AGB 201 GENETICS AND CYTOGENETICS 2+1

THEORY

Earlier concepts of heredity - Study of cell and cell organelles - Prokaryotes and Eukaryotes - study of mitosis and meiosis - cell cycle - cell theory - gametogenesis and fertilization.

Mendel's work and laws of heredity - Chromosomal theory of inheritance - Gene interactions - Multiple alleles - Multiple factor hypothesis - Penetrance, Expressivity, Pleiotropy, Modifiers, Phenocopy and Lethal Genes - Linkage and crossing over - Estimation of strength of linkage and crossing over value - two and three point test cross - Genetic map - Sex determination - sex linked, sex influenced and sex limited inheritance and cytoplasmic inheritance.

Experiments showing DNA as genetic material - DNA structure and replication - gene expression and regulation - modern concept of gene.

Chromosome structure – Types of chromosomes – Special chromosomes, variation in chromosome number and structure, its genetic and cytological implications.

Polyploidy - auto and allopolyploids - uses.

PRACTICAL

Study of genetic ratios of - Monohybrid, Dihybrid, Polyhybrid - incomplete dominance, gene interactions, Multiple alleles and Multiple factors.

Study of linkage, estimation of strength of linkage and recombination frequency in two point and three point test cross data and F_2 data - Drawing of genetic map - interference and coincidence.

Principles of microscopy - preparation of fixatives and stains - Pretreatment of materials for mitosis and meiosis - Study of mitosis and meiosis.

AGB 201 GENETICS AND CYTOGENETICS 2 + 1

LECTURE SCHEDULE

Theory

1. Concept of heredity – Vapour and fluid theory, Magnetic power theory, Preformation theory – Lamarck's theory, Darwin's theory, Germplasm theory and Mutation theory.
2. Define of genetics, heredity and inheritance.
3. Definition and brief history of Cytogenetics, structure of cell organelles – Difference between prokaryotes and Eukaryotes.
4. Chromosome structure, centromere, telomere, NOR, Satellite chromosome – karyotype and idiogram – types of chromosomes based on position of centromere.
5. Study of mitosis and meiosis – cells cycle.
6. Work of Mendel – Characters studied, his observations and interpretations – Reasons for his success – Law of dominance, Law of segregation and Law of independent assortment.
7. Rediscovery of Mendel's work, chromosomal theory of inheritance.
8. Definitions of gene, allele, homozygous, heterozygous, genome, phenotype, genotype, monohybrid, dihybrid, polyhybrid, backcross and test cross.
9. Types of dominance – Complete dominance, Incomplete dominance, Co-dominance and Over dominance with examples _ Lethal genes, Pleiotropy with examples; phenocopy penetrance and expressivity.
10. Epistasis Vs Dominance – epistatic and hypostatic genes, Types of epitasis – Non-allelic interaction without modification in Mendelian ratio – Bateson and Punnet's experiment on fowl comb shape.
11. Types of epitasis – 1. Dominant epistasis (12:3:1), 2. Recessive epistasis (9:3:4), 3. Duplicate dominant epistasis (15:1), 5. Duplicate recessive epistasis (9:7), 6. Dominant and recessive epistasis (13:3).
12. Multiple alleles – characteristic features, study of blood group, coat colour in rabbits and self incompatibility in plants.
13. Multiple factor hypothesis – Bilson-Ehle – Wheat kernel colour experiment – polygenes – Transgressive segregation.
14. Quantitative Vs Qualitative characters and modifiers.
15. Linkage – coupling and repulsion – experiment of Bateson and Punnet – chromosomal theory of linkage of Morgan – complete and incomplete linkage.
16. Crossing over – significance of crossing over – cytological proof for crossing over – Stern's experiment.
17. Mid semester examination.
18. Strength of linkage and recombination – two point and three point test cross – double cross over, interference and coincidence – genetic map.
19. Sex determination – chromosomal mechanism of sex determination and its types. Genic balance theory of sex determination of Bridges.
20. Sex linked inheritance – Cris cross inheritance – reciprocal difference – Holandric genes – sex influenced and sex limited inheritance – sed determination in plants – melandrium, papaya and maize.

21. Cytoplasmic inheritance – its characteristic features – examples for chloroplast, mitochondrial, plasmid and episomic inheritance.
22. DNS, the genetic material – Griffith's experiment – experiment of Avery, McCleod and McCarthy, confirmation by Hershey and Chase.
23. Structure of DNA – Watson and Crick model – Semi conservative model of DNA replication, Central dogma.
24. Gene expression – protein synthesis – transcription – role of mRNA, tRNA, rRNA.
25. Genetic code – translation – formation of polypeptide chain.
26. Regulation of gene expression – Operon model of jacob and Monad – structural genes and regulator genes.
27. Split genes, exons and introns – modern concept of gene – gene as cistron, muton and recon, complementation test.
28. Special chromosomes _Polytene, Lamp brush, B, Ring and Iso chromosomes.
29. Variation in chromosome structure – deletion and duplication – genetic and cytological implication.
30. Inversion and translocation – genetic and cytological implications.
31. Variation in chromosome number – Euploid, aneuploid _ types of euploids
32. Polyploid – auto and allopolyploids
33. Role of polyploidy in evolution of crops – wheat, cotton, Tobacco and Brassica
34. Types o aneuploids and their origin.

PRACTICAL

1. Principles of dominance, recessive, back cross, test cross, incomplete and co-dominance and lethal factor explaining with one model – principles of Chi-square test.
2. Study of the genetic ratios – monohybrid – incomplete dominance and test cross ratios and in combination of one or two above.
3. Dihybrid ratio – dominance, incomplete dominance and test cross ratios and in combination of one or two above.
4. Simple interaction of genes – comb character in fowls and Duplicate recessive epistasis.
5. Dominance epistasis and Recessive epistasis.
6. Duplicate and additive epistasis, duplicate dominant epistasis, duplicate recessive epistasis and dominant and recessive epistasis.
7. Multiple alleles and polygenic inheritance.
8. Estimation of linkage with F_2 and test cross data, coupling and repulsion, problems on tow point test cross.
9. Three point test cross – working out interference, coincidence and drawing genetic maps.
10. Principles of microscopy and types of microscopes.
11. principle of killing and fixing preparation of stains and preservatives.
12. Studying the stages of mitosis and meiosis
13. Study of mitotic pahses in root tips of onion / Aloe sp/ Arabidopsis.
14. Procedure for fixing and observing different meiotic phhases in the inflorescence of Maize and Pearl milelt.

15. Repeating the exercise.
16. Repeating the exercise with Maize, Pearl millet and procedure for making temporary slides to permanent slides.
17. Practical examination.

REFERENCE BOOKS

1. Chandrsekar. S.N. and S.V. Parthasarathy, 190. Cytogenetics and Plant Breeding. Varadachary and Co., Madras.
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UTILITY OF THE COURSE

Genetics begins with the simple concepts of Mendelian laws, and then ramifies into several highly specialized areas, e.g., biochemistry (DNA, gene action, regulation of gene action etc.), mathematics (quantitative genetics, population genetics), physiology (biochemical genetics) etc.

Knowledge of Genetics is basic to progress in agriculture, biology and medicine. From 1953 when the double helix model of DNA was discovered, progress in molecular genetics has been phenomenal. This in turn has made sophisticated methods of chromosome and genetic engineering available.

We are now on the threshold of major advances in our understanding the processes of growth, differentiation and development in living organisms.

Unusual opportunities are now open for transferring genes across sexual barriers.

The introduction of cytoplasm Inheritance has rendered the commercial exploitation of hybrid vigour possible in many economic plants.

Knowledge of population genetics is essential for undertaking improvements of polygenically controlled characters.

The gap in ability to utilize genetic manipulation techniques can be widened by undergoing this course.

TAMIL NADU AGRICULTURAL UNIVERSITY
Centre for Plant Breeding and Genetics
AGB 201 Genetics and Cytogenetics (2 + 1)
Mid Semester

Time : One hours Max. Marks: 20
Date: 17.02.2003

PART – A

1. Answer any **EIGHT** questions only **8 x 0.5 = 4**

1. Define over dominance
2. Define synapsis
3. Define Genetics
4. Who coined the word mitosis
5. Define monohybrid
6. What are lethal genes
7. Who proposed Vapour and fluid Theory?
8. Define Multiple alleles.
9. Who proposed mutation theory?
10. Who proposed Theory of natural selection?

PART – B

II. Distinguish the following (any **SIX** only) **8 x 0.5 = 4**

1. Dominance and incomplete dominance
2. Penetrance and expressivity
3. Euchromatin and Heterochromatin
4. Homozygous and Heterozygous
5. Test cross and Back cross
6. Epistatis and Dominance
7. Phenotype and Genotype

PART – A

III. Answer any **FIVE** questions only **5 x 2 = 10**

1. Write the significance of meiosis?
2. Explain germplasm theory?
3. What are Medal's laws of inheritance? Describe any one law in detail?
4. Explain Co-dominance with the example?
5. Write the difference between polygenic and oliogogenic traits?
6. Write about different types of epistatic interaction.

TAMIL NADU AGRICULTURAL UNIVERSITY

B. Sc. (Agriculture) Degree Programme

II Year IV Semester Final Theory Examinations May, 2003

AGB 201 Genetics and Cytogenetics (2 + 1)

(1999 Syllabus)

Time : 3 hours Max. Marks: 40

PART – A

(Answer any **Ten** questions Only) **(10 x ½ = 5)**

Define

A1. Gametes
A2. Multiple allele
A3. Genotypes
A4. Pure line
A5. Crossing over
A6. Polygenes
A7. Triploid
A8. Inversion
A9. Epistasis
A10. Lethal gene
A11. Reciprocal cross
A12. Heterozygous

PART – B

(Answer any **Five** questions Only) **(5 x 1 = 5)**

Differentiate:

B1. Back cross and test cross
B2. Sex limited and sex influenced character
B3. Euploid and aneuploid
B4. Inbred and pure line
B5. Autopolyploid and Allopolyploid
B6. Deletion and duplication

PART – C

(Answer any **Five** questions Only) **(5 x 2 = 10)**

Write short notes on:

C1. Law of segregation
C2. Endomitosis
C3. Sex linkage
C4. Transcription
C5. Genetic map
C6. Types of dominance

PART – D

(Answer any **Four** questions Only) **(4 x 5 = 20)**

D1. Describe the chromosome theory of inheritance with example.
D2. What is linkage? Explain the role of linkage and crossing over in evolution.
D3. Describe various types of chromosomal aberration and their role in evolution.
D4. Give an account of quantitative and qualitative characters and modifiers.
D5. What is DNA? How it differs from RNA?

Leture I

Concept of heredity – Vapour and fluid theory, Magnetic power theory, Preformation theory – Lamarck's theory, Darwin's theory, Germplasm theory and Mutation theory.

Concept of heredity:
Mendel's Principles of Heredity:

Mendel chose two plants differing in a pair of contrasting characters, eg., a plant with round seed-coat and another with wrinkled seed-coat, as parents for each of his experiments. He than confirmed that they bred true for several generation on self-fertilisation. He then crossed them and obtained the hybrid seeds. He found that the first generation (i.e., the first filial, or F_1 generation) hybrids were always uniform. He than self-fertilised the F_1 and obtained as large a number as possible of a second generation, or F_2. he found that the F_2 consisted of different kinds. He classified the F_2 according to the characters exhibited and counted the number of each class.

In the light of present knowledge, Mendel's principles of heredity can be expressed as follows:

In sexual reproduction, the individual (or zygote) is formed by the fusion of two gametes, one (the egg) from the mother and the other (the sperm) from the father.

The hereditary particles are called genes (or factors). The female gamete contributes one of gene from the mother and the male gamete, one of each kind of gene from the father.

A zygote carries therefore every gene in duplicate. These genes however do not blend but preserve their individualities.

When this individual forms its own gametes, the maternal and paternal members of each pair of genes segregate and pass to different gametes.

