from 900-1200 cc and averaging around 1000 cc.

Homo erectus roamed across all of the African and Eurasian continents. They came to occupy environments far beyond those that could be occupied by the australopithecines. They were, thus, far more adaptable than the australopithecines so much so that we are considering them to be a new stage of evolutionary progress beyond the australopithecines. At this stage of evolution there was cultural and behavioral progress as well. The significance of this cultural progress is that in order to adapt to the new and different ecological zones that it occupied, *Homo erectus* did not have to evolve into many different biological forms. Instead, *Homo erectus* adapted to these challenging environments primarily through learned behavioral means.

General Characteristics

Homo erectus represents an intermediate stage between *A.africanus* and Hominids of late Pleistocene. Compared to A.africanus, the skull is broader and is set lower. The cranial capacity of Java man ranged between 800 - 975 cc whereas that of Peking man, between 850 - 1300 cc. This clearly reveals that the Peking man had taken a step forward towards modern man in terms of cranial capacity. It is estimated

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that in a period approximately one million years ago, the Hominid brain size has literally doubled from 600 to 1300 cc. Probably the capacity to make tools has brought a selection for a larger and more complicated brain.

One characteristic feature of *Homo erectus* skull is its extreme platycephaly (flatness in the head). This flatness in head is less extreme in Peking man. The frontal region has a large supra-orbital torus, a continuous bar of bone. However, in Peking man there is a trend towards the separation of this torus into two large supra-orbital ridges. The frontal region in Java man is low, but Peking man exhibits the beginning of an expansion into a true vertical forehead. The cranial bones are generally thick walled. The foramen magnum is like the modern man and is clearly located in a position indicating that it was a habitually erect and bipedal Hominid.

The table below attempts to compare both the species of *Homo erectus.*

Trait	*Homo erectus javanicus*	*Homo erectus pekinensis*
Cranial capacity	775 - 975 cc	850 - 1300 cc
Platycephaly	Extreme	Less pronounced
Sagital keel	Extreme	Reduced
Supra-orbital	Huge torus region	Reduced, beginning to divide into separate ridges
Mandible	Large	Reduced, modern
Chin	Absent	Slight indication of the beginning
Diastema	Occasional	Absent
Canines	Some projecting	Non-projecting

The face of *Homo erectus* is broad and large, broad nasal aperture and nasal bridge is slightly depressed. Zygomatic bones are larger in relation to total facial dimensions. The Java specimens occasionally present a diastema and a slightly projecting and pointed canine in the upper jaw. No such diastema is noted in the lower jaw in Peking man. The canines reflect carnivorous nature of *Homo erectus.* The molars are morphologically more advanced over *A. africanus*, and exhibit **Taurodontism** - large pulp cavities and fused roots in molar teeth, as can be seen in cattle (hence the name Taurodontism). The palate is huge and parabolic. Chin and Simian shelf are absent.

The study of thigh bone in both the cases indicates that *H. erectus* was taller than Australopithecus. The Java man was 5 ft 8 inches tall while Peking man around 5 ft, 2 inches. There seem to be nothing simian about the straight and slender thigh bone. Linea aspera on the femur indicating vertical posture is present (refer to the notes on bipedalism). In the Peking man, the thigh bone is slightly shorter and bent.

The fossil remains of *Homo erectus*, particularly form Java and China demonstrate that the range of variation of many features falls within that of modern *Homo sapiens.* However, the fossil skulls are relatively thick as we find sometimes in modern man. Similarly, the teeth of *Homo erectus* were large, like those noted in contemporary American whites. The height and breadth of mandibles are larger.

During the lower and middle Pleistocene the early forms of Homo are believed to have evolved into *Homo erectus* 1.5 mya. *Homo erectus* had the requisite basic human cultural adaptation. It is believed that some kind of a vocal communication was possible even at this stage. He had a much wider geographical range and diversity of environments. He used fire, lived in caves, used animal skin for clothing and made shelters and was a big game hunter. It is believed that the beginning of rituals, formation of nuclear family

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and some other aspects of human socio-cultural system was developed by *Homo erectus*. By the time of *Homo erectus*, human evolution strangely developed its bio-cultural bases.

There are no fossils of *Homo erectus* from less than some 200,000 years ago. What happened to them? Estimates for a world population of *Homo erectus* have been made to around 1 million. Were they our ancestors? If so, once they had spread across the continents, did the populations in each region evolve into the various *Homo sapiens* groups of today? Or did a select band of erectus, perhaps from Africa, become founding population of *sapiens*, and was there a continental migration of humans? This is one of the hottest areas of debate in paleoanthropology.

Phylogenetic Implications

One might well ask: What caused the biological advance that led early Homo to evolve into *Homo erectus*? It is possible that we will never know for sure. One of the more plausible theories has been put forward by Grover S. Krantz in 1968. He points out that contemporary hunting and foraging groups with Stone Age technologies engage in what he terms persistence hunting. Humans are unique in their ability to pursue game over vast distances, literally driving their quarry into the ground. To persist despite fatigue, hunger, thirst and discouragement requires a tough minded commitment to a mental image of the success of the hunt. The ability to maintain this image for motivational purposes requires a memory of previous successes. One of the conspicuous aspects of the transition from the australopithecine stage to that of *Homo erectus* is the dramatic expansion of the size of the brain and the development of the cortex. Krantz suggests this evolutionary development underlies an improved memory and thus provided a selective advantage to those individuals who were consequently better able to keep memory images in their minds to motivate their hunting behavior.

Why did our ancestors adopt persistence hunting as a survival technique? Possibly because they were bipedal. Although bipedalism is a very useful adaptation to living in the tall grasses of savanna, and also makes tool use possible, as a form of locomotion, it is very slow. Indeed, it is far slower than the means of locomotion of the animals our ancestors hunted. Bipedalism, like all adaptive specialization, solved old problems but posed new ones in the process.

One does not have to stress persistence hunting as a selective pressure for the brain expansion and the increase in its organizational complexity that marked the transition to *Homo erectus*. After all, the information that we have about contemporary foraging groups suggests that only 20 to 30 percent of their diet is derived from hunting, and we have no reason to suspect that the ratio was much different in the case of our early ancestors. Nevertheless, the incorporation of regular, cooperative hunting into a primate social system must have had a profound and extensive impact. Virtually every aspect of social behavior must have been affected. And not only social behavior - the body itself quite probably changed significantly. These changes included physical adaptations to day time hunting like increased pigmentation of the skin, loss of fur on the body and increase in the number of sweat glands.

The next question is: Where exactly was the boundary between the australopithecines and *Homo erectus*? Clearly, many intermediate forms emerged as the australopithecines evolved. But nevertheless we find it useful to draw an arbitrary line, indicating as we have, those features that characterize the australopithecines, and those that are qualities of *Homo erectus*. The basis for separating these forms from each other is thus made clear. Also, in the same arbitrary manner, we can decide which forms are sufficiently similar to modern human beings, that we can now answer the question. Most australopithecines were sufficiently different from us to warrant being placed in a separate genus; even early Homo resembled the other australopithecines more than us. But *Homo erectus* has an overwhelming resemblance with us, from the neck down, and exhibits a brain that, in both size and complexity, widely overlaps the lower end of the range of variation of modern human brains. Thus we may, with assurance, refer to Homo erectus as an extinct ancestral form of human being.

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Homo erectus heidelbergensis (Mauer Jaw)

For quite some period of time, the presence of *Homo erectus* in Europe has been questioned. However, in the year 1907, a mandible (lower jaw) was discovered from a sand pit at Mauer, near Heidelberg, West Germany. The site of discovery abounds in animal fossils such as those of elephant, horse, wild boar, bison, wild cat, red deer, etc. It is ascribed to the period about 450,000 years ago. This jaw, called the Heidelberg jaw, is very massive, lacks chin, but the teeth are relatively small and resemble those of the Neanderthal man. This was designated as *Homo erectus*. This jaw is believed to represent a transitional stage between *Homo erectus* and *Homo sapien neanderthalensis*. In the year 1981, Stringer assigned a date of 0.35 to 0.45 million years to this jaw.

General Characteristics

1. It had all its teeth in place.
2. The jaw is very large and massive. The general profile is broad and square in shape. Its breadth is large compared to that of the modern man.
3. The condyloid processes are blunt and rounded and are at a higher level compared to the modern man. In this case, it resembles the jaw of the Gibbon.
4. The angle of the jaw is truncated and the meeting place of two sides of mandible is very thick. These characters resemble an ape's jaw.
5. The jaw is parabolic like that of the humans.
6. Dental series is continuous without a diastema as in the modern man.
7. The teeth are of ordinary size like that of the modern man. Incisors are normal and canines do not project.
8. The jaw displays more simian features whereas the teeth display more human features.

Phylogenetic Implications

Opinions vary as to whether the owner of the Mauer mandible, "Heidelberg Man", was a representative of *Homo erectus*. The jaw shows some similarities, yet it also has features that can be interpreted as Neanderthal. It might indicate that European *Homo erectus* evolved into Neanderthal people.

(c) Neanderthal Man

The term Neanderthal man is given to an assemblage of fossils showing certain common features which characterize the type of specimen viz., the skull cap with limb bones and ribs recovered from a cave in the Neander valley near Düsseldorf in Germany. Thus the term Neanderthal, in the strict sense, is used to describe a series of finds from Western Europe dating from the early part of the last glacial (about 170,000 years ago) to the rise of modern man around 35,000-40,000 years ago. These have also been called **Classic Neanderthals.** The term Neanderthal, however, has been extended to include all series of Hominid remains dating about 100,000 years to the rise of modern man. Such specimens have been reported from Asia, Europe and Africa. The Neanderthals not conforming to the Classic form are often referred to as **Progressive Neanderthals.**

Neanderthals are well known and generally consistent forms of Hominids represented by fossilized skeletal material found in caves throughout Europe including Germany, France, Belgium, Gibraltar, Italy, Yugoslavia, and Czechoslovakia; North Africa, Israel, Iraq and Uzbekistan and Central Asia. As hundreds of Neanderthal remains have been found in most parts of the world, it is not practically possible to describe all Neanderthal finds individually.

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It is now apparent that there are at least two distinct types of Neanderthal man: **Conservative type** and **Progressive type**. These two can be distinguished on the basis of their morphological features. The Conservative type has a flattened down brain case, a bun shaped protuberant occiput, marked projection of the jaws and practically no ape-like features.

The Progressive type in contrast had a laterally compressed and higher cranial vault, lower attachments of neck muscles and medium to fair development of the bony chin. The characteristics of the skeleton approximate to those of primitive but anatomically modern forms of man. We shall now take up both these types separately in detail and then discuss their relationship to *Homo sapiens*.

CLASSIC NEANDERTHALS - LA-CHAPELLE-AUX-SAINTS

Perhaps the best preserved of all the Neanderthal skeletons of the Classic or Conservative type is that of the old man of La-Chapelle-aux-Saints found in August 1908, in a small cave of Correze, France. The associated material includes flint tools of Mousterian culture with retouched blades, scrapers and keeled scrapers, conforming to the Aurignacian type. The fauna included bones of mammals like woolly rhinoceros, reindeer, wild horse, bison etc. The French paleontologist M. Boule described the fossil material during 1911-1913.

General Characteristics

The skull of the old man of La-Chapelle-aux-Saints is very large. The brain case is elongated and low, the supra orbital ridges are immense, the forehead is low and retreating, the occiput is protuberant and bun shaped and the whole braincase seems to have been flattened down. Face is long and projecting, the orbits are very large, nose is long and very broad, upper jaw is strongly prognathus, the mandible is powerful and the chin rudimentary.

A comparative evaluation of the features of face and brain-case with apes and the modern man indicates that apes differ from man in having larger face and smaller brain case. Neanderthal man, however, falls between modern man and chimpanzee both in facial projection and in the height of the cranial vault. The fossil human skull in both aspects is much nearer to the modern human type than to the chimpanzee. The crania of Conservative Neanderthals are long headed and low vaulted. The mastoid processes are small and rudimentary. These are, however, absent in apes and very well developed in modern man. The fossil skull exhibits bun-shaped and protruding occiput which is traversed from side to side by a torus for the attachment of neck muscles. In Anthropoid apes only a crest is present. This also indicates that the neck muscles and ligaments are hafted much higher up on the back of the skull than in modern man (an ape-like feature). The base of the skull exhibits many primitive features. The foramen magnum is situated farther back in the skull base than in modern man. But it is much anterior compared to ape condition. The lower jaw is massive, chin is rudimentary, but no simian shelf is present. The teeth are arranged in a very broad 'U' shaped (towards being parabolic) arch with the canines set in the curve and they are not projecting. The wear of the teeth indicates that the lower jaw, while chewing, was thrust forward and retracted in a different manner than in modern man.

The trunk and the limb skeletons are well preserved in La-Chapelle-Aux-Saints specimen. The spine is short and massive; the neck vertebrae have long, almost horizontal, spiny processes quite similar to chimpanzee and much different from modern man. These processes provide attachment for the muscles and ligaments supporting the over-balanced jaw. The spines of the neck vertebrae are backwardly projected. The thigh bones are strongly bowed forward as in apes. They are also short and massive with great articular heads. It is sufficiently marked to indicate an upright carriage. Tibiae or shin bones are short and strong. The upper surface of their articular head forms the lower part of the knee joint. They are sloped backwards indicating "bent-knee walking". On the basis of long bones, it is estimated that Classic Neanderthals were of low stature varying from 5 feet 1 inch to 5 feet 5 inches.

An estimate of the cranial capacity of Conservative Neanderthals indicates that it was not very small. The cranial capacity of Gibraltar skull belonging to a female works out to 1300 c.c. while that of the La Quina

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female skull is believed to be 1367 c.c. The cranial capacity of female skull from Neanderthal is assessed to be 1408 c.c. The old man of La-Chapelle-aux-Saints had a cranial capacity of about 1600 which is far above that of the average Europeans of today.

The endocranial casts of Neanderthal skull reveal some important features. The La-Chapelle-aux-Saints had a long, broad and low brain, the left lobe slightly larger indicating perhaps right handedness. The pattern of convolutions was simple unlike that of modern man. In Anthropoid apes the frontal lobes constitute about 32 percent of the cerebral surface. In case of modern man it works out to 43 per cent whereas in La-Chapelle-aux-Saints it is 36 percent. These estimates are believed to demonstrate an expansion of all the lobes in comparison to *Pithecanthropus*. The frontal lobe is slightly less expanded but its inferior frontal convolutions (the area of speech) are prominent. The parietal lobes are greatly expanded indicating greater sensory development and the better control of manual movements. The temporal lobe is well developed and exhibits a large auditory eminence. The visual and visuo-psychic areas are increased. Thus, Neanderthal man's brain was massive and primitive, but distinctly human.

The La Chapelle remains were one of the first major Neanderthal discoveries, even though over a half century had passed since the Neander valley find of 1856. None of these earlier materials provided substantive information about the Neanderthals prior to the discovery of the La Chapelle remains. The relatively complete skeleton of the latter, with good preservation and good archaeological context, offered one of the earliest opportunities for an extensive analysis of Neanderthal characteristics. The discoverers entrusted their find to France's world renowned paleontologist Marcellin Boule for description and analysis. The combination of the most complete known remains of Neanderthal and a scientist of Boule's ability would seem too ideal, but ironically, that was not to be the case.

Boule's description of La Chapelle was widely accepted as an accurate picture of what Neanderthals had been like. As reconstructed by Boule, Neanderthals had a less than fully upright posture, walked in an awkward, lumbering manner, were much inferior to modern humans intellectually, and in short had been primitive creatures barely deserving inclusion in the genus Homo. Boule believed Neanderthals to be an extinct sideline to the mainstream human evolution and to belong to a different species than modern humans. In support of his views he cited knee joints that could not be fully extended, great toes that diverged from the others to form an apelike grasping organ, neck vertebrae more chimpanzee like than human, and a simply curved spine without the modifications found in modern humans which allow a bipedal posture with the skull balanced on the top. Consequently, Boule believed Neanderthal skulls, with their big, projecting faces and long, low brain cases, were thrust forward in a basically ape-like position. In these and dozens of other ways, including purely speculative comments about their presumed mental abilities, Boule created an image of Neanderthal people that is still recognizable in the "cave men" that inhabit the bad novels, comic books etc. Unfortunately, for many years this was also an image largely accepted by many anthropologists. These interpretations, especially when considered with the fraudulent Piltdown remains from England, seemed to preclude a direct evolutionary connection between the Neanderthals and modern humans.

Boule's view was not, of course, universal. Still, it was not until the 1950s that the extent of the fundamental errors in Boule's recognition became clear. In 1955 it was found that the gait proposed by Boule simply would not work. It implied a center of gravity too far in the front of the body's axis of support and would have precluded standing still without falling over. The final blow however came in 1957 when W.L. Straus and A.J.E. Cave reanalyzed the La Chapelle fossil. The skeleton was that of a robust male 40 to 50 years old, who had suffered from severe and extensive arthritis. To Straus and Cave it was obvious that any lack of a fully upright posture was pathological in origin. Furthermore, they contended, "Boule had made numerous errors in the reconstruction of the foot, the knee, the skull, and other parts of the skeleton". They concluded that there was "no valid reason for the assumption that the posture of Neanderthal man of the fourth glacial period differed significantly from that of present day men". They did not claim a lack of significant morphological differences, but they did rather whimsically suggest that a properly bathed, shaved, and dressed Neanderthal would probably not be noticed in a New York subway! In effect, a faulty reconstruction of an arthritic individual had been accepted for decades as

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typical of the Neanderthal population. Ironically, even an accurate reconstruction would have left room for misinterpretation. The La Chapelle skull, by pure chance, is among the most robust and long headed Neanderthal skulls known.

Straus and Cave are restrained in criticizing Boule, noting that he was convinced of the "sideline" nature of the Neanderthals because he believed that the true ancestors of modern human had already been identified in contemporaneous, but more modern looking fossils. Some other writers have been less charitable. Brace has suggested that Boule was essentially "anti-evolutionary" in his views, due partly to his training under the catastrophist Cuvier, and that he either consciously or unconsciously slanted his report so that it supported what Brace has called "hominid catastrophism".

To us it seems likely that the interpretations were unconsciously affected by the expectations. When judgment was required in assessing any particular part of the fossil, Boule tended to see primitive, rather than more advanced characteristics. Boule's belief in contemporaneous but more modern fossils and his still puzzling neglect of the pathologies present, especially when combined with an unusually robust specimen, resulted in a long series of errors, which collectively resulted in a seriously inaccurate picture of Neanderthal morphology, posture, and locomotion. He then postulated an even more distorted figure of Neanderthal intellect, and the concept of the archetypal "cave man" thus created, flourished for the next four decades.

PROGRESSIVE NEANDERTHALS – MOUNT CARMELITE SKELETONS

Neanderthal material outside Western Europe has usually been referred to as Progressive because it is less extreme than the Conservative type and exhibits many features of modern man. However, such a line of distinction is not agreed upon by many scholars, particularly in view of the discovery of a very extreme form from Rhodesia. Generally people living during the progressive phase display greater variability, rendering the Classic and Progressive designations purely simplistic. It may be stated that the earliest Neanderthals were most extreme, having long low skull, bun-shaped or angular occipital bone, and well developed brow ridges and large faced as well as some minor specializations of the post-cranial skeleton. The later discoveries, however, show that they were more like modern man. Furthermore, the extreme and the modern forms existed side by side and have often been found in the same site. These facts indicate that the Neanderthal population was a very variable one. Ever since the discovery of these Progressive Neanderthals, particularly from Palestine, the interpretation of Neanderthal problem has taken a new turn. These Progressive Neanderthals are morphologically different from Classical Neanderthals and some of them are similar to modern man. The more important finds related to Progressive Neanderthals are the Mount Carmelite skeletons.

The Mount Carmel skeletons were found buried in the adjoining caves which yielded Levalloiso-Mousterian culture. In terms of the time schedule, it covers a considerable part of third interglacial and the beginning of the last glaciation. The discovery was made during the excavations of the joint expedition of American School of Prehistoric Research and British School of Archaeology in Jerusalem in 1931-1932. The work was conducted under the direction of Miss Dorothy Garrod and the material was exhaustively studied by Sir Arthus Keigh and Theodore McCown. The first cave (Skuhl) yielded fragmentary remains of ten individuals and considerable number of isolated specimens. The second cave (Tibun) yielded the skeleton of an adult female, mandible of an adult male, some bones and teeth. The Skuhl material represented the Progressive Neanderthals whereas the Tibun material represented the Conservative type. The *stone tool industries* were the same in both caves.

When the Progressive people of Skuhl are compared with the Conservative type, it is noted that Conservatives are short and stocky while the Mount Carmel men are tall and the women short to medium. Both have massive heads, the face is relatively larger in the Conservatives. However, this portion of the skull is not excessively developed in the Progressive Mount Carmelites. The Progressives have medium high vault and their foreheads are not receding. Their occiputs are not compressed vertically but project beyond the attachment of neck muscles. The eyebrow ridges in both types are very large. In Palestinians

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they separate into medial and lateral parts and are not continuous. The Conservative Neanderthals have long and prognathus face, while in Progressive, the face is of moderate length and is orthognathus. The orbits are high in the classic type whereas they are wide and not so high in Progressives. The nose is quite variable in its proportions (lateral margins are demarcated from the face) unlike the condition found in the Conservatives. The Conservatives have much deeper jaws in the sub nasal region and the chin eminence is rudimentary. In Progressives the jaws are of moderate depth and the chin, though variable, is better developed.

The teeth are larger in the Conservatives but the molars show certain primitive features in both types. Keith and Boule have worked out the cranial capacities. In the Conservatives it works out to 1400 cc. and in Mount Carmel skulls it ranges 1518-1587 cc. However, the female skulls at Tibun have 1271 cc. whereas the Skuhl female skulls range in their cranial capacities from 1300 to 1350 cc. The comparison of the endocranial casts of two types indicate that the size and form of the brain of the Mount Carmel people lack the primitive and simian features of the Conservative type. In their limb skeleton the Palestinians neither have shorter legs nor somewhat imperfect adaptation to the human erect posture. In these characteristics the Palestinians approach modern man.

Disappearance of Neanderthal Man

The Classic Neanderthals seem to have disappeared between 35,000 to 40,000 years ago, as we have no fossil evidence thereafter. According to one view, they are believed to have been driven out by the migrations of advanced modern forms which are now referred to as *Homo sapiens sapiens*. However, some others believe that Neanderthals gave rise to modern man. Brose and Wolpoff (1971), however, are of the opinion that there is a continuous development of Neanderthal man to *Homo sapiens sapiens*. They have noted that in certain sites there are continuous sequences showing a gradual transition in cultural remains from Neanderthal man to modern man. They however feel that the gradual increase in the number of specialized tools during the period indicates more efficient technology which developed by the end of the Neanderthal period. This must have reduced the selective pressures which favored large incisors and canines which were used for holding objects. Brace (1964) has also noted the tremendous amount of wear on the Neanderthal's teeth which he attributes to the use of teeth for holding objects. Binford (1968), however, suggests that differences in demographic patterns and the size of the breeding population must have been a major factor in the disappearance of Neanderthal man.

Phylogenetic Implications

As regards the phylogenetic implications there are two clear cut opinions:

1. That Progressive Neanderthals represent the Conservative type in the process of evolving into *Homo sapiens*.
2. That the Palestinians are hybrids between Neanderthal man and some variety of *Homo sapiens*.

These opinions indicate that Neanderthal man is our direct ancestor. However, there are two principal objections to this view:

1. Neanderthal man shows specialized traits which indicate an early and wide divergence from the main line of human evolution that leads to modern man.
2. Completely evolved modern types of fossil men were contemporary of Neanderthal man in Western Europe and hence they cannot be the descendants.

Recent discoveries of a whole series of human fossil finds exhibit in their teeth and skeletal characters the piecemeal replacement of Neanderthaloid specialization by modern morphology. The question of "Taurodontism" has also been invalidated by Senyurck (in Korn and Smith 1959) who has shown that molar teeth and enlarged pulp cavities are sporadically distributed throughout the human and infra-

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human Primates (both fossils and modern). According to Senyurck this may be a primitive and generalized feature rather than a specialization and thus this trait was not peculiar to Neanderthal man.

As regards the hybridity of the ancient Mount Carmelites, it has been argued that such a complete range of human evolution from Conservative type of Homo sapiens would not have been possible in such a short series and brief time and space. Hooton however feels that Neanderthal man should have changed into modern man by radical race mixture. The skeletal series from the caves of Skuhl and Tibun in Palestine demonstrate this to some extent. In his estimation some individuals, particularly the females, tend to reproduce almost in the Conservative parental type, while the males usually vary towards the Progressive parental stock.

Ever since the discovery of Neanderthal man in the year 1856, his precise evolutionary position has been a source of intense controversy and this discussion still continues. The most significant points are their supposed sudden disappearance from the fossil record, their origins and their relationship to modern man (Homo sapiens sapiens). The Classic Neanderthals were first reported from European sites and have been associated with Mousterian culture. Their sudden disappearance has been variously attributed to epidemics, conflicts with more advanced Homo sapiens, changes in the climate or their possible assimilation in the gene pool of the incoming migrants. In case they really disappeared perhaps there cannot be a single cause but a combination of circumstances seems to be responsible. However, the idea of disappearance has been questioned by Brace (1964). Now there are several hundred fossil specimens of Neanderthal man from Europe and the near East. In recent years there has been an enormous addition to the samples known from the Classic sites.

In general terms, the phylogeny of Neanderthal man may be summarized in three ways using three hypotheses. The first two views are widely considered valid but the third view is less well supported.

1. Neanderthal phase of man hypothesis
2. Pre-Neanderthal hypothesis
3. Pre-sapiens hypothesis

The Neanderthal Phase of Man Hypothesis: This is considered a unilinear view. According to this hypothesis, Neanderthals arose from middle Pleistocene predecessors by successive evolution. They passed through the Neanderthal phase and finally evolved into the modern man. This suggestion was first made by Schwalbe (1904). He believed that Neanderthals were a separate species, intermediate between ape and man. Later on Brace (1964) has suggested that the dental and masticatory evolutionary changes were brought out by the use of tools which brought in cranial morphological changes from Neanderthal to modern sapient forms. This hypothesis has been duly supported by Brose and Wolpoff (1971), Wolpoff (1980), Frayer (1978, 1984) and Smith and Ranyard (1980), Smith (1982, 1984), while working on the Central and East European material also observed local continuity and change between Neanderthal and later modern sapiens.

Pre-Neanderthal Hypothesis: This view holds that the Neanderthals arose from a pre-Neanderthal stock that progressively specialized towards cold resistance and was open to severe natural selection. The restricted gene flow resulted into a Classic Neanderthal isolate represented by La Chapelle Aux Saints and many others. This specialized Neanderthal offshoot represents a group sharing new traits of sub-specific taxonomic value. According to them, the parent line may have developed out of Europe and Africa as a possible source for the origin of pre-Neanderthal line as early *Homo sapiens*. The adherents to this view include Howell, Santa Luca, Stringer, Le Gros Clark etc.

Pre-Sapien Hypothesis: According to this view a European modern sapient lineage represented by Swanscombe and Steinheim existed separately from the Neanderthals, and ultimately gave rise to modern Europeans. Neanderthals, however, became extinct at the end of the early Wurm glaciation. This hypothesis was initially given by Boule (1911, 1913, and 1923). Later followers include Vallois (1954), Weiner (1958), Thoma (1965), Leakey (1972), Vecek (1978) and Saban (1982). The studies of Stringer

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(1974), Hublin (1982), Brauer (1984), Smith (1984), etc. however, proved that Swanscombe and Steinheim skulls did possess Neanderthal traits and were not anatomically modern. Therefore, it is felt that they cannot be considered as a separate lineage.

(D) Rhodesian Man

In June 1921, T. Zwigelaar, a Miner, found at Broke Hill in Northern Rhodesia, Africa, an almost complete cranium at about 60 feet below the ground level and about 10 feet above the floor of the cave. A human leg bone was also found at a distance of 3 feet from the cranium. More skeletal material was recovered by A. S. Armstrong in 1921 and later by H.Hrdlicka in 1925. The skeletal remains included a cranium, a left tibia, parts of the left femur, a sacrum, portions of pelvis and parts of the upper jaw of another individual. Thus, the skeletal remains represent cranial and post-cranial bones of atleast two individuals. On the basis of faunal and archaeological evidence as well as the Lead and Zinc content analysis of bones, it is concluded that Rhodesian man lived during early Pleistocene or late Pliocene. This approximates to the Solo man of Java with whom the present fossil material exhibits remarkable morphological similarities.

A glance at this skull indicates enormous true brow ridges and a low receding forehead, which are ape-like features. Rhodesian man has chimpanzee form of brow ridges which approach the gorilla type condition in their lateral extension and median development. The brow ridges are, in fact, much larger than in any other fossil man. The forehead is much lower and narrower than in Neanderthal man. However, it displays more vaulting than *Pithecanthropus*. The brain case is primitive but of human shape. It is very long and relatively narrow. The great length may be attributed to the forward projection of brow ridges and backward projection of occipital crest. The actual brain cavity length as worked out by Keith is only 17.1 cc i.e., 81.4 percent of the total skull length (this brain cavity length is 92 per cent of skull length in Europeans, 88.5 per cent in Australian aborigines, 86.0 percent in Neanderthal man, 84.0 percent in Pithecanthropus, 81.0 percent in chimpanzee and 75.0 percent in gorilla). Thus, in this respect Rhodesian man is closer to apes and inferior even to *Pithecanthropus erectus*. The Rhodesian skull, however, is low vaulted but exhibits human expansion of the parietal region. According to Keith, this parietal elevation overlies the part of brain controlling movement of the limbs and also indicates upright posture. The cranial capacity varies between 1280 and 1400 c.c. The skull is dolicocranial (cranial index 69.3). The foramen magnum is anteriorly placed and occupies a central position indicating perfect equilibrium of the skull on the vertebral column. The mastoid processes are of modern size. The greatest breadth of the skull is found near the base. Face is very large with cheek bones and strongly developed alveolar prognathism. The alveolar processes are very deep and the palate larger, wider and deeper than modern man's.

The teeth exhibit modern human proportions. The canines are reduced to the level of other teeth. The molars have remarkable breadth in comparison to their length. The third molars are reduced in size and degenerate. In the opinion of Keith, third molars show reduction even in chimpanzee. In Rhodesian man the pulp cavities of the teeth are not enlarged. In general the teeth are heavily affected by decay or caries. Alveolar abscesses at the roots of the molars and other teeth are noted. In the knee joint there are rheumatic changes which are believed to be due to the infection of the mouth. In addition to this there is a peculiar perforation (quarter of an inch in diameter) in front of the left ear hole and a larger eroded hole behind the ear passage. These perforations are connected by a curved tract of roughened and pitted bone. Yearsley feels that these lesions might be due to chronic septic conditions of the mouth which may have led to middle ear disease and complicated by mastoid abscess. In his view the hole in front of the ear may be due to a wound inflicted by some sharp implement during life. On the basis of his studies on these perforations, Mollson feels that these holes were made by the canine teeth of a large carnivore. Weidenreich, however, does not feel convinced. In his view the mastoid abscess theory is more plausible.

The limb skeleton include sacrum, pelvis and leg bones of the lower extremity. They are quite human with no ape-like features. They are, however, large and very robust with massive joints. Femur is straight like man and not bowed as in Neanderthals. Keith infers that Rhodesian man must have been a heavy and powerful fellow weighing about 208 lbs. He had an erect gait and was capable of extending the leg fully

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upon the thigh while walking. His stature works out to 5 feet and 10 inches. The sacrum is narrow and straight. The size and muscular markings of the bones suggest that these belong to a male individual. Thus, the skull and the limb skeleton which were separately recovered from two places are not fully compatible. Hrdlicka, after careful examination of the site and the find, feels that the limb bones do not belong to the owner of the skull. These belong to two individuals; one male and the other female. Radiometric and chemical analysis however suggest that they belong to contemporary individuals (Oakley 1950). Assuming that the skull and the limb skeleton belong to Rhodesian man (even to two different individuals), it can be said that he was an upright bipedal walker with primitive features of the skull.

Phylogenetic Implications

Woodward (1921) on the basis of the position of foramen magnum considers it to be human and assigns these to *Homo rhodesiensis*. Pycraft (1928), however, feels that he did not have erect posture as is indicated by the pelvis and assigned it to *Cyphanthropus rhodesiensis*. This contention however has not been accepted. Von Bonian feels that the skull, pelvis, femur and tibia indicate erect posture and places the Rhodesian man near the point of divergence of the Neanderthal and the modern man. Morant (1928), on the basis of metric study and comparisons, feels that Rhodesian man tends to resemble *Homo sapiens*, though it seems more closely related to Neanderthal man. In the opinion of Boule, Neanderthal man, Rhodesian man and modern Australians had a common origin. Rhodesian man may be the African variety of Neanderthals. In the opinion of Hooton there is sufficient likeness between Neanderthal and Rhodesian man so as to regard the latter as a variant of the former. In certain features Rhodesian man is inferior to Neanderthals and even to Pithecanthropus.

(E) Homo sapiens

CRO-MAGNON MAN

In 1868, M. Louis Lartet excavated the rock shelter of Cro-Magnon on the banks of Vezere in the village of Les Eyzies. This site has yielded several human skeletons. There were strata containing hearths and implements of the Aurignacian culture. The human remains were recovered from the highest strata of the deposit. This material consisted of a skull and some other bones of an old man and parts of the skeletons of four other individuals at a short distance from the old man's bones. These skeletons indicate the type of man that seems to have inhabited various places in Western Europe throughout the three upper Paleolithic periods. These skeletons exhibit features quite similar to modern man. It is proposed to consider the bones of the old man of Cro-Magnon as the type of specimen of the group.

The skull is massive and large in every dimension. Its length is 20.3 cms and breadth 15.0 cms. The height above the ear holes works out to 13.2 cms. The skull is Dolicocranial with an index of 73.7. It exhibits a pentagonal contour possibly owing to the marked projection of parietal mass. Some anthropologists refer to it as Dolico-pentagonal form. On the basis of the dimensions of skull the cranial capacity works out to 1660 c.c. This is about 150 cc, more than the modern European average. The forehead is broad and of moderate height and so also are the brow ridges. The occiput bulges behind. The skull is broad across the middle of the parietals. The face is relatively flat and very broad while the skull is narrow and long. This combination of a short broad face with a long narrow head is known as cranial disharmony. The orbits are extremely broad without rectilinear margin forming a quadrilateral outline. Owing to shortness of the face the orbits are very low. The cheek bones are large and protruding, nose is narrow, long and high (i.e., Leptorrhine). The maxilla exhibits pronounced prognathism. The alveolar border is prognathus. The palatine arch is that of medium size and narrow. The teeth are not extraordinary. The lower jaw is robust and that of modern conformation. The chin is prominent. The long bones indicate a stature of slightly over 5 feet 6 inches. Forearms are long in comparison to upper arms. The shins are longer in relation to thighs. The proportions of the limbs are referred to as Negroid proportion and they are not usual in modern Europeans. However, they have been noted frequently in the taller groups of the American Indians and in other races not considered Negroid. Another important

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feature is a type of flattening of the thigh bones called Platymeria and also side to side flattening of the shin called Platynemia. This is attributed to the habit of walking with the knees bent on unleveled ground. The muscular impressions are strongly marked.

Skeletons attributed to Cro-Magnon have been reported from Solutre, four miles West of Macon in East Central France. This is the typical site for the Solutrean period of the Palaeolithic age. On the basis of the original old man of Cro-Magnon, the French anthropologists refer to a race called "Cro-Magnon race" and attempted to fit into the Cro-Magnon type almost all finds of modern man belonging to late Paleolithic period. However, in this are included certain long headed but broad faced type of men who are believed to inhabit Dordogne, France and are believed to be the direct lineal descendents of ancient Cro-Magnon cave dwellers. Hooton, however, disagrees because the type referred to is very short in stature whereas the Paleolithic men were supposedly of great height. Quatrefages and Hamy referred to the existence of Cro-Magnon descendents in Dalecarlia, southern Sweden who form a special group known as `Dal Race'. This type has also been subsequently reported from various regions of Germany. Many individuals from these areas display a series of features corresponding to those found in descendents of Cro-Magnon race. Broca has noted the presence of structural affinities among the Basques, the Kabyles and the Guanchos (inhabitants of Canary Islands on the western coast of Africa). Denikar refers to the Cro-Magnon type as dark Mesocephalic, tall race called Atlanto-Mediterranean, which currently may be observed along the Atlantic and Mediterranean seacoasts of the Iberian Peninsula. However, the Guanchos of Canary Islands best represent the Cro-Magnon type. Thus, in general, the morphological characteristics of Cro-Magnon man suggest that he belongs to *Homo sapiens* with several distinctive characteristics. Many scholars believe that European upper Paleolithic man exhibited a wider range of skeletal variation in their form as represented by the remains of Chancelade, Grimaldi, Predmost, Combe-capelle, etc.

GRIMALDI MAN

Grimaldi fossil remains are referred to European Upper Paleolithic. The discovery was made in a cave near Mentone in Italy. In the Italian Riviera, just across the French frontier, there are red rocks of Grimaldi on the sea coast. On the cliffs of these rocks there are many caves where upper Paleolithic man lived. In one of the caves, called Grotte des Enfants, two skeletons were found in June 1901. Professor Verneau considered these skeletons as of Negroid type. The name of the cave seems to have been given on the basis of the skeletons of two children found in its upper strata. The excavation brought to light many habitation sites which were marked by hearths, implements and other evidences of human occupation. On the basis of faunal evidence (reindeer bones) they are referred to Pleistocene age. At the level of the second hearth from the top was located the skeleton of an old woman and at a depth of 9 feet were found the skeleton of a tall man. Verneau examined the skeleton of the tall man and assigned it to the Cro-Magnon type. The stature is estimated at 6 feet and 2-1/2 inches. The skeleton of this man was found lying on its back with a slab of red clay under his head and large stones around his feet. The associated implements refer to the Aurignacian culture.

At a depth of about 29 feet and at the level of the oldest hearth was found a grave containing two skeletons, belonging to an old woman and a boy of about 15 years. The old woman was buried with her arms doubled up under her chin and her knees flexed against her abdomen. The position of the skeleton suggests that she was lying on her right side. The boy's skeleton was slightly above and on to her right. The child's skeleton was in a loosely flexed posture with arms half bent and doubled up legs at right angles to the axis of the spine. The skeletons were stained with red ochre and the implements around the burial were of Aurignacian culture. It seems that they refer to the beginning of the Aurignacian culture.

On the basis of cranial morphology, the Grimaldi skulls are believed to have Negroid features. Both the skulls are long, narrow and high. Length of the female skull is 19.1 cms, breadth 13.1 cms and height 11.5 cms. The brow ridges are feebly developed as in the Negroid crania. The forehead is straight and well developed and slightly bulging. The skulls present a regular elliptically shaped contour with flattened parietal eminences. The cranial indices of old woman and of the adolescent boy are 68.5 and 69.2 respectively. The cranial capacity according to Keith works out to 1454 c.c. in case of the young boy and

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1265 c.c. in case of old woman. The face in both the cases is rather narrow and short. However, it must be noted that the boy was in his adolescence with jaws not fully developed. The orbits are very low in comparison to their breadth as in Cro-Magnon. Nose is depressed at the root, the bridge is low and broad and nasal aperture extremely wide. The nasal index in case of female is 63.6 and in case of boy 54.3, suggesting that the nose is platyrrhine. The alveolar margins in both the skulls are swollen and prognathus. The lower jaw is strong with its body very thick. The chin is not greatly developed. A strongly marked prognathism correlated with the prognathism of the upper jaw gives it a receding appearance. Most of these cranial and facial features are Negroid.

Grimaldi skeletons exhibit Negroid features in the proportion of limbs. The forearm and the leg are very long in relation to the upper arm and thigh respectively. The shaft of the femur has a pronounced curvature and is strongly bent. The estimated stature of the woman works out to 5 feet 3 inches and that of the boy 5 feet 1-1/2 inches.

The Grimaldi material is believed by many anthropologists as definitely Negroid and they are commonly accepted as a good evidence of Negroid race in Europe during Aurignacian times. However, on the basis of the actual study of the material many others disagree with this view and feel that they are simply variants of the Mediterranean race now inhabiting Southern Europe. Boule and Vallois maintain that the Grimaldi skeletons represent a human type which is comparable to the modern Negritic or Negroid type. According to them, the Grimaldi exhibit resemblances to South African types, the Bushmen and Hottentots, in terms of similar Dolicocephalic characters, same prognathism, same flattening of the nose, and same development of facial breadth, similar form of jaw and the great size of teeth. The differences, however, are in terms of the stature and height of the skull.

In the opinion of Hooton, the so called Negroid features of the Grimaldi merely constitute a part of the primitive complex of the so called Cro-Magnon man. The efforts to identify the Grimaldi material with Bushmen of South Africa and Hottentots are interesting but not altogether convincing. The common possession of some of the Negroid characters in the Grimaldi, together with similar Aurignacian stone industry with that of the Bushmen perhaps is the basis for the observation that Bushmen are descendents of upper Paleolithic artists of Europe. Elliot Smith and Arthur Keith, on the basis of their independent research work on the Grimaldi skeleton, have concluded that the Grimaldi merely represent the primitive type of Cro-Magnon stock and the Negroid resemblances may be just a coincidence.

CHANCELADE MAN

The name is derived from a rock shelter near Chancelade where the discovery was made on October 1, 1888. Chancelade is 4 miles from Perigueux and lies in the centre of the Dordogne region of South Central France. The deposits on the floor of the rock shelter were 5 feet 4 inches in depth and yielded the typical fauna and implements belonging to the Magdalenian period.

The skeleton lay on its left side in the deepest stratum with its arms folded on its breasts and knees doubled up against the body. The body was powdered over with red ochre (this type of burial is practiced by Eskimos). The skeleton is believed to belong to a man between 55 and 65 years of age. The stature was short. It was 5 feet 2 inches. The limb bones were very robust and fragmentary.

The skull of the Chancelade man resembles the crania of modern Eskimos. It is long and narrow (length 19.4 cms and breadth 13.5 cms) with a cranial index of 70.9. Supra orbital ridges are slightly developed with forehead bulging and rising vertically. The cranial capacity as estimated by Keith is 1530 c.c. The occiput is steep behind as in Brachycephals.

Face is long and moderately broad. The cheek bones are prominent and strongly developed, which give a flat appearance to the face. The nose is long and narrow and the nasal bridge is broken. However, on the basis of the photograph of the skull Keith claims that it is high and long and at an angle never seen in Eskimo crania or in typical Mongoloid faces. The jaws do not flare outward in Eskimos. Most of the upper teeth have been lost from wear and disease. The lower teeth are not very large, molars seem to be

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powerful and increase in size from first to the third. In modern man the third molar is smaller than the other two.

The post-cranial skeleton indicates that long bones are strong and massive. The muscular impressions are well marked indicating a muscular body. The upper limbs are longer unlike modern Europeans. Femur is slightly bent. The shaft of the tibia has a flattening in the transverse direction and is slightly platynemic. Foot is large, with first metatarsal distinctly separated from second toe, like in Neanderthal man.

Testup, who examined the skeleton, has shown its resemblances with Eskimos. These resemblances are based on the combination of short stature with a conspicuous dolicocranial skull, a fairly long and broad face, narrow nasal aperture, strongly developed masticatory apparatus, flat face marked by squared cheek bones, and presence of a medial ridge like elevation of the skull roof. On the basis of these similarities and cultural similarities like the use of bone tools, needles, harpoons and bone engravings, etc., it has been claimed that the Magdalenians, following the retreat of the ice sheet northwards at the close of glacial age, eventually reached North America and thus are the ancestors of modern Eskimos. However, this view has not been fully accepted. Hooton argues that the resemblances exist but these are like many Eskimo features found in the skulls of twelfth century Icelanders of Norwegian and Irish origin. However, the possibility of a Mongoloid admixture in the man of Chancelade cannot be denied. Keith, on the other hand, has suggested that Chancelade is a European type. Many scholars are of the view that it is another variety of European upper Paleolithic man - like Cro-Magnon - but of much lower stature.

1.7 THE BIOLOGICAL BASIS OF LIFE

THE CELL

All organisms are composed of cells. Some are composed of a single cell and are called unicellular organisms while others, like us, composed of many cells, are called multicellular organisms.

What is a Cell?

Unicellular organisms are capable of (i) independent existence and (ii) performing the essential functions of life. Anything less than a complete structure of a cell does not ensure independent living. Hence, cell is the fundamental structural and functional unit of all living organisms. Anton Von Leeuwenhoek first saw and described a live cell. Robert Brown later discovered the nucleus. The invention of the microscope and its improvement leading to the electron microscope revealed all the structural details of the cell.

Cell Theory

In 1838, Malthias Schleiden, a German botanist, examined a large number of plants and observed that all plants are composed of different kinds of cells which form the tissues of the plant. At about the same time, Theodore Schwann (1839), a British Zoologist, studied different types of animal cells and reported that cells had a thin outer layer that is today known as the 'plasma membrane'. He also concluded, based on his studies on plant tissues, that the presence of cell wall is a unique character of the plant cells. On the basis of this, Schwann proposed the hypothesis that the bodies of animals and plants are composed of cells and products of cells. Schleiden and Schwann together formulated the cell theory. This theory however, did not explain as to how new cells were formed. Rudolf Virchow (1855) first explained that cells divided and new cells are formed from pre-existing cells (Omnis cellulae cellula). He modified the hypothesis of Schleiden and Schwann to give the cell theory a final shape. Cell theory as understood today is:

(i) All living organisms are composed of cells and products of cells.

(ii) All cells arise from pre-existing cells.

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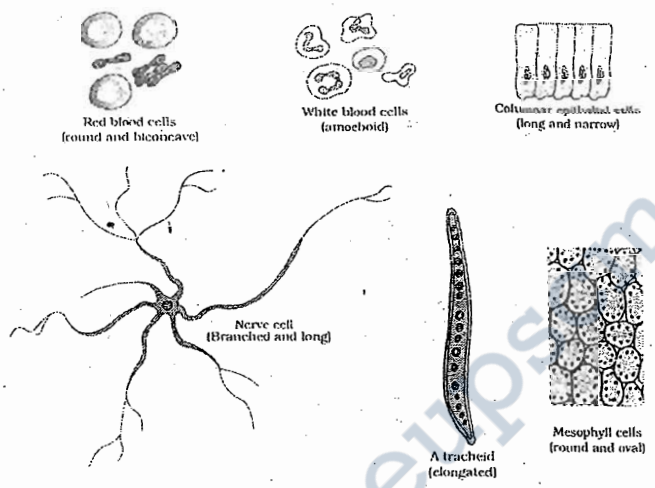
Cell – An Overview

A typical plant cell, has a distinct cell wall as its outer boundary and just within it is the cell membrane. The cells of the human cheek have an outer membrane as the delimiting structure of the cell. Inside each cell is a dense membrane bound structure called nucleus. This nucleus contains the chromosomes which in turn contain the genetic material, DNA. Cells that have membrane bound nuclei are called eukaryotic whereas cells that lack a membrane bound nucleus are prokaryotic. In both prokaryotic and eukaryotic cells, a semi-fluid matrix called cytoplasm occupies the volume of the cell. The cytoplasm is the main arena of cellular activities in both the plant and animal cells. Various chemical reactions occur in it to keep the cell in the 'living state'.

Besides the nucleus, the eukaryotic cells have other membrane bound distinct structures called organelles like the endoplasmic reticulum (ER), the golgi complex, lysosomes, mitochondria, microbodies and vacuoles. The prokaryotic cells lack such membrane bound organelles.

Ribosomes are non-membrane bound organelles found in all cells – both eukaryotic as well as prokaryotic. Within the cell, ribosomes are found not only in the cytoplasm but also within the two organelles – chloroplasts (in plants) and mitochondria and on rough ER.

Animal cells contain another non-membrane bound organelle called centriole which helps in cell division.



Cells differ greatly in size, shape and activities. For example, Mycoplasmas, the smallest cells, are only 0.3 μm in length while bacteria could be 3 to 5 μm. The largest isolated single cell is the egg of an ostrich. Among multicellular organisms, human red blood cells are about 7.0 μm in diameter. Nerve cells are some of the longest cells. Cells also vary greatly in their shape. They may be disc-like, polygonal, columnar, cuboid, thread like, or even irregular. The shape of the cell may vary with the function they perform.

Typical bacteria (1-2 m)

PPLO (about 0.1 m)

Viruses (0.02-0.2 m)

Prokaryotic Cells

The prokaryotic cells are represented by bacteria, blue-green algae, mycoplasma and PPLO (Pleuro Pneumonia Like Organisms). They are generally smaller and multiply more rapidly than the eukaryotic cells. They may vary greatly in shape and size. The four basic shapes of bacteria are bacillus (rod like), coccus (spherical), vibrio (comma shaped) and spirillum (spiral). The organization of the prokaryotic cell is fundamentally similar even though prokaryotes exhibit a wide variety of shapes and functions. All prokaryotes have a cell wall surrounding the cell membrane. The fluid matrix filling the cell is the cytoplasm. There is no well-defined nucleus. The genetic material is basically naked, not enveloped by a nuclear membrane. In addition to the genomic DNA (the single chromosome/circular DNA), many bacteria have small circular DNA outside the genomic DNA. These smaller DNA are called plasmids. The plasmid DNA confers certain unique phenotypic characters to such bacteria. One such character is resistance to antibiotics. In higher classes you will learn that this plasmid DNA is used to monitor bacterial transformation with foreign DNA. Nuclear membrane

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is found in eukaryotes. No organelles, like the ones in eukaryotes, are found in prokaryotic cells except for ribosomes. Prokaryotes have something unique in the form of inclusions. A specialized differentiated form of cell membrane called mesosome is the characteristic of prokaryotes. They are essentially infoldings of cell membrane.

Cell Envelop

Most prokaryotic cells, particularly the bacterial cells, have a chemically complex cell envelope. The cell envelope consists of a tightly bound three layered structure i.e., the outermost glycocalyx followed by the cell wall and then the plasma membrane. Although each layer of the envelope performs distinct function, they act together as a single protective unit. Bacteria can be classified into two groups on the basis of the differences in the cell envelopes and the manner in which they respond to the staining procedure developed by Gram viz., those that take up the gram stain are **Gram positive** and the others that do not are called **Gram negative** bacteria.

Glycocalyx differs in composition and thickness among different bacteria..It could be a loose sheath called the **slime layer** in some, while in others it may be thick and tough, called the **capsule**. The **cell wall** determines the shape of the cell and provides a strong structural support to prevent the bacterium from bursting or collapsing.

The plasma membrane is semi-permeable in nature and interacts with the outside world. This membrane is similar structurally to that of the eukaryotes.

A special membranous structure is the **mesosome**, which is formed by the extensions of plasma membrane into the cell. These extensions are in the form of **vesicles**, **tubules** and **lamellae**. They help in cell wall formation, DNA replication and distribution to daughter cells. They also help in respiration, secretion processes, to increase the surface area of the plasma membrane and enzymatic content. In some prokaryotes like cyanobacteria, there are other membranous extensions into the cytoplasm called chromatophores that contain pigments.

Bacterial cells may be motile or non-motile. If motile, they have thin filamentous extensions from their cell wall called flagella. Bacteria show a range in the number and arrangement of flagella. Bacterial flagellum is composed of three parts – **filament**, **hook** and **basal body**. The filament is the longest portion and extends from the cell surface to the outside.

Besides flagella, Pili and Fimbriae are also surface structures of the bacteria but do not play a role in motility. The **pili** are elongated tubular structures made of a special protein. The **fimbriae** are small bristle like fibres sprouting out of the cell. In some bacteria, they are known to help attach the bacteria to rocks in streams and also to the host tissues.

Ribosomes and Inclusion Bodies

In prokaryotes ribosomes are associated with the plasma membrane of the cell. They are about 15 nm by 20 nm in size and are made of two subunits - 50S and 30S units which when present together form 70S prokaryotic ribosomes. Ribosomes are the site of protein synthesis. Several ribosomes may attach to a single mRNA and form a chain called **polyribosomes** or **polysome**. The ribosomes of a polysome translate the mRNA into proteins.

Inclusion bodies: Reserve material in prokaryotic cells is stored in the cytoplasm in the form of inclusion bodies. These are not bounded by any membrane system and lie free in the cytoplasm, e.g., phosphate granules, cyanophycean granules and glycogen granules. Gas vacuoles are found in blue green and purple and green photosynthetic bacteria.

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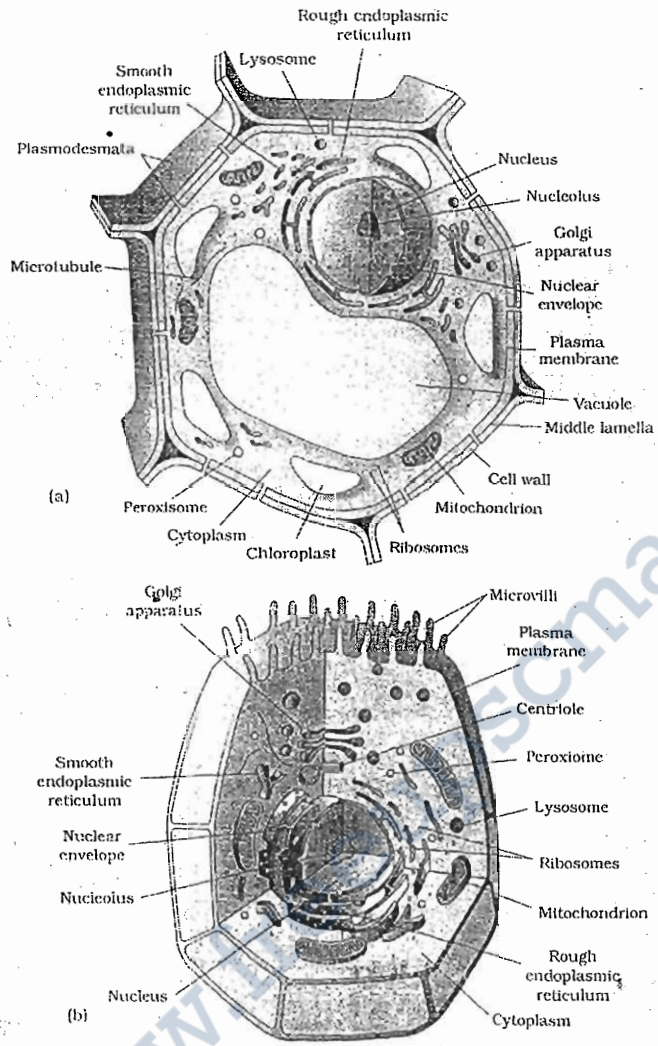
Eukaryotic Cells

The eukaryotes include all the protists, plants, animals and fungi. In eukaryotic cells there is an extensive compartmentalization of cytoplasm through the presence of membrane bound organelles. Eukaryotic cells possess an organized nucleus with a nuclear envelope. In addition, eukaryotic cells have a variety of complex locomotory and cytoskeletal structures. Their genetic material is organized into chromosomes. All eukaryotic cells are not identical. Plant and animal cells are different as the former possess cell walls, plastids and a large central vacuole that are absent in animal cells..On the other hand, animal cells have centrioles that are absent in almost all plant cells.



Let us now look at individual cell organelles to understand their structure and functions.

Cell Membrane

The detailed structure of the membrane was studied only after the advent of the electron microscope in the 1950s. Meanwhile, chemical studies on the cell membrane, especially in human red blood cells (RBCs), enabled the scientists to deduce the possible structure of plasma membrane. The membrane possesses lipids, proteins and carbohydrates. According to the Fluid Mosaic Model given by Singer and Nicolson, the lipids molecules, which are in a quasi-fluid nature, enables lateral movement of protein molecules. The fluid nature of the membrane is also important from the point of view of functions like cell growth, formation of intercellular junctions, secretion, endocytosis, cell division etc. One of the most important functions of the plasma membrane is the transport of the molecules across it.

Cell Wall

A non-living rigid structure called the cell wall forms an outer covering for the plasma membrane of fungi and plants. Cell wall not only gives shape to the cell and protects the cell from mechanical damage and infection, it also helps in cell-to-cell interaction and provides barrier to undesirable macromolecules. Algae have cell wall, made of cellulose, galactans, mannans and minerals like calcium carbonate, while in other plants it consists of cellulose, hemicellulose, pectins and proteins. The cell wall of a young plant cell, the primary wall is capable of growth, which gradually diminishes as the cell matures and the secondary wall is formed on the inner (towards membrane) side of the cell. The middle lamella is a layer mainly of

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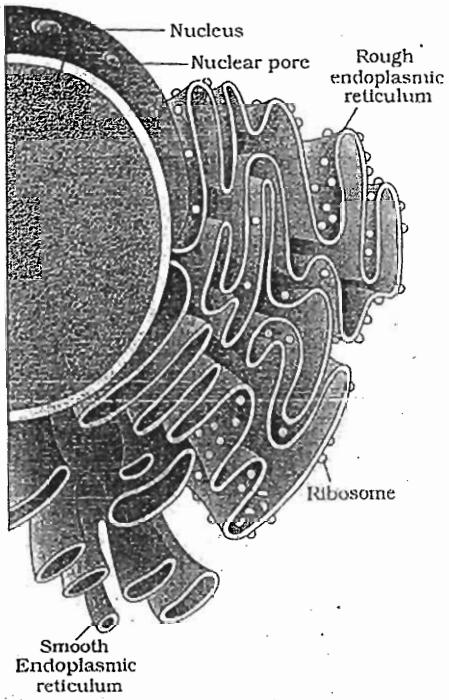
calcium pectate that holds or glues the different neighboring cells together. The cell wall and middle lamellae may be traversed by plasmodesmata which connect the cytoplasm of neighboring cells.

Endomembrane System



While each of the membranous organelles is distinct in terms of its structure and function, many of these are considered together as an endomembrane system because their functions are coordinated. The endomembrane system include endoplasmic reticulum (ER), golgi complex, lysosomes and vacuoles. Since the functions of the mitochondria, chloroplast and peroxisomes are not coordinated with the above components, these are not considered as part of the endomembrane system.

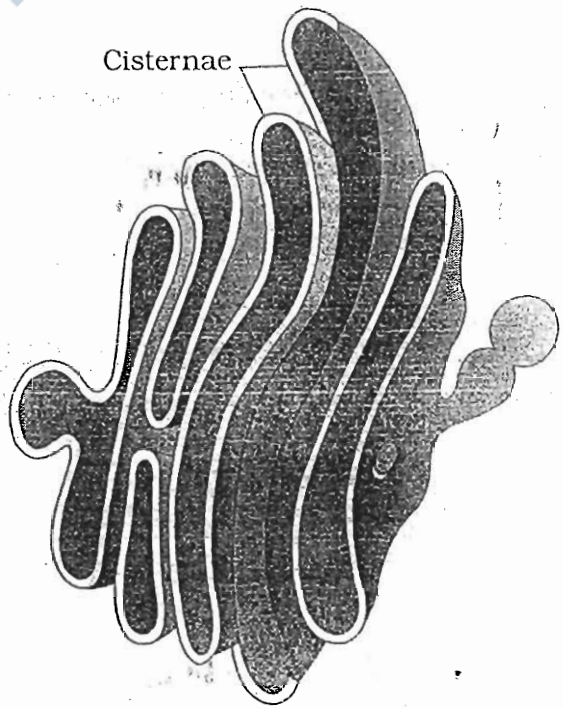
The Endoplasmic Reticulum (ER)

Electron microscopic studies of eukaryotic cells reveal the presence of a network or reticulum of tiny tubular structures scattered in the cytoplasm \that is called the endoplasmic reticulum (ER). Hence, ER divides the intracellular space into two distinct compartments, i.e., luminal (inside ER) and extra luminal (cytoplasm) compartments.

The ER often shows ribosomes attached to their outer surface. The endoplasmic reticulun bearing ribosomes on their surface is called rough endoplasmic reticulum (RER). In the absence of ribosomes they appear smooth and are called smooth endoplasmic reticulum (SER). RER is frequently observed in the cells actively involved in protein synthesis and secretion. They are extensive and continuous with the outer membrane of the nucleus. The smooth endoplasmic reticulum is the major site for synthesis of lipid. In animal cells lipid-like steroidal hormones are synthesised in SER.

Golgi Apparatus

Camillo Golgi (1898) first observed densely stained reticular structures near the nucleus. These were later named Golgi bodies after him. They consist of many flat, disc-shaped sacs or cisternae of 0.5μm to 1.0μm diameter (Figure 8.6). These are stacked parallel to each other. Varied number of cisternae is present in a Golgi complex. The Golgi cisternae are concentrically arranged near the nucleus with distinct convex cis or the forming face and concave trans or the maturing face.



The cis and the trans faces of the organelle are entirely different, but interconnected. The golgi apparatus principally performs the function of packaging materials, to be delivered either to the intra-cellular targets or secreted outside the cell. Materials to be packaged in the form of vesicles from the ER fuse with the cis face of the golgi apparatus and move towards the maturing face.

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This explains, why the golgi apparatus remains in close association with the endoplasmic reticulum. A number of proteins synthesized by ribosomes on the endoplasmic reticulum are modified in the cisternae of the golgi apparatus before they are released from its trans face. Golgi apparatus is the important site of formation of glycoproteins and glycolipids.

Lysosomes

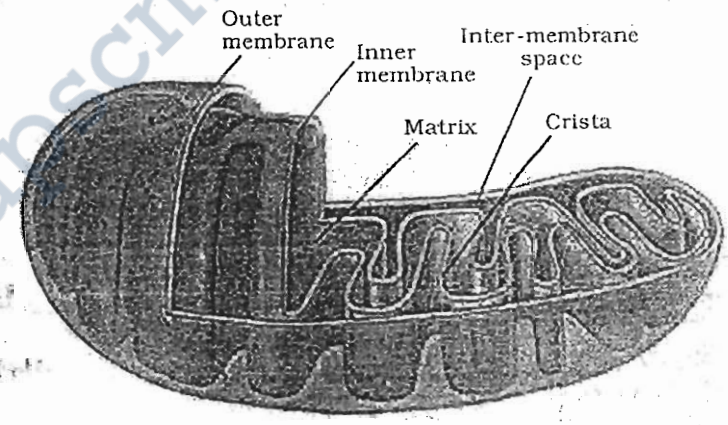
These are membrane bound vesicular structures formed by the process of packaging in the golgi apparatus. The isolated lysosomal vesicles have been found to be very rich in almost all types of hydrolytic enzymes (hydrolases – lipases, proteases, carbohydrases) optimally active at the acidic pH. These enzymes are capable of digesting carbohydrates, proteins, lipids and nucleic acids.

Vacuoles

The vacuole is the membrane-bound space found in the cytoplasm. It contains water, sap, excretory product and other materials not useful for the cell. The vacuole is bound by a single membrane called tonoplast. In plant cells the vacuoles can occupy up to 90 per cent of the volume of the cell. In plants, the tonoplast facilitates the transport of a number of ions and other materials against concentration gradients into the vacuole, hence their concentration is significantly higher in the vacuole than in the cytoplasm. In Amoeba the contractile vacuole is important for excretion. In many cells, as in protists, food vacuoles are formed by engulfing the food particles.

Mitochondria

Mitochondria (sing.: mitochondrion), unless specifically stained, are not easily visible under the microscope. The number of mitochondria per cell is variable depending on the physiological activity of the cells. In terms of shape and size also, considerable degree of variability is observed. Typically it is sausage-shaped or cylindrical having a diameter of 0.2-1.0μm (average 0.5μm) and length 1.0-4.1μm.



Each mitochondrion is a double membrane-bound structure with the outer membrane and the inner membrane dividing its lumen distinctly into two aqueous compartments, i.e., the outer compartment and the inner compartment. The inner compartment is called the matrix. The outer membrane forms the continuous limiting boundary of the organelle. The inner membrane forms a number of infoldings called the cristae (sing.: crista) towards the matrix. The cristae increase the surface area. The two membranes have their own specific enzymes associated with the mitochondrial function. Mitochondria are the sites of aerobic respiration. They produce cellular energy in the form of ATP, hence they are called 'power houses' of the cell. The matrix also possesses single circular DNA molecule, a few RNA molecules, ribosomes (70S) and the components required for the synthesis of proteins. The mitochondria divide by fission.

Plastids

Plastids are found in all plant cells and in euglenoides. These are easily observed under the microscope as they are large. They bear some specific pigments, thus imparting specific colours to the plants. Based on the type of pigments plastids can be classified into chloroplasts, chromoplasts and leucoplasts.

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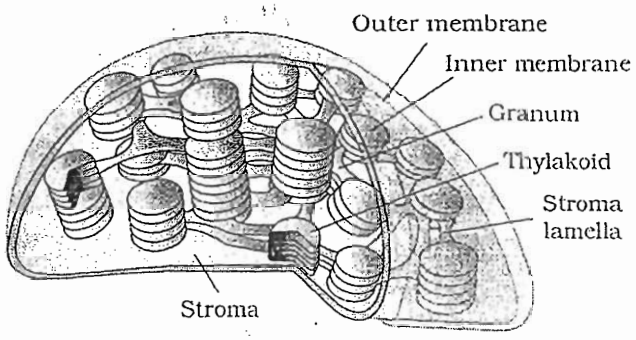
The chloroplasts contain chlorophyll and carotenoid pigments which are responsible for trapping light energy essential for photosynthesis. In the chromoplasts fat soluble carotenoid pigments like carotene, xanthophylls and others are present. This gives the part of the plant a yellow, orange or red colour. The leucoplasts are the colourless plastids of varied shapes and sizes with stored nutrients: Amyloplasts store carbohydrates (starch), e.g., potato; elaioplasts store oils and fats whereas the aleuroplasts store proteins.



Majority of the chloroplasts of the green plants are found in the mesophyll cells of the leaves. These are lens-shaped, oval, spherical, discoid or even ribbon-like organelles having variable length (5-10mm) and width (2-4mm). Their number varies from 1 per cell of the Chlamydomonas, a green alga to 20-40 per cell in the mesophyll.

Like mitochondria, the chloroplasts are also double membrane bound. Of the two, the inner chloroplast membrane is relatively less permeable. The space limited by the inner membrane of the chloroplast is called the stroma. A number of organised flattened membranous sacs called the thylakoids, are present in the stroma. Thylakoids are arranged in stacks like the piles of coins called grana (singular: granum) or the intergranal thylakoids. In addition, there are flat membranous tubules called the stroma lamellae connecting the thylakoids of the different grana. The membrane of the thylakoids encloses a space called a lumen. The stroma of the chloroplast contains enzymes required for the synthesis of carbohydrates and proteins. It also contains small, double-stranded circular DNA molecules and ribosomes. Chlorophyll pigments are present in the thylakoids. The ribosomes of the chloroplasts are smaller (70S) than the cytoplasmic ribosomes (80S).

Ribosomes

Ribosomes are the granular structures first observed under the electron microscope as dense particles by George Palade (1953). They are composed of ribonucleic acid (RNA) and proteins and are not surrounded by any membrane. The eukaryotic ribosomes are 80S while the prokaryotic ribosomes are 70S. Here 'S' stands for the sedimentation coefficient; it indirectly is a measure of density and size. Both 70S and 80S ribosomes are composed of two subunits.

Cytoskeleton

An elaborate network of filamentous proteinaceous structures present in the cytoplasm is collectively referred to as the cytoskeleton. The cytoskeleton in a cell are involved in many functions such as mechanical support, motility, maintenance of the shape of the cell.

Cilia and Flagella

Cilia (sing.: cilium) and flagella (sing.: flagellum) are hair-like outgrowths of the cell membrane. Cilia are small structures, which work like oars, causing the movement of either the cell or the surrounding fluid. Flagella are comparatively longer and responsible for cell movement. The prokaryotic bacteria also posses flagella but these are structurally different from that of the eukaryotic flagella.

Centrosome and Centriole

Centrosome is an organelle usually containing two cylindrical structures called centrioles. The centrioles form the basal body of cilia or flagella, and spindle fibres that give rise to spindle apparatus during cell division in animal cells.

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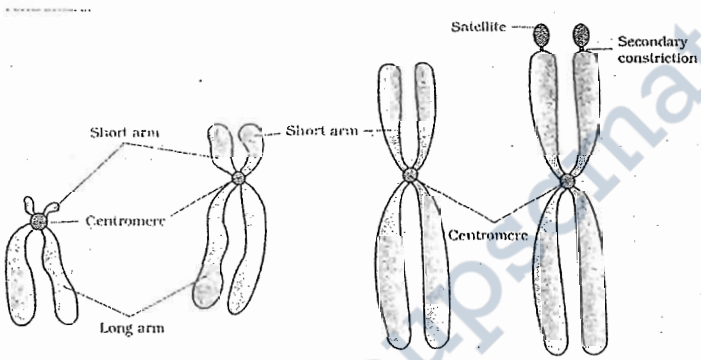
Nucleus

Nucleus as a cell organelle was first described by Robert Brown as early as 1831. Later the material of the nucleus stained by the basic dyes was given the name chromatin by Flemming. The interphase nucleus (nucleus of a cell when it is not dividing) has highly extended and elaborate nucleoprotein fibres called chromatin, nuclear matrix and one or more spherical bodies called nucleoli (sing.: nucleolus) Electron microscopy has revealed that the nuclear envelope, which consists of two parallel membranes with a space between (10 to 50 nm) called the perinuclear space, forms a barrier between the materials present inside the nucleus and that of the cytoplasm. The outer membrane usually remains continuous with the endoplasmic reticulum and also bears ribosomes on it. At a number of places the nuclear envelope is interrupted by minute pores, which are formed by the fusion of its two membranes. These nuclear pores are the passages through which movement of RNA and protein molecules takes place in both directions between the nucleus and the cytoplasm. The nuclear matrix or the nucleoplasm contains nucleolus and chromatin. The nucleoli are spherical structures present in the nucleoplasm. The content of nucleolus is continuous with the rest of the nucleoplasm as it is not a membrane bound structure. It is a site for active ribosomal RNA synthesis. Larger and more numerous nucleoli are present in cells actively carrying out protein synthesis.

During different stages of cell division, cells show structured chromosomes in place of the nucleus. Chromatin contains DNA and some basic proteins called histones, some non-histone proteins and also RNA. A single human cell has approximately two-meter long thread of DNA distributed among its forty six (twenty three pairs) chromosomes. Every chromosome essentially has a primary constriction or the centromere on the sides of which disc shaped structures called kinetochores are present.



Based on the position of the centromere, the chromosomes can be classified into four types. The metacentric chromosome has middle centromere forming two equal arms of the chromosome. The sub-metacentric chromosome has centromere nearer to one end of the chromosome resulting into one shorter arm and one longer arm. In case of acrocentric chromosome the centromere is situated close to its end forming one extremely short and one very long arm, whereas the telocentric chromosome has a terminal centromere.

Microbodies

Many membrane bound minute vesicles called microbodies that contain various enzymes, are present in both plant and animal cells.

DNA STRUCTURE

DNA, The Genetic Material

Several lines of indirect evidence have long suggested that DNA contains the genetic information of living organisms. Most important, results obtained using several different experimental procedures showed that most of the DNA is located in the chromosomes, whereas RNA and proteins are also abundant in the cytoplasm. Moreover, a precise correlation exists between the amount of DNA per cell and the number of sets of chromosomes per cells. That is, most somatic cells of diploid organisms, for example, contain exactly twice the amount of DNA as the haploid germ cells or gametes of the same species. Finally, the molecular composition of the DNA in all of the different cells of an organism is the same (with rare

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exceptions), whereas the composition of RNA and proteins varies both qualitatively and quantitatively from one cell type to another.

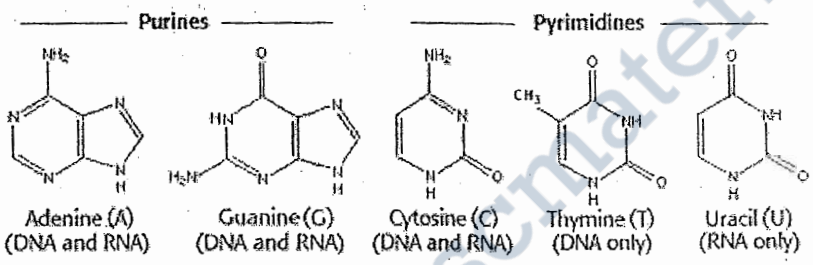
Although these correlations strongly suggest that DNA is the genetic material, they by no means prove it. Fortunately, direct evidence has established that the genetic information is encoded in DNA. Noteworthy of these experiments are those conducted by Frederick Griffith in 1928 and O.T. Avery, C.M. Macleod and M. McCarty in 1944. A.D. Hershey and M. Chase published additional direct evidence indicating that DNA is the genetic material in 1952.

Structure of DNA

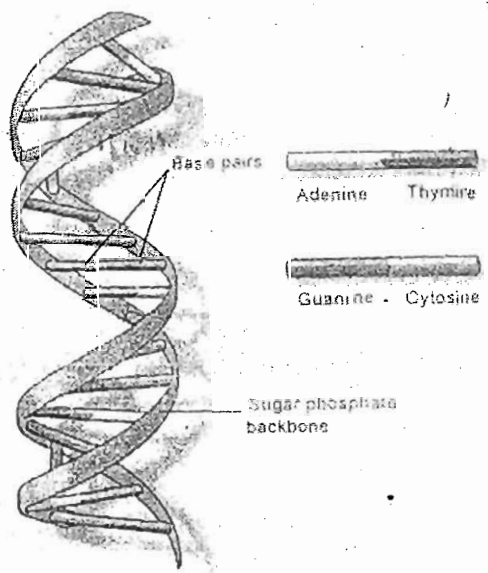
The genetic information of all living organisms, except the RNA Viruses, is stored in DN, and in what form is the genetic information stored? What features of the structure of DNA allow for the transmission of genetic information from generation to generation?

Nucleic Acids are macromolecules composed of repeating subunits called **nucleotides.** Each nucleotide is composed of

1. A Phosphate group
2. A five-carbon sugar (or pentose)
3. A cyclic nitrogen-containing compound called a base.



In DNA, the sugar is 2-deoxyribose (thus the name **deoxyribonucleic acid**); in RNA, the sugar is ribose (thus **ribo**nucleic acid). There are four different bases commonly found in DNA: **adenine, guanine, thymine,** and **cytosine.** RNA also usually contains adenine, guanine and cytosine, but has a different base, **uracil**, in the place of thymine. Adenine and guanine are double-ring bases called **purines**; *cytosine*, thymine and uracil are single-ring bases called **pyrimidines**. Both DNA and RNA, *therefore*, contain four different subunits or nucleotides, two purine nucleotides, and two pyrimidine nucleotides. RNA usually exists as a single stranded polymer that is composed of a long sequence of nucleotides. DNA, however, has one very important additional level of organization; it is usually a double-stranded molecule.



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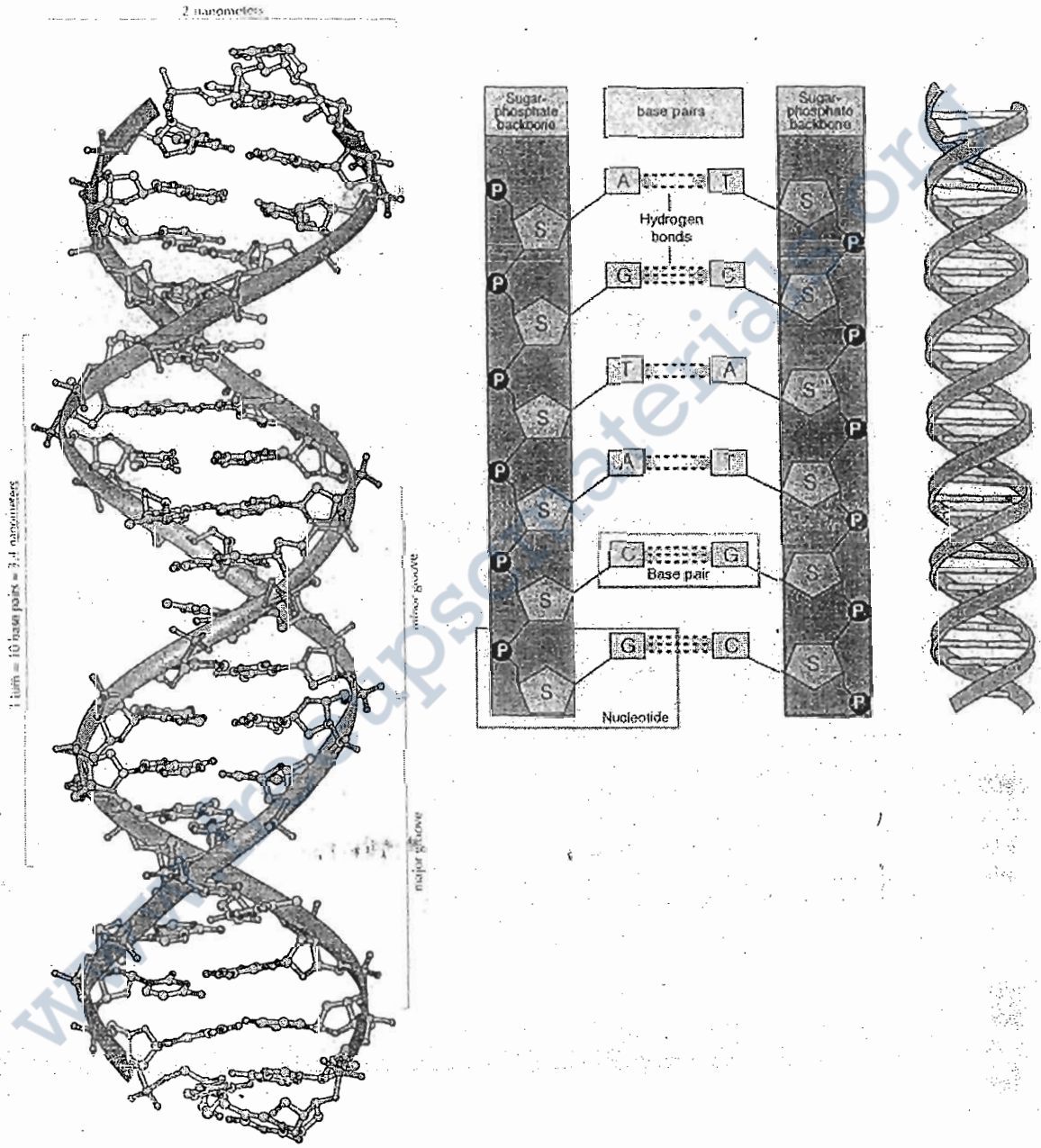
The Watson and Crick DNA Double Helix

The correct structure of DNA was first deduced by J.D. Watson and F.H.C. Crick in 1953. They proposed that DNA exists as a **double helix** in which the two polynucleotide chains are coiled about one another in a spiral. Each polynucleotide chain consits of a sequence of nucleotides linked together by **phosphodiester bonds,** joining adjacent deoxyribose moieties.



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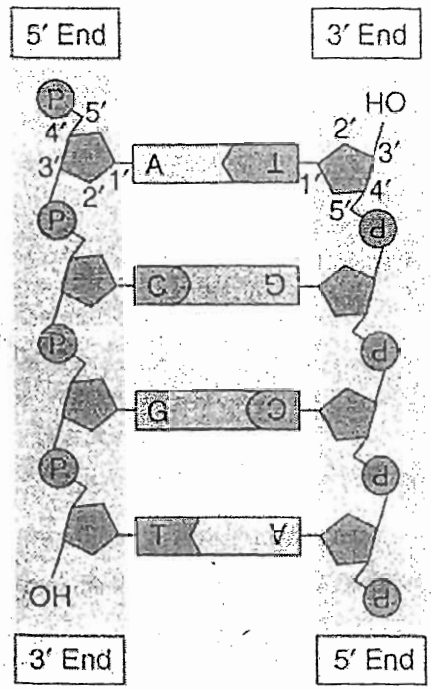
The two polynucleotide strands are held together in their helical configuration by **hydrogen bonding** between bases in opposing strands, the resulting base-pairs being stacked between the two chains perpendicular to the axis of the molecule like the steps of a spiral staircase. The base pairing is specific; **adenine is always paired with thymine, and guanine is always paired with cytosine.** Thus, all base-pairs consist of one purine and one pyrimidine. The specificity of base-pairing results from the hydrogen-bonding capacities of the bases in their normal configurations. In their most common structural configurations, adenine and thymine from two hydrogen bonds, and guanine and cytosine form three hydrogen bonds. Analogous hydrogen bonding between cytosine and adenine, for example, is not possible except when they exist in their rare structural states (when mutations occur).



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Once the sequence of bases in one strand of DNA double helix is known, the sequence of bases in the other strand is also known because of the specific base-pairing. The two strands of a DNA double helix are thus said to be **complementary (not identical).** It is this property, complementarity of two strands, that make DNA uniquely suited to sore and transmit genetic information (we shall discuss this in the next topic of DNA replication).

The base pairs in DNA are stacked 3.4 nanometers apart with 10 base-pairs per turn (360°) of the double helix. The sugar-phosphate backbones of the two complementary strands are **antiparallel;** that is they have **opposite chemical polarity.** As one moves unidirectionally along a DNA double helix, the phosphodiester bonds in one strand go from a 3′ carbon of one nucleotide to a 5′ carbon of the adjacent nucleotide, whereas those in the complementary strand go from a 5′ carbon to a 3′ carbon. This opposite polarity of the complementary strands is very important in considering the mechanism of replication of DNA.