Each gamete therefore has only one member of a pair genes existing in adult individuals.

The members of different maternal and paternal pairs of genes segregates independently and different gametes produced by the same individual may therefore contain different sets of genes.

These principles were summed up by Carl Correns, one of the rediscoveries of Mendel, in what are now known as Mendel's laws of heredity.

The first law is that hereditary factors (genes are found in pairs in mature individuals. They do not blend but separate or segregate unchanged during the formation

of gametes. The gametes therefore contain only one of a pair of factors responsible for each character. Even hybrids therefore produce gametes which are 'pure'.

The second law is that the members of different pairs of factors responsible for different characters segregate and recombine independently in different gametes.

Vapour and fluid theory:

Early Greek philosophers speculated that the hereditary informations of parents existed in the form of vapours of fluids. Pythagoras (500 B.C.) speculated that a moist "vapour" descended from the brain, nerves and other body organs of the male during the coitus and from these vapours an embryo was formed in the uterus of the female. According to him, the male transmitted all the characters of the embryo and the female does not. However, another Greek philosopher of the same age, Empedocles thought that both parents contributed equally to the embryo and each parent produces a "semen"' which arises directly from various body parts.

After 200 years, another Greek philosopher Aristotle forwarded a highly imaginative speculation that the seman of the male had certain vitalizing or "dynamic" effect and it was supposed to be highly purified blood. According to him, the female furnished the inert building materials, while the male gives the motion and new life to the material.

Magnetic power theory:

In the 17th century W. Harvey (1578-1657), after performing certain experiments on deer proposed the theory called magnetic power theory. He suggested that as iron by friction with a magnet possesses the magnetic properties, so that the uterus by the friction of coitus acquires some magnetic power to conceive an embryo.

Preformation theory:

Leonardo da vinci (1452-1519) revealed the fact that the male and female parents contribute equally to the heredity of the offspring. W. Harvey (1651) speculated that all living things (including man) originate form eggs and that the semen only plays vitalizing role. It was R. de Graaf (1641 – 1673) who observed that the progeny would possess the characteristics of both parents (*i.e.,* mother and father) and therefore, suggested that both the parents should contribute to heredity (biparental inheritance). The sperms of man and other mammals were observed first of all by A.V. Leeuwenhoek in 1677. The mammalian egg was discovered by Von Baer in 1828. N. Grew (1682) first of all reported the reproductive organs of plants. With the discovery of eggs and sperms the biologists of 17th century started to speculate that the new in dividual was completely preformed in miniature in the gamete. According to different workers such miniature preformed embryo was speculated to occur either in the egg or sperm. Swammerdam (1637-1680), for example, thought that a tiny preformed frog occurred in the animal hemisphere of the frog egg and that became simply larger by feeding on the food stored in the vegetal hemisphere of the egg. Another biologist, Hartsoeker (1695) published a figure showing a miniature man known as mankin or homunculus in the head of the

human spermatazoa. Such preformation theories had been supported by Leeuwenhoek (1632-1723), Malphigi (1673), Reaumur, Bonnet (1720-1793), Spallazzani (1729-1799) and other workers of 17th and early 18th centuries.

With the development of improved microscopy and other cytological techniques in 17th and 18th centuries, it became clear to biologists that neither the egg nor the sperm contained a preformed individual but that each was a relatively uniform, homogenous mass of protoplasm.

Lamarck's theory:

The French biologist Jean Bapthiste de Lamarck (1744-1829) proposed the theory that environmental changes cause modifications in organisms and that such modifications are transmitted to subsequent generation. He believed that environment acts directly on plants and indirectly on higher animals.

Lamarck said that changes in environmental conditions create new needs in animals. Conscious efforts of the animals to adapt to the environment involves the use of certain organs, thereby causing them to become large, strong and well-developed. Other organs are not used and so become smaller, weaker and less well-developed. Such bodily changes are called acquired characters since an animal achieves them by its own exertions to adapt to the environment. Acquired characters, according to Lamarck, are hen passed on to the offspring of the organism that acquired them, and new species originate by accumulation of these modification.

The giraffe dwells in the interior arid parts of Africa where there is not much herbage. According to Lamarck, the giraffe was obliged to feed on the leaves of tall trees and to strain itself continuously to reach them. Such exercise caused the necks and legs to grow in length. The increased length was inherited by the progeny, which, in turn, stretched their necks and legs and transmitted their increased length to their own offspering. Thus has evolved the present day six-metre high giraffe.

Detailed studies have failed to show that acquired characters are inherited. Most biologists have therefore abandoned the theory of inheritance of acquired characters, otherwise known as *Lamarckism.*

Darwin's Theory

In 1858, Charles Darwin (1809-1882) and Wallace independently proposed the *'Theory of Natural Selection'*. According to this theory, many more individuals of each species are born than can possibly survive and consequently there is always a struggle for existence. If hereditary differences occur within the wild species of plants, nature will eliminate some and select others.

Over-production, struggle for existence, hereditary variations and survival of the fittest are thus the important principles of the theory of natural selection.

Ten years after the publication of the *Origin of Species* (1859), Darwin adopted the doctrine of the inheritance of acquired characters but he proposed a new theory of how it happened. He modified the views of Spencer and proposed the 'Hypothesis of Pangenesis' (1868).

Darwin assumed that hereditary particles termed pangenes or gemmules, are produced by every part of the body during the life time of an organism and that, these assume the characters of the various parts of the body from which they were derived, together with whatever modifications the latter may have acquired. Eventually all the pangenes accumulate to form the germ cells which give rise to the new individual, thus ensuring the development of the parental characters and inheritance of acquired characters.

Germplasm theory

Weismann (1834-1914), a German zoologist, suggested in 1887 that a reduction in chromosome number took place during the formation of the egg and the sperm, and that the original number was restored when the egg and the sperm fused. In 1892, he suggested that the maternal and paternal chromosomes separated during the reduction division and that they recombined when the gametes united.

According to Weismann's *'Germplasm Theory of Heredity'*, the hereditary particles called *ids* (what we now call as genes) situated on *idants* (what we now call as chromosomes) constituted the germplasm. The germplasm is handed down form parent to offspring and it gives rise to the body or soma (somatoplasm) whose character it determine. The germplasm is independent of the body and whatever happens to this body has no effect on the germplasm which his contained within it.

According to Weismann, acquired characters cannot therefore be inherited. To prove this he cut off the tails of mice for twenty two generations and found that the progeny consisting of 1,592 individuals had tail of normal length.

The independence of the germplasm from the somatoplasm was shown by the ovary transplantation experiment in guinea pig. Ordinarily, when albino guinea pigs are mated with albinos, only albinos are produced. Castle and Phillips removed the ovaries of an albino guinea pig and grafted in their place the ovaries of a black guinea pig. The albino animal with the ovary of the black one was then mated with an albino. All the offspring were found to be black, thereby proving that the germplasm (i.e., the ovary from the black guinea pig) is not affected by the somatoplasm (i.e., the body of the albino).

Mutation Theory

Charles Darwin believed that evolution is due to natural selection of small hereditary variations occurring among individuals of any species. Bateson did not agree with Charles Darwin. He believed that evolution is due to large discontinuous variations. De Vries (1848-1935) introduced the term 'mutation' for these large, discontinuous changes in the genotypes and proposed the '*Mutation Theory'*, according to which sudden hereditary changes lead to evolution.

De Vries (1901) observed that the evening primrose *Oenothera lamarkiana,* a native of America, was growing wild in Holland. In a population of this weed, he observed some plants which differed in some characters from the typical *Oenothera lamarckiana.* Since it is a self fertilised species, he felt that these variants have arisen suddenly rather than as hybrids. He transplanted them to his garden and studied them for several years. He observed that variation continued to arise spontaneously and that these variations were inherited. He called these drastic changes as mutations and maintained that mutations play an important role in the evolution of new species.

Lecture II

Define of genetics, heredity and inheritance

GENETICS

1. **Genetics is the science of heredity and variation**.
2. **Genetics is a study of the mechanism of transmission of characters from parents to their offspring, origin of variation and gene action.**

It uses viruses ("virus genetics") microorganisms ("microbial genetics"), plants ("plant genetics"), animals ("animal genetics"), and man ("human genetics") as objects of study. The subject matter is the phenomenology and physiology of heredity ("classical genetics") as well as the nature of the genetic material and the storage of – genetics information, its replication, mutation, transmission, recombination, and translation into systems by which the genetic material mediates its control over metabolism and development and determines the reappearance of parental characters among progeny ("molecular genetics"). "Population genetics", as distinguished from studies of inheritance at the familial level, describes in mathematical terms the consequences of inheritance on the population level and attempts to predict the behavior of future generations it deals with the frequencies and interactions of genes in interbreeding populations and studies the agencies (e.g., mutation, natural and artificial selection, gene flow, migration, and change factors) which tend to alter gene frequencies and thus to cause evolutionary changes.

HEREDITY

That process which brigs about the biological similarity between parents and progeny.

It consists of the conservation of specificity during replication of the – genetics material (the storage of – genetics information) and is carried out by means of transcription and translation of the genetic information (- genetic transcription; genetic translation; genetic code). Genetics is the science of heredity.

INHERITANCE

Inheritance is the transmission of – genetic information from parents and ancestors to offspring.

The theory, that the cell is the basic unit of life, and all plants and animals are composed of one or more cells, was enunciated in 1833 by two German scientists, Schleiden and Schwann. That cells arise only from pre-existing cells is an equally important generalization made by another German scientist, Virchow in his 'Theory of Cell Lineage', proposed in 1858.

Reference: R. Rieger- 243, 267, 295., B.D.S – 868., DDS: 14

Every plant or animal starts its life only as a single cell. This gives rise to two new cells by division. Each of these again divides into two and the process is repeated. In multicellular organisms, a number of cells is thus formed and the appearance of the mature organism depends upon the arrangement of these cells. Growth in multicellular forms thus depends upon cell division accompanies generally by cellular enlargement and differentiation. In unicellular organisms, the division of cells is a process of asexual reproduction. It leads to an increase in the total number of individuals. In sexual reproduction, two cells unite to give rise to a new individual. Life is thus an uninterrupted succession of cells and what is inherited must therefore be contained in cells.

The first cell of a new individual arising from sexual reproduction is formed by the union of the egg nucleus from the female and the sperm nucleus from the male. The physical links between the parents and the offspring are thus the nuclei of the egg and the sperm, and the hereditary material passed on from one generation to another must therefore be contained in those nuclei. The nuclei are thus the carriers of heredity.

Cytogenetics

Cytogenetics is study of chromosomes in relation to genetics.

Essentially, the field of study comprises the behavior of the chromosme during mitosis and meiosis, their origin and their relation to the transmission and recombination of genes.

Lecture III
Chromosome structure, Centromere, Telomere, NOR, Satellite chromosome – karyotype and idiogram – types of chromosomes based on position of centromere.

Chromosome

The chromosome have been considered as the physical bases of heredity because they have a special organization, individuality, functions and are capable of self-reproduction. Their main chemical constituent is DNA, an universally accepted genetic or hereditary materials found to carry genetic informations from one generation to next generation. They occur in all living beings in a specific number and organization and usually fall into following categories.

a. Viral chromosomes

The chromosomes of viruses are called viral chromosomes. They occur singly in a viral species and chemically may contain either DNA or RNA. The DNA containing viral chromosomes may be either of linear shape (*e.g.,* T_2, T_3, T_4, T_5, bacteriophages) or circular shape (*e.g.,* most animal viruses and certain bacteriophages). The RNA containing viral chromosomes are composed of a linear, single-stranded RNA molecule and occur in some animal viruses(*e.g.,* poliomyelitis virus, influenza virus, etc.); most plant viruses, (*e.g*., tobacco mosaic virus, TMV) and some bacteriophages. Both types of viral chromosomes are either tightly packed within the capsids of mature virus particles (virons) or occur freely inside the host cell.

b. Prokaryotic chromosomes

The prokaryotes usually consists of a single, giant and circular chromosome in each of their nucloids. Each prokaryotic chromosome consists of a single circular, double-stranded DNA molecule, but has no protein and RNA around the DNA molecule like eukaryotes. Different prokaryotic species have different sizes of chromosome. Thus, the bacterium *Escherichia coli* has 100 long chromosome.

c. Eukaryotic chromosomes

The eukaryotes (plants and animals) usually contain much more genetic informations than the viruses and prokaryotes, therefore, contain a great amount of genetic material, DNA molecule which here may not occur as a single unit, but, as many units called chromosomes. Different species of eukaryotes have different but always constant and characteristic number of chromosomes.