DNA REPLICATION

Living organisms perpetuate their kind through reproduction. This may be simple duplication (cell fission) as in bacteria or complex modes of sexual reproduction as in higher plants and animals. IN all cases, however, reproduction entails the faithful transmission of the genetic information of the parents to the progeny. Since the genetic information in stored in the DNA, **the replication of DNA is central of all of biology.**

Significance of Specific Base Paring in DNA Replication

One question whose answer was immediately suggested by the double helix was the manner in which gene duplication occurs so precisely in cell division. Millions of cells undergo mitosis each day in complex organisms like humans, for the most part resulting in cells exactly identical to each other. The precision involved has long fascinated biologists. We now know this precision is the function of nitrogenous bases.

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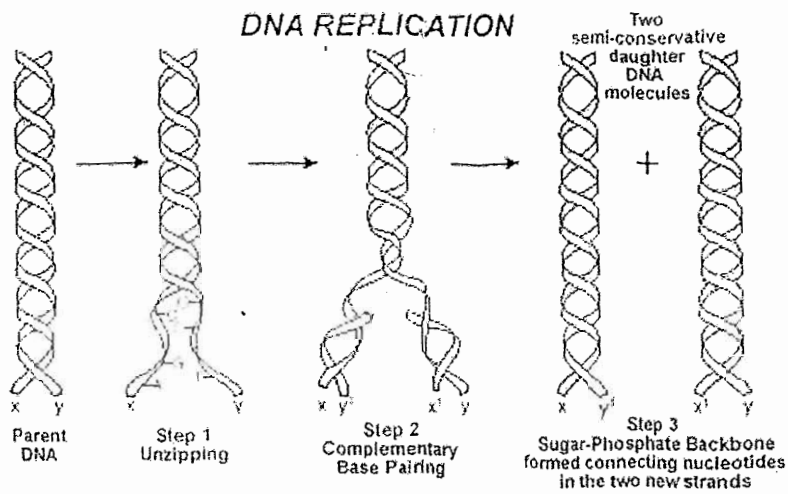

The one variable component of DNA is the nitrogenous base. The sugar and phosphoric acid are the same in all DNA. There are four different nitrogenous bases, however, which vary in sequence and quantity from organism to organism and from species to species. These four bases are adenine, thymine, cytosine and guanine; for simplicity we shall refer to them as A, T, C and G.

Even before the double helix was discovered, biochemists knew that the amount of A always equaled the amount of T in the DNA of any organism, and the amount of C always equaled the amount of G. With the clarification of the molecular nature of DNA, the reasons for this phenomenon and for the precision of duplication of genes became apparent.

The consistency of the equality C+T = A+G is a function of the size of the bases, the space available between the sugar molecules in the double helix, and the chemical nature of these bases. The general shapes of the four nitrogenous bases are given earlier. As we have seen earlier, because of their chemical structures, A and G are classified as purine bases and C and T are pyrimidine bases. As you can see, purines are larger molecules than pyrimidines. Within the diameter of the DNA helix, only purine-pyrimidine paring can occur; purine-purine would be too large to fit into the helix and pyrimidine-pyrimidine pairing would be too small. In addition, certain details of chemical structure of the bases, which we need not go into, allow linkage only of A with T and of C with G. This is known as **specificity** of base pairing. Because of specific base pairing, the strands of DNA are referred to as being **complementary** to each other in their base sequences.

Experiments indicate that when a cell is preparing to undergo division, the double helix within each linear chromosome unwinds in a progressive manner from one end. An enzyme has been found which causes the hydrogen bonds holding the bases together to break, resulting in single polynucleotide strands with the bases exposed. From the nucleoplasm, new molecules of the bases are bonded to the exposed bases. Because of the specificity of base-pairing, the original partners of the exposed bases are replaced by new but **chemically identical molecules.** Enzymes then cause the joining of the nucleotides into a chainlike molecule.

Each of the two new strands of polynucleotides will be the complement of its partner strand, and each will be identical to the original partners. The end results are two double helices that are exactly alike. They will separate in mitosis into daughter cells, which will therefore be genetically identical.

In higher organisms, cells contain chromosome paris rather than the single strand of DNA found in bacteria, and each chromosome consists of a double helix of DNA. During mitosis and meiosis, one of the double helices resulting from the synthesis described above will be in each of the sister chromatids. For this reason the sister chromatids may be considered genetically identical, barring events such as crossing over. Just as all chemical reactions that occur in a cell are under the control of enzymes, so are all the steps involved in the duplication of DNA, which is referred to as **replication.** This type of duplication, in which the resulting double helices consist of one old and one new strand, is frequently referred to as **semiconservative replication.**

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PROTEIN SYNTHESIS

Prior to the discovery of the double helix, some definite ideas had already been formed about the actual function of a gene. An essential class of macromolecules called proteins contributes to the structure of the cell and, in the form of enzymes, determines the metabolic reactions of a cell. In other words, the structure and function of a cell – which are actually its phenotype – are determined in great part by the proteins in the cell.

Based on experimentation in the year 1941, G. Beadle and E. Tatum have come out with the famous "**One Gene One Enzyme" Hypothesis.** They felt that there is a definite relationship between genes and those vital proteins that control metabolic reactions, enzymes:

"The development and functioning of an organism consists essentially of an integrated system of chemical reactions controlled in some manner by genes. It is entirely tenable to suppose that these genes which are themselves a part of the system, control or regulate specific reactions in the system either by acting directly as enzymes or by determining the specificities of enzymes."

In human, the relationship between genes and enzymes was discovered many decades before Beadle and Tatum's work. In 1901, a British physician, Sir Archibald Garrod, was studying a condition called alkaptonuria. Mendel's laws were just receiving their due attention in the scientific world at the time. Garrod noted that several patients suffering from the disease were offspring of consanguineous marriages. With the help of classical geneticists, Garrod concluded that alkaptonuria was a heritable disease, which was determined by a recessive gene.

Further study showed a number of heritable diseases that seemed to result form metabolic disorders, and so in 1908 Garrod published a book called **The Inborn Errors of Metabolism**, in which he linked these biochemical defects with genes. Since that time over 100 conditions have been shown to be due to either the deficiency or absence of specific enzymes.

Fortunately, a few of these conditions can be treated after detection. For example, a homozygous condition, phenylketonuria (PKU) exists in which the enzyme for the breakdown of an amino acid phenylalanine is missing. Undetected, PKU results in severe mental retardation and other defects. However, if the condition is detected in a homozygous baby, the mental retardation can be avoided by simply eliminating excess phenylalanine from the baby's diet. In other disorders, such as albinism, cures are not yet possible.

The Mechanism of Protein Synthesis

In attempting to discern the mechanism by which genes determine the production of enzymes and other proteins in a cell, biologists had to reflect on the fact that the synthesis of proteins generally occurs in the cytoplasm of the cell. The genetic information for the synthesis, however, resides in the DNA, which in higher organisms like the human beings remains in the nucleus, being bound up in the chromosomes. Thus, it appeared likely to scientists attempting to understand gene function that if genes remain in the nucleus, there must be some form of communication between the DNA and cytoplasmic factors involved in protein synthesis.

Evidence of a channel of communication between the nucleus and cytoplasm came from various areas of biology. Experiments in this context revealed that some particles leave the nucleus by traversing the nuclear membrane to enter the cytoplasm. The high magnification of powerful electron microscopes showed that the nuclear membrane is actually full of tiny pores through which material can pass.

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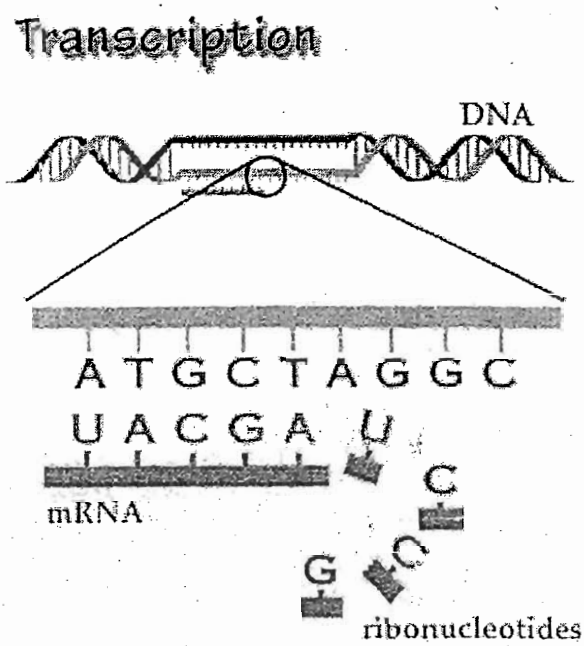
Messenger Molecules for the Gene: Transcription



Two French scientists at the Pasteur Institute, F. Jacob and J. Monod hypothesized the nature of the system of communication between chromosomal and cytoplasmic elements. They proposed that there must be a molecule that serves as a "messenger" to carry the genetic message from the gene to the ribosome, the organelle that was known to be associated with protein synthesis in the cytoplasm of cells.

Subsequent experimentation showed that there was such a messenger molecule, and that it was, in fact, a type of nucleic acid slightly different from DNA, called **ribonucleic acid, or RNA.** The major differences between DNA and RNA are that the latter has a different sugar, **ribose**, which accounts for its name. Also, RNA is single stranded (unlike DNA which double-stranded) and has uracil (U) instead of thymine (T) as one of its four bases. The initial evidence for **messenger ribonucleic acid (mRNA)** was obtained from studies that confirmed that RNA molecules do move from the nucleus to the cytoplasm.

mRNA is formed when a particular segment of DNA – that is, a gene – becomes active. IN a manner similar to the replication of DNA during cell division, an active gene serves as the template for the formation of a piece of RNA, which then contains a sequence of bases that is the complement of one of the strands of DNA and identical to the other strand of DNA. Each sequence of three bases on the mRNA is called a **codon.** (We shall discuss later why the codon is composed of a sequence of only three bases, no more or less). The process of copying an RNA strand from a DNA template is known as **transcription.** Usually only one strand of the DNA helix serves as s template for transcription.

Processing of mRNA

In eukaryotes, a number of chemical changes in the mRNA must take place following transcription before it is released into the cytoplasm. One of these changes is known as **capping,** during which the beginning of the mRNA chain is altered in a particular way. A part of this process involves the addition of methyl (CH_3) groups at one end and a addition of a number of adenines are added to the other end of the mRNA.

The functions of these particular chemical changes are still under study. Some scientists have proposed that they may be necessary in order for the mRNA to enter the cytoplasm, and that once they have occurred, they protect the mRNA from cytoplasmic enzymes which might degrade the RNA. Yet others propose that the caps and multiple adenine sequences (also known as poly-A) may serve to bind mRNA to other cytoplasmic factors involved in protein synthesis.

Some recent observations have revealed another fascinating aspect of mRNA processing. Technological advances have enabled molecular geneticists to determine every base of certain genes, and also of mRNA molecules transcribed from these genes. It was discovered that mRNA of a particular gene that is newly transcribed (i.e. still in the nucleus) has more bases than the mature mRNA of the gene found in the cytoplasm.

AT
GC

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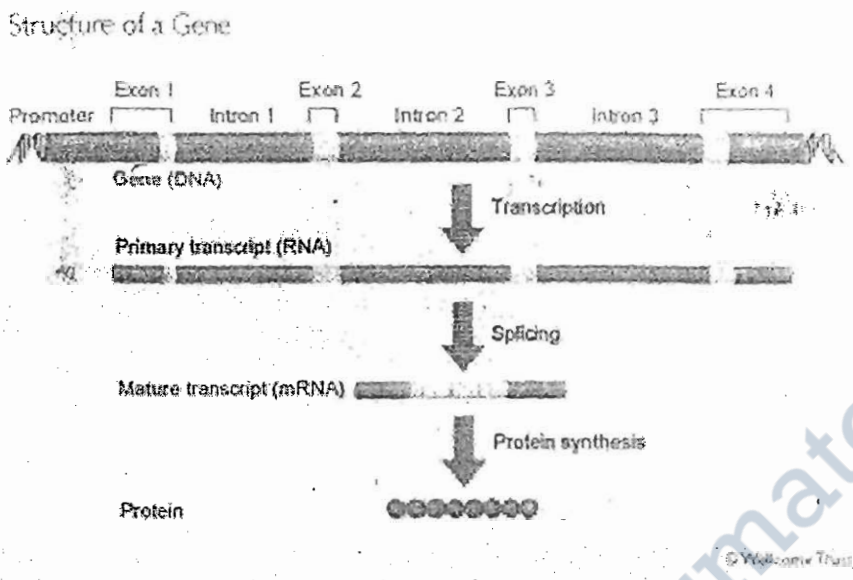
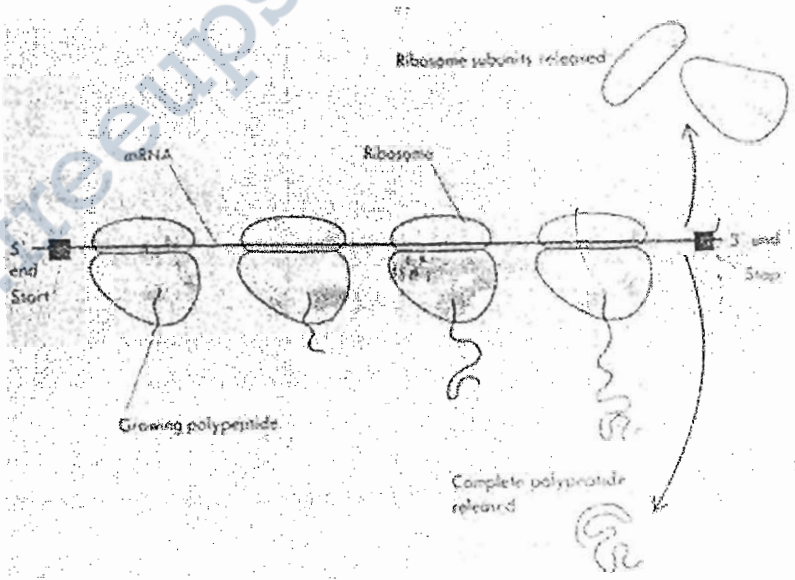
As more reports of discrepancies between the number of bases in mature mRNA and newly transcribed RNA, or the corresponding DNA, appeared, scientists realized that portions of the DNA transcribed into mRNA are actually removed, or **excised,** from the messenger molecule before it leaves the nucleus to take part in protein synthesis. Using a number of different techniques, scientists confirmed that this phenomenon occurred in cells of various eukaryotic organisms and some viruses. For example, these mRNA have been found in the genes for insulin, and hemoglobin, in mammals. A gene that determines Alpha-2 Collagen (a component of cartilage) is known to be over 50,000 bases long; yet its mature mRNA has fewer than 5,000 bases. No less than 50 regions of the newly transcribed collagen mRNA are removed.



The terminology, which has been most commonly used for a sequence of DNA that is transcribed but excised from the messenger molecule, is **intron**, and **exon** for the sequences that are retained. There is some evidence that their presence and excision are involved with the presence of a nuclear membrane and perhaps, with the movement of mRNAs from the nucleus into the cytoplasm. Since the mRNA precursor molecules found in nuclei are in various stages of processing, the population of these nuclear molecules is very heterogeneous. We refer to them as **heterogeneous nuclear RNA,** or **hnRNA.**

It is fascinating to contemplate the controls that must exist in cells of eukaryotes over the processing of mRNAs. How do enzymes that cut the mRNA molecule recognize the boundaries of introns from exons? After the introns are removed, how are the remaining pieces then spliced together in correct order? There is evidence that introns are excised in sequence. How is this controlled? The process in the above figure summarizes the above aspects of mRNA processing.



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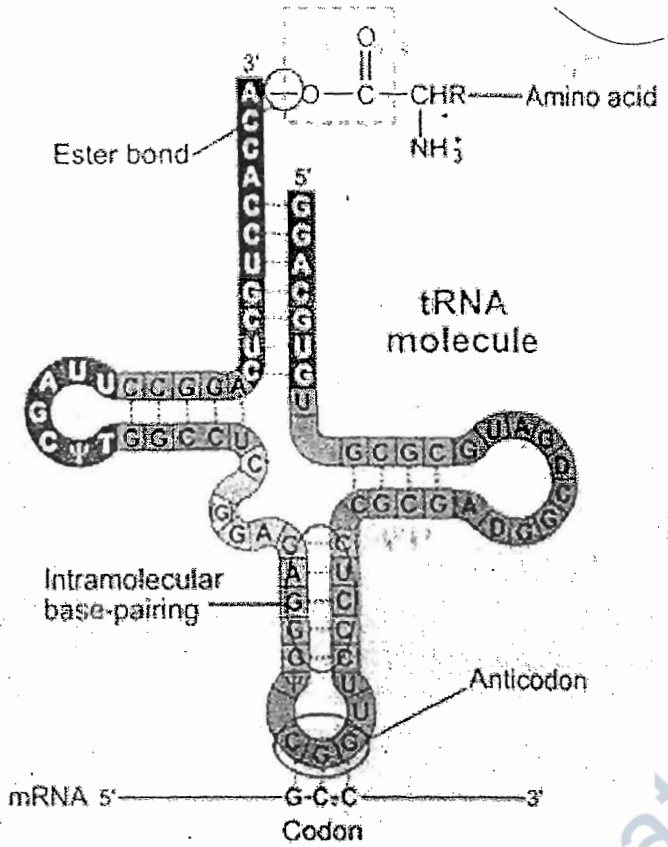

Translation of the Genetic Message

Once the mRNA has been transcribed and processed in eukaryotic cells, it then leaves the nucleus and becomes attached to ribosome particles in the cytoplasm.

Ribosomes: Ribosomes have been found to contain RNA (referred to as rRNA) and protein as well, and because they are the sites of protein synthesis in a cell, they have sometimes been called the "factories" of a cell. The figure shows diagrammatically the relationship between an mRNA strand and a group of ribosomes, often called **polyribosomes** or **polysomes.**

Ribosomes are actually very complex organelles of the cell. The rRNA is transcribed directly from the DNA, as is mRNA. Other genes determining ribosomal proteins are transcribed to give mRNAs. The messages enter the cytoplasm, and the proteins are synthesized. After the synthesis, newly synthesized ribosomal proteins are believed to reenter the nucleus to bind with the rRNAs. The organization of proteins and rRNA into ribosomal particles usually occurs around the areas of DNA that specify rRNA. The resulting clump of molecules forms a structure called a **nucleolus** as the term implies, nucleoli are found only in the nucleus. Therefore, they are present only in eukaryotic cells. In bacteria, ribosomes are formed and released into the surrounding cytoplasm.

tRNA: In the cytoplasm there is yet another class of DNA-determined RNA molecules which become involved in the process of protein synthesis when mRNA attaches to polysomes. These molecules, referred to either as **soluble RNA (sRNA)** or, more commonly as **transfer RNA (tRNA)** are quite different in form and function from mRNA. Recent evidence has indicated that the tRNA molecule is a very interesting one, featuring segments, which exist as double-stranded nucleic acid, while other regions are single-stranded. The double-stranded regions are not composed of two separate strands as in DNA, but indicate areas of complementary bases in different parts of the tRNA molecule that fold over and base pair. This results in a folded structure with a shape that sometimes resembles a cloverleaf.

There is evidence that after the tRNA is transcribed from DNA, some chemical changes and processing also occur. There is also modification of some of the bases, such as the addition of methyl groups. One end of all tRNA molecules contains the sequence CCA.

tRNA has other unique features: it is joined at the CCA end to a single molecule of amino acid, and at the apex of one of the loops found in tRNA there is a short sequence of bases (called the anticodon) that is the source of attraction between the tRNA and the mRNA. tRNA that has a sequence of bases at the loop complementary to a sequence on the mRNA forms a bond with the messenger similar to the bonding between the double strands of the DNA helix. The two other loops are believed to be the sites of attachment of the tRNA to the ribosome, and of the enzyme catalyzing the acceptance of an amino acid by the tRNA.

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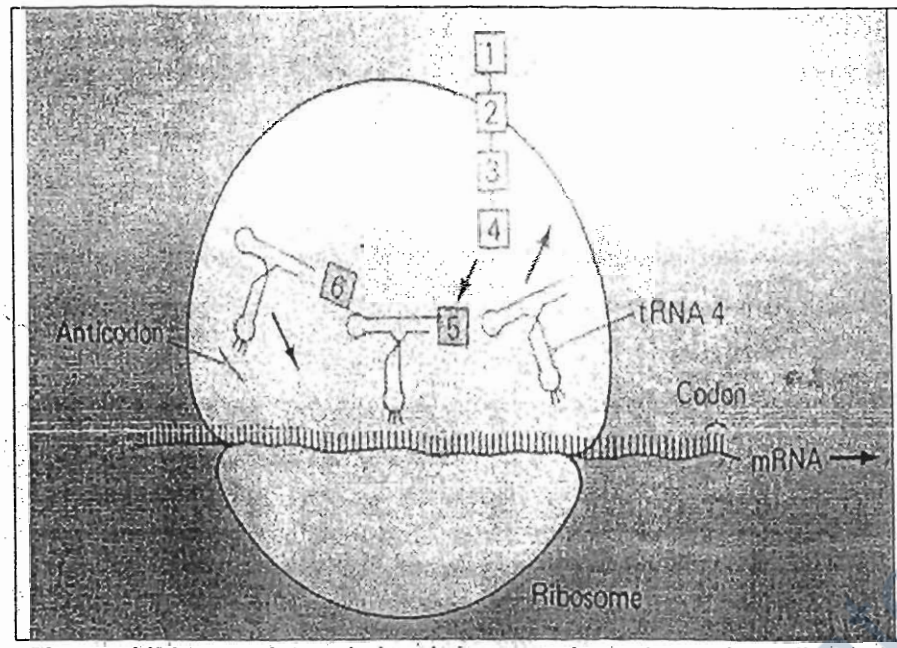
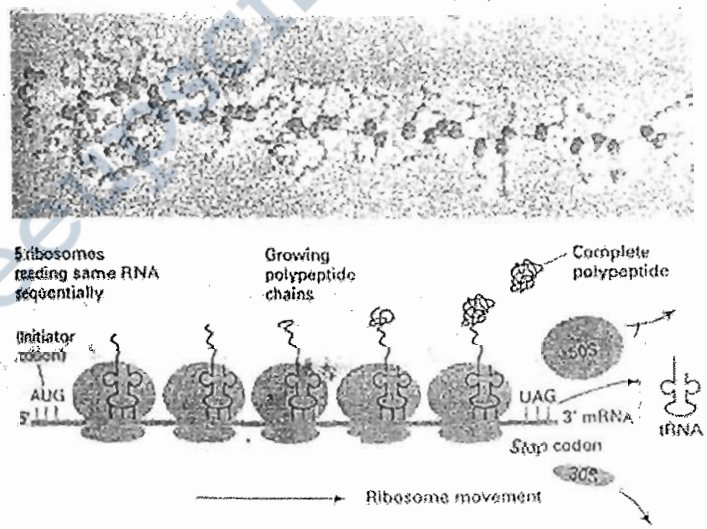
Several tRNA molecules become attached to the long messenger, and they bring their amino acids with them. The result of this interaction between mRNA and tRNA is progressively lengthening series of amino acid molecules lined up next to one another. This interaction is a gene-determined protein synthesis known as **translation.**



The interaction among mRNA, and ribosomes in the formation of a polypeptide chain, known as the translation phase of protein synthesis. This is a detailed schematic diagram of tRNA molecules bringing in and releasing amino acids as they are added to the growing polypeptide chain. The sequence of amino acids in the chain is indicated by numbers and is determined by the sequence of triplet cods in the mRNA.

The establishment of peptide bonds between the amino acids is followed by the enzymatic release of the amino acids from the tRNA, resulting in the formation of a chain of amino acids. This chain of amino acids is a polypeptide.

As the growing polypeptide is added to the next amino acid in the sequence, the amino acid is enzymatically released from the tRNA, which then returns to the cytoplasm to pick up another amino acid molecule, the same kind it had before. After the message has been completely translated, the polypeptide chain is released. The ribosome breaks up into two parts of different sizes, which reassemble when polysomes are formed, in preparation for further protein synthesis.



Other Aspects of Protein Synthesis

Most mRNAs are short-lived molecules; they are destroyed once their messages have been translated into protein molecules. This may seem wasteful, since each message is for a protein the cells need, but actually it is necessary in order to maintain a balance of reactions in the cell. If a particular message is allowed to be translated continuously, it could cause the finely tuned balance of metabolic reactions to be thrown off. For one thing, the same amino acids would be needed over and over again, depleting the supply of such molecules in the cytoplasm faster than they could be replaced. If a protein is needed in large quantities by the cell, it is obtained by the continual production of more mRNA bearing the same

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genetic message. In the case of some proteins, such as hemoglobin, mRNAs may be stable, and large numbers are stored in the red blood cells.

Also, it may be detrimental to a cell or an organism to have too much of a very potent substance. Consider, for example, the hormone adrenaline, which is synthesized in the adrenal glands (a pair of small glands situated on top of the kidneys). In times of emergency or excitement, one of the ways the body can respond is by producing adrenaline in response to signals from the nervous system. When circulated throughout the body in the bloodstream, adrenaline causes the symptoms of excitement so familiar to all of us: rapid breathing, dilation of the pupils, spurts of energy and strength, etc. Imaging how exhausting it would be in this state constantly! It is not only fortunate but imperative for our survival that our cells and tissues do not continually produce all the substances they are capable of producing.

tRNAs on the other hand are longer-lived in general and have been found to take part in several cellular functions besides protein synthesis and ribosome particles take part in more than one translation of mRNAs.

How Does a Cell Produce Correct Proteins?

Following the discovery of the steps involved in protein synthesis, a concerted effort was undertaken by many laboratories to determine how the cell is able to synthesize the correct protein. It is obvious that not all cells contain the same proteins. No two cells types look alike, and no two cell types function in the same way; for example, muscle cells are able to contract because of special contractile proteins that are present in great amount in their highly specialized cytoplasm, but not in the cytoplasm of other cells.

The number and sequence of amino acids in its polypeptide chains determine the individuality of a protein. Therefore, in order to determine that a specific protein will be made by a cell, the genetic material DNA must have in its substance the information for a particular number and sequence of amino acids. Because of the great number of proteins found in nature, it was evident to molecular biologists that the answer did not lie in the sugars and phosphoric acid of DNA. These subunits of DNA are the same in all cells and could not therefore account for any of the variability. What is variable in DNA is the sequence and number of nitrogenous bases in cells from organism to organism, although the proportional amounts and sequences of the four bases are the same in cells from the same individual. There must be a correlation, then, between the sequence of bases in DNA and the sequence of amino acids in proteins. In other words, the message for particular proteins must be encoded in some way in the sequence of nitrogenous bases.

The Genetic Code

There are 20 amino acids; there are four different bases in DNA. How many bases would form a code for a particular amino acid? It can be recognized that the code could not be a doublet code. With four bases to work with, there are only 16 possible combinations of two bases, for example, A-T, A-C, A-G, A-A, C-T, C-C, C-G, G-C etc. (Note that here we are discussing the sequence of bases on one strand of DNA, not the base pairs between the two partner strands of a double helix). Whatever the code is, it has to allow for at least 20 different combinations to cover the 20 different amino acids that exist.

It was evident that the code was probably a triplet code, which allows for 64 combinations. It is beyond the scope of our discussion to go into the many experiments conducted. It is enough to say that scientists have established that the genetic is a triplet code, and have completely solved the "meaning" of each of the 64 triplets. This has to stand as one of the great scientific achievements of all time.

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First position (5' end)	U		C		A		G		Third position (3' end)
U	UUU	Phe	UCU	Ser	UAU	Tyr	UGU	Cys	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu	UCA		UAA	Stop	UGA	Stop	A
	UUG		UCG		UAG	Stop	UGG	Trp	G
C	CUU	Leu	CCU	Pro	CAU	His	CGU	Arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile	ACU	Thr	AAU	Asn	AGU	Ser	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys	AGA	Arg	A
	AUG		ACG		AAG		AGG		G
G	GUU	Val	GCU	Ala	GAU	Asp	GGU	Gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu	GGA		A
	GUG		GCG		GAG		GGG		G

Amino acid names:

Ala = alanine	Gln = glutamine	Leu = leucine	Ser = serine
Arg = arginine	Glu = glutamate	Lys = lysine	Thr = threonine
Asn = asparagine	Gly = glycine	Met = methionine	Trp = tryptophan
Asp = aspartate	His = histidine	Phe = phenylalanine	Tyr = Tyrosine
Cys = cysteine	Ile = Isoleucine	Pro = proline	Val = valine

This table shows the complete unabridged "dictionary" of the genetic code. Note that the code contains U and not T in the triplets. This is due to the fact that geneticists refer to the triplets carried on the mRNA molecules as the components of the genetic code rather than the sequence in the DNA itself, because it was from work on mRNA that the code was deciphered. Each triplet representing the information for a particular amino acid is referred to as a codon.

Obviously 64 possible combinations in triplet sequence are more than enough to represent 20 amino acids, and indeed, there has been found a certain redundancy in the genetic code, whereby more than on triplet codes for a particular amino acid. Further, it has been found that the first two bases on the codon are usually the important ones, the third base may in some cases be any of the four bases, and the triplet would still code for the same amino acid as long as the first two bases are the same and have the same sequence. This concept is known as the **wobble hypothesis**, first formulated by Francis Crick.

Knowledge of the genetic code has helped to clarify some of the events in protein synthesis. The tRNA molecules interact with the codons on the mRNA because at the apex of one of the hairpin loops the molecules have a triplet which is complementary to the codon. This triplet on the tRNA, as was stated before, is called an **anticodon.** The only type of amino acid that each tRNA carries is the one specified by the codon complementary to the anticodon of the tRNA. The codons are not overlapping on the mRNA.

One can make the analogy of the translation of the sequence of triplets on a messenger molecule to the translation of words from one language to another in this manner the proteins synthesized by a cell are read from messages determined by a gene.

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Termination of the Message

Chromosomes contain numerous genes and we now know that each gene must consist of scores of nucleotides. What causes transcription of mRNA to occur only over the length of a particular gene and not overlap into neighboring DNA?

It appears that the message being formed ends at codons, which do not code for any amino acids. If you will refer to the table of Genetic Code again, you will see that there are three such codons which do not code for amino acids. It is thought that the positions of these termination condons (once called nonsense codons) serve to terminate the message. There is some evidence that they also cause the release of the polypeptide chains from the tRNA molecules.

The initiation of gene activity, that is, of the transcription of mRNA can result form "feedback" information, which the nucleus obtains from substances in the cytoplasm. The nature of the chemical state of the nucleoplasm, which must reflect the state of the cytoplasm from which it receives nutrients and replacement substances, could serve as a signal to trigger transcription of certain genes which code for new proteins required by the cell.

GENE MUTATION

In a species variations may be caused by changes in the environment, by changes in the heredity constitution or more commonly by a combination of both. If a variation appears only when the environment is changed and disappears when it is restored, it is not heritable and as it is not built into the genotype, it does not provide a permanent step on which evolutionary changes can be based. These are phenotypic variations and are known as **modifications**. They are defined as phenotypic differences between organisms of similar genotype. If however, changes occur which are independent of the environment and heritable, they are liable to provide a permanent step on which evolutionary changes may be built. Such changes occur in the genome of an individual and may be caused by recombination or crossing over and mutation. Recombination usually causes no remarkable variation because it merely redistributes existing genetic material among different individuals. However, mutations are sudden changes in genotype, involving qualitative or quantitative alterations in the genetic material itself.

Mutations occur frequently in the nature and have been reported in many organisms. In man, the mutations cause variation in hair, color, skin pigmentation and several somatic malformations.

KINDS OF MUTATIONS

There exists a lot of controversy about the possible kinds of mutations among geneticists. They have been classified variously according to different criteria as follows:

1. **Classification of Mutation according to type of Cells:** According to their occurrence in somatic and germinal cells, following types of mutations have been classified:

    a) **Somatic Mutations**: The mutations occurring in non-reproductive body cells are known as somatic mutations. The genetic and evolutionary consequences of somatic mutations are insignificant, since only single cells and their daughter cells are involved. If, however a somatic mutation occurs early during embryonic life, the mutant cells may constitute a large proportion of body cells and the animal body may be a mosaic for different types of cells. Somatic mutations have been often related with malignant (cancerous) growth.

    b) **Gametic Mutations**: The mutations occurring in gamete cells (e.g., sperms and ova) are called gametic mutations. Such mutations are heritable and of immense genetic significance. Genetic mutations form the raw material for natural selection.

2. **Classification of Mutations according to the Size and Quality:** According to the size, following two types of mutations have been recognized.

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a) **Point Mutation:** When heritable alterations occur in a very small segment of DNA molecule, i.e., a single nucleotide or nucleotide pair, then these types of mutations are called "point mutations". Point mutations may occur due to following types of sub-nucleotide changes in DNA and RNA:

1) Deletion mutations - The point mutation, which is caused due to loss or deletion of some portion (single nucleotide pair) in a gene is called deletion mutation.

2) Insertion or addition mutation - The point mutations that occur due to addition of one or more extra nucleotides to a gene or cistron are called insertion mutations. Certain chemical substances called mutagens can artificially induce the insertion mutations.

3) Substitution mutation - A point mutation in which a nucleotide is replaced by another nucleotide is called substitution mutation. Substitution mutations alter the phenotype of an organism variously and are of great genetic significance.

b) **Gross Mutation:** When changes involve more than one nucleotide pair, or entire gene, then such mutations are called gross mutations. Gross mutations occur due to rearrangements of genes within the genome.

3. **Classification of Mutation according to the Origin:** According to the mode of origin, following two kinds of mutations have been recognized:

a) **Spontaneous Mutations:** Spontaneous mutations occur suddenly in the nature and their origin is unknown. They have been reported in many organisms.

b) **Induced Mutations:** Mutations can be induced artificially in the living organisms by exposing them to abnormal environments such as radiation, certain physical conditions like temperature and some chemicals. The substances or agents that induce artificial mutations are called mutagens or mutagenic agents. Mutagenic agents are of the following kinds:

i) Radiations - The radiations which are important in mutagenesis are of two categories: The first type is ionizing radiation such as X-rays and gamma rays, alpha and beta rays, electrons, neutrons, protons and other fast moving particles. The second type is non-ionizing radiations such as ultraviolet and visible light.

ii) Temperature as mutagens - The rate of all chemical reactions is influenced by temperature. It is not surprising that temperature can be mutagenic. For example, an increase of 10°C temperature increases the mutation rate two or three fold. Temperature probably affects the thermal stability of DNA.

iii) Chemical mutagens - Many chemical substances have been responsible to increase the mutability of genes. Any chemical substance that effects the chemical environment of chromosomes is likely to influence, at least indirectly, the stability of DNA and its ability to replicate without error. A chemical mutation can cause mutation only when it enters the nucleus of the cell.

CHROMOSOMES

Chromosomes are complex structures located in the cell nucleus, they are composed of DNA, histone and non-histone proteins, RNA , and polysaccharides. They are basically the "packages" that contain the DNA. Normally chromosomes can't be seen with a light microscope but during cell division they become condensed enough to be easily analyzed at 1000X. To collect cells with their chromosomes in this condensed state they are exposed to a mitotic inhibitor which blocks formation of the spindle and arrests cell division at the metaphase stage.

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A variety of tissue types can be used to obtain chromosome preparations. Some examples include peripheral blood, bone marrow, amniotic fluid, and products of conception.

Under the microscope chromosomes appear as thin, thread-like structures. They all have a short arm and long arm separated by a primary constriction called the ***centromere***. The short arm is designated as ***p*** and the long arm as ***q***. The centromere is the location of spindle attachment and is an integral part of the chromosome. It is essential for the normal movement and segregation of chromosomes during cell division. Human metaphase chromosomes come in three basic shapes and can be categorized according to the length of the short and long arms and also the centromere location. ***Metacentric*** chromosomes have short and long arms of roughly equal length with the centromere in the middle. ***Submetacentric*** chromosomes have short and long arms of unequal length with the centromere more towards one end. ***Acrocentric*** chromosomes have a centromere very near to one end and have very small short arms. They frequently have secondary constrictions on the short arms that connect very small pieces of DNA, called stalks and satellites, to the centromere. The stalks contain genes that code for ribosomal RNA.

The diagrams called ideograms*, of G-banded chromosomes 1, 9, and 14 are typical examples of metacentric, submetacentric, and acrocentric chromosomes respectively. The ideogram is basically a "chromosome map" showing the relationship between the short and long arms, centromere (*cen*), and in the case of acrocentric chromosomes the stalks (*st*) and satellites (*sa*). The specific banding patterns are also illustrated. Each band is numbered to aid in describing rearrangements.

Normal human somatic cells have 46 chromosomes: 22 pairs, or ***homologs***, of autosomes (chromosomes 1-22) and two sex chromosomes. This is called the ***diploid*** number. Females carry two X chromosomes (46,XX) while males have an X and a Y (46,XY). Germ cells (egg and sperm) have 23 chromosomes: one copy of each autosome plus a single sex chromosome. This is referred to as the ***haploid*** number. One chromosome from each autosomal pair plus one sex chromosome is inherited from each parent. Mothers can contribute only an X chromosome to their children while fathers can contribute either an X or a Y.

CELL DIVISION

All organisms, even the largest, start their life from a single cell. Growth and reproduction are characteristics of cells, indeed of all living organisms. All cells reproduce by dividing into two, with each parental cell giving rise to two daughter cells each time they divide. These newly formed daughter cells can themselves grow and divide, giving rise to a new cell population that is formed by the growth and division of a single parental cell and its progeny. In other words, such cycles of growth and division allow a single cell to form a structure consisting of millions of cells.

Cell Cycle

Cell division is a very important process in all living organisms. During the division of a cell, DNA replication and cell growth also take place. All these processes, i.e., cell division, DNA replication, and cell growth, hence, have to take place in a coordinated way to ensure correct division and formation of progeny cells containing intact genomes. The sequence of events by which a cell duplicates its genome, synthesizes the other constituents of the cell and eventually divides into two daughter cells is termed **cell cycle**. Although cell growth (in terms of cytoplasmic increase) is a continuous process, DNA synthesis occurs only during one specific stage in the cell cycle. The replicated chromosomes (DNA) are then distributed to daughter nuclei by a complex series of events during cell division. These events are themselves under genetic control.

Phases of the Cell Cycle

A typical eukaryotic cell cycle is illustrated by human cells in culture. These cells divide once in approximately every 24 hours. However, this duration of cell cycle can vary from organism to organism and also from cell type to cell type. Yeast for example, can progress through the cell cycle in only about 90 minutes.

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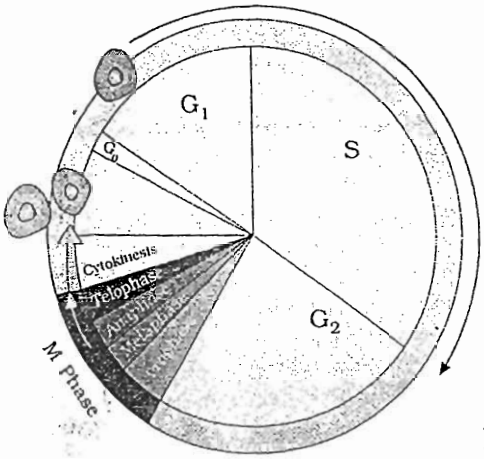
The cell cycle is divided into two basic phases: Interphase and M Phase (Mitosis phase)



The M Phase represents the phase when the actual cell division or mitosis occurs and the interphase represents the phase between two successive M phases. It is significant to note that in the 24 hour average duration of cell cycle of a human cell, cell division proper lasts for only about an hour. The interphase lasts more than 95% of the duration of cell cycle. The M Phase starts with the nuclear division, corresponding to the separation of daughter chromosomes (karyokinesis) and usually ends with division of cytoplasm (cytokinesis). The interphase, though called the resting phase, is the time during which the cell is preparing for division by undergoing both cell growth and DNA replication in an orderly manner. The interphase is divided into three further phases: G_1 phase (Gap 1), 0 S phase (Synthesis) and G_2 phase (Gap 2)

G1 phase corresponds to the interval between mitosis and initiation of DNA replication. During G1 phase the cell is metabolically active and continuously grows but does not replicate its DNA. S or synthesis phase marks the period during which DNA synthesis or replication takes place. During this time the amount of DNA per cell doubles. If the initial amount of DNA is denoted as 2C then it increases to 4C. However, there is no increase in the chromosome number; if the cell had diploid or 2n number of chromosomes at G1, even after S phase the number of chromosomes remains the same, i.e., 2n. In animal cells, during the S phase, DNA replication begins in the nucleus, and the centriole duplicates in the cytoplasm. During the G2 phase, proteins are synthesized in preparation for mitosis while cell growth continues.

M Phase

This is the most dramatic period of the cell cycle, involving a major reorganization of virtually all components of the cell. Since the number of chromosomes in the parent and progeny cells is the same, it is also called as equational division. Though for convenience mitosis has been divided into four stages of nuclear division, it is very essential to understand that cell division is a progressive process and very clear-cut lines cannot be drawn between various stages. Mitosis is divided into the following four stages: Prophase, Metaphase, Anaphase and Telophase

Prophase

Prophase, which is the first stage of mitosis, follows the S and G_2 phases of interphase. In the S and G_2 phases the new DNA molecules formed are not distinct but interwined. Prophase is marked by the initiation of condensation of chromosomal material. The chromosomal material becomes untangled during the process of chromatin condensation. The centriole, which had undergone duplication during S phase of interphase, now begins to move towards opposite poles of the cell. The completion of prophase can thus be marked by the following characteristic events:

- Chromosomal material condenses to form compact mitotic chromosomes. Chromosomes are seen to be composed of two chromatids attached together at the centromere.
- Initiation of the assembly of mitotic spindle, the microtubules, the proteinaceous components of the cell cytoplasm help in the process.

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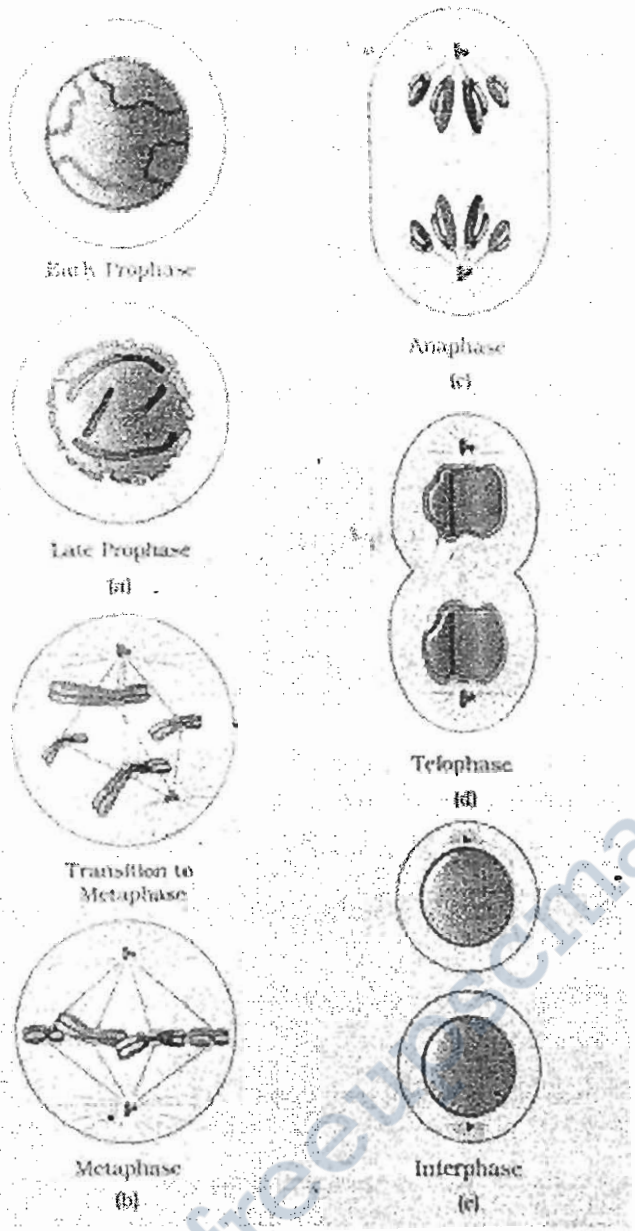

Cells at the end of prophase, when viewed under the microscope, do not show golgi complexes, endoplasmic reticulum, nucleolus and the nuclear envelope.

Metaphase

The complete disintegration of the nuclear envelope marks the start of the second phase of mitosis, hence the chromosomes are spread through the cytoplasm of the cell. By this stage, condensation of chromosomes is completed and they can be observed clearly under the microscope. This then, is the stage at which morphology of chromosomes is most easily studied. At this stage, metaphase chromosome is made up of two sister chromatids, which are held together by the centromer. Small disc-shaped structures at the surface of the centromeres are called kinetochores. These structures serve as the sites of attachment of spindle fibres (formed by the spindle fibres) to the chromosomes that are moved into position at the centre of the cell. Hence, the metaphase is characterised by all the chromosomes coming to lie at the equator with one chromatid of each chromosome connected by its kinetochore to spindle fibres from one pole and its sister chromatid connected by its kinetochore to spindle fibres from the opposite pole. The plane of alignment of the chromosomes at metaphase is referred to as the metaphase plate. The key features of metaphase are:

- Spindle fibres attach to kinetochores of chromosomes.
- Chromosomes are moved to spindle equator and get aligned along metaphase plate through spindle fibres to both poles.

Anaphase

At the onset of anaphase, each chromosome arranged at the metaphase plate is split simultaneously and the two daughter chromatids, now referred to as chromosomes of the future daughter nuclei, begin their migration towards the two opposite poles. As each chromosome moves away from the equatorial plate, the centromere of each chromosome is towards the pole and hence at the leading edge, with the arms of the chromosome trailing behind. Thus, anaphase stage is characterized the following key events:

- Centromeres split and chromatids separate.
- Chromatids move to opposite poles.

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Telophase

At the beginning of the final stage of mitosis, i.e., telophase, the chromosomes that have reached their respective poles decondense and lose their individuality. The individual chromosomes can no longer be seen and chromatin material tends to collect in a mass in the two poles. This is the stage that shows the following key events:

- Chromosomes cluster at opposite spindle poles and their identity is lost as discrete elements.
- Nuclear envelope assembles around the chromosome clusters.
- Nucleolus, golgi complex and ER reform.

Cytokinesis

Mitosis accomplishes not only the segregation of duplicated chromosomes into daughter nuclei (karyokinesis), but the cell itself is divided into two daughter cells by a separate process called cytokinesis at the end of which cell division is complete. In an animal cell, this is achieved by the appearance of a furrow in the plasma membrane. The furrow gradually deepens and ultimately joins in the centre dividing the cell cytoplasm into two.

Significance of Mitosis

Mitosis or the equational division is usually restricted to the diploid cells only. However, in some lower plants and in some social insects haploid cells also divide by mitosis. It is very essential to understand the significance of this division in the life of an organism. Are you aware of some examples where you have studied about haploid and diploid insects? Mitosis results in the production of diploid daughter cells with identical genetic complement usually. The growth of multicellular organisms is due to mitosis. Cell growth results in disturbing the ratio between the nucleus and the cytoplasm. It therefore becomes essential for the cell to divide to restore the nucleo-cytoplasmic ratio. A very significant contribution of mitosis is cell repair. The cells of the upper layer of the epidermis, cells of the lining of the gut, and blood cells are being constantly replaced.

MEIOSIS

The production of offspring by sexual reproduction includes the fusion of two gametes, each with a complete haploid set of chromosomes. Gametes are formed from specialised diploid cells. This specialised kind of cell division that reduces the chromosome number by half results in the production of haploid daughter cells. This kind of division is called meiosis. Meiosis ensures the production of haploid phase in the life cycle of sexually reproducing organisms whereas fertilisation restores the diploid phase. We come across meiosis during gametogenesis in plants and animals. This leads to the formation of haploid gametes. The key features of meiosis are as follows:

- Meiosis involves two sequential cycles of nuclear and cell division called meiosis I and meiosis II but only a single cycle of DNA replication.
- Meiosis I is initiated after the parental chromosomes have replicated to produce identical sister chromatids at the S phase.
- Meiosis involves pairing of homologous chromosomes and recombination between them.
- Four haploid cells are formed at the end of meiosis II.

Meiotic events can be grouped under the following phases:

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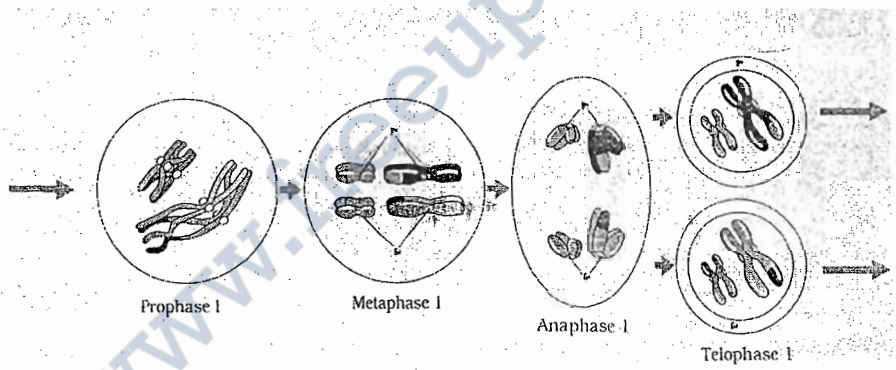
Meiosis I	Meiosis II
Prophase I	Prophase II
Metaphase I	Metaphase II
Anaphase I	Anaphase II
Telophase I	Telophase II

MEIOSIS I

Prophase I: Prophase of the first meiotic division is typically longer and more complex when compared to prophase of mitosis. It has been further subdivided into the following five phases based on chromosomal behaviour, i.e., Leptotene, Zygotene, Pachytene, Diplotene and Diakinesis.

During **leptotene** stage the chromosomes become gradually visible under the light microscope. The compaction of chromosomes continues throughout leptotene. This is followed by the second stage of prophase I called **zygotene.** During this stage chromosomes start pairing together and this process of association is called synapsis. Such paired chromosomes are called homologous chromosomes. Electron micrographs of this stage indicate that chromosome synapsis is accompanied by the formation of complex structure called **synaptonemal complex.** The complex formed by a pair of synapsed homologous chromosomes is called a bivalent or a tetrad. However, these are more clearly visible at the next stage. The first two stages of prophase I are relatively short-lived compared to the next stage that is **pachytene.** During this stage **bivalent** chromosomes now clearly appears as tetrads. This stage is characterized by the appearance of recombination nodules, the sites at which crossing over occurs between non-sister chromatids of the homologous chromosomes. Crossing over is the exchange of genetic material between two homologous chromosomes. Crossing over is also an enzyme-mediated process and the enzyme involved is called recombinase. Crossing over leads to recombination of genetic material on the two chromosomes. Recombination between homologous chromosomes is completed by the end of pachytene, leaving the chromosomes linked at the sites of crossing over.

The beginning of **diplotene** is recognized by the dissolution of the synaptonemal complex and the tendency of the recombined homologous chromosomes of the bivalents to separate from each other except at the sites of crossovers. These X-shaped structures, are called **chiasmata.**



The final stage of meiotic prophase I is **diakinesis.** This is marked by terminalization of chiasmata. During this phase the chromosomes are fully condensed and the meiotic spindle is assembled to prepare the homologous chromosomes for separation. By the end of diakinesis, the nucleolus disappears and the nuclear envelope also breaks down. Diakinesis represents transition to metaphase.

Metaphase I: The bivalent chromosomes align on the equatorial plate. The microtubules from the opposite poles of the spindle attach to the pair of homologous chromosomes.

Anaphase I: The homologous chromosomes separate, while sister chromatids remain associated at their

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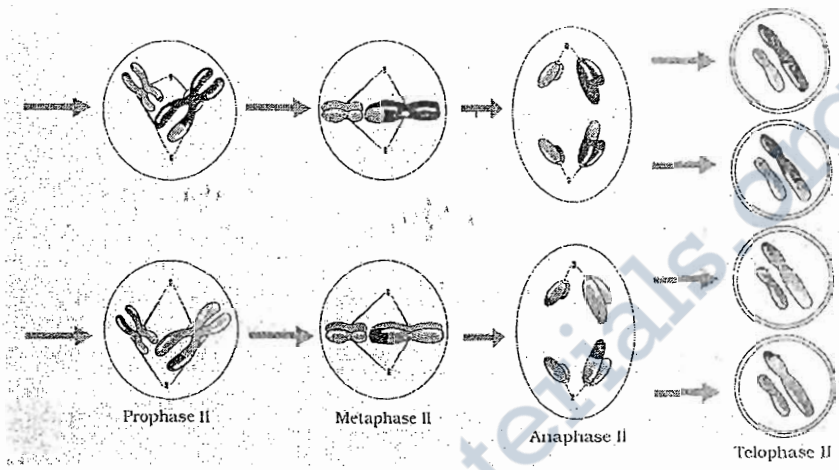
centromeres.

Telophase I: The nuclear membrane and nucleolus reappear, cytokinesis follows and this is called as diad of cells. Although in many cases the chromosomes do undergo some dispersion, they do not reach the extremely extended state of the interphase nucleus. The stage between the two meiotic divisions is called interkinesis and is generally short lived. Interkinesis is followed by prophase II, a much simpler prophase than prophase I.

MEIOSIS II

Prophase II: Meiosis II is initiated immediately after cytokinesis, usually before the chromosomes have fully elongated. In contrast to meiosis I, meiosis II resembles a normal mitosis. The nuclear membrane disappears by the end of prophase II. The chromosomes again become compact.



Metaphase II: At this stage the chromosomes align at the equator and the microtubules from opposite poles of the spindle get attached to the kinetochores of sister chromatids.

Anaphase II: It begins with the simultaneous splitting of the centromere of each chromosome (which was holding the sister chromatids together), allowing them to move toward opposite poles of the cell.

Telophase II: Meiosis ends with telophase II, in which the two groups of chromosomes once again get enclosed by a nuclear envelope; cytokinesis follows resulting in the formation of tetrad of cells i.e., four haploid daughter cells.

Significance of Meiosis

Meiosis is the mechanism by which conservation of specific chromosome number of each species is achieved across generations in sexually reproducing organisms, even though the process, per se, paradoxically, results in reduction of chromosome number by half. It also increases the genetic variability in the population of organisms from one generation to the next. Variations are very important for the process of evolution.

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9.1 HUMAN GENETICS – METHODS AND APPLICATIONS

Though the branch of human genetics was established as early as 1901, no significant work was done in the early years as man was found to be an unfavourable object for genetic studies because of the following reasons:

1. Controlled breeding experiments under standardized environment are impossible because of social hindrance.
2. Number of offspring of each marriage is very small so that the statistical computations are not possible.
3. Life span of man is long. As a result, the results cannot be studied for three or more generations.
4. Majority of human beings are genetically heterozygous for many characters. Therefore, it is difficult to get isogenic strains.

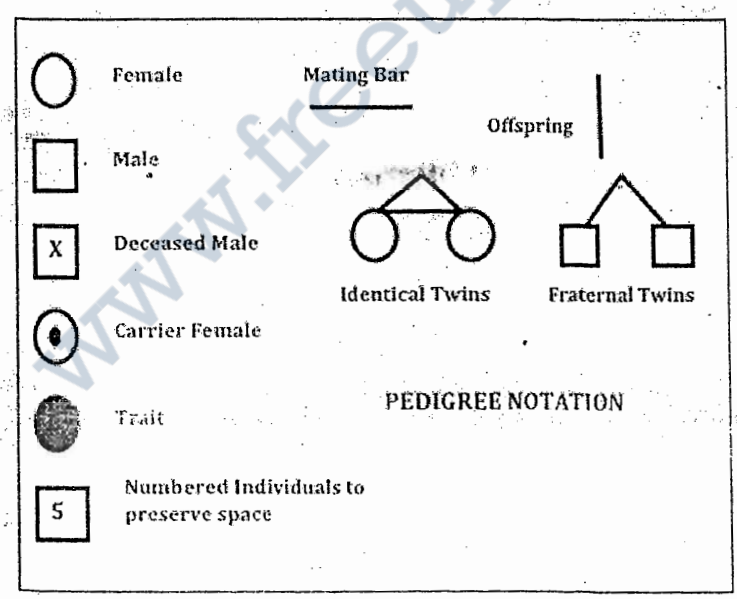
In recent years, the development of newer techniques has helped in understanding the mechanism of inheritance of a number of characters in man.

PEDIGREE ANALYSIS

Although human beings have, in the past, reproduced to the point of overpopulating our fragile planet, the relatively small numbers of offspring per generation in our families is a hindrance from the standpoint of genetic analysis. In addition, as geneticists themselves are obviously human beings with limited life spans, it is somewhat impractical to wait for three generations or more to appear.

There is nothing we can - or would - do about making experimental cases, but there is something we can do about establishing patterns of transmission of traits. Since we cannot go forward, that is, wait for future generations, we go backward; we gather information about all existing members of the family under study and as much information as possible about previous generations; and draw up what is referred to as a Pedigree chart, a family tree of sorts.

For the geneticist, however, information about the phenotype, in addition to names of the individuals, is essential if a pedigree is to be useful. The incidence of a particular condition in the pedigree will often indicate whether the gene involved is autosomal or X-linked, dominant or recessive.



Pedigree analysis is the tool most widely used for study and representation of the inheritance of human traits, and certain standard symbols have been established by geneticists in their publications. Traditionally, females are depicted by circles; males are depicted by squares. A mating is indicated by a horizontal bar connecting a circle and square, and the symbols for offspring are shown suspended from a line drawn perpendicular to the mating bar, as in the figure enclosed.

Family members expressing the trait under study are usually indicated by solid black symbols, as is one of the sons in the pedigree in the figure

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enclosed. Heterozygotes or carriers of the genes customarily are designated by a black dot in the middle of the symbol, as is the case of the mother in the pedigree chart given, or by coloring half the symbol black. If an individual is deceased, a cross is placed through the symbol, and if the phenotype of the individual is uncertain, a question mark is placed in the symbol.

The indication for identical and non-identical twins are given in the figure enclosed. If there are many offspring in a family, numbers are placed in the symbols to conserve space and simplify the pedigree chart. For example, number 5 in the circles and number 4 in the squares (related to each others as brothers and sisters), would indicate that there were five normal girls and boys in this sibship. Occasionally, an arrow pointing at a particular affected individual, as in the figure enclosed, is added to indicate that this is the person who brought the trait to the geneticist's attention. Such a person is called as a Proband, or Propositus if male, and Proposita if female. Sometimes, one can see pedigrees that are headed by a single person. It means that the mate is normal and believed to be of no consequence to the analysis of the pattern of transmission.

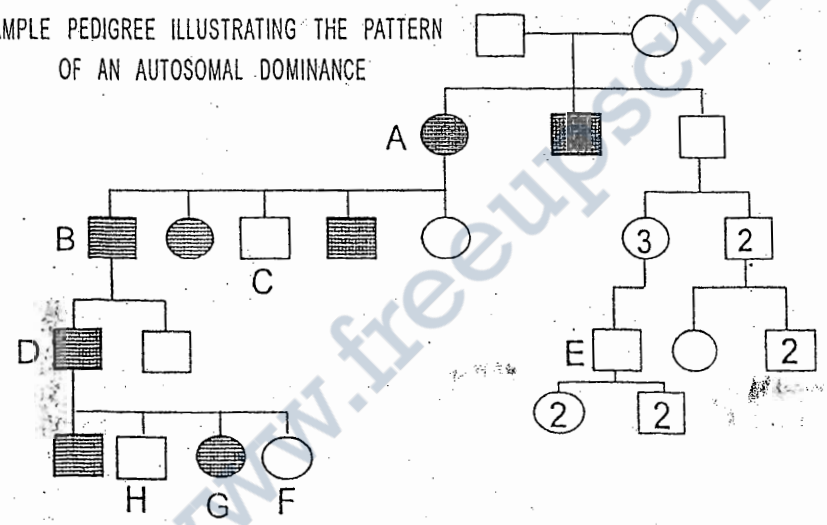
These are the usual symbols used in making up pedigrees. Occasionally, one finds variations in the charts drawn by different geneticists, but they are usually explained by an accompanying key.

Illustrating Inheritance in Pedigree Charts

1. Autosomal Dominance:

Let us turn our attention now to pedigrees which illustrate different kinds of inheritance. For the most part, we shall be using pedigrees drawn up in the study of inherited diseases rather than normal traits, simply because there are better documented pedigrees of diseases and abnormal conditions. Furthermore, an autosomal dominant gene regardless of the trait it determines, normal or abnormal, will show the pattern of transmission characteristic of all autosomal dominant genes.

SAMPLE PEDIGREE ILLUSTRATING THE PATTERN OF AN AUTOSOMAL DOMINANCE



In the figure provided, a sample pedigree of a family in which an autosomal dominant trait was transmitted to many in the kindred, certain characteristics of the pattern of transmission would lead a geneticist to conclude that the trait is determined by an autosomal gene (genes present on the chromosomes other than the sex chromosomes - X, Y). First, equal numbers of women and men are affected. Second, the affected man in the third generation (B) passed the trait to his son (D). As we discussed earlier, the Y chromosome is the only sex chromosome which a man transmits to his sons. Thus, the father could not have passed the condition to his sons if determined by an X linked gene, i.e., one that would be located on the man's X chromosome.

Along the same lines, the affected male in the fourth generation (D) produced an affected daughter in the fifth generation (G). This again would be impossible short of a mutation if the gene were an X linked dominant, since he would have passed the gene to all his daughters, who would all be affected. An X linked recessive would not be expressed in any daughter, since we must assume from the pedigree that

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the man's wife must have been normal, and she would have contributed a dominant normal allele to mask any recessive gene on the man's X chromosome.

This last point holds true for an autosomal gene as well as an X linked one. If the gene were an autosomal recessive, there is no way that the girls in the fifth generation can be affected if the mother is homozygous normal. Therefore, if the gene is neither X linked nor recessive, it must be autosomal dominant.

Additional evidence that the gene is dominant, lies in the fact that there is no skipping of generations in the expression of the trait, because the trait is expressed whenever the gene is present. Another characteristic of dominant traits is that every affected individual has an affected parent. In order for an offspring to have inherited the dominant gene from a parent, that parent must therefore also be affected.

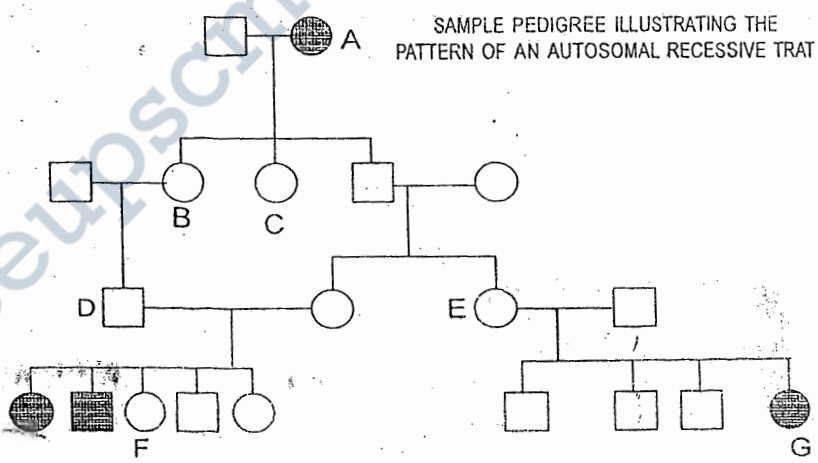
A third factor characterizing dominant traits is that when one generation does not express the trait, the trait is lost and will not reappear in future generations. This is so, of course, because the unaffected people must be homozygous recessive and would not have a dominant allele to transmit. The right side of the pedigree for the second, third, fourth and fifth generations illustrates this principle.

Hereditary conditions in humans known to be determined by autosomal dominant genes include achondroplastic dwarfism, Huntington's chorea (a neural degeneration disease), and nail patella syndrome, which is expressed as abnormal knees and nails.

2. Autosomal Recessiveness:

The figure given shows a pedigree of a family in which an inherited condition was found in a number of individuals in the fourth generation. The presence of affected daughters whose fathers and mothers are normal again indicates that the gene is not sex linked but autosomal. It could not be an X linked dominant because the parents are normal, and for the gene to be X linked recessive would require that the paternal X chromosome carry the gene, which would only be possible if the father was affected.

In addition, the rarity of the condition, and the fact that two different sibships of affected individuals were involved in which the parents are normal, argue strongly against the possibility that this may be an autosomal dominant trait. Because heterozygotes are always phenotypically determined by their dominant genes, the resulting masking of recessive genes leads to "skipping of generations" in the expression of the condition under study. This is one characteristic of the pattern of transmission that distinguishes recessive genes.



SAMPLE PEDIGREE ILLUSTRATING THE PATTERN OF AN AUTOSOMAL RECESSIVE TRAT

Furthermore, the pedigree shows that the affected son and his affected sister in the fourth generation are the offspring of two related individuals, specifically first cousins. It is not surprising to find consanguineous marriages when dealing with the recessive trait.

If a recessive trait is fairly rare, as are most abnormal conditions, the chances of two people being heterozygous for this are much greater if they are related than if they are not, since the two related people may have inherited the same gene from a common ancestor. The pedigree in the figure is

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somewhat unusual from this point of view because the father of the other sibship in the fourth generation was apparently unrelated to his wife, but by chance was a carrier for the same recessive gene. Examples of autosomal recessive conditions in humans are diseases such as sickle cell anaemia, Tay Sachs disease (a neural degenerative condition), and albinism. They are, of course, of concern because the genes may be transmitted by carriers who are phenotypically normal. In some cases, such as sickle cell anaemia and Tay Sachs disease, carriers may be identified with biochemical tests.

THE TWIN METHOD

The act of reproduction culminates by the act of union of egg and sperm, following the intricate maneuvers of meiosis. A number of cellular changes occur in the egg. After the union of the sperm and egg, there is a reconstitution of diploid number of chromosomes in the zygote. From here, the zygote undergoes repeated mitotic divisions to give rise to a fully developed child. Now we shall consider the formation of twins, the determination of zygosity and the estimates of heritability.

It was in 1876 that Francis Galton indicated the value of studying twins in Genetic studies. He also introduced the terms "nature" and "nurture". But two years earlier, in 1874, Camille Dareste gave the distinction between two types of twins - Monozygotic and Dizygotic. It was in 1924, Siemens provided the method for investigation of normal variability and determination of zygosity.

How Twins are formed?

There are two types of twins - Monozygotic and Dizygotic.

Monozygotic Twins: Monozygotic twins are named so because they originate from a single zygote. The zygote splits into two at an early stage and develops into two genetically identical individuals. It is a case of extreme duplication. Since both the members of the pair are developed from a single zygote, they are genetically identical. Hence monozygotic twins are also called as identical twins, and are of same sex.

Dizygotic Twins: Dizygotic twins result from the fertilization of two eggs by two sperms. The genetic relationship between these twins is no different from sibs. Hence dizygotic twins are also called fraternal twins. These twins may be of different sexes also.

The frequencies of twinning are not same in all populations. It is fairly high in Belgium, for example, touching a frequency of 1.79% while in South American population it recorded 0.8%.

Identical twins arising from a division of a single fertilized egg have genetic homogeneity. Any observed differences in their phenotype can be considered purely environmental in origin. By contrast, fraternal twins of the same sex coming from separate eggs may differ genetically and environmentally for any character. By measuring both kinds of twins for a particular character, we can evaluate the roles of Heredity and Environment. That is, on one hand, we have a genetically identical pair of the same age and sex raised in a single uterine environment, and on the other hand, we have a genetically dissimilar pair of the same age and sex also raised in a common uterine environment. Presumably, the difference between these two kinds of twins is only in the extent of their genetic similarity. If phenotypic similarities for a particular character are greater among identical twins than among fraternal twins, or conversely, if phenotypic differences are less among identical, we can ascribe this to the genetic similarity of the identical pairs and the genetic dissimilarity among the fraternal pairs. On the other hand, if phenotypic differences for a particular character are the same for both identical and fraternal twins, we can assume that genetic similarity or dissimilarity plays less of a role than the environmental differences which occur in both kinds of twins.

Concordance and Discordance: One of the ways the phenotypic similarities and differences for a character can be measured in twins is simply to note whether the character is present or absent in one or both members. If both possess the character or are free from it, the pair is concordant, or phenotypically similar. If only one member of the pair possess the trait, the pair is discordant or phenotypically dissimilar. The extent to which identical or fraternal twins differ in their degree of concordance offers a

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measure with which to assess the roles of environment and heredity. If, for example, 20 pairs each of both identical and fraternal twins are evaluated for a particular trait, and 12 pairs of identical twins (60%) show concordance as compared to only 4 pairs of fraternal twins (20%), the trait can be considered to have strong heredity element. Similarly, comparisons in this example between the degree of discordance for identical twins (40%) and for fraternal twins (80%) show that the genetic dissimilarity between the members of the latter group has contributed a greater share to the phenotypic dissimilarity. More equal concordance and discordance ratios between the identical and fraternal groups would, in turn, signify less emphasis on heredity similarity in determination of the trait and more emphasis on environmental factors.

DIAGNOSIS OF ZYGOSITY:

Diagnosis of zygosity implies the diagnosis of a twin pair either as Monozygotic or Dizygotic i.e., to know whether the twin pair are born from same zygote or from different zygotes. This diagnosis is extremely significant in the condition where the twins are of same sex and similar looks. There are basically three methods to diagnose the zygosity of twins.

Placental Method: Through this method absolute diagnosis of zygosity is little difficult. However, this method is sometimes used. In the uterus of the mother, the developing zygote is surrounded by two layers - an inner amnion and outer chorion. A third outer most layer constitutes the placenta. In case of the dizygotic twins, each of the members has completely different set of membranes, with exception of the placenta which might be fused. In the case of monozygotic twins, the nature of fetal membranes and extent of placenta vary,

1. A single amnion, chorion and placenta -
2. A separate amnion but single chorion and placenta.
3. Separate amnion, chorion, but single placenta.
4. Separate amnion, chorion and also placenta.

These are also referred to as:

i) Diamniotic, Dichorionic fused.

ii) Diamniotic, Monochorionic.

iii) Monoamniotic, Monochorionic.

Based on these conditions of placenta the zygosity of twins can be diagnosed.

The Similarity Method: Geneticists, over a period of time with their experiments, have brought out a list of traits in human beings where individual variations occur, like for example, the tissue antibodies. In order to diagnose the zygosity, if skin graft is made, only a monozygotic twin will be able to accept the skin for its pair. However, morphological features such as eye color, nose form, etc., are less reliable data to assess the zygosity.

DNA Fingerprinting: The DNA mapping for the monozygotic twins could be exactly similar whereas it may differ for the dizygotic twins. Based on this DNA fingerprint result, the zygosity of twins can be exactly diagnosed.

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HERITABILITY ESTIMATES

Variations between the human beings, which are phenotypic, are a universal phenomenon. However, the causes for variation may differ. There are basically three reasons for phenotypic variations.

i) Genetic variation.

ii) Environmental variation.

iii) Interaction between Genetics and Environment.

For example, the phenotypic stature of a human being depends both on the genotype and environment. Heritability can be defined as that proportion of the phenotypic variation in any population that can be attributed to genetic factors.

Estimation of heritability is not an easy task. There are several factors responsible for it.

i) Human beings are not experimental animals that can be selectively mated, highly inbred or raised under strictly controlled conditions.

ii) Phenotypic variation does not just depend upon heredity and environment alone but also the interaction between the two, which is difficult to conserve and also quantify.

Monozygotic twins (MZ) however, provide an excellent opportunity to estimate heritability. This is because the members constituting MZ twins are genetically similar. Since the twins have identical genes, any variation among them is largely due to the environmental effect. Most of the time, the MZ twins are reared in the same environment. However, when they are reared apart, a study of the differences between the two individuals can provide a fairly accurate measure of the genetic and environmental components of certain human characteristics.

As we already noted, if the pair constituting the twins are sharing a characteristic, they are concordant for that character. The percentage of concordance provides an estimate of the degree to which a particular character is genetically influenced i.e., the heritability estimate.

Generally, two types of estimation for the heritability of a particular trait are used by geneticists. One is based on the variance between the MZ and DZ twins and the other is based on the MZ twins reared apart and together. These two methods are discussed here.

Heritability Estimate based on the Variance between MZ and DZ Twins

This method is based on the comparison of twins for the degree of concordance and discordance. For example, the concordance rate for polio is high among the MZ twins and low for DZ twins. This shows that polio has a strong genetic susceptibility.

This concordance or discordance rate indicates within-the-pair variation for MZ or DZ twins. Once this within-the-pair variation is known, then the coefficient of correlation can be calculated by using the formula

$$(V_{DZ}-V_{MZ}) \ / \ V_{DZ}$$

Where, V_{DZ} = Within-the-pair variation of Dizygotic or Fraternal twins. It is also denoted as Vf.

VMZ = Within-the-pair variation of Monozygotic or Identical twins. It is also denoted as Vi.

It is obvious that for a character with strong genetic tendency, the value of V_{MZ} will be low, compared to the value of V_{DZ}. Hence, the value of the coefficient of correlation for a character with strong genetic tendency would be approaching the value one.

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If a particular character has a strong environmental influence, the value of V_{MZ} would be high and it may be as high as the value of V_{DZ}. Hence, here the value of the coefficient of correlation would be approaching the value zero.

By the values of genetic and environmental components of a character, we can calculate the heritability by using the formula

$$H = V_G / (V_G+V_E) = V_G / V_T$$

Here, H = Heritability

V_G = Genetic Variance

V_E = Environmental Variance

V_T = Total Variation

One can observe from the above formula that the genetic component of total variation is being calculated.

Heritability Estimates by Variance between MZ twins Reared Apart and Reared Together

Here, the magnitude of difference between MZ twins reared together and reared apart can be calculated. This is based on the assumption that, since the MZ twins are genetically similar, any variation among them is purely environmental in susceptibility. The coefficient of correlation in this case would be calculated by using the formula:

$$(V_A-V_T) \ / \ V_A$$

Where, VA is the variation of the MZ twins reared apart and V_T is the variation of the MZ twins reared together.

In case of a strong environmental influence, the coefficient of correlation would approach 1 (one) because the value of VA will be large and the value of V_T is small. Similarly, if there is a strong genetic influence, the value of VA would be small and the value of V_T will be large.

VIABILITY OF TWIN METHOD IN HERITABILITY ESTIMATES

From quite some time, the twin method is utilized to analyze the role of heredity and environment in bringing about human variation. Moreover, a number of human traits, both physical and behavioral, were analyzed using this method. However, the viability of this method is under consideration. This is because of the following reasons:

1. The twin method does not actually specify the genetic and environmental component, but rather realizes it. It does not recognize the nature, location and behavior of genes; neither does it recognize the physical, chemical nor biological components of the environment. Heredity and environment are rather treated as aggregates.

2. Heritability estimation of complex characters as intelligence and behavior are resented because they result not independently from genetic or environmental influence alone but an abstract interaction between the two, which cannot be easily quantified.

3. Twin method assumes that the MZ twins are exactly identical. However, evidences show that they do differ in a number of characters like size and vigor.

4. The twin method cannot be used to estimate the intra-uterine environmental component and influence. The difference in such environmental component may result in low concordance among MZ twins.

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5. Twin method cannot be applied to analyze traits like congenital malformations.
6. A majority of environmental factors are still being neglected. Hence the changes brought about by these factors may be falsely assumed to be genetic.
7. The sampling of the environment of twins need not be the environment of the population as a whole.
8. Inter population variations are not accounted for. For example, a particular trait may differ in one population for genetic reasons and the same trait in another population may differ due to environmental influence.
9. One of the most important drawbacks of the twin method is that twinning by nature in itself is a very rare phenomenon. In such a case, the generalizations of the findings based on this study are questionable in its objectivity.

The above stated limitations of the twin method or genetics of twins does not imply any lack of importance of this method. It has very much helped the physical anthropologists to understand and draw reasoning for the long standing "twin" concept of nature and nurture. However, additional methods and their findings should also be made use of, like for example, the foster child method.

CO-TWIN METHOD

We have already noted that twins can be monozygotic or identical or dizygotic or fraternal. The degree of similarity can be calculated on the basis of concordance.

In the twin method, the twins are combined and studied in order to understand the influence of heredity and environment. However, in the co-twin studies, the identical twin along with its co-twin and the fraternal twin along with its co-twin are investigated and compared. This technique is used to understand a number of abnormal behaviours, especially schizophrenia.

However, when the conclusions of the twin method and co-twin method are compared for same behaviour, disparities existed. This is due to the following reasons. Take for example, in the co-twin method; rates of schizophrenia are expressed in terms of concordance, the proportion of twin pairs in which both co-twins are diagnosed as schizophrenic. Whereas in the twin method, concordance is determined by "pair wise" analysis, whereby one simply calculates the proportion of all twin pairs in which both are affected. This analysis does not take into consideration the fact that in any given twin pair, either might have been independently registered a case. Thus the co-twins would be counted twice for the concordance rate whereas they should have been counted only once.

The limitations of this method are similar to that of twin method.

FOSTER CHILD

The foster child method is complementary to twin studies on the nurture - nature aspects, particularly of mental traits. It is another method to analyse the respective influence of heredity and environment in the development of a trait.

In this method, various groups of children are selected at random and are placed in different homes classified good, average and poor homes. Since group of children are randomly selected, the genetic factor of the trait studied, say, intelligence, is equally distributed in them. After a lapse of time, they are tested on different intelligence scales. If intelligence has an environmental component, children placed in good homes should score better than those in average and poor homes.

The foster child method is however not free from biases, both at selection and analysis level. Osborne in 1951 has given the following four requirements for using this technique.

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1. Foster children must be placed in the adoptive home sufficiently early to be relatively uninfluenced by the environment of their original home.
2. There must be little or no selective placement of the children.
3. Adequate sample of adoptive children from various social levels must be included.
4. The foster children must be from one population to eliminate variations due to ethnicity or race.

CYTOGENETIC METHOD

Cytogenesis is the study of chromosomes and the related disease states caused by abnormal chromosome number and/or structure. Chromosomes are complex structures located in the cell nucleus, they are composed of DNA, histone and non-histone proteins, RNA, and polysaccharides. They are basically the "packages" that contain the DNA. Normally chromosomes can't be seen with a light microscope but during cell division they become condensed enough to be easily analyzed at 1000X. To collect cells with their chromosomes in this condensed state they are exposed to a mitotic inhibitor which blocks formation of the spindle and arrests cell division at the metaphase stage.

A variety of tissue types can be used to obtain chromosome preparations. Some examples include peripheral blood, bone marrow, amniotic fluid, and products of conception. Although specific techniques differ according to the type of tissue used, the basic method for obtaining chromosome preparations is as follows:

- Sample log-in and initial setup.
- Tissue culture (feeding and maintaining cell cultures).
- Addition of a mitotic inhibitor to arrest cells at metaphase.

Stain chromosome preparations to detect possible numerical and structural changes.

CHROMOSOMAL AND KARYOTYPE ANALYSIS

Human beings have 23 pairs of chromosomes; twenty two pairs are called the autosomes and one pair of Sex chromosomes. The autosomes of man and woman have similar appearance but the sex chromosomes are different. In a female the two sex chromosomes are identical and are represented as XX. In male these are dissimilar and are represented as XY. The Y chromosome is smaller than the X and is the male determining chromosome.

The concept of Karyotyping and human Karyotype is already discussed in the chapter on Molecular Evolution of Primates. This method is one of the primary mechanisms through which the human chromosomes can be analyzed.

Karyotype is a systematized array of chromosomes of a single cell prepared either by drawing or by photography, with the extension in meaning that the chromosomes of a single cell can typify the chromosomes of an individual or even a species.

The term Ideogram means the diagrammatic representation of a karyotype, which may be based on measurements of chromosomes in several or many cells.

For identifying various regions of individual chromosomes, a new technique is being practiced since 1969. This involves staining the chromosomes with fluorescent dyes after certain treatments. The staining gives different patterns of bands and interbands (stained and unstained regions) along the length of chromosomes. The banding patterns of a particular chromosome remain constant for a particular treatment. At present, four such banding patterns are known which are represented as Q, G, C and R

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patterns. According to normal banding patterns, abnormalities in different chromosomes can easily be identified.

1. Q Banding: The Q banding are fluorescent bands observed on human chromosomes by staining with quinacrine (Q) mustard and observing with UV light. By this staining, Y chromosome becomes brightly fluorescent while the distal ends of each chromatid remain unstained.

2. G Banding: The G bands are produced by staining human chromosomes with Giemsa Stain. These bands occur on the same locations as the Q bands. Their staining does not require fluorescent microscopy.

3. C Banding: The C bands are regions of heterochromatin ('C' refers to Constitutive Heterochromatin). These are localized to particular sites on the chromosomes. For example, these are localized to centromere regions of a chromosome.

4. R Banding: This is also called Reverse Banding. The R bands are located in those regions of chromosomes that lie between the fluorescent Q bands. These appear as green; brightly fluorescent band with acridine orange staining.

Significance of Chromosomal analysis

Study of human chromosomes has helped a lot in correlating various human diseases, malformation and deformities with the abnormalities in the number and structure of chromosomes. These abnormalities may be in the autosomes or sex chromosomes. The various abnormalities, both numerical and structural aberrations are discussed in the chapter on Chromosomal disorders.