Reference: PSC & VKA: 26-36,

Chromosome number in some plant and animal species.

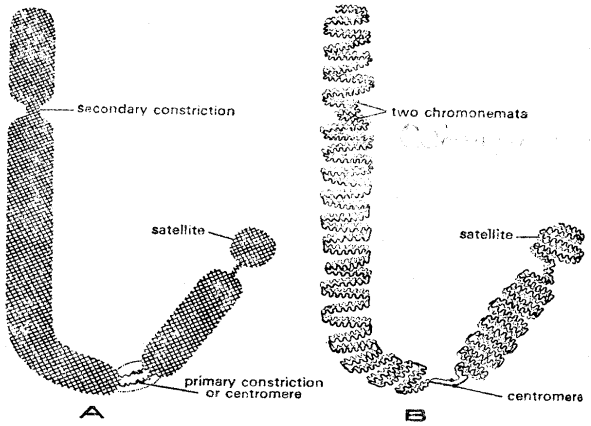
Common name	Scientific name	Chromosome number
Pea	*Pisum sativm*	14
Cabbage	*Brassica oleracea*	18
Radish	*Raphanus sativus*	18
Onion	*Allium cepa*	16
Indian corn	*Zea mays*	20
Sugar cane	*Saccarum officinarum*	80
Hydra	*Hydra vulgaris*	32
Mosquito	*Culex pipiens*	6
Fruit fly	*Drosophila melanogaster*	8
Man	*Homo sapiens*	46

Diploids and Haploids

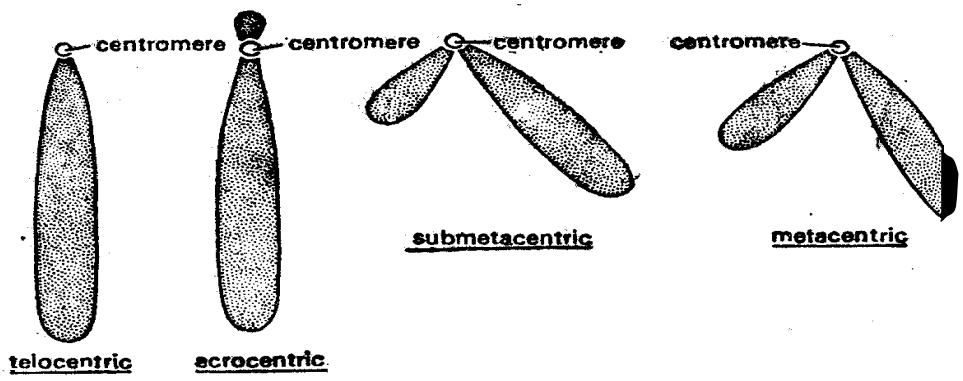
In contrast to prokaryotes, most eukaryotes are **diploids**, *i.e.,* each somatic cell of them contains one set of chromosome inherited from the maternal (female) parent and a comparable set of chromosomes (called homologous chromosomes) from the paternal (male) parent. The number of chromosomes in a dual set of a diploid somatic cell is called the **diploid number (2n).** The sex cells (sperms and ova) of a diploid eukaryotic cells contain half the number of chromosomal sets found in the somatic cells and are known as **haploid (n)** cells. A haploid set of chromosome is also called **genome.** The fertilization process restores the diploid number of a diploid species.

Morphology of the Eukaryotic chromosomes

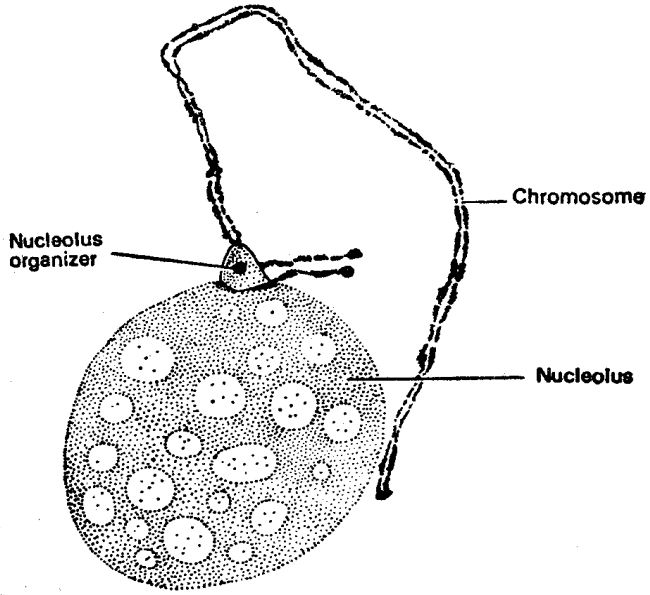
The eukaryotic chromosomes differ from the prokaryotic chromosomes in morphology, chemical composition and molecular structure. The shape of the eukaryotic chromosomes is changeable from phase to phase in the continuous process of the ell growth and cell division. They are thin, coiled, elastic, contractile, thread-like structure during the interphase (when no division of cell occurs) and are called chromatin threads. During metaphase stage of mitosis and prophase of meiosis, these chromatin threads become highly coiled and folded to form compact and individually distinct ribbon-shaped chromosomes.



These chromosomes contain a clear zone called kinetochore or centromere along their length. The number and position of centromeres is variable, but is definite in a specific chromosome of all the cells and in all the individuals of the same specie. Thus, according to the number of the centromere the eukaryotic chromosomes may be acentric (without any centromere), monocentric (with one centromere), dicentric (with two centromeres) or polycentric (with more than two centromeres). The centromere has small granules or spherules and divides the chromosomes into two or more equal or unequal chromosomal arms. According to the position of the centromere, the eukaryotic chromosomes may be rod-shaped (telocentric and acrocentric) J-shaped (submetacentric) and V-shaped (metacentric).



During the cell divisions the microtubules of the spindle are get attached with the chromosomal centromers and move them towards the opposite poles of cell. Besides centromers, the chromosomes may bear terminal unipolar segments called telomeres. Certain chromosomes contain an additional specialized segment, the nucleolus organizer, which is associated with the nucleolus.



Chemical structure of chromosomes

Chemically, the eukaryotic chromosomes are composed of deoxyribonucleic acid (DNA), ribonucleic acid (RNA), histone and non-histone proteins and certain metallic ions. The most important enzymatic proteins of chromosomes are phosphoproteins, DNA polymerase, RNA-polymerease, DPN-pyrophosphorylase, and nucleoside triphosphatase. The metal ions as Ca^{+} and Mg^{+} are supposed to maintain the organization of chromosomes intact.

Molecular structure of chromosomes

According to the recent and widely accepted theory of Duparaw (1965, 1970) and Hans Ris (1967) called unistranded theory, each eukaryotic chromosome is composed of a single, greatly elongated and highly folded nucleoprotein fibre of $100A^{O}$ thick. This nucleoprotein fibre in its turn is composed of a single, linear, double-stranded DNA molecule which remains wrapped in equal amounts of histone and non-histone proteins and variable amount of different kinds of RNA.

Kinds of chromosomes

The eukaryotic chromosomes have been classified into autosomes and sex chromosomes. The autosomes have nothing to do with the determination of sex and exceed in number than sex chromosomes. The sex chromosomes determine the sex of their bearer. They are usually two in number and are usually of two kinds: X chromosomes and Y chromosomes

Karyotype and idiogram

For cytogentical studies, when the chromosomes of a species are arranged according to their shape, size and structure, than that is called karyotype of that species. When the karyotype of a species are represented by the diagram then such diagrams are called idiograms.

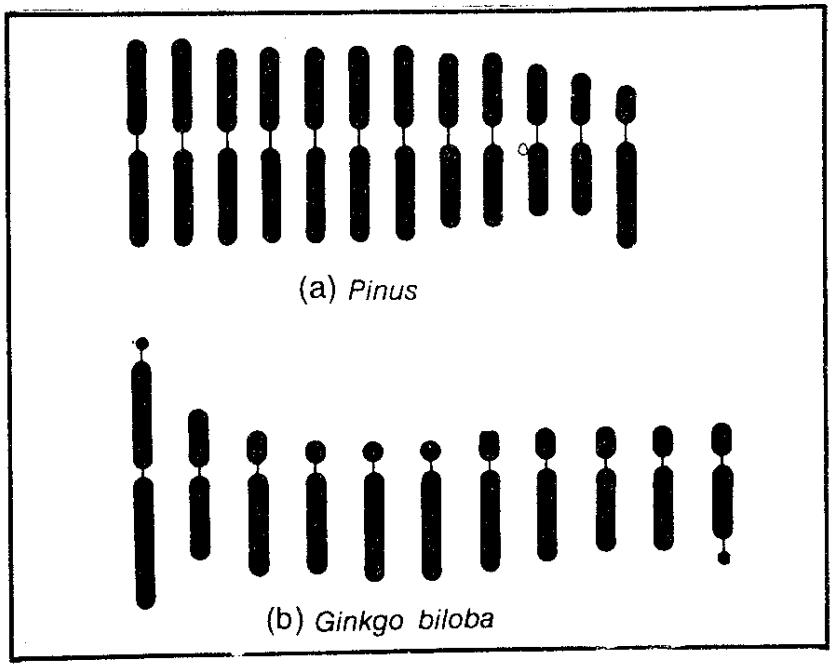
Special Types of Chromosomes

1. Polytene chromosomes – E.g. *D.melanogaster*
2. Lampbrush chromosomes
3. B-chromosomes
4. Holokinetic chromosomes

Genetic significance of chromosomes

The chromosomes are considered as the organs of heredity because of following reasons:

i. They form the only link between two generations.
ii. A diploid chromosome set consist of two morphologically similar (except the X and Y sex chromosomes) sets, one is derived from the mother and another form the father at fertilization.
iii. The genetic material, DNA or RNA is localized in the chromosome and its contents are relatively constant from one generation to the next.
iv. The chromosomes maintain and replicate the genetic informations contained in their DNA molecule and this information is transcribed at the right time in proper sequence into the specific types of RNA molecules which directs the synthesis of different types of proteins to form a body form like the parents.



(a) symmetric karyotype and (b) Asymmetric karyotype.
(Redrawn from Stebbins, 1971)

Lecture IV
Work of Mendel – Characters studied, his observations and interpretations – Reasons for his success – Law of dominance, Law of segregation and Law of independent assortment.

Mendel and his work:

Johann Mendel was the pioneer of classical geneticists. He was born in July 22, 1822 in Heinzendorf in Austrian Silesia, where his father, Anton Mendel was the owner of a small farm. He graduated from the Gymnasium in 1840. In his youth, he led a disastrous, poor, difficult and sad life. In the year 1846, Johann Gregor Mendel attended courses of agriculture, pomiculture and viniculture at the Philosophical Academy in Brunn. In the spring of 1856, he began experimental crossing of pea varieties.

On February 8, 1865, he delivered his first lecture on pea experiments to Brunn Naturals Science Society. In 1866 his paper "Experiments on plant hybridization" published in volume 4 of the proceedings of the Natural Science Society. In the same years, he began experiments with other plant species. In this paper, Mendel proposed some basic genetic principles. But unfortunately his remarkable piece of work remained unattended and unappreciated upto 1900. Mendel wasted his valuable six years on the hybridization experiments on this plant species, ruined his eye sight, but even then failed to confirm or even to test his theory. The results of these ill-fated experiments were published in 1869, in the Proceedings of the Natural Science Society, Brunn.