Chromosomal Abnormalities

Although chromosome abnormalities can be very complex there are two basic types: ***numerical*** and ***structural***. Both types can occur simultaneously.

Numerical abnormalities involve the loss and/or gain of a whole chromosome or chromosomes and can include both autosomes and sex chromosomes. Generally chromosome loss has a greater effect on an individual than does chromosome gain although these can also have severe consequences. Cells which have lost a chromosome are ***monosomy*** for that chromosome while those with an extra chromosome show ***trisomy*** for the chromosome involved. Nearly all autosomal monosomies die shortly after conception and only a few trisomy conditions survive to full term. The most common autosomal numerical abnormality is Down Syndrome or trisomy-21. Trisomies for chromosomes 13 and 18 may also survive to birth but are more severely affected than individuals with Down Syndrome. Curiously, a condition called ***triploidy*** in which there is an extra copy of every chromosome (69 total), can occasionally survive to birth but usually die in the newborn period.

Another general rule is that loss or gain of an autosome has more severe consequences than loss or gain of a sex chromosome. The most common sex chromosome abnormality is monosomy of the X chromosome (45, X) or Turner Syndrome. Another fairly common example is Klinefelter Syndrome (47,XXY). Although there is substantial variation within each syndrome, affected individuals often lead fairly normal lives.

Occasionally an individual carries an extra chromosome which can't be identified by it's banding pattern, these are called ***marker*** chromosomes.

Structural abnormalities involve changes in the structure of one or more chromosomes. They can be incredibly complex but for the purposes of this discussion we will focus on the three of the more common types:

- ***Deletions*** involve loss of material from a single chromosome. The effects are typically severe since there is a loss of genetic material.

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- ***Inversions*** occur when there are two breaks within a single chromosome and the broken segment flips 180° (inverts) and reattaches to form a chromosome that is structurally out-of-sequence. There is usually no risk for problems to an individual if the inversion is of ***familial*** origin (has been inherited from a parent.) There is a slightly increased risk if it is a ***de novo*** (new) mutation due possibly to an interruption of a key gene sequence. Although an inversion carrier may be completely normal, they are at a slightly increased risk for producing a chromosomally ***unbalanced*** embryo. This is because an inverted chromosome has difficulty pairing with it's normal homolog during meiosis, which can result in gametes containing unbalanced derivative chromosomes if an unequal cross-over event occurs.

- ***Translocations*** involve exchange of material between two or more chromosomes. If a translocation is ***reciprocal*** (balanced) the risk for problems to an individual is similar to that with inversions: usually none if *familial* and slightly increased if *de novo*. Problems arise with translocations when gametes from a balanced parent are formed which do not contain both translocation products. When such a gamete combines with a normal gamete from the other parent the result is an *unbalanced* embryo which is partially monosomic for one chromosome and partially trisomic for the other.

Numerical and structural abnormalities can be further divided into two main categories: ***constitutional*** those you are born with; and ***acquired***, those that arise as secondary changes to other diseases such as cancer.

Sometimes individuals are found who have both normal and abnormal cell lines. These people are called ***mosaics*** and in the vast majority of these cases the abnormal cell line has a numerical chromosome abnormality. Structural mosaics are extremely rare. The degree to which an individual is clinically affected usually depends on the percentage of abnormal cells. A routine Cytogenetic analysis typically includes the examination of at least 15-20 cells in order to rule out any clinically significant mosaicism.

These are just some of the more common abnormalities encountered by a Cytogenetic Laboratory. Because the number of abnormal possibilities is almost infinite, a Cytogeneticist must be trained to detect and interpret virtually any chromosome abnormality that can occur.

BIOCHEMICAL METHODS

Genes control the appearance of a phenotypic trait of an organism by exercising control on the developmental and biochemical activities of its cells. All these cellular activities are controlled by enzymes, whose synthesis is directly supervised by genes. A dominant gene is capable of synthesizing the required enzyme and expresses itself, whereas the recessive gene is unable to produce the functional enzyme and hence is unable to express itself. That genes express themselves through the synthesis of enzymes was demonstrated by Beadle and Tatum in 1941. They proposed 'one gene one enzyme' hypothesis. This hypothesis means that all the steps which transform a precursor substance to its end product that is ultimately expressed into a structural or functional phenotypic trait, constitutes a biosynthetic or metabolic pathway. Each step of biosynthetic pathway is catalyzed by a specific enzyme which in turn is synthesized under the control of a specific gene.

Biochemical genetics combines genetics and biochemistry to elucidate the nature of metabolic pathways and their control. These have been widely studied in man where any error in the metabolic pathway is expressed in the form of a disease.

Biochemical genetics was initiated by English physician A.E. Garrod when he noticed that some of the hereditary diseases in man are due to the effects of mutant genes on the metabolic systems. Five metabolic disorders have been noticed in man associated with the defective metabolism of Phenylalanine, an essential amino acid of dietary proteins, (Phenylketonuria, Alkaptonuria, Tyrosinosis, Albinism, and Goitrous Cretinism). Each of the steps of normal metabolism of phenylalanine is controlled by a specific

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enzyme. Various diseases are caused by mutated genes that block particular steps in biochemical reactions.

A number of procedures for separation of biochemical substances and their analysis have been developed. Most important of them are mentioned here:

1. **Separation and Identification of Proteins:** There are basically two methods to separate proteins and their identification.
   - **Gel-filtration:** This method separates proteins by their molecular weight. The mixture of protein is passed through a column filled with a polysaccharide called sephadex. Sephadex is present in the form of beads with inbuilt holes, and proteins with high molecular weight cannot enter through them, and thus are separated.
   - **Electrophoresis:** This involves the separation of proteins based on charge. Proteins being electrically charged, when put in an electric field, move towards positive or negative poles.

After the proteins are separated, their amino acids can be analysed.

2. **Sequencing of DNA:** DNA can be sequenced and analysed by Maxam - Gilbert method. The DNA is cleared by four restriction enzymes that cut DNA at different bases. Thus four sets of DNA are found, each having differing lengths of DNA fragments representing the position of enzymes.

IMMUNOLOGICAL METHOD

The ability of our bodies and cells to resist invasion by foreign objects, be they cellular, viral, or chemical, is the subject of interest of the increasingly important field of immunology. Like all life processes, the basis for immune reactions can be traced to gene action. Because of the profound influence of molecular genetics on all cellular studies, an area of genetics known as Immunogenetics has developed, which combines the two disciplines. We now know that immune reactions are the result of complex gene regulation in our white blood cells.

What is an Immune Reaction?

To the layperson, the term immune reaction generally brings to mind common situations such as allergies and the use of vaccinations to render a person immune to some disease. These are immune reactions, of course, but so far, blood-type incompatibility and transplantation rejection, which we will use as examples, are the two major types of immune reactions.

Blood-type incompatibility results from the presence of different factors, proteins, which are produced by certain white blood cells called plasma cells and release a circulate in our bloodstream. This production of proteins in reaction to the presence of foreign matter in one's body is known as the circulating or humoral type of immune reaction. Transplantation rejection, on the other hand, results from the recognition of foreign cells in the body and the stimulation of white blood cells known as lymphocytes and macrophages to literally attack the foreign tissue and destroy it. This manner of rejection of foreign cells is known as the cell-mediated type of immune reaction.

Antigens and Antibodies

Basically, an immune reaction involves the interaction of two factors, an outside agent that is unacceptable to the special cells of the body involved in immune reactions, and the body factor that responds to this invader. The "foreign" factors, called antigens, are usually organic substances or cells of some sort. When antigens enter the body, they elicit a reaction from the specialised cells of the host, which results in either the production of special proteins called antibodies or the proliferation of cells called phagocytes (as mentioned above), which literally attack the antigens.

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When antibodies are involved, a number of different kinds of reactions can occur: If the antigen is in solution, reaction with antibodies can cause the antigen to precipitate out of solution; or if cells are the antigenic agent, antibodies can cause them to break open, or lyse; finally, antibodies can cause the antigens to agglutinate, or stick together in clumps.

Circulating Antiboides, Blood Groups and Reaction to Disease

The definitions of antigen and antibody, then, are circular: an antigen is a factor that elicits the production of antibodies. An antibody is the protein, found in blood plasma, which is produced by cells of the body in reaction to the presence of an antigen and which specifically reacts to that antigen. Because they circulate in the plasma of blood, antibodies which participate in immune reactions are called circulating antibodies.

The Genetic Basis of ABO Blood Types: The clumping reaction, or agglutination, occurs when different types are mixed because type A individuals have anti-B antibody molecules present in their blood and type B people have anti-A antibodies. Type O individuals are found to have both anti-A and anti-B antibody molecules. On the other hand, type AB individuals have no anti-bodies to the ABO group of antigens at all.

Genetic Determination of Antibody Molecule

What has intrigued biologists is the fact that all plasma, or antibody-producing cells of an individual would of course be genetically identical, yet they are capable of producing only one of millions of different antibodies. By being able to respond specifically to each antigen that enters the body, every individual has the potential to produce a staggering number of different antibody molecules. Consider not only all the different micro organisms that can infect us, but also all the various organic substances and plant materials to which many people react.

The variations in the antibody are made use of in immunological method. Some antigen is injected into man and antibodies are separated and purified. Such antibodies have been studied and their amino acid compositions determined. It has been found that antibodies elicited by different individuals differ in amino-acid composition of different chains. Such antibodies, if injected into some experimental animal will behave as antigen and in the animal the antibody for human antibody will be formed. This anti-serum from the animal is taken which contains antibodies against human antibody. With this anti-sera blood samples can be tested and one will observe how humans differ in genetic variability.

RECOMBINANT DNA TECHNOLOGY

The terms "recombinant DNA technology," "DNA cloning," "molecular cloning,"or "gene cloning" all refer to the same process: the transfer of a DNA fragment of interest from one organism to a self-replicating genetic element such as a bacterial plasmid.

Would be possible to edit the genetic text, modifying it to produce a desired result? In the early 1970s, biochemists at Stanford University showed that genetic traits could indeed be transferred from one organism to another. In this experiment, the DNA of one microorganism recombined with the inserted DNA sequence of another, and thus had been edited to exhibit a very specific modification.

The actual editing, or insertion process, is painstaking, for it involves manipulating incredibly tiny pieces of incredibly tiny organisms. But the process can be explained in terms of editing a written text: scissors and "glue" are used to "cut" and "paste."

The methods used in rDNA technology are fairly simple. We take, for example, the gene for insulin production in humans and paste it into the DNA of Escherichia coli, a bacterium that inhabits the human digestive tract. The bacterial cells divide very rapidly making billions of copies of themselves, and each bacterium carries in its DNA a faithful replica of the gene for insulin production. Each new E. coli cell has inherited the human insulin gene sentence.

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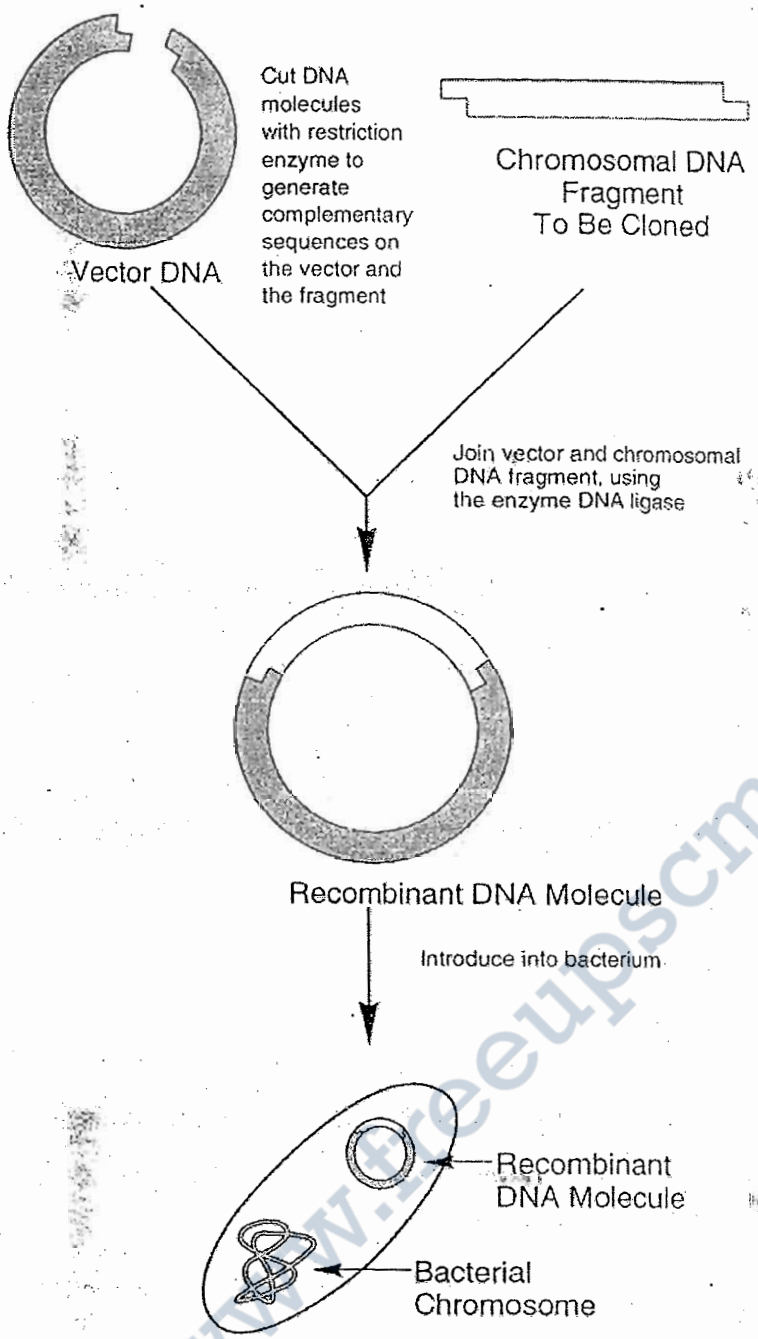

How do we transfer the gene embodying the instruction for insulin production? One approach would be to cut the appropriate gene from human DNA and paste, or splice, it into plasmid DNA, a special kind of DNA that takes a circular form and can be used as a vehicle for this editing job. Our "scissors" are the class of enzymes called restriction enzymes. There are well over a hundred restriction enzymes, each cutting in a very precise way a specific base sequence of the DNA molecule. With these scissors used singly or in various combinations, the segment of the human DNA molecule that specifies insulin production can be isolated. This segment is "glued" into place using an enzyme called DNA ligase. The result is an edited, or recombinant, DNA molecule. When this recombinant plasmid DNA is inserted into E. coli, the cell will be able to process the instructions to assemble the amino acids for insulin production. More importantly, the new instructions are passed along to the next generation of E. coli cells in the process known as gene cloning.

This highly simplified description of rDNA technology does not fully convey the enormous complexity and awesome economy and efficiency of genetic processes. But we can begin to understand how, by using rDNA, it is possible to produce substances of medical and economic value.

Of about 5000 recognised single gene defects in man, about 1500 have been cloned using recombinant DNA techniques. Recombinant DNA technique is used in another completely different way for analysis of structure and function of a gene. Here a DNA is interrupted by insertion of a foreign sequence and study of proteins under such circumstances allows a greater knowledge of functions of genes.

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9.2 MENDELIAN GENETICS IN MAN

Before we proceed with these topics, it is imperative to know Mendelian inheritance. The following discussion helps us understand the concept of inheritance and heredity.

THE MENDELIAN PRINCIPLES

The Austrian monk Gregor Mendel is considered as the pioneer of modern genetics. He was born on July 22, 1822 in Heinzendorf in Austrian Silesia.

The approach of Mendel was simple, logical, scientific mathematical and analytical, probably, because he was well aware about the failures of his predecessor hybridists. The pre-Mendelian hybridists made observations on plants and animals as a whole and study all variations in the hybrid at a time. Mendel however, concentrated on a particular character at a time and studied the pattern of inheritance of only one or two character or characters.

MENDEL'S CONSIDERATIONS

For his hybridization experiments Mendel had certain considerations in his mind about the choice of a suitable material, Mendel's considerations about the material were as follows:

1. **Variation:** The organisms that are to be chosen for the genetic experiments should have a number of detectable differences and at a time only single detectable character should be considered.
2. **Reproduction:** The chosen organisms should be sexually reproducing (i.e., by fusion of male and female sex cells) because only then, the offspring will be able to receive different characters from both the male and female parents.
3. **Controlled mating:** The chosen organisms should be able to mate in controlled or well-planned conditions. Because in genetic experiments sometimes we have to rear genetically pure parents by methods of controlled mating. One should maintain careful records of the offspring of many generations.
4. **Short life cycle:** The chosen organisms should have very short life cycles.
5. **Large number of offspring:** The organisms that have been chosen for the genetic experiments should produce large number of offspring after each successive mating, because it will help in deducing the correct conclusions.
6. **Convenience in handling:** The experimental species should be of a type that can be raised and maintained conveniently and inexpensively in the laboratory. For instance, the elephant will prove entirely useless material for genetic experiments than the Drosophila, pea plants, tomato, rats, guinea pigs, etc., which have been generally used and are still used in hybridization experiment.

MENDEL'S MATERIAL

Mendel found edible pea (Pisum sativum), a best material for his hybridization experiments. A pea plant has various contrasting characters among its different varieties such as a stem which may be tall or dwarf, cotyledons may be green or yellow; seeds may be round or wrinkled, seed coat may be colored or colorless; the unripe pods may be green or yellow; the ripe pods may be inflated or constricted between the seeds, flowers may have axial or terminal positions and the colors of flowers may be red or white. Besides these contrasting characters, the pea plant is a very satisfactory material for the hybridization experiments due to its flower structure. The flowers of pea plants are so constructed that the pollens of a flower normally fall on the stigma of the same flower and thus effect self-pollination or self-fertilization. Mendel easily prevented self-fertilization by removing the stamens from the flower buds. Because, for genetic experiments, cross fertilization was the fundamental necessity, so Mendel cross pollinated two desired pea plants differing in a particular pair of contrasting characters by placing the pollens of flower

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on the stigma of other flower whose stamens have already been removed. Further, to prevent artificially pollinated flower from the contamination by pollens of unknown source by insects, Mendel covered the flowers by bags.

MENDEL'S PROCEDURE

For getting exact results in breeding experiments, it was necessary for Mendel to rear genetically pure variety of pea plants for a single character. Mendel adopted self fertilization technique for it. For instance, to get pure character for tallness, he fertilized a tall pea plant for many generations till the resulted offspring always produced only tall plants. Likewise, he got genetically pure variety for dwarf pea plants. Mendel cross pollinated these two varieties of pea plants which were differing in a pair of contrasting characters, viz., tallness and dwarfness of the stem. When he made observations on the offspring of first generation, he found only tall plants. He allowed the self pollination of the offspring of first generation and made further observations on the offspring of second generation. He was astonished to note both tall and dwarf offspring in the second generation. This showed him that the character of dwarfness disappeared in first generation but again reappeared in second generation. Further, the tall and dwarf plants of second generation were always in the ratio of 3:1 (3 tall:1 dwarf). He self-pollinated the dwarf offspring of second generation and found only dwarf plants in the third generation. But when he self-pollinated the tall plants of second generation then he found that one-third (1/3) tall plants yield only tall plants in third generation, while rest two-third (2/3) tall plants yield tall and dwarf plants in the ratio of 3:1.

On the basis of the results of his experiments, Mendel formulated following three laws:

a) Law of Dominance.

b) Law of Segregation.

c) Law of Independent Assortment.

Actually Mendel himself did not postulate any genetic principle or laws; he simply gave conclusive theoretical and statistical explanations for his hybridization experiments in his research paper. However, it was Correns, the discoverer of Mendel's work, who thought that Mendel's discovery could be represented by these fundamental laws of heredity.

I. LAW OF DOMINANCE

The cross between the pea plants differing in single pair of contrasting characters is known as monohybrid cross. As we have already noticed, when Mendel made a monohybrid cross between tall and dwarf pea plants, then only tall pea plants appeared in the first filial generation (F1). But when the F1 progeny were allowed to be self-fertilized, both tall and dwarf characters appeared in the second filial generation or F2. This shows that in F1 hybrid, the character of tallness dominates or conceals the character of dwarfness. The character of dwarfness, which could ~~not~~ express itself in F1 generation, is called by Mendel as dominant character, while the character which remained unexpressed or latent had been called recessive. According to these results, Mendel formulated the law of dominance which states that "in crossing pure (homozygous) organisms for contrasting character of a pair, only one character of the pair appears in the first filial generation".

MECHANISM OF DOMINANCE

Mendel carefully studied the results of his experiments and it became evident to him that there is a clear-cut difference in the actual visible character and that something which caused its production. Because, the character cannot be present as such in sex cells (sperm and ova) which form the only link between a new individual and the parents. Therefore, there must be something which represent the characters and is responsible for their production. This something of Mendel is now called the factor or gene. The gene thus can be considered as the unit of inheritance that is transmitted in a gamete and determines or

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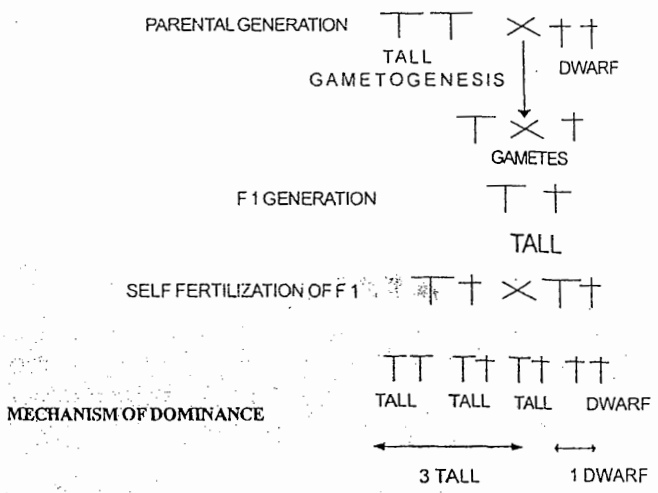
controls the development of a character by interaction with the other genes, the cytoplasm and the environment.

The cytological investigations have now established that the genes are the units of deoxyribonucleic acid (DNA) which along with ribonucleic acid (RNA) and other nucleoproteins constitute the thread-like stainable structures - the chromosomes. The chromosomes are specific in number, shape and size to a particular species.



A diploid cell has two sets of chromosomes that come from two different parents (male and female) via gametes (sperm and ova).

The chromosomes of similar size and nature often form pairs during meiotic cell division and such identical chromosomes are known as homologous chromosomes. Each character of a pair of contrasting characters is represented by a single gene. If the character for tallness is represented by the gene T and because each homologous chromosome of a pair can have single gene for tallness, so the character of tallness in a homozygous tall pea plant will be represented by a pair of genes 'TT'. Likewise the homozygous dwarf pea plant contains the gene 'tt' for the character of dwarfness.

During the gametogenesis, the homologous chromosomes with TT or tt genes are separated and each chromosome with T or t gene is passed to the gamete. The gamete of both parents unite during the process of fertilization and produce a new individual containing both tall (T) and dwarf (t) characters. This new individual of first generation (F1) contains two different genes of a contrasting pair of characters, therefore, it is known as heterozygote or hybrid. Because the hybrids of F1 have tall stems so the character of tallness (T) is considered as dominant and because the character for dwarfness could not express itself in F1 generation, it is considered recessive.

INCOMPLETE DOMINANCE

The law of dominance does not occur universally. For example, when a red flowered pea plant (RR) is crossed with white flowered pea plant (rr), then the F1 hybrid pea plants are found to have pink flowers. It shows that gene for red color could not dominate the gene for white color. The appearance of this intermediate character in F1 generation is known as incomplete dominance. This type of incomplete dominance has also been reported in snapdragons and many animals.

CO-DOMINANCE

In co-dominance, both the dominant and recessive characters occur side by side in F1 hybrids to give the mixed characters. The best example of co-dominance has been observed in cattle. When cattle of red coat are crossed with the cattle of white coat, the F1 heterozygote or hybrid is found to possess roan coat. In roan coat the red and white hairs occur in definite patches but no hair has intermediate color of red and white.

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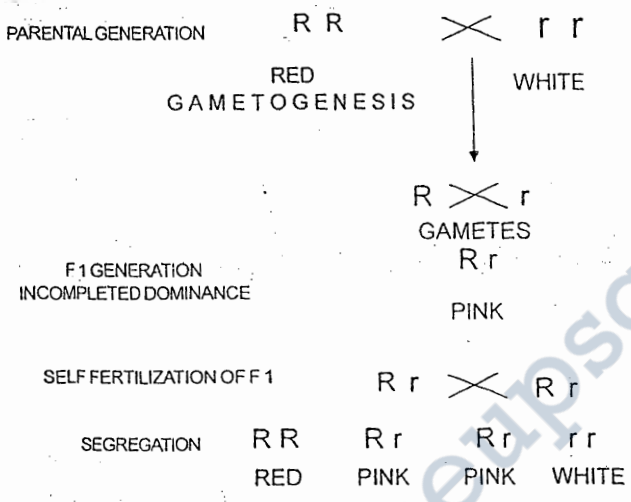
II. LAW OF SEGREGATION

The law of segregation is also known as law of purity of gametes. The law states that the hybrids or heterozygotes of F1 generation have two contrasting characters or allelomorphs of dominant and recessive nature. These characters though remain together for long time but do not contaminate or mix with each other and separate or segregate at the time of gametogenesis, so that each gamete receives only one character either dominant or recessive.

For the proper understanding of the Mendel's law of segregation, it will be helpful to study one of the Mendel's monohybrid cross. Mendel crossed a homozygous red flowered pea plant with a homozygous white flowered pea plant. The F1 heterozygotes or hybrids were found to be pink or purple flowered, thus showing the incomplete dominance of red color over white color. When the F1 hybrids were allowed to be self fertilized, they produce both colors (red or purple) and white flowered pea plants in F2 generation in the ratio of three and one (3:1). The reappearance of white color in F2 generation indicates the process of segregation.

MECHANISM OF SEGREGATION

The mechanism of segregation in above mentioned monohybrid cross between red and white flowers pea plants can be understood by assuming that the homozygous red flowered pea plant has the allele RR for Redness and white flowered pea plant has the alleles rr for whiteness. The pea plant with RR alleles produces the gametes with single allele R and pea plant with rr alleles produces the gametes with the allele r. The gametes of both unite to from a hybrid or heterozygote having the alleles Rr, both for redness and whiteness. Due to the phenomenon of incomplete dominance, the allele R for red color partially expresses itself in hybrids of F1, while the allele r for white color remains latent or recessive. Both the allele R and r remain together for long time but they do not affect each other. Neither they mix nor do they contaminate each other. Because a gamete can contain only one chromosome of a homologous pair, therefore, each gamete can carry single allele R or r. At the time of gametogenesis two types of gametes are produced by F1 hybrids in equal numbers. Half of the gametes carry the allele R and other half carries the allele r. These gametes during the process of fertilization can unite in three possible combinations, viz., RR, Rr, and rr to produce three types of individuals in F2 generation. Thus in F2, 75% individuals have Red colored flowers and 25% white flowers. The appearance of white color in F2 generation indicates that in the hybrid the allele (r) for white color remain along with allele (R) for red color but does not mix with it or contaminated by it and it separates or segregates during gametogenesis.



MECHANISM OF SEGREGATION

III. LAW OF INDEPENDENT ASSORTMENT

To formulate the law of dominance and law of segregation, Mendel considered monohybrid crosses in which single pairs of contrasting characters were considered at a time. But he tried to find out how different characters would behave in relation to each other in their inheritance from generation to generation. For this purpose Mendel crossed two varieties of pea plants which were differing in two pairs

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of contrasting characters. Because such crosses yielded dihybrids and at a time two pairs of contrasting characters had been considered in them, therefore, these crosses were known as dihybrid crosses.

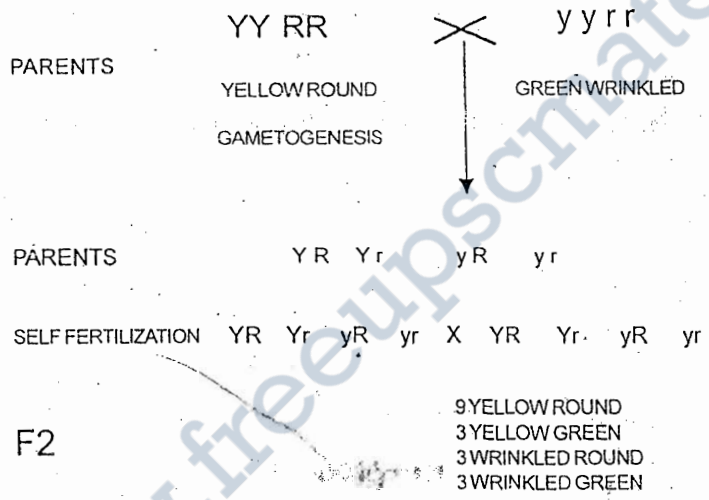
MENDEL'S DIHYBRID CROSS

In one of his hybridization experiment Mendel crossed a homozygous pea plant having yellow round seeds with the homozygous pea plant having green wrinkle seeds. The F1 hybrids were found to have yellow round seeds. When the F1 hybrids were allowed to cross among themselves, they produced four types of seeds in the ratio of 9: 3: 3: 1 given as follows:

1. Yellow Round - 9
2. Yellow wrinkle - 3
3. Green Round - 3
4. Green wrinkle - 1

Besides getting the ratio of 3:1 of the monohybrid crosses Mendel got the ratio of 9:3:3:1. This irregularity in the ratio of F2 offspring was explained by Mendel stating that "when the parents differ from each other in two or more pairs of contrasting characters or factors, then the inheritance of one pair of factor is independent to that of the other pair of factors". This is the Mendel's law of independent assortment.

MECHANISM OF INDEPENDENT ASSORTMENT



MECHANISM OF INDEPENDENT ASSORTMENT

The mechanism of independent assortment can be understood easily by assuming that the homozygous pea plant with yellow round seeds has the alleles YY and RR for the yellow color and roundness of the seed, respectively. Similarly the homozygous pea plant with green wrinkle seeds contains the allele's yy and rr for the green color and wrinkleness of seeds. The gametes which are produced by YYRR and yyrr plants are YR and yr types respectively. When both parents are crossed, the union of both types of gamete takes place to give the F1 hybrid (Yr Rr). The F1 hybrids have been found to contain yellow round seeds showing the dominance of allele Y for yellow color over the recessive allele y for green color and the dominance of allele R for roundness over the recessive allele r for wrinkleness of seed. Now the F1 hybrids have four types of alleles, viz., Y for yellow color, y for green color, R for round shape and r for wrinkleness of seed. During gametogenesis these four alleles may combine in following four combinations:

1. The allele Y may be associated with the allele R to give rise to YR combination.
2. The allele Y may be associated with the allele r to give Yr combination.
3. The allele y may be associated with the allele R to give yR combination.
4. The allele y may be associated with the allele r to give rise to yr combination.

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Thus, four types of alleles are assorted independently to produce four types of gametes, viz., YR, Yr, yR and yr. These four types of gametes (pollens or ovules) of F1 hybrid unite at random in the process of fertilization and produce sixteen types of individuals in F2 generation.

Thus the sixteen F2 individuals have the ratio of 9 yellow rounds: 3 yellow wrinkle: 3 green round: 1 green wrinkle. These results have proved the law of independent assortment and showed that each pair of contrasting characters behaves independently and bear no permanent association or relation with particular character. The allele Y was associated with the allele R in the parent but it does not always remain associated with it in the next generation.

The earlier emphasis for understanding the mechanism of heredity is in the individual. However, anthropological interest lies predominantly with evolution, and the individual is not the unit of evolution. It is not true that people change over time. They get taller and heavier; perhaps their hair changes color, along with all those other things that are variously labeled growth, development, and decline. Yet, even though an individual today is not the same individual he or she will be tomorrow, that person is not evolving. Nor is the fact that people produce offspring different from themselves in evolution, for no two individuals, whether contemporaries or not, are exactly alike. Variation is not evolution. Evolution is change that can lead to the development of new kinds of populations and the population is the unit of evolutionary change.

SINGLE FACTOR, MULTIFACTOR AND POLYGENIC INHERITANCE IN MAN

QUANTITATIVE GENETICS

Genetics is the study of variation. Without variation, genetic analysis at any level becomes difficult. In many cases, this genetic analysis of variation involves striking differences between the organisms (Red or White petals, Long or Short wings) etc. In other cases, however, biological variation is difficult to categorize. E.g., the height and weight in human beings are traits that vary continuously in the population. Distinct classes cannot be identified for such traits, making it difficult to analyze by conventional genetics methods. Quantitative genetics is the discipline that deals with these complex traits.

Quantitative Traits

Quantitative genetics is based on measurements of individuals within a population of organisms. The methods of quantitative genetics recognize two important facts...

1. Most quantitative traits involve contribution of many different genes - sometimes a single gene may exert primary influence but usually there are many equally influential genes. The existence of different alleles at each of these genes will produce genotypic variation which will in turn produce phenotypic variation for the trait.
2. Quantitative traits are often influenced by environmental factors. The environmental influence is to such an extent that quantitative trait will show phenotypic variation over and above that caused by genotypic differences.

The Multifactor Hypothesis

R.A. Fisher realised that the study of quantitative traits had to be reconciled with the Mendelian view of heredity. The problem was that no one could show directly that these traits were determined by single Mendelian factors. Fisher appropriated the seriousness of this problem and proposed a solution.

"Quantitative traits were not determined by single genes, but many and their alleles each had small and approximately additive effects".

Thus, a phenotype of an individual depended on it genotype at all the relevant loci, with each allele adding or subtracting a small amount. Traits that have these features are called **Polygenic Traits**. Fisher also proposed that many environmental factors influenced the trait by adding or subtracting effects in a manner similar to that of all the genetic loci. This

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combination of multiple environmental and genetic factors determined the phenotype of an individual.

Multiple Factor Hypothesis readily explains three features of quantitative traits:

1. The multiplicity of factors contributing to a trait accounted for the variability among phenotypes. This variability arises from the large number of genotypes and environments that are possible when many factors are involved.
2. Fisher's hypothesis suggested an explanation that if a trait is influenced by two types of alleles, plus and minus, defined according to whether they add or subtract an effect, Mendelian segregation should lead to many individuals carrying a mixture of plus or minus. Thus, extreme values of the trait should be rare, whereas intermediate values that are close to the mean should be common.
3. Fisher's principle explains why offspring of crosses between phenotypically different parents often had intermediate phenotypes. One parent might have a large number of plus alleles; the other a large number of minus alleles, and their offspring would have a mixture of both types. Such offspring would therefore be expected to be phenotypically in between the parents.

LETHAL GENE ACTION

One of the most important, but rarely stated, assumptions for a discussion on inheritance of any trait is the equal survival of all the gametes and zygotes produced as a result of segregation. This assumption is fulfilled in case of a majority of the genes. But some genes affect the survival of those zygotes or individuals in which they are present in the appropriate genotype. The effect of various genes on survival ranges from an increase in survival, through no effect to death of all individuals carrying the gene. Genes can be categorized into five types based on their effect on survival – Lethal, Semi-lethal, Sub-vital, Vital and Super-vital genes.

- **Lethal Gene:** A lethal gene is a gene that causes death of all the individuals carrying this gene in the appropriate genotype before they reach adulthood. The appropriate genotype for an allele would depend on its dominance relationship with its other allele (s). The stage of development at which a lethal gene produces its lethal affect varies considerably from one gene to the other - some cause death very early in development while others allow survival and development close to the reproductive age - E.g., Epiloia in man. Lethal genes can further be classified into Recessive lethals, Dominant lethals, Conditional lethals and Gametic lethals.
    - **Recessive Lethals:** Most of the lethal genes are recessive lethals. Their lethal effect is expressed only when they are in the homozygous state. The survival of the heterozygotes is unaffected. However, many other genes are known that have a dominant phenotypic effect in case of heterozygosity, but are recessive lethals - E.g., Achrondroplastic Dwarfism and Xeroderma pigmentosum (producing heavy freckles in heterozygotic condition). Other genes are also present which are recessive both in their phenotypic as well as lethal effects. In such cases, the heterozygous individuals have normal phenotype as well as normal survival and they cannot be differentiated from normal homozygotes. The lethal genes described so far reduce the survival of zygotes that carry them in the appropriate genotype - such lethals are called zygotic lethals and constitute a vast majority of lethal genes.
    - **Dominant Lethals:** These lethal genes reduce viability in the heterozygous conditions as well e.g., Epiloia in man - this gene causes abnormal skin growths, severe mental defects and multiple tumours in the heterozygotes so that they die before reaching adulthood. Hence, dominant lethals cannot be maintained in the population while the recessive lethals are maintained in the heterozygous state. Dominant lethals have to be produced in every generation through mutation.

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- **Conditional Lethals:** Some lethal genes allow normal development and survival, except under a specified condition that is necessary for their lethal effect. Such lethal genes that require a specific condition for their lethal action are termed as conditional lethals. E.g., some lethals require environment conditions like temperature, light, nutrition. Xeroderma pigmentosum in man - light and Phenylketonuria - nutrition. All the conditional lethals require specific genetic environment.

- **Gametic Lethals:** Some genes lead to inviability of gametes or make them incapable of fertilization; such genes are termed as gametic lethals. Gametic lethals lead to a drastic departure from the typical ratio expected in a segregating generation - this phenomenon is called segregation distortion or meiotic drive.

- **Semi Lethal Genes:** Semi lethal genes do not lead to the death of all the individuals that carry them in the appropriate genotype. However, they cause the death of more than 90% of the individuals; only less than 10% of the individuals survive.

- **Sub Vital Genes:** These mutant genes reduce viability of the individuals carrying them in the appropriate genotype as compared to that of the normal individuals. Such genes kill less than 90% of the individuals in which they are present.

- **Vital Genes:** These genes do not affect the survival of the individuals in which they are present. These genes neither enhance nor reduce the viability of the individuals carrying them. It does not imply that these genes are not necessary for survival of the concerned organism - it simply means that the survival of the organism is not affected by the fact that the concerned vital genes are present in homozygous or heterozygous state.

- **Super Vital Genes:** These mutant genes enhance the survival of those individuals that carry them in the appropriate genotype as compared to that of the wild type allele. Genes for disease resistance in a crop species may be regarded as super vital genes as they protect the individuals carrying them against the concerned disease and thus increase the chances of their survival in the presence of concerned disease.

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9.3 CONCEPT OF GENETIC POLYMORPHISM AND SELECTION

Genetic Polymorphism: Individuals in a population differ in different counts. There might be purely environmental differences like the person's language, there might be purely genetic differences like the blood type, or there might be differences due to interactions between heredity and environment, like the person's intelligence.

Usually phenotypic variations, i.e., the morphological variations are considered to be genetic variations. But, this is not true in all the cases. In fact, genetic differences far outweigh phenotypic variations. Many genetic variations are concealed at the levels of DNA, or Chromosome of the cells and are not necessarily expressed as phenotypic variations which are amenable to direct observation. This variation in a population in the genetic material is generally termed as Genetic Polymorphism.

Polymorphic traits are discrete and discontinuous genetic characters which are controlled by single genes and are less influenced by environmental factors. These polymorphic traits are different from those phenotypic traits which are graded or continuous variations that are controlled by many genes and are strongly modified by environment. An example for the earlier case is the ability to taste PTC and for the latter case, the height and skin color of a person.

The populations which are characterized by individuals with discrete, discontinuous genetic traits are said to be polymorphic and those which lack such individuals are said to be monomorphic. A population with both tasters of PTC and non-tasters for PTC is said to be polymorphic and those with only the tasters or only the non tasters is said to be monomorphic. The concepts of polymorphic or monomorphic populations can be applied to both the genotype as well as the phenotype.

Definition: Genetic polymorphism can be defined as the occurrence of more than one morph (a gene) in a population. In order to be classified as genetically polymorphic trait, a character must be present in a frequency greater than 1%. If it is present in a frequency below this level, it is supposed that it is arising due to mutation and selection is not operating on it. It is only above the ratio of 1% that we can be sure to say some selection is involved in its maintenance and it is not because of recurrent mutations.

Source of Genetic Polymorphism: Any population is not entirely dependent on mutation for genetic polymorphism to develop, though it is the ultimate source. A small rate of mutation cannot maintain the observed genetic polymorphism in a population. The supply of genetic polymorphism upon which natural selection acts, is provided by a variety of genetic combinations built up over many generations by the flow of genes from other populations, the migration, genetic recombination and as a result of interaction between mutations and natural selection. Thus, genetic polymorphism can be brought about by activity of many factors.

Levels of Genetic Polymorphism: Genetic polymorphism can be seen in different levels of the heredity material of the population. It can be seen at the level of the cell, DNA, Proteins, Chromosomes, etc. At the cellular level, we can consider the polymorphism of the antigenic molecules. For example, the antigens in the human ABO blood systems are polymorphic. There can be four conditions for this antigen complex - A, AB, B, and O. Chromosomes have a capability to undergo rearrangements. These include deletions, insertions, additions, inversions and translocations. These rearrangements usually characterize differences in the chromosomal behavior and can also provide adaptive advantages to the populations housing them. There are also instances of chromosomal fusions. Different types of chromosomal rearrangements may involve drastic shifts in the character manifested, thus providing opportunities to agents of selection to act and effect major evolutionary consequence.

Genetic polymorphism can also be seen at the level of protein molecules. There are many instances where there are structural differences in the molecules of proteins. These polymorphic proteins also might provide an adaptive advantage to the populations having them. Similar is the case with certain enzymes.

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GENETIC POLYMORPHISM AND SELECTION

Most of the inherited differences in human beings represent differences in the structure of DNA. These differences in the DNA are due to the changes in the sequence of bases. It has been found out that all such differences in the DNA may not be important for natural selection to act to affect Darwinian fitness. However, too much of genetic variation or polymorphism might increase the effect of genetic load and the latter is responsible for thinning of the population. Due to the excess presence of genetic polymorphism, the average fitness rate of the population might get affected.

According to Kimura, the vast genetic polymorphisms at the gene and protein level involve only alteration of structure, being equivalent at functional level. Such variants, being functionally equivalent, would respond similarly to selective forces. Such polymorphisms are referred to by Kimura as "Selectively Neutral" polymorphisms.

Contrarily, for some other scientists, polymorphisms have a selective basis and might be useful in some way for different shades of environment. It is also useful for inbreeding and prevents a gradual build up of homogeneity in the population which is considered to be deleterious. According to these scientists, genetic polymorphism is very advantageous to the population and hence is not Selectively Neutral as according to Kimura.

Moreover, certain characters in the population are more or less permanently polymorphic. Sex is one such polymorphic feature. Stable polymorphic features are maintained in the population by stabilizing natural selection. Stabilizing selection removes from the population those individuals that deviate from the population mean. Thus individuals not perfectly male or female are removed from the population so that population shows only two discrete individuals with reference to sex - male or female.

Polymorphism may be in a transitory phase in the sense that certain populations with shifting environmental demands shift their population for some character in the direction of environmental change. At a time when one form has not replaced the other completely, a polymorphic population may exist. But gradually over the time this transient population may be replaced by monomorphism by directional natural selection.

Natural selection prefers heterozygotic condition when it considers them to be superior to the homozygotes. Polymorphism provides a population with alternative set of traits of a character. In a stable environment it may be of little significance. But under conditions when the environment is changing, populations with different polymorphic traits can explore for an adaptive advantage. A majority of polymorphic traits exist in the human populations. Most of their potentialities can only be assessed in the future environments.

MENDELIAN POPULATION

The term "Population" means different things to different persons. In its dictionary sense, the term implies a collection of times. To a biologist, it is an aggregation of similarly adapted organisms. A geneticist uses the term to signify a "spatial - temporal group of conspecific (i.e., belonging to the same species) interbreeding individuals" - Mather and Gregg. Population geneticists restrict the definition, further, to a population of genes rather than to that of individuals. The population of genes under consideration is termed a Gene Pool and represents the total collection of all the allelic forms that are carried by all the gametes of a population. It is the total genetic information possessed by a population. The basic unit of such a population has been termed variously as panmictic by Sewall Wright, Local Mendelian Population by Dobzhansky and Gamodeme by Gilmour and Gregor. The term that is mostly in vogue today for a genetic population is the Deme. A deme represents a local population of interbreeding, or potentially inbreeding, individuals in a given locality; that is, any gamete in this population has an equal chance to unite with any gamete of the opposite sex in the same population and result in a viable zygote. An ideal deme is, of course, a panmictic one.

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Successful reproduction requires sexual behavior culminating in copulation, fertilization, normal development of the fetus, and offspring that are normal and healthy, capable of reproducing in turn. A number of conditions can prevent closely related populations from exchanging genes by preventing successful reproduction. One of the many ways this may occur is if the sexual behavior of the male, such as the mating dance of birds, fails to stimulate the female of a closely related population. Several forms of reproductive isolations are already discussed in the chapter on Modern Synthetic theory.

Of course, successful reproduction of a population requires a rate of reproduction sufficient to sustain the population. The number of individuals produced in each generation must be great enough to compensate for deaths due to accident, predation, disease, etc. However, the rate of reproduction cannot be so great that the population will increase to the point where it can no longer be supported by its food sources or other elements in the environment. In other words, a successful reproductive rate is one that maintains a balance between population size and the potentials and limitations of the environment.

The largest natural population is the species, the members of which are potentially capable of successful reproduction among themselves but not with members of other species. Species can be broken down into smaller reproductive populations, which are to some degree, and often temporarily, isolated from one another.

Phenotype in a Mendelian Population: Just as one can speak of the phenotype of an individual, it also is possible to speak of the phenotype of a whole population. Since a population is made up of varied individuals, such a description must be handled statistically. For example, the average stature of a population can be calculated and the variation from that average noted. It is also possible to calculate the percentage of blood type O, blue eyes, red hair, and so on, and emerge with a statistical profile.

Genotype in a Mendelian Population: It also is possible to speak of the collective genotype of a population. This is known as the gene pool. The frequencies of genes in a gene pool can be calculated by formulae which we shall discuss later. Each individual can be thought to contribute one cell to the gene pool. From these cells the genes can be extracted and tallied. It may be noted, for example, that if the alleles for PTC tasting in a population are tallied, 41.3 percent of them are dominant (T) and 58.7 percent are recessive (t). On further examination it might be found that 17.1 percent of the genotypes that emerge from the gene pool of PTC genes are homozygous dominant, whereas 48.4 percent are heterozygous and 35.4 percent are homozygous recessive. The frequency of these genotypes makes up a statistical statement about the genotypes of the population.

POPULATION GENETICS

The need to understand the behavior of genes in the gene pool rather than in individuals is important to several areas of theoretical and applied biology.

Take the case of the agricultural practice of breeding for improvement of cultivated or domesticated stocks of plants and animals.

1. It was observed that variety and vigor could be maintained only if there existed a certain degree of heterozygosity in the strains that are crossed. The question naturally arose; How does one ensure the optimum amount of heterozygosity or desirable traits in stocks of interest? To answer this, it is necessary to know how genotypes are maintained in populations, as well as the factors that affect their maintenance. Such knowledge is expected to lend to the development of more rational methods of stock improvement.

2. A second area that benefits from knowledge of genes in a population is human genetics. For obvious reasons, man is not amenable to genetic experimentation in the conventional manner. However, it has been possible to glean a great deal about the distribution and dynamics of human genes by combining analyses of pedigree charts and of distributions of genes in finite populations. The methods of population genetics have been particularly fruitful in the detection of rare inherited

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disorders and of infrequently occurring but hereditary behavioral aberrations. One of the obsessions of the biologists has been to understand how these deleterious alleles continue to exist in the population generation after generation.

3. Perhaps the greatest impact of population genetics has been in stimulating a search for realistic models that explain the process of evolution. Since evolution represents a shift in the genetic structure of a population, the most obvious way to understand evolution is to figure out how gene frequencies remain stable or change in response to environmental persuasions. This brings up the intriguing questions about how a strain manages to adapt itself to vastly different environments. Such adaptation is not the feat of an individual, but of an entire population. How is this achieved? Adaptation follows changes in several gene loci and is hardly expected to occur in a single individual. Imperceptible alterations in allele distribution in members of an inbreeding population can, however, become included in a large section of the population over a stretch of time. Once again, studying alterations in individual genotypes is not enough to provide an understanding of the dynamics of allele distribution in an evolving population.

4. A very useful spin-off of investigating the behavior of genes in populations has been the development of sophisticated mathematical methodologies which are of intrinsic interest even to mathematicians not concerned about their immediate practical applications. Mathematical models have been presented from time to time and refined as required to handle the complexities of dealing with several variables of alleles and environmental and other governing factor. These models try to depict the essence of the rules that govern genes in population, and even if they are not exact representatives of the actual situations, they are useful for furnishing scaffolding which can be utilized to design further experiments and test the parameters and tenets of hypotheses. Indeed, many of the simple deductions from the studies of population genetics have emerged from a mathematical approach to biological problems. In each instance, a statistical population of genes, rather than of specific individuals, is taken into consideration. The most significant model that provided the initial impetus to an intelligent study of genes in the population is the one known as the Hardy-Weinberg Model or Principle.

To understand how genes behave in a population, geneticists study the effects of factors that affect the genetic heritage of populations both spatially and temporally. The bewildering diversity of living forms is sufficient evidence that the genetic heritage of a population is not static. However, to study the dynamics of alleles in changing populations, a reference model would be convenient. Hardy and Weinberg provided such a standard against which deviations in population structure can be measured. The Hardy-Weinberg model represents a static genetic population, which is one in which genes undergo no change, so that its allele and genotype frequencies are in equilibrium, generation after generation.

Hardy, a mathematician, and Weinberg, an experimental biologist, independently arrived at the conclusion that, if certain disruptive factors are not in operation, the relative proportions of alleles of a gene locus remain in a constant proportion to each other in every succeeding generation. The Hardy-Weinberg law states that in a panmictic - randomly mating - population, without disruptive influences, the frequencies of alleles of a gene remain unchanged in every generation, but if this equilibrium is disturbed in some manner, the genotype frequencies reach a new equilibrium value in one or more generations, depending on the nature of the disrupting influence.

Although such an ideal population, or conditions, hardly exist, if at all, in natural environments, the Hardy-Weinberg law has served as a fundamental theorem in population genetics by supplying a norm or standard against which actual framework of gene frequencies can be measured.

In summary, population genetics deals with the dynamics of genes in potentially inbreeding communities. The frequency of genes and genotypes, and, therefore, the genetic structure of a population, remains stable from generation to generation, provided evolutionary agents do not disrupt them. Subtle changes occur, however, due to the disruptive forces that, in time, contribute to the segregation of a section of a

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gene pool into a population with an altered genetic structure. If the alteration is significant enough to prevent breeding with the parent population, the new set is considered to have acquired the status of a species. In short, an evolutionary change has taken place.

With this overview of population genetics and Mendelian population we proceed to brief descriptions of some of the fundamental concepts, methodologies and findings in this aspect of genetics which has generated tremendous interest in biologists and mathematicians.

GENETIC EQUILIBRIUM

A population is said to be evolving if the frequencies of its alleles are changing; it is not evolving if these frequencies remain constant. This latter situation is termed Genetic Equilibrium.

Certain conditions must be met with if the population is to remain in a genetic equilibrium.

1. **Random Mating:** Random mating denotes a union between gametes without reference to their genotypes. In random mating populations (panmictic), every gamete is assumed to have an equal opportunity of fusing with any other gamete of the opposite sex, resulting of course in viable and fertile offspring. In natural populations this condition may not be present. For, although any member of this population has the potentiality to breed with any other member of the opposite sex, the logistics may prevent such panmixis, with mating restricted within members confined to each other's vicinities. If such a population is further constrained in choosing partners, due to inbreeding, a significant alteration in gene frequencies will be noticed in comparison of those in the wider or bulk population. So, gene frequencies of non-panmictic and inbreeding populations will not be representative of those for the total gene pool of the species.

2. **Large Population Size:** Gene frequencies calculated from populations below a certain sample size may not be representative of the entire species. There will be significant sampling errors if the populations are not large enough. More over in the populations which are small, sudden random changes might have a significant evolutionary impact by drastically changing the gene frequencies.

3. **Lack of evolutionary forces:** Forces of evolution like mutation, migration, natural selection and random drift may alter the frequencies of the genes and alter the genetic structure of the population. How these alter the gene frequencies will be shortly discussed. However, Hardy-Weinberg Equilibrium assumes that no evolutionary forces are active in the population.

4. **Mating should be equally fertile:** The mating in the populations must produce the same number of viable offspring.

Deviation from the equilibrium shows that one or a combination of these conditions is not being met.

HARDY - WEINBERG EQUILIBRIUM

As already stated, mathematical calculations were independently made to understand whether a population is in genetic equilibrium, or not, for the first time in 1908 by Godfrey Hardy and Wilhelm Weinberg. Hence, genetic equilibrium is also termed Hardy - Weinberg equilibrium. The concept of Hardy-Weinberg Equilibrium can also be described algebraically. This results in the following general formula for the Hardy-Weinberg equilibrium.

$$p^2 + 2pq + q^2 = 1$$

Where p = [A] and q = [a], from which it follows that [AA] = p^2; [Aa] = 2pq; [aa] = q^2

Here 'A' and 'a' are the allelic forms of one gene. Brackets are used to indicate "frequency of". The model of Hardy - Weinberg equilibrium presents a hypothetical situation of no change. It is a fact that

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populations do change that makes the model important. By being able to specify under what conditions a static condition would exist, we have already seen the requirements for it to exist.

In the above model, let us take the example of the pea plant experiment already discussed in the beginning of this chapter on Mendelian inheritance. In the first filial generation we have observed that the four possible genotypes in the offspring (after a cross between a genetically pure tall plant (TT) and a genetically pure dwarf plant (tt) are TT Tt Tt tt

Here,

The frequency for the alleles TT i.e., [TT] = ¼
The frequency for the alleles Tt i.e., [Tt] = 2/4
The frequency for the alleles tt i.e., [tt] = 1/4

This pattern of allelic occurrence has already been discussed in the phenotypic ratio of 3:1.

However, in the context of Genetic Equilibrium, the same allelic occurrence can also be interpreted in a different manner using the Hardy-Weinberg equation, mentioned above.

Thus the allelic frequencies of TT, Tt and tt add up to 1. This is an ideal model for the genetic equilibrium.

However, no population is a perfect ideal type. There are certain forces which are responsible to bring about a change or disturbance in the Hardy - Weinberg equilibrium. These forces are discussed hereunder.

CAUSES AND CHANGES IN GENE FREQUENCY

Natural populations are subjected to forces that prevent a population from maintaining an invariant genetic equilibrium. Most studied of such forces are mutation, migration, selection, random drift and inbreeding. We shall now consider each of these forces in turn.

1. MUTATION

Variation is the basic ingredient for evolution. The larger the genetic variation available in a population, the greater the raw material at the disposal of selection agents. The primary source of genetic variation is, of course, gene mutation. Forward and reverse mutations are continuously adding new genetic material to a population. Variation can also be the outcome of genetic recombination events and of alterations in number and/or structural organization of genetic units larger than the unit of gene - such as chromosomes.

Recurrent mutation is a one way mutation by which mutation takes place from one allele to another in only one direction, i.e., A to a or a to A, and no reverse mutation occurs. Such persistent alterations of one allele into another would obviously erode the frequency of the original allele in the gene pool and increase that of the newly formed allele. However, under natural conditions, the rate of mutation in higher organism is extremely low and cannot possibly be responsible by itself in altering the frequencies of genes in a significant manner. Nevertheless, the rate at which this slow conversion of one allele into another takes place can be calculated and the value obtained is termed as the mutation pressure. For example, if the initial frequency of A is p_o and "u" is the rate of mutation from A to a, in the first generation, the frequency of a becomes u X p_o and that of A is lessened and becomes p_o - u p_o, i.e., p_o (1-u) in one generation and for n generation, it will be p_o (1-u)n. This is, usually, of a very low value - say in a 1000 generations, an allele A with frequency p = 0.9 may change to 0.899.

When both forward and reverse mutations occur, the effective proportion of alleles at any time will be given by the difference in rates of the mutation in the opposite directions. So, if mutation rate of A to a = u and mutation rate of a to A = v, in such a population, an equilibrium between frequencies of A and a will be reached when u = v, or the value of p_o (1-u) remains constant over a period of generations.

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2. SELECTION

There is a natural discrimination against survival and / or mating success of certain allelic combinations in particular environments, so that only one or more specific combinations are favored. These combinations turn out to be the ones that allow the strain to adapt itself to specific niches. In other words, there is a subtle competition among genotypes, only some of which provide optimum traits for surviving successfully in preferred situations. In short, certain combinations of genes undergo a process of selection, the idiosyncrasies of the environmental niche acting as the selection pressure.

Particular genes of the genetically fitter individuals will, therefore, increase in frequency. Such fitness for survival is what Darwin indicated as the main criterion for adaptive evolution. The traits that make the successful offspring well adapted are presumed to accumulate over generations and slowly but inevitably change the genetic structure of a population. The alteration may result in the delineation of a species or of a higher category - genus, family, and phylum.

Selection may thus be viewed as

1. A process that generates greater diversity in genotypes or
2. A means of reducing variability by elimination of genotypes Sub optimal in fitness for adaptation.

Selection may be induced by a variety of physical, chemical and biological agents. Temperature, light intensities, availability of food in general or minerals in particular, presence or absence of predators and a host of other factors may act as selection pressures, persuading the direction in which selection will occur.

Measure of Selection Force

Selection may be measured by its fitness for the environment. The fitness of a genotype is an expression of the ability to produce viable and fertile offspring - i.e., of its reproductive success. Another way of indicating the reproductive ability of a genotype is by assuming a value termed selective value or adaptive value.

The selective value of a genotype is the measure of offspring produced by it, relative to those of another. If, for instance, a genotype produces 100 viable and fertile offspring, and another only 70, the reproductive capacity of the latter is less than that of the former by 30 percent. In abstract terms, the ratio of offspring of a genotype that are viable and fertile as compared to those of other genotypes is designated by the letter "w". The most successful genotype can be assigned a value of 1 for "w", while the least successful, or lethal genotypes with w = 0. Other values range between 1 and 0. The discriminatory selective force which lessens the adaptive value of a genotype from the ideal is termed the selection coefficient, "s". In the above example, `w' for the most successful genotype has no selection coefficient working against it, so that s = 0, while for the less successful genotypes is 0.3. In other words w = 1-s and s = 1-w.

Usually, the most favored genotype is assigned the value of 1 and others are assessed relative to this norm.

Selection occurs on whole genotypes of individuals and not against individual alleles. However, if several individuals have certain combinations of alleles in common, one may speak of selection for or against some of these allelic combinations. An allele a for instance may be selected for in a particular setting of genes, while it may be selected against in another. The selective value, therefore, depends really on the interrelationships in the expression of all the genes in a genotype and not specifically on one or more alleles.

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3. MIGRATION AND ISOLATION

Mutation, by itself, is not a very effective force in changing frequencies of alleles in a population, primarily due to its very slow rate. A much faster way of altering gene frequencies is provided by migration. A group of individuals from another population, which can interbreed with the recipient population, and which has an allelic frequency pattern distinct from that of the population with which it is merging, will quickly affect the original allelic frequencies in the recipient group. The difference in the recipient and migrant allele frequencies is a consequence of adaptation to particular environmental niches over long periods of time. To become optimally adapted to a locale, certain alleles are favored and retained in larger proportions than others which are selected against. As conditions in different environs require different traits for optimum efficiency of living and reproducing, such isolated populations develop characteristic differences in the allelic frequencies of the same gene.

Let us consider a group of migrants entering an isolated population. The following figures are to be considered. We will consider the frequency of an allele say "a".

Total number of Recipient population = M

Total number of migrant population = m

Frequency of the allele a in the recipient population = q_a

Frequency of the allele a in the migrant population = q_b

Hence, the

Total number of alleles a in the recipient population = Mq_a

Total number of alleles a in the migrant population = mq_b

Hence, the

Total frequency of the allele a in the mixed population $Q = q_a+q_b$

Total number of the alleles a in the mixed population = Mq_a+mq_b

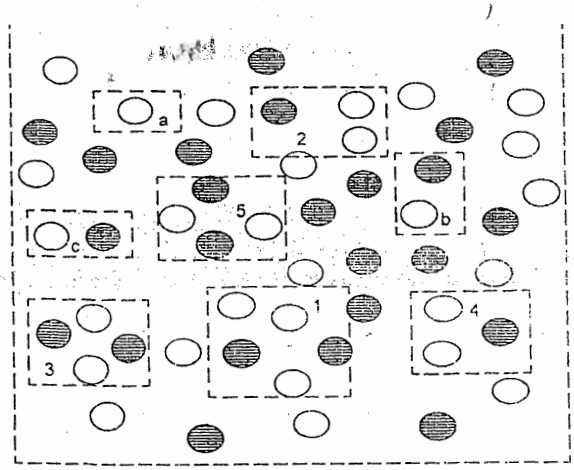
Thus the change in the frequency of the allele after migration can be denoted by $Q^\wedge = q_b - q_a$

If the frequency of allele a in the migrants is higher than that in the recipient, the value of q^ will have a positive value and in the converse case, a negative one. The differences between the two frequencies are a measure of migration and are called as the migration coefficient.

Migration is often referred to in terms of migration-pressure of gene flow and is a measure of the migration coefficient and difference in gene frequencies. Migrations between isolated populations of the same species bring about a greater variation within that species.

4. GENETIC DRIFT

Any population, no matter how large, is a finite one (of N individuals), and may not necessarily reflect the pattern of allele distribution in the more global gene pool. In the figure given the large square represents the total gene pool, and the smaller squares (1, 2, 3, 4, and 5) represent various portions of this larger pool. One will notice that not all the squares give a correct picture of the ratio of black and white circles for the entire pool. It is also evident that the smaller the chunk chosen,



A SCHEMATIC REPRESENTATION OF RANDOM DRIFT

LARGER SAMPLES : 1, 2, 3, ... SMALLER SAMPLES : a, b, c .....

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the greater is the chance of it being an atypical segment.

Such a typical sets of a gene pool may lead to an altered genetic structure of the population. The extent of deviation is seen to occur within certain predictable limits. The results are not very much unlike those in inbred populations especially in the tendency of one or more alleles to become fixed, or become homozygous in most members of the population. Such fixation or homoallelism is more likely to occur in smaller population.

Sewall Wright's Studies:

Sewall Wright was the first to investigate the extent and effect of random genetic drift. For this reason the drift is often referred to as the "Sewall Wright Effect".

Sewall Wright developed mathematical models for assessing deviations in gene frequencies in generations. According to this model, the deviation from mean frequencies of alleles can be represented by the equation

$$\Sigma = pq/N$$

Where Σ (Sigma) represents standard deviation, p and q are frequencies of alleles A and B, and N is the size of the population.

5. INBREEDING

One of the chief conditions for realizing the Hardy-Weinberg equilibrium is that the population under consideration breeds randomly. In actual circumstances, though, ideal random mating is a rarity. The reasons for this are not difficult to imagine. Although, there may be a large settlement of individuals of the same interbreeding species in a locality, the mating between individuals are usually dictated by proximity, choice and other factors, and are restricted, to a small group of individuals in the immediate neighborhood. So, although, theoretically the entire population is at the disposal of an individual, it rarely uses this wide opportunity. As a result, two extreme types of mating happen to be quite common. These are inbreeding or mating between closely related individuals, and out crossing - or mating between very much unrelated individuals.

Both forms of mating have been used in selections, for the specific purpose of producing desirable varieties of plants and animals. When close relatives are bred together, homozygosity is attained very easily in several loci. If this breeding is continued systematically for several generations, the progeny will separate out into separate stocks, each homozygous for specific alleles. These are the true-breeding stocks of breeders. A defect of this procedure is that together with the desirable alleles, many other non-desirable (deleterious to the organism) alleles also become homozygous. The total effect of such homozygosity is a weaker, or less successful, or less fit member of the species. Inbreeding, thus, lowers the fitness or adaptive value of species. If suddenly called upon to cope with a new set of circumstances, such an inbred stock would be unable to do so, since many of the genes that could have helped in this new situation have been lost.

One method in nature, therefore, of maintaining sufficient genetic variability in a population is to ensure crossbreeding. Many mechanisms have been evolved which promote cross pollination or cross fertilization. The offspring of cross bred individuals have been found empirically to be stronger or better products than those of inbred lines. This fact was exploited by breeder geneticists in producing unusually vigorous progeny from widely different parents. This excess superiority or vigor, or increase in degrees of expression of several traits, has been named heterosis, or hybrid vigor, and has been exploited commercially.

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CONSANGUINEOUS AND NON-CONSANGUINEOUS MATING

The simplest type of mating is called as Random Mating. In this type of mating, an individual of one sex has an equal probability of mating with any individual of the opposite sex in the population. This type of mating is also called as Panmixis.

However, random mating is prevented by certain factors. Such factors, to name some are the mate preferences and social factors. Random mating forms the foundation of population genetics as was discussed by Hardy Weinberg Law. A deviation from random mating is Inbreeding.

Inbreeding is a departure from random mating and it is the mating between genetically related individuals. It is a situation of Genetic Consanguinity. It was discovered by the geneticists that close inbreeding is detrimental to the offspring and cross breeding is, on the other hand, beneficial. Perhaps for the same reason, the laws and customs of the people still forbid marriage of certain near relatives.

The closest type of inbreeding in the bisexual organisms or the hermaphrodites is Self fertilization. In the unisexual organisms, the closest type of inbreeding is Back Cross. This is the crossing to a single homozygous parental type. (Refer the topic on Mendel's laws). Self fertilization tends to make a population pure breeding according to the Mendel's laws of genetics. In inbreeding, in the genepool of the population, the frequency of heterozygotes decrease in size and that of homozygotes increase. Inbreeding, thus, does not give opportunity to the formation of heterozygotes. This is because there is no chance of new genes or alleles entering the genepool.

The chief advantage of cross fertilization over inbreeding is the greater opportunity for appearance of new and better combination of genes. Continuous inbreeding in the absence of cross fertilization will result in accumulation of injurious recessive genes because these rare recessive genes so far were concealed by normal dominant genes. The effect of inbreeding is more likely to be injurious than beneficial.

Hybridization or Gene flow is a means by which genes already present in some population are introduced into the other. It may result in genetic combinations which are entirely new. When we say that new genes are introduced in a population, it also means that genetic variations are introduced in it. This process is also called as Migration or Admixture.

Hybridization results when individuals from outside contribute their genes to the pool of a population and alter their gene frequency as a consequent. There may be a series of migrations between adjacent populations resulting in appearance of genes from a point of high frequency to that of a point of low frequency. A person may pass the gene to some one who will in turn repeat the process. This results in the spread of genes to vast distances while carriers themselves may remain relatively stable.

Such a kind of a Gene flow where a gene is spreading from a local population results in a gradually decreasing frequency along the line. This line indicates the direction and magnitude of Gene flow. This frequency gradient is called as a Cline. An example of this gradient is the epicanthic fold which flows from Himalayas to Orissa.

Usually well separated populations are connected by numerous intermediate groups through which genes can flow and give rise to a geographic gradient of gene frequency between two polar populations. Such gradients in gene frequency are known as Geno Cline and a gradient of trait is called as a Pheno Cline.

The migration of population into new territories may lead to contacts accompanied by gene exchanges between diverse populations formerly separated by great distances.

The formation of new gene pools by interbreeding two populations is a slow process. Populations may live side by side for hundreds of years and yet may retain genetic diversity. This can be possible only by the cultural factors like endogamy. Such factors restrict gene diffusions in one direction.

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Hybridization may also mean admixture of two or more genetically dissimilar population to form hybrids. Two new populations when come in contact, it is very rare they form a new panmictic unit immediately. This feature is particularly slow if the populations differ greatly in their physical features and social customs.

Hybridization or Gene flow is a basic evolutionary mechanism which brings about a change in the gene frequencies of a population by disturbing the Hardy Weinberg Equilibrium. It breaks the isolation and makes the gene frequencies remain similar. Isolation, resulting from the absence of Gene flow may permit genetic drift to cause a divergence. Gene flow slows the process of population differentiation as the genetic drift speeds it up. Gene flow hence can also be referred to as a stabilizing mechanism.

GENETIC LOAD

Genetic Polymorphism is not necessarily an unqualified boon to a natural population. The vast genetic diversity that is present obviously includes several alleles which do not impart optimum fitness to the population, and may, in fact, be quite harmful for its existence. Such deleterious genes are a sort of genetic liability to the population. This liability tends to decrease the potential optimum fitness of a population and is known as the genetic load of the population. Crow defines genetic load as the "proportion by which fitness of the average genotype in the population is reduced in comparison with the best genotype".

Types of Genetic Load

Three major types of genetic loads originate and are maintained in a population.

1. **Mutational Load:** It is the slicing away of the genetic variability of a species due to the occurrence of recurrent mutations. Deleterious genes are eliminated in an optimally fit population but beneficial or advantageous genes may also be eliminated, thus leading to genetic burden.

2. **Segregational Load:** It is represented in those situations where the heterozygotes are more successful in efficient living than the homozygotes. The inferior homozygotes which are continuously being segregated in each generation represent the liability the population carries without any immediate and obvious utility value. Segregational load can cause greater lowering of genetic variability than mutational load.

3. **Incompatibility Load:** It is encountered when certain genotypes are unable to survive in the environment of specific parental genotypes. The conception of an Rh positive child to an Rh negative mother is a common example of this type of genetic load. In this situation Rh hemolytic disease of the new born or Erythroblastosis foetalis is caused by Rh factor incompatibility between a pregnant woman and her fetus. It can occur only when the mother is Rh negative and the father is Rh positive. Because the Rh factor is genetically dominant, the resulting fetus may be Rh positive. The disease develops if fetal Rh factor enters the mother's circulatory system through a breach in the placenta, usually late in the pregnancy or during delivery. The mother's immune system treats the fetal Rh factor as a foreign protein and produces antibodies against it. The antibodies can pass through an intact placenta into the fetal circulation and cause a potentially fatal anemia by destroying the RBC.

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GENETIC EFFECTS OF CONSANGUINEOUS AND COUSIN MARRIAGES

NON RANDOM MATING

Genetic recombination does not by itself alter allele frequencies. However, any consistent bias in mating patterns can alter the genotypic proportions. By affecting the combination frequencies of genotypes, nonrandom mating causes deviations from the Hardy Weinberg expectations. It therefore sets the stage for the action of natural selection.

Basically, there are three models of nonrandom mating – positive assortative mating, negative assortative mating and inbreeding.

Positive Assortative Mating: This occurs when individuals of like phenotype mate more often than expected by random mating predictions. Because individuals with like phenotypes are also similar to some degree in genotypes as well, the result of positive assortative mating increases the amount of homozygosity in the population and reduces heterozygosity (P^2 and Q^2 greater than expected; 2pq less than expected). Ex. Stature and IQ or Eye color.

Negative Assortative Mating: This involves mating with an individual who is phenotypically dissimilar. Theoretically, if this occurs more than expected by random mating predictions, it should increase the amount of heterozygosity in the population while correspondingly reducing homozygosity.

INBREEDING

Inbreeding, also called consanguinity, occurs when relatives mate more often than expected. Such mating will increase the amount of homozygosity, since relatives who share close ancestors will more than likely also share similar genes. Inbreeding occurs less frequently than predicted under random mating because all societies have some form of incest taboo, especially nuclear family. Inbreeding reduces the variability among the offspring thereby reducing the reproductive fitness. In some societies, due to the small number of potential mates available, inbreeding among fairly close relations such as cousins is actively encouraged or is unavoidable. Ex. Pitcairn Island and Tahitian wives. There are some areas, on the other hand, where inbreeding is avoidable but it is still actively encouraged. In some parts of Japan among certain social classes, first cousin marriages make up almost 10% of all marriages, and in Andhra Pradesh of India, among certain castes, uncle/niece marriages also make up about 10% of marriages

However, such considerable inbreeding is an exception rather than a rule among human populations and most groups actively work very hard at maintaining exogamy. As a general rule, then, most human populations do not inbreed if they can at all help it. Inbreeding has important medical consequences in addition to its effect on genetic equilibrium. When relatives mate, their offspring have increased probability of inheriting an allele in homozygous dose.

Any departure from random mating naturally leads to complications in the relationships between allele frequencies and genotype frequencies. If a heterozygote Aa is self-fertilized, it will produce three kinds of progeny AA, Aa and aa, in the ratio of 1:2:1. At this stage, the frequency of the heterozygote is 0.5. If self-fertilization is continued for another generation, the homozygotes will breed true but the heterozygotes will again segregate, reducing their frequency to 0.25. With every succeeding generation of self-fertilization, the frequency of the heterozygotes drops off by 50%, reaching 0.008 by generation seven and 0.001 by generation ten. At this stage, the population is 99.9% homozygous.

The process of repeated self-fertilization illustrates the general effects of a form of nonrandom mating called inbreeding. Inbreeding occurs when the mates are genetically related. Self-fertilization is the extreme example, but other examples such as the mating of full siblings, first cousins, parents and offspring, and half siblings have the same effect, namely, to increase the frequency of homozygotes and decrease the frequency of heterozygotes. As a result, the Hardy Wienberg method does not hold.

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9.4 CHROMOSOMES AND CHROMOSOMAL ABERRATIONS IN MAN

Diagnosis

Preparing chromosomes for analysis is relatively simple. Circulating blood lymphocytes are generally used, except in the fetus, in whom amniocytes from amniotic fluid or chorionic villus cells from the placenta are used instead. The cells are cultured in the laboratory with phytohemagglutinin to stimulate cell division. Colchicine is then added to arrest mitosis during metaphase, when each chromosome has replicated into two chromatids attached at the centromere. The cells, which are spread onto microscope slides, are then stained. Chromosomes from single cells are usually photographed, their images cut out of the print and pasted onto a piece of paper, forming a karyotype. Computer imaging can also be used to produce a visual display of the chromosomes.

Chromosome staining is performed by G (Giemsa) or Q (fluorescent) banding techniques. Additional staining procedures and techniques for extending chromosome length have greatly increased the precision of cytogenetic diagnosis.

New molecular techniques use DNA probes (which can have fluorescent tags) to locate specific genes or DNA sequences on the chromosomes. *F*luorescent *I*n *S*itu *H*ybridization (FISH) is used to identify the organization of genes and to look for deletions, rearrangements, and duplications of the chromosomes.

Karyotype nomenclature is as follows. The normal male is designated as 46,XY and the normal female as 46,XX. In Down syndrome due to an extra chromosome 21 (trisomy 21), the notation is 47,XY,21+ for a male and 47,XX,21+ for a female. In Down syndrome due to a translocation (two chromosomes that are stuck together), the typical 14/21 "balanced translocation carrier" mother is written as 45,XX, t(14q;21q). The translocation chromosome (t) is formed from 14q and 21q (in which q is the long arm); the short arms (p) are lost. For a deletion of the short arm of chromosome 5 (as in the 5p deletion syndrome), the female karyotype is 46,XX,5p-.

Each arm of a chromosome is divided into one to four major regions, depending on chromosomal length; each band, positively or negatively stained, is given a number, which raises as the distance from the centromere increases. For example, 1q23 designates the chromosome (1), the long arm (q), the second region distal to the centromere (2), and the third band (3) in that region.

Chromosomal aberrations have been classified into two types depending upon the type of changes they bring about i.e., changes in the chromosomal structure and in the chromosomal number. Apart from this, you are expected to also know the various types of chromosomes in the human body, the insight into which will provide a better understanding of the various chromosomal disorders in man.

In all the sexually reproducing organisms, chromosomes exist in homologous pairs. In the humans, the organism of our interest, there are 23 pairs of chromosomes (total 46 in number). Not all these chromosomes are similar in their structure and function. They differ in their structures and vary in their functional attributes. These functional aspects of a chromosome depend upon the genes each chromosome carries. All the genes present on the same chromosome are said to be "linked" and form a linkage group.

In many sexually reproducing organisms, including the humans, one pair of chromosomes found in the male of the species does not fit the description of homologous chromosomes. The members of this unique pair neither look alike nor seem to carry genes that are allelic. But they are of vital importance because the very existence of the species depends on their presence, for they determine the sex of the individual, they are the sex chromosomes and the others are termed as autosomes.

The tiny chromosome that determines the maleness in the human beings is the Y chromosome. When the Y is absent, the individual is female.

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The X and Y chromosomes segregate as a homologous pair during gametogenesis. Following spermatogenesis, half the mature sperms therefore contain Y chromosomes, and half carry the X chromosome. Since the mature ovum, or egg cell, can have only the X chromosome, the sperm which fertilizes an egg will determine the sex of the child conceived. The Y bearing sperm determines that the child is a boy and the X bearing sperm determines that the child is a girl. This means that the father is the parent whose germ cells determine the sex of the children.

With this background, we shall proceed with the discussion on the various types of chromosomal disorders in human being.

NUMERICAL AND STRUCTUAL ABERRATIONS

Numerical aberrations of chromosomes may involve the entire set, a condition called euploidy, or individual chromosomes, called aneuploidy. Aberrations involving individual chromosomes i.e., aneuploidy, are indicated by the suffix - somy. Thus, the presence of an extra chromosome 21 is called trisomy 21. An individual possessing only one rather than a pair of 21 chromosomes would be monosomic 21.

Although polyploidy (extra set of chromosomes) is known to be a viable condition in other organisms, such as some plants, it seems to be fatal to the humans. There are however a number of conditions found in humans resulting from aneuploidy of both autosomes and sex chromosomes.

Structural changes require breaks in the chromosomes. More than one break can occur in a single chromosome or set of chromosomes, and the broken parts may then reunite in new arrangements. Any broken end may unite with any other broken end, thus potentially resulting in new linkage arrangements. The loss or addition of a chromosome segment may also occur in the process. More than one type of aberration may occur at the same time. For example, a section may be broken off and lost during the formation of an inversion or translocation; thus simultaneously producing a deficiency.

SEX CHROMOSOMAL ABERRATIONS

KLINEFELTER'S SYNDROME

Non disjunction produces gametes with extra sex chromosomes; one would also expect trisomy of the sex chromosomes if the gametes containing an extra sex chromosome took part in fertilization. Conditions resulting from trisomy of the sex chromosome in fact occur even more frequently than the Turner's syndrome.

If a sperm from nondisjunction were to fertilize a normal X bearing one, and if XX ovum rising from nondisjunction in oogenesis were to be fertilized by a normal Y bearing sperm, the zygotes in each case would be XXY.

This imbalances in the number of sex chromosomes leads to a series of abnormalities known collectively as Kleinefelter's Syndrome.

An extra X chromosome in addition to the usual male XY chromosome complement (47, XXY) has been associated with the abnormal male syndrome described by H.F. Kleinefelter. It is estimated to occur in 1 per 500 live births. Individuals with this syndrome are phenotypically males but with some tendency toward femaleness, particularly in secondary sex characteristics. Such features as enlarged breasts, underdeveloped body hair, small testes, and small prostrate glands are a part of the syndrome. Presumably, the XXY constitution originates either by fertilization of an exceptional XX egg by a Y sperm or of an X egg by an exceptional XY sperm. Studies of Kleinefelter's syndrome and Turner's syndrome indicate that the Y chromosome in human beings determines male sex.

The most common karyotype for Kleinefelter's syndrome is 47,XXY, but the symptoms of the syndrome will usually occur whenever more than one X chromosome is present along with a Y chromosome. More

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complex karyotypes associated with the Kleinefelter's syndrome include XXYY, XXXY, XXXYY, XXXXY, XXXXYY and XXXXXY. Mental retardation is usually found when there are more than two X chromosomes. Thus this syndrome brings about both physical and reproductive anomalies.

TURNER'S SYNDROME

This monosomic condition has a chromosome number of 44 autosomes and one X chromosome. The chromosome anomaly is associated with an abnormal female phenotype described in 1938 by H.H. Turner and associates and known as Turner's syndrome. It occurs in about 1 in 2500 live females' births. More than 90 percent abort spontaneously. These adults have virtually no ovaries, have limited secondary sexual characteristics, and are sterile. Microscopic sections of the ovaries show fibrous streaks of tissue representing remnants of ovaries. Affected females have short stature, low set ears, webbed neck, and a shield like chest. Mental deficiency is not usually associated with this syndrome. Since there is a loss of one sex chromosome (X) thus resulting in one less chromosome in the karyotype i.e., 45 instead of 46, Turner syndrome can also be represented as (45, X), whereas the normal is (46, XX).

These patients i.e., the X monosomics probably originate from exceptional eggs or sperm with no sex chromosome or from the loss of a sex chromosome in mitosis during early cleavage stages, after an XX or XY zygote has been formed. Most of the cases of somatic Turner's phenotype have normal X chromosome and a fragment of a second X chromosome. Both arms of the second X chromosome are apparently necessary for normal ovarian development and differentiation. Individuals with only the long arm of the second X chromosome are short in stature and show other somatic symptoms of Turner's syndrome, whereas those with only the short arm of the second X have normal stature and do not show as many signs of Turner syndrome. This indicates that the Turner's phenotype is mostly controlled by genes on the short arm of the X chromosome.

A Y chromosome also occurs in some individuals with one X and a fragment of Y, not including the short arm of the Y chromosome. These individuals have streak ovaries but are normal in phenotype. This suggests that male determining genes are in the short arm of the Y chromosome, and those that prevent the Turner's phenotype is in the Y long arm as well as the X short arm. Major features of the Turner's syndrome are characterized by defective development of the testes, sterility, and limited male secondary sexual characteristics, along with somatic features of the Turner's phenotype.

The Turner's syndrome individuals result from nondisjunction of the sex chromosomes, both male and female. In both the cases, if the Nullisomic gametes (0) were involved in fertilization with an X bearing gamete from the other parent, the resulting zygote would be XO.

XXX SUPER FEMALES

Fertilization of XX germ cells by X bearing gametes results in XXX persons, or "super females". There is no characteristic phenotype associated with this anueploid condition. The level of intelligence and development of the reproductive system vary from normal to subnormal.

XYY SYNDROMES

Another type of abnormal sperm is created by the nondisjunction of Y chromatids. The result is a spermatid without sex chromosomes and a spermatid with two Y chromosomes. Fertilization involving either of the two abnormal sperms results in an imbalance of sex chromosomes in the zygote. The 0 sperm leads to Turner's syndrome when it fertilizes a normal X bearing egg. The YY sperm results in an interesting condition, XYY, which has been referred in the past as the Criminal syndrome, since at one time it was thought that persons possessing the XYY complement tended to commit acts of violence. The association of the XYY chromosome complement with aggressive behavior stems from some studies in the mid 1960s of patients in mental institutions and prisons. These studies indicated that a relatively large percentage of inmates were XYY.

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Persons with sex chromosome abnormalities should not be confused with transsexuals, those who undergo surgery for a change in sexual phenotype. The latter are usually genetically and phenotypically normal but have deep rooted psychological problems accepting their given sex. Still other individuals are true intersexes in the sense that they possess both ovaries and testes. They are called hermaphrodites.

INTERSEX STATES

Conditions in which the appearance of the external genitalia is either ambiguous or at variance with the person's chromosomal or gonadal sex.

The genitalia form during the first 3 monts of gestation via a cascade of events initiated by the fetal karyotype and mediated largely by the sex steroids. Aberrations in this cascade can produce genital ambiguities or inconsistencies, resulting in intersex states. Classification is most conveniently based on gonadal histology.

Female pseudohermaphrodites have ovaries and normal female internal genitalia but ambiguous external genitalia; they are genetically normal females with a 46,XX karyotype. The ambiguous external genitalia result from exposure to excessive amounts of androgens in utero. The offending androgen may be exogenous (eg., progesterone given to the mother to prevent miscarriage) but is more commonly endogenous, eg., resulting from an enzymatic block in steroidogenesis due to genetic aberrations on chromosome 6 (adrenal virilism or adrenogenital syndrome)

Male pseudohermaphrodites have gonadal tissue that is only testicular and usually have a 46,XY karyotype. The external genitalia are usually ambiguous, but this is variable and a female phenotype is seen in the complete form of the testicular feminization syndrome (androgen insensitivity syndrome). Etiology is complex, but in general terms the disorder arises from inadequate production of androgen or inadequate response to androgen

True hermaphrodites have both ovarian and testicular tissue, and mixed masculine and feminine genital structures depending on whether ovarian or testicular tissue predominates. In the USA, most true hermaphrodites have a 46,XX karyotype, but the pattern can be quite variable. Rarely, in true hermaphrodites the external genitalia is fully masculinized.

Patients with **mixed gonadal dysgenesis** have both testicular tissue and primitive gonadal tissue called streaks. These patients usually have a 46,XY/45,XO mosaic karyotype and ambiguous genitalia, and as adults they tend to be short of stature. When the streaks are bilateral, the disorder is termed pure gonadal dysgenesis. Such patients appear phenotypically as females.

Assignment of appropriate sex is paramount. Generally, female pseudohermaphrodites are assigned as females. Male pseudohermaphrodites are assigned according to their genital development and hormonal activity. True hermaphrodites are also best assigned according to their genital development, but most have been reconstructed as males - an attractive option if the child has a normally descended testis to provide hormonal function at puberty. Male pseudohermaphrodites with full-blown testicular feminization syndrome must be assigned as females, but for many others, male assignment is appropriate.

AUTOSOMAL ABERRATIONS

DOWN'S SYNDROME

It is one of the most familiar conditions in humans. Down's syndrome results from aneuploidy - trisomy 21. Hence the person with Down's syndrome has 47 instead of 46 chromosomes because of the presence of one extra chromosome number 21. This is the reason why Down's syndrome can also be represented as (47, +21). Originally studied by Langdon Down in 1866, it had been termed Mongoloid Idiocy or Mongolism because individuals with trisomy 21 were considered to have certain facial characteristics

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that resembled oriental features. The condition is now referred to as Down's syndrome which removes the nuances of associating mental retardation with any particular race of people.

The presence of an extra copy of one of the smallest chromosomes in trisomy 21 results in a wide variety of abnormalities in almost every area of the body, differing in combinations and degree of severity from patient to patient. One of the most constant characteristics which allows doctors to identify Down's syndrome in very young children, even in new born babies, is the line found on the palms of both hands, which are known as dermatoglyphic patterns. The presence of a "simian crease" and a tendency for every finger to have a loop in the print region, rather than whorls or arches, distinguish the Down's child from a normal child. Because of various defects, such as in the cardiovascular system, the life expectancy of an individual with Down's syndrome is shorter than normal. One study sets the life expectancy at 16 years.

Other phenotypic characteristics of this condition are short stature (about 4 feet tall) and an epicanthic fold (thus the earlier name "mongolism"), broad short skulls, wide nostrils, large tongues with distinctive furrowing, stubby hands (particularly the fifth digit) and a general loose jointedness, particularly in the ankles. The patients are characterized as low in mental ability, but they can be trained in routine mechanical skills. Through the investigation of J. Lejeune in 1959, Down's syndrome became the first chromosomal disorder to be described in humans.

The incidence of Down's syndrome increases dramatically with maternal age. This increase is thought to result from the age of the egg cell that is fertilized to produce the zygote. The human female is born with an estimated 400,000 gametes and will not produce any new ones in her lifetime. Therefore, if a woman is 45 years old, her gametes are also 45 years old. Increasing age is believed to result in a tendency for a particular aberration of meiosis called Nondisjunction to occur. There has been little correlation found between the incidence of Down's syndrome and paternal age.

Nondisjunction: Nondisjunction can occur before either the first or the second meiotic division. The basic event in Nondisjunction is just what the term implies: The chromosomes do not separate i.e., they do not "disjoin". This results in aneuploidy in the daughter cells.

Gamete containing the extra chromosome will produce an individual with Down syndrome. Nullisomic gametes (gametes that are missing a particular chromosomes altogether) are presumably nonfunctional or lethal. If nondisjunction occurs in germ cells that are normal, it is called Primary nondisjunction. If however, a trisomic individual reproduces the nondisjunction resulting in aneuploidy in the germ cells is called Secondary nondisjunction.

Down syndrome occurs once in about 700 live births in Europe alone. About one in six of the Down's syndrome children born alive die within the first year. The average expectation of life is 16.2 years. The cost of training and maintaining Down's syndrome cases in the United States is estimated at $1 billion per year. Emotional stress in families with Down's syndrome children and adults is also a factor in their care. The need for effective counseling and prevention is readily apparent.

Amniocentesis for detecting Aneuploidy

Chromosomal abnormalities are sufficiently well understood to permit genetic counseling. A fetus may be checked in early stages of development by karyotyping the cultured cells obtained by a process called amniocentesis. A sample of fluid is withdrawn with a needle from the amniotic sac. Fetal cells are cultured and, after a period of two to three weeks, chromosomes in dividing cells can be stained and observed. If three number 21 chromosomes are present, Down's syndrome is confirmed. The risk for mothers less than 25 years of age to have the trisomy is about 1 in 1500 births; at the age of 40, 1 in 100 births; at 45, 1 in 40 births. Pregnant women above 40 years are thus the special high risk groups.

PATAU SYNDROME

This syndrome was described by K. Patau in 1960. It results when there is a trisomy of chromosome number 13. It can also be represented by its karyotype (47, +13). Patau syndrome occurs in about 1 in

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20,000 newborns. It is rare in children and non-existing in adults because the severe symptoms result in early death. Most of the deaths occur within the first three months after birth, but a few victims have lived for as long as five years.

The symptoms of the syndrome include small brain, apparent mental deficiency, deafness and numerous other external and internal abnormalities.

EDWARD'S SYNDROME

It is first described by J.H. Edwards and his colleagues in 1960. It is the result of trisomy of chromosome number 18. Thus its karyotype is (47, +18).

The symptoms include mental deficiency and multiple congenital malformations involving virtually every organ system. Most infants with this syndrome die at an early age, some 90 percent within their first six months. Nearly all are deceased before they reach one year, but a few have been reported to be alive in their teen years.

The incidence of Edward's syndrome is 1 in 8000. Thus far, only a few cases have been observed, and it is not known whether racial or other population groups differ in incidence. In general, the incidence of this deformity is greater among infants of older women, as expected if the cause of this trisomy is primary nondisjunction in meiosis.

CRUI-DE-CHAT SYNDROME

This is a syndrome resulting from the deletion chromosomal aberration. Chromosomal deletions are usually lethal resulting in zygotic loss, still births, or infant deaths. Sometimes infants with small chromosome deficiencies, however, survive long enough to permit observation of some of the abnormal phenotypes they express.

L.Lejeune and his colleagues discovered a chromosome deficiency in humans that has been associated with the term Crui de chat syndrome. The name of this syndrome came from a plaintive catlike mewing cry from small weak infants with the disorder. Other characteristics are microcephaly (small head), broad face and saddle nose, widely spaced eyes with epicanthic folds, unique facial features, and physical and mental retardation. IQs of Crui de chat children studied are in the range of 20-40. The chromosome deficiency is in the short arm of chromosome 5 and is designated 5p-. A karyotype (46,XX,5p-) is represented for the patient.

Crui de chat patients die in infancy or early childhood and do not transfer the chromosome deletion to offspring. This chromosome deficiency however has been shown by Lejeune and others to become involved sometimes in a reciprocal translocation and thus to be transmitted. When the short arm of chromosome 5 became translocated to chromosome 15, the heterozygous translocation was carried in a normal healthy parent.

GENETIC IMPRINTS IN HUMAN DISEASES

In order to have a better conceptual understanding on the concept of genetic imprinting, it is important to have a look back on Mendelism again. While discussing Mendelism, we have noted the experiment on pea plants. We have observed that a cross between a pure tall with a pure dwarf plant gives rise to heterozygous tall plant in the first filial generation. What we did not note then was which of the plants was a male and other was a female. Mendel wanted to know what could be the affect when a tall female plant was crossed with a dwarf male plant. The result was a tall plant in the first generation. He then wanted to observe the affect when sexes of the plants was reciprocated i.e., he crossed a tall male plant with a female dwarf plant and noted no difference in the phenotype of the offspring. On the basis of those observations, Mendel formulated the theory of equivalence. This theory says that there is no difference in the phenotype of the trait even in case of a reciprocal cross. Later this theory was extended to a number of plants and animals, including human beings.

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However, it is to be noted that the law of equivalence is not universal. This law might be applicable to some genes within the same animal and may not be applied to other genes or chromosomes. Take for example, the sex-chromosomes in the human beings. The male sex chromosomes have a different phenotypic effect compared to the female sex chromosomes. Moreover, there are a number of genes which have "imprinted" or "stamped" sex markers, which is male or female, though they are responsible for a same trait. This leads to a situation where the gene for a same character from a female differ in behavior compared to the gene from a male. This situation is an exception to the law of equivalence and the phenomenon is called Genetic Imprinting. Genetic imprinting is also termed as Genome imprinting. It can be seen both in autosomes and sex chromosomes.

A clear understanding of the concept would be possible with an illustration. In the human beings there is an autosomal dominant disease called Huntington's chorea. Since it is dominant, any of two parents can suffer from this disorder. Hence, the source of the gene for the disease among the offspring can be paternal or maternal. It is observed that in both the cases of origin, paternal or maternal, the symptoms of the disease are same. However, there is a minor difference between the two cases of origin with respect to the behavior of the genes. It is observed that in case of paternal origin the time of initiation and severity of the disease vary when compared to the maternal origin. This proves the fact that though genes have similar structure and function they are differentially "stamped" or "imprinted" according to the source of origin. This is genetic imprinting. Thus, we can now define genetic imprinting as the differential inheritance of genetic material from the mother versus the father.

Mechanism: One of the theories proposed to give an explanation of Genetic Imprinting is the Selective Methylation of DNA.

DNA Methylation is a usual process to control functioning of genes. This is brought about by an enzyme called DNA methylase. This enzyme adds a Methyl group (CH3) to cytosine. If cytosine in one strand is methylated, it would pass through Guanine to the other strand.

It has been discovered that the Genes coming from the mother are usually more methylated than the ones coming from the father. This degree of methylation of genes brings about qualitative and quantitative differences among them. This is the site of genetic imprinting. Excessive methylation results in inhibiting the activity of gene and thus is responsible for a number of disorders.

The concept of genetic imprinting suggests that in certain cases a genetic defect will only produce a phenotype if inherited from a particular parent. We will consider the most important diseases.

If there is a chromosomal deletion in a region concerned with placental development, it may have no effect if inherited maternally but may cause a failure of placental development if inherited paternally. The good examples of the above case are the Prader - Willi and Angelman syndromes. The Prader-Willi syndrome is caused by deletion of chromosome - 15, in position 11, 12 and 13, and is characterized by developmental delay, obesity and hypogonadism. The individuals with this syndrome are always derived paternally. In the case of Angelman syndrome, the deleted chromosome is always maternally inherited. The characteristics of disease are happy disposition, mental retardation, large mouth, protruding tongue and seizures.

Another genetic disease, hereditary Glomus Tumor is seen only in the individuals who have inherited the disease from their father.

Characteristics

1. Imprinted Genes Express Variously: The imprinted male and female genes can have different magnitude of expression and time of expression.

2. Genetic Imprints are Erasable: Imprinting is done in the Gonadal tissues, testes of a male and ovaries of a female. Such markings are erased during gamete formation in the ovary and testes of offspring and he or she stamps his or her marking on the genes.

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3. Gene Imprinting is not a Rule: This phenomenon is discovered only in some male and female genes. A majority of the genes are unimprinted, so that it is difficult to make any difference between male and female genes.

4. Genetic Imprinting is Species Specific: A gene imprinted in one species may not be imprinted in another.

The differences between maternally and paternally derived genes are significant in studying the differential inheritance. Genetic imprinting helps us to know the effect of deletions in different sexes and variation of the disease according to different sexes. A most recent discovery of a new disease due to this phenomenon is Myotonic dystrophy that affects the growing fetus.

GENETIC SCREENING

Genetic screening may be used in populations at risk for a particular genetic disorder. Genetic screening is only appropriate when the natural history of the disease is understood; the screening tests are valid and reliable; sensitivity, specificity, false-negative, and false-positive rates are acceptable; and effective therapy is available. A sufficient benefit must be derived from a screening program to justify its cost.

Heterozygote screening: Screening a susceptible population (eg, Tay-Sachs disease in Ashkenazic Jews, sickle cell anemia in blacks, thalassemia in various ethnic groups) may be appropriate because of the high frequency of heterozygotes. Heterozygote screening can determine if a person is a carrier for a specific disorder. If the partner is also a heterozygote, the couple is at risk of having an affected child. Screening allows the couple to make informed reproductive choices.

Presymptomatic genetic screening: Presymptomatic genetic screening may be appropriate for persons with a family history of a dominantly inherited disorder (eg, Huntington's disease, breast cancer). Identifying a definite carrier of the genetic disorder may allow the patient to make informed decisions (eg, monitoring in the case of breast cancer, reproductive choices in the case of Huntington's disease or adult polycystic kidney disease).

Prenatal diagnosis: Amniocentesis (refer above), chorionic villus sampling, umbilical cord blood sampling, maternal blood sampling, maternal serum screening, and fetal visualization with ultrasound and radiography are useful in prenatal diagnosis. Common reasons for prenatal screening include maternal age > 35 yr, a family history of a condition that can be diagnosed by prenatal techniques, abnormal maternal serum screening results, and certain complications of pregnancy.

Newborn screening: Screening for phenylketonuria, galactosemia, and hypothyroidism in the newborn allows prophylaxis (i.e., special diet or replacement therapy) to be initiated early enough to prevent severe complications.

GENETIC COUNSELING

Genetic counseling consists of educating prospective parents either suffering from or suspected to be heterozygous for some genetic disease on the risk of their children suffering from the same disease. It is hoped that such parents would voluntarily abstain from producing children; this may be encouraged by creating a suitable social environment. It is relatively easy for a trained clinician to identify people suffering from a genetic disease. But identification of carriers for genetic disease is much more difficult and in many cases not possible. Analysis of family pedigree also provides information on the likelihood of an individual being a carrier for a genetic disease.

In a broad sense, Genetic counseling involves:

1. Genetic screening of a disorder and explanation to the patients or their parents the genetic or medical implications of the disease.

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2. Calculation of the likelihood of recurrence of such genetic disorder in the family.

3. To suggest ways in which the occurrence of the genetic disorder can be controlled.

Once genotypes of both the prospective parents become known, it is a simple matter to work out the probability of their child inheriting the disease. If parents heterozygous for a genetic disease decide to produce a child, it is now possible to advise them if their child has inherited the disease through appropriate tests done about two months after conception. This is achieved through amniocentesis; the cultured fetal cells may be used for determining their karyotype, levels of the critical enzymes and restriction patterns of DNA. Such an antenatal diagnosis is now feasible for more than 35 genetic diseases and for a variety of chromosomal defects. The purpose of such a diagnosis is premature termination of abnormal fetuses. This approach has helped to reduce the incidence of thalassemia from 30 to 2 per year in most of Europe. However, antenatal diagnosis may sometimes be misused. For example, it may be used to selectively abort fetuses of one sex. There were some newspaper reports that some parents in India are using amniocentesis to selectively abort female fetuses, obviously to save themselves from the hardships and sufferings they must face to arrange the exceedingly high amounts of dowry for their daughters. If such a practice becomes widespread, its consequences for the society should be obvious to even the most dimwitted.

Genetic counseling and antenatal diagnosis provide definite relief to the possible parents and reduce the frequency of genetically defective individuals in the population. However, it is unlikely that these measures would eliminate the deleterious alleles from a population. This is so because most genetic defects are recessive and heterozygotes for such alleles. Thus even after a total ban on reproduction by the homozygotes for such recessive alleles they would remain in the population through the heterozygotes; therefore even such an extreme selection would lead to only a slow decline in their frequency. Further, it is not likely that all the couples in any society will willingly submit themselves, at least in the foreseeable near future, to these procedures. But genetic counseling has become a routine aspect of medical practice in most developed countries.

It has been advocated that defective genes may be corrected through sophisticated genetic techniques either during the early stages of embryo development (embryo therapy) or in specific tissues of the adult patient (patient therapy); such an approach is referred to as genetic surgery. Embryo therapy would involve...

1. In vitro fertilization of egg

2. Production of several copies of the normal allele of the defective gene

3. Introduction of this DNA into the zygote or in the cells of the developing embryo

4. Integration of DNA, preferably in place of the defective allele, so that it may function normally.

Some of these steps have been tried in mice and rabbit and are being constantly refined. It is possible that embryo therapy may become practically feasible sometime in future, but its desirability is another issue for social debate.

The aim of patient therapy is to introduce the normal gene into the critical tissue of the patient that is affected by a genetic disease, i.e., the tissue where the concerned gene is required to express itself the most, e.g., pancreas in the case of diabetes. The steps involved in patient therapy would be similar to those of embryo therapy. But in this case, cells from the concerned tissues may have to be exercised and treated in vitro to correct their genetic defects and then reintroduced into the tissue where they may function normally. Techniques for isolation, identification and multiplication of many human genes are now available, and for many others they are likely to be developed soon. The techniques for gene transfer in eukaryotes are being refined and it may not remain a great problem in the near future.

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A suggestion has also been made to use highly specific chemical mutagens that will correct the defect in the concerned gene. Such a directed mutagenesis however is a dream that may be more difficult to fulfill than patient and embryo therapies through DNA mediated genetic modifications.

Genetic screening and counseling may also lead to certain problems. The cases of mistaken paternity, the problem of confidentiality, delayed counseling are important among them.

The various proposals on eugenics involve a number of religious, social, moral and political issues which have to be discussed and resolved. The issues involved in genetic surgery and in negative eugenics are relatively more straight forward than those involved in positive eugenics. For example, the concept of "superior type" is likely to vary from one society to another and even from one individual to another within the same society. More importantly, over a period of time, the same society may change its social concepts and values. This leads us directly to the discussion on genetics and bioethics.

HUMAN DNA PROFILING

DNA (Deoxyribonucleic acid) is a chemical structure that forms chromosomes. A piece of a chromosome that dictates a particular trait is called a gene.

Structurally, DNA is a double helix: two strands of genetic material spiraled around each other. Each strand contains a sequence of bases (also called nucleotides). A base is one of four chemicals (adenine, guanine, cytosine and thymine).

The two strands of DNA are connected at each base. Each base will only bond with one other base, as follows: Adenine (A) will only bond with thymine (T), and guanine (G) will only bond with cytosine (C). Suppose one strand of DNA looks like this:

A-A-C-T-G-A-T-A-G-G-T-C-T-A-G

The DNA strand bound to it will look like this:

T-T-G-A-C-T-A-T-C-C-A-G-A-T-C

Together, the section of DNA would be represented like this:

T-T-G-A-C-T-A-T-C-C-A-G-A-T-C
A-A-C-T-G-A-T-A-G-G-T-C-T-A-G

What is DNA Fingerprinting?

The chemical structure of everyone's DNA is the same. The only difference between people (or any animal) is the order of the base pairs. There are so many millions of base pairs in each person's DNA that every person has a different sequence.

Using these sequences, every person could be identified solely by the sequence of their base pairs. However, because there are so many millions of base pairs, the task would be very time-consuming. Instead, scientists are able to use a shorter method, because of repeating patterns in DNA.

These patterns do not, however, give an individual "fingerprint," but they are able to determine whether two DNA samples are from the same person, related people, or non-related people. Scientists use a small number of sequences of DNA that are known to vary among individuals a great deal, and analyze those to get a certain probability of a match.

How is DNA Fingerprinting Done?

1. Performing a Southern Blot
2. Making a Radioactive Probe

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3. Creating a Hybridization Reaction
4. VNTRs

Southern Blot

The Southern Blot is one way to analyze the genetic patterns which appear in a person's DNA. Performing a Southern Blot involves:

1. Isolating the DNA in question from the rest of the cellular material in the nucleus. This can be done either chemically, by using a detergent to wash the extra material from the DNA,or mechanically, by applying a large amount of pressure in order to "squeeze out" the DNA.
2. Cutting the DNA into several pieces of different sizes. This is done using one or more restriction enzymes.
3. Sorting the DNA pieces by size. The process by which the size separation, "size fractionation," is done is called gel electrophoresis. The DNA is poured into a gel, such as agarose, and an electrical charge is applied to the gel, with the positive charge at the bottom and the negative charge at the top. Because DNA has a slightly negative charge, the pieces of DNA will be attracted towards the bottom of the gel; the smaller pieces, however, will be able to move more quickly and thus further towards the bottom than the larger pieces. The different-sized pieces of DNA will therefore be separated by size, with the smaller pieces towards the bottom and the larger pieces towards the top.
4. Denaturing the DNA, so that the entire DNA is rendered single-stranded. This can be done either by heating or chemically treating the DNA in the gel.
5. Blotting the DNA. The gel with the size-fractionated DNA is applied to a sheet of nitrocellulose paper, and then baked to permanently attach the DNA to the sheet. The Southern Blot is now ready to be analyzed.

In order to analyze a Southern Blot, a radioactive genetic probe is used in a hybridization reaction with the DNA in question. If an X-ray is taken of the Southern Blot after a radioactive probe has been allowed to bond with the denatured DNA on the paper, only the areas where the radioactive probe binds will show up on the film. This allows researchers to identify, in a particular person's DNA, the occurrence and frequency of the particular genetic pattern contained in the probe.

Making a Radioactive Probe

1. Obtain some DNA polymerase. Put the DNA to be made radioactive into a tube.
2. Introduce nicks, or horizontal breaks along a strand, into the DNA you want to radiolabel. At the same time, add individual nucleotides to the nicked DNA, one of which is radioactive.
3. Add the DNA polymerase to the tube with the nicked DNA and the individual nucleotides. The DNA polymerase will become immediately attracted to the nicks in the DNA and attempt to repair the DNA, starting from the one end and moving toward the other end.
4. The DNA polymerase begins repairing the nicked DNA. It destroys all the existing bonds in front of it and places the new nucleotides, gathered from the individual nucleotides mixed in the tube, behind it. Whenever a G base is read in the lower strand, a radioactive *C base is placed in the new strand. In this fashion, the nicked strand, as it is repaired by the DNA polymerase, is made radioactive by the inclusion of radioactive *C bases.
5. The nicked DNA is then heated, splitting the two strands of DNA apart. This creates single-stranded radioactive and non-radioactive pieces. The radioactive DNA, now called a probe is ready for use.

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Creating a Hybridization Reaction

1. Hybridization is the coming together, or binding, of two genetic sequences. The binding occurs because of the hydrogen bonds between base pairs. Between a A base and a T base, there are two hydrogen bonds; between a C base and a G base, there are three hydrogen bonds.

2. When making use of hybridization in the laboratory, DNA must first be denatured, usually by using heat or chemicals. Denaturing is a process by which the hydrogen bonds of the original double-stranded DNA are broken, leaving a single strand of DNA whose bases are available for hydrogen bonding.

3. Once the DNA has been denatured, a single-stranded radioactive probe can be used to see if the denatured DNA contains a sequence similar to that on the probe. The denatured DNA is put into a plastic bag along with the probe and some saline liquid; the bag is then shaken to allow sloshing. If the probe finds a fit, it will bind to the DNA.

4. The fit of the probe to the DNA does not have to be exact. Sequences of varying homology can stick to the DNA even if the fit is poor; the poorer the fit, the fewer the hydrogen bonds between the probe and the denatured DNA. The ability of low-homology probes to still bind to DNA can be manipulated through varying the temperature of the hybridization reaction environment, or by varying the amount of salt in the sloshing mixture.

VNTRs

Every strand of DNA has pieces that contain genetic information which informs an organism's development (exons) and pieces that, apparently, supply no relevant genetic information at all (introns). Although the introns may seem useless, it has been found that they contain repeated sequences of base pairs. These sequences, called Variable Number Tandem Repeats (VNTRs), can contain anywhere from twenty to one hundred base pairs.

Every human being has some VNTRs. To determine if a person has a particular VNTR, a Southern Blot is performed, and then the Southern Blot is probed, through a hybridization reaction, with a radioactive version of the VNTR in question. The pattern which results from this process is what is often referred to as a DNA fingerprint.

A given person's VNTRs come from the genetic information donated by his or her parents; he or she could have VNTRs inherited from his or her mother or father, or a combination, but never a VNTR either of his or her parents do not have. Because VNTR patterns are inherited genetically, a given person's VNTR pattern is more or less unique. The more VNTR probes used to analyze a person's VNTR pattern, the more distinctive and individualized that pattern, or DNA fingerprint, will be.

Practical Applications of DNA Fingerprinting

1. Paternity and Maternity

Because a person inherits his or her VNTRs from his or her parents, VNTR patterns can be used to establish paternity and maternity. The patterns are so specific that a parental VNTR pattern can be reconstructed even if only the children's VNTR patterns are known (the more children produced, the more reliable the reconstruction). Parent-child VNTR pattern analysis has been used to solve standard father-identification cases as well as more complicated cases of confirming legal nationality and, in instances of adoption, biological parenthood.

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2. Criminal Identification and Forensics

DNA isolated from blood, hair, skin cells, or other genetic evidence left at the scene of a crime can be compared, through VNTR patterns, with the DNA of a criminal suspect to determine guilt or innocence. VNTR patterns are also useful in establishing the identity of a homicide victim, either from DNA found as evidence or from the body itself.

3. Personal Identification

The notion of using DNA fingerprints as a sort of genetic bar code to identify individuals has been discussed, but this is not likely to happen anytime in the foreseeable future. The technology required to isolate, keep on file, and then analyze millions of every specified VNTR pattern is both expensive and impractical. Social security numbers, picture ID, and other more mundane methods are much more likely to remain the prevalent ways to establish personal identification.

Problems with DNA Fingerprinting

Like nearly everything else in the scientific world, nothing about DNA fingerprinting is 100% assured. The term DNA fingerprint is, in one sense, a misnomer: it implies that, like a fingerprint, the VNTR pattern for a given person is utterly and completely unique to that person. Actually, all that a VNTR pattern can do is present a probability that the person in question is indeed the person to whom the VNTR pattern (of the child, the criminal evidence, or whatever else) belongs. Given, that probability might be 1 in 20 billion, which would indicate that the person can be reasonably matched with the DNA fingerprint; then again, that probability might only be 1 in 20, leaving a large amount of doubt regarding the specific identity of the VNTR pattern's owner.

1. Generating a High Probability

The probability of a DNA fingerprint belonging to a specific person needs to be reasonably high--especially in criminal cases, where the association helps establish a suspect's guilt or innocence. Using certain rare VNTRs or combinations of VNTRs to create the VNTR pattern increases the probability that the two DNA samples do indeed match (as opposed to look alike, but not actually come from the same person) or correlate (in the case of parents and children).

2. Problems with Determining Probability

A. *Population Genetics*

VNTRs, because they are results of genetic inheritance, are not distributed evenly across all of human population. A given VNTR cannot, therefore, have a stable probability of occurrence; it will vary depending on an individual's genetic background. The difference in probabilities is particularly visible across racial lines. Some VNTRs that occur very frequently among Hispanics will occur very rarely among Caucasians or African-Americans. Currently, not enough is known about the VNTR frequency distributions among ethnic groups to determine accurate probabilities for individuals within those groups; the heterogeneous genetic composition of interracial individuals, who are growing in number, presents an entirely new set of questions. Further experimentation in this area, known as population genetics, has been surrounded with and hindered by controversy, because the idea of identifying people through genetic anomalies along racial lines comes alarmingly close to the eugenics and ethnic purification movements of the recent past, and, some argue, could provide a scientific basis for racial discrimination.

B. *Technical Difficulties*

Errors in the hybridization and probing process must also be figured into the probability, and often the idea of error is simply not acceptable. Most people will agree that an innocent person should not be sent to jail, a guilty person allowed to walk free, or a biological mother denied her

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legal right to custody of her children, simply because a lab technician did not conduct an experiment accurately. When the DNA sample available is minuscule, this is an important consideration, because there is not much room for error, especially if the analysis of the DNA sample involves amplification of the sample (creating a much larger sample of genetically identical DNA from what little material is available), because if the wrong DNA is amplified (i.e. a skin cell from the lab technician) the consequences can be profoundly detrimental. Until recently, the standards for determining DNA fingerprinting matches, and for laboratory security and accuracy which would minimize error, were neither stringent nor universally codified, causing a great deal of public outcry.

GENE MAPPING & GENOME STUDY

The Human Genome Project (HGP) was formally launched in October 1990 in USA. The project was co-sponsored by the Department of Energy (DOE) and the National Institute of Health (NIH). It was envisaged as a US$3 billion, 15 year - effort. The completion of the first draft sequence or map representing about 90 per cent of the human genome was announced on June 26, 2000. The gigantic task has been accomplished a year ahead of schedule. It is undoubtedly a milestone in the march of sciences & technology. The then US President Bill Clinton has hailed it as "the most important and most wondrous map every produced by humankind'. The draft sequences will form the basis for obtaining the high quality finished sequence. It will also be a valuable tool for researchers hunting disease genes. A complete, high quality DNA reference sequence is expected to be achieved by 2003, two years earlier than originally envisaged. It may be achieved even sooner. In the year 2003 the 50th anniversary of unraveling of DNA structure by James Dewey Watson (1928) - and Francis Harry Compton Crick (1916) - will also be celebrated. It is expected that discoveries made based on the sequences will have profound impact on the better understanding of the state of good health and diseases. This information is likely to revolutionize the diagnosis, prevention and treatment of most, if not all, human diseases. Scientists in many countries are now engaged in developing tools and applications for the new data in such wide-ranging fields as medicine, agriculture, bioremediation and industrial enzymology.

The draft sequence is like a jumble of parts of a functional device - to make the device with a rough idea of how to assemble the parts. It contains gaps and errors. It provides scientists with a road map to an estimated 90% genes on every chromosome. All HGP data have been placed in the public domain (on the internet).

We will now briefly describe the concepts involved, genesis and implications of the HGP.

Genome may be defined as the complete genetic information carried by an individual or the range of genes found in a given species. The carriers of genetic information inside animal and plant cells are chromosomes, threadlike bodies. Chromosomes get their name from the fact that they take certain strains more heavily than do other parts of the cell, the term being derived from the Greek `Chromos' (color) and 'soma' (body). Chromosome number is characteristic of a species. For example humans have 46 chromosomes, mosquitoes 6, frogs 26, earthworms 36, chimpanzees 48, amoebas 50 and horses 64. Human chromosomes comprise 22 matched pairs called autosomes and two sex chromosomes (X and Y). Every cell of our body has the same number of chromosomes except the reproductive cells (eggs are sperm), which have exactly half the usual number. Chromosome comes from both parents and a new individual life is formed by the fusion of two reproductive cells. Each chromosome consists of one very large strand of DNA (deoxyribonucleic acid) and proteins. The DNA is coiled and folded to produce a compact body. DNA, the chief constituent of chromosome, contains in chemically coded form, all the information needed to build, control and maintain a living organism. The structure of DNA, first proposed by J.D. Watson and F. H. C. Crick in 1953, is made of a double helix of two chains coiled around each other. Each chain is made up of alternating pentose sugar (deoxyribose) and phosphate groups, with an organic base attached to each pentose group. There are four possible bases: adenine (A), guanine (G), cytosine (C), and thymine (T). The bases join with each other by hydrogen bond to form base pairs that link the two strands of the DNA molecule like the rungs of a twisted ladder. The pairs are formed in a specific

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way; adenine links with the thymine and cytosine with guanine. Hereditary information is preserved from generation to generation as a specific sequence of bases. It is the bases (G, C, T and A) which form the letters of the genetic alphabet. A set of three bases called codon represents a particular amino acid, the building block of a protein. The particular order of As, Ts, Cs and Gs is extremely important because the order underlines all of life's diversity. In fact, this order directs whether an organism is human or another species. In a human genome about 3 billion bases are arranged along the DNA molecule in a practically infinite variety of sequence. The human genome can be seen as an incredible book written with 3 billion letters with four alphabets.

A functional unit of DNA corresponding to a specific sequence of the genetic code is known as a gene, a term first coined by W.L. Johannsen in 1909. Genes are the basic physical and functional units of heredity. Human genes vary widely in length, often extending over thousands of bases. Structural genes code for individual polypeptide while regulatory genes control the activity of the structural genes. Often the information content within a gene of higher organism may not be continuous. In such case different parts of a gene called exons are joined together after removing the unwanted sequences (introns) to make a continuous stretch of information in the form of messenger RNA molecule, which in turn is translated into a protein. Scientists do not know how many genes are there in the human genome. It used to be believed that the human genome has 100,000 genes. But recently this number has been questioned - predictions ranges from 30,000 to 1, 20,000 genes.

Till 1971 only 15 human genes had been localized to specific chromosomes. However, the ability to map genes was substantially increased with the development of the recombinant DNA technology particularly with the use of restriction enzymes to cut DNA molecules into small fragments.

The HGP was conceived by US Department of Energy (DOE). It was an outcome of the DOE's long-term mission to develop improved technologies for measuring; the effect of low level exposures to radiation and other energy related agents-especially the effect of such exposure on cancer risk. A brief outline of the project history is given below.

1984: The seeds of the HGP were sown in a conference at Alta, Utah. The conference jointly organized by the Office of the Health and Environmental Research of DOE and the International Commission for Protection against Environmental Mutagens and Carcinogens addressed a single question: "Does modern DNA research offer a way of detecting tiny genetic mutations - and, in particular, of observing any increase in the mutation rate among the survivors of the Hiroshima and Nagasaki bombings and their descendants".

1986: The office of the Health and Environmental Research of DOE initiated the process for considering the feasibility of a dedicated human genome program. An international conference was organized in March 1986 to assess the desirability and feasibility of implementing such a project. The participants agreed that ordering and eventually sequencing DNA representing the human genome were not only desirable but also achievable. The DOE initiated several pilot projects.

1987: The Health Effects Research Advisory Committee recommended in its report that DOE and the nation commit to a large, multi-disciplinary scientific and technological under taking to map and sequence the human genome.

1988: The US National Research Council (NRC) brought out a report entitled "Mapping and Sequencing the Human Genome". The NRC report recommended that the US support this research effort. This report also recommended a major role for the National Institute of Health (NIH). It also presented an outline for a multiphase plan. The NIH established the office of Genome Research in the Director's Office, which was upgraded to the National Centre for Human Genome Research in 1989 and subsequently the National Human Genome Research Institute. The US Congressional office of Technology Assessment (OTA) and the National Academy of Science studied the issue.

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Goals of HGP

- Map the entire human genome.
- Determine the complete sequence of the 3 billion base pairs that make up human genome or defines the human species.
- Identify all the genes in human DNA.
- Store this information in database.
- Develop tools for data analysis.
- Address the ethical, legal and social issues resulting from the project.

All organisms are related through similarities in DNA sequence and so understanding of the functioning of non-human genomes, often lead to new knowledge about human biology. As a part of the HGP the scientists are also carrying out parallel studies of several non-human organisms like the common human gut bacterium, Escherichia coli, the fruit fly and the laboratory mouse.

Though the HGP started as US project it has become a worldwide research effort. The "Universal Declaration on Human Genome and Human Rights" by the UNESCO recognizes that research on human genome and the resulting capabilities and research would open up immense prospects for the whole humankind. Today, some 17 countries are participating in the project. The USA, the country which initiated the project, is playing a major role. Significant contributions are coming from the Sanger Centre in the United Kingdom and research centers in Germany, France and Japan. The other countries are: Australia, Brazil, Canada, Denmark, Israel, Italy Korea, Mexico, The Netherlands, Russia and Sweden. European Union is also taking part in the project. A considerable amount of financial support for the project is coming from the private companies. The Muscular Dystrophy Association of France and the Welcome Trust of the UK are also supporting the project. Some developing countries including India are directly or indirectly participating through studies of molecular biology techniques for research and studies of region specific organisms. The Indian contribution to the genome analysis programs is not very significant.

An international organization, the Human Genome Organization, (HUGO), was established in Geneva, Switzerland in 1988 with the following objectives:

- "To assist with the co-ordination of research on the human genome and in particular to foster collaboration between scientists with a view to avoiding unnecessary competition or duplication of efforts and to co-ordinate this research with parallel studies in model organisms."
- "To co-ordinate and to facilitate the exchange of data and biomaterials relevant to human genome research and through training program, encourage the spreading of the related technologies".
- "To encourage public debate and provide information and advice on the scientific, ethical, legal and commercial implication of human genome project".

Criticism

When the project was first announced there was a big hullabaloo. Among the critics were prominent molecular biologists. Some of the objections raised by the critics were:

- The project was viewed as colossal waste of time, money and human resources on a monotonous, technology-driven exercise.
- The mega project would take major chunk of already scarce research funds away from the traditional small science of individual investigator-initiated research projects.

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- The complete sequence of each gene is simply unnecessary and tedious. The search for disease genes may be accomplished by approaches without involving arduous task of complete sequencing.
- Most part of the genome may simply be evolutionary baggage with no useful function.
- A host of ethical, legal and social issues were also raised: Who should have access to the genetic information? If handled carelessly genetic information could threaten us with discrimination by potential employers and insurers. How are we going to ensure privacy and confidentiality of genetic information! Personal knowledge of genetic susceptibility can make it possible for more accurate diagnoses, preventive interventions, intensified screening, life-style changes and early and effective treatment. But then there is the risk of anxiety, unwelcome changes in personal relationships and danger of stigmatization. How are the "products" of HGP to be patented and commercialized!
- How are the judicial, medical, education communities and the general public to be effectively educated about the implication of HGP! To address the ethical and other social issues the NIH-DOE joint Working Group on Ethical, Legal and Social Implications of Human Genome Research was created in 1990.

But there are also strong supporters of the project. Some of the arguments in favor of the project are:

The project has been likened with the American drive to put a man on the moon during the 1960's. It was projected as panacea for all diseases. It would be possible to unveil and eventuality to subvert, the causes of thousands of human diseases. "It represents nothing less than the ultimate scientific response to the Socratic dictum "know thyself" - Professor James Trefil of George Mason University.

Benefits

- Technology and resources developed by the HGP will make possible for identifying the most fundamental causes of diseases. Earlier detection of genetic predispositions to diseases will be possible by virtue of rapid, improved and more specific diagnostics tests. Greater understanding of genetic predisposition to disease will also make possible avoidance of environmental conditions that may trigger diseases. It has been projected that eventually it would be possible to find out that genetic disease a person is at risk for and fix it by putting in a gene of appropriate sequence.
- Taking advantage of new capabilities and resources developed by the HGP studies for sequencing the genome of bacterial useful in energy production, environmental remediation, toxic waste reduction and industrial processing have already been initiated. It will be possible to explore new energy sources.
- It will be possible to assess health damage and risks caused by radiation exposure (including low-dose exposure), mutagenic chemicals and cancer-causing toxins. The likelihood of heritable mutations can be reduced.
- As a part of HGP the scientists are also studying the genomes of other organism. Such competitive studies will enable scientists to ascertain the function of thousands of unknown genes. It will be possible to develop new insights about human evolution and the relationship among the three kingdoms of life-archaeo-bacteria, eukaryotes and prokaryotes.
- The knowledge generated by the GP will help in environmental protection by detecting bacteria and other organisms that may pollute air, water, soil and food.
- It will be possible to develop disease, insect and drought resistant crops.
- Healthier, more productive, disease-resistant farm animals will be developed.
- Bio-pesticides will be developed.

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The project has both positive and negative aspects just like in the case of atomic energy. There are many disturbing issues, which need to be resolved but then there are also immense possibilities which humankind may exploit for a better future. The draft sequence of the human genome is the beginning of a biological revolution. The practical and economic applications of the HGP are destined to usher in dramatic growth. Its impact that will be felt in medicine and health care alone is inestimable. But then the DOE report states: "The genome project itself offers no promises of cancer cures or quick fixes for Alzheimer's disease, no detailed understanding of genius or schizophrenia. But if we are ever to uncover the mysteries of carcinogenesis, if we are every to known how biochemistry contributes to mental illness and dementia, if we every hope to really understand the processes of growth and development, we must first have a detailed map for the genetic landscape. That's what the Human Genome Project promises. In a way, it's rather prosaic step, but what lies beyond is breath taking".

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9.5 RACE AND RACISM

CONCEPT OF RACE

Theodosus Dobzhansky, the pioneering researcher on Reproductive isolation has defined race as a "group of population which is reproductively isolated to the extent that the exchange of genes between them is absent or so slow that the genetic differences are not diminished." Implied in this definition are four features of a race.

1. Group of Population: All populations have their own gene pool with differing gene frequencies. If race is defined in terms of gene frequencies, all populations would qualify to be called a race. Race is higher in hierarchy than populations, and consists of many populations that have gene frequencies related to one another.

2. Genetic Differences: The group of populations forming a race has some genes in very high frequency and some in very low frequency and these genetic differences characterize a race. Such differences in gene-frequency arise partly because of natural selection and chance.

3. Reproductively Isolated: The group of populations forming races is reproductively isolated. This isolation maintains the genetic differences. This isolation is however, not complete. Whenever races expand their range and come into contact of other races, they potentially hybridize and a new gene frequency is set up.

4. Race is a Biological Concept: Race is different from religious, political or cultural groups. Race is purely a biological concept and occurs not due to any cultural superiority but genetic differences.

RACE & RACISM

Since the development of the concept of race, it has been subjected to a lot of controversy. For some, it exists as a biological phenomenon and for others as a cultural one. To better understand the controversy, we shall see both the connotations of the term "Race".

Race as a Biological Concept

To understand why the racial approach to human variation has been so difficult, we must first understand the race concept in strictly biological terms. Briefly, a race may be defined as a population of a species that differs in the frequency of some gene or genes from other population of the same species. Simple and strait forward though such a definition may seem there are three very important things to be noted about it. First, it is arbitrary - there is no agreement on how many genetic differences it takes to make a race. For some people, differences in the frequency of one gene are sufficient, while for others, differences in frequencies of several genes are necessary. The number of genes and the ones needed to make a race are still open to debate.

The second thing to note but the biological definition of race is that it does not mean that any one race has exclusive possession of any particular gene or genes. In human terms, the frequency of the gene for blood group `O' may be high in one population and low in another, but it is present in both. Races are genetically "open", meaning that gene flow takes place between them. Thus one can see the fallacy of any attempt to identify "pure" races; if gene flow cannot take place between two populations, then they are not races, they are separate species.

The third thing to note about the biological definition of race is that individuals of one race will not necessarily be distinguishable from those of another. In fact, as we have just noted with respect to humans, the differences between individuals within a population may be greater than the differences between populations. This follows from the genetic "openness" of races; no one race has an exclusive monopoly on any particular gene.

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As a device for understanding human variation, the biological race concept has serious drawbacks. One is that the category is arbitrary to begin with, which makes agreement on any given classification difficult. Perhaps if the human species were divided into a number of relatively discrete breeding populations, this would not be such a problem, but even this is open to debate. What has happened though is that human populations have grown in the course of human evolution, and with this growth has gone increased chances for contract and gene flow between populations. Since the development of food production, which began about 10,000 years ago, the process has accelerated so that differences between human populations today may be less clear-cut than back in the days of Homo erectus, or even the Neanderthals.

If this is not enough of a problem, things are complicated even more because humans are so complicated genetically. Thus the genetic underpinnings of phenotypic traits that traditionally are the subject of racial studies are often poorly understood. To compound the problem, "race" exists as a cultural, as well as biological, category. In various different ways, cultures define religious, linguistic, and ethnic groups as races, thereby confusing linguistic and behavioral traits with physical traits. They also adopt emotional attitudes toward them.

A major result of all this has been a lot of debate about what "race" is and is not, as well as the number of human races that can be defined. Often forgotten is the fact that a race, even if it can be defined, is the result of the operation of evolutionary processes. Because it is these processes rather than racial categories themselves in which we are really interested, a growing number of anthropologists have become convinced that the race concept is of no particular utility. Instead, they prefer to study the distribution and significance of specific, genetically based characteristics, or else the characteristics of small breeding populations that are, after all, the smallest meaningful units in evolution.

Race as a Cultural Concept

Why has it been impossible to agree upon a definition of race? The answer to this question lies in the fact that the radical racial categories people use are often very revealing indicators of their social values. The way in which we see other people is culturally conditioned. Recognizing the race of another is something we learn in the process of growing up in a particular culture. Some cultures recognize many more shades of skin color than do Americans, who tend to dump people into the categories white and black. Brazilians have about 500 different racial labels (Kottak, 1974), each level corresponding to some phenotypical trait. Because the gradations are so subtle, one individual may have several different racial labels during the course of his or her lifetime.

Concepts of race are so ambiguous and so much a product of cultural bias that race, as the term is traditionally used, has no clear biological meaning. Thus, by using the term, one runs the risk of having it interpreted in ways that were not intended. No anthropologist would deny that race is an important way of organizing human variation in cultural terms. But culturally defined categories cannot be translated into objective, clear, biological categories. For this reason, physical anthropologists are gradually abandoning the use of racial categories in studying human variation.

Relevance of the Concept of Race Today

The biological and scientific reasoning behind the conception of race is today sadistically trashed. The objective and scientific reasoning behind the biological concept of race is increasingly substituted by subjective and unscientific bases of culture. The growth of "Racism" in recent decades on the basis of cultural and intellectual superiority has led to the new concept of "ethnic cleansing" as seen in former Yugoslavia. People are being increasingly differentiated on the basis of unscientific concepts like "purity and superiority of blood", "fine color", "superior mental and physical abilities" and "cultural superiority". In other words, race and concept of race has paved the way for ethnocentrism.

In 1952, the UNESCO has made a declaration regarding the concept of race.

1. All human beings belong only to one species - Homo sapiens.

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2. There is no doubt some differences in the physiological anatomy either because of hereditary trail or environment but generally both affect it.
3. The change in heredity is because of mutation or cross marriage.
4. Race cannot be grouped on the basis of nationality, religion, geographical, cultural and linguistic factors.
5. The present day classification is based on anatomical and morphological differences and not on any ground of inferiority and superiority.
6. Intelligence does not play any role in classification of the races.
7. Culture differences are not the cause of racial differences.
8. The so called "pure races are no where to be found either these days or in the past". There has been intermingling of races going on.
9. The human beings are equal and they deserve equal treatment.

Owing to these difficulties in comprehending the scientific objectivity and failure to translate them into objective facts, the concept of race is being abandoned today. Of late, the concept of race has only become a myth and not a reality.

BIOLOGICAL BASIS OF MORPHOLOGICAL VARIATION OF NON-METRIC AND METRIC CHARACTERS

The most noticeable biological variations among human populations are those which are on the surface. They are also many biological variations we do not see. Certain of these variations may be explainable as adaptations to differing physical or social environment or as consequences of other physical or cultural changes. According to Boyd, a criterion or a base which is selected for classifying the human populations into various races should fulfill the following conditions:

1. The parameter should be objective without any bias of individual variations.
2. The criteria should be least or should not be influenced by the environment; that is the parameter chosen should be non-adaptive.
3. The parameter or the criteria should not undergo mutations.
4. The genetic principles of the character which is considered for the racial classification should have known genetic principles, which mean it should be inherited in a Mendelian pattern.

The criteria which are chosen for the racial classification to a majority are phenotypic in nature. These criteria are the morphological bases of racial classification. There are other criteria which have the physiological characters with well-known genetic principles. These characters are the serological or genetic bases for the racial classification.

MORPHOLOGICAL CRITERIA

The morphological criteria can be anthroscopic or morphoscopic, in the sense that the parameters can be discriminated on the basis of vision. For e.g., skin color, nose form etc. A morphological character can also be anthropometric or morphometric in the sense that the parameter is measurable. It includes stature, Nasal index etc.

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I. SKIN COLOR

Human populations differ in average skin colour. Many people consider skin colour as the most important indicator of racial distinction. But anthropologists note that skin colour is not a good indicator of race. For e.g., extremely dark skin is found most commonly in Africa. However there are natives of Southern India whose skin is as dark as or darker than that of the Africans. Yet these people are not closely related to Africans either genetically or historically.

Nevertheless, the very fact that there are people in the world with the markedly different skin colours leads us to the question of how we can explain this wide range of hues. We now know that skin colour is influenced by a pigment called Melanin. The more Melanin there is in the skin, the darker the person will be. Furthermore, the amount of melanin in the skin seems to be related to the climate in which a person lives. According to Gloger, the populations of birds and mammals living in warm, humid climates have more melanin than do populations of the same species living in cooler, drier areas. Dark pigmentation seems to have at least one specific advantage in the tropical climate. Melanin protects the sensitive inner layers of the skin from the sun's damaging ultra-violet rays. Therefore dark skinned people living in sunny areas are safer from sun-burn and skin cancers than light skinned people. Presumably the light coloured skin must be having some benefits; otherwise, through the process of natural selection human populations would all tend to have relatively dark skin colour. Although the light skinned people are more susceptible to sun-burn and skin cancer, light skin absorbs the ultra-violet radiation which facilitates the body's production of Vitamin-D. Thus the light colour of people in temperate latitudes maximizes ultra-violet penetration, perhaps ensuring production of sufficient amounts of Vitamin-D for good health, where as the darker skin of people in tropical latitudes minimizes it, thereby preventing illness from too much of production of Vitamin-D.

The population of the world can be classified into three groups on the basis of skin colour.

1. **Leucoderms:** The leucoderms are the white-skinned people. Apart from the Europeans, who form the best example for this skin color, the Western Asiatics, the North Africans, and Polynesians are the other examples. Caucusoids are also included in this group.

2. **Xanthoderms:** The xanthoderms are the yellow skinned people. For example, Mongoloids, Armenoids etc.

3. **Melanoderms:** Melanoderms are dark skinned people and the dark color can be accounted to abundant deposition and profuse diffusion of melanin and hence the name. For example, The Negroid.

II. HAIR

Hair is one of the most important criteria applied for racial classification. In fact it is one of the oldest criteria employed for this purpose. The criterion of hair can be further divided into various characteristics conveyed by it. These are the form, colour, texture, quantity, cross-section, and hair whorl.

1. **HAIR FORM:** Depending upon the form of hair, three broad groups can be identified.

   a) Leiotrichy or Straight Hair - Mongoloids.

   b) Cymotrichy or Wavy Hair - Peoples of West Asia, some parts of Africa and Europe.

   c) Ulotrichy or Woolly Hair - Negroes, Andamanese, Bushmen, and Melanesians etc.

   It has been noted that the climatic or the environmental conditions influence the hair form. A moist and a warm climate is said to be responsible for woolly hair, whereas the dry and a cold climate is

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responsible for straight hair. But many anthropologists are in a view that climatic conditions do not influence the hair form.

2. **HAIR COLOUR:** A wide range of colours are exhibited by hair. The colour of the hair is because of the presence of pigments present in the cortex of the hair. The range of colour extends from dark hair to light or white hair. Environment is said to have no effect at all on the colour of the hair. Among the North Europeans the colour of the hair ranges from light brown to red. Some Negroid populations in Africa and Melanesia possess hair with a yellow tinge. The oceanic Negroids possesses dark red hair and blonde hair is found only among the people of Europe in the Baltic area.

3. **HAIR TEXTURE:** Based on the texture of the hair, it can be divided into coarse hair, medium hair or fine hair. The thickness of the hair ranges between 5 - 125 microns. According to Garn, hair can be classified accordingly.

   Fine hair - 56 microns

   Medium hair - 57 - 84 Microns

   Coarse hair - 85 and above Microns

4. **QUANTITY:** The quantity of the hair on the head can be further divided into following types based on the amount of hair present:

   a) SCANTY

   b) MEDIUM

   c) THICK

   d) VERY THICK

   e) RICH

   This criterion is an anthroposcopic or morphoscopic criterion since it can be ascertained visually by mere observation. One point to be noted is that when the quantity of hair is being considered, the quantity of hair on the various portions of the body should also be taken into account.

5. **CROSS SECTION:** Based on the cross section of hair, it can be classified into Circular or Oval. The circular cross section can be noticed among the hair of the Mongoloids and an oval cross section can be observed among the Negroids. This criterion is very rarely considered for the purpose of racial classification.

6. **HAIR WHORL:** The occiput of the head reflects the whorls of the hair. The hair whorls can be either Clock Wise or Anti-Clock Wise. Though possession of one whorl is common in one individual, rarely more than one whorl in a single individual can also be observed.

III. STATURE

Scientists have suggested that the body build of many mammals may vary according to the temperature of the environment in which they live. According to Bergmann, the smaller size populations of a species inhabit the warmer part of its geographical range and the large size populations inhabit the cooler areas. The rationale behind this theory is that the long limbed body type that is often found in tropical region provides more surface area in relation to body mass and thus facilitates the dissipation of body heat. In contrast, the chunkier short limbed body type found among the residents of cold regions help to retain body heat by reducing the amount of surface area to body mass. It is not clear, however, whether

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differences in body build between populations are due solely to natural selection or different conditions of cold or heat. Some of the variations may be induced during the life-span of the individual.

Although the adaptation to heat and cold may have some effect on the varying height of human populations, such variation may also be produced by other conditions. Physical or psychological stress, nutrition, medical care and other factors unrelated to temperature may affect the human stature. As a result of our increased social and geographic mobility, mating between different kinds of people are fairly common now-a-days across cultures and continents. Increased height in the off-springs of these matings may be partially a consequence of the hybrid vigour so produced.

According to the general scale of stature, the following list is tabulated.

CATEGORY	MALE (cm)	FEMALE (cm)
Very Short	0 – 150	0 – 140
Short	150 – 160	140 -159
Medium	161 – 170	151 – 159
Tall	180 – 185	168 – 178
Very Tall	186 +	178 +

IV. HEAD FORM

The head form can be expressed in three indices - Cephalic index, Length-height index and Breadth-height index. It was Anders Retzius, who noted that there was a great deal of variation in the head form among different races. He decided that the shape of the head was an important criterion for classifying the people.

Cephalic Index = Breadth of Head / Length of Head x 100

According to Broca, the following is the classification of the head forms depending upon the Cephalic index:

TYPE	CEPHALIC INDEX
Dolicocephalic	- 75.00
Sub-dolicocephalic	75.01 – 77.77
Mesaticephalic	77.78 – 80.00
Sub-brachycephalic	80.01 – 83.33
Brachycephalic	83.34 - +

Broca's scale of classification is not widely accepted because his calculations are based on the measurements he has taken on a skeletal skull, when actually for thickness the epicranial tissues are to be considered for this calculation.

The following is the classification based on Joseph Denicker's scale.

TYPE	CEPHALIC INDEX
Hyper - dolicocephalic	- 75.90
Dolicocephalic	76.00 – 77.00
Sub-dolicocephalic	78.00 – 79.00
Mesaticephalic	80.00 – 81.00
Sub-brachycephalic	82.00 – 83.00
Brachycephalic	84.00 – 85.00
Hyper-brachycephalic	86.00 - +

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There is much of ambiguity and debate regarding the cephalic or head form for the purpose of racial classification. Moreover, since the recent discovery that head form to some extent is determined by birth, this criterion for racial classification is considered to be of secondary importance.

V. FACE FORM

Like the stature, the facial structure may also be affected by environment. This was proved by the experiments conducted on rats by Reisenfeld. Temperature related effects are also evident in the structure of human faces. The people living in cold climates have higher, narrower noses, than warmer regions. This difference may be due to low absolute humidity associated with cold air rather than with cold itself. A relatively narrow nose may be more efficient humidifier and heater of cold air than a broad nose.

Faces are of different shapes - round, oval, square, etc. For scientific purposes, faces have been classified on the basis of facial index. It is calculated as given below:

Facial Index = Morphological facial length/bizygomatic breadth x 100

TYPE	CEPHALIC INDEX
Hyper Euryprosopic	- 78.90
Euryprosopic	79.00 – 83.90
Mesoprosopic	84.00 – 87.90
Leptoprosopic	88.00 – 92.90
Hyper Leptoprosopic	93.00 - +

There is a harmonic relation between the face and the head. A long face is generally associated with long head while a Brachycephal has a broad face. But in some cases exceptions are noticed in these arrangements. This condition is known as disharmony. The Cro-Magnon people in the history possessed this type of disharmonic face. The condition of long head and broad face is generally seen amongst the Eskimos.

VI. JAW PROFILE

The lower jaw shows a varying degree of conditions - Prognathism is the condition in which the lower jaw is protruded outwards and downwards as in the Negroids. When the face does not show any trace of protrusion, it is known as Orthognathism. Sometimes the alveolar regions of the upper and lower jaw project forward and these conditions are known as Alveolar Prognathism. The modern people are characterized by orthognathism.

VII. NOSE FORMS

Three different morphological conditions can be considered with regards to the nose.

Nasal Index: Nasal index can be calculated by the following formula:

Nasal Index = Nasal breadth / Nasal length x 100

NOSE TYPE	SKELETAL MEASUREMENT	LIVING FORM MEASUREMENT
Leptorrhine (Caucasoid)	....- 47.9	...- 70.0
Mesorrhine (Mesorrhine)	48.0 – 52.9	71.0 – 84.9
Platyrrhine (Negroid)	53.0 - +	85.0 – 99.0

Nasal Root: Nasal root is the junction of nasal bones with the frontal bones of the skull. It bears a depression of varying depth. It is of three kinds - low, medium and deep.

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Nasal bridge: The two nasal bones join laterally all along their lengths. Both bones are rectangular in shape. They thus form an angle while they join with each other. This is the nasal bridge.

The Nasal Profile depends upon nasal bridge and roots. The various profiles are: Straight, Concave, Convex or Concavo-Convex.

Narrow and broad nasal bridges are found in primitive human groups. Negroes have depressed root with a broad bridge. Australians have much lower root than Negroid, their bridges are also much low and narrower. Caucasoids have the highest bridge.

SEROLOGICAL AND GENETIC CRITERIA

In this section, we shall begin with a discussion of the genetically determined bio-chemical characteristics in which different individuals in the same population - indeed, different members of the same family - often differ from one another. These differences are of interest genetically, anthropologically, and from an evolutionary perspective because they reflect the presence or absence of the same genes.

Simple, genetically determined bio-chemical variations can be identified and counted within populations and populations can then be compared in respect to the frequencies of the genes which determine these variants. The development of this branch of human genetics has resulted from the importance of bio-chemical variants in medicine. Anthropologists do not often focus on the frequently complex relationships of genetic variations to health and disease. Usually, they seek common variants of known or unknown pathological significance, which if they occur, can be used to trace the historic connection between people.

SEROLOGY

We know by the traditional methods of Physical anthropology, and even from causal observations that main characteristics such as skin colour, nose size and hair form differ among human groups. But in structures such as skin, nose and hair, genetic potential is expressed to a varying extent and is different throughout the course of development. The relationship between them can be traced through a particular base sequence in the DNA of a chromosome and the end result can rarely be identified in the genetics of these quantitative characteristics. On the other hand, through Serology, the examination of components of blood specimens, it is possible to identify very small qualitative differences.

Blood is a complex substance. If it is collected in a syringe and allowed to stand, it separates into two parts. First, a reddish mass composed of red blood cells and of fibrinogen that causes them to clot and second, a clear yellow fluid blood-serum. However, if a chemical which prevents clotting is added to a fresh specimen and the blood is then separated into components by spinning it in a centrifuge, fibrinogen and the serum remain together and constitute blood plasma, while the red cells collect at the bottom of the tube. There is also a thin third layer composed of the white cells. All these components of blood contain many varying substances. The Serum contains a wide variety of serum proteins. The red cells contain haemoglobins and have blood group substances on their surfaces, and the intermediate Buffy-coat (the white cells) is a valuable material for the study of the tissue antigens.

ANTIGENS AND ANTIBODIES

When blood group substance from one individual is introduced into the blood stream of another, a number of substances called antigens on the surfaces of the red blood cells of the transfusion blood, react with the substances called antibodies in the host blood serum. This reaction is similar to that of blood serum with bacteria which stimulate the production of antibodies, capable of combining with the bacterial substance and hence serving as anti-bacterial agents. Although there are several kinds of interaction all help protect against repeated attacks of the disease, caused by a specific kind of bacteria. The study of such interactions between substances and serum antibodies is called immunology because

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in case of the bacteria and the viruses the interaction of the antibodies with bacteria and viruses destroy these organisms, thus affording immunity to disease.

The study of such reactions in humans has permitted the classification of human red cell antigens. These antigens are inherited according to Mendelian principles and the individual keeps the same kind throughout the life. Antibodies however are not usually inherited but are formed in the gamma-globulin molecules of serum exposed to a particular antigen.

POLYMORPHISM

The minor differences in the amino acid sequence of proteins in the blood serum and in enzymes are caused by corresponding differences in the nucleic acids that code for these molecules. Many of the variants are the result of a single variation in the base sequence of the DNA. Some of these variants reach high frequency in some human populations and are called Polymorphisms. A polymorphism may be defined as a gene in which a variant allele has a frequency above 10% in some human populations. There are at least 20 such polymorphic variations in 13 or 14 different human blood group systems, a number of additional polymorphisms among the 23 or so distinguishable serum proteins and at least 4 among the several score of known human haemoglobin variations. There are also normally occurring human variations in the nature of some enzymes. Besides polymorphisms tested in blood, there are a few that affect the substances in the other body fluids, such as one amino acid - in urine and the secretor factor and other components of saliva.

Analysis of the pedigrees of polymorphisms shows whether they are due to allelic or closely linked genes. Those that are allelic or linked constitute a system (e.g., The most familiar system is the ABO system which has four different blood groups A, B, AB, O). There is no evidence of linkage between the different blood group systems and most of them are known to be inherited independently of one another. There are also independent in evolution. Gene frequencies of one system can change while those of another remain the same. Hence two populations may be more similar with respect to one system than another. There are enough mutually independent systems of blood group antigens to permit anthropologists to classify the present day populations into meaningful patterns that reflect historical relationships over centuries. When national or tribal groups are classified according to the frequencies with which they posses the genes for different antigens, fewer and smaller differences are usually found to exist among the neighbouring groups and those with common origins than among groups with widely separated origins.

THE ABO BLOOD GROUPS

Interests in blood groups developed from the practice of transfusion of blood from one person to another. Such experiments began at least as early as the 17th century and were usually uneventful. But in a few instances, the recipient of the blood died following the transfusion. In 1900, Karl Landsteiner, who latter received Nobel prize for this work, discovered why. He noticed that when the red cells from the blood of one individual are introduced into the serum of another, the cells may clump - a reaction called Agglutination.

Landsteiner and his students in 1945 found that they would group the humans into four types on the basis of these reactions. The four types can be explained by the presence or absence of either or both of two substances (the A and B antigens) on the cells. Agglutination was soon associated with the transfusion accidents. For instance, transfusing A type blood into B group patient could produce a severe reaction. The reason why transfusion of blood of the wrong type produce serious and sometimes fatal results is that human blood contains antibodies as well as antigens. The red cells of the donor are agglutinated if the recipient has antibodies to their antigens. Fortunately, blood owners of type `O' have red cells that are not normally clumped and destroyed by anyone else's serum, so that the blood of group `O' persons called universal donors can be given in an emergency to individuals of other type.

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BLOOD GROUP	ANTIGEN	ANTIBODY
A	A	b
B	B	a
AB	A,B	No antibody
O	No antigen	a, b

To study the genetic bases of traits we begin by analysing the phenotype of the trait. Phenotypes can be seen and measured but we cannot directly observe the controlling genes. Once the various phenotypes are known, it is possible to figure out the number of different alleles that are involved in their inheritance by making up a pedigree. A pedigree is an analysis of the phenotypes of related persons.

The A B O blood groups were earliest traits to be analysed by pedigrees. Landsteiner was able to identify the phenotypes A, B, AB and O by observing the pattern of clumping when one person's blood cells were mixed with the serum of another.

Once the Phenotypes were known, scientists worked to determine genes that govern inheritance of different types. Mating between type `O', type `A' and type `B' individuals yielded more children of types A and B than of `O'. This indicated that the alleles controlling A and B Phenotypes are dominant over the type `O' allele. Since children of parent of type A and parent of type B showed AB phenotypes, it was assumed that both A and B alleles are dominant. Ultimately the existence of three alleles A, B and O was proposed to account to the known phenotypes. There were thus six possible genotypes - AA, AO, BO, BB, AB and OO. But there only four phenotypes because `O' is recessive to both A and B alleles so that AO and BO are expressed as A and B phenotypes.

GENOTYPE	PHENOTYPE
AA	A
AO	A
BB	B
BO	B
AB	AB
OO	O

The world distribution of A B O blood group frequencies is now better known than that of any other human polymorphism. In general the gene for the blood group `O' is at a minimum in the centre of the European, Asian and African landmass and reaches its highest frequency among the American-Indians. The gene for the blood group A also tends to be somewhat peripheral in distribution, where as a gene for group B is centrally distributed although not predominant in any population. Among Australian aborigines, the B gene is found only in the North where it may have been introduced from New Guinea or elsewhere. The B gene probably did not occur in the New World prior to Columbus. Formerly it was assumed that the distribution of ABO blood group genes nearly reflected the movements and mixtures of peoples with different gene frequencies. It now seems more reasonable that different ratios of these genes occur under different conditions. For example, the relatively high ratios of the gene for blood group B may occur in the areas of urban civilization due to some advantages of its possessors in resistance to epidemic diseases such as Plague and small pox, which spread among densely settled populations.

Blood group B is very much prevalent in Asiatic people with a maximum concentration in the central Asian countries among the mongoloids (35 - 37%). The frequency of this blood group decreases as we proceed Westwards, where it shows a 25% concentration in Egypt, 22% in Ukraine, 15% in Europe and shows a very less concentration of about 2% in American population and is completely absent in North and South American Indians.

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Blood group A has the highest frequency in central and Western Europe, with 45% itself in England. Blood group `O' is prevalent in North and South American populations where it reaches a rate of 96% in the former. In the world statistics, blood group `O' is common followed by blood group A which is trailed by blood group B.

MNS SYSTEM

Karl Landsteiner, the discoverer of the blood groups, continued his experiments and in 1927 he and one of his associates, Levine, discovered that when rabbits were injected with human blood, a few of the animals produced antibodies capable of reacting with some but not all human blood specimens. The antigen in the human blood thus identified was designated as M. A second substance N which occurs in some individuals who have M and in all non-M individuals was soon detected by Landsteiner and Levine. The antigens N and M differ from A and B substances in that the corresponding antibodies are not found in human blood. To test for these human antigens, the antibodies were prepared in the Rabbit. The antibodies to most human antigens are prepared in animals because the antibodies do not occur naturally in human blood. There are two alleles M and N. An individual with an M gene on each of a pair of chromosomes is of type of M; likewise an individual homozygous for N is of type N, but the blood of heterozygous individuals reacts with both kinds of antisera and they are of type of MN. There is thus no Mendelian dominance and the alleles M and N are expressed equally and are codominant. The MN system gene frequencies like those of ABO system vary throughout the world suggesting that different genotypes are advantageous under different conditions.

More recently, two additional antisera anti-S and anti-s have been prepared. One or the other or both of these react with all blood specimens. S and s are closely related to M and N because the frequency of S is not the same in individuals of type M, MN and N. The system may therefore be referred to as the MNSs or simply the MNS system.

GENOTYPE	PHENOTYPE
MMSS	MS
MMSs	MS
MNSS	MNS
MNSs	MNS
MMSS	MS
MMss	Ms
NNSs	NS
NNss	Ns
NNSS	NS

Rh System

Landsteiner and his associates, especially Philip Levine and A.S.Weiner, continued to make discoveries. In 1939, Levine and R.E. Stetson noted an unusual case of a woman patient whose serum agglutinated (her blood group was of "O") blood of some other group O persons. The following year, Landsteiner and Wiener, injected rabbits with the blood of Rhesus monkeys and found that the resulting anti-rhesus antibodies in the rabbit blood serum agglutinated not only the blood of other monkeys but also that of some humans. It was then shown that the same people react to both kinds of anti-serum, rabbit and human. The antigen they show is designated Rh, because it occurs in the blood of Rhesus monkeys. A series of subtypes of Rh antigen have since been established.

The alleles R and r are respectively necessary for the Rh +ve and Rh -ve individuals. Among the Mongoloids, Rh _ve is very rare and among the whites it is relatively higher.

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ADDITIONAL BLOOD TYPES AND THE SECRETOR FACTOR

Not only there are sub-groups of Rh, but there is at least one important subgroup A2 of A, and there are also several subtypes of M and N. In addition, there are a large number of other antigens that are independent of the previously mentioned systems or related in still unknown ways. These are usually named for the individual in whom the antigen was first found. New types continue to turn up from time to time. They often are discovered when an individual receives repeated transfusions for some diseases and then develops serum antibodies for an antigen of a type he or she lacks but which was in one or more of the transfusions.

The sector factor is also of value in genetic comparison of populations. In some individuals there are substances called secretors. Evidence of their ABO status is found in saliva and in other body fluids. In other persons of the same group the substances are not dissolvable in water and do not occur in the saliva. Besides anti-A and anti-B sera that can be used to test the saliva of A, B and AB individuals, there are anti-H antibodies that react with saliva of some group `O' individuals. Persons lacking A or B antigens have the corresponding antibodies (anti-A, anti-B) respectively. In the other blood group systems, antibodies are not normally found in individuals who lack the antigen. These other systems therefore have less immediate significance for blood transfusions. Nevertheless, from the anthropological standpoint these factors are equally interested.

Taken in aggregate, the blood groups permit geneticist to plot the frequency distribution of a considerable number of human beings. William C. Boyd, one of the first and strongest advocates of the application of genetic methods to anthropology, has used the distribution of blood group genes to divide the human species into races. Most anthropologists would now prefer to study the various blood group systems in their variations, distributions and changes with reference to ecological settings, disease, and migrations, without invoking any concept of race that applies to overall appearance. Analyses of geographic distribution of blood group are a substitute for and not an adjunct to, racial classification and its interpretation.

THE BLOOD GROUP SYSTEMS IN THE HUMANS

BLOOD GROUP	YEAR OF DISCOVERY	NUMBER OF KNOWN ANTIGENS
ABO	1900	5 or more
MNSs	1927	29
P	1927	3
Rhesus	1940	28 or 30
Lutherian	1945	2 or 3
Kell – Cellano	1946	9
Lewis	1946	3
Duffy	1950	2
Kidd	1951	2
Deigo	1955	1
Auberger	1961	1 or more
X 5	1962	1
Sciana	1962	2
Dombrock	1965	1

HAEMOGLOBIN

A molecule of the protein haemoglobin consists of four polypeptide chains, which are normally present in two pairs. Different kinds of such chains exist and each is designated by a Greek letter. Variant haemoglobins are normally assigned English letters. Thus adult normal haemoglobin-A has two alpha and two beta chains. Several other variants are discussed below.

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The haemoglobin molecule consists of four polypeptide chains. There are several kinds of polypeptide chains in haemoglobin. The principle kind of adult haemoglobin is haemoglobin A (HbA). The foetal haemoglobin is represented as HbF. The HbA has a mutant form HbS, found in the persons suffering from Sickle-celled anaemia. Another polymorphic form of haemoglobin, HbC is also found which differs from the normal HbA, in its amino acid sequence. HbE is another polymorphic form of haemoglobin formed due to the mutation in HbA. HbC and HbE are related to a malarial parasite affecting those different from HbA and HbS.

A blood protein called Glucose-6-Phosphate dehydrogenase is found in the RBC. Certain populations show the deficiency of this enzyme. They are found in Africa, Asia, Indonesian Islands, Burma and India etc. The distribution of this enzyme in the populations is correlated with the infection by a malarial parasite Plasmodium falciparum. The deficiency of this enzyme has a sex-linked inheritance. The G-6-PD deficiency is found in people whose ancestors have lived in the areas of the world with a high incidence of Malaria. Individuals who inherit the deficiency are better equipped, than non-deficient persons, to resist malarial infections.

RACIAL CRITERIA, RACIAL TRAITS IN RELATION TO HEREDITY AND ENVIRONMENT

People owe their differences to genetic and environmental factors. Both these interact so that the degree of manifestation of genetic factors depends on the environment. To a greater extent such interaction has played an important role in producing differences between people which, in turn has allowed human life to exist in a wide range of environments than would have been possible without this variety. The question of significance of the persistence of this variety has been much debated. Explanations have been put forth in reference to natural selection which ensures that the members of the species conform to the best type. However, the complexity of environments and relation to organisms are of paramount importance which has ultimately resulted into polymorphic states.

As a gene may influence many characters and a character may be influenced by many genes, the facts of heredity are very complicated. Man is a world-wide species. The original roots of racial diversity must have been partly in the production of types suited to particular conditions. However, man has tended to maintain and magnify, for many reasons, superficial differences in features. For example, skin colour is believed to reflect racial differences. Human communities remain distinct partly because they contain individuals whose genes endow them with different physical capacities. The pattern of human life is not based upon communities composed of identical individuals. In fact we have varied capacities, motivations, satisfactions and hence opinions.

Since man has been able to control the conditions around him, the question of what constitutes his 'environment' assumes a new aspect. The extremes of some climatic conditions may stimulate the inhabitants to find ways to isolate themselves from heat or cold. Living in centrally heated and air conditioned buildings has its own hazards for pharynx and respiratory system. This may set up selection pressures which are slightly different than others who are subjected to the normal hazards of climate. Infectious disease constitutes another environmental factor. Naturally, there are differences between people in the pattern of their resistance to infection. Thus, environment constitutes a very wide range which refers to the sum total of social, economic, moral, intellectual and all other patterns of learned behaviour which influence the genes along with the behaviour pattern of human beings. This however is always coupled with all the external physical forces that affect the life of an organism.

Both genetic and environmental factors influence most of our characteristics. Some categories can be recognised which include elements of both heredity and environment. These may broadly be categorised as (i) differences due to accident, (ii) differences due to adaptive response during the lifetime of the individual, and (iii) differences due to cultural influences.

Generally, people differ in power of their adaptation because of differences in the degree of the hereditary control which is not easily altered by environment. On the other hand, there are characteristics like those

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of brain which are subject to hereditary influences but greatly affected by the environment. The endocrine system, muscles or blood are intermediate between these extremes. Thus there are some features having greater while others having lesser capacity to adapt during development. These powers of adaptation are inherited which help in responding various organs to environmental influences.

Human differences may arise due to accident. Some people are born a little wiser than others. Similarly children born to mothers who are affected by German measles during the early months of pregnancy suffer from serious defects in the heart, brain, bones and elsewhere. The features also differ in their susceptibility to such afflictions. The complication of pregnancy leads to a variety of clinical conditions `ranging' from cerebral palsy through epilepsy, mental deficiency, behaviour disturbances and reading disturbances.

People depend on the environment for food, water, air, warmth and many other factors. They are greatly influenced by the climate. It is suggested that there is a high incidence of congenital malformations in areas containing igneous rocks with a relatively high radioactivity.

The relationship of climate and disease is a specially interesting and important area. Diseases due to prevalence of parasites and intermediate hosts in certain areas are major problem of homeostasis e.g., the prevalence of malaria, hookworms, yellow fever, dysentery, sleeping sickness and other tropical conditions. Resistance to all these depend to a large extent upon hereditary factors and on the capacity to survive with parasites or worms. Naturally we depend on the special conditions and many organs of the body including brain enable an individual to tolerate his diseased condition. Selection certainly operates and that is why people inhabiting tropical countries differ from those of temperate zones (those zones are devoid of intermediate hosts and hence the diseases mentioned above are not prevalent). Some of the differences between humans are undoubtedly due to the fact that survival depends largely on the capacity to resist infection. Sickle cell gene provides the best example where change of one amino acid in the haemoglobin reduces its value as an oxygen carrier. This is fatal when the individual is homozygous. However, in heterozygous condition it confers some protection against the malarial parasite. Possibly there are variations in the capacity to resist all infective diseases (by production of anti-bodies). Resistance to infection by diphtheria bacilli is partly the result of the presence of a hereditary factor. Similarly the familiar distribution of Poliomyelitis suggests the involvement of hereditary factors. Very little is known about the capacity to resist even the more ordinary infections.

In addition to the above, every habitat imposes a particular climatic regime on man which varies widely and especially so because man can make appropriate adjustments with his habitat. Human body is sensitive to many ailments that go to make up a climate of the place. This sensitivity and responsiveness help to maintain homeostasis. Under the conditions of extreme heat or cold, the heat regulatory system acts to maintain the body temperature within certain limits. At high altitudes it is the respiratory system which keeps the pressure of oxygen and carbon dioxide of the body fluids adjusted within stable limits. In addition to the stresses imposed by thermal factors and the low barometric pressure (at high altitudes) other environmental hazards are provided by excess of short wave radiation including both ultraviolet light and ionizing radiations. The efficient adaptation of human body to climatic change is necessary for the attainment of bodily comfort, performance of physical work without undue fatigue, performance of skilled work involving alertness and dexterity and attainment of normal growth and development.

As regards response to heat, the total number of sweat glands varies in different individuals, though striking differences between different racial groups do not seem to exist. In an investigation by Weiner, South African Bantus and Europeans were found to have very similar sweat gland count. In Europeans the density of glands has a distribution in decreasing order as follows: it is maximum in the upper limb (dorsum of hand>fore arm>upper arm), followed by the lower limb (foot>leg>thigh) and then over the trunk, abdomen and thorax. In Indians and Africans, the gradients are similar.

The complex changes that make up physiological acclimatisation to heat have been demonstrated in peoples of different races living in hot climate (in Nigerians, Chinese, Indians, Malayans living in Malay,

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Kalahari Bushmen, South African Bantus as well as in Europeans in the tropics and in hot deserts). This physiological acclimatisation enables man to carry out his active life under a wide variety of hot conditions and makes it possible for our species to occupy very different kinds of tropical and equatorial habitats. It seems that genetic selection of various bodily characters for life and diverse climate has been superimposed on this physiological plasticity.

There are indications that acclimatisation to cold develops gradually. Increased tolerance to cold can be acquired. The Australian aborigines can sleep naked with little apparent discomfort at air temperatures of about 32oF and radiant temperatures of 50oF. These conditions could not be tolerated and shivering and discomfort were apparent under the same conditions amongst the investigators. Similarly, Bushmen of the Kalahari sleep in extremely cold conditions with only a slight protection of single covering and small fire. A high degree of cold tolerance can be acquired by Europeans and many Norwegians. Eskimos are stated to have a far greater tolerance to cold in the hands than White men. This ability of Eskimos to use the hands efficiently at low temperatures is associated with an increased blood flow. However, the role of thyroid, like the adrenal gland, is involved in cold adjustment. Thus, the available evidence points strongly to the existence of acquired cold tolerance in which physiological adjustments play a large part.