Rediscovery of mendel's work

Mendel's research paper remained dormant and unnoticed by the scientific world until 1900. During these intervening thirty four years many developments occurred in biology which prepared the way for the rediscovery of Mendel's work.

It was in the beginning of 20^{th} century that three botanists, namely Hugo de Vries, working on Oenothera ; Correns working on Xenia, peas and maize and Von Tschermak working on various flowering plans, independently drawn the conclusions like Mendel. Later these botanists came across the research paper of Mendel and rediscovered it in 1900. Mendel's original paper was republished in Flora, 89, 364 (1901). Bateson confirmed Mendel's work by a series of hybridization experiments.

Mendel's Work

From the original research paper of Mendel it was obvious that Mendel was well acquainted with the scientific literature related to hybridization of his time. His approach was simple, logical, scientific, mathematical and analytical. Mendel, concentrated his attention on a particular character and at a time, he studied only one character of the hybrid. Further, unlike other hybridists, he designed his hybridization experiments to record the number of different types of the progeny.

Reference: PSC & VKA: 73-81

Mendel's Selection of the Experimental Plant

He found the plants of family Leguminosae such as peas and beans, most suitable materials for his experiments, because these plants 1) were easy to culture in open ground or in pots ; (2) had short growth period and life-cycle ; (3) had self-pollinating flowers of peculiar structure ; (4) had contrasting heritable characters, and (5) might produce fertile hybrids on artificial cross pollination.

Mendel found edible pea (*Pisum sativum*) a best material for his hybridization experiments, because its various available varieties (about 34) showed clear cut differences. Mendel used a total of following seven pairs of characters :

1. The shape of the seed – Round and full : Irregularly-shaped and Wrinkled.
2. The colour of the cotyledons – Yellow : Green
3. The colour of the seed coat – Grey to buff : White
4. The shape of the ripe pod – Simply inflated : Constricted between the seeds.
5. The colour of the unripe pods – Light to dark green : Yellow
6. The position of the flowers on the stem – Axial : Terminal.
7. The length of the stem – Tall or long : Short or dwarf.

Besides pea, Mendel has also used following plant species in his hybridization experiments ; *Zea mays, IpomoeaPhaseolus* sp., *Dianthus* sp.

Mendel's Experimental Observations and Results

To understand the observations and results of Mendel's hybridization experiments, one has to become familiar about few genetic terms, which have been coined by W. Bateson between the years 1902-1909.

Parental (P) generation: The plants with unlike characteristics in which the artificial cross is made are called parental (P) generation and the progeny obtained from such a cross is called **hybrid**.

In genetical language, a hybrid can be defined as an individual which results from the crossing of the two individuals differing atleast in one set of the character.

According to the number of pairs of contrasting characters in the parental generation, the resultant hybrids are called as follows.
Monohybrids (have one pair of different character).
Dihybrids (have two pairs of different characters) or
Polyhybrid (have more than two pairs of different characters).

The process through which the hybrids are produced is called hybridization and such a hybridization cross may be a monohybrid cross, dihybrid cross or polyhybrid cross depending upon the kind of hybrid it produces. The resultant hybrids of P generation is the first filial generation or $\mathbf{F_1}$. When $\mathbf{F_1}$ progeny is allowed to self-fertilize or cross-fertilize among themselves, they produce the second filial generation or $\mathbf{F_2}$. The subsequent generation which may be produced by self-fertilization are called $\mathbf{F_3}$, $\mathbf{F_4}$, $\mathbf{F_5}$, $\mathbf{F_6}$, etc.

Mendel's observations of the hybrids of F_1 revealed that hybrid plants of F_1 contained the character of only one parent, none of them displayed any intermediate character of both parents. Those characters which were transmitted unchanged and expressed in the hybrid in the hybridization process were called as **dominant characters** and those which became latent in the process were called as **recessive characters** by Mendel. He tested each of the seven pairs of contrasting characters for the phenomenon of the dominance and recessiveness and found following characters as dominant.

Dominant and Recessive Characters is Pea.

Character	Dominant	Recessive
Seed form	Round or Smooth	Wrinkle
Cotyledon colour	Yellow	Green
Seed coat	Grey	White
Shape of unripe pod	Inflated	Constricted
Colour of unripe pod	Green	Yellow
Flower position	Axial	Terminal
Stem length	Tall (long)	Dwarf (Short)

Mendel's observations of F_2 progeny further revealed the fact that the recessive character which was depressed or concealed in F_1 hybrids reappeared in the F_2 offspring in the definitely expressed average propagation of three to one, so that among each four plants of F_2, three displayed the dominant character and one the recessive character. This 3:1 F_2 ratio was observed to occur in the monohybrid cross for each of seven pairs of characters. Mendels original F_2 results for seven pairs of character has been tabulated.

Results of Mendel's Original Crosses for Seven Pairs of Characters.

S.No.	Structure	Character	Dominant	Recessive	Ratio in F_2
1.	Seed	Form	5,474 Round	1,850 Wrinkled	2.96 : 1
2.	Cotyledon	Colour	6,022 Yellow	2,001 Green	3.01 : 1
3.	Seed coat	Form	882 Inflated	299 Constricted	2.95 : 1
4.	Seed coat	Colour	705 Grey	224 White	3.15 : 1
5.	Unripe pods	Colour	428 Green	152 Yellow	2.82 : 1
6.	Flowers	Position	651 Axial	207 Terminal	3.14 : 1
7.	Stem	Length	787 Long	277 Short	2.84 : 1

When the recessive hybrids of F_2 (which were forming 25 percent of total F_2 progeny) were self-pollinated, they always produced F_3 offsprings with only recessive characters. From remaining 75 percent F_2 hybrids, 25 per cent hybrids yielded F_3 offsprings with only dominant characters, while remaining 50 per cent dominant hybrids of F_2 yielded the F_3 offsprings displaying the dominant and recessive characters in 3 : 1 ratio.

Mendel's explanations and predictions

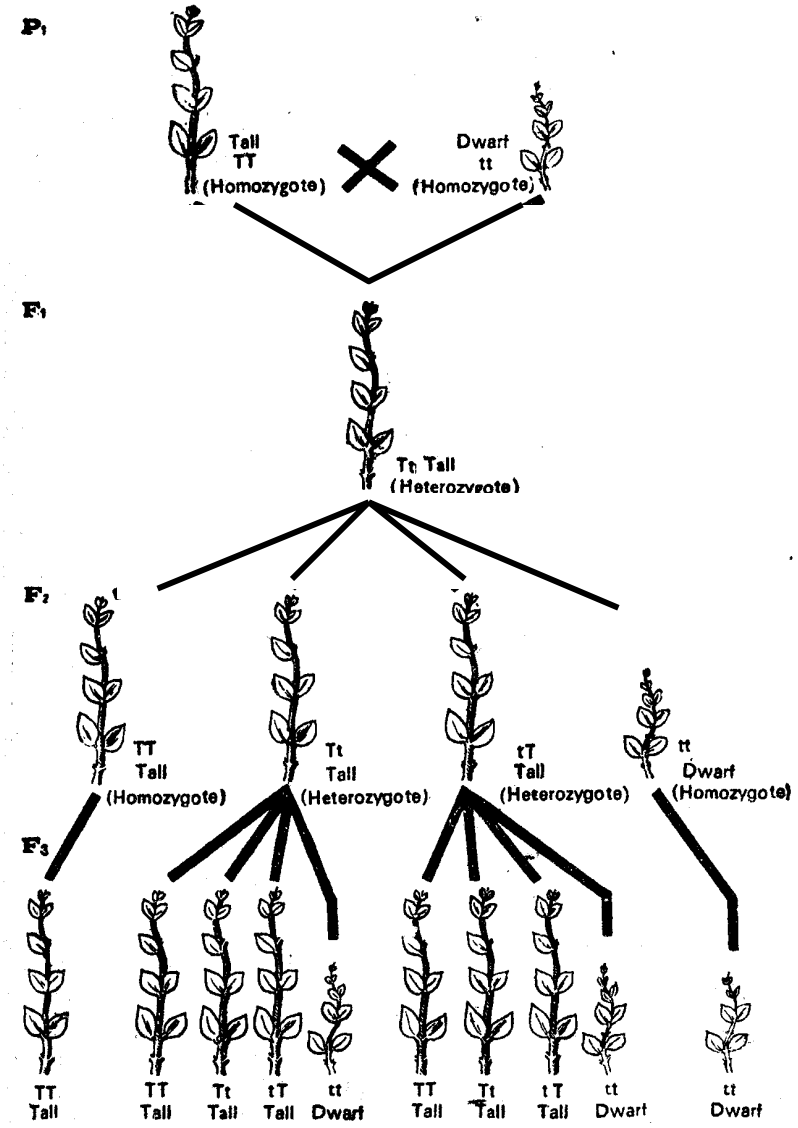
At the time Mendel was conducting his hybridization experiments, the cytology was in its very primitive state and the gymnastics performed by the chromosomes during gametogenesis and fertilization had yet to be observed and interpreted. Mendels brilliance of mind is revealed by his explanations and predictions which have been forwarded by him for the results of his hybridization experiments. Behind the characters and their mode of inheritance from generation to generation, he visualized the determinants of the characters, the factors or elements (named as genes by Johannsen in 1909). According to Mendel, each male and female parent contained a pair of such hereditary factors, and each parent passed only one factor of a pair to their offsprings. Thus, each parental character was though not be transmitted directly to the offsprings but by some indirect mean and that is by hereditary factors which determine the characters. Further, he predicted that each factor retained its individuality from generation and it was not modified in the hybrid. The factors contributed by the parents united randomly to produce the characters of a hybrid. Thus, indirectly, he predicted about the reduction division during gametogenesis and the physical hereditary mechanism, both were unknown to the scientific world at that time.

Mendel's Laws

Mendel himself did not postulated any genetical principle or laws as erroneously described various text books of genetics. He simply gave conclusive theoretical and statistical explanations for his hybridization experiments in his research paper. It was Correns the discoverer of Mendel's work who thought that Mendels discovery could be represented by the two laws of heredity. These laws of heredity are 'law of segregation" and the law of independent assortment" or "law of free recombination." The phenomenon of dominance has been considered erroneously as the law of dominance in some of the text books of genetics.