Differences in the average physique of different populations are of significance in climatic adaptations as they conform to the ecological rules of Bergmann (1847) and Allen (1877). According to Bergmann's rule "within the polytypic warm blooded species the body size of a subspecies usually increases with decreasing temperature of its habitat". Allen's rule states that "in warm blooded species there tends to be an increase in the relative size of the protruding organs such as ears and the tail with increasing temperature of the habitat". Several studies have demonstrated that human body size and shape tend to follow these rules. The mean body weight in hot regions is demonstrably lower than that in temperate and cooler climate. Roberts has shown that the lower limbs tend to be longer in hotter climates. The dimensions of the trunk also become less in hotter climates.

The variation in the subcutaneous fat covering in different populations has not been adequately studied. American Negroes have a smaller range of skin fold thickness than American Whites. Possibly Eskimos have thicker fat covering than Negroes.

Owing to the differences in adult physique, growth pattern also shows some relationship to climatic variation. The growth period is prolonged and maturation somewhat delayed in warm periods. A relatively greater height per unit body weight is attained by a delayed skeletal maturation. Coon, Garn and Birdsell have emphasised that Mongoloid face exhibits features adapted to life in extreme cold, e.g., the reduction of brow-ridges and the frontal sinuses, flattening and widening of orbital regions to permit more fat padding and the reduction of nasal prominence. Amerindians and Eskimos are not affected by cold in spite of their long periods of exposure to cold. Climatic variables have a high correlation with the shape of the nasal aperture. It has been suggested that the nose shape may be more concerned functionally with moistening the inspired air rather than with heat exchange. Perhaps because of this reason narrow nose is found in both hot and cold deserts (having excessive dryness). The activity of the cilia of the respiratory epithelium is more reduced by drying than by heating or cooling.

The above discussion indicates some functional advantages of certain body characters and their regional occurrence in certain people. It seems that regional selection has proceeded on Darwinian lines and climatic differences may be because of genetic differences. On the basis of twin studies we know that the variations in body shape, size, fat deposition, growth pattern, etc., are all determined largely by the genetic constitution than by purely environmental factors. On the other hand, certain genotypes owing to the multifactor recombination like that of nose shape or limb length and trunk length ratio remain unaffected on change of environment. It has also been suggested that exposure to high temperature during the growth period can result in morphological changes conferring higher resistance to heat stress. This capacity to make immediate responses may have resulted into rapid selection and establishment of appropriate growth patterns in some populations genetically. Each major racial group (Caucasoid, Mongoloid, Negroid or Australia) occupies a wide range of climate possibly because of the physiological

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acclimatisation and partly because of the existence of Bergmann and Allen body size differences and partly to technological adjustment to climatic conditions.

Man's cultural adaptation has provided an additional safeguard. The very spread of man over contrasting habitats would have been impossible if biological adaptation was not supplemented by technological measures. The history of the technology of housing, heating, cooling and clothing suggests that housing demands a high insulation of walls for facing extreme cold demands. Similarly, roof and floor must have been adjusted to make the best use of space in warming appliances and to provide efficient ventilation. The winter habitation of Central Eskimo and those of the Alaska and Greenland display a remarkable mastery of these principles. Man's technological understanding of hot climate had undergone much development by the beginning of the historical period. Humans have developed appropriate building methods to deal with the complex factors of hot, dry or humid conditions. Such methods are always related to physiological principles and standards of comfort and efficiency. Like housing we may examine clothing-including footwear and hand wear - in terms of heat exchange in different climates both hot and cold.

The implications of heredity and environment in the ethnic differentiation of man are not easy to evaluate. However, the nature of the probable mechanism or processes involved in the evolution of ethnic groups of man can be appreciated if we postulate that originally the ancestral human population (which has genetically heterogeneous) inhabited a circumscribed region of the earth. However, gradually individual families or groups of families dispersed over great distances at different times from the original ancestral habitation. Some of these groups became geographically isolated from one another and remained as such for generations. These isolated groups must have been subjected to natural selection, mutation, isolation, genetic drift, hybridisation and social selection. We have already discussed all these factors in detail. However, the implications of natural selection in the context of the interaction of heredity and environment need to be considered again. Natural selection preserves through the action of environment such variations that arise and are beneficial to the organism under its conditions of life (such variations are said to be adaptive). Since natural selection neither has a purposeful design nor foresighted planning, therefore, its results are always relative to the particular environment in which the organisms are living and to their structures and habits. Adaptive traits, therefore, are adaptive only in relation to the environment where structure and function fit harmoniously.

We know that the skin colour is an adaptive character. It has selective value. In human population darkly pigmented people are geographically distributed in high temperature, sunlight and humidity regions while lightly pigmented people are in regions of lower temperature-sunlight-humidity conditions. The marked gradients shown by pigmentation confirm Gloger's rule which states that melanin pigmentation tends to increase in the warm and humid part of the species range. It seems that black pigmentation is certainly promoted by high humidity and high temperature whereas aridity with high temperature promotes the formation of reddish, yellowish and reddish-brown pigment.

Nose is another adaptive character. Thomson and Buxton, on the basis of their research work, have stated that air at higher temperatures is breathed more easily through a broad nose than the narrow one. Because a broad nose allows maximum exhalation of hot air from the lungs, therefore, in hot climates (where heat dissipation is essential) a broad nose will be at selective advantage. On the other hand, it has been argued that narrow nose represents an adaptation to cold climate where it is necessary that the temperature of the air must be adjusted to the proper warmth and humidity. Kenny has shown that most human desert dwellers have a slender nose which can provide cooling and saturation of the inspired air before its exposure to the respiratory surfaces. Weinger stated that the broad nose is correlated rather more significantly with external absolute humidity than with air temperature and humidity.

It has further been observed that populations living in extremely cold regions, e.g., Arctic area, Siberia, Alaska and Greenland are relatively short and tend to be well padded with fat. Their surface area is less as compared to those who have been living in regions of high temperature. The latter naturally would radiate as much heat as possible while the former as little. Body weight is also correlated with mean

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temperature. Thus the high body weight is associated with high temperatures whereas medium body weight to more temperate regions. Accumulation of fat in cold climate provides insulting qualities which affect body heat loss. A decrease of surface is naturally advantageous in maintaining body height in the cold environment. A large body size thus serves to reduce heat radiation and a small body size increases it. These facts are in accordance with Bergmann's rule which in this context states that smaller sized members of a population are to be found in the warmer parts of the range and the larger sized members in the cooler areas.

Thus it is clear that the action of natural selection varies under different conditions and the rate at which it operates depends upon the character or quality involved. It may, however, be emphasised that even the organisms possessing certain selective advantages certainly need the cooperation of their fellowmen in order to be preserved. In fact, cooperation is an integral part of the process without which the survival itself would be seriously jeopardised. Thus, it is the cohesive effect of natural selection as represented by the cooperative aspect which ensures the perpetuation of the species or group. The `Favoured Races' are preserved by cooperation. Cooperation has thus certainly played a role in the evolution of man and also in the interaction of heredity and environment.

RACIAL CLASSIFICATION AND DIFFERENTIATION

One of the most striking features of human species is its awesome variety. In the preceding chapter we have discussed the factors that are responsible for the formation of races. We have noted that races are manifestation of the on-going evolutionary processes and apart from genetic factors, environment, nutrition and culture also play an important role in their formation. We have also seen the various criteria employed by raciologists in the classification of human populations. We shall now focus our attention to the different types of racial / ethnic groups in the world, their characteristics and distribution.

History

The first scientific attempt towards classification of the human populations can be credited to the Swedish naturalist Carolus Linnaeus. He has classified our species Homo sapiens into four groups

1. Homo americanus
2. Homo europeans
3. Homo asiaticus
4. Homo afer (Africans)

In 1775, John Frederick Blumenbach, a German biologist has classified the human populations into five groups or races on the basis of skin color. They are...

1. Caucasian or White: Europeans (except Lapps and Finns). Also found in North Africa, West Asia.
2. Mongolian or Yellow: Includes the Lapplanders and Finns, Eskimos of Americas, Asia
3. Ethiopian or Black: Includes all of Africa except North Africa where Caucasians are present.
4. American or Red: Non Mongolian populations of America
5. Malayan or Brown: All the inhabitants of Pacific.

A number of different classifications have been proposed by different scientists including Pickering in 1848, Topinard in 1885, Quatrafages in 1889 and Deniker in 1889 and many others. For example, Montandon, in 1933 has identified five races of mankind – Pygmoid, Veddoid, Negroid, Australoid and

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Europoid. In 1925, Ottenberg attempted to classify the races on the basis of ABO blood group system and has identified six races...

1. European: Norwegians, Italians, Germans, English, French, Swedes, Greeks, Anglo-Saxon Americans.
2. Intermediate: Russians, Turks, Arabs, Armenians, Polish Jews.
3. Hunan: Japanese, Southern Chinese, Romanian Jews, Hungarians.
4. Hindu–Manchu: Koreans, Manchu, Northern Chinese, Gypsies, Hindus
5. Afro-Malaysian: Negroes, Malayans, African Americans.
6. Pacific-Americans: Amerindians, Icelanders, Australian Aborigines.

In 1946 Weiner proposed another classification system based on the frequencies of alleles for ABO system, MNS system, Rh blood factor. He classification identifies six groups of peoples on this basis into Caucasoid, Negroid, Mongoloid, Asiatic, Australian and Amerindian.

In 1931 and subsequently in 1947 Hooton, an American anthropologist have proposed the now widely accepted classification of races into three major groups – Caucasoid, Negroid and Mongoloid – with each one of them further divided into different subtypes.

It is surprising to know that till date more than twenty to thirty different classifications have been given by different scientists at different times. This highlights the lack of agreement amongst them as to what constitutes a race and how many races can be identified in the world. Disagreements exist as different anthropologists use different criteria for classification of races.

We shall now look at the most popular classification of the human races into three races, Caucasoid, Mongoloid and Negroid, with many subtypes. The following table gives a general idea about their racial features.

Trait	Caucasoid	Negroid	Mongoloid
Skin Color	Light reddish white to olive brown.	Brown to brown-black.	Light yellow to yellow-brown.
Hair (Head)	Color: Light blond to dark brown Texture: Fine to medium Form: Straight to wavy	Color: Brown-black Texture: Coarse Form: Curly to wooly	Color: Brown to brown-black Texture: Coarse Form: Straight
Hair (Body) Quantity	Moderate to profuse	Slight	Sparse
Eye	Light blue to dark brown in color	Brown to brown-black in color	Brown to dark brown in color. Presence of epicanthic fold
Head Form	Dolicocephalic to brachycephalic	Predominantly dolicocephalic	Predominantly brachycephalic
Face	Narrow to Medium-broad	Medium-broad to narrow. Presence of prognathism.	Medium-broad to very broad. High cheek bones.
Nose	Leptorrhine to mesorrhine. High bridge.	Platyrrhine. Low bridge.	Mesorrhine to platyrrhine. Low to medium bridge.
Stature	Medium to tall	Very short to tall	Medium to short
ABO Blood Group	High incidence of A	High incidence of A and B	High incidence of A
Rh Factor	Highest frequency of Rh –ve	Moderate frequency of Rh –ve	Rare occurrence of Rh –ve

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Let us now discuss each of these major races in some detail.

1. CAUCASOID

The Caucasoid include many ethnic groups and racial elements. The following are some of the features of Caucasoid race in general.

1. Skin Color: White, olive, light to dark brown
2. Flat wavy to curly hair
3. Light colored hair with occasional black hair
4. Medium to fine textured hair with rare occurrence of coarse hair
5. Moderate or abundant body and facial hair
6. Dolicocephalic to brachycephalic head
7. Leptorrhine to mesorrhine nose, rarely platyrrhine
8. High nasal bridge
9. Not so prominent cheek bones
10. Absence of prognathism
11. Thin lips
12. Pronounced or medium chin
13. High forehead
14. Lighter shade of eye color.

The ethnic groups which fall into this category are Mediterranean, Nordic, Alpine, East Baltic, Dinaric, Armenoid, Keltic, Lapp, Indo-Dravidian, Polynesian and Ainu.

A. Mediterranean

Though this group derives its name from the fact that they originally inhabited the shores of Mediterranean Sea, today they have migrated to places as far as Asia. They can be seen in Portugal, Italy, France, Greece, Turkey, North Africa, Arabia, Iran, Afghanistan, Pakistan and India.

Sub-Types: There are three sub-types of the Mediterranean group – Ibero-Insular, Littoral and Irano-Afghan.

The Ibero-Insular sub-type, also called Classic Mediterranean or Basic Mediterranean, are characterized by light brown skin color, wavy or curly black hair, dolicocephalic to mesocephalic head, leptorrhine nose, narrow and oval face with pointed chin, dark eyes and somewhat delicately build bodies of medium stature. They are distributed in the whole of Mediterranean basin including Spain, Portugal, France and Italy. They are also sporadically distributed across eastern, central and north-western Europe. This sub-type can also be seen in Egypt, Morocco and other parts of Arabia including Palestine.

The Littoral, also called Atlanto-Mediterranean group is characterized by dark skins, very wavy to curly hair which is dark in color, dolicocephalic to mesocephalic head, retreating forehead, well developed brow-ridges or supra-orbital ridges, straight nose, long face with slightly prominent cheek bones, usually medium tall stature who are more robustly built than their classic counterparts. They are found amongst

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the populations of North Africa, Iraq, Palestine, Eastern Balkans, some parts of Arabia, sparsely scattered in Spain, British Isles and Portugal.

The Irano-Afghan, also called the Indo-Afghan sub-type is characterized by very light brown skin color, black and wavy hair, abundant facial hair, dolicocephalic to mesocephalic head, finely cut, leptorrhine nose with convex bridge, long and narrow face, dark eyes and tall to medium statures. They are distributed across Iraq, Iran, Afghanistan, Pakistan and North-west India.

B. Nordic

The typical representatives of this group are the Scandinavians. People belonging to this group can be seen in Baltic region, Northern Germany, Northern France, parts of Netherlands and Belgium, British Isles and USA.

The main characteristic features of this group are the presence of reddish-white skin, slightly wavy and rarely curly hair with variable hair color including reddish brown, golden brown and ash brown, fine to medium hair texture, sparse or medium body and facial hair, mesocephalic head, usually straight, long and prominent nose, long and narrow face with strongly developed brow ridges, prominent chin, thin or medium lips, blue or grey eyes and tall stature.

C. Alpine

This group is characteristic to populations living in Central Europe, Balkans, Denmark, Norway, North Italy etc. They are sporadically distributed across Europe. Alpine populations are an admixture of Nordic and Mediterranean groups. These people are referred to as the Beaker Folk or Round Barrow Men or Bronze Age Men and are considered a cross between the Nordic and Mediterranean groups.

These groups are characterized by reddish white skin, wavy hair, large, brachycephalic head, well developed brow ridges, flattened occiput, leptorrhine nose, long and narrow face, square chin and large jaw, blue or blue-grey eyes, very tall and muscular and robust bodies

D. East Baltic

East Baltic groups can be found in Baltic States, Russia, North Eastern Germany, Poland, and Finland.

The group is characterized by having a creamy white skin color, light-ash blond or ash brown colored straight hair, medium to coarse texture of hair, moderately developed facial hair with sparely distributed body hair, brachycephalic head with high and broad forehead, flat occiput, square face with prominent cheekbones, square lower jaws with well developed chin, prominent mesorrhine nose which is convex in profile, low nasal bridge with snubbed tip, light blue or grey eye color and short to medium stature bodies.

E. Dinaric

The Dinaric, also called Adriatic or Illyrian, are distriburted across Dinaric Alps region in Yugoslavia, Albania, Austrian Tyrol and sporadically in Central Europe.

These Groups exhibit a combination of racial elements characteristic of Alpine, Nordic, Armenoid and Atlanto-Mediterranean. They are characterized by lighter skin color, straight to wavy hair form which is sometimes curly with medium brown to dark brown color and medium texture; abundant facial and body hair; brachycephalic head, with very high and extremely flat occiput; sloping forehead, well developed brow ridges; leptorrhine nose and a convex nasal profile; fleshy nasal tip; longer and narrower face, heaver and projecting chin; brown or lighter eye color and tall stature bodies.

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F. Armenoid

The Amernoids live in Turkey, Palestine and Syria. They can also be found in Iran and Balkan States. These groups are a considerable mix of Classic Mediterranean, Alpine, Nordic and Indo-Afghan racial types.

The phenotypes of this group include olive skin, dark brown to black hair which is wavy or curly in form, with coarse to medium texture; abundant facial and body hair; thick eyebrows; brachycephalic head, high head with vertical occiput and sloping forehead; prominent, leptorrhine nose with high and broad nasal root and convex bridge, fleshy and depressed tip; long and narrow face with well developed cheek bones, medium prominence of chin, moderately prominent lips and medium to tall stature. In these groups the body has a tendency to obesity.

G. Keltic

Keltic groups are distributed in Iceland, Scotland and Wales. They also are sporadically distributed across England and Western parts of Europe.

They are characterized by pale white skin, medium to dark brown skin that can be red; usually wavy or curly and straight hair; mesocephalic head; leptorrhine nose, with a convex profile; long and narrow face and compressed cheek bones; blue or grey colored eyes and tall stature.

H. Lapp

The Lapps are distributed across North Scandinavia, Sweden, Norway, Northern Finland and North West Russia. Some people classify the Lapps with Mongoloid groups.

They are characterized by having graying yellow to yellowish brown skins; dark brown hair which is slightly wavy; sparse body and facial hair; mesorrhine nose with a concave profile and snubbed tip; moderately broad and very short face, moderately prominent cheek bones; narrow forehead and faintly developed brow ridges; occasional presence of epicanthic fold and short stature.

I. Indo Dravidian

They are concentrated in South and Central India. These groups are predominantly Caucasoid and some mixture of Classic Mediterranean and Australoid elements can be seen.

They are characterized by light to dark brown skin; usually black and slightly wavy skin; plentiful hair on the head with medium distribution of facial and body hair; dolicocephalic head; rounded forehead; mesorrhine nose with depressed nasal root and straight profile; narrow and medium length face; moderately developed brow ridges and medium stature.

J. Polynesian

The Polynesian groups live in Polynesian Islands of the Pacific including New Zealand, Samoa, Marquesan and Hawaii. They are a composite race as they are essentially white people with both Mongoloid and Negroid mix.

They are characterized by light brown to yellow brown skin color; dark brown to black hair color which is usually wavy and sometimes straight; sparse body and facial hair; predominantly brachycephalic head with occasional occurrence of dolicocephalic and mesocephalic head; high forehead which is slightly sloping and narrow; flat occiput; prominent nose with slightly depressed root and convex profile; long and broad face with prominent cheek bones; moderately developed brow ridges and well developed chin; lips with medium fullness; medium to dark brown eyes with rare occurrence of mongoloid fold or epicanthic fold; tall stature and muscular body build.

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K. Ainu

Ainu's distribution is in Northern Japan, Yezo and South Sakhlain. They represent a very ancient stock of Japan who is predominantly Caucasoid with considerable Mongoloid mix. They resemble the Australian Aborigines in many features.

They are characterized by light brownish white skin; dark brown to black hair which is wavy; abundant hair on the body and face – hence are also sometimes referred to as "Hairy Ainu" mesocephalic head; mesorrhine nose with slightly depressed nasal root and convex profile; short face with medium breadth – mesoprosocpic and orthognathus; well developed jaw and chin; thin lips; medium to dark brown eyes which are horizontal and widely opened; occasional epicanthic fold; thickly set bodies with medium or short stature.

2. NEGROID

Some common features of Negroid are

1. Wooly or frizzly hair on the head which is black in color
2. Dark brown to black skin color with yellowish brown in some groups
3. Broad and flat nose with low bridge and root
4. Dark brown to black colored eyes
5. Thick and fleshy lips
6. Small ears with no lobe
7. Marked facial prognathism
8. Predominantly long head with protruding occiput and rounded head profile
9. Small brow ridges

The Negroid are further divided into African Negro and Oceanic Negro.

A. African Negro

This group is further divided into five sub-groups – True Negroes, Nilotic Negroes or Nilote, Bantu, Bushmen Hottentots and Negrillo.

True Negroes are characterized by dark brown skin color; wooly hair; tall stature, platyrrhine nose; dolicocephalic head with bulging forehead and prognathous face; thick lips; short legs and long arms. They are distributed across West Africa and Guinea Coast.

Nilotic Negroes or Nilotes are characterized by very dark skin color, very tall stature, slim figure, long legs, dolicocephalic head, wooly hair, platyrrhine nose, less marked facial prognathism and retreating forehead. Some examples of Nilotic Negroes are the Shilluk, Dinka and Kavirondo. They are distributed across Upper Nile Valley and Eastern Sudan.

Bantu are the numerous Bantu speaking people who inhabit Central and Southern Africa. It is not yet clearly ascertained the different ethnic groups who belong to this category. Their characteristic features show a wide range of variation. Their skin color varies from being dark chocolate to yellowish brown and black; hair is Negroid i.e., wooly, typically dolicocephalic head, stature is medium to tall. They possess generally narrower and more prominent nose than the true Negroes.

Bushmen-Hottentots group is composed of both the Bushmen and Hottentots who are physically similar with very little difference in their characters. They only differ from each other in their culture. The

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Hottentots are known as Khoi Khoi and the Bushmen as Khuai or San. While the Hottentots are found in South West Africa, the Bushmen are predominantly found in the Kalahari. They are characterized by having short hair on the head which coils up to spiral knots, leaving bare spaces between them – a hair form termed as pepper-corn, hair color is black; sparse facial and body hair; light to brownish yellow skin in Bushmen and light reddish yellow skin in Hottentots; dark brown to black eye color; the Hottentots are slightly taller than the Bushmen with average height of around 4.5 feet. Dolicocephalic head amongst the Hottentots and mesocephalic head amongst the Bushmen is seen. Face is shorter, squarer and orthognathus in the Bushmen while it is more elongated, triangular and progrnathus in the Hottentots. Prominent cheek bones, small chin, bulbous forehead, little developed brow-ridges; frequently lobeless ears, usually thick lips; platyrrhine nose with very broad and flat root, low bridge with concave nasal profile; narrow eyes, slanting with internal epicanthic fold, small hands and feet. One prominent characteristic feature amongst these groups is steatopygia, which is an immense deposit of fat in the buttocks), especially the women of Hottentots.

Negrillo, also called the African Pygmy are very short in stature with an average height of around 4.6 feet. They are characterized by short legs and long arms; steatopygia in the women, mesocephalic head; prognathic face with narrow chin; very flat and broad nose; very short hair on the head which is wooly and dark rusty brown in color and body hair yellowish brown to black. They are represented by the Akka, Ba Twa, Bam Bute and inhabit the equatorial forests of the Congo basin.

Pygmies are found in various regions of the world. Apart from the Congo basin, they are also found in Malay Peninsula, Sumatra, Andaman islands, Philippines, New Guinea etc. They can thus be categorized into African Pygmy, Oceanic Pygmy and Asiatic Pygmy. The last two are also termed the Negritos.

B. Oceanic Negroes

Oceanic negroes include the Negritos – Asiatic and Oceanic and Malenisian-Papuan subgroups.

The Negritos are further divided into Andamanese, Semang, Aeta (Asiatic) and Tapiro (Oceanic) groups. The Andamanese inhabit the Andaman Islands of India, Semang in central region of Malay Peninsula and East Sumatra, Aeta are the inhabitants of Philippine Island and the Tapiro live in New Guinea. It is a general belief that all the pygmies of the work are genetically interrelated and they constitute a common pygmy race. However recent researchers have shown that they don't constitute a race as such and the formation of Pygmy is attributed to a large extent to environmental factors. They are characterized by mesocephalic to brachycephalic heads, round faces, short and flattened noses. Skin color is dark chocolate brown and sometimes sooty black. Hair on the head is short and face and body hair is scanty.

Malenesian-Papuans inhabit New Guinea, Fiji, Admiralty Island, New Caledonia etc. They are characterized by a typical dolicocephalic head with high retreating forehead; heavy and continuous brow ridges and prognathous face. Broad nose, depressed nasal root with convex profile, platyrrhine nose in some; usually medium stature; black and wooly hair; abundant facial hair (scanty in some) are other features. Their stature varies from medium to tall with average height of 5.6 feet.

3. MONGOLOID

The following are characteristics of the Mongoloid group...

1. Hair which is black, straight and coarse
2. Yellow or yellow brown skin color
3. Scanty body and facial hair
4. Broad and flat face with prominent cheek bones
5. Obliquely set eyes with narrow slit-like openings with internal epicanthic fold

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This group is further sub-divided into four main groups based on their geographical locations – The Classic or Central Mongoloid, The Arctic or Northern Mongoloid or Eskimoids, The Southern or Indo-Malayan Mongoloids and The American Indians.

A. Classic or Central Mongoloids are found in Siberia, sporadically in Northern China, Mongolia and Tibet. The representative ethnic groups are Mongoloid tribes like Buriat, Koryak, Goldi, Gilyak and some Tibetans and northern Chinese. They are characterized by brachycephalic head, projecting occiput, rounded forehead, nose with no nasal depression and a very low root, straight to concave nasal profile, strongly developed cheek bones, yellow to yellow brown skin color, hair on the head straight, black and coarse, sparsely distributed facial and body hair, dark brown eyes which are obliquely set with a complete epicanthic fold.

B. Arctic or Eskimoid groups are distributed in northern Asia, Arctic coast of North America, Greenland, Labrador and Western Alaska. They are represented by the Eskimo, Chukchi, Kamchatdales, Yakuts, Samoyedes etc. They are characterized by yellowish skin color, straight black hair, large broad and flat face with prominent cheek bones; narrow and prominent nose; remarkably small hands and feet; large trunks and relatively short legs. Eyes are black and often straight; occasional occurrence of a complete epicanthic fold. The Western Eskimos are taller than the Alaskan Estimos. Head form ranges from brachycephalic to mesocephalic.

C. Indonesian-Malay Mongoloid are distributed across Southern China, Indo-China, Burma, Thailand, Malay Peninsula, Dutch East Indies, Philippines, Japan and interior of Malay Archipelago. They are characterized by an average stature of around 5.2 feet; brachycephalic to mesocephalic head; mesorrhine to platyrrhine nose; straight and black colored hair with occasional reddish tinge; scanty body and facial hair; light yellow brown to dark yellow brown skin; medium to dark brown colored eyes with internal epicanthic fold; short face and prominent cheek bones.

D. The American Indian or the Amerindian groups are predominantly mongoloid with some non-mongoloid racial strains like Caucasoid, Australoid and some Negroid elements. They are distributed in different parts of North, Middle and South America. They have black and rarely dark brown hair which is coarse, straight and sometime slightly wavy; scanty distribution of body and facial hair; yellow brown to red brown skin color; dark brown to black eye color; complete mongoloid fold is almost absent; broad face and prominent cheek bones; prominent chin; strongly developed brow ridges; forehead which slopes and face exhibits prognathism. Nose is predominantly mesorrhine and the stature is variable. Head form can either be brachycephalic or mesocephalic.

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9.6 AGE, SEX AND POPULATION VARIATION

In the chapter on Races and Racial Classification we have observed that human populations vary in morphological and genetic characteristics. Apart from this, it has been observed that human populations also vary greatly in physiological characteristics like haemoglobin level, fat level, blood pressure and sense perception. This variation in physiology is not only seen between different populations, but also among the members of the same population. These physiological characters vary so much that they are different in same person during different periods of time. In short, the physiological aspects in man vary depending on age, sex and population.

VARIATION IN HAEMOGLOBIN LEVEL

The Red Blood Corpuscles (RBC) of blood carries Haemoglobin. Haemoglobin has a complex molecular structure. It is made of a heme group which contains iron and four protein chains.

Metabolic Limit of RBC: The haemoglobin level in a cell fluid is approximately 35 grams per decilitre (gm/dl). The level of Haemoglobin (Hb) never rises above this level. This level is called the metabolic limit of RBC. Maximum concentration is seen in RBC.

In 1977, Owen has studied the variation in Hb level according to Age. He, along with Pearson, studied Quebec society in Canada and has indicated that from the age of 20 years to 60 years the level of Hb among the males is maintained at 15.3 gm/dl. The females reach the highest level of Hb, which is 13.5 gm/dl, by the age of 10 - 14. After the age of 14, the boys improve their Hb level by attaining 15.3 gm/dl whereas females more or less remain stagnant. This difference of 2 gm/dl between males and females can be attributed to the menstrual losses among the females. Moreover, there is lesser number of RBCs in a female. For this reason, Allan and Girdwood in 1982 have indicated that an Hb level of 12 gm/dl among males is considered to be a situation of Anemia whereas it is a normal situation among females.

There is a growing interest in ethnographical diversities in Hb level in different populations. It has been observed that there is a relative influence on Hb level by the activity level of the population. Those cultures which demand higher activity from their peoples have selective pressure on higher level of Hb. At the same time, culture and socio-economic statuses also influence Hb level. Since the nutritional levels of people are more often than not cultural adaptations, they significantly influence the Hb level among the people in a population. In most of the ethnic groups, it has been found that there is a gradual increase in Hb level till age of 30. Among the Americans, a constant value is maintained throughout the life and it gradually starts falling only in the old age.

Recent studies conducted in this context indicate that there is a growing secular trend in Hb level. The rise in Hb level in primitive societies is greater than the level prevalent in advanced societies several years ago. This trend is similar in all the ethnic groups.

GLUCOSE 6 PHOSPHATE DEHYDROGENASE – G6PD

There are some populations and individuals who show a deficiency of G6PD, which is an enzyme. G6PD deficiency is a heritable disorder. People with this deficiency exhibit a response in their blood called haemolytic response to some drugs and food items like fava or broad beans, which are common foods in the Middle East. If an individual with this deficiency consumes these food items, it may result in a condition called haemolytic anaemia or Favaism. The deficiency of this enzyme results in a condition where the subject's erythrocytes or red blood cells show low levels of reduced glutathione. Glutathione Stability Assay or Test is used to detect people with this problem. G6PD deficiency is an X-linked disorder and is caused by a single gene. Middle Eastern populations exhibit this condition with over 60% of incidence reported among the Kurdish Jews. In Africa, especially the equatorial region, the frequency of this problem is around 10-20% and the trait is also common in some parts of Greece and Iran. Thailand and Micronesian populations also exhibit this trait with incidence rates of around 10%.

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TRANSFERRINS

Transferrins are beta globulin factors present in the serum of blood and they are capable of binding the ferric ions in the blood tissue. They are also called Siderophilins. Normal individuals exhibit the presence of only one transferring band denoted by TfC. However, amongst some other groups, especially the Australian Aborigines, there is an extra band of transferring TfD. Transferrin is determined by a single gene. A third variant of transferring is also reported amongst some East Asian populations and Lapps and this variant is represented as Dem. Usually, transferrin polymorphism is determined by many alleles at a single locus in an individual's genome.

HAPTOGLOBINS

There are two globulins that are capable of biding free haemoglobin that has escaped from the RBCs. This binding is crucial as it prevents haemoglobin from damaging the kidneys by passing through glomeruli. The haptoglobins are found in the blood serum and can be separated by electrophoresis. In 1955, Smithies recognized that the patterns of hapt0globins are not same in all individuals and there are three types – Type 1-1, 2-2 and 2-1. Family studies have indicated that the above patterns are inherited and possibly more than one allele is involved. These alleles (recognized later to be two) are named Hpy^1 and Hpy^2. Population studies indicate that the frequency of Hpy^1 gene is around 40% in Western Europe. Its incidence is much higher in tropical Africa. It has lower values in South Africa (30%). In Asia, its frequency is much lower – about 10%. The selective forces at work are not clearly known. In some infections and inflammatory diseases, higher levels of haptoglobins are found.

VARIATION IN BODY FAT

There is a unique distribution of fat, especially subcutaneous fat over the body. Heredity and environment have a significant role to play in establishing the fat level of the person.

Subcutaneous fat layer has age related changes. Fat begins to be laid in the fetus at about 34 weeks and increases from then on until birth and until nine months after the birth.

When the child is attaining prepubescent height, i.e., from 9 months to 8 years, the fat level shows negative velocity. This decrease in the fat level is due to the breakdown of fat by the growth hormone. This fact is also supported in cases where a deficiency of growth hormone brings about an increase in fat level. The fat content present around bones and muscles decrease with increase in their size. The fat level of the body also shows sex related variations. While both boys and girls show reduction in fat level till the age of 8 years, the decrease in girls is comparatively slow.

An increase in the fat level of the body marks the period from 8 years to puberty, in both the sexes. It is only at adolescence that a temporary halt in increasing fat level is seen among the boys and this loss of fat is gained back after 20 years. This temporary halting among the boys is seen in trunk and limbs while there is no such loss among the girls.

The nutrition level and the level of activity are other facts with a socio-economic dimension which control fat level. Increase in nutrition and decrease in activity level increases the fat level of the body. This is the reason why in industrial societies fat related disorders are very common. Genetics also control the fat level deposition to an extent. In some ethnic groups, there is an increased deposition of fat in thighs and buttocks, a condition called steatopygia which occurs in Bushmen and Hottentots populations.

PULSE RATE

Pulse is the number of heart beats per minute. The pulse is measured at the wrist, neck, temple, groin, behind the knees, or on top of the foot. In these areas, an artery passes close to the skin. Measuring the pulse can give very important information about the health of a person. Any deviation from normal heart rate can indicate a medical condition. Fast pulse may signal the presence of an infection or dehydration. In emergency situations, the pulse rate can help determine if the patient's heart is pumping.

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The pulse measurement has other uses as well. During exercise or immediately after exercise, the pulse rate can give information about the fitness level and the health of a person.

Pulse rate varies according to the age of persons, whether they exercise or not and other physiological conditions in the body.

For resting heart rate:

- newborn infants; 100 to 160 beats per minute
- children 1 to 10 years; 70 to 120 beats per minute
- children over 10 and adults; 60 to 100 beats per minute
- well-trained athletes; 40 to 60 beats per minute

Resting heart rates that are consistently high (tachycardia) and resting heart rates that are below the normal values (bradycardia) may indicate a problem.

Changes in the oxygen pressure, high altitude areas and other metabolic demands on the body may also cause change in the pulse rates.

VARIATION IN BLOOD PRESSURE

The blood pumped by heart into blood vessels for its distribution to different parts of the body must leave the heart at some pressure because it has to overcome the resistance offered by frictional and other forces. The pressure at which blood leaves heart is called systolic blood pressure (SBP) and it gives an estimate of contractility of the heart. As heart expands or relaxes, these pressure drops, which give a measure of diastolic blood pressure (DBP). It is at this pressure that heart receives blood. A rise in the SBP indicate increased contraction whereas a rise in the DBP indicates decreased relaxation of the heart, both being harmful if they cross optimum levels. Blood-pressure is indicated in fraction: the upper number indicating SBP and the lower number indicating DBP. Though a pressure of 120/80 is considered an average for the adults, values as low as 115/70 is also considered normal. For the SBP, 100 plus age is also considered normal. Researches show positive correlation between blood-pressure and age, sex, heredity, body composition, and social status.

Knowledge about blood-pressure at the population level is of utmost importance in the assessment of health status, particularly in the detection of cardiovascular diseases. The variations in blood pressure according to age is high during growth and development up to adolescence, thereafter it shows a rhythmic variation on a daily basis. The blood pressure in the neonates during first day averages 70/50. There occurs increase in blood pressure during the next several months to approximately 90/60. The rate of increase is higher during this period. Thereafter, it remains more or less same throughout the adult period.

Using family studies, it has been possible to underline factors for blood pressure variation in a population. It has been estimated that 16% of blood pressure variation is due to environmental factors, 48 percent of additive genetic factors, and 36 percent to dominance. Heredity is indicated in causation of blood-pressure because it has been shown to run in families, though environment seems to be the precipitating agent.

According to Kaplan (1978), the populations can be arranged into three groups on the basis of variation of the blood pressure.

a) Hypertensive: Systolic blood pressure > 169 mm Hg

Diastolic blood pressure > 95 mm Hg

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b) Borderline cases: S.B.P. 140 - 160 mm Hg

D.B.P. 90 - 95 mm Hg

c) Normotensives: S.B.P. < 140 mm Hg

D.B.P. < 90 mm Hg

In a majority of rural Indian population the percentages of the normotensives have been almost always found to be more than 95%. In many cases, percentage of normotensives and hypertensives has been found to be higher than borderline. This indicates that in majority of the populations, persons either are normotensives or hypertensives. The cardiovascular system perhaps has the ability because of homeostatic mechanisms, to sustain changes due to stress and strain of environment. This seems to have a threshold limit. Once it is crossed, there is no scope of return and a normotensive becomes hypertensive. This may be reason for low percentage of borderlines compared to hypertensive patients.

On the contrary, studies conducted indicate that in industrialised population the proportion of normotensives in comparison to hypertensives is not so high. Hypertensives are around 5% subjects showing increase in both SBP and DBP. Another feature of the industrialised society is a small but significant group of borderlines. This is probably because the homeostatic mechanisms are disturbed in such societies due to nutritional and life style reasons. Among the nutritional reasons, absence of green vegetables and fruits and inclusion of fat to a great extent in the diet have been the most glaring reasons for the increase in percentage of border-line cases and hypertensive.

In cases of both rural and urban populations, it has been found that the variations of the blood pressure tend to be lower in the household samples than that of hospital. It is probably due to natural settings of the house, free from anxiety and tension. Blood pressure, as it is a multifactoral condition, may tend to recover in situations of normal, tension-free emotions.

Scientists associated with such population studies of blood pressure variation outside India are Boyce Attenborough, Harrison - Thornabrook and Sinneff. The group has been involved in recording population variation in blood pressure since 1970. They have analysed populations of New Guinea etc. and have reached to the same conclusions that in addition to heredity, diet and stress and strain of life are the two most important causative environmental factors of high blood-pressure.

VARIATION IN SENSE PERCEPTION

There have been some population studies of physiological variations that include survey of different sensory mechanisms. Among the various sensory mechanisms, the most thoroughly surveyed are ability to taste phenylthiocarbamide and red and green colour-blindness. The ability to taste PTC is an autosomal polymorphic trait (Vogel and Motulsky). Tasters with genotypes TT, Tt have the ability to taste the substance while non tasters are homozygous for the recessive allele 't'.

The defective colour vision has long been used as a genetic marker in the study of human variation. Red-green colour blindness is an X-linked trait in which individuals fail to distinguish red and green from other colours. Various populations inside and outside our country have been surveyed like Turkey, Balkans, Nigeria, Myanmar, etc. In India, such studies have mainly been conducted in South India.

Most of the studies related to PTC tasting have found sex-differences in the trait: females are found to be more sensitive to the taste of PTC than males and the difference is statistically significant in Balkan, Burmese and Nigerian population. Though females are more sensitive than males, the difference does not appear to be significant one. Muslims of India also do not show sex differences in ability to PTC tasting.

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9.7 CONCEPTS AND METHODS OF ECOLOGICAL ANTHROPOLOGY

CONCEPT OF ECOLOGICAL ANTHROPOLOGY

Every organism, human and non-human, responds to structural and functional characteristics of their environment. Adaptations result from exposure to physical and chemical factors in the environment, from interaction with other species and from the interaction of individuals within the same species. Evolutionary change through the mechanism of natural selection involves the replacement of individuals with one type of adaptation by those with another. This adaptation involves a slow adjustment to environmental change and is studied at both the individual level as well as at the population level.

Ecological anthropology concentrates on the mechanisms of human adaptability. It is unique in its way because it integrates the findings from ecology, physiology, social and cultural anthropology and geography around the set of problems set or posed by the human habitats. This problem oriented approach lends itself to the integration of data and at the same time orients the researcher toward answering the question, how do humans adapt?

The answer to this question is found through the study of human adaptability, which emphasizes the human response to environment. This adaptability is manifested in both socio-cultural and physiological terms. Socio-cultural responses such as clothing, shelter and various forms of social organization help to articulate the adjustment of our species to the environment. Physiological changes require changes in organic structure and function and therefore take longer to come into operation than socio-cultural responses. Both types of responses provide a more rapid mechanism for improving survival chances than the genetic change, which accumulates over several generations. Hence a need arises where so much varying facts which are both biological and socio-cultural, need to be integrated in order to understand the concept of human adaptability. This is exactly what ecological anthropology does. It integrates the facts regarding human physiology, geography, genetics, society and culture in order to throw invaluable light on the way in which human species try to maintain equilibrium with the environment by adapting to it. Ecological anthropology tries to explore the multilevel ways in which humans adjust to their surrounding by both biological and non-biological processes.

The interaction of the social and biological approaches to the study of adaptability was facilitated by the increasing acceptance of the ecosystem concept. This concept, derived from the study of biological ecology concept, views all organisms as part of ecological systems and subject to the same physical laws. Using this framework, one can refer to human beings as the third order consumers in the food chain or view the interaction between two human populations as mutualistic. The ecosystem concept makes it possible to apply a greater body of data to explanatory models of human behavior than is possible from strictly social or cultural approach. Ecological anthropology recognizes this fact.

METHODS OF ECOLOGICAL ANTHROPOLOGY

A major influence on ecological anthropology came from general or biological ecology. Ecological study in its broadest sense, applies to the dynamic relation between living and nonliving parts of an ecological system. The ecosystem concept provided a conceptual framework more satisfactory to some scientists than the behaviour/ social structure equation stressed by Steward. By studying human population as parts of ecosystems, attention can be paid to human adaptability - physiological, cultural and behavioural.

The research strategy of ecological anthropology is to study a wide range of human responses to environmental problems, to social constraints, and to past solutions of environmental problems. Little and Morren have succinctly expressed the strategy as "ecological anthropologists are concerned with those cultural and biological responses, factors, processes and cycles that affect or are directly connected with the survival, reproduction, development, longevity or spatial positions of people".

An ecosystem includes all the organisms in a given area, interacting with the physical environment, so that a flow of energy leads to a clearly defined tropic structure, biotic and material cycles. Ecosystems are

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said to be self-maintained and self-regulating. The concept of homeostasis, which in the past has been defined as the tendency for biological systems to resist change and to remain in a state of equilibrium, has led to an overemphasis on static considerations and to an evaluation of man's role as basically disruptive. Recently certain scholars have defined homeostasis as the maintenance of system properties.

The ecosystem quality leads naturally to use the systems analysis techniques. Systems analysis has become a useful approach for anthropologists since it begins with a holistic model of components and interrelations of an ecosystem, essentially a qualitative and descriptive process with which anthropologists feel at ease. Systems theory provides a broad framework for analyzing empirical reality and for cutting across disciplinary boundaries. Essentially, systems theory is a perspective that bears a great deal of similarity to anthropological holism; a system is an integral whole and no part can be understood apart from the entire system.

Clifford Geertz was perhaps the first anthropologist to argue for the ecosystem as a viable unit of analysis in cultural anthropology. In taking a whole systems approach, Geertz notes that historical and political factors must also be included, if one is to explain, say, agricultural development.

Roy Rappaport and Andrew Vayda have given the strongest impetus to an ecosystem approach in the field of cultural anthropology. In fact, they prefer the term ecological anthropology because they feel that the emphasis on culture suggested by the term cultural ecology obscures the applicability of principles from biological ecology to the study of human adaptation. Given that humans are but one species in nature, subject to the same laws as other species, use of the principles, methods and analytical tools of the ecological sciences would greatly add to our understanding of our own species. Vayda and Rappaport believe that anthropologists should not hesitate to adopt biological units as units of study since this allows a more comprehensive approach to ecological studies. Ecologists have shared various areas of interest with anthropology; ways of defining territorial rights, ways of establishing group identity and mechanisms for establishing buffer zones. All these can be viewed ecologically as regulating behaviour or serving a homeostasis function.

Bennett criticized the use of biological analogies, but concedes that Rappaport's study is important as a concrete demonstration of the fact that the behaviour of men toward each other, as well as toward nature, is part of ecosystems. Bennett finds the distinction between cultural ecology and the ecosystems approach artificial. The choice of one approach over the other depends on the size and complexity of the group under study. Among small tribes with primitive technologies, the ecosystem approach can be employed since much of the human environment interactions in such tribes are embedded in cultural traditions. On the other hand, in larger, complex and technologically advanced cultures, institutions and technology have created a distance between the population and its environment. Studies of modern societies must investigate these institutions and the processes of decisions that affect nature and humans. In these contexts, Bennett argues the ecosystem approach does not work well because it cannot research the dynamic processes of institutions and the conscious processes of choice among alternatives due to the complexity of the systems.

Homeostasis and dynamic equilibrium do not imply changelessness. On the contrary they require constant adjustment of system parts and even some change in structure. In other words, while systems have lower order mechanisms geared at the maintenance of stability, they also have higher level, less specialized mechanisms that can reorder the system to assure its survival.

So far the studies with ecological anthropological approach have concentrated on isolated populations of Alaskan Eskimos, South American Yanomamo Indians, New Guinea tribesmen, and Miskito Indians of Central America. The choice of such small isolated groups was made because it permitted easier monitoring of the interaction between the human population and their environment. Monitoring the complex relationships between such environmental stresses as disease, low energy and food availability, heat, cold and altitude in technologically complex societies would be far more difficult task. It is likely that once a clearer understanding of the human ecology of simpler cultures has been reached, ecological

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anthropologists will be able to more easily study human response to environmental degradation, urban pollution and other contemporary stresses.

The Geographers consistently show a greater sophistication in their analyses of the physical environment, while anthropologists deal with cultural components of the analyses with greater insight and facility. Although anthropologists have no difficulty dealing with the micro scale, since it has been their traditional unit of research, there is concern over the applicability of these micro studies to the large context of human behaviour and human adaptation.

It is certainly premature to expect the ecosystem perspective to resolve most of our questions about how humans adapt. Close cooperation will be required between biological and behavioural scientists to generate an integrated study of people in ecosystems. In the future, studies are likely to be most fruitful when they integrate the general systems approach with the study of how actors develop their own individual strategies. There is no reason why both perspectives cannot be used, and there is evidence that researchers have already begun to balance a concern for the individual with a concern for the population. One way to overcome the tendency towards static equilibrium models might be to study how populations adapt to certain kinds of stress. By studying the response of individuals to hazards, we can answer such questions as does stress lead to changes in the structuring of the population? Are cultural patterns changed? How do people perceive the severity of the stress to which they are responding? How does the human population adjust to termination of the stress? These questions are more likely to be productive in outlining systemic interrelations in populations experiencing changing situation is than in those with stable situations. This however presents even greater challenges to researchers. As a way of dealing with these challenges, Alland has proposed what he calls structural ecology, which integrates cognitive / structural and bio ecological aspects of human adaptation.

THE ETHNO-ECOLOGICAL APPROACH

A very different approach to the study of human environment relations grew out of developments in the field of sociolinguistics. This general approach has been termed ethnoscience and deals with the study of various cultural perceptions of the world and how people order those perceptions through their given language. Ethnoscience has given rise to subfields dealing with specific domains of culture, such as ethnobotany, ethnozoology, and ethnoecology. The aim of ethnoecological approach is to provide a better understanding of how people perceive their environment and how they organize these perceptions.

BIO-CULTURAL ADAPTATIONS – GENETIC AND NON GENETIC FACTORS

What is Ecology?

Ecology is the study of organisms in relation to the surroundings in which they live. These surroundings are called the environment of the organism. This environment is made up of many different components, including other living organisms and their effects, and purely physical features such as climate and soil type.

Ecologists, those who study ecology, are always aiming to understand how an organism fits into its environment. The environment is of supreme importance to an organism and its ability to exist in the environment where it lives will determine its success or failure as an individual.

Thus ecology deals with the problem of how growing and multiplying beings maintain themselves in a constantly changing but, ever restricted environment. It proceeds, in other words, with the conception of life as a continuous struggle for adjustment of organisms to environment, a struggle initiated and continued essentially by the differential modes of change of these two components of the life process. In the ecological view, life is not an individual but, an aggregate phenomenon. Hence the underlying assumption of ecology is that adjustment to environment is a mutual, in fact a communal function. The adjustment of a population to its physical world occurs not through the independent actions of many

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individuals but through the coordination and organization of individual actions to form a single functional unit.

Human Ecology

Human ecology, as plant and animal ecology, represents a special application of the general viewpoint to a particular class of living things. It involves both recognition of the fundamental unity of animate nature and awareness that there is differentiation within that unity. Man not only occupies a niche in nature's web of life, he also develops among his fellows an elaborate community of relations comparable in many important respects to the more inclusive biotic community. In at least one of its aspects the human community is an organization of organisms adjusted or in process of adjustment to a given unit of territory. Hence the rise of human ecology has meant a logical extension of the system of thought and the techniques of investigation developed in the study of the collective life of lower organisms to the study of man. Human ecology may be defined as the study of the form and the development of the community in human population.

Ecology as applied to man differs in important respects from its application to other forms of life or even to life as a whole. Man is an organism, to be sure, and as such he has much in common with other forms of organic life. But at the same time, he is capable of an extraordinary degree of extensive control over his surroundings, as manifested in the degree to which he modifies and reconstructs his environment through invention and the use of tools, and again in the complex cooperative arrangements entered into with his fellow men. Furthermore, man's great faculty for devising and accumulating methods of coping with life situations is evidence of dynamics in human behaviour that is without counterpart elsewhere in the animate world. It is this that constitutes man as an object of special inquiry and makes possible a human ecology as distinct from a general ecology.

It is necessary to keep the phenomenon of human culture in proper perspective. When man by nature of his culture-producing capacity, is regarded as an entirely unique type of organism the distinction has reached a point of overemphasis. Human behaviour, in all its complexity, is but a further manifestation of the tremendous potential for adjustment inherent in organic life. Thus, if we look upon culture as a totality of the habitual ways of acting that are general in a population and are transmitted from one generation to the next, there exists for human ecology no peculiar problems other than those involved in the fact of its complexity. The term simply denotes the prevailing techniques of adjustment by which a population maintains itself in its habitat. The elements of human culture are therefore identical in principle with the appetency of the bee for honey, the nest building activities of birds, and the hunting habits of carnivores.

ADAPTABILITY, ADJUSTMENT AND ACCLIMATISATION

The term "adaptation" is at the heart of ecological approach. Organisms, human and non-human, respond to structural and functional characteristics of their environment. Adaptations result from exposure to physical and chemical factors in the environment, from interaction with other species, and from the interaction of individuals within the same species. Evolutionary change through the mechanisms of natural selection involves the replacement of individuals with one type of adaptation by those with another. This type of evolutionary or genetic adaptation involves a slow adjustment to environmental change and is studied at level of population.

Individuals, however, can respond to changes in their environment by morphological and functional adjustments. Ricklefs differentiates between regulatory, acclimatory and developmental adjustments. All three types operate by a process known as negative feedback. This type of feedback seeks to maintain a stable relationship between the organism and the surroundings. To be effective, a response must be of the proper magnitude and occur at a time and rate that is appropriate in relation to the stimulus that elicited the adjustment.

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Regulatory Responses: These occur rapidly and reflect the physiological and behavioural flexibility of an organism. Virtually all behaviour is a form of regulatory response that either serves to maintain a stable relationship to the environment or permits adjustment to changes in that environment. Cultural strategies of clothing and shelter are among the most common regulatory mechanisms that enhance human chances to survive and live in relative comfort in a variety of environments.

Acclimatory Responses: These take longer to come into operation because they require a change in organismic structure. They occur when an external stimulus is present for a sufficient amount of time. They are usually reversible when the situation that produced the organismic change ends. For example, muscle enlargement as a result of frequent and demanding physical exercise is reversed when the individual begins to lead a more sedentary life.

Developmental Responses: These are not reversible and occur during the growth and development of an individual. They are adjustments of the organism to the environmental conditions prevalent during the developmental period. During this developmental period human organisms have the ability to mold themselves to prevalent environmental conditions - genetic plasticity. For example, a child growing at high altitudes will develop larger lungs and chest capacity to adjust to prevalent low oxygen conditions. After the growth period, a non native will be unable to achieve a larger chest cavity. Developmental adjustments are of limited value for short term environmental adjustments, but provide a more flexible adjustment to prevalent conditions than the genetic change. The development flexibility of the human population provides a more rapid mechanism for improving survival chances and enhancing reproduction than genetic change accumulated over several generations.

The ecosystem is a fundamental ecological unit that refers to associated species of living organisms in a nonliving physical environment and to the structural and functional relationships among them. In the study of human adaptability, the ecosystem is the total situation within which adaptability occurs. Because human populations have spread over the totality of the earth, this context of adaptability varies a great deal. A population in a specific ecosystem adjusts to environmental conditions in specific ways that represent both present and past conditions. In other words, a desert population that has existed in that environment for several millennia will differ significantly in its responses to desert conditions than a population that migrated there only in the last generation. A population that has existed longer in a particular environment is more likely than a recently settled population to have developed physiological and even genetic characteristics aimed at coping with environmental constraints, such as extreme heat. The more recent inhabitants will have physiological and cultural adjustments attuned to another environment. When they begin adjusting to the new environment, a process of change will be initiated that may take several generations, and the final result may or may not be much like the adjustments of the original inhabitants. This is particularly true when native populations are available from whom to borrow a variety of forms of adjustment. The newcomers may borrow some of the practices of the original inhabitants in order to achieve a satisfactory adjustment to their new habitat.

The above scenario suggests that human adaptability can proceed along any one of many paths. In the absence of borrowing and diffusion of ideas, the population may innovate and develop new forms of adjustment. If the new adjustment pattern conflicts with the previous practices and yet provides a workable solution that does not threaten the survival and well being of the individual or group, some form of compromise may emerge. If the exposure to stress is continual, physiological change of a permanent kind (development adjustment) may provide a more adequate adjustment than regulatory forms. The human body is able to adapt by genetic, physiological and behavioural and cultural means. The purpose of these various levels of adjustment is to enhance adaptability through a flexible hierarchy of response.

Human adaptability, therefore, refers to ecological success as measured by demographic, energetic or nutritional criteria. Demographic criteria often used are: (1) balance between natality and mortality, (2) morbidity or incidence of disease and (3) the population's rate of reproduction. Energetic criteria can be relative or absolute. Relative energetic efficiency is used as an indication of the adequacy of a technology.

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Efficient subsistent technologies have been noted to be sustainable at low levels of population density. In the past it has been more common to use total energy harnessed as indicative of success. Nutritional criteria provide a good index of adaptability since food consumption reflects knowledge of resources, capability to exploit them and capacity to achieve given level of work capacity.

However, all these criteria are only indices of adaptability and do not constitute any firm measure of fitness. Fitness refers to reproductive success. The more "adapted" a species is in its environment, the greater the opportunity for individuals of the population to survive and reproduce and then to occupy the territory. It is much easier to define energetic failure or inefficiency and nutritional inadequacy than to determine reproductive success. The problem of over population raises numerous questions about the adequacy of fitness as an index of human adaptability.

MAN'S PHYSIOLOGICAL RESPONSES TO ENVIRONMENTAL STRESSES

HOT DESERTS

Arid lands, or deserts, are characterized by low and random distribution of rainfall, high levels of solar radiation, high daytime temperatures, high levels of evapotranspiration, and a consequent scarcity of plant cover during most of the year.

Human populations appear to have neither genetic nor developmental adaptations for living in dry heat areas. They depend instead on acclimatory and behavioural adjustments to facilitate their occupation of these regions. Acclimatization to desert conditions actually occurs within a week or two. However, it is the problems of locating water, storing it, and minimizing its loss that is central to human adaptation to arid conditions.

Desert Ecosystems: Deserts are not homogeneous, but vary a great deal in characteristics. Generally speaking, they can be said to be areas of very low rainfall and high evaporation where vegetation is scanty. Not only is the precipitation low, it is infrequent and largely erratic as well. True or extreme deserts are said to be those with less than 100 mm of rainfall. They lie beyond the equatorial rainfall belt and in latitudes where drying trade winds blow throughout the year. Deserts may also appear in areas dominated by the subtropical high pressure belt on the leeward side of high mountains where moisture-laden air is blocked from crossing; in areas far removed from sources of oceanic moisture; and in locations on the leeward side of cold currents. Winds that pass over cold water reach land with lowered temperatures, but are warmed as they pass over the land and act as drying agents when they pass over moist land. Each type of desert is characterized by unique variations in daytime temperatures, level and seasonal distribution of rainfall and vegetation.

Deserts can be areas of surprising contrasts. In daytime, temperatures may rise to 43°C., while at night they may drop to an uncomfortable zero degree temperatures. Surface heating is rapid, but cooling is just as rapid when the sun sets because of high albedo (that is surface reflectivity) and clear skies. Weathering is slow and absence of vegetative cover makes every detail visible. Large and small closed basins dramatically cut by surface runoff and erosion is common.

Adjustment to Dry Heat: Before a human population can exploit the resources of a desert, its individual members must be able to cope with the hot and drying stresses of the ecosystem on their organisms. An individual in the desert is constantly threatened with dehydration and physiologically dangerous heat loads. Thermal sweating helps reduce heat gain through evaporation of moisture at the skin surface. When air and ground temperature are below 33.3 °C., human beings lose heat by radiation to cooler ground or by convection to cooler air. In the desert, however, daytime temperatures are often higher and sweating is the only effective mechanism. To become cool, we pay with loss of body water. The human body appears dry in the desert because evaporation is so rapid that as sweat nears the surface, it disappears (insensible evaporation). No changes in the human metabolism are needed to maintain this cooling system, but one must obtain an equal amount of liquid to replace what is lost.

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Water requirements cannot be represented by an absolute figure, but are a function of ambient temperature, air humidity, diet, and level of activity. A body at rest that is protected from the sun requires at least five ounces of water per hour to remain in body fluid equilibrium. Less water intake means the body would utilize stores of tissue liquid, eventually resulting in dehydration and thermostatic deregulation. Exposure to solar radiation adds to his equilibrium requirement an extra sixteen ounces per hour; and walking an additional twelve ounces per hour. Water requirements increase proportionately according to increase in activity level and temperatures. Reduction of activity and adoption of a relaxed posture that increases surface area help compensate for increases in heat load in the human body.

Physiological Adjustments: To date, physiological studies tend to agree that human populations acclimatize to desert conditions within a week or two. Very little difference in heat adaptation has been noted between desert natives and non natives. A diminished pulse rate, lower rectal temperatures, increases in the rate of sweating and ability to work in the heat are remarkably similar among human populations. Acclimatized individuals show a reduction in the salt concentration of sweat, which prevents deficiencies associated with cardiovascular inadequacy and violent muscular cramps. Urine volume is also reduced as a homeostatic compensation for increased dehydration. These few factors represent the bulk of physiological changes that occur. Adolph has raised questions about the limitations of the desert ecosystem on the amount of physical work that could be achieved, but concluded that these limitations are not severe as long as people have enough water. Further work is needed to establish whether alleged limitations on the amount of work that is possible to achieve might not be associated with work schedules that are inappropriate to desert living.

Body size, shape, and composition can also influence the heat exchange process. The "ideal" body type for desert conditions is tall, with long, lean extremities and low subcutaneous fat. Tallness maximizes the surface area to weight ratio for enhanced cooling, while lean body composition minimizes the presence of insulating fat that may limit heat flow from core to shell to cool the body. The Nilotic peoples of Sudan are often cited as an example of optimal human form for desert environments. However, one could also argue that such greater surface area would also maximize evaporation of sweat and expose those individuals to dehydration unless factors intervened to ameliorate that possibility. In fact, social and cultural patterns enter in to provide exactly such protection.

While there is some positive correlation between ideal body types and different body temperatures, factors such as migration, nutrition, and disease preclude a perfect association between body shape and environment. In desert areas today we find peoples of all shapes, sizes and degrees of leanness. The major adjustments of human mammals are not small size and nocturnal habits, nor complex physiological adjustments as among camels; they are cultural and behavioural in nature. The problem of how to cope with aridity may be overcome by activity patterns, clothing, shelter, diet and other similar adjustments.

Summary: To survive in any ecosystem, populations must take into account the existing constraints to life. In arid lands the challenge to human inhabitants lies in determining how much control to exercise over water resources and how to make optimal use of the water obtained. Most commonly, groups have scattered widely over these areas, thus keeping the densities per unit areas down. Kalahari hunter gatherers even today maintain effective population controls through dietary, social and cultural adjustments.

Cyclical changes in aridity necessitate flexible form of social organization, a nomadic way of life, or modifications in the technology of water use. We can expect that when severe drought strikes in frequent cycles, the population will adjust their lifestyles to the driest periods of the cycle. This has been clearly the case among the populations of the Kalahari. When the driest episodes are infrequent or unpredictable, however, populations will adjust to normal years and experience serious stress during dry periods.

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Individual survival must also be assured by behaviours that conserve water and at the same time permit constant cooling evaporation through effective sweating. Acclimatory adjustments through diminished pulse rates, reduced salt concentration in sweat, and intensified rates of insensible perspiration increase comfort. Most of the adjustments, however, are social and cultural. Clothing and shelter reduce the heat load of the body by providing insulation and promoting heat loss. Activity schedules are designed to avoid exposure to strong sun and drying winds and at the same time conserve energy and fluids. These adjustments to the requirements of the thermoregulatory system prevent dehydration, heat stroke and possible death.

COLD HIGH ALTITUDE CLIMATES

Mountain Ecosystems: Since the prehistoric times, human populations have occupied high altitude zones despite the numerous problems initially encountered by persons entering such areas. Anyone who has grown and developed at sea level will, when first exposed to high altitudes, experience symptomatic discomfort, reduced work capacity, accelerated breathing, higher haemoglobin levels, and higher arterial pressure. It is not surprising to find relatively small numbers of peoples living at high altitudes. Some ten to twenty five million people, i.e., less than one percent of the earth's population, currently make it their home. The majority of the areas occupied are within forty degrees of the equator where solar insolation is greater and, all things being equal, biotic productivity is higher. These areas include the South American Andes, the mountains of Ethiopia, the Caucasus of southern Russia, the Asian Himalayas, and the Rocky mountains of the United States.

High altitudes have been the subject of intensive research in human biology and ecology. In most other biomes, cultural practices can play a major buffering action, but in the high altitude biome, hypoxia (low oxygen pressure) provides one of the few situations in which behavioural regulatory adjustments play a secondary role to acclimatory and developmental adjustments. Human biologists saw in these regions an ideal setting to study potential genetic and physiological adaptations. These studies were stimulated by the important writings of Carlos Monge who postulated that Andean natives were biologically different from, and possessed adaptive characteristics not found among lowlanders. Since that time numerous workers have sought to test the validity of Monge's hypotheses.

Adatpation to Hypoxis

Hypoxia, or low oxygen pressure, is the most important stress with which populations living at high altitude must cope. Cultural practices are not able to increase the amount of oxygen available to human subjects, but some of its associated problems have been minimized through appropriate diets, work patterns, and cultural attitudes towards reproduction. It was thought at one time that populations native to a high altitude had genetic adaptations that permitted them to reproduce and live with no apparent difficulty where outsiders experienced serious malfunctioning. It now appears that there is no direct evidence of population-level adaptation to hypoxia in man, but only indirect evidence, based on inferences from individual and infra individual adaptations. Human adaptations to hypoxia appear to reflect the general genetic plasticity common to all of mankind, rather than distinctive genetic attributes.

The partial pressure of oxygen decreases with increasing altitude. Hypoxia results whenever either physiological, pathological, or environmental conditions cannot deliver an adequate supply of oxygen to the tissues. Since air is compressible, air at high altitudes is less concentrated and under less pressure. At forty five hundred meters the partial pressure of oxygen is decreased by as much as 40 percent, in comparison to pressure at sea level. This reduces the amount of oxygen finally available to the tissues.

Adaptations to high altitude hypoxia results in a series of modifications in body functioning that are oriented toward increasing the supply of oxygen. Because some of the adjustments to hypoxia are developmental, it is not surprising to find different adaptive mechanisms in sea level and highland populations. Sea level or lowland populations utilize the less efficient response of increased pulmonary ventilation, in contrast to the developmentally acquired advantage of increased lung volume, to achieve the same adaptive result.

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The adaptive responses to hypoxia are directed toward increasing both the availability of oxygen and the pressure of oxygen at the tissue live. Some of the physiological mechanisms that facilitate adaptation to hypoxia operate along the oxygen pressure gradient in the body and permit oxygen to reach tissues despite the low atmospheric pressure. Other mechanisms operate at the level of the tissues and include enlargement of the capillary bed.

At high altitudes oxygen pressure can be 40 percent or more below sea level pressure. Without adaptive mechanisms, the tissues of a man living at 4,000 and above meters would have an oxygen tension inadequate to diffuse oxygen and make it available for cell metabolism. At high altitude, where that pressure is already low to start with, the organism adjusts by shortening the distance the oxygen must travel. This is accomplished by an increase in the number of capillaries.

An increase in the pulmonary ventilation is a common response to high altitude. Natives have a moderately higher breathing rate, i.e., 20 to 40 percent, than sea level populations, and newcomers to high altitudes will quickly begin to hyperventilate (breathe rapidly). Highland natives have a larger lung volume and greater residual lung volume (that is, the volume of air that remains in the lungs after maximum expiration). This increased volume is a developmental adjustment accomplished by the proliferation of alveolar units and an increase in alveolar surface area during childhood.

Polycythemia refers to an increase in the number of red blood cells and in the amounts of haemoglobin in persons acclimatized to high altitude. It does not appear to be present in uterine life, but develops rapidly within a few days after birth. Prolonged exposure to hypoxia has a stimulating effect on bone marrow growth. As a result, more red blood cells are produced and there is an increase in the total volume of red blood cells in the body (i.e., the thickening of the blood). Associated with the increased production of cells, there is a decrease in blood plasma so that blood contains a larger volume of red blood cells and haemoglobin. This means that a greater quantity of oxygen can now be carried which helps reduce the oxygen gradient between arterial and venous blood and aids the buffer capacity of blood for carbon dioxide.

Work Capacity and Diet: Accounts of the great physical capacity of natives at high altitudes are somewhat exaggerated. Although there is little doubt that natives perform better than newcomers at high altitudes, carefully designed experiments have not shown any significant superiority in natives when they move to lowland areas. Good physical training and lifelong exposure to high altitude act to increase maximal oxygen consumption. Grover finds the Andean native physiologically similar to the athlete; both share a high aerobic or maximal work capacity. The native accomplishes this by exposure to atmospheric hypoxia and an active life, and the athlete by subjecting himself to strenuous exercise that subjects the muscles to tissue hypoxia.

Childhood development at high altitude, on the other hand, enhances adult work capacity. There might be a genetic factor involved to their lower body weight, their vigorous life in the pursuit of subsistence, and their characteristic high carbohydrate diet. It has been shown that a high carbohydrate diet enhances endurance of hard work through increase of muscle glycogen.

Body tissues require a constant dietary supply of carbohydrate to provide energy. Absorption of ingested carbohydrate in the intestine is dependent on the time the carbohydrate is in contact with the absorbing surface, and on the supply of enzymes for the oxidation of carbohydrate. Once oxidized, carbohydrate is absorbed into the bloodstream as glucose, galactose and fructose. The last two are later transformed to glucose in the liver. Glucose is stored as glycogen for storage and is reconverted to glucose when needed by body.

Low levels of glucose and problems of carbohydrate mal absorption are apparently widespread health problems in highland natives. It is not clear; however, to what extent the general reduction in food utilization is a result of hypoxia.

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Reproduction and Maturation: Hypoxia appears to affect reproduction. It was found that moderately high altitudes where most Andean populations live have only a mild effect on reproductive performance. A more dramatic effect on reproduction is reflected in lower birth weights and increased postnatal mortality. Although short term difficulties in spermatogenesis and menstruation are experienced upon moves to high altitudes, men and women native to the area appear normal in these respects.

There appears to be no hypoxia stress on the fetus during pregnancy, possibly as a result of an increase in the size of the placenta. There is also a decrease in the birth weight relative to the placenta, which facilitates delivery of oxygen to the fetus without unduly increasing the demands on the mother.

In addition to the birth weight being low, the growth rate and development of children at high altitude is slow. This may be related to the demands of chest and bone marrow development during growth. During childhood important changes occur in alveolar area and diffusion capacity that facilitate the diffusion of oxygen. Maturation is delayed at high altitude, and sexual dimorphism (the differentiation between males and females as manifested in body size and muscle size) appears to occur only after the sixteenth year.

Maladaptations: Not all individuals native to high altitude adapt to hypoxia, and some lose their adaptation. This is known as chronic mountain sickness and involves loss of normal stimulation of breathing. This leads to low oxygen pressure in the alveoli and in arterial blood. To compensate, the body experiences excessive Polycythemia (that is, the thickening of blood). The only cure involves bleeding the person every two or three weeks, but the best solution is to leave the high altitude zone. Since high altitude natives also sometimes engage in seasonal migration to lowlands, they may also experience pulmonary edema on returning to altitude (that is, build-up of fluid in the lungs which interferes with the transfer of oxygen from the air to the bloodstream). Following several weeks at sea level, even natives may experience this temporary illness upon return to high altitudes.

Cold Stress

Next to hypoxia, cold is the most significant stress felt by high altitude populations. However, it is more amenable to management than hypoxia by appropriate cultural practices.

Long term exposure to cold stress may lead to acclimatization. The responses of the habituated demonstrate an increased peripheral blood flow to the extremities that prevents cold injury, limits heat loss through the hand, and permits easier hand functioning in cold conditions.

Such a difference in response to cold appears to be developmental. Studies of foot exposure to cold air at 0°C in the Andean highlands showed that both native adults and children maintain warmer skin temperatures than whites, but the differences between the young and the adult studied suggest that this ability increases through the developmental period. If this is the case, the neonate may experience unusually severe cold stress. Researchers have found that brown adipose tissue (BAT) is present in human infants and plays important roles in maintaining high core temperatures and in promoting non-shivering thermogenesis. Normal fat as a percentage of the body weight is at its lowest at birth. Between birth and age one, however, it increases six fold, while the stores of brown adipose tissue are slowly depleted.

Summary: Human adaptation to high altitude zones involves a combination of behavioural and physiological adaptations that have made it possible for populations to survive in this vertically zonated environment. The major constraint is hypoxia and numerous developmental adaptations are particularly valuable. An enlarged capillary bed, Polycythemia, decreased alveolar arterial oxygen gradient, and other mechanisms are involved in facilitating survival in an oxygen scarce environment.

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9.8 EPIDEMIOLOGICAL ANTHROPOLOGY

CONCEPT OF EPIDEMIOLOGICAL ANTHORPOLOGY

Over the past two decades increasing interest has emerged in the contributions that social sciences like anthropology might make to epidemiological study of health and disease. There are several reasons for this increasing interest. Primary among these has been the rise of chronic, non-infectious diseases as important causes of morbidity and mortality among westernized societies during the 20th century. Generally speaking, the chronic, non-infectious diseases are influenced by a number of lifestyle variables, which are themselves strongly influenced by social and cultrual factors. The understanding of the effects of behavioral factors, thus require an understanding of the social and cultural forces on different conditions of health and disease. Equally, there is a growing awareness that considerations of human behavior and its social and cultural determinants are important for understanding the distribution and control of infectious diseases. Related to this expansion of the epidemiological interest in the behavioral realm has been the development of etiological models which focus on the psychological, biological and socio-cultural characteristics of hosts, rather than exclusive concern with exposure to a particular agent. Both epidemiology and anthropology have undergone parallel evolution. Epidemiologists have been forced to define and measure complex social and cultural processes hypothesized to have an impact on health and disease states. Anthropologists, on the other hand, have become increasingly interested in the potential contributions of their discipline to resolve the day to day problems of specific populations, and especially so within the realms of disease occurrence and heath care. These convergent interests have created the opportunity for mutually beneficial cross-discplinary study – Epidemiological Anthropology.

As epiodemiology has expanded its concerns to encompass the full range of human disease and disorder, researchers have been forced to grapple with complex assemblages of psychological, social, cultural, demographic and genetic factors in their quest to identify etiologic relationships and improve health services. The recognition that causal assemblages often include phenomena as acculturation, anomie, ethnicity, and poverty has led epidemiologists to look to anthropology in explaining the relationship of social and cultural processes to health. The interdisciplinary research in epidemiological anthropology has revealed that any human disease or disorder is the result of many factors within what may be described as a "causal web", a web of determinants. These webs, their extent varying between populations depending on their size and density, include...

- Exogenous Factors – Biotic and Non-Biotic
- Endogenous (Genetic) Factors
- Demographic Factors
- Behavior, as governed by social, cultural and psychological factors

It is the goal of epidemiological anthropology to identify and measure the relative importance of factors within the causal web of disease. Since almost all the diseases are caused, at least in part, by behavior of individuals, epidemiological anthropology must be a behavioral science. Epidemiological anthropology, as a specialization within the parent discipline of medical anthropology, grew essentially with these concerns.

HEALTH AND DISEASE

Health could be defined theoretically in terms of certain measured values; for example, a person having normal body temperature, pulse and breathing rates, blood pressure, height, weight, acuity of vision, sensitivity of hearing, and other normal measurable characteristics might be termed healthy. Biological criteria of normality are based on statistical concepts.

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Health might be defined better as the ability to function effectively in complete harmony with one's environment. Implied in such a definition is the capability of meeting—physically, emotionally, and mentally—the ordinary stresses of life. In this definition health is interpreted in terms of the individual's environment. Health involves more than physical fitness, since it also implies mental and emotional well-being.

The definitions of illness and disease are equally difficult problems. Despite the fact that these terms are often used interchangeably, illness is not to be equated with disease. A person may have a disease for many years without even being aware of its presence. Although diseased, this person is not ill. Similarly, a person with diabetes who has received adequate insulin treatment is not ill. An individual who has cancer is often totally unaware of having the disorder and is not ill until after many years of growth of the tumour, during which time it has caused no symptoms. The term illness implies discomfort or inability to function optimally. Hence it is a subjective state of lack of well-being produced by disease. Regrettably, many diseases escape detection and possible cure because they remain symptomless for long years before they produce discomfort or impair function.

Disease, which can be defined at the simplest level as any deviation from normal form and function, may either be associated with illness or be latent. In the latter circumstance, the disease will either become apparent at some later time or will render the individual more susceptible to illness. The person who fractures an ankle has an injury—a disease—producing immediate illness. Both form and function have been impaired. The illness occurred at the instant of the development of the injury or disease. The child who is infected with measles, on the other hand, does not become ill until approximately 10 days after exposure (the incubation period). During this incubation period the child is not ill but has a viral infectious disease that is incubating and will soon produce discomfort and illness.

Disease, defined as any deviation from normal form and function, may be trivial if the deviation is minimal. A minor skin infection might be considered trivial, for example. On the eyelid, however, such an infection could produce considerable discomfort or illness. Any departure from the state of health, then, is a disease, whether health is measured in the theoretical terms of normal measured values or in the more pragmatic terms of ability to function effectively in harmony with one's environment.

Disease may be acute, chronic, malignant, or benign. Of these terms, chronic and acute have to do with the duration of a disease, malignant and benign with its potentiality for causing death.

An acute disease process usually begins abruptly and is over soon. Acute appendicitis, for example, is characterized by the sudden onset of nausea, vomiting, and pain usually localized in the lower right side of the abdomen. It usually requires immediate surgical treatment. The term chronic refers to a process that often begins very gradually and then persists over a long period. For example, ulcerative colitis—an inflammatory condition of unknown cause that is limited to the colon—is a chronic disease. Its peak incidence is early in the second decade of life. The disease is characterized by relapsing attacks of bloody diarrhea that persist for weeks to months. These attacks alternate with asymptomatic periods that can last from weeks to years.

The terms benign and malignant, most often used to describe tumours, can be used in a more general sense. Benign diseases are generally without complications, and a good prognosis (outcome) is usual. A wart on the skin is a benign tumour caused by a virus; it produces no illness and usually disappears spontaneously if given enough time (often many years). Malignancy implies a process that, if left alone, will result in fatal illness. Cancer is the general term for all malignant tumours.

Diseases usually are indicated by signs and symptoms. A sign is defined as an objective manifestation of disease that can be determined by a physician; a symptom is subjective evidence of disease reported by the patient. Each disease entity has a constellation of signs and symptoms more or less uniquely its own; individual signs such as fever, however, may be found in a great number of diseases.

Classifications of diseases

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Classifications of diseases become extremely important in the compilation of statistics on causes of illness (morbidity) and causes of death (mortality). It is obviously important to know what kinds of illness and disease are prevalent in an area and how these prevalence rates vary with time. Classifying diseases made it apparent, for example, that the frequency of lung cancer was entering a period of alarming increase in the mid-20th century. Once a rare form of cancer, it had become the single most important form of cancer in males. With this knowledge a search was instituted for possible causes of this increased prevalence. It was concluded that the occurrence of lung cancer was closely associated with cigarette smoking. Classification of disease had helped to ferret out an important, frequently causal, relationship.

The most widely used classifications of disease are (1) topographic, by bodily region or system, (2) anatomic, by organ or tissue, (3) physiological, by function or effect, (4) pathological, by the nature of the disease process, (5) etiologic (causal), (6) juristic, by speed of advent of death, (7) epidemiological, and (8) statistical. Any single disease may fall within several of these classifications.

In the **topographic classification**, diseases are subdivided into such categories as gastrointestinal disease, vascular disease, abdominal disease, and chest disease. Various specializations within medicine follow such topographic or systemic divisions, so that there are physicians who are essentially vascular surgeons, for example, or clinicians who are specialized in gastrointestinal disease. Similarly, some physicians have become specialized in chest disease and concentrate principally on diseases of the heart and lungs.

In the **anatomic classification**, disease is categorized by the specific organ or tissue affected; hence, heart disease, liver disease, and lung disease. Medical specialties such as cardiology are restricted to diseases of a single organ, in this case the heart. Such a classification has its greatest use in identifying the various kinds of disease that affect a particular organ.

The **physiological classification** of disease is based on the underlying functional derangement produced by a specific disorder. Included in this classification are such designations as respiratory and metabolic disease. Respiratory diseases are those that interfere with the intake and expulsion of air and the exchange of oxygen for carbon dioxide in the lungs. Metabolic diseases are those in which disturbances of the body's chemical processes are a basic feature. Diabetes and gout are examples.

The **pathological classification** of disease considers the nature of the disease process. Neoplastic and inflammatory disease are examples. Neoplastic disease includes the whole range of tumours, particularly cancers, and their effect on human beings.

The **etiologic classification** of disease is based on the cause, when known. This classification is particularly important and useful in the consideration of biotic disease. On this basis disease might be classified as staphylococcal or fungal, to cite only a few instances. It is important to know, for example, what kinds of disease staphylococci produce in human beings. It is well known that they cause skin infections and pneumonia, but it is also important to note how often they cause meningitis, abscesses in the liver, and kidney infections. The sexually transmitted diseases syphilis and gonorrhea are further examples of diseases classified by etiology.

The **epidemiological classification** of disease deals with the incidence, distribution, and control of disorders in a population. Epidemiology is one of the important sciences in the study of nutritional and biotic diseases around the world. The United Nations supports, in part, the World Health Organization, whose chief function is the worldwide investigation of the distribution of disease. In the course of this investigation, many observations have been made that help to explain the cause and provide approaches to the control of many diseases.

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INFECTIOUS AND NON INFECTIOUS DISEASES

DISEASES OF GENETIC ORIGIN

Certain human diseases result from mutations in the genetic complement (genome) contained in the deoxyribonucleic acid (DNA) of chromosomes. A gene is a discrete linear sequence of nucleotide bases (molecular units) of the DNA that codes for, or directs, the synthesis of a protein; and there may be as many as 100,000 genes in the human genome. Proteins, many of which are enzymes, carry out all cellular functions. Any alteration of the DNA may result in the defective synthesis and subsequent malfunctioning of one or more proteins. If the mutated protein is a key enzyme in normal metabolism, the error may have serious or fatal consequences. More than 5,000 distinct diseases have been ascribed to mutations that result in deficiencies of critical enzymes.

Mutations that occur in the DNA of somatic (body) cells cannot be inherited, but they can cause congenital malformations and cancers; however, mutations that occur in germ cells—i.e., the gametes, ova and sperm—are transmitted to offspring and are responsible for inherited diseases. Each gamete contributes one set of chromosomes and therefore one copy (allele) of each gene to the resultant offspring. If a gene bearing a mutation is passed on, it may cause a genetic disorder.

Genetic diseases caused by a mutation in one gene are inherited in either dominant or recessive fashion. In dominantly inherited conditions, only one mutant allele, which codes for a defective protein or does not produce a protein at all, is necessary for the disorder to occur. In recessively inherited disorders, two copies of a mutant gene are necessary for the disorder to manifest; if only one copy is inherited, the offspring is not affected, but the trait may continue to be passed on to future offspring. In addition to dominant or recessive transmission, genetic disorders may be inherited in an autosomal or X-linked manner. Autosomal genes are those not located on the sex chromosomes, X and Y; X-linked genes are those located on the X chromosomes that have no complementary genes on the Y chromosome. Females have two copies of the X chromosome, but males have an X and a Y chromosome. Because males have only one copy of the X chromosome, any mutation occurring in a gene on this chromosome will be expressed in male offspring regardless of whether its behaviour is recessive or dominant in females. Autosomal dominant disorders include Huntington's chorea, a degenerative disease of the nervous system that usually does not develop until the carrier is between 30 and 40 years of age. The delayed onset of Huntington's chorea allows this lethal gene to be passed on to offspring. Autosomal recessive diseases are more common and include **cystic fibrosis, Tay-Sachs disease, and sickle cell anemia**. X-linked dominant disorders are rare, but X-linked recessive diseases are relatively common and include **Duchenne's muscular dystrophy and hemophilia A**.