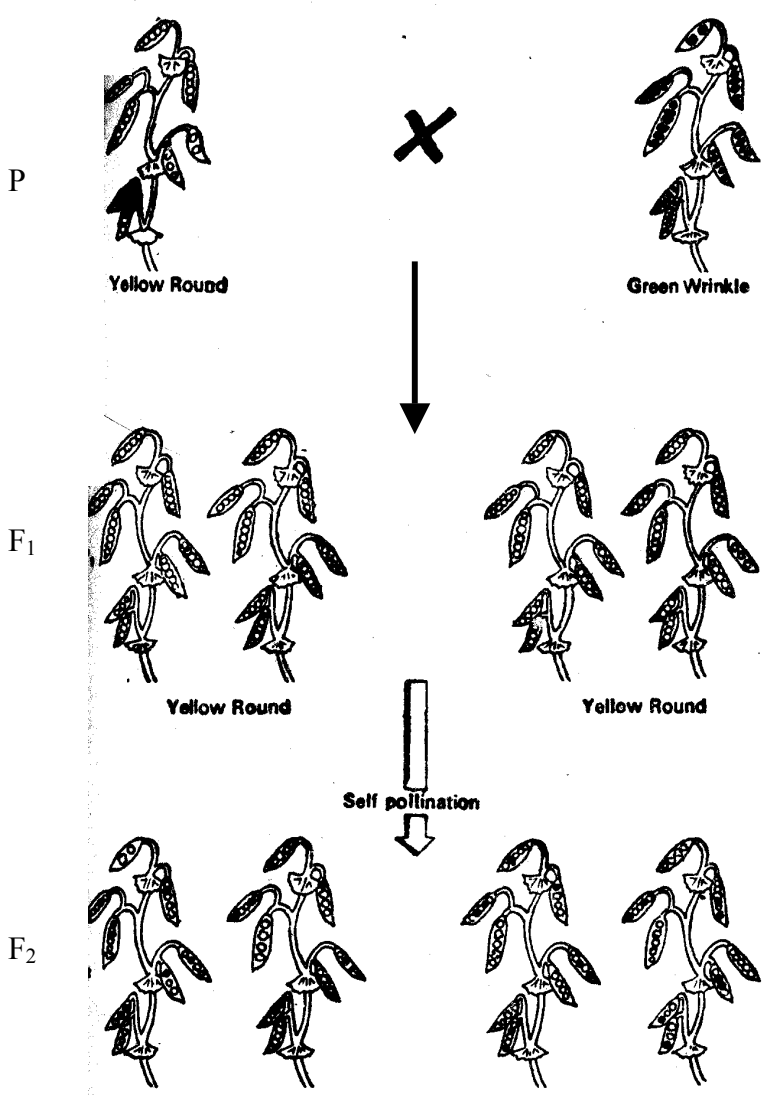
Mendel's Law of segregation

Mendel;s first law of inheritance, the law of segregation or law of purity of gametes states that in a heterozygote a dominant and a recessive allele remain together throughout the life (from the zygote to the gametogenesis stage) without contaminating or mixing with each other and finally separate or segregate from each other during gametogenesis, so that, each gamete receives only one allele either dominant or recessive. For example, the F_1 hybrids (Tt) of a monohybrid cross between tall (TT) and dwarf (tt) pea plant (Fig 8.1) have one dominant allele (T) for tallness and one recessive allele (t) for dwarfness. This genotype of F_1 hybrids remains the same from the unicellular zygote stage to the gametogenesis stage of multicellular adult plant. These F_1 hybrids by self-fertilization produce tall and dwarf plants in the ratio of 3 : 1. It means that tall and dwarf alleles though, remain together for long time but does not contaminate or mix with any one and both alleles segregate to produce gametes which either having dominant allele T or recessive allele t. These gametes unite to produce the 3 : 1 phenotypic ratio in F_2.



Mendel's Law of Independent Assortment

Mendel's law of independent assortment or recombination off genes states that when the gametes are formed the members of the different pairs of factors (genes) segregate quite independently of each other and that all possible combinations of the factors (genes) concerned will be found among the progeny.



F_2 phenotypic ratio 9 Yellow Round, 3 YW, 3 GR and 1 GW

Lecture V
Definitions of gene, allele, homozygous, heterozygous, genome, phenotype, genotype, backcross and test cross. Chromosomal Theory of Inheritance

Gene

Mendel Postulated that 'Characters' are determined by 'elements' found in the sex cells or gametes. The character and its determinant are thus different and Bateson coined the word '*factor'* for that which determines a character. The Danish geneticist Johannsen recognised that there is something in the gametes and in the fertilised egg that determines a character and he proposed the word '*gene'* for it.

Gene can be defined as the hypothetical unit of inheritance located at a fixed position (i.e., the locus) on a chromosome which by interaction with the other genes, the cytoplasm and the environment controls the development of a character.

Allele

Allele is defined as one of a pair (or series) of forms of a gene situated at the same locus of homologous chromosomes.

Homozygote and Heterozygote

Mendel recognised that a gamete can possess only one of a pair of alleles, for example, either **R** or **r** and not both. An individual formed by the union of like gametes is said to be pure. The British geneticist Bateson introduced the term *homozygote* (*homo* = same; *zygos* = yolk) for an organism in which the two genes at the same locus of homologous chromosomes are identical. The true-breeding round-seeded pea plant is formed by the union of an egg with **R** and a sperm, also with **R** and is represented as **RR**, and the true-breeding wrinkled-seeded plant formed by the union of an egg and a sperm, each with **r**, is represented as **rr**. An individual formed by the union unlike gametes is said to be a *hybrid*. Bateson called this a *heterozygote* (*hetro* = different; *zygos* = yolk) because the two genes at the same locus of homologous chromosomes are not identical. The round seeds of the first hybrid generation (i.e., F1) formed by the union of a gamete with **R** and another gamete with **r** are therefore heterozygous and are represented as **Rr**.

Genome

A haploid set of chromosome is called as genome.

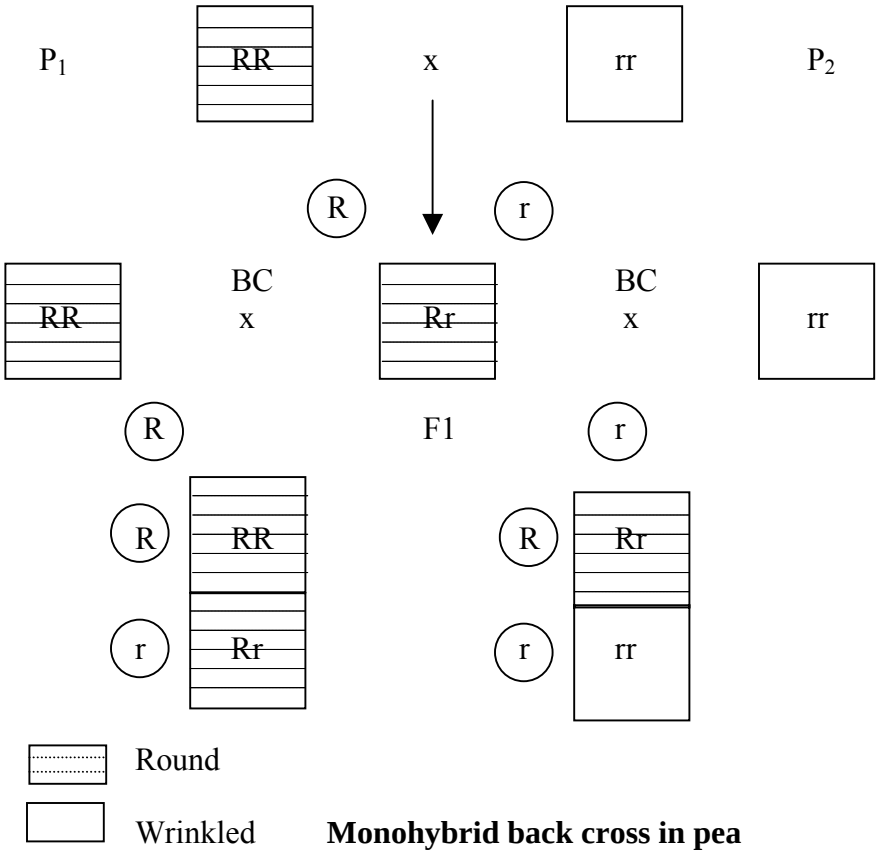
Phenotype and Genotype

Johannsen clearly brought out the difference between the visible character and the invisible gene that is responsible for the character. He coined the world *phenotype* (*pheno* = appear) for the visible character of an individual and the word *genotype* for the

heredity of a plant or animal. The genotype of an individual can be determined by observing its phenotype, but as two different genotypes may possess the same phenotype because of the phenomenon of dominance, it can be confirmed only by studying the ancestry or the progeny of the individual. Thus, for example, in pea, two different genotypes, **RR** and **Rr**, have the same phenotype, *viz.,* round seeds, but wherease the former produces nothing but round seed, the latter produces round seeds and wrinkled seeds in the ratio of 3 round : 1 wrinkled.

Back cross and Test cross

Back cross is a cross between a hybrid and either of its parents whereas test cross is a cross between hybrid and a recessive homozygote.



Monohybrid back cross in pea

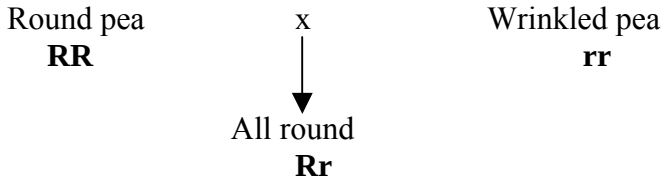
That individuals which are hybrid (heterozygous) for one pair of alleles produce two kinds of gametes in approximately equal numbers can also be shown by crossing the hybrid with its own recessive parent (i.e., back cross) or with any other recessive individual (i.e., test cross).

On crossing an F_1 hybrid with the recessive parent, about half the offspring formed show the dominant character and the other half show the character. For example,

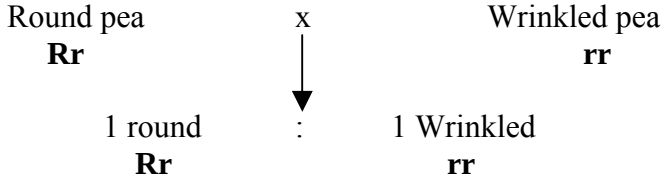
seeds obtained from a cross between a plant raised from the hybrid round seed, and the wrinkled-seeded parent are in the ratio of 1 round : 1 wrinkled.

As the recessive parent produces only one kind of gametes, all with the recessive allele, an 1 : 1 ratio is possible only if the F1 hybrid produces two kinds of gametes, one kind with the dominant allele and the other kind with the recessive allele, in approximately equal numbers.

That an individual exhibiting the dominant character is a pure (homozygous) or a hybrid (heterozygous) one can be found out if it is test crossed with a recessive individual. If the individual is a pure dominant, all the offspring will exhibit the dominant character, e.g.,



If the individual is a hybrid, the offspring will be in the ratio of 1 dominant : 1 recessive, e.g.,



Chromosomal theory of inheritance.

Mendel's Laws of Inheritance assume that the hereditary materials are particles called genes found in the cells of all living organisms. Genes have neither been seen nor analysed chemically but it is estimated that the diameter of one genes assuming it to be a spherical particle, is something like 6 millimicrons (0.000006 millimeter). Genes are thus fundamental units of life, just like atoms which are the ultimate units of matter.

Mendel established the existence of genes without knowing anything about chromosomes, in fact, several years before chromosomes had been named or described in detail.

The regular and precise longitudinal division of the chromosomes into two identical halves and the distribution of the two halves to the two daughter cells by mitosis, the neat separation of chromosomes and the reduction in the number of chromosomes from the diploid (2n) to the haploid state (n) during the formation of gametes by meiosis and the restoration of the diploid number of chromosomes in the zygote by fertilisation showed that the chromosomes are of great importance to the cell.

Hypothesis of Sutton and Boveri

The hypothesis that the Mendelian genes must be carried on the chromosomes was put forth simultaneously, but independently, in 1902 by Sutton, an American biologist, and Boveri, a German cytologist.

The chromosomes maintain their individual identity, just as do genes. In favourable materials, each pair of chromosomes can be seen to be different from every other pair. Similarly genes have an individuality as can be inferred from the specific effects produced by each gene.

Chromosomes are found in pairs, each member of which has been derived from one of the two parents. The facts of inheritance can be satisfactorily explained only on the assumption that genes also occur in pairs, one member of each pair being contributed by one parent and the other by the other parent.

Proofs for the theory of Sutton and Boveri

The first definite suggestion that a chromosome determines a character came from McClung, an American zoologist, when he discovered that the male grasshoppers differ from the females in the absence of one chromosome. The female has an even number of chromosomes, all the chromosomes being in pairs. All the eggs produced by the XX female are alike in having a single X chromosome each. The male, however, has an odd number of chromosomes, one of the chromosome being always without a partner. Two types of sperms are produced in equal numbers by the XO male, one type with the odd X chromosome and the other without it. Since the eggs are all alike and the two kinds of sperms are equal in number, the ratio of 1 female : 1 male observed in the offsprings is possible only if eggs fertilised by sperms with the X chromosomes develop into females and those fertilised by sperms without the X chromosome develop into males. That the X chromosome determines sex is seen from the fact that the two types of sperms differ only in that, one type has a X chromosome while the other lacks it.