Most genetic disorders can be detected at birth because the child is born with characteristic defects. Thus these abnormalities are congenital (existing at birth) genetic disorders. A few genetic defects, such as **Huntington's chorea** mentioned above, do not become manifest until later in life. Hence it may be said that most but not all genetic diseases are congenital.

Conversely, some congenital diseases are not genetic in origin; instead they may arise from some direct injury to the developing fetus. If a woman contracts the viral disease ***German measles (rubella)*** during pregnancy, the virus may infect the fetus and alter its normal development, leading to some malformations, principally of the heart. These malformations constitute a congenital disease that is not genetic.

DISEASES OF IMMUNE ORIGIN

The immune system protects against infectious disease, but it may also at times cause disease. Disorders of the immune system fall into two broad categories: (1) those that arise when some aspect of the host's immune mechanism fails to prevent infection (immune deficiencies) and (2) those that occur when the immune response is directed at an inappropriate antigen, such as a noninfectious agent in an allergic

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reaction, the body's own antigens in an autoimmune response, or the cells of a transplanted organ in graft rejection.

The immune system may fail to function for many reasons. Many immunodeficiency disorders are caused by a genetic defect in some component of the system and thus usually manifest early in life. Some deficiencies, however, are acquired through the action of infectious agents such as viruses, through the action of immunosuppressive agents used to treat various medical conditions, and through the effects of certain disease processes such as cancer. Both inherited and acquired immune deficiencies suppress one or many aspects of the immune response, rendering the affected individual unable to resist infection unless treated by administration of immunoglobulins or by bone marrow transplant.

Inherited immune disorders undermine the immune response in a variety of ways: B lymphocytes may be unable to produce antibodies, phagocytes may be unable to digest microbes, or specific complement components may not be produced.

Acquired immune deficiency syndrome (AIDS) is caused by infection with the human immunodeficiency virus (HIV), which destroys a certain type of T lymphocyte, the helper T cell. An infected individual is susceptible to a variety of infectious organisms, including those called opportunistic pathogens, which may live benignly in the human body and cause disease only when the immune system is suppressed. Certain diseases such as Kaposi's sarcoma and Pneumocystis carinii pneumonia, which until recently were rarely encountered by clinicians, have become prevalent in the AIDS population and are often the cause of mortality.

Allergies

The immune system may react to any foreign substance, and consequently it can respond to innocuous materials in the same way that it responds to infectious agents. If the foreign material poses no threat to the individual, an immune response is unnecessary, but it nevertheless may ensue. This misplaced response is called an **allergy**, or hypersensitivity, and the foreign material is referred to as an **allergen**. Common allergens include pollen, dust, bee venom, and various foods such as shellfish. What causes one person and not another to develop an allergy is not completely understood.

An allergic response occurs in the following manner. On first exposure to the allergen, the person becomes sensitized to it—that is, develops antibodies and specific T cells to the allergen. An allergic reaction does not usually accompany this initial event. When reexposure occurs, however, symptoms of the allergic response appear. These symptoms range from the mild response of sneezing and a runny nose to the sometimes life-threatening reaction of **anaphylaxis**, or anaphylactic shock, symptoms of which include vascular collapse and potentially fatal respiratory distress.

Autoimmune Disorder

Immune responses can be mounted against proteins that belong to the host, giving rise to autoimmune diseases. Although the immune system naturally generates antibodies to its own cells, mechanisms exist to keep this activity in check. Two mechanisms that prevent the immune system from mounting an attack against the host's own tissues have been identified. The first involves the elimination of self-reactive lymphocytes during their development and maturation in the thymus, a lymphoid organ in the chest. Self-reactive lymphocytes present in these cell populations are destroyed when they encounter the self-antigen to which they react. Because this protective selection process is not highly efficient, some self-reactive lymphocytes survive, exit the thymus, and enter the blood and tissues. Outside the thymus a second line of defence against immune self-destruction is afforded in which self-reactive lymphocytes lose their ability to react to self-antigens when they are encountered in blood and tissues. This state is referred to as immunologic ignorance. Autoimmune diseases arise when this mechanism fails and self-reactive lymphocytes are activated by self-antigens in the host's own tissues, often with devastating effects. Systemic lupus **erythematosus, thyroiditis, insulin-dependent diabetes mellitus,** and **rheumatoid arthritis** are examples of this type of disorder.

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Graft Rejection

Transplantation of organs and cells from one individual to another has become an important medical treatment. As are other forms of therapy, it is accompanied by certain risks. Each individual's cells have a spectrum of genetically determined cell surface protein antigens, the major histocompatibility complex (MHC) antigens, or human leukocyte antigens as they are referred to in humans. MHC antigens determine a person's tissue type just as red blood cell antigens determine blood type. Aside from siblings, especially identical twins—having the same form of each gene is extremely small. Even parents will have different tissue antigens from their children.

These differences in tissue antigens pose an obstacle to transplantation because it is highly likely that foreign donor tissue will introduce antigens in the recipient that will trigger an immune response leading to tissue death and rejection. However, by careful matching of the MHC type of donor and recipient, rejection can be diminished or avoided. Because perfect matching is possible only between identical twins or very close relatives, many transplants occur between less closely matched tissue types, and success is achieved with the administration of powerful immunosuppressive drugs.

DISEASES OF ABNORMAL CELL GROWTH

The growth of cells in the body is a closely controlled function, which, together with limited and regulated expression of various genes, gives rise to the many different tissues that constitute the whole organism. For the most part, control of cell growth persists throughout life except for episodic instances such as healing of an injured tissue. In this situation the growth of a localized group of cells is accelerated to reconstitute the tissue to its previous state of normal structure and function, following which tightly regulated growth resumes. Such areas of increased cell growth are referred to as **hyperplasias**; they consist of expanded numbers of normal-appearing cells and, depending on the duration of growth, can result in an enlargement of tissues and organs. In general, hyperplasias arise to meet special needs of the body and subside once these needs are met. Hyperplasias are the result of the sustained impact over time of stimulatory influences together with a loss of growth-inhibitory factors that are normally found within or around cells. As long as the loss of inhibition of cell growth is temporary, the capacity for enhanced cell proliferation when necessary has obvious advantages. However, if cells permanently lose their ability to respond to growth-inhibitory factors, their growth becomes irrepressible, and cancer may result.

Malignant and Benign Tumours

Diseases arising from uncontrolled cell growth and behaviour collectively constitute the second most common cause of human death (the most common cause being heart disease). The significance of this incidence is placed in proper perspective by a consideration of the following facts. While cancer arises at all stages of life, its incidence (number of cases) increases with age, reaching a peak between 55 and 74 years. This fact, together with the increasing longevity of the general population and improved diagnostic modalities that enable clinicians to detect cancers with greater frequency, tempers the notion that the incidence of cancer is increasing.

In addition to cancers—malignant tumours that may eventually kill the host—there are benign tumours that rarely produce serious disease. The two types of tumours are collectively referred to as neoplasms (new growths), and their study is known as **oncology**. Tumours are referred to as malignant or benign based on the structural and functional properties of their component cells and their biological behaviour. The cells and tissues of malignant tumours differ from the tissues from which they arise. They exhibit more rapid growth and altered structure and function, including stimulation of new blood vessel growth (angiogenesis) and a capacity to invade adjacent normal tissues, enter the blood vascular system, and spread (metastasize) to distant sites. The properties of malignant tumour cells serve to enhance and support their proliferation and extension throughout the body tissues and organs, eventually leading to death of the host. In contrast, the cells and tissues of benign tumours tend to grow more slowly and in general closely resemble their normal tissues of origin. When the structure and function of benign tumour cells are morphologically and functionally indistinguishable from those of normal cells, their growth as a

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tumour mass is the sole feature indicative of their neoplastic nature. It is hoped that a greater understanding of malignant cell growth and behaviour will lead to the development of novel cancer therapies based on tumour cell biology that will complement or replace the current treatments of surgical extirpation (complete excision), chemotherapy, and radiation.

Epidemiology of Cancer

Epidemiological studies of the worldwide incidence of cancers have identified striking differences among countries and population groups. For example, the incidence of and death rates for skin cancer are much higher in Australia and New Zealand than in the Scandinavian countries—presumably because of the marked differences between these two regions in total annual hours of exposure to sunlight. The importance of environmental influences is highlighted by comparing the incidence of and death rates for cancers among populations in different geographic regions. For example, prostate and colon cancer rates in Japanese persons living in Japan differ from the rates in Japanese persons who have emigrated to the United States, the rates of their offspring born in California, and the rates of long-term white residents of that state. These rates are much lower among Japanese living in Japan than they are in white Californians. However, the rates for each type of tumour among first-generation Japanese immigrants are intermediate between the rates in Japan and those in California, suggesting that environmental and cultural factors may play a more important role than genetic ones.

Genetics of Cancer

The irreversibility of the structural and behavioral changes of cancer cells has long been recognized and has favoured the postulate that they are probably due to permanent genetic alterations. This postulate remained speculative until the discovery in 1979 that **oncogenes** (cancer-causing genes) are derived from proto-oncogenes (normal growth-regulatory cellular genes). When proto-oncogenes become mutated or deregulated, they are converted to oncogenes, which are capable of causing the malignant transformation of cells, including those of humans. Cellular proto-oncogenes code for proteins involved in cell regulation. Thus, changes in the structure of proto-oncogenes and their conversion to oncogenes results in the synthesis of abnormal proteins that are incapable of carrying out their usual growth-regulatory functions. In identifying the genes involved in the development of cancer, researchers discovered a group of cellular genes—tumour-suppressor, or suppressor, genes—whose protein products normally negatively regulate cell growth by suppressing cell proliferation, thus counterbalancing the growth-stimulatory effects of proteins synthesized by proto-oncogenes. Genetic analyses of various animal and human cancers have demonstrated that, in the majority, alterations of oncogenes and suppressor genes were often simultaneously present. These analyses suggest that multiple genetic alterations involving growth-stimulatory and growth-inhibitory genes are required for the induction of malignancy. Such discoveries have ushered in a new era in cancer biology and may well lead to the eventual control, cure, and prevention of malignant diseases.

Heredity and Environment

The many causes of cancer include intrinsic factors, such as heredity, and extrinsic factors, such as environment and lifestyle. Hereditary causes of cancer are less common and are due to the inheritance of a single mutant gene that greatly increases the risk of developing a malignant tumour. Such cancers include (1) a childhood tumour of the eye, **retinoblastoma**, and a bone tumour, **osteosarcoma**, both of which involve the loss of a tumour suppressor gene, and (2) familial **adenomatous polyposis**, in which all patients develop colon cancer by age 50. The most common types of cancer that occur sporadically, such as cancers of the breast, ovary, colon, and pancreas, also have been documented to occur in familial forms. The children in such families appear to have a two to threefold increased risk of developing a particular tumour, but the transmission pattern is unclear. A still rarer hereditary cause of cancer is an inherited deficiency in the ability to repair DNA. Patients with this defect (known as **xeroderma pigmentosum**) are particularly sensitive to sunlight and develop skin cancer during early adolescence because of unrepaired mutations induced by ultraviolet (UV) light.

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Although the environment contains many agents that can cause cancer in humans, the extent to which they contribute to the human disease is often difficult to assess. For example, the link between tobacco smoking and lung cancer is clear; however, little is known about the cause of cancer of the prostate, the most common form of cancer in males, despite the fact that many factors—including age, race, male hormone, increased consumption of dietary fat, and a genetic basis—have been implicated.

Three categories of carcinogens (chemical or physical agents that mutate DNA) that induce cancer in experimental animals and humans have been identified in the environment: (1) chemicals, (2) radiant energy, and (3) oncogenic viruses.

DISEASES OF ENDOCRINE ORIGIN

Endocrine Glands are a group of ductless glands that regulate body processes by secreting chemical substances called hormones. Hormones act on nearby tissues or are carried in the bloodstream to act on specific target organs and distant tissues. Diseases of the endocrine system can result from the oversecretion or undersecretion of hormones or from the inability of target organs or tissues to respond to hormones effectively.

It is important to distinguish between an **endocrine gland**, which discharges hormones into the bloodstream, and an **exocrine gland**, which secretes substances through a duct opening in a gland onto an external or internal body surface. Salivary glands and sweat glands are examples of exocrine glands. Both saliva, secreted by the salivary glands, and sweat, secreted by the sweat glands, act on local tissues near the duct openings. In contrast, the hormones secreted by endocrine glands are carried by the circulation to exert their actions on tissues remote from the site of their secretion.

The endocrine system, in association with the nervous system and the immune system, regulates the body's internal activities and the body's interactions with the external environment to preserve the internal environment. This control system permits the prime functions of living organisms—growth, development, and reproduction—to proceed in an orderly, stable fashion; it is exquisitely self-regulating, so that any disruption of the normal internal environment by internal or external events is resisted by powerful countermeasures. When this resistance is overcome, illness ensues.

Other body tissues may also function as endocrine organs. Examples include the lungs, the heart, the skeletal muscles, the kidneys, the lining of the gastrointestinal tract, and bundles of nerve cells called nuclei. While all nerve cells are capable of secreting neurotransmitters into the synapses (small gaps) between adjacent nerves, nerve cells that regulate certain endocrine functions—for example, the nerve cells of the posterior pituitary gland secrete neurohormones directly into the bloodstream.

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Glands and hormones of the human endocrine system		
gland or tissue	principal hormone	function
testis	testosterone	stimulates development of male sex organs and secondary sex characteristics, including facial hair growth and increased muscle mass
ovary	estrogens (estradiol, estrone, estriol)	stimulate development of female sex organs and secondary sex characteristics, maturation of ovarian follicles, formation and maintenance of bone tissue, and contraction of the uterine muscles
	inhibin (folliculostin)	inhibits secretion of follicle-stimulating hormone from the pituitary gland
	progesterone	stimulates secretion of substances from the lining of the uterus (endometrium) in preparation for egg implantation in the uterine wall
	relaxin	induces relaxation of pubic ligaments during childbirth to facilitate infant delivery
thyroid gland	thyroxine	stimulates cellular metabolism, lipid production, carbohydrate utilization, and central and autonomic nervous system activation
	triiodothyronine	stimulates cellular metabolism, lipid production, carbohydrate utilization, and central and autonomic nervous system activation
adrenal gland, medulla	epinephrine (adrenaline)	stimulates "fight or flight" response, increases heart rate, dilates blood vessels in skeletal muscles and liver, increases oxygen delivery to muscle and brain tissues, increases blood glucose concentrations, and suppresses digestion
	norepinephrine (noradrenaline)	stimulates "fight or flight" response, increases heart rate, constricts blood vessels, increases blood glucose concentrations, and suppresses digestion
adrenal gland, cortex	cortisol	activates physiological stress responses to maintain blood glucose concentrations, augments constriction of blood vessels to maintain blood pressure, and stimulates anti-inflammatory pathways
	aldosterone	regulates balance of salt and water in the body
	androgens	contribute to growth and development of the male reproductive system and serve as precursors to testosterone and estrogen
pituitary gland, anterior lobe	corticotropin (adrenocorticotropin, ACTH)	stimulates growth and secretion of cells of the adrenal cortex; increases skin pigmentation
	growth hormone (GH; somatotropin)	stimulates growth of essentially all tissues in the body
	thyrotropin (thyroid-stimulating hormone)	stimulates secretion of thyroid hormone and growth of thyroid cells
	follicle-stimulating hormone (FSH)	stimulates maturation of egg follicles in females and development of spermatozoa in males
	luteinizing hormone (LH; interstitial cell stimulating hormone, ICSH)	stimulates rupture of mature egg follicles and production of progesterone and androgens in females and secretion of androgens in males
	prolactin (PRL; luteotropic hormone, LTH; lactogenic hormone; mammotropin)	stimulates and maintains lactation in breast-feeding mothers

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pituitary gland, posterior lobe	oxytocin	stimulates milk ejection during breast-feeding and uterine muscle contraction during childbirth
	vasopressin (antidiuretic hormone, ADH)	regulates fluid volume by increasing or decreasing fluid excretion in response to changes in blood pressure
pituitary gland, intermediate lobe	melanocyte-stimulating hormones (MSH)	stimulate melanin synthesis in skin cells to increase skin pigmentation; may also suppress appetite
hypothalamus	corticotropin-releasing hormone (CRH)	stimulates synthesis and secretion of corticotropin from the anterior pituitary gland
	growth hormone-releasing hormone (GHRH)	stimulates synthesis and secretion of growth hormone from the anterior pituitary gland
	thyrotropin-releasing hormone (TRH)	stimulates and regulates secretion of thyrotropin from the anterior pituitary gland and may modulate neuronal activity in the brain and spinal cord
	gonadotropin-releasing hormone (GnRH)	stimulates synthesis and secretion of follicle-stimulating hormone and luteinizing hormone from the anterior pituitary gland
	prolactin-inhibiting factor (PIF; dopamine)	inhibits secretion of prolactin from the anterior pituitary gland
	somatostatin	inhibits secretion of growth hormone from the anterior pituitary gland, inhibits secretion of insulin and glucagon in the pancreas, and inhibits secretion of gastrointestinal hormones and secretion of acid in the stomach
	gastrointestinal neuropeptides	hormones secreted from the stomach and pancreas that stimulate hypothalamic secretion of neuropeptides, such as neuropeptide Y, gastrin-releasing peptide, and somatostatin, that regulate appetite, fat storage, and metabolism
pancreatic islets of Langerhans	glucagon	maintains blood glucose concentrations by stimulating release of glucose from the liver and production of glucose from amino acids and glycerol
	insulin	stimulates glucose uptake and storage in adipose, muscle, and liver tissues
	somatostatin	inhibits glucagon and insulin secretion from the pancreas and inhibits secretion of gastrointestinal hormones and secretion of acid in the stomach
	pancreatic polypeptide	inhibits contraction of the gallbladder and secretion of exocrine substances from the pancreas
parathyroid gland	parathyroid hormone (parathormone)	increases serum calcium concentrations by stimulating release of calcium from bone tissue, reabsorption of calcium in the kidneys, and production of vitamin D in the kidneys; inhibits reabsorption of phosphate in the kidneys
	calcitonin	decreases serum calcium concentrations by promoting uptake of calcium into bone tissue and excretion of calcium in the urine
skin, liver, kidneys	calciferols (vitamin D)	maintain serum calcium concentrations by increasing absorption of calcium and phosphate in the intestines and reabsorption of calcium and phosphate in the kidneys; mobilizes calcium from bone in response to parathyroid hormone activity
stomach	gastrin	stimulates secretion of acid and pepsin in the stomach and contraction of the pyloric region of the stomach near the small intestine to increase motility during digestion

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duodenum	cholecystokinin (CCK; pancreozymin)	stimulates release of bile from the gallbladder into the intestine and stimulates secretion of pancreatic juices into the intestine; may induce satiety
	secretin	stimulates secretion of water and bicarbonate from the pancreas into the duodenum; inhibits secretion of gastrin in the stomach, delaying gastric emptying
	gastric-inhibitory polypeptide (GIP)	inhibits secretion of acid into the stomach; stimulates secretion of insulin from the pancreas
	vasoactive intestinal peptide (VIP)	stimulates dilation of blood vessels and secretion of water and electrolytes from the intestine; modulates immune functions
pineal gland	melatonin	regulates circadian rhythm (primarily in response to light and dark cycles) and release of gonadotropin-releasing hormone from the hypothalamus and gonadotropins from the pituitary gland
kidneys	renin	regulates blood pressure and blood flow by catalyzing conversion of angiotensinogen to angiotensin I in the kidneys
multiple tissues	insulin-like growth factors (somatomedins)	stimulate growth by mediating secretion of growth hormone from the pituitary gland
	prostaglandins	regulate many physiological processes, including dilation and constriction of blood vessels, aggregation of platelets, and inflammation

DISEASES OF NEUROPSYCHIATRIC ORIGIN

Diseases of neuropsychiatric origin afflict large segments of the population. For example, a total of about 2.8 million persons in the United States alone suffer from three major psychiatric diseases—schizophrenia, major depression, and mania—and three major neurological disorders—Alzheimer's disease, Huntington's chorea, and Parkinson's disease.

The key function of the nervous system is to collect information about the body and its external environment, process the information, and coordinate the body's responses to that information. This complex function depends on each nerve cell (neuron) receiving signals from other neurons and transmitting this input to still other neurons. This critical input and output of communication (signaling) between neurons is mediated by chemical transmitter molecules (neurotransmitters). **Neurotransmitters** are synthesized by nerve cells and released from one cell to another across a narrow gap between the two neurons known as the synapse. Eight different major neurotransmitters and a large number of neuropeptide molecules (which serve to modulate the effects of neurotransmitters) have been identified. Different types of nerve cells respond to different neurotransmitters and neuropeptides. Chemical signaling between nerve cells is rapid and precise and can occur over long distances. The precision is due to receptor molecules, which are activated following their recognition and binding of specific neurotransmitters. In some types of nerves the synapses do not possess receptors, in which case interneuronal communication is achieved by electrical transmission. In many neuropsychiatric diseases alterations in the levels of transmitter substances appear to play a major role in pathogenesis.

Psychiatric Diseases

Mental illnesses affect the very fabric of human nature, robbing it of its various facets of personality, purposeful behaviour, abstract thinking, creativity, emotion, and mood. Those suffering from mental disorders exhibit a spectrum of symptoms depending on the severity of their disease. These diseases include **obsessive-compulsive personality disorder, dementia, schizophrenia, major depression, and manic disorders.**

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Schizophrenia in its severe form is a catastrophic mental illness that begins in adolescence or early adult life. It is relatively common, occurring in about 1 percent of the general population worldwide. Because the incidence of schizophrenia among parents, children, and siblings of patients with the disease is increased to 15 percent, it is believed that heredity plays an important role in the genesis of the disease. However, other studies suggest that nongenetic factors are also influential. The biochemical basis of the disease may be an excess of the neurotransmitter substance dopamine, as high levels of dopamine and its metabolites, as well as increased dopamine receptors, are found in the brains of persons with schizophrenia. Further evidence for this hypothesis is that the drugs most effective in treating the disease are those that have a high capacity to block dopamine receptors.

Pathological disturbances of mood, ranging from severe depression to manic behaviour, are common forms of mental illnesses. **Severe depression** is characterized by despondency, diminished interest in most or all activities, weight fluctuation not due to dieting, disruption in sleep patterns, psychomotor agitation or retardation, feelings of worthlessness, excessive quiet, and recurrent thoughts of death or suicide. **Manic behaviour** involves a period in which an expansive, elevated, or irritable mood persists abnormally. During this episode symptoms such as increased talkativeness, distractibility, decreased need for sleep, inflated self-esteem, and excessive involvement in pleasurable yet risky activities may be present. **Major depression** is associated with decreased brain levels of the neurotransmitters norepinephrine and serotonin, and the most effective therapy consists of drugs that inhibit the breakdown of these compounds. The neurochemical alterations in mania are less clearly understood, but it is well established that drugs effective in the treatment of mania are those that antagonize dopamine and serotonin. Although mood disorders have a familial background, the evidence for a genetic component is not convincing.

Neurological Diseases

The three neurological diseases considered in this section—Alzheimer's disease, Huntington's chorea, and Parkinson's disease—are age-related, and to varying degrees they manifest as deterioration of mental function that involves the loss of memory and of acquired intellectual skills. This deterioration is referred to as **dementia**. Because dementia can result from many causes, other features of each disease must be present before a definitive diagnosis can be made.

Alzheimer's disease is the most common form of dementia, being responsible for about two-thirds of the cases of dementia in patients over 60 years of age. Women are affected twice as often as men. More rarely there are familial forms of the disease that have an early onset affecting individuals in the fourth and fifth decades of life. Early manifestations include memory loss, temporary confusion, restlessness, poor judgment, and lethargy. A failure to retain new information and a deterioration of social relationships often ensue. In some patients paranoia and delusions, which worsen during the night, are the first symptoms of the disease. Whatever the onset, the last stages are characterized by intellectual vacuity and loss of control over all body functions. The brains of patients with Alzheimer's disease are characterized by the loss of neurons, which, as the disease progresses, becomes severe and leads to decreased brain size and weight. A variety of genetic factors have been identified in the different forms of Alzheimer's disease. The rare cases of the early familial forms of the disease are linked to three different genetic defects found on three different chromosomes—chromosomes 1, 14, and 21. Another gene on chromosome 19 is believed to play a part in the more common late-onset cases.

Huntington's chorea occurs at the rate of about 5 per 100,000 individuals. It affects both sexes equally and usually becomes manifest in the fourth decade of life. The disorder is characterized by uncontrolled movements (chorea), dementia, and death within 20 years after onset. The symptoms worsen until the patient becomes totally incapacitated and bedridden. Huntington's chorea is a hereditary disease passed on as an autosomal dominant trait (see above Diseases of genetic origin). Because of its highly regular familial inheritance, the disease is often traceable to the original carriers who introduced the defective gene. The recent localization of the Huntington's chorea gene to chromosome 4 and its cloning will allow identification of the gene product, insight into the mechanism responsible for the disease, and perhaps

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effective treatment. It will also permit the disease to be diagnosed in fetuses as well as in children before the onset of symptoms.

Parkinson's disease is a motor disorder characterized by the onset of a rhythmic tremor, muscle rigidity, difficulty and slowness in movement, and stooped posture. As the disease progresses, the face of the patient becomes expressionless, the rate of swallowing is reduced, leading to drooling, and depression and dementia increase. The prevalence of Parkinson's disease is estimated to be about 160 per 100,000 persons in the general population, with about 16 to 19 new cases per 100,000 appearing each year. Men are slightly more affected than women, and there are no apparent racial differences. The disease appears typically in the sixth and seventh decades, although occasionally it can begin as early as the third decade. Parkinson's disease has no known cause. A marked decrease in the level of dopamine, a major neurotransmitter, has been noted in the brains of patients with Parkinson's disease, and this change has been attributed to the loss of so-called dopaminergic neurons that normally synthesize and use dopamine to communicate with other neurons in parts of the brain that regulate motor function.

INFECTIOUS DISEASES

Infection is the invasion of the body by various agents—including bacteria, fungi, protozoans, viruses, and worms—and its reaction to them or their toxins.

Infections are called subclinical until they perceptibly affect health, when they become infectious diseases. Infection can be local (e.g., an abscess), confined to one body system (e.g., pneumonia in the lungs), or generalized (e.g., septicemia). Infectious agents can enter the body by inhalation, ingestion, sexual transmission, passage to a fetus during pregnancy or birth, wound contamination, or animal or insect bites. The body responds with an attack on the invader by leukocytes, production of antibodies or antitoxins, and often a rise in temperature. The antibodies may result in short-term or lifelong immunity. Despite significant progress in preventing and treating infectious diseases, they remain a major cause of illness and death, particularly in regions of poor sanitation, poor nutrition, and crowding.

The most important barriers to invasion of the human host by microorganisms are the skin and mucous membranes (the tissues that line the nose, mouth, and upespiratory tract). When these tissues have been broken or affected by earlier disease, invasion by microorganisms may occur. These microorganisms may produce a local infectious disease, such as boils, or may invade the bloodstream and be carried throughout the body, producing generalized bloodstream infection (**septicemia**) or localized infection at a distant site, such as **meningitis** (an infection of the coverings of the brain and spinal cord). Infectious agents swallowed in food and drink can attack the wall of the intestinal tract and cause local or general disease. The conjunctiva, which covers the front of the eye, may be penetrated by viruses that cause a local inflammation of the eye or that pass into the bloodstream and cause a severe general disease, such as or smallpox. Microorganisms can enter the body through the genital tract, causing the acute inflammatory reaction of gonorrhea in the genital and pelvic organs or spreading out to attack almost any organ of the body with the more chronic and destructive lesions of syphilis. Even before birth, viruses and other infectious agents can pass through the placenta and attack developing cells, so that an infant may be diseased or deformed at birth.

From conception to death, humans are targets for attack by multitudes of other living organisms, all of them competing for a place in the common environment. The air people breathe, the soil they walk on, the waters and vegetation around them, the buildings they inhabit and work in, all can be populated with forms of life that are potentially dangerous. Domestic animals may harbour organisms that are a threat, and wildlife teems with agents of infection that can afflict humans with serious disease. However, the human body is not without defences against these threats, for it is equipped with a comprehensive immune system that reacts quickly and specifically against disease organisms when they attack. Survival throughout the ages has depended largely on these reactions, which today are supplemented and strengthened through the use of medical drugs.

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Categories of Organisms: The agents of infection can be divided into different groups on the basis of their size, biochemical characteristics, or manner in which they interact with the human host. The groups of organisms that cause infectious diseases are categorized as bacteria, viruses, Chlamydia, rickettsias, mycoplasmas and ureaplasmas, fungi, and parasites.

Bacteria

Bacteria can survive within the body but outside individual cells. Some bacteria, classified as **aerobes**, require oxygen for growth, while others, such as those normally found in the small intestine of healthy persons, grow only in the absence of oxygen and, therefore, are called **anaerobes**. Most bacteria are surrounded by a capsule that appears to play an important role in their ability to produce disease. Also, a number of bacterial species give off toxins that, in turn, may damage tissues. Bacteria are generally large enough to be seen under a light microscope. **Streptococci**, the bacteria that cause scarlet fever, are about 0.75 micrometre (0.00003 inch) in diameter. The **spirochetes**, which cause syphilis, leptospirosis, and rat-bite fever, are 5 to 15 micrometres long. Bacterial infections can be treated with **antibiotics**.

Bacterial infections are commonly caused by pneumococci, staphylococci, and streptococci, all of which are often **commensals** (that is, organisms living harmlessly on their hosts) in the upper respiratory tract but that can become virulent and cause serious conditions, such as pneumonia, septicemia (blood poisoning), and meningitis. The **pneumococcus** is the most common cause of lobar pneumonia, the disease in which one or more lobes, or segments, of the lung become solid and airless as a result of inflammation. **Staphylococci** affect the lungs either in the course of staphylococcal septicemia—when bacteria in the circulating blood cause scattered abscesses in the lungs—or as a complication of a viral infection, commonly influenza—when these organisms invade the damaged lung cells and cause a life-threatening form of pneumonia. Streptococcal pneumonia is the least common of the three and occurs usually as a complication of influenza or other lung disease.

Pneumococci often enter the bloodstream from inflamed lungs and cause **septicemia**, with continued fever but no other special symptoms. Staphylococci produce a type of septicemia with high, swinging fever; the bacteria reach almost any organ of the body, the brain, the bones, and especially the lungs, and destructive abscesses form in the infected areas. Streptococci also cause septicemia with fever, but the organisms tend to cause inflammation of surface lining cells rather than abscesses—for example, pleurisy (inflammation of the chest lining) rather than lung abscess, and peritonitis (inflammation of the membrane lining the abdomen) rather than liver abscess. In the course of either of the last two forms of septicemia, organisms may enter the nervous system and cause streptococcal or staphylococcal meningitis, but these are rare conditions. Pneumococci, on the other hand, often spread directly into the central nervous system, causing one of the common forms of meningitis.

Staphylococci and streptococci are common causes of skin diseases. Boils and **impetigo** (in which the skin is covered with blisters, pustules, and yellow crusts) may be caused by either. The staphylococcus also can cause a severe skin infection that strips the outer skin layers off the body and leaves the underlayers exposed, as in severe burns, a condition known as **toxic epidermal necrolysis**. In the 1980s causes of severe **streptococcal necrotizing fasciitis**, causing disease and death, began to be noticed. Streptococci can be the cause of the red cellulitis of the skin known as **erysipelas**.

Some staphylococci produce an intestinal toxin and cause food poisoning. Streptococci settling in the throat produce a reddening toxin that speeds through the bloodstream and produces the symptoms of **scarlet fever**. Streptococci and staphylococci also can cause toxic shock syndrome, a potentially fatal disease.

Meningococci are fairly common inhabitants of the throat, in most cases causing no illness at all. As the number of healthy carriers increases in any population, however, there is a tendency for the meningococcus to become more invasive. When an opportunity is presented, it can gain access to the bloodstream, invade the central nervous system, and cause **meningococcal meningitis** (formerly called cerebrospinal meningitis or spotted fever). Meningococcal meningitis, at one time a dreaded and still a

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very serious disease, usually responds to treatment with penicillin if diagnosed early enough. When meningococci invade the bloodstream, some gain access to the skin and cause bloodstained spots, or purpura. If the condition is diagnosed early enough, antibiotics can clear the bloodstream of the bacterium and prevent any from getting far enough to cause meningitis. Sometimes the septicemia takes a mild, chronic, relapsing form with no tendency toward meningitis; this is curable once it is diagnosed. The meningococcus also can cause one of the most fulminating of all forms of septicemia, meningococcemia, in which the body is rapidly covered with a purple rash, purpura fulminans; in this form the blood pressure becomes dangerously low, the heart and blood vessels are affected by shock, and the infected person dies within a matter of hours. Few are saved, despite treatment with appropriate drugs.

Haemophilus influenzae is a microorganism named for its occurrence in the sputum of patients with influenza—an occurrence so common that it was at one time thought to be the cause of the disease. It is now known to be a common inhabitant of the nose and throat that may invade the bloodstream, producing meningitis, pneumonia, and various other diseases. In children it is the most common cause of acute epiglottitis, an infection in which tissue at the back of the tongue becomes rapidly swollen and obstructs the airway, creating a potentially fatal condition. H. influenzae also is the most common cause of meningitis and pneumonia in children under five years of age, and it is known to cause bronchitis in adults. The diagnosis is established by cultures of blood, cerebrospinal fluid, or other tissue from sites of infection. Antibiotic therapy is generally effective, although death from sepsis or meningitis is still common. In developed countries where H. influenza vaccine is used, there has been a great decrease in serious infections and deaths

Viruses

Viruses are not, strictly speaking, living organisms. Instead, they are nucleic acid fragments packaged within protein coats that require the machinery of living cells to replicate. Viruses are visible by electron microscopy; they vary in size from about 25 nanometres for poliovirus to 250 nanometres for smallpox virus. Vaccination has been the most successful weapon against viral infection; some infections may be treated with antiviral drugs.

Viruses of the **Herpesviridae family** cause a multiplicity of diseases. Those causing infections in humans are the **varicella-zoster** virus (VZV), which causes **chickenpox and herpes zoster** (shingles); the **Epstein-Barr virus**, which causes infectious mononucleosis; the **cytomegalovirus**, which is most often associated with infections of newborn infants and immunocompromised people; and **herpes simplex virus**, which causes cold sores and herpetic venereal (sexually transmitted) diseases.

There are two serotypes of herpes simplex virus, HSV-1 and HSV-2. HSV-1 is the common cause of cold sores. The primary infection usually occurs in childhood and is without symptoms in 50 to 80 percent of cases. Between 10 and 20 percent of infected individuals have recurrences precipitated by emotional stress or by other illness. HSV-1 can also cause infections of the eye, central nervous system, and skin. Serious infections leading to death may occur in immunocompromised persons. HSV-2 is associated most often with herpetic lesions of the genital area. The involved area includes the vagina, cervix, vulva, and, occasionally, the urethra in females and the head of the penis in males; it may also cause an infection at the site of an abrasion. The disease is usually transmitted by sexual contact. In herpetic venereal diseases, the lesions are small, red, painful spots filled with fluid, and quickly rupture, leaving eroded areas that eventually become scabbed. These primary lesions occur from two to eight days after exposure and may be present for up to three weeks. Viral shedding and pain usually resolve in two weeks. When infections recur, the duration of the pain, lesions, and viral shedding is approximately 10 days.

Chlamydia

Chlamydia are intracellular organisms found in many vertebrates, including birds and humans and other mammals. Clinical illnesses are caused by two species, **C. trachomatis and C. psittaci.** The former is a frequent cause of genital infections in women, and, if an infant passes through an infected birth canal, it can produce disease of the eye (conjunctivitis) and pneumonia in the newborn. Young children

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sometimes develop ear infections, laryngitis, and upper respiratory tract disease from Chlamydia. Such infections can be treated with erythromycin.

C. psittaci produces **psittacosis**, a disease that results from exposure to the discharges of infected birds. The illness is characterized by high fever with chills, a slow heart rate, pneumonia, headache, weakness, fatigue, muscle pains, anorexia, nausea, and vomiting. The diagnosis is usually suspected if the patient has a history of exposure to birds and is confirmed by blood tests. Mortality is rare, and specific antibiotic treatment is available.

Rickettsias

The rickettsias are a family of microorganisms named for American pathologist Howard T. Ricketts, who died of typhus in 1910 while investigating the spread of the disease. The rickettsias, which range in size from 250 nanometres to more than 1 micrometre and have no cell wall but are surrounded by a cell membrane, cause a group of diseases characterized by fever and a rash. Except for Coxiella burnetii, the cause of Q fever, they are intracellular parasites, most of which are transmitted to humans by an arthropod carrier such as a louse or tick. C. burnetii, however, can survive in milk, sewage, and aerosols and can be transmitted to humans by a tick or by inhalation, causing pneumonia in the latter case. Rickettsial diseases can be treated with antibiotics.

Humans contract most rickettsial diseases only when they break into a cycle in nature in which the rickettsias live. In murine typhus, for example, Rickettsia mooseri is a parasite of rats conveyed from rat to rat by the rat flea, Xenopsylla cheopis; it bites humans if they intrude into its environment. Scrub typhus is caused by R. tsutsugamushi, but it normally parasitizes only rats and mice and other rodents, being carried from one to the other by a small mite, Leptotrombidium (previously known as Trombicula). This mite is fastidious in matters of temperature, humidity, and food and finds everything suitable in restricted areas, or "mite islands," in South Asia and the western Pacific. It rarely bites humans in their normal environment, but if people invade its territory en masse it will attack, and outbreaks of scrub typhus will follow.

The spotted fevers are caused by rickettsias that spend their normal life cycles in a variety of small animals, spreading from one to the other inside ticks; these bite human intruders and cause African, North Asian, and Queensland tick typhus, as well as Rocky Mountain spotted fever. One other spotted fever, rickettsialpox, is caused by R. akari, which lives in the body of the ordinary house mouse, Mus musculus, and spreads from one to another inside the house mite Allodermanyssus sanguineus. This rickettsia is probably a parasite of wild field mice, and it is perhaps only when cities push out into the countryside that house mice catch the infection.

Mycoplasmas and Ureaplasmas

Mycoplasmas and ureaplasmas, which range in size from 150 to 850 nanometres, are the smallest free-living microorganisms. They are ubiquitous in nature and capable of causing widespread disease, but the illnesses they produce in humans are generally milder than those caused by bacteria. Diseases due to mycoplasmas and ureaplasmas can be treated with antibiotics.

Mycoplasma pneumoniae is the most important member of its genus. M. pneumoniae is associated with 20 percent of all cases of pneumonia in adults and children over five years of age. Patients have fever, cough, headache, and malaise and, upon physical examination, may be found to have pharyngitis (inflamed throat), enlarged lymph nodes, ear or sinus infection, bronchitis, or croup. Diagnosis is established by chest X-rays and blood tests. Although treatment with erythromycin or tetracycline may shorten the illness, it can last for weeks.

Mycoplasmas may also cause a red, bumpy rash—usually on the trunk or back—that is occasionally vesicular (with blisters). Inflammation of the heart muscle and the covering of the heart (pericardium) is rare but can be caused by mycoplasmas. About one-fourth of the people infected with these organisms

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experience nausea, vomiting, diarrhea, and cramping abdominal pain. Inflammation of the pancreas (pancreatitis) or the liver (hepatitis) may occur, and infection of the brain and spinal cord is a serious complication.

Ureaplasmas can be recovered frequently from the genital areas of healthy persons. The organism can cause inflammation of the urethra and has been associated with infertility, low birth weight of infants, and repeated stillbirths. In general, however, ureaplasma infections are mild. Tetracycline is the preferred treatment once the organism has been established as the cause of infection by microscopic examination of urethral secretions.

Fungi

Fungi are large organisms that usually live on dead and rotting animal and plant matter. They are found mostly in soil, on objects contaminated with soil, on plants and animals, and on skin, and they may also be airborne. Fungi may exist as yeasts or molds and may alternate between the two forms, depending on environmental conditions. Yeasts are simple cells, 3 to 5 micrometres (0.0001 to 0.0002 inch) in diameter. Molds consist of filamentous branching structures (called hyphae), 2 to 10 micrometres in diameter, that are formed of several cells lying end to end. Fungal diseases in humans are called mycoses; they include such disorders as histoplasmosis, coccidioidomycosis, and blastomycosis. These diseases can be mild, characterized by an upper respiratory infection, or severe, involving the bloodstream and every organ system. Fungi may cause devastating disease in persons whose defenses against infection have been weakened by malnutrition, cancer, or the use of immunosuppressive drugs. Specific types of antibiotics known as antifungals are effective in their treatment.

Parasites

Among the infectious parasites are the protozoans, unicellular organisms that have no cell wall, that cause such diseases as malaria. The various species of malarial parasites are about 4 micrometres (0.0002 inch) in diameter. At the other extreme, the tapeworm can grow to several metres in length; treatment is designed either to kill the worm or to dislodge it from its host.

The worm **Ascaris lumbricoides** causes ascariasis, one of the most prevalent infections in the world. Ascaris lives in the soil, and its eggs are ingested with contaminated food. The eggs hatch in the human intestine, and the worms then travel through the bloodstream to the liver, heart, and lungs. They can cause pneumonia, perforations of the intestine, or blockage of the bile ducts, but infected people usually have no symptoms beyond the passage of worms in the stool. Specific treatment is available and prognosis is excellent.

Infections are also caused by whipworms, genus **Trichuris, and pinworms, Enterobius vermicularis,** each popularly named for its shape. The former is parasitic in the human large intestine and may cause chronic diarrhea. The latter can be found throughout the gastrointestinal tract, especially in children, and can cause poor appetite, loss of weight, anemia, and itching in the anal area (where it lays its eggs). Both conditions are easily diagnosed and treated with drugs.

Modes of Survival of Infectious Agents

Infectious agents have various methods of survival. Some depend on rapid multiplication and rapid spread from one host to another. For example, when the measles virus enters the body, it multiplies for a week or two and then enters the bloodstream and spreads to every organ. For several days before a rash appears, the surface cells of the respiratory tract are bursting with measles virus, and vast quantities are shed every time the infected person coughs or sneezes. A day or two after the rash appears, the amount of antibody rises in the bloodstream, neutralizing the virus and stopping further shedding. The patient rapidly becomes noninfectious but already may have spread the virus to others. In this way an epidemic can rapidly occur. Many other infectious agents—for example, influenza virus—survive in this manner. How such viruses exist between epidemics is, in some cases, less clear.

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The picture is different in more-chronic infections. In tuberculosis there is neither overwhelming multiplication nor rapid shedding of the tubercle bacillus. Rather, the bacilli remain in the infected person's body for a long period, slowly forming areas of chronic inflammation that may from time to time break down and allow them to escape.

Some organisms form spores, a resting or dormant stage that is resistant to heat, cold, drying, and chemical action. Spore-forming organisms can survive for months or years under the most adverse conditions and may not, in fact, be highly infectious. The bacterium that causes tetanus, Clostridium tetani, is present everywhere in the environment—in soil, in dust, on window ledges and floors—and yet tetanus is an uncommon disease, especially in developed countries. The same is true of the anthrax bacterium, Bacillus anthracis. Although usually present in abundance in factories in which rawhides and animal wool and hair are handled, it rarely causes anthrax in employees. Clostridium botulinum, the cause of botulism, produces one of the most lethal toxins that can afflict humans, and yet the disease is one of the rarest because the microorganism depends for its survival on its resistant spore.

In contrast to these relatively independent organisms, there are others that cannot exist at all outside the human body. The germs of syphilis and gonorrhea, for example, depend for survival on their ability to infect and their adaptation to the human environment.

Some organisms have complicated life cycles and depend on more than one host. The malarial parasite must spend a portion of its life cycle inside a mosquito, while the liver fluke Fasciola hepatica, an occasional human parasite, spends part of its life in the body of a land animal such as a sheep, part in a water snail, and part in the open air as a cyst attached to grass.

Commensal Organisms

All of the outer surfaces of the human body are covered with microorganisms that normally do no harm and may, in fact, be beneficial. Those commensal organisms on the skin help to break down dying skin cells or to destroy debris secreted by the many minute glands and pores that open on the skin. Many of the organisms in the intestinal tract break down complex waste products into simple substances, and others help in the manufacture of chemical compounds that are essential to human life.

The gastrointestinal tract is considered in this regard to be one of these "outer" surfaces since it is formed by the intucking, or invagination, of the ectoderm, or outer surface, of the body. The mouth, nose, and sinuses (spaces inside the bones of the face) are also considered to be external structures because of their direct contact with the outside environment. Both the gastrointestinal tract and the mouth, nose, and sinuses are heavily populated with microorganisms, some of which are true commensals—living in humans and deriving their sustenance from the surface cells of the body without doing any harm—and others of which are indistinguishable from disease germs. The latter may live like true commensals in a particular tract in a human and never cause disease, despite their potential to do so. When the environment is altered, however, they are capable of causing severe illness in their host, or, without harming their host, they may infect another person with a serious disease.

It is not known why, for example, the hemolytic streptococcus bacterium can live for months in the throat without causing harm and then suddenly cause an acute attack of tonsillitis or how an apparently harmless pneumococcus gives rise to pneumonia. Similarly, it is not understood how a person can harmlessly carry Haemophilus influenzae type B in the throat but then become ill when the organism invades the body and causes one of the most severe forms of meningitis. It may be that external influences, such as changes in temperature or humidity, are enough to upset the balance between host and parasite or that a new microbial invader enters and, by competing for some element in the environment, forces the original parasite to react more violently with its host. The term lowered resistance, often used to describe conditions at the onset of infectious disease, is not specific and simply implies any change in the immune system of the host.

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A microorganism's environment can be changed radically, of course. If antibiotics are administered, the body's commensal organisms can be killed, and other, less-innocuous organisms may take their place. In the mouth and throat, penicillin may eradicate pneumococci, streptococci, and other bacteria that are sensitive to the drug, while microorganisms that are insensitive, such as Candida albicans, may then proliferate and cause thrush (an inflammatory condition of the mouth and throat). In the intestinal tract, an antibiotic may kill most of the microorganisms that are normally present and allow dangerous organisms, such as Pseudomonas aeruginosa, to multiply and perhaps to invade the bloodstream and the tissues of the body. If an infectious agent—for example, Salmonella—reaches the intestinal tract, treatment with an antibiotic may have an effect that differs from what was intended. Instead of attacking and destroying the salmonella, it may kill the normal inhabitants of the bowel and allow the salmonella to flourish and persist in the absence of competition from other microorganisms.

Ecology of Infectious Diseases

Family Patterns: Humans are social animals. As a result, human social habits and circumstances influence the spread of infectious agents. Poorer families tend to live in more-crowded conditions, which facilitate the passage of disease-causing organisms from one person to another. This is true whether the germs pass through the air from one respiratory tract to another or whether they are bowel organisms that depend for their passage on close personal hand-to-mouth contact or on lapses of sanitation and hygiene. The composition of the family unit is also important. In families with infants and preschool children, infection spreads more readily, for children of this age are both more susceptible to infection and, because of their undeveloped hygiene habits, more likely to share their microbes with other family members. Because of this close and confined contact, infectious agents are spread more rapidly. Distinction must be made between disease and infection. The virus of poliomyelitis, for example, spreads easily in conditions of close contact (infection), but it usually causes no active disease. When it does cause active disease, it attacks older people much more severely than the young. Children in more-crowded homes, for example, are likely to be infected at an early age and, if illness results, it is usually mild. In less-crowded conditions, young children are exposed less often to infection; when they first encounter the virus at an older age, they tend to suffer more severely. The difference between infection and disease is seen even more rapidly in early childhood, when infection leads more often to immunity than to illness. Under high standards of hygiene, young children are exposed less frequently, and fewer develop immunity in early life, with the result that paralytic illness, a rarity under the former conditions, is seen frequently in older children and adults. The pattern of infection and disease, however, can be changed. In the case of the poliomyelitis virus, only immunization can abolish both infection and disease.

Population Density: Density of population does not of itself determine the ease with which infection spreads through a population. In New York City, with its many high-rise dwellings, the density of the population per square mile is much greater than in some of the world's older cities, but the hepatitis virus, for example, spreads much faster in the latter. A family in a New York City apartment may never see the inhabitants of most of the other apartments in the block, while neighbours in an ancient Asian city are in daily contact. In New York state, the incidence of infectious hepatitis has been shown to vary inversely with population density—to be lowest in New York City, higher in other urban areas of the state, and highest in rural areas. This pattern reflects the population's chances of contact and social habits, as well as its density.

Social Habits: The vampire bats of Brazil, which transmit paralytic rabies, bite cattle but not ranchers, presumably because ranchers are few but cattle are plentiful on the plains of Brazil. Bat-transmitted rabies, however, does occur in humans in Trinidad, where herdsmen sleep in shacks near their animals. The mechanism of infection is the same in Brazil and Trinidad, but the difference in social habits affects the incidence of the disease. During the early 20th century in Malta, goats were milked at the customers' doors, and a Brucella species in the milk caused a disease that was common enough to be called Malta fever. When the pasteurization of milk became compulsory, Malta fever almost disappeared from the island. (It continued to occur in rural areas where people still drank their milk raw and were in daily contact with their infected animals.) Important alterations in environment also occur when children in a

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modern community first go to school. Colds, coughs, sore throats, and swollen neck glands can occur one after the other. In a nursery school, with young children whose hygiene habits are undeveloped, outbreaks of dysentery and other bowel infections may occur, and among children who take their midday meal at school, foodborne infection caused by a breakdown in hygiene can sweep through entire classes of students. These are dangers against which the children are protected to some extent at home but against which they have no defense when they move to the school environment. Changing food habits among the general population also affect the environment for humans and microbes. Meals served in restaurants, for example, offer a greater danger of food poisoning if the standard of hygiene for food preparation is flawed. The purchase and preparation of poultry—which is often heavily infected with Salmonella—present a particular danger. If chickens are bought fresh from a farm or shop and cooked in an oven at home, food poisoning from eating them is rare. If poultry is purchased while it is deep-frozen and then not fully thawed before it is cooked, there is a good chance that insufficient heat penetration will allow the Salmonella—which thrive in the cold—to survive in the meat's centre and infect the people who eat it.

Temperature and Humidity: At a social gathering, the human density per square yard may be much greater than in any home, and humidity and temperature may rise to levels uncomfortable for humans but ideal for microbes. Virus-containing droplets pass easily from one person to another, and an outbreak of the common cold may result. In contrast, members of scientific expeditions have spent whole winters in the Arctic or Antarctic without any respiratory illness, only to catch severe colds upon the arrival of a supply ship in the early summer. This is because viruses, not cold temperatures, cause colds. During polar expeditions, the members rapidly develop immunity to the viruses they bring with them, and, throughout the long winter, they encounter no new ones. Their colds in the summer are caused by viruses imported by the crew of the supply ship. When the members of the expedition return on the ship to temperate zones, they again come down with colds, this time caught from friends and relatives who have spent the winter at home.

Migration: Movement into a new environment often is followed by an outbreak of infectious disease. On pilgrimages and in wars, improvised feeding and sanitation lead to outbreaks of such intestinal infections as dysentery, cholera, and typhoid fever, and sometimes more have died in war from these diseases than have been killed in the fighting. People entering isolated communities may carry a disease such as measles with them, and the disease may then spread with astonishing rapidity and often with enhanced virulence. A traveler from Copenhagen carried measles virus with him to the Faroe Islands in 1846, and 6,000 of the 8,000 inhabitants caught the disease. Most of those who escaped were old enough to have acquired immunity during a measles outbreak 65 years earlier. In Fiji a disastrous epidemic of measles in 1875 killed one-fourth of the population. In these cases, the change of environment favoured the virus. Nearly every person in such "virgin" populations is susceptible to infection, so that a virus can multiply and spread unhindered. In a modern city population, by contrast, measles virus mainly affects susceptible young children. When it has run through them, the epidemic must die down because of a lack of susceptible people, and the virus does not spread again until a new generation of children is on hand. With the use of measles vaccine, the supply of susceptible young children is reduced, and the virus cannot spread and multiply and must die out. An innocent change in environment such as that experienced during camping can lead to infection if it brings a person into contact with sources of infection that are absent at home. Picnicking in a wood, a person may be bitten by a tick carrying the virus of one of several forms of encephalitis; as he swims in a canal or river, his skin may be penetrated by the organisms that cause leptospirosis. He may come upon some watercress growing wild in the damp corner of a field and may swallow with the cress almost invisible specks of life that will grow into liver flukes in his body, giving him fascioliasis, an illness that is common in cattle and sheep but that can spread to humans when circumstances are in its favour.

Occupation and Commerce: In occupational and commercial undertakings, people often manipulate their environment and, in so doing, expose themselves to infection. A farmer in his fields is exposed to damp conditions in which disease microorganisms flourish. While clearing out a ditch, he may be infected with leptospires passed into the water in rats' urine. In his barns he may be exposed to brucellosis if his

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herd of cattle is infected or to salmonellosis or Q fever. Slaughterhouse workers run similar risks, as do veterinarians. A worker in a dock or tannery may get anthrax from imported hides; an upholsterer may get the disease from wool and hair; and a worker mending sacks that have contained bone meal may contract the disease from germs still clinging to the sack. Workers in packing plants and shops often are infected from the raw meat that they handle; they are sometimes regarded as carriers and causes of outbreaks of Salmonella food poisoning, but as often as not they are victims rather than causes. Workers in poultry plants can contract salmonellosis, more rarely psittacosis or a viral infection of the eye, from the birds that they handle. Forestry workers who enter a reserve may upset the balance of nature of the area and expose themselves to attack from the undergrowth or the trees by insect vectors of disease that, if undisturbed, would never come into contact with humans. Whenever people manipulate the environment—by herding animals, by importing goods from abroad, by draining a lake, or by laying a pipe through swampy land, and in many other seemingly innocent ways—they run the chance of interfering with microbial life and attracting into their own environment agents of disease that they might not otherwise ever encounter.

NUTRITIONAL DEFICIENCY RELATED DISEASES

The most significant nutrition-related disease is chronic **undernutrition**, which plagues more than 800 million people worldwide. Undernutrition is a condition in which there is insufficient food to meet energy needs; its main characteristics include weight loss, failure to thrive, and wasting of body fat and muscle. Low birthweight in infants, inadequate growth and development in children, diminished mental function, and increased susceptibility to disease are among the many consequences of chronic persistent hunger, which affects those living in poverty in both industrialized and developing countries. The largest number of chronically hungry people live in Asia, but the severity of hunger is greatest in sub-Saharan Africa. At the start of the 21st century, approximately 20,000 people, the majority of them children, died each day from undernutrition and related diseases that could have been prevented. The deaths of many of these children stem from the poor nutritional status of their mothers, as well as the lack of opportunity imposed by poverty.

Only a small percentage of hunger deaths is caused by starvation due to catastrophic food shortages. During the 1990s, for example, worldwide famine (epidemic failure of the food supply) more often resulted from complex social and political issues and the ravages of war than from natural disasters such as droughts and floods.

Malnutrition is the impaired function that results from a prolonged deficiency—or excess—of total energy or specific nutrients such as protein, essential fatty acids, vitamins, or minerals. This condition can result from fasting and anorexia nervosa; persistent vomiting (as in bulimia nervosa) or inability to swallow; impaired digestion and intestinal malabsorption; or chronic illnesses that result in loss of appetite (e.g., cancer, AIDS). Malnutrition can also result from limited food availability, unwise food choices, or overzealous use of dietary supplements. (*See* the table of select nutrient-deficiency diseases.)

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Select nutrient-deficiency diseases		
disease (and key nutrient involved)	**symptoms**	**foods rich in key nutrient**
xerophthalmia (vitamin A)	blindness from chronic eye infections, poor growth, dryness and keratinization of epithelial tissues	liver, fortified milk, sweet potatoes, spinach, greens, carrots, cantaloupe, apricots
rickets (vitamin D)	weakened bones, bowed legs, other bone deformities	fortified milk, fish oils, sun exposure
beriberi (thiamin)	nerve degeneration, altered muscle coordination, cardiovascular problems	pork, whole and enriched grains, dried beans, sunflower seeds
pellagra (niacin)	diarrhea, skin inflammation, dementia	mushrooms, bran, tuna, chicken, beef, peanuts, whole and enriched grains
scurvy (vitamin C)	delayed wound healing, internal bleeding, abnormal formation of bones and teeth	citrus fruits, strawberries, broccoli
iron-deficiency anemia (iron)	decreased work output, reduced growth, increased health risk in pregnancy	meat, spinach, seafood, broccoli, peas, bran, whole-grain and enriched breads
goitre (iodine)	enlarged thyroid gland, poor growth in infancy and childhood, possible mental retardation, cretinism	iodized salt, saltwater fish
Source: Gordon M. Wardlaw, *Perspectives in Nutrition* (1999).		

PROTEIN ENERGY MALNUTRITION

Chronic undernutrition manifests primarily as protein-energy malnutrition (PEM), which is the most common form of malnutrition worldwide. Also known as protein-calorie malnutrition, PEM is a continuum in which people—all too often children—consume too little protein, energy, or both. At one end of the continuum is **kwashiorkor**, characterized by a severe protein deficiency, and at the other is **marasmus**, an absolute food deprivation with grossly inadequate amounts of both energy and protein.

An infant with marasmus is extremely underweight and has lost most or all subcutaneous fat. The body has a "skin and bones" appearance, and the child is profoundly weak and highly susceptible to infections. The cause is a diet very low in calories from all sources (including protein), often from early weaning to a bottled formula prepared with unsafe water and diluted because of poverty. Poor hygiene and continued depletion lead to a vicious cycle of gastroenteritis and deterioration of the lining of the gastrointestinal tract, which interferes with absorption of nutrients from the little food available and further reduces resistance to infection. If untreated, marasmus may result in death due to starvation or heart failure.

Kwashiorkor, a Ghanaian word meaning the disease that the first child gets when the new child comes, is typically seen when a child is weaned from high-protein breast milk onto a carbohydrate food source with insufficient protein. Children with this disease, which is characterized by a swollen belly due to edema (fluid retention), are weak, grow poorly, and are more susceptible to infectious diseases, which may result in fatal diarrhea. Other symptoms of kwashiorkor include apathy, hair discoloration, and dry,

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peeling skin with sores that fail to heal. Weight loss may be disguised because of the presence of edema, enlarged fatty liver, and intestinal parasites; moreover, there may be little wasting of muscle and body fat.

Kwashiorkor and marasmus can also occur in hospitalized patients receiving intravenous glucose for an extended time, as when recovering from surgery, or in those with illnesses causing loss of appetite or malabsorption of nutrients. Persons with eating disorders, cancer, AIDS, and other illnesses where appetite fails or absorption of nutrients is hampered may lose muscle and organ tissue as well as fat stores.

Treatment of PEM has three components. (1) Life-threatening conditions—such as fluid and electrolyte imbalances and infections—must be resolved. (2) Nutritional status should be restored as quickly and safely as possible; rapid weight gain can occur in a starving child within one or two weeks. (3) The focus of treatment then shifts to ensuring nutritional rehabilitation for the long term. The speed and ultimate success of recovery depend upon the severity of malnutrition, the timeliness of treatment, and the adequacy of ongoing support. Particularly during the first year of life, starvation may result in reduced brain growth and intellectual functioning that cannot be fully restored.

CARBOHYDRATES

Under most circumstances, there is no absolute dietary requirement for carbohydrates—simple sugars, complex carbohydrates such as starches, and the indigestible plant carbohydrates known as dietary fibre. Certain cells, such as brain cells, require the simple carbohydrate glucose as fuel. If dietary carbohydrate is insufficient, glucose synthesis depends on the breakdown of amino acids derived from body protein and dietary protein and the compound glycerol, which is derived from fat. Long-term carbohydrate inadequacy results in increased production of organic compounds called ketones (a condition known as ketosis), which imparts a distinctive sweet odour to the breath. Ketosis and other untoward effects of a very-low-carbohydrate diet can be prevented by the daily consumption of 50 to 100 grams of carbohydrate; however, obtaining at least half of the daily energy intake from carbohydrates is recommended and is typical of human diets, corresponding to at least 250 grams of carbohydrate (1,000 calories in a 2,000-calorie diet). A varied diet containing fruits, vegetables, legumes, and whole-grain cereals, which are all abundant in carbohydrates, also provides a desirable intake of dietary fibre.

ESSENTIAL FATTY ACIDS

There is also a minimum requirement for fat—not for total fat, but only for the fatty acids linoleic acid (a so-called omega-6 fatty acid) and alpha-linolenic acid (an omega-3 fatty acid). Deficiencies of these two fatty acids have been seen in hospitalized patients fed exclusively with intravenous fluids containing no fat for weeks, patients with medical conditions affecting fat absorption, infants given formulas low in fat, and young children fed nonfat milk or low-fat diets. Symptoms of deficiency include dry skin, hair loss, and impaired wound healing. Essential fatty acid requirements—a few grams a day—can be met by consuming approximately a tablespoon of polyunsaturated plant oils daily. Fatty fish also provides a rich source of omega-3 fatty acids. Even individuals following a low-fat diet generally consume sufficient fat to meet requirements.

VITAMINS

Although deficiency diseases have been described in laboratory animals and humans deprived of single vitamins, in human experience multiple deficiencies are usually present simultaneously. The eight B-complex vitamins function in coordination in numerous enzyme systems and metabolic pathways; thus, a deficiency of one may affect the functioning of others.

Vitamin A: Vitamin A deficiency is the leading cause of preventable blindness in children and is a major problem in the developing world, especially in Africa and Southeast Asia; in the poorest countries hundreds of thousands of children become blind each year due to a deficiency of the vitamin. Even a mild deficiency can impair immune function, thereby reducing resistance to disease. Night blindness is an

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early sign of vitamin A deficiency, followed by abnormal dryness of the eye and ultimately scarring of the cornea, a condition known as xerophthalmia. Other symptoms include dry skin, hardening of epithelial cells elsewhere in the body (such as mucous membranes), and impaired growth and development. In many areas where vitamin A deficiency is endemic, the incidence is being reduced by giving children a single large dose of vitamin A every six months. A genetically modified form of rice containing beta-carotene, a precursor of vitamin A, has the potential to reduce greatly the incidence of vitamin A deficiency, but the use of this so-called golden rice is controversial.

Vitamin D: Vitamin D (also known as vitamin D hormone) is synthesized in the body in a series of steps, starting in the skin by the action of sunlight's ultraviolet rays on a precursor compound; thus, without adequate food sources of vitamin D, a deficiency of the vitamin can occur when exposure to sunlight is limited. Lack of vitamin D in children causes rickets, a disease characterized by inadequate mineralization of bone, growth retardation, and skeletal deformities such as bowed legs. The adult form of rickets, known as osteomalacia, results in weak muscles as well as weak bones. Inadequate vitamin D may also contribute to the thinning of bones seen in osteoporosis. Individuals with limited sun exposure (including women who completely cover their bodies for religious reasons), elderly or homebound persons, and those with dark skin, particularly those who live in northern latitudes, are at risk of vitamin D deficiency. Vitamin D is found in very few foods naturally; thus fortification of milk and other foods (e.g., margarine, cereals, and breads) with the vitamin has helped protect those populations in which sun exposure is inadequate. Supplemental vitamin D also may help protect against bone fractures in the elderly, who make and activate vitamin D less efficiently even if exposed to sunlight.

Vitamin E: Vitamin E deficiency is rare in humans, although it may develop in premature infants and in people with impaired fat absorption or metabolism. In the former, fragility of red blood cells (hemolysis) is seen; in the latter, where deficiency is more prolonged, neuromuscular dysfunction involving the spinal cord and retina may result in loss of reflexes, impaired balance and coordination, muscle weakness, and visual disturbances. No specific metabolic function has been established for vitamin E; however, it is an important part of the antioxidant system that inhibits lipid peroxidation; i.e., it protects cells and their membranes against the damaging effects of free radicals (reactive oxygen and nitrogen species) that are produced metabolically or enter the body from the environment. The requirement for vitamin E is increased with increasing consumption of polyunsaturated fatty acids. People who smoke or are subjected to air pollution may also need more of the vitamin to protect against oxidative damage to the lungs.

Vitamin K: Vitamin K is necessary for the formation of prothrombin and other blood-clotting factors in the liver, and it also plays a role in bone metabolism. A form of the vitamin is produced by bacteria in the colon and can be utilized to some degree. Vitamin K deficiency causes impaired clotting of the blood and internal bleeding, even without injury. Due to poor transport of vitamin K across the placenta, newborn infants in developed countries are routinely given the vitamin intramuscularly or orally within six hours of birth to protect against a condition known as hemorrhagic disease of the newborn. Vitamin K deficiency is rare in adults, except in syndromes with poor fat absorption, in liver disease, or during treatment with certain anticoagulant drugs, which interfere with vitamin K metabolism. Bleeding due to vitamin K deficiency may be seen in patients whose gut bacteria have been killed by antibiotics.

B1 **Thiamin:** Prolonged deficiency of thiamin (vitamin B_1) results in beriberi, a disease that has been endemic in populations where white rice has been the staple. Thiamin deficiency is still seen in areas where white rice or flour constitutes the bulk of the diet and thiamin lost in milling is not replaced through enrichment. Symptoms of the form known as dry beriberi include loss of appetite, confusion and other mental symptoms, muscle weakness, painful calf muscles, poor coordination, tingling and paralysis. In wet beriberi there is edema and the possibility of an enlarged heart and heart failure. Thiamin deficiency can also occur in populations eating large quantities of raw fish harbouring intestinal microbes that contain the enzyme thiaminase. In the developed world, thiamin deficiency is linked primarily to chronic alcoholism with poor diet, manifesting as Wernicke-Korsakoff syndrome, a condition with rapid eye movements, loss of muscle coordination, mental confusion, and memory loss.

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B2 **Riboflavin:** Riboflavin (vitamin B_2) deficiency, known as ariboflavinosis, is unlikely without the simultaneous deficiency of other nutrients. After several months of riboflavin deprivation, symptoms include cracks in the skin at the corners of the mouth, fissures of the lips, and an inflamed, magenta-coloured tongue. Because riboflavin is readily destroyed by ultraviolet light, jaundiced infants who are treated with light therapy are administered the vitamin. Milk, milk products, and cereals, major sources of riboflavin in the diet, are packaged to prevent exposure to light.

Niacin: Symptoms of pellagra develop about two months after niacin is withdrawn from the diet. Pellagra is characterized by the so-called three Ds—diarrhea, dermatitis, and dementia—and, if it is allowed to progress untreated, death ensues. Pellagra was common in areas of the southern United States in the early 1900s and still occurs in parts of India, China, and Africa, affecting people who subsist primarily on corn. The niacin in corn and other cereal grains is largely in bound form, unable to be absorbed well. Soaking corn in lime water, as practiced by Native American populations for centuries, frees bound niacin and thus protects against pellagra. In addition, unlike other cereals, corn is low in the amino acid tryptophan, which can be converted in part to niacin. Sufficient high-quality protein (containing tryptophan) in the diet can protect against niacin deficiency even if intake of niacin itself is inadequate.

B6 **Vitamin B_6:** Vitamin B_6 (pyridoxine and related compounds) is essential in protein metabolism, the synthesis of neurotransmitters, and other critical functions in the body. Deficiency symptoms include dermatitis, microcytic hypochromic anemia (small, pale red blood cells), impaired immune function, depression, confusion, and convulsions. Although full-blown vitamin B_6 deficiency is rare, marginal inadequacy is more widespread, especially among the elderly, who may have a reduced ability to absorb the vitamin. People with alcoholism, especially those with the liver diseases cirrhosis and hepatitis, are at risk of deficiency. A number of drugs, including the tuberculosis drug isoniazid, interfere with vitamin B_6 metabolism.

Folic acid: Vitamin B_{12} and folic acid (folate) are two B vitamins with many closely related functions, notably participation in DNA synthesis. As a result, people with deficiencies of either vitamin show many of the same symptoms, such as weakness and fatigue due to megaloblastic anemia, a condition in which red blood cells, lacking sufficient DNA for cell division, are large and immature. Deficiency of folic acid also causes disruption of cell division along the gastrointestinal tract, which results in persistent diarrhea, and impaired synthesis of white blood cells and platelets. Inadequate intake of the vitamin in early pregnancy may cause neural tube defects in the fetus. Thus, women capable of becoming pregnant are advised to take 400 micrograms (μg) of folic acid daily from supplements, fortified foods (such as fortified cereals), or both—in addition to consuming foods rich in folic acid such as fresh fruits and vegetables (especially leafy greens) and legumes. The cancer drug methotrexate interferes with folic acid metabolism, causing side effects such as hair loss and diarrhea. Folic acid deficiency may also result from heavy use of alcohol, which interferes with absorption of the vitamin.

Vitamin B_{12}: Deficiency of vitamin B_{12} (cobalamin), like folic acid, results in megaloblastic anemia (large, immature red blood cells), due to interference with normal DNA synthesis. Additionally, vitamin B_{12} maintains the myelin sheath that protects nerve fibres; therefore, an untreated deficiency of the vitamin can result in nerve degeneration and eventually paralysis. Large amounts of folic acid (over 1,000 μg per day) may conceal, and possibly even exacerbate, an underlying vitamin B_{12} deficiency. Only animal foods are reliable sources of vitamin B_{12}. Vegans, who eat no foods of animal origin, are at risk of vitamin B_{12} deficiency and must obtain the vitamin through fortified food or a supplement. For people who regularly eat animal products, deficiency of the vitamin is unlikely, unless there is a defect in absorption. In order to be absorbed, vitamin B_{12} must be bound to intrinsic factor, a substance secreted by the stomach. If intrinsic factor is absent (due to an autoimmune disorder known as pernicious anemia) or if there is insufficient production of hydrochloric acid by the stomach, absorption of the vitamin will be limited. Pernicious anemia, which occurs most often in the elderly, can be treated by injections or massive oral doses (1,000 μg) of vitamin B_{12}.

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Pantothenic acid: Pantothenic acid is so widespread in foods that deficiency is unlikely under normal circumstances. Deficiency has been seen only in individuals fed semisynthetic diets deficient in the vitamin or in subjects given a pantothenic acid antagonist. Symptoms of deficiency include fatigue, irritability, sleep disturbances, abdominal distress, and neurological symptoms such as tingling in the hands. Deficiency of the vitamin was suspected during World War II when prisoners of war in Asia who exhibited "burning feet" syndrome, characterized by numbness and tingling in the toes and other neurological symptoms, responded only to the administration of pantothenic acid.

Biotin: Deficiency of biotin is rare, and this may be due in part to synthesis of the vitamin by bacteria in the colon, although the importance of this source is unclear. Biotin deficiency has been observed in people who regularly eat large quantities of raw egg white, which contains a glycoprotein (avidin) that binds biotin and prevents its absorption. A rare genetic defect that renders some infants unable to absorb a form of biotin in food can be treated with a supplement of the vitamin. Long-term use of certain anticonvulsant drugs may also impair biotin absorption. Symptoms of deficiency include skin rash, hair loss, and eventually neurological abnormalities.

Vitamin C: Vitamin C, also known as ascorbic acid, functions as a water-soluble antioxidant and as a cofactor in various enzyme systems, such as those involved in the synthesis of connective tissue components and neurotransmitters. Symptoms of scurvy, a disease caused by vitamin C deficiency, include pinpoint hemorrhages (petechiae) under the skin, bleeding gums, joint pain, and impaired wound healing. Although rare in developed countries, scurvy is seen occasionally in people consuming restricted diets, particularly those containing few fruits and vegetables, or in infants fed boiled cow's milk and no source of vitamin C. Scurvy can be prevented with relatively small quantities of vitamin C (10 milligrams [mg] per day), although recommended intakes, which aim to provide sufficient antioxidant protection, are closer to 100 mg per day. Disease states, environmental toxins, drugs, and other stresses can increase an individual's vitamin C needs. Smokers, for example, may require an additional 35 mg of the vitamin daily to maintain vitamin C levels comparable to nonsmokers.

IRON

Iron deficiency is the most common of all nutritional deficiencies, with much of the world's population being deficient in the mineral to some degree. Young children and premenopausal women are the most vulnerable. The main function of iron is in the formation of hemoglobin, the red pigment of the blood that carries oxygen from the lungs to other tissues. Since each millilitre of blood contains 0.5 mg of iron (as a component of hemoglobin), bleeding can drain the body's iron reserves. When iron stores are depleted a condition arises known as microcytic hypochromic anemia, characterized by small red blood cells that contain less hemoglobin than normal. Symptoms of severe iron deficiency anemia include fatigue, weakness, apathy, pale skin, difficulty breathing on exertion, and low resistance to cold temperatures. During childhood, iron deficiency can affect behaviour and learning ability as well as growth and development. Severe anemia increases the risk of pregnancy complications and maternal death. Iron deficiency anemia is most common during late infancy and early childhood, when iron stores present from birth are exhausted and milk, which is poor in iron, is a primary food; during the adolescent growth spurt; and in women during the childbearing years, because of blood loss during menstruation and the additional iron needs of pregnancy. Intestinal blood loss and subsequent iron deficiency anemia in adults may also stem from ulcers, hemorrhoids, tumours, or chronic use of certain drugs such as aspirin. In developing countries, blood loss due to hookworm and other infections, coupled with inadequate dietary iron intake, exacerbates iron deficiency in both children and adults.

IODINE

Iodine deficiency disorders are the most common cause of preventable brain damage, which affects an estimated 50 million people worldwide. During pregnancy, severe iodine deficiency may impair fetal development, resulting in cretinism (irreversible mental retardation with short stature and developmental abnormalities) as well as in miscarriage and stillbirth. Other more pervasive

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consequences of chronic iodine deficiency include lesser cognitive and neuromuscular deficits. The ocean is a dependable source of iodine, but away from coastal areas iodine in food is variable and largely reflects the amount in the soil. In chronic iodine deficiency the thyroid gland enlarges as it attempts to trap more iodide (the form in which iodine functions in the body) from the blood for synthesis of thyroid hormones, and it eventually becomes a visible lump at the front of the neck known as a goitre. Some foods, such as cassava, millet, sweet potato, certain beans, and members of the cabbage family, contain substances known as goitrogens that interfere with thyroid hormone synthesis; these substances, which are destroyed by cooking, can be a significant factor in persons with coexisting iodine deficiency who rely on goitrogenic foods as staples. Since a strategy of universal iodization of salt was adopted in 1993, there has been remarkable progress in improving iodine status worldwide. Nonetheless, millions of people living in iodine-deficient areas, primarily in Central Africa, Southeast and Central Asia, and even in central and eastern Europe, remain at risk.

ZINC

A constituent of numerous enzymes, zinc plays a structural role in proteins and regulates gene expression. Zinc deficiency in humans was first reported in the 1960s in Egypt and Iran, where children and adolescent boys with stunted growth and undeveloped genitalia responded to treatment with zinc. Deficiency of the mineral was attributed to the regional diet, which was low in meat and high in legumes, unleavened breads, and whole-grain foods that contain fibre, phytic acid, and other factors that inhibit zinc absorption. Also contributing to zinc deficiency was the practice of clay eating, which interferes with the absorption of zinc, iron, and other minerals. Severe zinc deficiency has also been described in patients fed intravenous solutions inadequate in zinc and in the inherited zinc-responsive syndrome known as acrodermatitis enteropathica. Symptoms of zinc deficiency may include skin lesions, diarrhea, increased susceptibility to infections, night blindness, reduced taste and smell acuity, poor appetite, hair loss, slow wound healing, low sperm count, and impotence. Zinc is highest in protein-rich foods, especially red meat and shellfish, and zinc status may be low in protein-energy malnutrition. Even in developed countries, young children, pregnant women, the elderly, strict vegetarians, people with alcoholism, and those with malabsorption syndromes are vulnerable to zinc deficiency.

CALCIUM

Almost all the calcium in the body is in the bones and teeth, the skeleton serving as a reservoir for calcium needed in the blood and elsewhere. During childhood and adolescence, adequate calcium intake is critical for bone growth and calcification. A low calcium intake during childhood, and especially during the adolescent growth spurt, may predispose one to osteoporosis, a disease characterized by reduced bone mass, later in life. As bones lose density, they become fragile and unable to withstand ordinary strains; the resulting fractures, particularly of the hip, may cause incapacitation and even death. Osteoporosis is particularly common in postmenopausal women in industrial societies. Not a calcium-deficiency disease per se, osteoporosis is strongly influenced by heredity; risk of the disease can be lessened by ensuring adequate calcium intake throughout life and engaging in regular weight-bearing exercise. Sufficient calcium intake in the immediate postmenopausal years does appear to slow bone loss, although not to the same extent as do bone-conserving drugs.

FLUORIDE

Fluoride also contributes to the mineralization of bones and teeth and protects against tooth decay. Epidemiological studies in the United States in the 1930s and 1940s revealed an inverse relationship between the natural fluoride content of waters and the rate of dental caries. In areas where fluoride levels in the drinking water are low, prescription fluoride supplements are recommended for children older than six months of age; dentists also may apply fluoride rinses or gels periodically to their patients' teeth. Fluoridated toothpastes are an important source of fluoride for children and also for adults, who continue to benefit from fluoride intake.

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SODIUM

Sodium is usually provided in ample amounts by food, even without added table salt (sodium chloride). Furthermore, the body's sodium-conservation mechanisms are highly developed, and thus sodium deficiency is rare, even for those on low-sodium diets. Sodium depletion may occur during prolonged heavy sweating, vomiting, or diarrhea or in the case of kidney disease. Symptoms of hyponatremia, or low blood sodium, include muscle cramps, nausea, dizziness, weakness, and eventually shock and coma. After prolonged high-intensity exertion in the heat, sodium balance can be restored by drinking beverages containing sodium and glucose (so-called sports drinks) and by eating salted food. Drinking a litre of water containing two millilitres (one-third teaspoon) of table salt also should suffice.

Chloride is lost from the body under conditions that parallel those of sodium loss. Severe chloride depletion results in a condition known as metabolic alkalosis (excess alkalinity in body fluids).

POTTASSIUM

Potassium is widely distributed in foods and is rarely deficient in the diet. However, some diuretics used in the treatment of hypertension deplete potassium. The mineral is also lost during sustained vomiting or diarrhea or with chronic use of laxatives. Symptoms of potassium deficiency include weakness, loss of appetite, muscle cramps, and confusion. Severe hypokalemia (low blood potassium) may result in cardiac arrhythmias. Potassium-rich foods, such as bananas or oranges, can help replace losses of the mineral, as can potassium chloride supplements, which should be taken only under medical supervision.

WATER DEFICIENCY - DEHYDRATION

Water is the largest component of the body, accounting for more than half of body weight. To replace fluid losses, adults generally need to consume 2 to 4 litres of fluid daily in cool climates, depending on degree of activity, and from 8 to 16 litres a day in very hot climates. Dehydration may develop if water consumption fails to satisfy thirst; if the thirst mechanism is not functioning properly, as during intense physical exercise; or if there is excessive fluid loss, as with diarrhea or vomiting. By the time thirst is apparent, there is already some degree of dehydration, which is defined as loss of fluid amounting to at least 1 to 2 percent of body weight. Symptoms can progress quickly if not corrected: dry mouth, sunken eyes, poor skin turgor, cold hands and feet, weak and rapid pulse, rapid and shallow breathing, confusion, exhaustion, and coma. Loss of fluid constituting more than 10 percent of body weight may be fatal. The elderly (whose thirst sensation may be dulled), people who are ill, and those flying in airplanes are especially vulnerable to dehydration. Infants and children with chronic undernutrition who develop gastroenteritis may become severely dehydrated from diarrhea or vomiting. Treatment is with an intravenous or oral solution of glucose and salts.

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10. CONCEPT OF HUMAN GROWTH AND DEVELOPMENT

CONCEPT OF HUMAN GROWTH AND DEVELOPMENT

In day to day parlance the terms "growth" and "development" are used interchangeably. However, the two terms are conceptually different, though both denote the dynamic aspects of life process. The dynamic aspect of life process starts with the union of sperm and egg. The zygote thus formed after fertilization undergoes a series of changes which include the multiplication of cells, the expansion of the cells, structural and functional differentiation of the cells, maturation and acquisition of a variety of new functions. A close look at these various aspects of life process includes both quantitative and qualitative changes. The quantitative change constitutes growth and the qualitative change constitutes development.

The term "growth" denotes the net increase in the size of tissues and the multiplication of the cells. It also includes the expansion in the size of cells. On the other hand, the term "development" includes processes involving functional specialization, acquisition of a variety of skills, process of maturation, etc. Like the emergence of a butterfly from a cocoon, qualitative change is marked by the appearance of new phenomena that could not have been predicted from earlier functioning. Speech is one such phenomenon.

Human development is the scientific study of how people change and how they stay the same over time. In some ways, people show an underlying continuity or consistency, from one time of life to another. In other ways they change. This study basically focuses on an analysis of the following:

i) How people change throughout life?

ii) What people need to develop normally?

iii) How people react to the many influences upon and within them?

iv) How people can best fulfill their potential as individuals and as species?

The study of human development draws on many disciplines, including psychology, sociology, anthropology, biology, education and medicine.

Aspects of Growth and Development: Human development is a complex phenomenon. One reason for such complexity is that growth and development occur in different aspects of the self. There are three aspects of Growth and Development - physical, intellectual and personality and social. Each aspect of development affects the other.

1. **Physical Development:** Changes in the body, the brain, sensory capacities are all part of physical development. They exert a major influence on the intellect and personality.