Morgan's proof for the chromosome theory

The discovery of sex-linked genes by Morgan in 1910 furnished another proof for the chromosomal theory of inheritance. He showed that the transmission of the recessive gene for white colour of the eye in Drosophila melanogaster depends upon the sex which carries the gene initially.

In a cross between a red-eyed female and a white-eyed male, the F_1 flies of both sexes are red-eyed. Of the F_2 offsprings, all the females are red-eyed, whereas half the males are red-eyed and the other half are white-eyed. The F_2 shows a segregation of 3 red : 1 white, but strangely enough, the white-eyed flies are always male.

In the reciprocal cross between a white-eyed female and a red-eyed male, the F_1 females are red-eyed. In the F_2 generation, one half of the females and males are red-eyed and the other half white eyed.

The different results from the reciprocal crosses can be explained only on the assumption that the gene for colour of the eyes is located on the X chromosome. Morgan thus showed that the distinctive pattern of inheritance eof sex-linked genes parallels the transmission of the X chromosome.

Morgan's Theory

The work of Morgan and Bridges firmly established the fact that specific genes are borne on specific chromosomes. Study of linkage and crossing over in Drosophila melanogaster by Morgan, Sturtevant, Muller and Bridges threw more light on the genes on the one hand and the chromosome on the other.

From the co-oridnated genetic and cytological studies on Drosophila, Morgan postulated that genes are arranged in a linear order along the length of the chromosome, each gene having a fixed place on the chromosome, and its allele a corresponding position on the homologous chromosome. He also put forward the hypothesis that the degree of linkage depends upon the distance between the linked genes in the chromosome. This led to a new field dealing with mapping of chromosomes.

Lecture VI

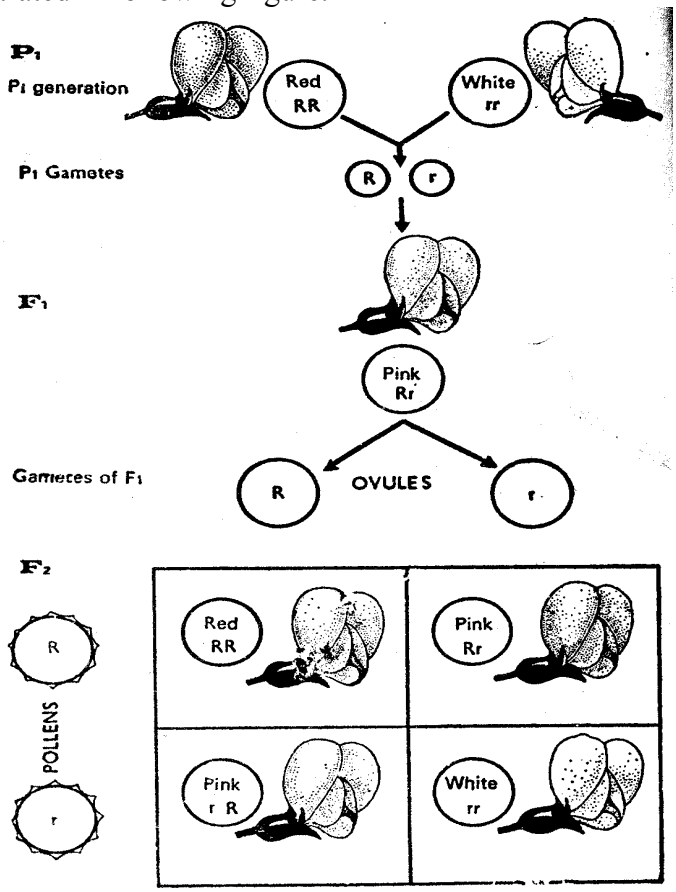
Types of dominance – Complete dominance, Incomplete dominance, Co-dominance and Over dominance with examples – Lethal genes, Pleiotropy with examples – Phenocopy, penetrance and expressivity.

INCOMPLETE DOMINANCE

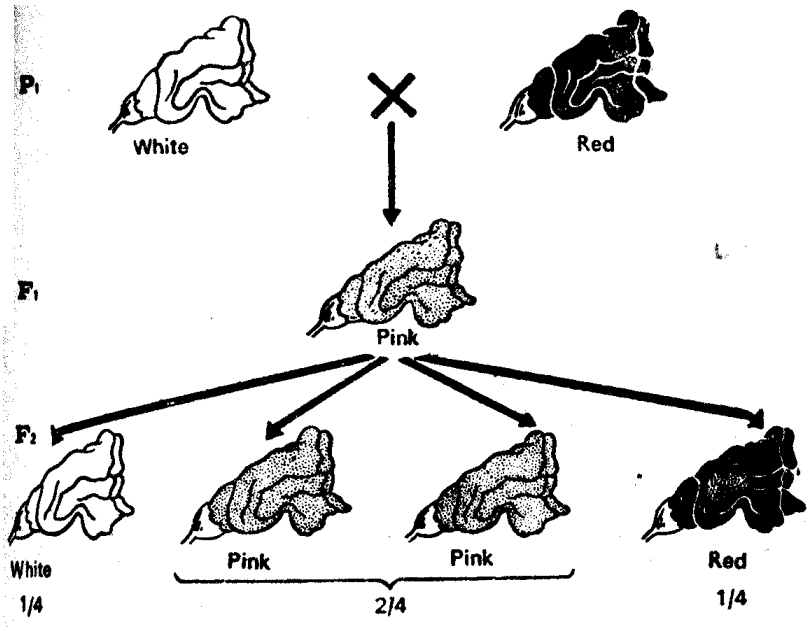
When a dominant allele does not mask completely the phenotypic expression of the recessive allele in a heterozygote, then a blending of both dominant and recessive traits takes place in the F_1 and F_2 heterozygotes. This phenomenon is know as incomplete or partial dominance. In such cases, the blending occurs only in the phenotype of the F_1 heterozygotes and the alleles maintain their individual identities and segregate from each other during gametogenesis. The F_1 gametes produce F_2 progeny having the phenotypic and genotypic ratios of 1 : 2 : 1.

Examples of incomplete dominance in plants

1. When a homozygous red flowered pea plant is crossed with a homozygous white flowered pea plant, the F_1 heterozygotes are found to have pink flowers. When the F_1 pink flowered heterozygotes are self crossed, they produce a F_2 progeny having identical phenotypic and genotypic ratio of 1 red (RR) : 2 pink (Rr) : 1 white (rr), as has been illustrated in following figure.



2. The snapdragons (*Antirrhinum majus*) and four-o'clock plants (*Mirabilis jalapa*) also provide good examples of incomplete dominance. When a homozygous red flowered snapdragon or four-o'clock plant is crossed with a homozygous white flowered snapdragon or four-'clock plant, they produce a F_1 progeny of pink flowered heterozygotes. The F_1 heterozygotes produce the F_2 progeny with identical phenotypic and genotypic ratios of 1 : 2 : 1 as has been illustrated for snapdragon in following diagram.

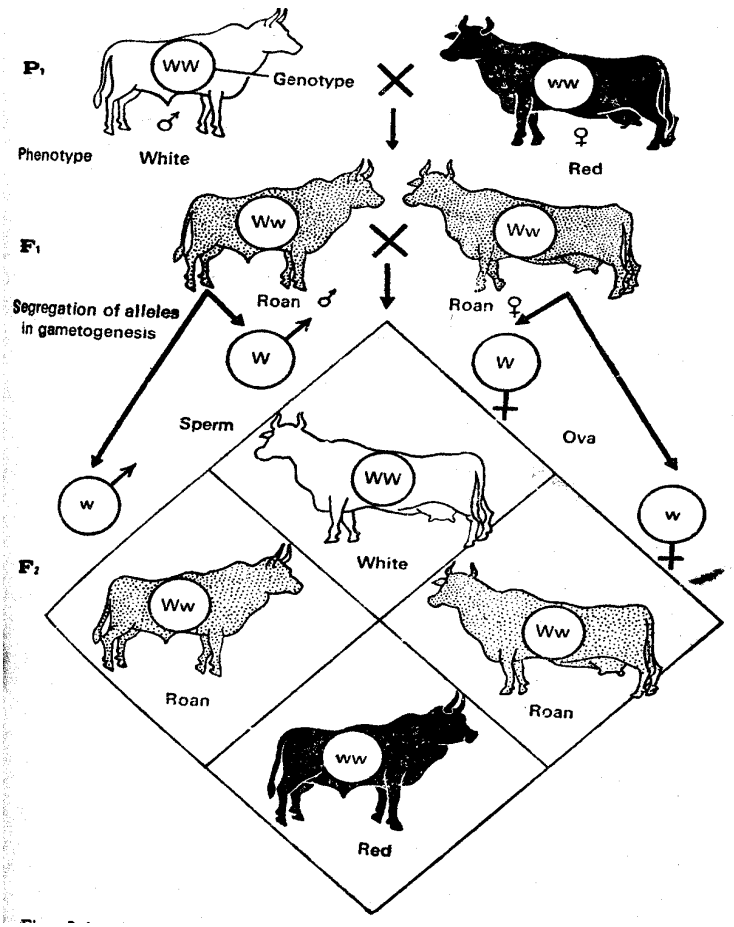


CO-DOMINANCE

In the phenomenon of co-dominance, both dominant and recessive alleles lack their dominant and recessive relationships and both have capability to express them phenotypically, in the heterozygous condition. In a heterozygote of co-dominant nature, the dominant and recessive traits occur side by side. The F_1 heterozygotes produce a F_2 progeny in the phenotypic and genotypic ratios of 1 : 2 : 1 like the incomplete dominance.

Example of Co-dominance

The best example of co-dominance is found in cattles. In one breed of cattle a genotype of WW will be expressed phenotypically in white coat colour, while the genotype of ww into a red coat colour. When a white-coated cattle is crossed with a red-coated cattle, the F_1 heterozygote are found to have a phentoype of redish gray or roan colour. The roan coat of a F_1 heterozygote has no hair of intermediate colour between red and white, but rather has a mixture of red hair and white hairs. The F_1 heterozygotes produce a F_2 progeny of phenotypic and genotypic ratios of 1 : 2 : 1 as has been illustrated.



OVER DOMINANCE (HETERO DOMINANCE)

When the heterozygotes have a more extreme phenotype than either of the corresponding homozygotes (homozygous parents), then it is usually referred to as over-dominance, super-dominance or hetero-dominance (Serra, 1959). For example, there is heterodominance, when the heterozygote Aa between a pair of factors which control size is bigger than the homozygotes AA or aa. This type of allelic relation which implies interaction between the alleles, or of these with other factors of the genotype, may be found in quantitative characters and especially those such as size, production, vigour, etc., which are of importance in the breeding of animals and plants.

LETHAL GENE

The term lethal is applied to those changes in the genome of an organism which produce effects severe enough to cause death. Lethality is a condition in which death of a certain genotype occurs prematurely. The fully dominant lethal allele kills the carrier individual both in its homozygous and heterozygous conditions. It occasionally arises by mutation from a normal allele. The individuals with a dominant lethal allele die before they can produce the progeny. Therefore, the mutant dominant lethal allele is removed from the population in the same generation in which it arose.

The recessive lethal allele kills the carrier individual only in homozygous condition. They may be of two kinds (I) one which has no obvious phentoypic effect in hetrozygotes and (ii) one which exhibits a distinctive phenotype when in heterozygous condition.

The completely lethal genes usually cause death of the zygote, later in the embryonic development or even after birth or hatching. Complete lethality, thus is the case where no individual of a certain genotype attain the age of reproduction. However, in many cases lethal genes become operative at the time the individuals become sexually mature. Such lethal genes which handicap but do not destroy their possessor are called subvital, sublethal or semilethal genes. The lethal alleles modify the 3 : 1 phenotypic ratio into 2 : 1.

Types of Lethality

Lethality may manifest itself in different nuclear phases. In diploid organism lethality may act during the diplophase, which follows zygote formation, or it may kill the gametes or other haploid cells. The former constitute zygotic lethality, the second is gametic lethality and in plants also gametophytic lethality. Haplolethality, whether the haploid phase is prolonged or limited to the gamets, occurs when only one dose of a particular gene is present and there arises no question of dominance.

Examples of lethal alleles or complete lethality in plants

1. In snapdragons three types of plants occur : 1. Green plants with chlorophyll ; 2. Yellowish green plants with carotenoids, usually are referred to as golden or auria plants and 3. White plants without any chlorophyll. The homozygous green plants have the genotype CC and the homozygous white plant has the genotype cc. The auria plants have the genotype Cc because they are heterozygotes of green and white plants. When two such auria plants are crossed, the F1 progeny has identical phenotypic and genotypic ratio of 1 : 2 : 1 (*viz.,* 1 green (CC) : 2 auria (Cc) : 1 white (cc). But the white plants because lack chlorophyll pigment, therefore die to modify the ratio of 1 : 2 : 1 into 1 : 2 or 2 : 1. In this case the homozygous recessive genotype (cc) is lethal.

F_1 heterozygote :	Auria		Auria	
	Cc	x	Cc	
F_2 :	1 CC	:	2 Cc	: 1 cc
	Green		Auria	White (lethal)
	Or 1 CC	: 2 Cc	or 1 : 2	

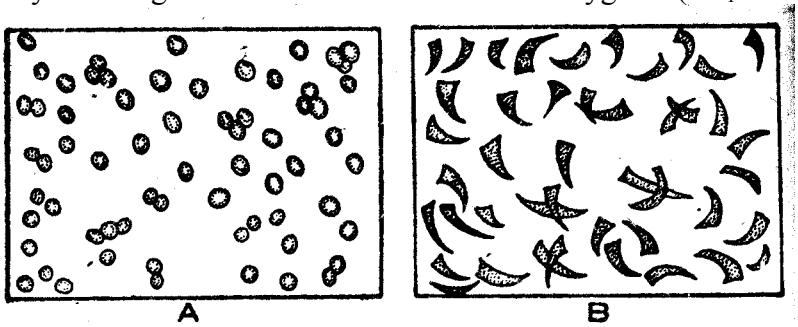
2. In maize (*Zea mays)* the amount of chlorophyll is controlled by a recessive allele (g) which exhibits a lethal effect in homozygous (gg) and in heterozygous condition (Gg) has phenotype similar to homozygous condition for dominant gene GG. It modifies 3 : 1 phenotypic ratio into 2 : 1.

F_1 heterozygote :	Green		Green	
	Gg	x	Gg	
F_2 :	1 GG	:	2 Gg	: 1 gg
	Green		Green	White (lethal)
	Or 1 GG	: 2 Gg	or 1 : 2 or 2 :1	

Lethal Genes in Man

In man several hereditary diseases have lethal effects. Few important lethal genes of man are following:

1. **Congenital ichthyosis** – One of the most typical cases of a recessive lethal gene in man is expressed in congenital ichtyosis. At birth children afflicted with this disease have a crusted leathery skin with deep fissures down to the subcutaneous tissue; the fissures lead to bleeding, infection and death. Congenital ichthyosis occurs only when there occur homozygous condition for its recessive lethal genes.
2. **Amaurotic idiocy** – A recessive allele in homozygous condition causes a fatal disease called Amaurotic idiocy in juvenile stage. Bearers of this genotype begin to lose their eye sight between the age of four to seven years. The complete blindness is followed by mental degeneration and finally death before adolescence.
3. **Cooley's anemia** – Among certain African tribes, a co-dominant gene Hb1g in homozygous condition produces a syndrome (disease) called sickle cell anemia which leads to death, generally at least by late adolescence. In the blood of such persons the erythrocytes become distorted, many being essentially sickle-shaped. such cells not only impede circulation by blocking capillaries, but also cannot properly perform their function of carrying oxygen and carbon dioxide to and from the tissues. Under normal conditions, heterozygous (Hb_1^A Hb_1^S) manifest none of these symptoms, being outwardly indistinguishable from the normal homozygotes (Hb_1^A Hb_1^A).



Human blood smear showing normal (A) and sickle erythro cytes (B) (after Levine, 1969).

Besides these cases of complete lethality in man, there are cases of sublethal genes. Sublethal genes produce less than 50% mortality. The examples of sublethal genes of man are following:

1. Retinoblastoma – It is a human disease which is caused by a dominant mutant gene and is charcterized by the growth of tumours in eyes. This gene causes mortality in 50% childern only.

2. Epilopia – This disease is caused by a dominant lethal gene in heterozygous condition. It is characterized by mental deficiency, tumoors in organs and abnormal growth of the skin. Persons suffering from epilopia disease die in childhood but some of them survive and produce children.

3. Huntingdon's chorea – Huntingdon's chorea is lethal disease of man which is characterized by involuntary jerking of the body and a progressive degeneration of the nervous system, accompanied by gradual mental and physical deterioration. The patients of this disease die in the age of forty of forty five. This disease in caused by a dominant lethal gene.

PLEIOTROPY

Until now we have observed that a specific gene has a specific effect upon a specific phenotypic trait or in other words, each gene (allele) has its relation with a single phenotypic trait. But a single gene often influences more than one phenotypic trait. However, it may be that one gene may cause evidently well marked expression of some phenotypic trait (major effect) than the others with less evident phenotype (secondary effect). Most genes have their multiple effects and are called pleiotropic genes. The phenomenon of multiple effect (multiple phenotypic expressions) of a single gene is called pleiotropism.

Examples of Pleiotropism

1. In *Drosophila* the recessive gene for vestigial wings cause vestigial wings in homozygous condition. However, careful observations show that other traits as well are affected – (I) the tiny wig like balancer behind the wings : (ii) certain bristles ; (iii) the structure of the reproductive organs ; (iv) egg production is lowered, and (v) longevity is reduced.

2. In human, the gene for disease phenylketonuria has pleiotorpic effect and produces various abnormal phentoypic traits, collectively called syndrome. For example, the affected individuals secrete excessive quantity of amino acid phenylalanine in their urine, cerebrospinal fluid and blood. They become short stature, mentally deficient, with widely spaced incisors, with pigmented patches on skin, with excessive sweating and with non-pigmented hairs and eyes.

PHENOCOPY

The alteration of the phenotype, by nutritional factors or the exposure to environmental stress during development, to a form imitating that characteristically produced by a specific gene. Thus rickets due to a lack of vitamin D would be a phenocopy of vitamin-D-resistant rickets.

PENETRANCE AND EXPRESSIVITY

The ability of a given gene or gene combination to be expressed phenotypically to any degree is called penetrance. In other words, penetrance refers to the statistical regularity with which a gene produces its effect when present in the requisite homozygous (or heterozygous) state. It is of following two kinds:

1. Complete Penetrance

Most dominant and recessive genes in homozygous conditions and many completely dominant genes even in heterozygous condition give their complete phenotypic expressions. Such genes which always produce the expected phenotype are called to have complete penetrance. If only 70 per cent of the individuals of a stock homozygous for a certain recessive gene show the character phenotypically, the gene is said to have 70 per cent penetrance.

Examples of Complete Penetrance

1. In pea, the alleles (RR) for red flowers and the alleles (rr) for white flowers have complete penetrance in homozygous conditions.

2. In *Drosophila* the recessive alleles for vestigial wings in homozygous conditions have complete penetrance.

3. In guinea pigs the dominant allele 'B' for black coat has complete penetrance both in homozygous and heterozygous conditions.

2. Incomplete Penetrance

Some genes in homozygous as well as in heterozygous conditions fail to provide complete (cent per cent) phenotypic expression of them. Such genes are called to have incomplete penetrance.

Examples of Incomplete Penetrance

1. Polydactyly in man is thought to be produced by a dominant gene P. the normal condition with five digits on each limb is produced by the recessive genotype (pp). some heterozygous individuals (Pp) are not polydactylus and therefore has a penetrance of less than 70%.

2. In, man the tendency to develop diabetes mellitus (a condition in which there is an excess of sugar in the blood) is controlled by certain genes. However, not everyone carrying the genes for diabetes actually develops the conditions, for the genes have incomplete penetrance.

EXPRESSIVITY

A trait though penetrant, may be quite variable in its phenotypic expressions. The degree of effect produced by a penetrant genotype is called expressivity.

Example of expressivity

In man the polydactylous condition may be penetrant in the left hand (6 fingers) and not in the right (5 fingers) ; or it may be penetrant in the feet and not in the hands.

Lecture VII

Epistasis Vs Dominance – epistatic and hypostatic genes, Types of epitasis – Non-allelici interaction without modification in Mendelian ration – Bateson and Punnet's experiment on fowl comb shape.

Interaction of two pairs of alleles

When an individual forms gametes, the two members of each pair of alleles always separate from each other but the separation in one pair of alleles is independent of the separation in any other pair of alleles. Gametes, therefore, always contain any one allele of each of the several pairs of alleles found in an individual. At fertilisation, these gametes recombine at random to give rise to new individuals.

When different pairs of alleles influence the same character of an individual, it is likely that the expressions of these genes interact. As two different genes interact and affect the same character, such a genetic interaction is said to be intergenic or nonallelic. In nonallelic interactions different genes located on the same or different chromosomes interact with one another for the expression of a single phenotypic trait of an organism.

Intergenic or nonallelic interactions may suppress or mask the action of a gene at another locus or modify partially or completely the effect of another gene. This nonallelic interaction is otherwise called *epistasis.*

Definition: - A kind of interaction between genes belonging to different pairs of alleles, the dominant allele in one of the pairs preventing the dominant allele in the other pair from expressing itself. Thus, the gene A may be epistatic over B. B is then said to be hypostatic to A.

We shall now consider a few cases of independently transmitted pairs of alleles that are not independent in their expression.

Intergenic Non-epistatic interaction (9: 3 : 3 : 1 Ratio)

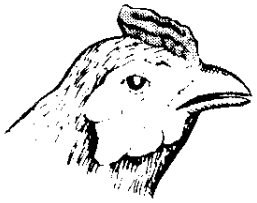
A classical case of two pairs of alleles affecting the same characteristic and producing in the F_2 four different phenotypes in the ratio of 9 : 3 : 3 : 1 was discovered in fowls by Bateson and Punnett.

Each breed of pultry possesses a characteristic type of comb. The Wyandotte breed has a comb known as the 'rose' comb, the Brahma has a 'pea' comb, the Leghorn has a 'single' comb and the Malay breed has a comb known as the 'walnut' comb. Each of these breeds true.

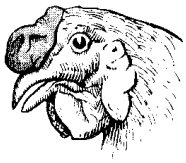
Reference: DDS: 91-94, PSC & VKA: 150

Rose: AAbb
or
Aabb

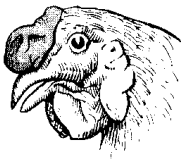
X



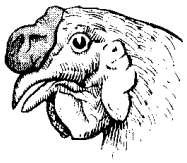
Pea: aaBB
or
aaBb



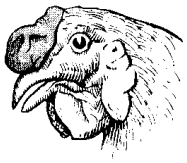
Walnut: A-B-



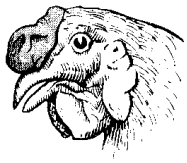
Walnut: A-B-



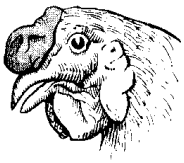
Walnut: A-B-



Walnut: A-B-

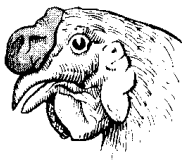


Walnut: A-B-



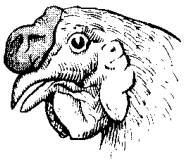
Walnut: A-B-

Rose: AAbb
or
Aabb

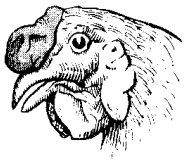


Walnut: A-B-

Rose: AAbb
or
Aabb



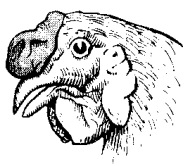
Walnut: A-B-



Walnut: A-B-

Pea: aaBB
or
aaBb

Pea: aaBB
or
aaBb



Walnut: A-B-

Rose: AAbb
or
Aabb

Pea: aaBB
or
aaBb

Single -aabb

Crosses between rose-combed and single-combed types show that rose is dominant to single comb and that there is a segregation of 3 rose : 1 single comb in the F_2. In matings between pea-combed and single-combed birds, pea comb is found to be dominant over single comb and a 3 : 1 ratio appears in the F_2.