2. **Intellectual Development:** Changes in the mental abilities - such as learning, memory, reasoning, thinking and language - are aspects of intelligence development. These changes are closely related to motor and emotional development.

3. **Personality and Social Development:** Personality - the unique way in which each person deals with the world, expresses emotions, and so on and social development - relationship with others - affect both the cognitive aspects and the physical aspects of functioning.

Individual Differences in Growth and Development: Although people typically proceed through the same general sequence of development, there is a wide range of individual differences in the timing and expression of developmental changes. The range of individual differences increases as people grow older. Normal children pass the same milestones in development at nearly the same ages. Later in life, experiences and environment exert more influence.

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Not only rates, but also the results of development vary. People differ in height, weight and body build, in health and energy levels, in comprehension of complex ideas, and in emotional reactions.

Influences on Development: Development and Growth are subject to many influences. Some influences are purely individual and others are common to an entire group. People's own behavior and life style also influence their development. All these factors are discussed in detail later in this chapter.

Influences on development can be both internal and external. Internal influences on development originate with heredity - the inborn genetic endowment that people receive from their parents. External influences, or environmental influences, come from people's experiences with the world outside the self.

Rate of Growth: Rate of growth is also called growth velocity and it can be defined as the increment in growth at a period of time. Measurement of growth velocity helps in early assessment of retarding factors of growth and also the prediction of ultimate growth.

Catch-up Growth: It is also called as **compensatory growth**. After illness or starvation, which is a period characterized by slow growth, there has been found a tendency in the young subjects to bridge the deficit as soon as possible and catch-up with the original growth curve. This is known as catch-up growth.

Catch-up growth may be complete or incomplete. It depends on the severity of the stress and the period of stress.

Principles of Growth: Growth and Development are not haphazard or idiosyncratic. Rather, it progresses along logical lines that are similar for all human beings. Three guiding principles are at work in growth and development both before and after birth.

1. **Top-to-Bottom Development:** This principle, also called the Cephalocaudal principle ("head to tail", from Greek and Latin roots), dictates that development proceeds from the head to the lower parts of the body. An embryo's head, brain and eyes develop first; the head of a two month old embryo is half the length of the entire body. By the time of birth, the head is only one-fourth the length of the body but is still disproportionately large; it becomes less so as the child grows. Further more, infants learn to use the upper parts of the body before the lower parts. Babies can see objects before they can control the trunk, and they can use their hands to grasp long before they can walk.

2. **Inner-to-Outer Development:** According to this principle, which is also called Proximodistal principle ("near to far", from Latin roots), development proceeds from the central part of the body to outer parts. The embryonic trunk and head develop before the limbs, and the arms and legs before the fingers and toes. Babies first develop the ability to use their upper arms and upper legs (which are closest to central axis), then their forearms and forelegs, then their hands and feet, and finally their fingers and toes.

3. **Simple-to-Complex Development:** The third rule of development states that in acquiring all practical skills, we progress from the simple to the complex. Children could sit with support before they can sit alone.

Some gerontologists distinguish between primary ageing - a gradual, inevitable process of bodily deterioration that begins early in life and continues through the years; and secondary ageing - the results of disease, abuse and disuse - factors that are often avoidable and under people's control. Older people may not be able to stop their reflexes from slowing down or their hearing from becoming less acute; but by eating sensibly and keeping physically fit; many can and do stop the secondary effects of ageing.

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STAGES OF HUMAN GROWTH

PRENATAL DEVELOPMENT

Prenatal development proceeds according to genetic instructions from a single cell to an extremely complex being. This development before birth is called Gestation. It takes place in three stages: Germinal, Embryonic and Fetal. Some highlights of each stage are discussed.

1. **Germinal Stage (Fertilization to 2 Weeks):** During the Germinal Stage, the organism divides, becomes more complex, and is implanted in the wall of the Uterus. Within 36 hours of fertilization, the single cell zygote enters a period of rapid cell division. After 72 hours of fertilization, it has divided into 64 cells. This cell division continues until the original cell has become 800 billion or more cells that make up the adult human body.

   While the fertilized egg is dividing, it is also making its way down the fallopian tube to the uterus, a journey of 3 or 4 days. Meanwhile, its form has changed to a fluid filled sphere, a blastocyst, which then floats freely in the uterus for a day or two. Some cells around the edge of the blastocyst cluster on one side to form the embryonic disk, a thickened cell mass from which the baby will develop. This mass is already differentiating into two layers. The upper layer, the ectoderm, will become the nails, hair, teeth, sensory organs, the outer layer of skin, and the nervous system, including the brain and the spinal cord. The lower layer, the endoderm, will develop into the digestive system, liver, pancreas, salivary glands, and respiratory system. Later, a middle layer, the mesoderm, will develop and differentiate into the inner layer of skin, muscles, skeleton, and excretory and circulatory system.

   During the germinal stage, other parts of the blastocyst develop into the nurturing and protective organs; the placenta, the umbilical cord, the amniotic sac. The placenta, which has several important functions, is connected to the embryo by the umbilical cord, through which it delivers oxygen and nourishment to the embryo and removes its body wastes. The placenta also helps to combat internal infection and protects the unborn child from various diseases. It produces the hormones that support pregnancy, prepare the mother's breasts for lactation, and eventually stimulate the uterine contractions that will expel the baby from her body. The amniotic sac, a fluid filled membrane, encases the baby, protecting it and giving it room to move.

   The Trophoblast, the outer cell layer of the blastocyst, produces tiny threadlike structures that penetrate the lining of the uterine wall. In this way, the blastocyst burrows into the wall of the uterus until it is implanted in a nesting place where it will receive nourishment from the mother's body. Upon implantation, the blastocyst has about 150 cells; when it is fully implanted in the uterus, it is an embryo.

2. **Embryonic Stage (2 to 8-12 Weeks):** During the embryonic stage - the second stage of gestation - the major body systems (respiratory, alimentary, and nervous) and organs develop.

   Because of its rapid growth and development, the embryo is very vulnerable to environmental influences. Almost all the developmental birth defects occur during the critical first three month period, or trimester of pregnancy.

3. **Fetal Stage (8-12 Weeks to Birth) :** With the appearance of the first bone cells at about 8 weeks, the embryo begins to become a fetus, and by 12 weeks it is fully in the fetal stage, the final stage of gestation. From now until birth, the finishing touches are put on the various body parts and the body changes in form and eventually grows about 20 times in length.

   The fetus is far from being a passive passenger in its mother's womb. It kicks, turns, flexes its body, somersaults, swallows, hiccups and sucks its thumb. It responds to both sound and vibrations, showing that it can hear and feel.

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Even within the womb, each of us is unique. Fetuses' activity varies in amount and kind, and their heart rates vary in regularity and speed. Some of these patterns persist to adulthood, supporting the notion of "inborn temperament".

Birth: Birth is both a beginning and an end; the climax of all that has happened from the moment of fertilization through nine months of growth in the womb. The uterine contractions that expel the fetus begin as mild tightening of the uterus, each lasting 15 to 25 seconds.

Stages of Child Birth:

Child birth, or labor, takes place in three overlapping stages. The first stage, which is the longest, lasts an average of 12 to 24 hours for a woman having her first child. During this stage, uterine contractions cause the cervix to widen until it becomes large enough for the baby's head to pass through, a process called dilation. At the beginning of this stage, the contractions occur about every 8 to 10 minutes and last 30 seconds. Toward the end of the labor they may come every 2 minutes and last 60 to 90 seconds.

The second stage, which typically lasts about one and a half hour begins when the baby's head begins to move through the cervix into the vaginal canal, and it ends when the baby emerges completely from the mother's body. At the end of this stage the baby is born. The umbilical cord, which is still attached to the placenta, is cut and clamped.

During the third stage, which lasts only a few minutes, what is left of the umbilical cord and the placenta are expelled.

With the birth of the child, the prenatal stage of development is complete. This is followed by stage of infancy.

INFANCY

The time between birth to an age of 3 years falls under the stage of Infancy.

The Neonate: The first four weeks of life are the neonatal period, a time of transition from intrauterine life - when a fetus is supported by a mother's body - to an independent existence.

Physical Characteristics: An average neonate, or newborn, is about 20 inches long and weighs about 7 1/2 pounds. The size at birth is related to such factors as race, sex, maternal nutrition etc.

In their first few days, neonate loses as much as 10% of their body weight, primarily because of a loss of fluids. They begin to gain weight again at about the fifth day and are generally back to birth weight by the tenth to the fourteenth day.

The neonate's head may be long and misshapen because of "molding" that eased its passage through the mother's pelvis. This temporary molding was possible because the body's skull bones are not yet fused; they will not be completely joined for 18 months.

Newborns are quite pale and have a pinkish cast because of the thinness of skin; this barely covers the blood flowing through their capillaries.

Circulatory System: Before birth, mother and baby have independent circulatory systems and separate heartbeats; but the fetus's blood is cleansed through the umbilical cord, which carries blood to and from placenta. At birth, the baby's own system must takeover. The neonate's heartbeat is still fast and irregular, and blood pressure does not stabilize until about the 10th day.

Respiratory System: The fetus gets oxygen through the umbilical cord, which also carries away carbon dioxide. The newborn, which needs much more oxygen, must now get it independently. Most infants start to breathe as soon as they emerge into the air.

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Gastrointestinal System: The fetus relies on the umbilical cord to bring food and carry body wastes away. The newborn has a strong suckling reflex to take in milk and has gastrointestinal secretions to digest it.

Temperature Regulation: The layers of fat that develop during the last months of fetal life enable healthy full term infants to keep their body temperature constant despite changes in air temperature. They also maintain body temperature by increasing the activity.

Growth: Growth is faster during the first three years and especially during the first few months - than it even will be again. Children's early physical growth and muscular development make possible the rapid reflex/motor advancements of this period.

At five months, the typical baby's birth weight doubles. By one year, babies weigh three times their birth weight. During the second year, this rapid growth tapers off; the child gains 5 or 6 pounds and by the second birthday weighs about four times his or her birth weight. During the third year, the gain is even less about 4 to 5 pounds.

The same pattern holds true for height, which increases by about 10-12 inches during the first year, making the typical one year old about 30 inches tall. The average 2 year old has grown about 6 inches and is 3 feet tall; another 3 to 4 inches will be added in the third year.

As young children grow in size, their shape also changes. The rest of the body catches up with the head, which becomes proportionately smaller until full adult height become leaner; the 3 year old is slender.

Teething usually begins around 3 or 4 months, but the first tooth may not arrive until sometime between 5 and 9 months of age, by age 2 1/2, they have a mouthful - 20.

Motor Control: The eye coordination starts developing between 2-4 weeks and by 3-4 months; infants follow the objects with steady movement of eyes.

Hand to mouth coordination develops slightly later; the baby is able to feed himself only after 18 months. Hard skills begin to develop from 2 years when he could perform crude skills.

The infants learn to control body in the sitting position from age of 5 months. From the age of 9 months, the infant makes early stepping movements.

Language development starts by one month, when the infant attends to curious sounds. By nine months he produces monosyllable sounds. By ten months he understands speech and responds to it. True speech develops between 1-2 years.

CHILDHOOD

The stage of childhood falls between the years 3 to 12. We shall further classify this stage into Early Childhood (3-6 years) and Middle Childhood (6-12 years).

1. **EARLY CHILDHOOD:** At this stage of human development the physical, intellectual, emotional and social factors continue to intertwine to make each person unique.

   Physical Growth and Change: Physical changes may be less obvious during early childhood than during the first 3 years of life. However, they are nonetheless important, and they make possible dramatic advances in motor skills and intellectual development.

   At about age 3, boys and girls begin to lose their chubbiness and begin to take on the slender, athletic appearance of children. As children's abdominal muscles develop, their potbellies slim down. The trunk, arms and legs all grow longer. The head is still relatively large, but other parts of the body continue to catch-up as body proportions steadily become more adult.

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Within that overall pattern, children show a wide range of individual and sex-related differences. Boys tend to have more muscles per pound of body weight than girls, while girls have more fatty tissue. Boy's slight edge in height and weight normally continue until puberty.

Structural and Systemic Changes: The changes in children's appearance reflect important internal development. Muscular and skeletal growth progresses, making children stronger, cartilage turns harder, giving the child a firmer shape and protecting the internal organs. These changes, coordinated by the maturing brain and nervous system, allow a proliferation of both large and small muscle motor skills. In addition, the increased capacities of the respiratory and circulatory systems improve stamina and, along with the developing immune system, keep children healthier.

By the age of 3, all the primary teeth are in place and so children can chew anything they want to. The permanent teeth, which will begin to appear before age 6, are developing; therefore, if thumb sucking persists past the age of 5, it can affect how evenly the teeth come in.

Motor Skills: With the child's stronger bones and muscles, greater lung power, and improved coordination between senses, limbs and central nervous system, it can do more and more of the things it wants to do. Between the ages of 3 and 6, children continue to make important advances in motor development.

Large Muscle Coordination: At 3, children can walk a straight line and stand on one foot but only for about a second. At 4, they could hop on one foot, and could catch a ball bounced to them, with hardly any misses. At 5, they could jump nearly 3 feet.

Such motor skills - advanced far beyond the reflexes of infancy - are required for sports, dancing and other activities that begin during middle childhood and may last a life time.

Small Muscle and Eye-Hand Coordination: Children at age 3 can draw a nearly straight line and a recognizable circle. At 4, they can cut a line with scissors, draw a person, and make designs and crude letters. With their small muscles under control, children are able to tend to *more of* their own personal needs and so have a sense of competence and independence. By age 2 or 3 they use one hand more than the other.

Development of Language: At the beginning of early childhood, children can give and follow commands that include more than one step. And they can name familiar things like pets, body parts, and people. Their linguistic skills progress rapidly through early childhood. Speech becomes more adult once children pass the age of 3. Children over 3 use plurals and the past tense, and they know the difference between "I", "You" and "We".

Between ages 5 and 6, children use longer and more complex sentences. They can define simple words, and they know some opposites. They use more conjunctions, prepositions, and articles.

Between 6 and 7 years of age, children's speech becomes quite sophisticated. They now speak in grammatically correct compound and complex sentences, and they use all parts of speech.

2. **MIDDLE CHILDHOOD:** Compared with the pace of physical and intellectual development in early childhood, development between the ages of 6 and 12 may seem to be low. Physical growth slows down considerably except for the growth spurt toward the end of this period and while motor abilities continue to improve, changes are less dramatic than they were earlier. But development at these ages is still highly significant. Intellectual growth is substantial. If the early childhood is made of small children, middle childhood is composed of almost adults.

Physical Development: Both boys and girls gain an average of 7 pounds and 2 to 3 inches a year until the adolescent growth spurt, which begins at about 10 for girls. Then, girls are on average taller and heavier than boys until the boys begin their spurt at about age 12 or 13 and overtake the girls.

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Individual children vary widely, however - so widely that if a child who was of exactly average height at his seventh birthday grew not at all for two years; he would still be just within the normal limits of height attained at age nine. Also, growth rates vary with race, national origin and socio-economic level. Given the wide variance in size during middle childhood, we have to be careful about assessing children's health or identifying possible abnormalities in physical growth.

Motor Development: Middle childhood children are physically active and their motor abilities increase with age. The boys tend to run faster, jump higher and show more strength than girls. After age 13, the gap between sexes widens, boys improve and girls stay the same or decline.

Development of Language: Language develops quickly in middle childhood. Children can understand and interpret communications better, and they are better able to make themselves understood. Children develop increasingly complex understanding of syntax by 9 years.

During childhood, one can observe the disorganized behavior gradually changing into an organized communication skills.

ADOLESCENCE

A Developmental Transition: Adolescence is generally considered as beginning at puberty, the process that leads to sexual maturity, when a person is able to reproduce. Puberty is a part of a long and complex process that begins even before birth. The biological changes that signal the end of childhood produce rapid growth in height and weight, changes in body proportions and form, and attainment of sexual maturity. But adolescence is also a social and emotional process. It has been said that "adolescence begins in biology and ends in culture".

Physical Development: The biological changes that signal the end of childhood include the adolescent growth spurt, the beginning of menstruation for girls, the presence of sperm in the urine of males, the maturation of organs involved in reproduction, and development of secondary sex characteristics.

During puberty, the reproductive functions mature, the sex organs enlarge, and secondary sex characters appear. The process takes about 4 years and begins about 2 years earlier for girls than for boys. Girls, on an average, begin to show pubertal change at 9 or 10 years of age, achieving sexual maturation by 13 or 14. Normal girls, however, may show the first signs as early as age 7 or as late as 14. The average age for boys' entry into puberty is 12, with sexual maturity coming at age 14. But normal boys may begin to show changes from ages 9 to 16.

The physical changes of adolescence unfold in a sequence that is much more consistent than their actual timing, though even this order varies somewhat from one person to another. Some people move through puberty very quickly, while for the others the process takes much longer.

Puberty occurs in response to changes in the body's hormone system, which is triggered by some physiological signal, possibly related to a critical weight level. Whatever the signal is, its response in a girl is for her ovaries to sharply step up their production of the female hormone estrogen; and in a boy, for his testes to increase the manufacture of androgens, particularly testosterone. Both boys and girls have both types of hormones, but girls have higher level of estrogen and boys have a higher level of testosterone. As early as age 7, the levels of these sex hormones begin to rise, setting the events of puberty in motion. Estrogen stimulates growth of the female genitals and development of breasts; androgens stimulate the growth of the male genitals and body hair.

Hormones are also closely associated with emotions, specifically with aggression in boys and with both aggression and depression in girls.

The Adolescent Growth Spurt: An early sign of maturation is the `Adolescent Growth Spurt', a dramatic increase in height and weight. It generally begins to appear in girls between ages 9 ½ and 14 ½ and in boys between ages 10 ½ and 16. It typically lasts about 2 years. Soon after the spurt ends, the young

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person reaches sexual maturity. Most girls reach their adult height by age 14 or 15, and most boys by age 18.

Before the growth spurt, boys are typically only about 2 percent taller than girls. Since girls growth spurt usually occurs earlier than that of boys, there is a period of several years when girls are taller, heavier, and stronger. After the growth spurt, boys are larger again, now by about 8 percent. The growth spurt in boys is more intense, and its later appearance allows for an extra period of growth, since growth goes on at a faster rate before puberty.

Boys and girls grow differently during adolescence. A boy becomes larger overall, his shoulders are wider, his legs are longer relative to his trunk, and his fore arms are longer relative to both his upper arms and his height. A girl's pelvis widens during adolescence to make child bearing easier, and layers of fat are laid down just under the skin, giving her a more rounded appearance.

In both sexes, the adolescent growth spurt affects practically all skeletal and muscular dimensions. The changes, which are greater in boys than in girls, follow their own time tables, so that parts of body may be out of proportion for a while. The result is the familiar teenage awkwardness that accompanies unbalanced, accelerated growth.

Primary Sex Characteristics: The primary sex characteristics are the organs necessary for reproduction. In the female, the sex organs are the ovaries, uterus and vagina; in the male, the testes, prostrate gland, penis, and seminal vesicles. The gradual enlargement of these body parts occurs during puberty, leading to sexual maturation.

The principle sign of sexual maturity in girls is menstruation. In boys, the principal sign is the presence of sperm in the urine. Both the onset of menstruation and the first appearance of sperm in the urine are highly variable. Another sign of puberty in a boy is the occurrence of an ejaculation of semen while he is asleep, known as nocturnal ejaculation (wet dream).

Secondary Sexual Characters: The secondary sex characteristics are physiological signs of sexual maturation that do not directly involve the sex organs. They include the breasts of females and the broad shoulders of males. Other secondary sex characteristics involve changes in the voice, skin texture, and body hair. The timing of these signs is variable, but the sequence is fairly consistent.

The first sign of puberty for girls is usually the budding of the breasts. The nipples enlarge and protrude; the areolae, the pigmented areas surrounding the nipples enlarge.

Various forms of hair growth, including pubic hair and axillary (arm pit) hair, also signal maturation.

The skin of adolescent boys and girls becomes coarser and oily, and the increased activity of sebaceous glands causes outbreaks of pimples and blackheads. Acne is more common in boys than in girls and seems to be related to increased amounts of the male hormone testosterone. The voices of both boys and girls deepen, partly in response to growth of the pharynx and partly - especially in boys - in response to the production of male hormones.

The most dramatic sign of a girl's sexual maturity is Menarche - the first menstruation, or monthly shedding of tissue from the lining of the womb. Menarche occurs fairly late in the sequence of female development. On an average, a girl first menstruates at the age of 12 ½, after her growth spurt has slowed down.

MATURITY

Maturity is a long process which includes young adulthood (20 - 40 years) and middle adulthood or middle age (40 - 65 years).

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1. **YOUNG ADULTHOOD:** During this stage, people make many of the decisions that will affect the rest of their lives - their health, their happiness, and their success. The interactions among the various aspects of development - physical, intellectual and social - are striking.

   Physical Development: A typical young adult is a fine physical specimen. Strength, energy and endurance are now at their peak. From the middle twenties, when most body functions are fully developed, until about age 50, declines in physical capabilities are usually so gradual that they are hardly noticed.

   The 20 year old tend to be taller. Between ages 30 and 45, height is stable, and then it begins to decline.

   The peak of muscular strength occurs sometime around 25 - 30 years of age; it is followed by 10% loss of strength between ages 30 and 60. Manual dexterity is most efficient in young adults; agility of finger and hand movements begins to lessen after the mid-thirties.

   The senses are also sharpest during young adulthood. Visual activity is keenest at about age 20 and does not begin to decline until about age 40. A gradual hearing loss typically begins before age 25; after age 25, the loss becomes more apparent, especially for higher pitched sounds. Taste, smell and sensitivity to pain and temperature generally show no diminution until about age 45 to age 50 or later.

2. **MIDDLE ADULTHOOD:** From young adulthood through the middle years, biological changes generally take place so gradually that they are hardly noticed - until one day a 45 year old man realizes that he cannot read the telephone directory without eye glasses.

   Physical functioning and health are usually still good, though not at the peak level of young adulthood. Most people take changes in reproductive and sexual capacities - menopause and the male climacteric - in stride, and some experience a kind of sexual renaissance.

   Physical Changes: Although changes in sensory and motor capabilities during mid-life are real and affect people's concept of themselves and their interaction with others, these changes are usually fairly small, and most middle aged people compensate well for them.

   Vision, Hearing, Taste and Smell: Throughout life, the lens of the eye becomes progressively less elastic, so that its ability to focus is diminished; this process is usually noticed for the first time in middle age. Many people now need reading glasses for presbyopia, the far sightedness associated with aging. Bifocals - eyeglasses in which lenses for reading are combined with lenses for distant vision - help people make the adjustment between near and far objects. Middle aged people also experience a slight loss in sharpness of vision; and because the pupil of the eye tends to become smaller, they need about 1/3rd more brightness to compensate for the loss of light reaching the retina. Nearsightedness, though, tends to level off in these years.

   There is also a gradual hearing loss during middle age, especially with regard to more high-pitched sounds; this condition is known as presbycusis. After about age 55, hearing loss is greater for men than for women. However, most hearing loss during these years is not even noticed, since it is limited to levels of sound that are unimportant to behavior.

   Taste sensitivity begins to decline at about age 50, particularly the ability to discriminate "finer nuances of taste". Since the taste buds become less sensitive, foods that may be quite flavorful to a younger person may seem bland to a middle aged person. Sensitivity to smell holds up well; it is one of the last senses to decline.

   Strength, Coordination and Reaction Time: Although strength and coordination decline gradually during the middle years, the loss is so small that most people barely notice it. A 10% reduction in

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physical strength from its peak during twenties does not mean much to people who rarely if ever exert their full strength in daily life.

People who lead sedentary lives lose muscle tone and energy, and so they become even less inclined to exert themselves physically. A sedentary life style has recently emerged as the major co-relate to deaths from heart attacks. People who become active early in life reap the benefits of more stamina and more resilience after age 60.

Reaction time slows down by about 20% on the average, between ages 20 and 60. Complex motor skills, which increase during the childhood and youth, gradually decline after people have achieved full growth. But the decline does not necessarily result in poorer performance.

Physiological Changes: The most common physiological changes of midlife include a diminished ability to pump blood; reduced kidney functioning; less enzyme secretion in the gastrointestinal tract, leading to indigestion and constipation; weakening of the diaphragm; and in male, enlargement of the prostrate gland, which may cause urinary or sexual problems. Some of these changes are a direct result of aging. Still, behavioral factors and lifestyle, dating from youth, often affect their timing and extent. People age at different rates and the decline of the body systems is gradual.

Reproductive and Sexual Capacity: One fundamental change of middle age - the decline of reproductive capacity - affects men and women differently. Women's ability to bear children comes to an end, and although men can continue to father children, they begin to experience lessened fertility and, in some cases, a decrease in potency.

Menopause: The biological event of menopause occurs when a woman stops ovulating and menstruating and can no longer bear children. Menopause is generally considered to have occurred one year after the last menstrual cycle. This typically happens between ages 45 to 55, at an average age of 51.

The Male Climacteric: Although men can continue to father children till late in life, some middle aged men experience a decrease in fertility and frequency and increase in impotence.

The male climacteric is a period of physiological, emotional and psychological change involving a man's reproductive system and other body systems. It generally begins about ten years later than a woman's climacteric.

SENESCENCE

The onset of senescence - the period of lifespan marked by decline in body functioning associated with aging - varies greatly. One 80 year old man can hear every word of a whispered conversation, while another has trouble hearing words shouted at him.

Physical Changes: Although sensory and psycho-motor abilities decline with age, there is a great degree of individual variation. Losses of vision or hearing have particularly strong psychological consequences as they deprive people of their activities, social life and independence.

Vision: Older adults report that they have trouble doing a variety of activities dependent on vision. The problems regarding vision stem from deficits in five areas : speed in processing what they see, near vision, light sensitivity, dynamic vision and visual search.

Farsightedness usually stabilizes at about age 60, and with the help of lenses, older people can see well. After 65, however, serious visual problems that affect daily life are all too common. Many older adults have trouble perceiving depth or color. About 16% develop cataract, cloudy areas in lens of eye that prevent light from passing through and thus cause blurred vision.

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Glaucoma, another frequent cause of blindness, occurs when fluid pressure builds up, damaging the eye internally.

Hearing: Hearing loss is very common late in life; about 3 out of 10 people between ages 65 and 74 and about half of those between 75 and 79 have it to some degree. Because older people tend to have trouble hearing high-frequency sounds, they often cannot hear what other people are saying, especially when there is a competing noise or buzz.

Taste and Smell: When older people complain that their food does not taste good anymore, it may be because they have fewer taste buds in tongue and also because the olfactory bulb - the organ in brain that is responsible for the sense of smell - has withered.

Sensitivity to Cold and Heat: An older person's body adjusts more slowly to cold and becomes chilled more easily than that of a younger person. Exposure to outdoor cold and to poorly heated interiors may lower body temperature - a serious risk for the aged. Older people cannot cope as well with heat, either, and they often have trouble working in warm temperatures.

Strength, Coordination and Heat: A general slowing down - which affects the quality of responses as well as response time - may result from environmental deprivation and depression, and from neurological changes. Because reflex responses are slowed, loss of bladder and bowel control sometimes occurs.

It takes longer for them to assess the environment, take in all the pertinent information, and make decisions. Slowed information processing shows up everywhere in their lives. The combination of slower information processing, slower reaction time and less efficient sensor motor coordination makes independence more risky.

Other Physical Changes: Many of the changes associated with aging are readily apparent to even the most casual observer - and of course to the elderly themselves. The skin becomes paler, taking on a parchment like texture and losing elasticity. As some subcutaneous fat and muscle disappear, the inelastic skin tends to hand in folds and wrinkles. The hair becomes thinner, what is left turns white, and it sprouts in new places.

People may shrink in size as the disks between their spinal vertebrae atrophy. Osteoporosis, a thinning of the bone that affects some women after menopause, may cause a hump at the back of neck. The chemical composition of bones changes causing a greater chance of fractures.

All the body systems and organs are more susceptible to disease, but the most serious change affects the heart. After age 55, its rhythm becomes slower and more irregular; deposits of fat accumulate around it and interfere with its functioning and blood pressure rises. The digestive system remains relatively efficient : the smooth muscles of the internal organs continue to operate well, and the liver and gall bladder holds up well when obesity is present, it affects the circulatory system, the kidneys, and sugar metabolism; it contributes to degenerative disorders and tends to shorten life.

Reserve Capacity: The human body has the equivalent of money in the bank for a rainy day. Normally, people do not use their organs and body systems to the limit; but extra capacity is available for extraordinary circumstances. This backup capacity, which lets body systems function in times of stress, is called reserve capacity; it allows each organ to put forth 4 - 10 times as much effort as usual. Reserve capacity helps to preserve homeostasis, the maintenance of vital functions within their optimum range.

With age, however, reserve levels drop. Although the decline is not usually noticeable in everyday life, older people cannot respond to the physical demands of stressful situations as quickly or efficiently as they used to. People need reserve capacity just to survive as pedestrians; but because older people cannot call upon fast reflexes, vigorous heart action, and rapidly responding muscles to get out of harm's way, they are more likely to be victims of accidents. In general, then, as reserve capacity diminishes, people may become less able to care for themselves and more dependent on others.

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FACTORS AFFECTING GROWTH AND DEVELOPMENT

GENETIC FACTORS

Genetic factors control growth and development right from the formation of an egg or a sperm. These genetic factors control the various stages of growth and development through the life of an individual. The genetic factors and their influence on different stages are discussed.

We have already observed that the human growth and development follows a coordinated sequence. Different characters appear at different stages of growth. These characters in the organism are controlled by genes. Hence, for a coordinated growth, it is essential for the different genes to be activated at specific time periods. This process requires a regulated chain reaction. This regulated chain reaction is called as **Induction**. The term implies that when one part of the growing embryo is differentiated, it releases substances or "Inducers" for differentiation of another part of the growing embryo. Thus the inducers are responsible for stimulating growth in different parts of the embryo. This leads to a fast organization of the growing embryo.

Inducers are proteins by nature. They act on the cell membranes and thus bring gradual changes in the functional and structural configurations of the cells. These changes are brought about by activating the genes within the cells. The cells of different regions are thus gradually differentiated to form an embryo with different observable regions.

The cell differentiation can only happen when the genes of different cells are activated at specific functional sites. Based on the functions associated with cell differentiation, genes can be classified into two types:

1. **Housekeeping Genes:** These genes synthesize proteins that are required by all types of cells. For example, the proteins required for the formation of cell membrane. These genes, thus, are activated by inducers in all types of cells.
2. **Luxury Genes:** These are related with certain special functions associated with cells differentiations. Hence they are activated in only certain types of cells. For example, the cells which synthesize insulin are activated by inducers responsible for this function.

Cell Division: For any division to occur, the duplication of DNA is essential. Moreover, the synthesis of proteins depends on MRNA. Without such genetic changes, the cells can neither increase in size nor can they be successfully be differentiated. Thus DNA and MRNA control the growth process.

The duplication of DNA is regulated by DNA-polymerase enzyme and MRNA by RNA-polymerase.

This transcription of DNA and RNA is actually controlled by proteins called Transcriptional Factors. They help the DNA or RNA to interact with the concerned - polymerase enzyme. Thus genetic factors control growth.

Apart from this, genes also control the metabolism of body which in turn is responsible for normal growth and development. Certain special genes called Homeotic genes control differentiation of the various organs of the body. They are responsible to give identity to the organs and their internal development and functional specialization.

Role of Heredity: Heredity is a dominant factor controlling growth. It controls growth and development at the following levels:

1. **Phenotype:** The pattern of morphological aspects of children depends on the phenotype of the parents. Thus genes control greatly, though not exclusively, the pattern of growth and development.
2. **Twins:** We have already noted the significance of genetics of twins in the concerned chapter.

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3. **Sex:** The sex of the child is governed exclusively by genetics. Thus even the sex related growth patterns are controlled by genetics.

Apart from this, heredity controls Biorhythms, Dental Eruption and Skeletal maturity.

ENVIRONMENTAL FACTORS

Human beings though depend on Genetics for basic pattern of growth; environment plays a significant role in controlling growth and development. Its influences are seen in following circumstances.

1. **Migration:** Migration of the human population changes the environment of the group. These changes bring about adaptive modifications in the body and thus influence the growth pattern and direction of development.

2. **Climate:** The climatic factors affect the rate of growth. For example, among the Eskimos, the rate of growth is less compared to their African counterparts. In humid and tropical climate, the rate of growth tends to be faster than in cold environment. However, as Tanner (1992) has noted, there is a considerable individual variation among the human beings with respect to this phenomena.

3. **Emotions:** This aspect of social environment plays a significant role in growth and development. An optimal rate of growth is not seen among children coming from orphanages and broken homes. Anxiety, insecurity and lack of emotional support from the family prejudice the regulation of hormones responsible for growth.

Environmental influence is more on people during developmental period during pregnancy and sensitive period after delivery when they are in process of learning. Moreover, environment has more impact on homozygous people than heterozygotes.

BIOCHEMICAL FACTORS

The entire process of human growth and development is largely a part of interplay between various hormones secreted by the endocrine glands. They are very crucial for normal growth and development. Hormones are active for growth in the stages of prenatal, postnatal and adolescent periods.

HORMONES		
PRE-NATAL GROWTH	POST-NATAL GROWTH	ADOLESCENCE
1. Thyroxin 2. Chorionic Gonadotropin (CG)	1. Thyroxin 2. Growth Hormone (GH)	1. Thyroxin 2. GH 3. Leutinizing Hormone (LH) 4. Follicle Stimulating Hormone (FSH)

Thyroxin: From the above table, it is obvious that thyroxin is active in all stages of growth. From the 12th week of Gestation, the human fetus secretes thyroxin. Thyroxin is responsible for:

1. Increase in permeability of cell membrane for absorption of substances which are responsible for growth.
2. Increasing the number of mitochondria that provide energy for growing cells.
3. Activating DNA and RNA for synthesis of proteins.
4. Increasing responsiveness of tissues to other hormones.

Chorionic Gonadotropin: This hormone is responsible for the stimulation of gonads for secreting other hormones like testosterone. This is responsible for gonadal differentiation and secretion of growth factors responsible for normal growth of fetus.

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Growth Hormone: It is secreted by fetal pituitary gland. It is responsible for:

1. Formation of joints of bones.
2. Cell division.
3. Proliferation of cartilaginous tissue.
4. Increase in muscles.
5. Increase in proteins.
6. Fat synthesis.

Reproduction Promoting Hormones: The follicle stimulating hormone and leutinizing hormone are responsible for normal sexual maturity.

FSH: In females it is responsible for development of ovarian follicle, secretion of estrogen and growth of secondary sexual characters, while in males it is responsible for spermatogenesis, growth of testes etc.

LH: In females, LH is responsible for ovulation and in males for secretion of testosterone and secondary sexual characters.

Apart from these, there are also many other biochemical factors controlling growth and development.

NUTRITIONAL FACTORS

Nutrition is considered to be a most important factor affecting growth. Since a proper definition for "optimal growth" is unavailable, the concept of "optimal diet" is less understood. Moreover, the nutritional requirements for individual to individual vary; hence a single dietary regime cannot be prescribed for normal development.

The daily requirement for basal metabolism in the first eighteen months of life averages to about 55 calories / kg. There after, the basal requirement gradually lessens to about 30 calories/kg in an adult. The requirement for activity may vary tremendously per individual at different ages and for the two sexes. In a premature infant the requirement for growth may double as compared to the normal new born.

The distribution of total caloric requirement for boys and girls at different ages is given below: (Watson & Lowrey, 1951).

TOTAL CALORIC REQUIREMENT PER DAY (CAL/KG)				
AGE	**8 WEEKS**	**10 MONTHS**	**4 YEARS**	**ADULT**
Basal Metabolism	55	55	30	25
Specific Dynamic Actions	7	7	6	6
Caloric Loss in Excreta	11	10	8	6
Muscular Activity	17	20	25	10
Additional Energy for Growth	20	12	10	0

Malnutrition may cause serious impairment of growth. Malnutrition refers to effects of an inadequate intake of calories. It may also result from diseases which decrease appetite and assimilation.

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Children suffering from mal-nutrition grow slow. If malnutrition is prolonged for a long time during childhood, the genetic potential for growth is not attained, and the final stature is reduced.

Tempo of growth is the first to be effected by under-nourishment. Adult size is affected even less severe than adult shape. The first five years of a child is a period when he is at a maximum risk from malnutrition.

CULTURAL AND SOCIO-ECONOMIC FACTORS

Socio-economic factors play a significant role in controlling development and growth. In 1972, Tanner has observed that there is a height difference among the children of unskilled laborers and managerial class. There is also a difference in time of maturity. Causes of such differences are discussed below:

1. **Nutritional Deficiency:** Nutritional deficiency among weaker sections may start right from pregnancy. A poor state of economy of the weaker sections deprives a child of a balanced diet. Moreover, illiteracy of the parents may also lead to this problem.

2. **Home Environment:** The home conditions are a causative factor for growth retardation according to Tanner. They reflect intelligence and personality of the parents that provide emotional needs for normal growth and development of children.

3. **Faulty Family Budgeting:** A low economic condition of the family is a fountainhead of all deprivations they suffer. Family budgeting never reflects child centeredness.

4. **Minor Illnesses:** Due to malnutrition, the children are prone to illnesses like cold, measles etc. that have a long-term effect on normal development.

5. **Smoking:** This habit, both in mothers and fathers, has impact on the fetus. They are born stunted and this difference prevails throughout the life.

However, due to the developments happening in the fields of society and economy, a secular trend is now obvious in growth patterns. Improved nutrition, improved and clear environment and practices like exogamy have helped develop this new secular trend.

AGEING AND SENESCENCE

BIOLOGICAL AND CHRONOLOGICAL LONGEVITY

The term ageing simply implies growing older, and changes which are related to age, regardless of when in the life span they occur. Senescence however implies changes which occur during the later years of life span when there is a functional decline.

Strechler in 1962 has defined senescence as "the changes which occur during the post-reproductive period and it results in decreased survival capacity on the part of the individual organism."

Ageing process is an increased liability to die, an increasing loss of vigor, with increasing chronological age, with the passage of time.

Senescence and ageing are gradual processes. They are generally initiated at molecular level. The process is advanced much before the external symptoms appear. This process of ageing follows seven steps:

1. From the time of the zygote itself, organisms accumulate a number of substances, which over a period of time cause damage to bio-molecules like DNA within the cell and bio-molecules outside the cell.

2. The damage to bio-molecules impairs intra-cellular and inter-cellular functioning.

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3. The efficiency of the cell reduces as a result of this functional impairment.
4. The functional impairment may also lead to cell death.
5. Due to these functional impairments, reduction in the cell efficiency and cell death, the tissues, organs and organ systems loose their functional capacities too.
6. This deterioration of tissues and organs lead to the deterioration of the organism itself.
7. Deterioration of the organism leads to an increased probability of death.

Characteristics: Ageing and senescence have four characteristics. They are:

1. The changes associated with ageing increase the probability of organism's death and hence these changes are deleterious.
2. Ageing decreases the animal's capacity to cope with the environment.
3. The changes associated with ageing are cumulative. This implies that though death is sudden, the processes that lead to death are gradual.
4. The processes associated with ageing and senescence are inescapable. They are the fundamental and intrinsic characteristics of life.

THEORIES AND OBSERVATIONS

Many theories have been put forth to explain the causes of ageing. Some of the important ones are explained here. The theories of ageing can be broadly classified into three groups. The first group of theories attempts to explain the process of ageing as result of intracellular processes and their errors in biochemical processes. The second group of theories concentrate on extra cellular processes that lead to ageing and the last group of theories concentrate on numerous regions of body that go into explaining the process of ageing.

CELLULAR THEORIES

Theory of Hayflick Limit

1. In 1965, Leonard Hayflick reported that the human lung fibroblasts divide and their division is limited.
2. The number of cell divisions is roughly related to the age and life span of the species.
3. The longer the life span of a species, the greater is the number of divisions.
4. If cells divide indefinitely, they are abnormal and / or cancerous.
5. Senescence is due to the loss of cell functions that occur before cells reach their maximum division point - the Hayflick Limit.
6. There is a built-in biological clock which decides the cell's capacity to divide the nucleus.
7. Senescence sets in tissues that are more sensitive and then secondarily the other tissues of the body.
8. The potential victim in the case of senescence is the endocrine system.

Theory of Free Radical Reaction

1. Free radicals of oxygen are produced in the cells which cause damage and death of a cell.
2. Certain enzymatic reactions are responsible for generating super oxide radical (O2).

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3. In the presence of water, this super oxide radical is converted into highly reactive hydroperoxyl radical (HO2).
4. This radical (HO2) reacts primarily with unsaturated lipids present in the cell to form the pigment lipofuscin - the "Age Pigment".
5. The free radicals are especially harmful when they react in the cell membrane causing the obstruction of the passage through which diffusion of nutrients and excretion of toxins happen.
6. This leads to cell senescence and cell death.
7. This theory was proved by Lester Packer and James R. Smith.

Theory of DNA Error

1. Errors in DNA replication process may result from time to time and the errors might also crop up as a result of mutations.
2. If the damage to DNA is undetected by DNA repair system or damages are faster than the action of repair system, these errors accumulate.
3. The accumulated DNA errors impair cell functions and impede the synthesis of essential enzymes.
4. Thus cell senescence sets in and this leads to cell death.
5. This theory is supported by Ronald Hart and Richard Setlow.

The Error Catastrophe Theory

1. This concept of "Error Catastrophe" was given by Leslie Orgel and supported by Robin Holliday.
2. Certain types of errors during the process of DNA replication produce a great number of subsequent errors. Thus error in replication would be a "error catastrophe".
3. A mutation in DNA polymerase enzyme gene, which is responsible for DNA replication, would produce a new enzyme which produces further mistakes.
4. Such defective enzymes produced will fill the entire cell with a mosaic of defective proteins.

Theory of Missing Factors

1. This theory was propounded by Dr. M. S. Kanungo, a leading gerontologist of India.
2. Ageing is not due to the malfunctioning of a specific gene or genes but due to non-functioning of genes responsible for maintaining adulthood.
3. The malfunctioning may be due to endogenous factors (mutations) and exogenous factors (nutrition, stress).
4. Thus the reproductive phase of individuals depletes certain factors which are responsible for maintaining adulthood.
5. These missing factors accelerate ageing and process of senescence.

EXTRA CELLULAR THEORIES

Immunological Theory

1. This theory was supported by Burnet and Walford.
2. The efficiency of the immune system (spleen, lymph nodes, bone marrow and thymus) decrease after attainment of adulthood.

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3. The efficiency of thymus gland is especially significant. Its degeneration (after puberty) programs events that lead to ageing of the animal.
4. Immunity can also determine ageing by production of auto-immune responses - the mutations where antibodies are produced against ones own tissues - triggering a fast process of ageing.

Collagen Theory:

1. About 40% of the body protein is composed of collagen which is present in extra cellular spaces.
2. Collagen is made of three polypeptide chains which form a super helix.
3. The three chains are supported by non-covalent bonds.
4. As age advances, the three chains are further stabilized by covalent bonds.
5. Because of this change in the property of collagen, a healthy intracellular environment is not kept and the movement of products is thus obstructed.
6. This leads to accumulation of toxins and under nourishment of cell and thus to cell senescence.

MODERN THEORIES

Programmed Ageing Theory

1. Bodies age according to a normal development pattern built into every organism.
2. This program is only subject to minor modifications.
3. This pattern of ageing is predetermined and inborn.
4. This inborn mechanism of programmed ageing is supposed to be confined to Chromosome-1 or Chromosome-21.

Wear and Tear Theory

1. Body ages because of continuous use - aging is the result of accumulated "insults" to the body.
2. As time passes, when cells grow older, they are less able to repair or replace damaged components.
3. Internal and external stressors aggravate the wearing down process.

HUMAN PHYSIQUE AND SOMATOTYPES

HUMAN PHYSIQUE

An analysis of human populations reveals that the human beings are characterized by different constitutions. Human constitution or physique is consistent. It is consistent in the sense that the various aspects of its structure, function or behavior are almost permanent and do not show any change in the individual as time lapses.

The human physiques can be identified and classified into different somatotypes based on morphological, physiological and behavioral characteristics. This aspect of human physique and its classification into different somatotypes is reserved for the later part of discussion.

The human physique is influenced by numerous factors like environment and genetics. However, it is clear today, after the recent developments in the science of human genetics, that the human physique is largely determined by the genetic constitution of the individual. This is the reason why we have stated earlier that it cannot be subjected to change easily during the life span of the individual.

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SOMATOTYPES

Though we have noted that physiological and behavioral aspects also govern the human physique, morphology has been the most popular level through which human beings are somatotyped. Broadly, four systems of classification are employed for human somatotyping.

Sheldon and others in 1940 devised a method to analyze and quantify human body form and called it as somatotyping. According to them, "Somatotypes are morpho-phenotypic ranges along constantly recognizable characteristics and are the functional end products of the whole genetic and developmental complex".

The Somatotype is aimed at providing a sort of identification tag to an individual and may be regarded as an attempt towards general human taxonomy or classification.

1. **Viola System:** This system is developed by an Italian physician Viola. This system recognizes four somatotypes, based on the measurements and relative indexing of individuals. These somatotypes are :

   A. **Longitype:** Have relatively long limbs compared to the trunk, massive thorax compared to the abdomen, and greater transverse diameters compared to antero-posterior ones.

   B. **Brachitype (Broad type):** Have characters opposite to longitype.

   C. **Normative:** This type constitutes those individuals who fall in between longitype and brachytype categories.

   D. **Mixed Type:** This category includes those individuals, who exhibit characters of different types in different parts of the body, i.e., they may be brachitype in one part, longitype in another and normotype in yet another part of the body.

   The Viola system of somatotyping has however fallen into disuse.

2. **Kretschmer's System:** This system is developed by Kretschmer, a German psychiatrist. This system is based on anthroposcopic inspection. There are three somatotypes according to this system.

   A. **Pyknic Somatotype:** Broad, round and fat, sturdy and stocky.

   B. **Leptosome Somatotype:** Long, thin and linear.

   C. **Athelete Somatotype:** Heavily muscled, large thorax and shoulders and narrow hips.

   The Kretschmer's system is today outmoded. This is because this system classified human beings into fixed, discrete classes though intermediary between these are easily recognized. Moreover, this system lacked any detail compared to Viola's system.

3. **Sheldon's System:** This system recognized three basic components of physique, viz., endomorphy, mesomorphy and ectomorphy. Each individual has varying degrees of development of these three components. The somatotype is always written in three numerals; the first indicating the development of endomorphy, the second the mesomorphy and the third representing ectomorphy. The numerals fall between 1 and 7, the scale chosen by Sheldon to indicate the level of development.

   Sheldon's observations were based on 4000 undergraduate male students in the age range of 16 - 20 years by taking photographs. For the purpose of morphological observations, Sheldon divided the body components into five areas:

   1. Head, face and neck

   2. Thorasic trunk

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3. Arms, shoulders and hands
4. Abdominal trunk, and
5. Legs and feet.

Each component was scored from 1 to 7 as the somatotype of the subject. Each somatotype was thus represented by a three digit combination, i.e., 7-1-1 as extreme endomorph, 1-7-1 as extreme mesomorph and 1-1-7 as extreme ectomorph.

Brief description of each component follows as under:

1. **Endomorphy:** Reflects general softness and roundness of the body and its various forms. Proximal parts are relatively massive than distal ones, tapering of extremities, abdomen predominating over thorax, soft body contours, relatively small hands and feet.
2. **Mesomorphy:** Reflects general massiveness and sturdiness of the musculo-skeletal system of the body, highly developed limb muscles with distal segments of the extremities relatively more prominent. Strong and highly muscular thorax which predominates over abdomen with small antero-posterior diameter of trunk than the transverse one.
3. **Ectomorphy:** Reflects thin and lean body with weak muscles and thin skeletal diameters, pointed and sharp body projections, long and slender extremities with little muscles over them.

Critical evaluation:

1. This method is subjective.
2. This method is developed on white males of limited age range, ignoring all the other ages, races and females.
3. Ignores role of environment on human physique and considers the latter to be static from birth to death.
4. Overall body size and shape of the subject is ignored.

4. **Heath - Carter Method of Somatotyping:** Heath, in 1963, critically examined the short comings of Sheldon's method and came out with certain modifications and later in collaboration with Carter, gave her own method of somatotyping in 1967. This method differs from Sheldon's method in the sense that it evaluates the physique at a given time compared to the unchanging somatotype of Sheldon. The ratings of three primary components of physique are assigned as the basis of following anthropometric measurements. - a) Height, b) Body weight, c) Supra-spinal skin fold, d) Calf skin fold, e) Humerus diameter, f) Femur diameter etc.

1. **First component (Endomorphy):** Refers to relative flatness in individual physiques, as well as to relative leanness.
2. **Second component (Mesomorphy):** Refers to relative muscular skeletal development per unit height.
3. **Third component (Ectomorphy):** Refers to relative linearity of individual physiques. Evaluates the form and degree of longitudinal distribution of first and second components.

Evaluation:

1. Is it highly objective.
2. An excellent tool to explore spatial-temporal variations in human body form.

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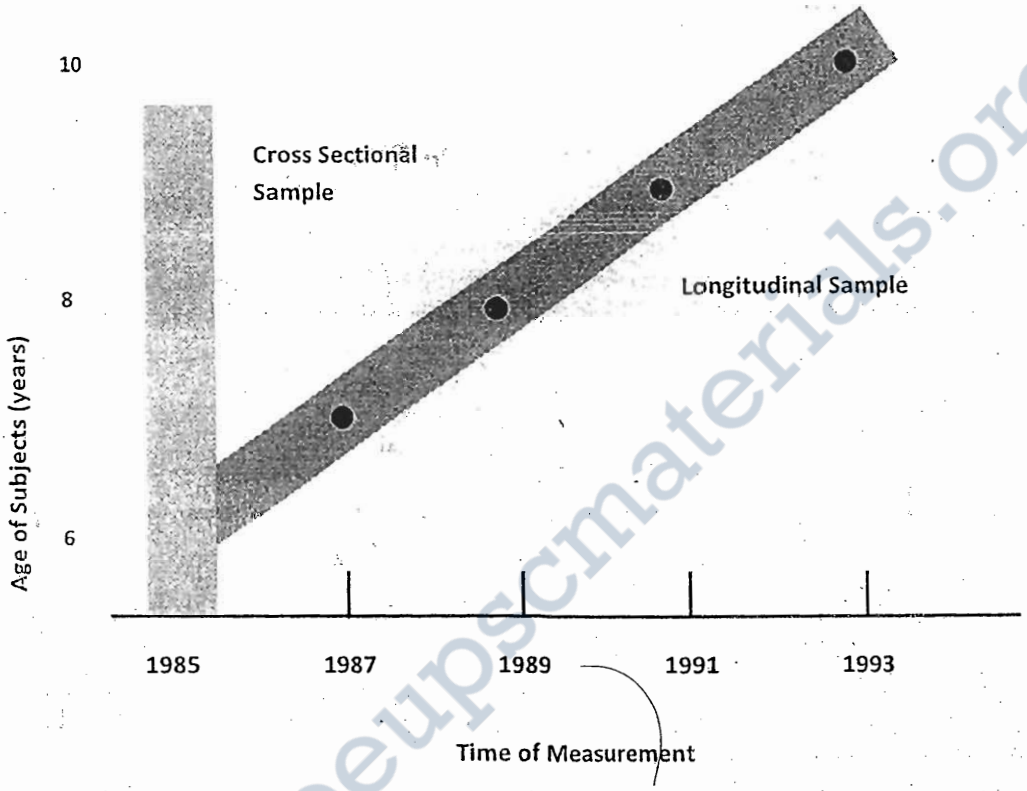
3. An easy, accurate and efficient method.
4. Method workable in field as well as laboratory.

Human somatotyping has not only a theoretical importance but also an enormous applied value. The somatotype technique is today increasingly used in the field of kinanthropometry, anthropology of sports. By comparing somatotypes of different regions, the role of environment can also be easily deduced.

METHODOLOGIES FOR GROWTH STUDIES

Information about growth and development is most commonly gathered by *cross sectional* or *longitudinal studies.* In some cases, *sequential designs* are used.



In a **Cross Sectional Study,** people of different ages are assessed on one occasion. This kind of study provides information about *differences* in development among different age groups, rather than *changes* with age in the same person (which longitudinal studies show).

In a **Longitudinal Study**, researchers measure the same people more than once to see changes in development over time. The researchers may measure one characteristic, such as vocabulary size, IQ, height or aggressiveness. Or they may look at several aspects of development, to find interrelationships among factors. Since the same people are measured more than once, this design gives a picture of the *process* of development.

Cross-sectional studies, then, look at **differences** among groups of people; longitudinal studies assess **changes** undergone by one or more persons. Each design has strengths and weaknesses.

The advantages of cross-sectional method include speed and economy; it is faster and cheaper than the longitudinal method. In addition, it does not lose subjects who drop out. Among its drawbacks is its masking of differences among individuals, since it looks at group averages. Its *major* disadvantage is that

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it cannot eliminate cohort, or generational, influences on subjects born at different times. Cross-sectional studies are sometimes misinterpreted as yielding information about developmental *changes* in groups or individuals; but such information is often misleading and may contradict longitudinal research. For example, it would be incorrect to conclude from the cross-sectional study that intellectual functioning declines in later years. This may be so, but longitudinal data would be needed to determine whether there were actual age changes. All that the cross-sectional method can show is that there were age *differences* in performance.

The great strength of *longitudinal* studies is their sensitivity to individual patterns of change, since data about individual patterns of change can be tracked. Also, they avoid cohort effects within a study – although longitudinal studies done on one particular cohort may not apply to a different cohort. Longitudinal studies, however, are time consuming and expensive. Another shortcoming is probable bias in the sample; people who volunteer tend to be of high than average socioeconomic status and intelligence, and those who stay with the project over time tend to be more competent than those who drop out. Also, the results can be affected by repeated testing; people tend to do better in later tests because of a "practice effect".

Sequential Studies

The **cross-sequential study** is one of several strategies designed to overcome the drawbacks of longitudinal and cross-sectional studies. This method combines the other two: people in a cross-sectional sample are tested more than once, and the results are analyzed to determine the differences that show up over time for the different groups of subjects.

Some important research on intellectual functioning in adulthood employs sequential techniques. These techniques seem to provide a more realistic assessment than either the cross-sectional method (which tends to overestimate the drop in intellectual functioning in later years) or the longitudinal method (which tends to underestimate it).

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11 Social Demography

11.1 Relevance of Menarche, Menopause and other Bioevents to Fertility

Fecundity and Fertility: Fecundity refers to the "capacity of a man, a woman, or a couple to participate in reproduction - i.e., the capacity to produce a live child. Fertility, on the other hand, refers to "the actual reproductive performance - whether applied to an individual or a group". While there is no direct measurement of fecundity, fertility can be studied from the statistics of births. The fertility of an individual is limited by his/her own fecundity-the physiological capacity to reproduce. The fecundity of an individual may be quite normal, yet the fertility performance may be low, for the term fecundity is biological, whereas fertility indicates the actual level of reproductive performance determined by social, cultural, psychological and even economic factors. Fecundity also refers to maximum fertility level that can be attained.

Sterility: While a man or a woman or a couple who has given birth to at least one live child, is considered fertile, one who has not had a single child is sterile. The term sterility may be used in connection with individuals or groups, consisting of either men or women or both. Sterility can be natural/involuntary or artificial/ voluntary.

Reproductive Span: The biological limits imposed on child bearing by such factors as age and sex can be easily recognized. Only women can conceive and give birth to children and that too within certain age limits. A woman becomes biologically fecund with the onset of menstruation. The onset of menstruation is called Menarche. Her capacity to bear children comes to an end with the onset of Menopause, when menstruation ceases. On the basis of research evidence, women can bear children from age of 15 to 44 years. The reproductive span of men is not well defined, nor is it easily identifiable, though it is generally found to be considerably longer than that of women.

Theoretical Maximum Fertility: Theoretically, during the physiologically limited child bearing period, a woman would get 37 children, if she gave birth to one child every ten months over a period of 31 years. Such a phenomenon is however very rare. There is a gap between fecundity and actual fertility. This gap is because of the operation of many physiological or biological factors along with social as well as cultural factors.

Adolescent Sterility: The fecundity of females is not uniform throughout the reproductive span of 30 to 35 years. Then woman's age is an important factor when her reproductive capacity is considered. Female fecundity is at a low level during the early stages of puberty. A certain degree of adolescent sterility (sterility among the adolescent age group) or sub-fertility is observed for girls between the ages of 13 and 18 years. Adolescent sterility or sub-fertility occurs because regular ovulatory cycles are not generally firmly established for the first two or three years after the onset of menstruation. The interval between menarche and the attainment of full biological maturity to bear children is therefore called the period of adolescent sterility. A woman's capacity to bear children reaches the highest level at 20 to 25 years, after which it starts declining first slowly and then after the age of 38 quite rapidly and reaches zero level at about 50 years.

Post-Partum Sterility: During the reproductive span of women, there are certain periods of temporary sterility. After the birth of a child, the woman is generally sterile for some period, as the menstrual cycle is not resumed, or if it is reestablished, the earlier cycles are un-ovulatory. This post-partum period in which menstruation is not resumed is known as the post partum amenorrhea period. During this period, the possibility of conception is very rare and hence this period of temporary sterility is known as the post partum sterility. It has been reported in many Indian studies that as a result of breast-feeding the post partum amenorrhea period is longer.

Reproductive Wastage: Reproductive wastage in terms of abortions and stillbirths also act as physiological factors affecting fertility.

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FERTILITY PATTERNS AND DIFFERENTIALS

It has been observed that the levels and patterns of fertility vary considerably in various sub-groups of the same population. These subgroups may be based on residence, whether urban or rural, social and economic status in terms of educational attainment, occupations, income, size of land holding, religion caste, race, etc. A study of differential fertility is useful in identifying the factors which determine fertility levels among various sub-groups. The information so provided gives us some idea of the future proportion of each group in the total population and also helps us to project more accurately the future population size of the entire country. A study of differential fertility is also important from the point of view of the implementation of family planning programs because it helps us to identify high fertility groups on which the program's efforts can be concentrated. In this connection, it is interesting to note that in the 1930s, when birth rates were at the lowest levels, research was concentrated on those groups which had the lowest fertility with a view to uncovering clues for the formation of policies for the encouragement of high fertility among these very groups.

Differential fertility can be understood as a result of following factors.

ECOLOGICAL FACTORS

(a) **Regional Differences in Fertility:** The fertility rates of various regions or States or provinces within one country may differ widely. In India, for instance, there is a great deal of variation in the fertility levels of various States and Union Territories.

(b) **Rural-Urban Residence and Fertility:** Numerous studies have been conducted on fertility differentials according to rural-urban residence. Towards the end of the last century, in the low fertility areas of the world, it was found that the fertility of those residing in cities was lower than that of rural residents and these differentials were more or less stable. However, when national birth rates declined these fertility differentials widened in a more pronounced manner among urban upper classes than among others. Rural-urban differences narrowed down again when birth rates rose after the Second World War.

SOCIO-ECONOMIC FACTORS

a. **Educational Attainment and Fertility:** The educational attainment of couples has a very strong bearing on the number of children born. Educational attainment, especially of women, is one of the indicators of modernization and the status of women in society. In low fertility countries, historically the relationship between fertility and the educational attainment of the wife has been a negative one, in the sense that the higher the educational level, the lower was the family size.

b. **Economic Status and Fertility:** General studies in the past have highlighted the inverse relationship between the economic status of the family and fertility. This traditional relationship is now undergoing substantial changes as far as the developed countries are concerned.

c. **Occupation of Husband and Fertility:** In developed countries occupation, especially that of the husband is used as an indicator of social economic status, and differential fertility is studied according to the occupation of the husband. Studies conducted in Europe around 1970 indicated that the wives of farmers and farm workers recorded a higher fertility than the wives of men engaged in non agricultural occupations. Even amongst those engaged in agriculture, the fertility of the group of farm workers was higher than that of the group of farmers. These differences were more pronounced in France and the United States than in the other countries. Manual workers were also found to have, on an average, more children than non manual workers. In India, some studies have tried to investigate the relationship between the occupation of the husband and fertility. It was generally observed that the wives of those engaged in professional jobs had the lowest fertility.

d. **Employment of Wife and Fertility:** It has been found in several studies that the gainfully employed women have a smaller number of children than those who are not employed. Though this

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relationship has been quite distinct in many industrialized countries, the exact nature of this relationship is not yet known. Is it that women who have smaller number of children tend to take paid jobs outside their homes, or is it that those who have already paid jobs restrict their family size to the very minimum so they can work? It is difficult to say anything very firmly.

e. **Religion, Caste, Race and Fertility:** Religion is considered to be an important factor affecting fertility. The study of differential fertility of various religions as well as ethnic groups has important social and political implications. In a democratic society where each person has the right to vote, the size of a particular religious, caste or ethnic group may be an important factor in determining the political power-structure. Several studies have been conducted in developed as well as developing countries to investigate the influence of affiliation to a particular religion on the fertility behavior of the people. At one time, all the religions of the world, except Buddhism, were pro-natalist or "populationist". The injunctions laid down in various religions indicate the importance of high fertility. Some illustrations are: "Be fruitful, multiply and replenish the earth" (Judaism and Christianity); "Marry a woman who holds her husband extremely dear and who is richly fruitful" (Islam); "Make the bride the mother of good and fortunate children, bless her to get ten children and make the husband the eleventh one" (Hinduism). It may however be pointed out that all these religions have their roots in the distant past, which was a period of high mortality. As such, the emphasis on prolific fertility was a functional adjustment to high mortality in order to ensure the continuation of the group. Resistance to human interference with fertility is common to all religions. As pointed out by Lorimer and Osborn this resistance has been less persistent in those religious groups where there was an absence of a central authority as in the case of Judaism and the Protestant sect of the Christian religion. This same reason, namely the absence of a central authority, may be attributed to the absence of opposition to birth control among the Hindus.

Various studies in the West, which is essentially dominated by the Christian-Hebraic tradition, show that the fertility of Catholics has exceeded that of non-Catholics in almost every country and socioeconomic group. The studies conducted in the United States and Canada have clearly pointed out that the fertility of Roman Catholics has been higher than that of either Protestants or Jews. Recent findings indicate that in most of the European countries as well as in Canada, differentials in fertility by religious affiliation are narrowing. The reason for this narrowing of differences may be due to modifications in the religious sanctions relating to the ideology and practice of birth control and the weakening of the influence of religious doctrines and traditions. It may also be that the influence of other socioeconomic factors such as urban residence, educational attainment etc. on fertility is on the wane with the result that fertility differentials are narrowing.

India may be taken as an illustration from the developing countries. In most of the studies conducted in India to study the fertility behavior of various religious groups it was observed that the Muslims had a higher fertility rate than the Hindus or the Christians.

Some sociologists are of the opinion that minority religious groups may tend to have higher fertility rates to gain more political power. This however does not hold true in all cases. Two minority communities, that is, the Zoroastrian community in India and the Jewish community in the United States, have always shown lower fertility rates than the majority group. Other socio-economic factors such as the educational attainment of the wives, the status of women, the degree of modernization of the community, etc., are found to affect the fertility of the subgroups in a particular society.

From this discussion it is obvious that it is difficult to determine the exact role of religion in influencing fertility and that the role of religion in causing fertility differentials though not negligible is quite limited.

As caste is an important indicator of social status in India, attempts have been made in several studies to observe fertility differentials by caste. In a study conducted in Lucknow City, it was observed that

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upper caste Hindus had, on an average 3.8 live births while the lower caste Hindus had on an average 4.1 births.

In multi-racial societies differences in fertility have been found among various racial groups. In the United States, for example, the Negroes show a higher fertility than the white community. These differences, however have been explained away by socio-economic factors.

Inter-relationships between Socio-Economic Variables Affecting Fertility: In the discussion on differential fertility only one variable at a time has been taken into consideration with a view to explaining the differences in fertility among various sub-groups in any population. It must however be recognized that all these variables are closely interrelated. For example, when the husbands are highly educated, they tend to have educated wives. Higher educational levels are related to higher incomes and more gainful occupations leading to higher standards of life. In a community in which women are educated, the influence of religious dogma is likely to be less rigid, while if the community has a high proportion of illiterate women, religious influences tend to be strong and affect fertility. Age at marriage is also closely associated with the educational attainment of wives.

When fertility differentials are studied in relation to various ecological, socioeconomic and cultural factors, it is found that, in many developed countries, they are narrowing, and the latest trend is towards uniformity. On the other hand, in a developing country like India, fertility differentials have in recent times become increasingly pronounced. Amos Hawley has identified three phases in the trends in fertility differentials. In the first phase, various socio-economic classes either had identical fertility rates or the relationship between socioeconomic status and fertility was positive. In the next phase, which began with industrialization and the resulting socioeconomic development, it was the highly educated urban elite group which first began to show declines in fertility thus showing a wave of fertility differentials. In the final phase, which is in existence now in many Western countries, there appears to be a positive association between socio-economic status and fertility. India is at present in the transitional stage of declining fertility and therefore differentials among various socio-economic classes are becoming increasingly more pronounced.

11.2 Demographic Theories – Biological, Social and Cultural

WHAT IS DEMOGRAPHY?

Demography is the study of human populations, their growth and decline due to changing patterns of migration, fertility and mortality and characteristics such as the Sex-ratio, dependency ratio, and age-structure. The subject is sometimes divided for further elucidation into 'formal demography', meaning the formal statistical analysis of population parameters and dynamics, and 'population studies', the wider investigation of the causes and consequences of population structures and change. It is in the latter area that many demographers have interests which overlap with those of sociologists and anthropologists and in their investigation demographic analysis forms an important component in the description and understanding of human societies.

The methodology of demography consists of analysis of databases of official statistics from births, deaths, and marriage registration, and from population censuses. Demographers seek ultimately to produce population projections that is forecasts not only of the size of the population over coming decades, but also its changing age-structure, which can be important for social policy and labor-market policy. For example, if the dependent population (children under school-leaving age and people over retirement age) is growing relative to the population of working age which has to support it financially, there may be major implications for taxation, social insurance, and fiscal policy. If the population of working age is declining in absolute numbers, there may be a case for government policy to encourage a larger percentage (of women especially) to enter employment. Thus demographic statistics and analyses provide the essential underpinning for many other types of study. For this reason population censuses were the very first type of systematic social enquiry to be developed.

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Analyses of vital statistics (statistics of births, deaths, marriages etc., of a country) do however have their limitations. In particular, they cannot supply information on the motivations, value-systems, or aims and preferences underlying changes in the birth-rate, which is a key factor in population growth. In recent years, there have been concerted efforts to develop and carry out interview surveys on fertility orientations and behavior. These cover issues such as the preferred number of children in a family, the effects of household income and women's employment on their fertility, attitudes to contraception and its use-all factors affecting the timing and spacing of births.

ANTHROPOLOGY AND DEMOGRAPHY

Demographic behavior is a part of individual's whole socio-psychological behavior, which has a decisive effect on the possibility of some demographic event. The socio-economic system of the society guides demographic behavior and by employing concepts of demography, socio-economic conditions can be clearly understood.

The foregoing discussion reflects the relation between anthropology and demography. Moreover, the relation between these two sciences becomes even clearer when one realizes that a majority of problems connected with population are classified as social problems. Values are fundamental to any society and functioning of any culture. Hence, demography cannot afford to ignore the study of social values which provide the context in which every human population grows and thrives to survive. This demands a holistic approach, especially drawing from the principles, concepts and methodology of anthropology. Even the *success and failure of family planning programs are* determined by socio-cultural consciousness of people. Brown and Selznick have noted that the areas of concern of both sociology and demography are common - social organization, collective behavior, urbanization, primary groups etc. This reveals the inseparable relationship between anthropology and demography and prevents any underestimation of their functional interdependence.

SOCIAL DEMOGRAPHY

Social demography is a field of study concerned with the analysis of how social and cultural factors are related to population characteristics. Its major focus is the impact of social and cultural factors on demographic features of society such as patterns of marriage and child bearing, the age-structure of the population, life-expectancy, and so forth. In addition however, social demography also encompasses examination of the social consequences of demographic change. Since the demographic characteristics of a society or social group are themselves social phenomena, and the immediate product of the social (but also biological) events of birth and death, in one sense the demographic study of any human population is a form of social demography. However, whereas demography itself is primarily concerned with determining and measuring population characteristics and the interrelationship between demographic variables, social demographers seek to understand and explain these demographic patterns. In so doing they draw on the expertise of anthropology, sociology as well as of demography.

The three main variables underlying population change are fertility, mortality, and migration, variables themselves associated with factors such as age at marriage, the proportions marrying, contraceptive use, levels and types of morbidity, rural-urban migration, and so forth. All receive attention from social demographers, who seek to understand these processes in terms of a range of standard social factors such as the levels and distribution of income, levels of education, the position of women, religion, and economic development.

THEORIES OF DEMOGRAPHY

MALTHUSIAN THEORY OF POPULATION

Thomas Robert Malthus attempted a population-resource relation in his Malthusian Principle of Population. The Malthusian theory of population is an economic approach to demography and is an empirical statement (since it is based on the population experience of West European Countries). The

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theory is based on two assumptions. The first assumption is that food is always necessary for mankind. The implications of this assumption are 1) Population growth is limited by the means of subsistence 2) Population growth increases in a proportion to the increasing means of subsistence (i.e. food) and 3) Population growth increases with increasing means of subsistence unless moderated by certain "Checks".

The second assumption of the Malthusian theory is that the passion between both the sexes will remain at the same levels of intensity throughout human history. The implications of this assumption are 1) birth rates will remain at relatively high levels (due to undiminished passion between sexes) and 2) population growth will proceed in a geometric ratio. According to Malthus, these two assumptions are immutable (unchangeable) principles and are also opposed to each other. The relation between these two principles will be such that while food production grows in an arithmetic ratio (1, 2, 3, 4,), the population will grow in a geometric ratio (1, 2, 4, 8,). As a result of this kind of relationship, the gap between population and food availability will continuously widen. This widening gap will have two consequences. The first consequence is the increasing stress of rising population on the natural resources which provide sustenance. This will lead to breakdown of the natural ecosystems triggering natural catastrophes.

The second consequence of the widening gap between food availability and rising population is the widening gap between the rich and the poor. The rich will try to control more resources but at the same time they do not produce many offspring fearing decline of their standard of living.

The Malthusian catastrophe (natural catastrophes in the form of famines and floods) will be inevitable unless some checks come into operation. The checks can be preventive and positive. The positive checks are largely outside the control of man and include war, disease, poverty etc. The preventive checks are within the control of Man and include factors such as moral restraint, birth control measures including abortion and vice such as adultery, and pre-marital sex. According to Malthus, man should take active steps by resorting to preventive checks to prevent the Malthusian catastrophe (breakdown of resource systems) from occurring. Though Malthus talked about birth control as one of the preventive steps, he was not convinced about contraception. According to him, contraception would not lead to delaying of marriage and hence would increase birth rates in the long run (on the assumption that lack of sex would drive people to work hard for preparing the ground for marriage, but with contraception people can indulge in sex early and also marry early).

Though the population theory of Malthus initiated theory building in population-resource relationships and also brought into sharp focus the importance of stable land-man ratios, a series of criticisms have been leveled against it. For e.g., it has been pointed out that Malthus could not envisage how technological revolutions would lead to greater provision of sustenance, thereby freeing Man from the threat of Malthusian catastrophes. It is also argued than Malthus mixed up a purely biological instinct (the desire to have sex) with a purely social instinct (the desire for children) by invoking the principle of undiminished passion between sexes. In addition, the arithmetic vs. geometric ratios for food growth and population growth has failed empirical validity in different regions of the world. The theory is also criticized on two other counts - its greater emphasis on positive checks and the unethical nature of preventive checks.

THEORY OF OPTIMUM POPULATION

The advocates of Optimum Population Theory, especially Edwin Canan, hold the view that population should grow to a desired / optimum level. Any increase in the population beyond this level is harmful to the society. The stage of optimum population would reach when the society is able to run its minimum and essential services. The situation can be optimum or satisfactory when the death rates equal birth rates.

This theory is based on the assumption that there is a close relation between population size and economic development. Economic development presupposes optimum utilization of all the resources and population constitutes the human resources which are employed to exploit both natural resources as well as capital. In countries where there is less population, in terms of labor, population growth is necessary

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to ensure use of all the available resources. This situation where the available resources are fully utilized is "Optimum Population". In the event of continued population growth beyond the stage of optimum population, then it would result in disturbance of balance of resources for production and economic development. This is because such growth would ensure a situation where more labor is available than what is "ideally" needed.

The theory of optimum population is based on the following two principles:

1. The rates between total population and working population remained constant even though there is an increase in the population.

2. The availability of natural resources and capital do not change proportionate to change in population. This results in operation of Law of Diminishing Returns.

CLASSICAL THEORY OF DAVID RICARDO

Ricardo adopted classical economic model to population growth. According to him, in early stages, the growing demand for labor pushes up the population growth rate. As the labor supply increases, the wages begin to decline. Ultimately, equilibrium is established between the demand and supply of labor. Once this equilibrium is established, capital accumulation in the economic system stops which leads to everyone receiving the same wage. This acts as a deterrent to further population growth because, beyond this level, population growth would lead to universal poverty.

THEORY OF HERBERT SPENCER

The British philosopher, Spencer, proposed a natural law of population growth based on the relationship between individuation and genesis. According to this theory, Man's interest in reproduction decreases with increasing socio-cultural development because of the antagonism between individuation and genesis. Hence as the society evolves, its fecundity will also decrease. This is a natural law which therefore absolves Man from any responsibility for controlling his numbers, as Nature achieves this by reducing Man's reproductive interests with increasing cultural development.

THEORY OF KARL MARX

Marx approaches the problem of population growth dialectically and his theory of population is a critique of capitalism. The two classes which make up a capitalist society respond differently to population growth according to Marx. The rich own the means of production and do not increase their numbers rapidly because labor is not an asset to their class. The poor, on the other hand, do not own any other means of production except labor, the only asset the class possesses. As a result, the poor tend to accumulate labor through rapid population growth. Eventually, the poor uproot the capitalist order by a revolution and assets are equally owned by all. Population growth is therefore slowed down. But, population growth can be slowed down by adopting the Communist system instead of letting a class revolution uprooting the capitalist system. The communist method of production would therefore solve the problem of rapid population growth.

CYCLICAL THEORY OF CORRADO GINI

According to Corrado Gini, the growth of human population follows the cycle of growth of the individual. Hence, it follows that the first stage of rapid growth would eventually be replaced by a second stage of slow growth.

Stage 1: This is the stage of rapid population growth. Gini maintains "every nation in its youth is simple and undifferentiated in its structure and has a high rate of fertility, because each generation springs from the people who are hereditarily most prolific i.e. highly fecund". Organizations become more and more complex as the numbers increase. The structure of the society becomes more and more complicated as

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industrial and commercial activities grow. At this stage, attempts are made to check population either through colonialism or war.

Stage 2: In this stage the population decreases. Gini gives two reasons for this trend.

1) Colonization or war resulting in loss of young and energetic people

2) Increase in the number of upper-class population which is less prolific than the lower class population.

The reason for this decline is biological and hence the decline in fertility is due to a decline in fecundity. As the lower class people move up the ladder to become members of upper class, even they reflect the lowered fertility rate. Gini notes "Even the sterility of these climbers is not a consequence of social conditions surrounding their climbing; it is rather a result of weakening of the reproductive instinct and is an inevitable phase of cycle of population growth". He continues his argument while saying that there would be a decline in quality of individuals due to decline in population and this is the result of change in the hereditary quality of individuals. The rise and fall of population is determined by an inevitable natural growth and he thus invoked some sort of "mystical" biological changes which determined both the qualitative and quantitative changes in human populations. Moreover, these biological changes were beyond the control of human beings. Thus, his theory analogizes population growth to human cycle of birth, growth and death. Corrado Gini's theory is supported by Bertrand Reed's conception of population growth which he represented graphically by letter 'S'. According to this school of mathematical theorists on demography, the growth of population is slow initially, gradually increases later and steadily till it reaches a mid-point.

THOMAS SADLER'S DESTINY AND FECUNDITY PRINCIPLE

Michel Thomas Sadler's theory was actually put forth as a critique of Malthusian theory on population. He opined that nature's laws about population were just the opposite of those which were enunciated by Malthus. He put forth his principle in his book "The law of population" as "the fecundity of human beings is in inverse ratio of the conversion of their numbers".

Labor and Privacy, according to Sadler are two important pre-requisites for maintaining population growth. The desire to put in labor and hard working attitudes are decreasing according to him with growth and advancement of civilization. This is mainly because machines started replacing the humans. Lack of privacy emerges as a consequence of shortage in accommodation. Because of reduction in labor and lack of privacy, there will be a reduction in population fertility rate. He thus concludes that nature, contrary to Malthusian conception, is helping mankind rather than having a negative approach towards population. Though this theory has been criticized on a number of grounds, especially on the ground that Sadler was confused with the distinction between fertility and fecundity, it remains significant because it was successful in removing pessimism which was created by Malthusian theory.

DIET THEORY OF THOMAS DOUBLEDAY

Thomas Doubleday drew a correlation between the diet of the people and population growth. He believed that man's increase in numbers was inversely proportional to his food supply. That is, the better the food supply, the slower the increase in numbers. In all those societies where food supply is in shortage, fertility rates are usually higher, contrary being the case in societies where there is good food supply. Population, Doubleday maintains, rapidly increases where people get very little food. He says "There is in all societies a constant increase going on amongst that portion of it which is worst supplied with food, instead among the poorest. He further extended his theory to dietary habits. He says that the non-vegetarians have less capacity to reproduce. He gives this as a reason for low density of population among pastoralist societies. Similarly, fertility is bound to be highest where population is purely vegetarian while it is moderately high among people who are both vegetarian and non-vegetarian.

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Jouse de Castro's Theory of Protein Consumption which he elucidated in his book "Geography and Hunger" also points to similar conclusions as Doubleday's. Castro too has come out with a negative correlation between protein intake and birth rates of various countries. He concludes that poor countries are highly populated because they have undernourished population which is more fertile.

THEORY OF DEMOGRAPHIC TRANSITION

Relatively, the Theory of Demographic Transition is a recent one. This theory is more rational as it is based on the demographic experience of many nations, especially those of the west. A majority of sociologists and an equal number of economists as well as demographers believe that the population dynamics in any country pass through a series of stages, each stage having its peculiarity. Though scholars are divided about the classification and grouping of these stages, the Theory of Demographic Transition is widely accepted as a framework through which the processes of population can be well understood.

Though there are many contributors to and proponents of this theory, it was however Notestein in the year 1945, who gave it a natural form. The theory postulates a three stage sequence of birth and death rate as typically associated with economic development.

First Stage of Demographic Transition:

According to this theory, death rates are high in the first stage of an agrarian economy on account of poor diets, primitive sanitation and absence of effective medical aid. Birth rates are also high in this stage as a consequence of widespread prevalence of illiteracy, absence of knowledge about family planning techniques, early age of marriage and as a consequence of deep-rooted social beliefs and customs about the size of the family, attitude towards children, etc. Moreover, in a primitive society there are economic advantages of a large family size. "Children contribute at an early age . . . and are the traditional source of security in the old age of parents. The prevalent high death rates, especially in infancy, imply that such security can be attained only when many children are born." In such a society the actual rate of growth of population is not high since high birth rate is balanced by high death rate. It is a stage of high growth potential but of low actual growth.

Second Stage of Demographic Transition:

Rise in income levels enables the people to improve their diet. Economic development also brings about all-round improvement including the improvement in transport which makes the supply of food regular. All these factors tend to reduce death rate. Thus in the second stage, birth rate remains high but death rate begins to decline rapidly. This accelerates the growth of population. High growth potential of the first stage is realized in the high actual growth in the second stage as a consequence of decline in death rate. High birth rate and falling death rate contribute to the growth of the average size of the family in the second stage.

The Third Stage of Demographic Transition:

Economic development further changes the character of the economy from an agrarian to a partially industrialized one. With the growth of industrialization population tends to shift away from rural areas towards industrial and commercial centers. Growth of urban population "with the development of economic roles for women outside the home, tends to increase the possibility of economic ability that can better be achieved with small families, and tends to decrease the economic advantage of a large family. One of the features of economic development is typically increasing urbanization, and children are usually more of a burden and less of an asset in an urban setting than in a rural." The consciousness to maintain reasonable standard of living tends to reduce the size of family in an industrialized economy; since the death rate is already low, this is possible only if birth rate falls. Thus, the characteristics of the third stage are low birth rate, low death rate, small family size and low growth rate of population. This is the stage of incipient decline of population.

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These three stages reveal the transformation of a primitive high birth and high death rate economy into a low birth and low death rate economy. When an economy shifts from the first stage to the second stage of demographic transition, an imbalance is created in the economy as a result of falling death rate but relatively stable birth rate. Historically it has been observed that death rate can be controlled more easily because the measures to reduce death rate are exogenous in nature and hence readily acceptable to the people. But the reduction of birth rate can be brought about by operating on endogenous factors, like changing social attitudes and customs, beliefs and dogmas about the size of the family, about marriage, etc. This requires a much longer time than the fall of death rate. Consequently, birth rates tend to fall after a time lag. The second stage of demographic evolution has, therefore, been termed as the stage of population explosion. This stage is the most hazardous period for a developing economy. The decline in death rate in the second stage, therefore, creates an imbalance which requires a period of transition for adjustment. Thus, the theory is termed as the theory of demographic transition. During the period of transition the demographic factors get out of harmony. A new constellation of demographic forces is brought about which changes the character of society, birth and death rates become balanced at a lower level as a result of which growth rate of population also declines.

11.3 Biological and Socio-ecological Factros Influencing Fecundity, Fertility, Natality and Mortality

FERTILITY

Births or fertility occupies a central place in the study of population because of the following reasons:

1. Fertility is responsible for biological replacement of life and maintenance of human society.
2. The growth of population depends to a very large extent on fertility.
3. The society replenishes itself through fertility.
4. Fertility is a positive force counteracting the negative force of mortality or deaths.

MEANING OF FERTILITY

1) Lewis and Thompson: - "Fertility is generally used to indicate the actual reproductive performance of a woman or groups of women".

2) Bernard Benjamin: - "Fertility measures the rate at which a population adds to itself by births and is normally assessed by relating the number of births to the size of some section of population ... as an appropriate yardstick of potential fertility".

3) Barclay: - "The fundamental notion of fertility is an actual level of performance in a population, based on the number of live births that occur".

FECUNDITY, FERTILITY AND NATALITY

Fecundity is the capacity to conceive or bear children. It is defined as a "biological potential or the physiological capacity to participate in reproduction." The absence of fecundity is called sterility or infecundity. It is the potential level of performance (or the physical capacity for bearing children) of the population. Fertility on the other hand refers to the actual reproductive performance of individual or group. It follows that fertility of an individual will be limited by the physiological capacity to reproduce i.e., fecundity.

Natality is the birthrate, which is the ratio of total live births to total population in a particular area over a specified period of time; expressed as childbirths per 1000 people (or population) per year.

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FACTORS DETERMINING FERTILITY

A causal analysis of fertility may involve a great number of factors and complicated chain reactions. It is therefore appropriate to make a systematic classification of the mechanisms which directly affect fertility through which all other factors must operate. An excellent categorization of these mechanisms has been devised by Kingsley Davis and Judith Blake. Their most basic concept is that the birth of a child is not possible unless...

1) Sexual intercourse has occurred
2) Intercourse has resulted in pregnancy
3) Pregnancy has been brought to successful term.

Building on this concept, they have devised as list of eleven variables which directly affect fertility. They term these "intermediate variables" since any other variables which may affect fertility must ultimately act through one of these.

I. Factors Affecting Exposure to Intercourse:-
   A. Those governing the formation and dissolution of unions in the reproductive period.
      1. Age of entry into sexual unions
      2. Permanent celibacy; proportion of women never entering into sexual unions
      3. Amount of reproductive period spent after or between unions
         i. when unions are broken by divorce, separation or desertion
         ii. when unions are broken by death of husband
   B. Those governing the exposure to intercourse within unions
      a. Voluntary abstinence
      b. Involuntary abstinence (from impotence, illness, and unavoidable but temporary separations)
      c. Coital frequency (excluding periods of abstinence)

II. Factors Affecting Exposure to Conception:-
      d. Fecundity or infecundity, as affected by involuntary causes
      e. Use or non-use of contraception
         a. by mechanical or chemical means
         b. by other means
      f. Fecundity or infecundity, as affected by voluntary causes (sterilization etc).

III. Factors Affecting Gestation and Successful Parturition:-
      g. Fetal mortality from involuntary causes
      h. Fetal mortality from voluntary causes.

Now we shall look into these variables in some detail.

1. **Age of Entry into Sexual Unions**: In the European nations' culture a couple is not supposed to marry until the husband is able to support a wife and family. In pre-industrial Europe, the age of marriage was relatively low but began to rise gradually. Ohlin has suggested that the rise in the age at marriage could plausibly be explained by the decline in mortality, since the average man now had to wait longer before inheriting land or advancing from his apprenticeship. In many Asian nations, on the other hand, the age at first marriage has always been very early, since marriages are arranged and the husband is not expected to support his family entirely by his own efforts.

2. **Permanent Celibacy:** A rather high proportion of permanent celibates are frequent in the nations which have a late average age at marriage.

3. **Amount of Reproductive Period Spent after or between Unions:** To some extent in all societies actual fertility is reduced below the biologically maximum level because part of the reproductive period is spent after or between sexual unions. Where monogamy is institutionalized it is almost inevitable that a certain proportion of widows never remarry, since there are almost always considerably more widows than widowers, and many widowers prefer to marry never-married

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women. Periods of separation between marital unions are also important in some societies in reducing fertility. Many legal marriages and consensual unions breakup and the breakup may be caused by marital incompatibility, or in many cases may occur simply because the woman does not want to incur the choice of an additional pregnancy.

4. **Voluntary Abstinence**: Certain primitive societies enjoy periods of voluntary abstinence on special ceremonial occasions. Almost all societies enjoy a period of abstinence during late pregnancy and also during the early postpartum period. The former has no detrimental effect on fertility and the latter has little since almost all women have very low biological fecundity during this time. Of the various forms of voluntary abstinence, the "rhythm period" probably has the greatest effect on fertility. This method of birth control demands abstinence in the days before and around the time of ovulation, which generally occurs around the mid-point of the menstrual cycle. When properly practiced, it will reduce the conception rates rather drastically.

5. **Involuntary Abstinence:** In a few societies a large proportion of men must absent themselves from their wives periodically to obtain gainful labor.

6. **Frequency of Intercourse:** Much theoretical evidence suggests that this variable may be rather important in determining differences in fertility between individuals, whether or not it affects the fertility of different populations is another nation. It is possible, however, that factors such as diet, temperature, humidity and the prevalence of certain enervating diseases may have effects on the average frequency of sexual intercourse in different populations. This is clearly a field where we need much more information than we now have.

7. **Fecundity or Infecundity as Affected by Involuntary Causes:** Several factors may affect the probability of conception, given the fact that intercourse occurs at a specified frequency. On a world wide basis perhaps the chief of these is the incidence of venereal disease. Venereal disease affects the fecundity of both men and women. Extreme hunger has also been found to cause amenorrhea (and hence temporary sterility) in women and a reduced sperm count in men. Modern medical science has made considerable progress in reducing the proportion of persons who are involuntarily childless. The use of antibiotics in case of venereal diseases has been very important in this respect. Another advance has been the use of artificial inseminations to impregnate women whose husbands are sterile. New drugs have recently been developed which stimulate ovulation and allow certain women to conceive who otherwise would probably never be able to do so.

8. **Use or Non-use of Contraception:** According to popular belief, contraception is the most important of all the intermediate variables effecting fertility. Actually, although there is no doubt that contraception is very influential in reducing levels of fertility, it is definitely not so overwhelming a contribution that the other variables can be ignored.

9. **Fecundity or Infecundity as Affected by Voluntary Causes:** The surgical operations of tubectomy in females and vasectomy in the males provide an individual permanent freedom from further parenthood. On a worldwide basis, prolonged breast feeding is one of the most important means by which a woman may temporarily reduce her fecundity. Women are sterile during their period of postpartum amenorrhea and a short period of an ovulatory cycle following the resumption of their menses. Prolonged lactation has a pronounced effect on the length of the period of post partum sterility. According to Robert Potter, the period of post partum sterility averages 13 months in a population which engages in prolonged lactation, and only four months in a population with no lactation.

10. **Fetal Mortality from Involuntary Causes:** On average, about 20% of all known pregnancies are spontaneously aborted. There is much individual variation in the proportion of pregnancies which miscarry, but little is known how populations may vary in this respect.

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11. **Fetal Mortality from Voluntary Causes:** Induced abortion is one of the most important means of birth control. Primitive methods of abortions have been practiced through out human history.

FACTORS ASSOCIATED WITH LONG-TERM DECLINE IN FERTILITY IN DEVELOPED COUNTRIES

In this section the factors which have brought about a change from high fertility to low fertility in developed countries will be discussed in detail.

Motivational Factors: Motivational factors have played an important role in bringing about a change from high fertility to low fertility. Demographers are of the opinion that over the years tremendous changes have occurred in the attitudes of couples towards reproduction. It appears that they have moved away from a strong positive desire to have several children to a strong motivation for a limited family. It is difficult to provide research evidence in support of this shift in the attitudinal and motivational forces which came in the wake of the social and economic conditions arising out of the Industrial Revolution. It is however quite evident that these forces operated at the level of individual couples, who translated into action the desire for a small family, for the social atmosphere was not favorable to birth control, nor were there any effective means of contraception. The State in most and the Church in all the cases did not approve of birth control. In fact, as Gunnar Myrdal put it, "All the forces of organized society-the law, officialdom, the clergy, educators, the press, the medical profession- were mobilized to prevent birth control from spreading". Yet the strong motivation of individual couples drove them to the achievement of their desired goal of having small families.

Economic and Social Factors: The phenomenon of fertility decline in the now developed countries is very complex. Several interacting and overlapping economic and social factors were responsible for the transition from high to low fertility. It follows therefore that no single factor can be held responsible for this fertility decline. In what follows, some economic and social factors are reviewed in the context of fertility declines. These are: (i) industrialization; (ii) urbanization; (iii) rising levels of living and increased costs of bringing up children; (iv) family functions and structure; (v) relationship between mortality and fertility; and (vi) social mobility. The process of industrialization began towards the middle of the eighteenth century, first in England and later throughout Europe and North America. It brought in its wake several far-reaching economic and social changes, which in turn brought about fertility declines. Most important, the process of industrialization initiated the process of modern economic growth; the per capita productivity increased and real income rose. Advancements in science and technology further improved the productivity of labor, for they created conditions in which workers received better education and training, worked shorter hours as a result of social reforms, and had better nutrition because of increased availability of food supplies. Several structural changes also took place about the same time. The share of agriculture to total product and that in the labor force decreased; there was a corresponding rise in the share of industry and other non-agricultural sectors. Industrialization was accompanied by urbanization. Declines in mortality were registered because of agricultural, economic and social development that came in the wake of industrialization.

Several change accompanied growing industrialization and urbanization, which had implications for fertility decline. Of particular interest are the changes which look place in the structure and functions of the family - the basic unit of society. The family lost its function as an economic unit, in the sense that it ceased to be a producing unit and became only a consumer unit. With the introduction of laws which prohibited child labor and made education compulsory, the economic usefulness of children to their parents was drastically reduced. In fact, they became a liability because of the increasing costs and lengthening duration of education. At the same time, there were declines in mortality, especially infant and child mortality; more children survived and the burden of bringing them up fell entirely on the nuclear family. Parents soon realized that, because of declining mortality, there was no need to have a large number of children in the hope that a few at least would survive. They therefore had fewer children. The advantages of rising real incomes flowing from industrialization were in danger of being nullified by large families, especially because of the rising costs of bringing up children. A large family was therefore seen as a threat to maintenance of a certain standard of life, and couples responded to this

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threat by having a smaller number of children. Rising costs of child rearing was thus an important factor in fertility declines in developed countries. Certain measures initiated by the Governments of various countries also contributed to changes in the attitudes of parents towards their children. Financial responsibility for medicines and medical treatment, provision of old age security, etc., which were originally shouldered by the family, were taken over by the State in many countries. Children therefore were no longer the only source of old age security. With the spread of education among women, social attitudes to women, as well as the attitudes of women to themselves, underwent profound changes. It was realized that a woman need not be restricted to her age-old role of homemaker and bearer of children. Women began to participate in gainful employment which provided an alternative to childbearing and child-rearing. Education was also responsible for bringing about a rational outlook, free from religious dogma; and this rational outlook facilitated the acceptance of the idea of fertility control. Moreover, flowing from educational opportunities and rising prosperity was the aspiration on the part of the individual to rise in the social scale. Too many children were perceived to be an obstacle in the attainment of this objective to climb the social ladder; and the natural result was the limiting of the size of the family.

According to Frank W. Notestein, the noted demographer, the growth of a huge and mobile city population largely changed the corporate family way of traditional society; instead came individualism, which was characterized by increasing personal aspirations to move upward. Large families became "progressively a difficult undertaking; expensive and difficult for a population ever increasingly freed from old taboos and increasingly willing to solve its problems rather than accept them." Notestein pointed out in 1953 that the decline in fertility in the West occurred as a result of the growth of an urban industrial society. He concluded that the development of technology was the underlying factor for fertility transition. He also pointed out that "industrialization and urbanization resulted in little development of a rational and secular point of view; the growing awareness of the world and modern techniques through popular education, improved health, and the appearance of alternatives to early marriage and child-bearing as a means of livelihood and prestige of women".

The reasons for the recent declines and low current levels of fertility in most of the developed countries may be summarized as follows: (1) Development of improved methods of fertility control and increasing use of the most effective methods; (2) Liberalized abortion laws and extensive grounds and facilities for abortion; (3) Decreasing desire for large families; (4) Rising costs of rearing child; (5) The increasing trend of women's employment in paid jobs outside the home: (6) Instability and changes in the values attached to the rewards and penalties of parenthood in the context of other needs and aspirations.

FACTORS RELATED TO HIGH FERTILITY IN DEVELOPING COUNTRIES

In order to understand the factors which are responsible for high fertility, it is necessary to study the social, cultural and economic conditions prevalent in the countries with high fertility, which may then be compared with the conditions in the countries where fertility has declined. The following section considers the same factors that were identified earlier for fertility declines in low fertility countries, but in a different context.

Motivational Factors and Factors Relating to Family Structure and Functions: Two general explanations are put forward for the high fertility in some countries. The first explanation is that high fertility is a fundamental adjustment to high mortality and that high fertility is necessary for group survival when mortality is high. When infant and child mortality rates are high, this consideration becomes even more important, because to have a large number of children in those circumstances becomes necessary in view of the fact that the chances of survival of children to adulthood are slender. Even when infant and child mortality rates begin to decline following improved health services, this fact does not become immediately evident to the people.

The other explanation is that high fertility is also an adjustment "to the central importance in community life of familial and kinship ties. In preindustrial societies, all activities are centered on kinsmen and

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children and a great deal of occupational co-operation is required from them to the large tasks that are to be carried out. In fact, in such societies, economic and social relationships overlap.

The production and consumption of goods and services, leisure-time activities, assistance in illness and old age and several other activities, which are normally entrusted to various non-familial institutions in complex societies, fall in the domain of the family and kinship groups in pre-industrial societies.

In such a social structure children have a great economic, social, cultural as well as religious value. They become economically useful by the age of six or seven, and therefore are not an economic liability for their parents, but are, in fact, economic assets. In most pre-industrial societies, great importance is attached to the procreation of male children, for sons extend the family line. Amongst the Hindus, a son is essential, for only he can ceremoniously kindle the funeral pyre and thus affect the salvation of his father's soul. He is also responsible for performing religious services for his ancestors. Children are generally considered to add to the wealth and prestige of the family for with the increase in the number of children and consequent increase in relatives and grandchildren come more political power and additional economic resources for the family.

Even when children migrate to urban areas, they continue to add to the family income because of strong family ties and their sense of duty towards parents which is ingrained in traditional societies. In such societies, biological parents may not be called upon to provide for the basic needs of their children, for the family is jointly responsible for all the children born into it. There is, therefore, no economic motivation for restricting the number of children.

In most traditional societies, a fatalistic attitude to life is ingrained and fostered from childhood. Such an attitude acts as a strong influence against any action that calls for the exercise of the right of the self determination with reference to reproduction. It is for this reason that, when people are asked how many children they would like to have, they are sometimes known to reply: "It is not for us to decide," "Children are the gifts of God," etc. Religious institutions also generally promote high fertility rates which are definitely incorporated in the tenets of Hinduism, Judaism, Islam and Christianity, especially among the Roman Catholics.

Economic, Social and Other Factors: The motivational factors mentioned above are supported by the low level of economic and social development which exists in most developing countries today in spite of the fact that the process of industrialization has already commenced in these countries. In most developing countries, there is still widespread poverty, and the literacy rates are low. The status of women is also quite low, leading to their unquestioning acceptance of excessive child-bearing without any alternative avenues for self-expression. The general low level of living leads to an apathetic state of mind, and there is hardly any desire to improve the standard of life. Lack of education acts as a constraint on rational and secular living, and the influence of religious dogmas persist. The result of all these factors is that the size of the family grows without any inhibiting influences.

Taking all these factors into account, the governments of most developing countries have launched official family planning programs to educate their people to accept, in keeping with the changing times, the small family norm. Though these family planning programs cannot be a substitute for economic and social development, and are definitely not meant to be so, they can be quite effective in augmenting the control of fertility.

DIFFERENTIAL FERTILITY

It has been observed that the levels and patterns of fertility vary considerably in various sub-groups of the same population. These subgroups may be based on residence, whether urban or rural, social and economic status in terms of educational attainment, occupations, income, size of land holding, religion caste, race, etc. A study of differential fertility is useful in identifying the factors which determine fertility levels among various sub-groups. The information so provided gives us some idea of the future proportion of each group in the total population and also helps us to project more accurately the future

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population size of the entire country. A study of differential fertility is also important from the point of view of the implementation of family planning programs because it helps us to identify high fertility groups on which the program's efforts can be concentrated. In this connection, it is interesting to note that in the 1930s, when birth rates were at the lowest levels, research was concentrated on those groups which had the lowest fertility with a view to uncovering clues for the formation of policies for the encouragement of high fertility among these very groups.

Differential fertility can be understood as a result of following factors.

ECOLOGICAL FACTORS:

(c) **Regional Differences in Fertility:** The fertility rates of various regions or States or provinces within one country may differ widely. In India, for instance, there is a great deal of variation in the fertility levels of various States and Union Territories.

(d) **Rural-Urban Residence and Fertility:** Numerous studies have been conducted on fertility differentials according to rural-urban residence. Towards the end of the last century, in the low fertility areas of the world, it was found that the fertility of those residing in cities was lower than that of rural residents and these differentials were more or less stable. However, when national birth rates declined these fertility differentials widened in a more pronounced manner among urban upper classes than among others. Rural-urban differences narrowed down again when birth rates rose after the Second World War.

SOCIO-ECONOMIC FACTORS:

f. **Educational Attainment and Fertility:** The educational attainment of couples has a very strong bearing on the number of children born. Educational attainment, especially of women, is one of the indicators of modernization and the status of women in society. In low fertility countries, historically the relationship between fertility and the educational attainment of the wife has been a negative one, in the sense that the higher the educational level, the lower was the family size.

g. **Economic Status and Fertility:** General studies in the past have highlighted the inverse relationship between the economic status of the family and fertility. This traditional relationship is now undergoing substantial changes as far as the developed countries are concerned.

h. **Occupation of Husband and Fertility:** In developed countries occupation, especially that of the husband is used as an indicator of social economic status, and differential fertility is studied according to the occupation of the husband. Studies conducted in Europe around 1970 indicated that the wives of farmers and farm workers recorded a higher fertility than the wives of men engaged in non agricultural occupations. Even amongst those engaged in agriculture, the fertility of the group of farm workers was higher than that of the group of farmers. These differences were more pronounced in France and the United States than in the other countries. Manual workers were also found to have, on an average, more children than non manual workers. In India, some studies have tried to investigate the relationship between the occupation of the husband and fertility. It was generally observed that the wives of those engaged in professional jobs had the lowest fertility.

i. **Employment of Wife and Fertility:** It has been found in several studies that the gainfully employed women have a smaller number of children than those who are not employed. Though this relationship has been quite distinct in many industrialized countries, the exact nature of this relationship is not yet known. Is it that women who have smaller number of children tend to take paid jobs outside their homes, or is it that those who have already paid jobs restrict their family size to the very minimum so they can work? It is difficult to say anything very firmly.

j. **Religion, Caste, Race and Fertility:** Religion is considered to be an important factor affecting fertility. The study of differential fertility of various religions as well as ethnic groups has important social and political implications. In a democratic society where each person has the right to vote, the

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size of a particular religious, caste or ethnic group may be an important factor in determining the political power-structure. Several studies have been conducted in developed as well as developing countries to investigate the influence of affiliation to a particular religion on the fertility behavior of the people. At one time, all the religions of the world, except Buddhism, were pro-natalist or "populationist". The injunctions laid down in various religions indicate the importance of high fertility. Some illustrations are: "Be fruitful, multiply and replenish the earth" (Judaism and Christianity); "Marry a woman who holds her husband extremely dear and who is richly fruitful" (Islam); "Make the bride the mother of good and fortunate children, bless her to get ten children and make the husband the eleventh one" (Hinduism). It may however be pointed out that all these religions have their roots in the distant past, which was a period of high mortality. As such, the emphasis on prolific fertility was a functional adjustment to high mortality in order to ensure the continuation of the group. Resistance to human interference with fertility is common to all religions. As pointed out by Lorimer and Osborn this resistance has been less persistent in those religious groups where there was an absence of a central authority as in the case of Judaism and the Protestant sect of the Christian religion. This same reason, namely the absence of a central authority, may be attributed to the absence of opposition to birth control among the Hindus.

Various studies in the West, which is essentially dominated by the Christian-Hebraic tradition, show that the fertility of Catholics has exceeded that of non-Catholics in almost every country and socioeconomic group. The studies conducted in the United States and Canada have clearly pointed out that the fertility of Roman Catholics has been higher than that of either Protestants or Jews. Recent findings indicate that in most of the European countries as well as in Canada, differentials in fertility by religious affiliation are narrowing. The reason for this narrowing of differences may be due to modifications in the religious sanctions relating to the ideology and practice of birth control and the weakening of the influence of religious doctrines and traditions. It may also be that the influence of other socioeconomic factors such as urban residence, educational attainment etc. on fertility is on the wane with the result that fertility differentials are narrowing.

India may be taken as an illustration from the developing countries. In most of the studies conducted in India to study the fertility behavior of various religious groups it was observed that the Muslims had a higher fertility rate than the Hindus or the Christians.

Some sociologists are of the opinion that minority religious groups may tend to have higher fertility rates to gain more political power. This however does not hold true in all cases. Two minority communities, that is, the Zoroastrian community in India and the Jewish community in the United States, have always shown lower fertility rates than the majority group. Other socio-economic factors such as the educational attainment of the wives, the status of women, the degree of modernization of the community, etc., are found to affect the fertility of the subgroups in a particular society.

From this discussion it is obvious that it is difficult to determine the exact role of religion in influencing fertility and that the role of religion in causing fertility differentials though not negligible is quite limited.

As caste is an important indicator of social status in India, attempts have been made in several studies to observe fertility differentials by caste. In a study conducted in Lucknow City, it was observed that upper caste Hindus had, on an average 3.8 live births while the lower caste Hindus had on an average 4.1 births.

In multi-racial societies differences in fertility have been found among various racial groups. In the United States, for example, the Negroes show a higher fertility than the white community. These differences, however have been explained away by socio-economic factors.

Inter-relationships between Socio-Economic Variables Affecting Fertility: In the discussion on differential fertility only one variable at a time has been taken into consideration with a view to explaining the differences in fertility among various sub-groups in any population. It must however be

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recognized that all these variables are closely interrelated. For example, when the husbands are highly educated, they tend to have educated wives. Higher educational levels are related to higher incomes and more gainful occupations leading to higher standards of life. In a community in which women are educated, the influence of religious dogma is likely to be less rigid, while if the community has a high proportion of illiterate women, religious influences tend to be strong and affect fertility. Age at marriage is also closely associated with the educational attainment of wives.

When fertility differentials are studied in relation to various ecological, socioeconomic and cultural factors, it is found that, in many developed countries, they are narrowing, and the latest trend is towards uniformity. On the other hand, in a developing country like India, fertility differentials have in recent times become increasingly pronounced. Amos Hawley has identified three phases in the trends in fertility differentials. In the first phase, various socio-economic classes either had identical fertility rates or the relationship between socioeconomic status and fertility was positive. In the next phase, which began with industrialization and the resulting socioeconomic development, it was the highly educated urban elite group which first began to show declines in fertility thus showing a wave of fertility differentials. In the final phase, which is in existence now in many Western countries, there appears to be a positive association between socio-economic status and fertility. India is at present in the transitional stage of declining fertility and therefore differentials among various socio-economic classes are becoming increasingly more pronounced.

MORTALITY

Historically, the factor of mortality has played a dominant role in determining the growth of population, the size of which fluctuated in the past mainly in response to variations in mortality. The increase in the population of European countries following the Industrial Revolution in the seventeenth century was mainly due to a decline in the death rates. The developing countries which are undergoing a typical demographic transition have also been affected initially by the fall in the death rates. In fact, the single most important contribution of demography has been the revelation of the fact that sharp declines in mortality rates, rather than any rise in the fertility rates, have been responsible for bringing about a rapid growth of population.

The study of mortality is useful for analyzing current demographic conditions as well as for determining the prospects of potential changes in mortality conditions of the future. The public health administration depends heavily on the study of mortality, for statistics on death in the population cross-classified by age, sex and the cause of death are of great value for the formulation, implementation and evaluation of public health programs. Statistics on deaths also form the basis of the policies of insurance companies.

It is of course possible to study mortality from several angles, for various biological, social, economic and cultural factors affect the health of an individual and consequently the mortality rate in society. These factors affecting mortality can be classified under heredity, constitution and environment. When mortality is viewed from the demographic point of view, it is studied to determine changes in the population size and structure, rather than from the medical angle. The demographic study of mortality therefore does not usually take into consideration the genetic factor. The remaining two factors, namely, the constitutional and the environmental factors provide the basis of a demographic analysis of mortality. Of the various constitutional factors - including the physical, physiological, anatomical and psychological characteristics of man - the most important for the demographic study of mortality are age and sex. The environmental factors affecting mortality include the natural physical surroundings of the individual as well as his/her social and economic environments and personal habits.

CONCEPT OF MORTALITY

The study of mortality deals with the effects of death on population. Though the meanings of terms "life" and "death" are obvious, a scientific study of demographic processes calls for formal definitions.

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The United Nations and the World Health Organization have defined death as follows: "Death is the permanent disappearance of all evidence of life at any time after birth has taken place (post natal cessation of vital functions without capacity of resuscitation)." A death can thus occur only after a live birth, and the span between birth and death is life.

The above definition of death does not include any death prior to a live birth, which has been defined by the United Nations as follows: "Live birth is the complete expulsion or extraction from its mother of a product of conception, irrespective of the duration of pregnancy, which, after such separation, breathes or shows any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles, whether or not the umbilical cord has been cut or the placenta is attached; each product of such a birth is considered live-born." It is, therefore, evident that any death prior to a live birth is not considered as a death. Thus, abortions and still births are referred to, not as deaths, but as fetal deaths. Any expulsion of the fetus, either spontaneous or induced, which occurs before the fetus becomes viable, that is, capable of independent existence outside its mother, is known as an abortion. When a birth does not have any of the characteristics included in either of these two definitions of live birth or abortion, it is known as a still birth.

SEX AND AGE PATTERNS OF MORTALITY

SEX DIFFERENCES IN MORTALITY: It has been observed that in most countries of the world, mortality conditions differ for males and females. The general experience is that females have an overall advantage over males with respect to mortality. For most countries, the average expectation of life at birth is higher for females than for males. The gap between the average expectation of life for females and males is wider in the developed than in the developing countries. In developed countries over a period of about 70 years the gap between female and male average life expectancy has widened. In most countries of the world, the crude death rates as well as the age specific death rates are higher for males than for females. There is a great deal of evidence to indicate that as far as mortality is concerned, males are at a definite disadvantage.

The phenomenon of male and female longevity has been studied by several social scientists. John Graunt, the father of demography, had observed as far back as the seventeenth century "Physicians have two women patients to one man and yet more men die than women. The question which has been investigated by some demographers is whether these differences between females and males are biologically determined or whether they arise because of the different roles played by them in society.

What are the underlying factors which are responsible for the widening gap between female and male longevity? Is it again the constitutional difference between man and woman, which equips the female with better resistance to degenerative diseases? Social scientists like Esterline and Conrad conclude that there is little to support a biological explanation of the gaps between female and male longevity. In their opinion, the reason may be traced to the different roles played by them in the society. It is pointed out that man as the breadwinner of the family has to undergo more physical and emotional stresses and strains; and therefore a great deal of his physical as well as mental energy is spent in competitive struggle to maintain a higher standard of living and achieving higher social status. It is argued that the age at retirement also presents different problems for males and females. While for the typical housewife there is practically no change in her routine and habits throughout her life time, retirement involves drastic changes in the life of the breadwinner and head of the family. He has to face a loss of status and the prospect of an inactive life. These developments have significant repercussions on his emotional and physical well being. This question regarding the longevity of females and males however has yet to be fully answered. More extensive research, covering both the biological and social aspects of the problem will have to be conducted before any firm conclusion can be arrived at.

THE AGE PATTERN OF MORTALITY: Age is an important variable in any analysis of mortality, for death rates vary with age and sex. It is for this reason that any meaningful study of mortality has to be undertaken with respect to both age and sex, and the data cross classified accordingly. The age specific

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death rate is a measure that is most appropriate for this purpose and may be defined as the number of deaths of persons of a given age per 1.000 during a given year. The computation is similar to that of the crude death rate.

The typical age specific mortality curve in countries of high mortality is roughly U-shaped, which indicates that mortality is very high at both the extremes of the life span, that is, in infancy and in old age. The shape of the age specific mortality curve of countries with low mortality is roughly J - shaped, the difference in the shape of the two curves being due to the difference in the infant mortality rate of these two countries. In countries with low mortality, the age specific mortality curve has a broader base, which indicates that low mortality rates extend over a large number of age groups. A similar pattern of the age specific mortality curve is observed for females, the only difference being that the values of the age specific death rates are lower for most of the age groups because of lower female mortality.

INFANT MORTALITY: The study of infant mortality gains importance especially because mortality during the first year of life is invariably high for all countries irrespective of whether the overall levels of mortality are high or low.

The trends in infant mortality rates of developed and developing countries indicate that some overlapping of these rates has occurred between these two regions. This phenomenon has raised an important issue regarding the utility of infant mortality rate as an indicator of social and economic development. As has been earlier observed in several countries, where infant mortality rates are similar to those of developed countries have not registered any remarkable progress in social and economic development.

FACTORS AFFECTING INFANT MORTALITY: A variety of factors affecting infant mortality are customarily classified as biological and socioeconomic or environmental factors, though these two categories should not be treated as watertight compartments, for there is a great deal of interaction between the two. At times it is even possible to modify biological factors by introducing changes in socioeconomic factors. For the sake of convenience, however, these two types of factors will be discussed separately.

As has been pointed out earlier, the recent spectacular decline in general mortality has affected all age groups to some extent but the magnitude of the decline has varied at different ages. The reduction in mortality was considerably greater in the younger age groups than in the older age groups. In general it may be said that the low level of infant mortality appears to be associated with low level of general mortality.

The level of mortality is very high in the first few hours, days and weeks of life. The reasons for infant deaths at the earlier and later stages of infancy differ to a certain extent. Hence, in a study of infant mortality, infant deaths are carefully grouped into two categories according to the age at death. The first category consists of those infants who die before they complete four weeks of life. The other category consists of those infants who die between 28 days and 365 days of their life. The rate based on the first period is known as the neo-natal mortality rate, while that on the second period is referred to as the post-neo-natal mortality rate. Factors which affect fetal and neo-natal deaths are primarily endogenous, while those which affect post-neo-natal deaths are primarily exogenous.

Endogenous Factors: The endogenous factors are related to the formation of the fetus in the womb and are therefore mainly biological in nature. Among the biological factors affecting fetal and neo-natal infant mortality rates, the important ones are the age of the mother, the birth order, and the period of spacing between births, prematurity, weight at birth and the fact of multiple births.

If a graph of fetal and neo-natal mortality rates is drawn with respect to these factors, it would more or less resemble a U-shaped curve. The maturity of an infant at birth has also been found to be an important factor affecting neo-natal and infant mortality rates. It has been observed that the weight of the baby at birth is also an important factor affecting neo-natal and post-neo-natal deaths.

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It may be concluded from this discussion that the causes of fetal and neo-natal deaths so far considered arise mainly out of genetic factors, and may be traced back to the intrauterine life of the fetus and to the damage occurring during the process of birth.

Exogenous Factors: Social, cultural, economic and environmental factors are also found to affect infant mortality, especially during the post-neo-natal period. Post-neo-natal deaths are therefore mainly due to various epidemics caused by communicable diseases both of the digestive system such as diarrhea and enteritis, and of the respiratory system, such as bronchitis and pneumonia, as well as by faulty feeding patterns and poor hygiene. The underlying environmental factors include crowding and congestion, unsanitary surroundings, lack of proper sunshine and fresh air, etc.

Illegitimacy is also an important factor contributing to a high infant mortality rate. The difference between infant mortality rates of legitimate and illegitimate births is usually found to be quite marked. The reason for this difference is quite obvious. A child conceived and born out of wedlock is generally unwanted both by the mother as well as society. Consequently such a child does not receive the care, in terms of nutrition and other facilities that it needs.

One interesting feature of the role of endogenous and exogenous factors in determining infant mortality rates is worth noting. In countries where infant mortality rates are very low, a higher proportion of infant deaths occur during the neo-natal stage because being developed they have been successful in almost completely eliminating the environmental factors responsible for such deaths. The main causes of infant mortality in these countries are, therefore, mainly genetic or biological in nature. On the other hand in countries where infant mortality rate are high the majority of infant deaths occur after the neo-natal stage and are due mainly to environmental factors.

REASONS FOR HIGH MORTALITY IN THE PAST

Death rates all over the world were very high and fluctuating till the 19th century. The main reasons for such high mortality rates were: (1) Acute and chronic food shortages, causing famines and conditions of malnutrition; (2) Epidemics; (3) Recurrent wars; and (4) Poor sanitary conditions.

1. **Famines and Food Shortages:** In the pre-industrial phase, man had limited control over his environment, and his food supply was profoundly affected by changes in weather conditions, such as droughts, floods, severe winters and scorching summers. Agricultural production was also limited by other conditions such as inefficiency of labor, pests and by plant diseases. Even when harvests were good, food could not be stored for the future difficult times to come because of inefficient methods of storage, nor could it be transported, to scarcity areas, since easy and cheap means of transportation were not available. Besides acute famines, conditions of severe malnutrition, resulting from continuous insufficient food supply, prevailed in all countries, the state of malnutrition weakening millions of people to such an extent that they fell an easy prey to infection. Men suffering from food shortages, both in terms of quality and quantity, were unable to work efficiently. They thus lowered their own as well as their community's income.

2. **Epidemics:** Since the beginning, mankind has suffered from communicable diseases such as typhoid, dysentery, small-pox, malaria, typhus, tuberculosis, pneumonia, yellow fever, plague, etc. as well as from childhood communicable diseases such as enteritis, measles, whooping cough, scarlet fever, diphtheria, etc. All these diseases were quite common until fairly recent times, and took a heavy toll of life. These diseases tended to spread rapidly in densely situated areas through personal contacts, community use of contaminated water and food supply and as a result of the migration of persons and the movements of disease carrying flies from place to place. Until 150 to 200 years ago, scientific knowledge about diseases did not exist, and nobody understood how and why an outbreak of a particular epidemic took place and how and why it spread. Even in the seventeenth and early eighteenth centuries, a majority of the people did not receive any medical attention. Those who did had to expose themselves to such harmful methods as purging and bleeding.

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3. **Recurrent Wars:** Throughout mankind's history, war has been an important factor affecting the population size. The effect of war on human populations is two-fold. First, death came to military personnel in the battlefield. Soldiers also died of wounds received in battle. Deaths among military personnel were also caused by deprivation and diseases associated with wars. Some wars indirectly caused heavy civilian casualties through the spread of diseases carried by armies, through plunders and various other forms of social and economic disorganization. Hans Zinsser, the eminent epidemiologist, is of the opinion that the courses of history has been shaped less by the acts of men than by chronic and epidemic diseases with which they had to contend as well as by superstitions and ignorance concerning medicine and sanitation. In the Franco-Prussian War of 1870-71, 13,000 amputations had to be performed, of which 10,000 were fatal. Napoleon was utterly helpless in his fight against typhus, pneumonia, dysentery and scurvy.

4. **Poor Sanitary Conditions:** Throughout most of mankind's history, sanitary conditions have been extremely poor. There was very little knowledge of the medical value of cleanliness. In preindustrial times, the standard of living was low: the personal hygiene of the people was inadequate and communal sanitary facilities were absent. All these factors contributed to extremely filthy environments, leading to epidemics and all kinds of diseases. Even in the early stages of industrialization, large sections of the population lived in overcrowded dark houses close to factories. These houses were poorly-ventilated, damp and lacked light and sunshine, and had poor bathing facilities and worse toilet amenities. Working conditions in factories were appalling. The concept of personal cleanliness was practically unknown. Most people not only accepted dirt and unsanitary conditions of life as being normal, but even obstructed the introduction of hygiene. Stink and nasty odors were taken for granted. The use of soap was almost unknown in the thirteenth century.

It may thus be concluded that food shortages, various types of death dealing epidemics and unsanitary hygienic conditions resulted in high levels of mortality from the beginning of the history of mankind. In recent years, however, man has triumphed to a great extent over these factors. As a result, mortality throughout the world has substantially declined.

CAUSES OF MORTALITY DECLINE IN DEVELOPED COUNTRIES

In Europe, North America and Oceania, continuous economic progress resulting from Agricultural and Industrial Revolutions, has been the main reason for the reduction in mortality rates, which first began to decline rather weakly in the seventeenth century, and then with an increasing tempo throughout the eighteenth and nineteenth centuries. In the twentieth century, mortality has continued to decline at a slower rate. Some ten factors have been enumerated as having contributed towards the reduction in mortality rates in the developed Countries.

One important development affecting mortality in European countries since the eighteenth century was the increase in the supply of food. With the Agricultural Revolution, which began in England around 1700 and spread throughout Europe and the European settlements abroad, the productivity of land and labor began to increase, the risk of crop failure was reduced and the supply of food became fairly steady. The system of rotation of crops, the development of improved strains of plants and animals, the introduction of new crops and improved farm machinery were some of the important outcomes of the agricultural revolution. With the increase in the quantity and quality of food, mortality rates in Europe and European overseas settlements came down. Further advances in agricultural technology during the nineteenth and the twentieth centuries, which included the introduction of artificial fertilizers and the mechanization of agriculture freed labor from the necessity of working on land for food production, so that they could engage themselves in different types of manufacturing activities.

With the development of the Steam engine, it became possible to transport and distribute surplus food from one area to another where scarcity conditions prevailed. The frequency of localized famines was thus arrested. Better methods of storing food also helped to mitigate the serious effects of famines.

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The real income of the people gradually increased, and the availability of enough nutritious food brought about mortality declines. In fact better diet was probably the main cause of the substantial fall in mortality from tuberculosis in England and Wales.

The advances in technology and improvements in the standards of living aided the fall in mortality in many other ways. Heavy and better clothing to combat severe winters became available and health services and medical research activities were extended.

Another important factor which contributed to the reduction in the mortality was improvements in sanitary conditions and public health measures. Sanitary reforms were introduced in England in the nineteenth century following the Sanitary Reform Movement which was started to combat the many evils of the Industrial Revolution. State Governments, which had so far not taken much interest in introducing sanitary reforms, began to do so by introducing several public health measures and sanitary improvements, such public utilities for the provision of water supply, purification of water, sewage disposal, etc. By the beginning of the twentieth century, most of the cities in the Western world and Russia had developed the drainage system and provided pure and protected water. The disinfection of water by chlorination came into practice at the turn of the century, with the result that many communicable waterborne diseases such as cholera, diarrhea and dysentery were brought under control.

Social reforms in the nineteenth and twentieth centuries lessened the hazards of working in the factory early in life. Legislations bearing on the number of working hours and minimum wages was enacted, and working conditions improved because of the various safety devices that were introduced. The social security system provided benefits like old-age pensions, health insurance, medical care, unemployment insurance, etc. The introduction of free and universal compulsory schooling resulted in mass literacy and the inclusion of personal and social hygiene in school curricula helped in creating a greater awareness of the necessity of health among the people.

In the late nineteenth century, the development of asepsis (precautionary exclusion of pathogenic micro organisms) and antisepsis (killing or inhibiting the growth of micro organisms already present) helped in the reduction of mortality. Aseptic and antiseptic surgery was introduced and the germ theory was accepted in various other aspects of life such as preservation of food through pasteurization and sterilization.

The development of immunology was yet another cause of the declines in mortality. Immunization against small-pox was introduced first. This was followed by the development of vaccines for chickenpox, cholera, sheep anthrax, hydrophobia and diphtheria. Prophylactic antitoxins were also introduced against tetanus, typhoid, yellow fever, scarlet fever, poliomyelitis, influenza, Measles, whooping cough etc.

Advances in chemotherapy (use of drugs to cure or inhibit the progress of diseases) began in the late 1930s. Sulfonamides and penicillin in the treatment of respiratory and urinary tract infections came to be widely used in the control and cure of various kinds of diseases. As a consequence of the use of these broad spectrum antibiotics, mortality from communicable diseases declined substantially. After the Second World War, there have been considerable declines in mortality following the use of anti-tuberculosis drugs. The development of effective insecticides such as DDT has greatly reduced the incidence of insect-borne diseases such as typhus and malaria. Anti malaria campaigns have practically wiped out this disease from many countries. It has also indicated that there has been a change in the virulence of the disease-causing organisms and/or an increase in the resistance of the human host. Mortality from small-pox, scarlet fever and diphtheria is believed by some writers to be affected by such factors. The reason for the disappearance of one of the biggest killers, plague, from Western Europe since the middle of the seventeenth century has continued to remain a mystery.

FACTORS RESPONSIBLE FOR RECENT MORTALITY DECLINES IN DEVELOPING COUNTRIES

The rapid declines in mortality experienced by many developing countries after the Second World War have been largely due to public health and disease-control measures, which were imported from the

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developed countries and therefore have been independent of economic development. These include DDT spraying; the use of antibiotics like penicillin and vaccines like the B.C.G. Scientific communication and international co-operation have made it possible for developing countries to import techniques developed by the economically advanced countries and apply them in mass public health programs at a relatively small cost. The assistance provided by the World Health Organization to eradicate such mass killers as malaria, small-pox, etc., has been helpful. Thus, though the inverse relationship between economic development and mortality continues to hold true, the link between the two has considerably weakened. It should however be noted that further declines in mortality in the developing countries will depend to a large extent on their economic development which will help them -to get the maximum benefits from health programs.

MORTALITY DIFFERENTIALS: Important variations in the levels of mortality are evident for different sub-groups of the population even in the same country. For instance, the rural areas and urban areas of the same country have widely different death rates. In addition to mortality differentials by geographical residence, differentials due to other demographic and socioeconomic factors may also be observed within the boundaries of a particular country. Such factors include occupation, income level, educational attainment, sex, age and marital status.

URBAN-RURAL DIFFERENTIALS: The limited available data on this point indicates that the urban-rural differences in mortality levels are at present very small in industrialized countries. In the past however important differences in the mortality levels of urban and rural areas existed in these countries. Prior to the twentieth century, mortality was generally higher for the urban areas of many countries of Europe and North America and the differences were usually quite substantial. In the United States of America, mortality conditions in 1830 were far worse in large cities than in small cities or in rural areas. In England and Wales, the experience was similar. In 1841, while the average expectation of life for England and Wales was about 40 years, it was 35 years for London and 35 years and 24 years respectively for Liverpool and Manchester - all Industrial cities. In India, according to the Nineteenth Round of the National Sample Survey (July 1964 to July 1965), the crude death rates for rural and urban areas were 13.01 and 7.97 per thousand population respectively, indicating a lower mortality for urban India. The Sample Registration System also indicates that, for the period July 1974-June 1975, the urban and rural crude death rates were 9.6 and 16.1 per thousand of population respectively.

OCCUPATIONAL AND OTHER DIFFERENTIALS: It is now well recognized that man's surroundings and the occupation from which he earns his livelihood have an important bearing on his health habits status. The study of the relationship between a man's occupation and the risk of mortality is important from many points of view. A man's occupation is related to his education; his income depends on his occupation. Both income and education may influence his diet, his housing conditions and habits. As it is not feasible to study individually the effects of all these factors on mortality, researchers have often taken one factor as an index of all the others. The factor most studied in this context is occupation, or social status based on occupation. In England and Wales, systematic studies since 1851 have been conducted on mortality differentials based on occupational class and social class. An analysis of the data for five social classes covered in these studies indicates that in 1921-23 and 1930-32 mortality rates for males aged 20 to 64 years rose progressively from social class I (professional) to social class V (unskilled). By 1950 however a change was observed in these relationships, and males in social class II (intermediate occupations) were found to have the lowest rates' while social class IV (partly skilled occupations) however continued as before to have a substantially higher mortality than the other classes. It is obvious that persons working in different occupations are exposed to different types of risks. For instance, while workers in coal mines are more likely to suffer from tuberculosis of the lungs and lung cancer, those in sedentary occupations face the risk of heart diseases.

The educational attainment of parents, especially that of mothers, has been found to have a significant relationship with the levels of infant mortality. In Hungary a strong inverse relationship was noted between the educational status of parents and the levels of infant mortality. For infants of mothers who had never attended school, this rate was 95 per thousand births, while for those with mothers who had

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thirteen or more years of schooling it was 27. In Greater Bombay it was observed in 1966 that the levels of infant mortality were highly influenced by the literacy and educational attainment of the mothers. The infant mortality rate was the highest for infants of illiterate mothers and the lowest for infants whose mothers had either passed the matriculation examination or had higher educations.

MARITAL STATUS AND MORTALITY: In countries where studies on mortality in relation to marital status have been conducted it was found that mortality rates were always lower for married males and females than for the unmarried of the same sex and age. According to demographers and sociologists, the reasons for this phenomenon may be found in the fact that marriages are selective with respect to the health status of persons for those who are healthy are more likely to get married, with the result that the risk of dying is also less. Besides, married persons are generally more secure and protected, and they usually lead a more sober life than those who are unmarried. All such factors are thought to contribute to lower mortality rates among married persons.

SOCIAL CONSEQUENCES OF MORTALITY

It is possible that the dramatic decline in mortality since the end of the 19th century has evoked some changes in social structure than any other single development of the period. However, there has been so little research that any discussion must be speculative.

Contemporary citizens of developed nations rarely encounter death, except among the aged. This situation contrasts greatly to that which prevailed in these nations formerly. In the following paragraphs it is attempted to discuss some social consequences of mortality.

1. **Changes in the Institutions of Mourning:** A seemingly direct consequence of the reduction in the frequency of bereavement is a decline in the institutions of mourning. In his book Death, Grief and Mourning, the English anthropologist Geoffrey Gorer points out that at the beginning of the 20th century there were strict rules of etiquette that the bereaved must observe toward others and that others should show toward him. At present, however, neither the bereaved nor the circle of his acquaintances knows quite how to act toward the other, and in fact a common reaction is to try to deny the very existence of bereavement.

2. **Impact on the Character of Religion:** Another apparent consequence of decline in mortality is a change in the character of religion. In their book Popular Religion, Schneider and Dornbusch point out a sharp decline in the emphasis placed on how religion will benefit one in the next world, and a marked increase in the emphasis on how religion will aid one in this world. Evidently, the intensity of popular feeling concerning an afterlife has waned. Probably a very important reason for wanting an afterlife is to reunite oneself with friends and relations who have already died. In a high-mortality society, persons of all ages have many close friends and relatives who have recently died; in a low-mortality society, only the elderly find themselves in this position. Thus mortality decline should reduce the general concern with immortality. It is also possible that extremely negative attitudes towards traditional religions may also abate with the reduction of mortality. This seemingly self-contradictory action might be based thus: supposing that the experience of prematurely loosing one's parent, spouse, or child would provoke in certain individuals severe doubt that there can be a deity who is both benevolent and omnipotent, the smaller the number of persons who have such an experience, the fewer will be the number who will develop or tend to cling to extremely negative attitudes toward such a belief.

3. **Changes in the Family Structure:** A third possible consequence of mortality decline may be a change in family structure. When there is a large probability of early widowhood and orphan hood, it is hazardous for a nuclear family - i.e., a married couple and its children - to isolate itself too far from its kin group. This is because the death of either the father or the mother would make it very difficult for the orphaned children to receive proper rearing and support. Thus, in high-mortality societies we commonly see the nuclear family strongly dependent on some larger kin group. By contrast, in the low mortality developed nations of today, the nuclear family often lives at a considerable distance

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from other kin, and its ties with relatives, although present, are considerably weaker than they would be in a society with high mortality. In turn, the possibility of a larger number of relatively isolated nuclear families has important implications for the process of economic development. A high economic level is not possible without an elaborate division of labor, and much geographical mobility is necessary if very specialized occupational positions are to be filled by the best possible people. Hence, a high level of mortality, by impeding the possibility of a relatively isolated and independent nuclear family, also hinders the process of economic development.

4. **Changes in the Intensity of Interpersonal Ties:** A fourth possible consequence of high mortality may be reduced intensity of certain interpersonal ties. In a society wherein many children will die before reaching the age of five, parents may frequently steel themselves for the possibility of their child's early death by forbidding themselves to develop a strong emotional attachment. The same may also apply to marriage. Arranged marriages are common in high mortality societies, whereas in societies with low mortality, marriage is commonly contracted by free choice to a person for whom one has a strong emotional commitment. In a society in which a strong love relationship might soon be disrupted by death, there should be less dissatisfaction with a system of arranged marriages than in societies where mortality is low. Conversely, one may speculate that the pressure for easy divorce may increase as mortality declines, since the number of years one must expect to live jointly with one's spouse becomes so much longer.

5. **Difference in Orientation in Time:** It may be hypothesized that when mortality is high, individuals tend to have a weaker orientation toward the future and a stronger orientation toward the present than when mortality is low. If so, this may have further effects on the degree of achievement motivation in the society, since achievement always involves a sacrifice of present values for future goals. Furthermore, where mortality is high, parents may be loath to make sacrifices for the future success of their children, since the probability of the child's living to maturity is by no means certain.

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12. APPLICATIONS OF ANTHROPOLOGY

ANTHROPOLOGY OF SPORTS

Sports have developed to be a part of human culture as recreational activities. Though these activities constitute the cultural aspects, the biological aspects of man considerably influence the performance in any sporting event. Physical factors like body size, body proportions, physique, and nutrition influence the performance. Most of these traits are acquired through heredity and are also influenced by environment to a considerable extent. Apart from this, human psychological factors like motivation, training and nature also play a vital role in molding the "sportive personality".

The above discussion makes it obvious that it requires an inter-disciplinary approach, combining human biological, cultural and psychological aspects, to understand the environment of sports. This is the foundation for anthropological role in the field of sports.

The study of sports is a specialized sub-discipline in physical anthropology. This specialization is named "Kinanthropometry". It evaluates physical structure of individuals in relation to gross motor functions. It also includes the study of such aspects as maturation, nutrition and body composition.

The term "kinanthropometry" was coined by Bill Ross in 1972. It was first considered in Olympic Scientific Congress at Quebec in 1976, prior to Montreal Olympic Games in 1978. The driving force behind most initiatives for development of kinanthropometry has been UNESCO when it founded an International Working Group on kinanthropometry at Brasilia. This working group has been working under the International Council of Sports Science and Physical Education, a division of UNESCO.

There are various factors that influence the performance of an individual in sports which depend on both heredity and environment. However, genetics has a greater role to play in the formation of a phenotype. Phenotypic variations in size, physique, body composition, metabolic powers, strength, speed and skill, cardio-vascular adaptations are governing forces behind a sportsman's performance. Environment can shape a genotype into a fit type by way of training and motivation. Kinanthropometry aims at selecting the fit genotypes which help individuals attain their fullest potentialities.

Apart from muscle strength, a sport will also need coordinated body movements. Anthropometric studies of individuals can help select players who can have better potentialities in a particular sport than others. It has been observed that training and other extragenic influences can change one's morphological status only within the narrow limits set by genotype. It is difficult to change the capacity of the genotype in order to maintain altered levels of different bio-chemical determinants. It is thus imperative to lay more emphasis on the genetically determined morpho-physiological status of the individual.

By making use of the techniques of kinanthropometry - a comprehensive approach to assess an individual's physique - anthropologists were able to classify humans into different somatotypes and suggest the correct sport for them. Koherausch made the following classification in 1929.

1. **Slender type**: Best runners, jumpers and hurdlers.
2. **Medium type**: Best boxers, ball players and swimmers.
3. **Massive type**: Best weight lifters, throwers and wrestlers.

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In 1931 Arnold provided a classification of sports events and specified morphological characteristics within those types.

i) **Gymnast type**: Individuals with relatively long hulk coupled with breadth.

ii) **Wrestler type**: Individuals with mighty forms, with great breadth of shoulders and pelvis coupled with greater depth and breadth of chest.

iii) **Pentathlon type**: Individuals with medium to slender body types with relatively longer legs and less body breadth.

The composition of body is also an important morphological characteristic to be considered for sports. The body composition is well conceived when we consider the muscular, skeletal, fatty tissues. The body composition is dependent on the environmental influence, sex, socio-economic conditions, occupation, genetic make-up, nutrition and exercise.

Body composition studies on sportsmen have great importance. It has been observed that those athletes who are less fatty but are heavy due to musculature perform better in certain competitive sports; while those with substantial amount of fatty tissue require increased energy due to inert weight, thus causing endurance in activities like jumping, running etc. But in events like swimming and other water sports, the moderate quantity of fat aids performance by providing extra buoyancy and reduction of heat loss.

Apart from physique and body composition, the favorability of one somatotype over other depends on flexibility of training, motivation factors and the psyche. Anthropology also is helpful in redesigning sports equipment to suit the particular somatotypes.

NUTRITIONAL ANTHROPOLOGY

Anthropology has been contributing enormously to the field of nutritional sciences. It has been especially contributing to defining the nutritional status of persons, especially by making use of the techniques of anthropometry. This area of defining the nutritional status is especially significant because the earlier methods were highly technical and were waiting for an internal consensus for their viable use. The significance of anthropological role and the underlying assumption for use of anthropometry in nutritional assessment of population lies in the fact that though heredity contributes to growth, the genotype is capable of different growth potentials in different environments. This led to the development of specialized sub-discipline within applied physical anthropology, called **Nutritional Anthropometry**.

Nutritional Anthropometry makes use of three basic measurements - Age, Weight and Height. By use of these three basic values, anthropologists calculate the indices and compare these indices with reference population or person.

There are basically four indices to calculate nutritional status. Each index is given a specific nomenclature.

1. Weight Height Index:

- It represents the weight of the observed person compared with that of a reference person of same height.
- This index is also called Wasting when it is deficit.

2. Height for Age Index:

- It represents the height of the observed person compared with that of a reference person of same age.
- Deficiency in this index is known as Stunting.

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3. **Weight for Age Index:**

- It represents the weight of the observed person compared with that of a reference person of same age.
- Deficiency in this index is known as underweight.

These three indices are commonly used for nutritional assessment of children. There exists a geographical variation in the values of these indices. The fourth index, however, is also used for adults.

4. **Body-Mass Index (BMI):**

- This is an index of weight/height (wt/ht) in which weight is taken in grams and height in centimeters.

Nutritional anthropologists have provided indicators in terms of cut-off points to assess nutritional status.

INDICES	NOMENCLATURE FOR DEFICIT INDEX	PERCENTAGE OF REFERENCE MEDIAN
Weight – for – Height	Wasting	< 80%
Height – for – Age	Stunting	< 90%
Weight – for – Age	Underweight	< 80%

From the above table it is clear that anthropologists gave 80% as a cut-off point for weight-for-height and weight-for-age indices. However, it is 90% for height-for-age index because any serious reason for stunted growth might interfere with life. Thus the cut-off point for the index is raised.

Apart from this, anthropologists have also provided a scale of critical limits for weight-for-height index, for easy assessment of nutritional status.

Form of Malnutrition	Weight for Height Index
Severe under-nutrition	< 60%
Mild under-nutrition	60-80%
Normal	80-110%
Mild form of over-weight	110-120%
Over weight	120-130%
Obesity	130-140%
Severe obesity	140% - +

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DESIGNING OF DEFENSE AND OTHER EQUIPMENTS

Anthropometry is the branch of physical anthropology concerned with the measurement of the human body. Anthropometric surveys provide information on the range and variation of body shape and size and they are important issues because they can significantly affect the utility of equipment, clothing, or work space.

There was a time when equipments were designed with little concern for the physical characteristic of the users. Physical anthropologists as experts of human anatomy were first involved in the designing of defense equipments during World War II. Since then anthropometric research has played significant role in engineering design of many technologies, from jet-fighter ejection seats to analyzing human posture in zero gravity based Skylab experiences.

The term Anthropometry was coined by Queflect (1871). It was however Martin, a German Scientist, who published in 1928 the famous title "Lehubuch der Anthropologie". The revised edition of this book in three volumes, co-authored by Salter is still a significant work (1957). Knussmann (1990, 1992) has edited two volumes based on Martin's work.

Designing of any product or equipment considering human variability is a complex one, and it needs participation of three groups of people, viz. the users, the anthropologists and the manufacturers. Role of anthropologists is crucial, particularly if the efficiency of equipment is dependent on human variability. Anthropologists are supposed to provide basic data on human variability in such a simple manner that it is easy to understand and ready to apply.

Three factors that collectively determine the quality of man-machine relationship are efficiency, safety and comfort. Designs that do not consider human variations lead to poor job performance, low job-satisfaction, waste of time and increased morbidity.

Gun Turrets: Anthropometric data has had been very reliably taken and intelligently applied by anthropologists for Air force. It improved flying efficiency of the pilots thus saving much money on procurement of large number of pilots. After 1942, its spread to other fields of human activities has improved work efficiency by reducing discomfort of people. It is not that anthropometric data has not been used in military services, but it was primarily used for physical or medical description.

A gun-turret is a movable enclosure containing the gunner, wearing protective clothing and equipment, a pair of machine guns and a sighting mechanism. The gun turret is so designed that the gunner has all the free movement of his body needed. It has to be scientifically designed because any extrusion from an aircraft adds air-resistance. Hence, such areas as offering resistance must be reduced to minimum without compromising efficiency of the gunner.

Such improvements in US gun-turrets greatly increased efficiency of crewmen, reduced their discomfort of long occupancy in a cramped enclosure and insured effective means of escape from an aircraft in emergency, or in removal of a casualty. With the increased efficiency of crewmen, the losses to the air force were effectively curtailed.

Cockpit Size and Seat Configuration: Scientists have made study of cockpit space and established parameters for cockpit size in different types of air-craft. They were also instrumental in designing various seat configurations for both fighters and bombers. Such improvisation largely aided in reducing cockpit fatigue and discomfort by proper body support.

Flight Clothing: Application of anthropometric data in the flight clothing has been vital. Individual designers have their own schedule of sizes so that clothing sometimes fit no one. Flying Helmets is one such problem. To provide correct size-control, anthropologists have sculptor-carved wooden head forms

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in four statistically derived sizes extra-large, large, medium and small. Sets of these head forms are supplied to helmet manufacturers as standards.

Physical anthropologists are also concerned with designing of oxygen masks and make its correct fit with the help of a set of seven statistical sizes and shapes of sculptured face forms. Similar is the case with the garments. Body sizes of females are also taken to procure flight clothing and other garments for service women.

Such anthropological efforts bring about two sided advantage - operational efficiency and economic benefits - the latter to agencies such as governments and manufacturers. Such studies indicate the vast number of sizes in which a garment would have to be fabricated, the values of dimensions necessary for each size and the number of garments required in each size.

Jet Engines: Anthropometric engineering is applied in the Jet engines and it is perhaps the most important engineering program. The jets fly over altitude above 50,000 feet. At such altitudes, human body can swell up due to reduced atmospheric pressure. Dr. J.P. Henry, a medical physiologist, invented a concept called the "Partial Pressure Suit" - a one piece perfectly fitting, non-stretch garment with air tubes connected to it so that when air pressure dropped, air could be introduced in the spaces within the clothing that could prevent muscles from expanding. The unit served the purpose but there were severe sizing dilemmas. Each suit has to fit like skin from neck to wrists and ankles, but there were no anthropological data.

Once the garments are available a "fit test" is conducted and they are checked for conformity to the specified dimensions by trying them on a sample of fifty or more subjects, selected to show the full range of body size in the target population. The subject after he dons it goes through all the motions required to show whether that size actually is comfortably fit.

It was found that stature and weight generally yield the highest correlations with other bodily dimensions and could become the diagnostic dimensions for complex fitting garments. Numerous sizing programs have been tested in the forces of all the countries the world over.

The Ejection Seat and Car Passenger Safety: Originally this used to be a simple metal bucket seat mounted in the aircraft so that in an emergency, the pilot could fire the seat and himself out of the aircraft, and after freeing himself from the seat, takes to his parachute. This seat was invented by the Germans during World War II.

Any such device must take into account the centre of gravity to avoid excessive rotation of the seat as it enters the air stream at high speed. The centre of gravity is determined by the man-equipment-seat combination. Since limbs are flayed in the air, it is important to know the centre of gravity of the limbs. For such purposes, centre of gravity of limbs are known from a large sample of population. Later on, many studies were conducted for knowing moments of inertia of living subjects in a variety of fixed positions on a compound pendulum, nude and with full-pressure suit. Such data has helped refine crew accommodation in the space capsules as well as cockpits and seats of advance fighter aircrafts and automobiles. This has reduced the severity of damage during accidents.

Design Requirements: Design requirements may be classified into three groups:

a) Work space design

b) Clothing and personal equipment design

c) Component and devices

Workspace design includes designing of any space for human occupancy during work, recreation, rest, education, travel, treatment etc. Designing aims to ensure that operator has adequate work space and proper location of controls, displays and devices. Designing of automobile interior, aircraft cockpits,

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seating apparatus, doors, tunnels etc. are some of the examples where workspace designing is needed. The measurements required in designing workspace include reach limits, body clearance, eye location etc.

Clothing and personal equipment design includes designing of garments, press suits, helmets and gloves etc. The designing of such things assure proper fit and minimize restriction of movements. The body measurements that is generally required for designing clothing and personal equipment includes circumferences, body contours, limb movements etc.

Designing of components and devices include designing of small appliances such as knobs, levers, switches, handholds etc.

FORENSIC ANTHROPOLOGY, METHODS AND PRINCIPLES OF PERSONAL IDENTIFICATION AND RECONSTRUCTION

The term "Forensic Anthropology" implies the application of anthropological and medical knowledge to questions of law. This science is used in detection of crime. The term comes from the Latin word "*Forensis*" which means "Court of Law".

Forensic anthropology is an applied specialization within physical anthropology. Physical anthropologists, with their insight into the human anatomy would be able to contribute considerably in the field of crime. Forensic anthropology, as an applied discipline in physical anthropology, was recognized by C.G. Show in 1972, and he defines the role of physical anthropologist as *"a person with specialized knowledge of human sexual, racial, age and individual variations to the problems of medical jurisprudence".*

The two most important aspects of Forensic Anthropology are...

1. The identification of decomposed or mutilated bodies.
2. Analysis of skeletal and fragmentary remains.

According to **Chaurasia**, the Forensic Anthropologist tries to answer the following questions while analyzing the remains at the crime scene.

1. Whether the bones are human or non-human?
2. Whether they belong to one or many humans?
3. What would be the sex?
4. What is the age?
5. What is the race of the person?
6. What is the cause of death?
7. What is the time of death?
8. What are the other distinguishing characteristics in the skeletal remains that may lead to personal identification?

The evidences for the Forensic anthropologist to work include bone, teeth and personal belongings. Apart from this, the Forensic anthropologist makes use of his techniques in genetics (e.g., DNA Fingerprinting) to identify the victim as well as the culprit. Even the trivial proportions of Saliva, Semen, blood or scratches of skin tissue can come handy for Forensic anthropologist to identify the persons involved.

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Relevance of Anthropology in Crime Studies

An anthropologist is an experienced specialist in the field of comparative anatomy. Their knowledge in the field of understanding human evolution and non-human primates has been of considerable help in the field of crime analysis. Their specialization in understanding racial affiliation, construction of a stature from broken bones and assessment of understanding postmortem skeletal alteration helps crime studies for a positive identification. Apart from this, an anthropologist's specialty in excavation techniques and mapping, have made a considerable change in field of evidence recovery.

Forensic anthropology, as a specialized field of physical anthropologist, made a considerable progress since last three decades. Much of its growth depends on new developments in its methodological areas. There is a growing need for associating anthropologists in field of evidence discovery and recovery in the modern world and this trend continues to grow in the near future.

PERSONAL RECONSTRUCTION AND IDENTIFICATION

IDENTIFICATION OF SEX

Sexual dimorphism in the pelvic girdle of the female is seen owing to the natural selection of birth process and larger heads during birth.

Pelvic Girdle	Typical Male	Typical Female
Sub Pubic Angle	< 90 Degrees	> 90 Degrees
Pubic Shape	Triangular	Rectangular
Sub Pubic Angle Shape	Convex	Concave
Greater Sciatic Notch	< 68 Degrees	> 68 Degrees
Sacrum	Small & Curved	Large & Straighter

Males are usually larger and have more rugged areas for muscle attachments than do females of the same species. However, populations differ in skeletal size and robusticity - Asian Indians are smaller and more gracile than Australian aborigines.

DIMORPHISM IN CRANIUM

Cranium	Typical Male	Typical Female
Muscle Attachment Areas	More Pronounced	Less Pronounced
Supra Orbital Torus (Ridge)	More Pronounced	Less Pronounced
Frontal Bone	Slanting	Globular
Supra Orbital Rim	Rounded	Sharp
Palate	Deep	Shallow

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DETERMINATION OF AGE

- During growth, the skeleton and dentition undergo regular changes that allow the determination of age at death

Dental Eruption

- The determination of ages at which the deciduous and permanent dentition erupts is useful in identifying age to approximately 15 years
- The third molar (wisdom tooth) erupts after this time, but is so variable in age of eruption (if it erupts at all) that it is not a very reliable age indicator

Bone Growth

- Postcranial bones are preceded by a cartilage model that is gradually replaced by bone, both in the primary growth centers (diaphyses) and in the secondary centers (the ends of the bones, or epiphyses)
- The initial ossification centers are very small, and are only rarely encountered by forensic anthropologist
- The bone continues to grow until the epiphyses fuse with diaphyses
- Because this fusion occurs at different times in different bones, the age of an individual can be determined by which epiphyses have fused and which have not
- The characteristic undulating appearance of the unfused surfaces of bone helps differentiate it from the mature long bone (smooth) or merely a broken end of a bone (sharp and jagged)
- Females mature more quickly than males, so usually 1 to 2 years must be subtracted from the ages
- Once a person has reached physiological maturity (by early 20s), the determination of age becomes more difficult
- Several techniques are used, including
    - Progressive changes in the pubic symphyseal face (the most common technique) in sternal ends of the ribs
    - Ectocranial (outside the cranium)and endocranial (inside the cranium) suture closures
    - Cellular changes determined by microscopic examination of the cross section of various long bones
- Degenerative changes, including arthritis and osteoporosis, can aid in the determination of relative age, but should not be used by themselves, as injury and certain diseases can cause changes that mimic old age in bones

Pubic Symphyseal Face

- The pubic symphyseal face in the young is characterized by a surface with ridges and furrows but undergoes regular metamorphosis from age 18
- By mid-30s it has a more finely grained face and perhaps still containing remnants of the ridge and furrow system

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- By mid 50s and 60s, there will be bony outgrowths often developing on the outer rims of the symphyseal face
- This was developed by T.W. Todd, McKern and Stewart

DETERMINATION OF RACE

- Racial identification is usually difficult because for many traits there is more variation within races than between races

FEATURE	NEGROID	CAUCASOID	MONGOLOID
Incisors	Blade	Rarely Shoveled	Shoveled
Cranial Shape	Dolicocranic	Mesocranic	Brachycranic
Nasal Root	Wide, Rounded	Narrow, Pinched	Medium, Tented
Nasal Aperture	Platyrrhiny	Leptorrhiny	Mesorrhiny
Zygomatic Bone	Medium	Retreating	Projecting
Ear Opening	Round	Round	Oval
Facial Shape	Prognathic	Orthognathic	Medium

ESTIMATION OF STATURE

Stature is usually calculated by measuring the length of long bones by Trotter, Gleser and Genoves. Because these formulae are derived for each sex of various racial groups, the sex and race of the unknown individual must be known before formulae can be reliably applied.

Femur Length (mm)	Stature (cm)
452	169
456	170
461	171
465	172
469	173

(Data drawn from White Males with known stature at time of death)

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PHOTO SUPERIMPOSITION

- Photo superimposition is employed when a probable identification has been made
- The skull is superimposed onto a photograph of the known individual
- If enough landmarks fall on the same position on the skull and on the photograph, the forensic scientist has made a identification
- This technique is also used to reinforce the decision made in some of the cases

FACIAL RECONSTRUCTION IN HUMAN IDENTIFICATION

- Facial reconstruction, also termed facial reproduction is a process used when other identification procedures have been unsuccessful
- Two different methods of producing a face on the skull are employed
  - A portrait of the individual using clues from the bones of the face
  - A more direct 3D method of applying clay to the skull
- These techniques employ both science and art
- The physical anthropologist discovers the age, sex and race of the skull, but there is no direct evidence from bone that indicates the eye color, hair color and style, lip form, or degree of wrinkling or fleshiness in the individual
- There is a great deal of subjectivity in the rendering of the finished product,
- An exact reproduction is however not expected and only a likeness is achieved
- Erasers or blocks of clay marking tissue depths are commonly glued to the skull (depths are arrived at experimentally from cadavers)
- Clay strips, graduated to the various tissue depths, then "fill in the dots" between erasers and the face is "fleshed out"
- The eyes, nose and sometimes ears are then fashioned according to various guidelines, and a wig is usually added
- If required, features for aging are also added

APPLIED HUMAN GENETICS

PATERNITY DIAGNOSIS

A parentage test is conducted to determine whether two individuals have a biological parent-child relationship. A paternity test establishes genetic proof whether a man is the biological father of an individual, and a maternity test establishes whether a woman is the biological mother of an individual.

DNA analysis (known as genetic fingerprinting of individuals), offers a more reliable way to determine the genetic parent, although older methods have included ABO blood group typing, analysis of various other proteins and enzymes, or using HLA antigens. The current techniques for paternity testing are using polymerase chain reaction (PCR) and restriction fragment length polymorphism (RFLP).

DNA testing is currently the most advanced and accurate technology to determine parentage. In a DNA parentage test, the probability of parentage is 0% when the alleged parent is not biologically related to

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the child and the probability of paternity typically greater than 99.9% when the alleged parent is biologically related to the child. More information on DNA Profiling is already presented in the relevant topic in this volume.

Postnatal DNA testing:

- Blood collection and testing
- Buccal swab (cheek swab) collection and testing
- Umbilical cord collection and testing
- Other sample collection and testing (semen, tissue, hair, etc.)

Prenatal DNA testing:

- **Amniocentesis:** This test is performed in the second trimester, anywhere from the 14th-20th weeks of pregnancy. During this procedure, the doctor uses ultrasound to guide a thin needle into the uterus, through the abdomen. The needle draws out a small amount of amniotic fluid, which is tested. Risks include a small chance of harming the baby and miscarriage. Other side effects may include cramping, leaking of amniotic fluid, and vaginal bleeding.
- **Chorionic Villus Sampling (CVS):** This test consists of a thin needle or tube which a doctor inserts from the vagina, through the cervix, guided by an ultrasound, to obtain chorionic villi. Chorionic villi are little finger-like pieces of tissue attached to the wall of the uterus. The chorionic villi and the fetus come from the same fertilized egg, and have the same genetic makeup. This testing can be done earlier in pregnancy from the 10th-13th weeks.

GENETIC COUNSELING

Genetic counseling is already discussed in the chapter on Human Gentics in this volume.

EUGENICS

Eugenics may be considered to be the social attitude that aims at improving the quality of future genetic constitutions by restraints of production. This goal has been the plea of many well known geneticists. Common sense dictates that persons endowed with good or desirable qualities of body, mind and social attitudes should populate the earth at a greater rate, while those with inferior traits should desist from adding to the load of deleterious genes. However, the difficulty and the danger in using planned mating to improve the human race arise because of the fundamental uncertainty as to what constitutes an improvement. The specifications for what may be regarded as "superior" qualities are likely to be highly subjective.

Eugenics deals with the application of the laws of genetics to the improvement of human race. The term "eugenics" was coined by an English scientist Francis Galton in 1885. The science of eugenics can also be defined as a science of well born, improving the inborn qualities of race and obtaining the better heritage by judicious breeding.

Eugenics and Euthenics: The betterment of human society can be achieved by following two inter-related methods:

1. By one of the method we can deal with the already existing human beings. The improvement of already existing human beings can be achieved by improving the environmental conditions, e.g., by subjecting them to better nutrition, better unpolluted ecological conditions, better education and sufficient amount of medical facilities. This type of method of improving the human race is known as "Euthenics".

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2. By another method, we can improve the future generations by improving the germplasm of existing individuals. This type of method is known as "Eugenics". Eugenics believes in artificial selection of physically and mentally sound individuals and discouragement of defective individuals for the inheritance of their defective germplasm to the future generations. In other words, eugenics seeks the measures to preserve the best type of germplasm and to eliminate defective germplasm from the human society by applying the laws of inheritance to man.

Basis of Eugenics: The science of eugenics roots largely in the fact that the human being is an animal subject to the same laws and principles that govern the rest of the animal kingdom. It has been observed that the basic genetic mechanism of man, mice, higher plants and molds appear to be the same and in principle, the genetics of man is not unlike the genetics of a virus.

For a long time, man was considered a peculiarly awkward object of genetic investigation because his long life cycle and comparatively small individual progenies are indeed unfavorable to the use of certain standard research techniques of the geneticists. Moreover, we are neither able to subject ourselves to rigorous experimental conditions nor do we desire to do so. In the standard genetic approach, closely controlled mating and standardized environment are prerequisite. Where man is concerned, these experimental ideas are not feasible. Taking into consideration of above mentioned difficulties, the geneticists evolved the technique of studying the family known as "pedigree analysis", which we shall discuss eventually in this chapter.

Need for Eugenics: The development of ill organisms including the human individuals depends on both heredity and environmental factors. For the best development, good heredity and good environment are essential. Even in the best environment there is little possibility for change of defective hereditary traits. The need of eugenics is apparent from the stand point of education, sociology and civilization. The aim of geneticists should be of increasing the normal and gifted population and at the same time decreasing the abnormal and deficient populations.

Eugenics and Human Betterment: Both hereditary and environmental factors play a significant role in the development of the organisms. For better type of development, both good hereditary and suitable environment are necessary. When we consider the future welfare of the human race then the following two factors alarm us greatly:

1. The declining birth rates are among the normal and superior people (those having best germplasm)
2. A relative rapid increase of the abnormal and defective individuals (those having defective germplasm).

For the betterment of future generation, it is necessary to increase the population of outstanding people and to decrease the population of abnormal and defective people by applying the principle of eugenics. Eugenics can be applicable by adopting following two methods.

a) By encouraging the marriages between desirable persons.

b) By discouraging the marriages between undesirable persons.

A) **Positive Eugenics:** The positive eugenics attempts to increase consistently better or desirable germplasm and thus to preserve best germplasm of the society. The percentage of desirable traits can be increased by adopting following measures:

1. **Early marriage of those having desirable traits:** It is most commonly observed fact that the highly placed persons of the society often have great ambitions for the future life. In achieving their ambitious goals, they often devote the best part of their youth and they are able to marry in late age (i.e., 30 to 35 years). The biological and psychological investigations have revealed that the aged persons often lack in necessary amount of emotional warmth for the sexual activities and

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moreover, their germplasm also loses its vigor. Therefore, some laws should be formulated to prevent the late marriages of highly endowed persons by applying high taxation on them and at the same time, the young persons having the best hereditary traits should be encouraged for early marriages.

2. **Subsidizing the fit:** Because the highly endowed persons lead a well planned life and, to avoid unnecessary difficulties in nursing the children, they often prefer to have small number of children. Therefore, the selected young men and women of best eugenic value should be encouraged to increase their birth rate. Moreover, H.J. Muller has suggested that such persons not only should increase their family size but through artificial insemination, the outstanding man can serve as fathers to many more children than would be otherwise possible. Artificial insemination is already widely practiced to permit those women whose husbands are sterile or have some serious hereditary afflictions to bear children. The sperms and eggs of outstanding persons can be stored for future use by quick freezing and storing them in deep freeze. These germ cells thus can be stored for 100 or more years. Very recently the scientists felt the urgent need of establishment of sperm and egg banks to protect these precious germ cells from radiation. The germ cells could be collected during early adulthood and stored in lead lined containers in the deep freeze. In this state, the germ cells would not be subjected to radiation exposure.
3. **Education:** In a society, the people should be educated about the basic principles of human biology, human genetics, eugenics and sex. The children should be instructed about basic laws of health and they should be encouraged to develop a physically and mentally healthy body. Moreover, sex should be free from the wide spread confusion, narrow minded concepts and religious and ethical bindings. The children ignorant about the facts of sex may do more harm to society than otherwise.
4. **By avoiding germinal waste:** The wastage of best type of germplasm can be avoided by adopting following measures:
5. The selection of marriage partners should be made with intelligence.
6. The social hindrance which does not allow the teachers, nuns and priests to get married, must be removed. By adopting such measures the wastage of best type of germplasm due to lack of opportunities can be prevented.
7. Wars must be avoided because in wars the best germplasm of the society is wasted.
8. **Genetic counseling:** Genetic counseling can do great benefit to human society. The role of the genetic counselor is to inform concerned individuals of the nature of mutant condition that concern them. If it is inherited in a Mendelian fashion, then the probability of producing affected offspring can be calculated. The final decision for taking a risk is entirely the responsibility of the individual involved. It cannot be made by the counselor.
9. **Improvement of environmental conditions:** Both heredity and environment have inter-related role in the development of eugenically better persons. Therefore, every person should get better food, living conditions, education and medical guidance, etc., so his or her hereditary traits can do their best development.
10. **Promotion of genetic research:** Our knowledge about genetics is not sufficient enough because we still have little information about various human disease and metabolic disorders which are generally related with the genes. Therefore, the research in the field of cytogenetics should be increased so that we can learn more and more about the man.

B) **Negative Eugenics:** Negative eugenics attempts to eliminate the defective germplasm of the society by adopting following measures:

1. **Sexual separation of the defective:** Defective persons may have various sex-linked diseases such as night blindness, hemophilia, color blindness, etc., and various other defective traits which

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may be regulated by dominant or recessive genes. The increase of germplasm of the persons having such defective traits in the population can be checked by keeping them away and separated from the society.

2. **Sterilization:** Sterilization is the best means to deprive an individual from his power of reproduction without interrupting with any of his normal functions. Sterilization is based on surgical operation of sperm duct or vas deferens in males and oviducts or fallopian tubes in females. The former is known as vasectomy and the later is splengectomy or tubectomy. The family planning movement in India has adopted the sterilization as the tool for controlling the rate of vastly increasing population and in that case, sterilization is euthenical in its application than eugenical.

3. **Control of immigration:** Through immigration there are enough chances that undesirable or defective genes of different races and nationalities may intermingle with the normal germplasm of the populations. Therefore, the immigration rules must be strict and the persons with undesirable hereditary traits must not be allowed to migrate from one place to another.

4. **Regulation of marriage:** Presently, most human societies are money-minded and for the marriage relationship, the wealthy or highly placed persons which, however, may contain several defective genetic traits, are more preferred than those which have eugenically sound hereditary traits but having no money and which fail to reach to highest status of the society due to lack of opportunities. Some rules must be enacted to encourage the marriages among desirable mates.

DNA TECHNOLOGY IN DISEASES AND MEDICINE

DNA technology has made a significant contribution in all the three spheres of medicine - prevention, diagnosis and treatment. The various techniques in this method are discussed.

1. **DNA Probes:** DNA probes are short segments of DNA that recognize complementary sequences in DNA and thus allow identification of specific DNA sequences. This method is especially useful in diagnosis. DNA probes can hybridize with specific DNA sequences and allow the identification of specific parasite. Earlier, the identification of a parasite required the culture of blood or stool sample in different media. But with development of this technique, the identification of the parasite is easily accomplished. DNA probes have been developed for Leishmania, Trypanosoma, Plasmodium, Schistosoma, Wuchereria and other human parasites, DNA probes can also be used to identify viruses which were earlier difficult to culture.

2. **Gene Therapy:** The method of gene therapy is already discussed in the chapter on Human Genetics. It is especially useful in introduction of functional genes in individuals suffering from non-functioning of some of their genes.

3. **Production of Hormones and Proteins:** DNA technique is today used for elaboration of many hormones and proteins. Earlier, these were obtained from various types of animals by killing them. Today, with DNA technique, the genes responsible for their production can be introduced into bacteria by vectors. These genetically altered bacteria produce greater amounts of these substances. Hormones like insulin, growth hormone, proteins like urokinase are industrially produced today using this technique.

4. **Production of Synthetic Vaccines:** The conventional vaccines are inactivated germs or their antigens. The use of such vaccines always has a danger of contamination. Synthetic vaccines are produced by separation of pure antigens using mono-clonal antibodies. These are specific antibodies produced by Lymphocytes when they hyridize with the concerned cell. The resulting hybridoma (of Lymphocyte and the cell) can synthesize antibodies continuously. Such antibodies can be used for diagnosis, therapy and also prevention. Synthetic vaccines can also be produced by transferring genes for certain antigens into bacteria. Bacteria produce antibodies in large quantities which can be used as vaccines. The vaccine for Hepatitis virus is manufactured this way.

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