When, however, a rose-combed fowl is crossed with a pea combed one, all the F_1 birds show the walnut comb. When the F_1 walnut combed birds are bred together, there appears in the F_2 9 walnut : 3 rose : 3 pea : 1 single comb.

These results can be interpreted as follows: The rose comb is due to a gene R and the pea is due to another gene P. The walnut comb is due to the presence of both the dominant genes, R and P and the single comb is due to their recessive alleles, r and p.

The breeding behaviour of the different genotypes of the F_2 is summarised.

F_2			Breeding behaviour
Phenotype	Genotype	Ratio	
Walnut	RRPP	1	All the progeny walnut-combed
	RRPp	2	3 walnut (RP) : 1 rose (Rp)
	RrPP	2	3 walnut (RP) : 1 pea (rP)
	RrPp	4	9 walnut : 3 rose : 3 pea : 1 single
Rose	RRpp	1	All the progeny rose-combed
	Rrpp	2	3 rose (Rp) : 1 single (rp)
Pea	rrPP	1	All the progeny pea-combed
	rrPp	2	3 pea (rP) : 1 single (rp)
Single	rrpp	1	All the progeny single-combed

The above example depicts a case of non-epistatic intergenic interaction in which two genes that determine the same character produce a new phenotype by mutual non-epistatic interaction.

Difference between dominance and epistasis

The phenomenon of dominance involve intra-genic or inter-allelic gene suppression, or the masking effect which one allele has upon the expression of another allele at the same locus, while the phenomenon of epistasis involves inter-genic suppression or the masking effect which one gene locus has upon the expression of another. The classical phenotypic ratio of 9:3:3:1 observed in the progeny of dihybrid parents becomes modified by epistasis into ratios which are various combinations of the 9:3:3:1 groupings.

Lecture VIII

Types of epistasis

1. Dominant epistasis (12:3:1),
2. Recessive epistasis (9:3:4)
3. Duplicate dominant epistasis (15:1)
4. Duplicate recessive epistasis (9:7)
5. Dominant and recessive epistasis (13:3)
6. Duplicate genes with cumulatie effect (9: 6 : 1)

1. Dominant Epistasis (12 : 3 : 1 Ratio)

In *Sorghum*, pearly grains are shining, translucent and oily white, and chalky grains are not shining but opaque and dull white. When a plant with pearly grains and another with chalky grains are crossed the F_1 is pearly. In the F_2 there is a segregation into 3 pearly : 1 chalky. The gene for pearly grains can be represented by Z and the gene for chalky by z.

The colour of the grains may be either red or white. When a plant with red grains is crossed with one with white grains, the F_1 is red and the F_2 shows a segregation of 3 red : 1 white. The gene for red grains is represented by W and white by w.

When the colour of the grain is white, it is possible to say whether it is pearly or chalky, but when the colour is red, it is not possible to find out whether it is pearly or chalky. One character, the red colour of the grain, masks another character, the pearliness

When two non-allelic genes affect the same part or trait of an organism, it is likely that the expression of one covers up or hides the expression of the other. A gene which thus masks the expression of another gene which is not its allele is said to be **epistatic** to it and the gene which is hidden is said to be **hypostatic**. Epistasis is the dominating influence of one gene over another which is not its allele and is similar to dominance expect that it occurs between different genes instead of between the members of an allelic pair. Dominant epistasis is also called Dominant suppressor.

The gene W is epistatic to those for pearliness Z and chalkiness z and so long as it is present, pearlines or chalkiness cannot be distinguished from one another.

Where this gene W is lacking, i.e., where the genotype is ww, the grains will be pearly if Z is present and chalky if Z is absent.

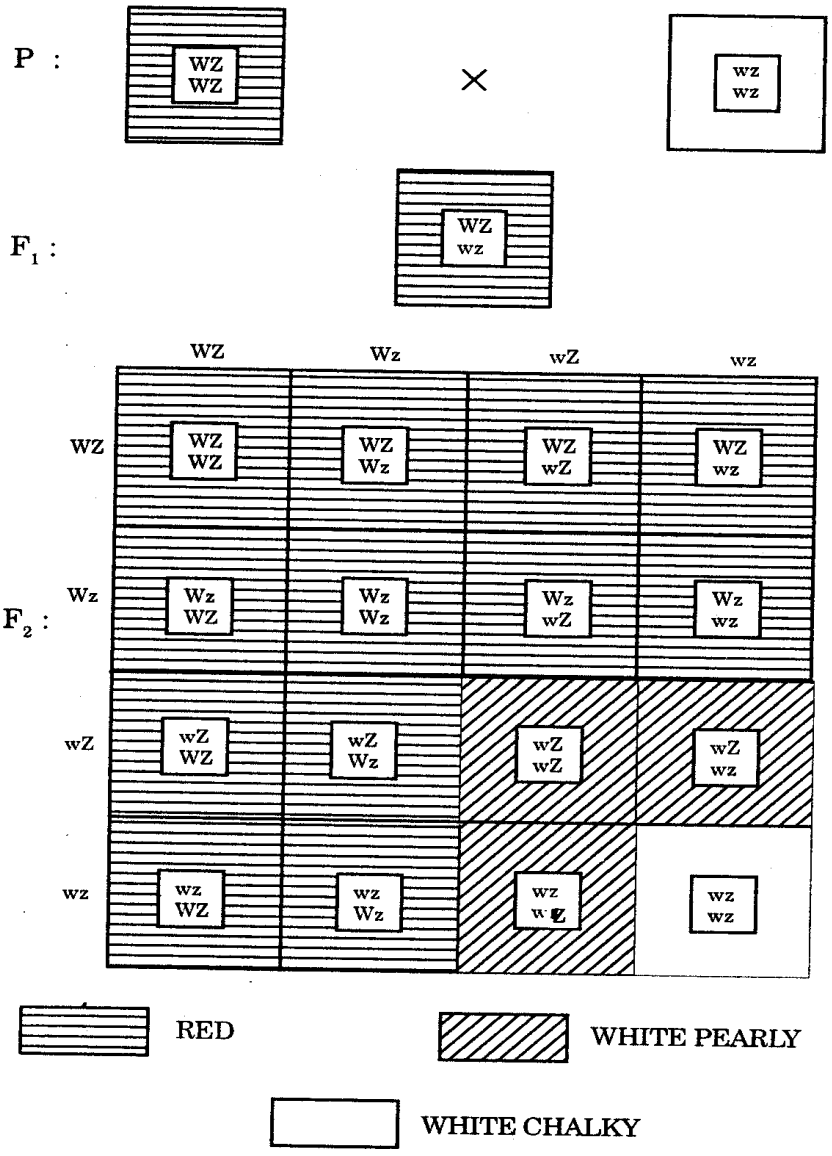
Thus W masks every thing that is hypostatic to it so that Z which segregates quite independently of W produces a visible expression only when W is absent.

Reference: DDS: 94 - 110

When a plant with red grains is crossed with one with white chalky grains, the F_1 is red and the F_2 segregates as follows:

3 W	3Z	=	9 WZ	Red
	1z	=	3 Wz	
1 w	3Z	=	3 wZ	White pearly
	1 z	=	1 wz	White chalky

The F_2 thus shows a ratio of 12 red : 3 white pearly : 1 white chalky.



Checkerboard showing the 12 : 3 : 1 ratio in Sorghum

The breeding behaviour of the different genotypes of the F_2 is summarised.

Inheritance of colour of grain in Cholam (Sorghum).

F_2			F_3
Phenotype	Genotype	Ratio	
Red	WWZZ	1	All red
	WWZz	2	All red (3 WZ : 1 Wz)
	WwZZ	2	3 red (WZ) : 1 white pearly (wZ)
	WwZz	4	12 red : 3 white pearly : 1 white chalky
White pearly	wwZZ	1	All white pearly
	wwZz	2	3 white pearly (wZ) : 1 white chalky (wz)
White chalky	wwzz	1	All white chalky

Epistasis in Cucurbita

An excellent example of the epistasis of one gene over another has been recorded in summer squash (*Cucurbita pepo)* by Sinnott and Durham.

The colour of the fruit in summer squash may be white, yellow or green. In crosses between white and yellow and between white and green, white is always found to be dominant. In crosses between yellow and green, yellow is found to be dominant.

When a particular plant with white fruits is crossed with one with green fruits, the F1 has white fruits. In the F2 there are 12 white : 3 yellow : 1 green.

Evidently, the gene W for white is epistatic to the gene Y for yellow and y for green and so long as this gene W is present, the colour of the fruit is only white. Where the gene W is absent, the colour of the fruit will be yellow if gene Y is present and green if gene Y is absent.

P	White	X	Green
	WWYY		**wwyy**
F1		White	
		WwYy	
F2	9 **W – Y –**	=	12 white
	3 **W – yy**		
	3 **wwY -**	=	3 yellow
	1 **wwyy**	=	1 green

(The dashes denote the dominant alleles which may be either homozygous or heterozygous. For example, wwY – may be wwYY or wwYy).

Thus, in dominant epistasis a dominant allele of one gene masks the expression of a dominant or recessive allele of another gene.

2. Recessive Epistasis (9 : 3 : 4 Ratio)

In a cross between a cholam (*Sorghum)* plant with blackish purple leaf sheath and another with brown leaf sheath, the F_1 is found to be blackish purple. In the F_2 there is segregation into 3 blackish purple : 1 brown. The gene for blackish purple can therefore be represented by P and brown by p.

In another cross between a blackish purple and a brown plant, the F_1 is found to be reddish purple. The F_1 is expected to be blackish purple because blackish purple is dominant to brown. Actually however, the F_1 is reddish purple. Evidently, there is another gene which is responsible for converting the blackish purple colour into reddish purple. This gene is called a supplementary gene, Q and it adds to the effects of blackish purple.

In the F_2 there is a segregation into 9 reddish purple : 3 blackish purple : 4 brown.

The gene P is responsible for the blackish purple colour and its allele p for the brown colour. When the supplementary gene Q is found in combination with the gene P for blackish purple, the leaf sheath is reddish purple. The gene Q, however, has no phenotypic expression of its own and plants will therefore be brown whether they possess Q or not, if they lack the gene P.

P	Blackish purple	X	Brown	
	PPqq		**ppQQ**	
F_1	Reddish purple			
	PpQq			

Genotypic ratio in the F_2

1 **PP**	1 **QQ**	=	1 **PPQQ**	Reddish purple
	2 **Qq**	=	2 **PPQq**	Reddish purple
	1 **qq**	=	1 **PPqq**	Blackish purple
2 **Pp**	1 **QQ**	=	2 **PpQQ**	Reddish purple
	2 **Qq**	=	4 **PpQq**	Reddish purple
	1 **qq**	=	2 **Ppqq**	Blackish purple
1 **pp**	1 **QQ**	=	1 **ppQQ**	Brown
	2 **Qq**	=	2 **ppQq**	Brown
	1 **qq**	=	1 **ppqq**	Brown

Phenotypic ratio in the F_2

3 **P**	3 **Q**	=	9 **PQ**	Reddish purple
	1 **q**	=	3 **Pq**	Blackish purple
1 p	3 **Q**	=	3 **pQ**	Brown
	1 **q**	=	1 **pq**	

This type of epistasis is called recessive epistasis, since the recessive allele of one gene masks the phenotypic expression of the dominant or recessive allele of another gene.

3. Duplicate dominant epistasis (15 : 1)

In jowar (Sorghum), plants with starchy grains breed true.

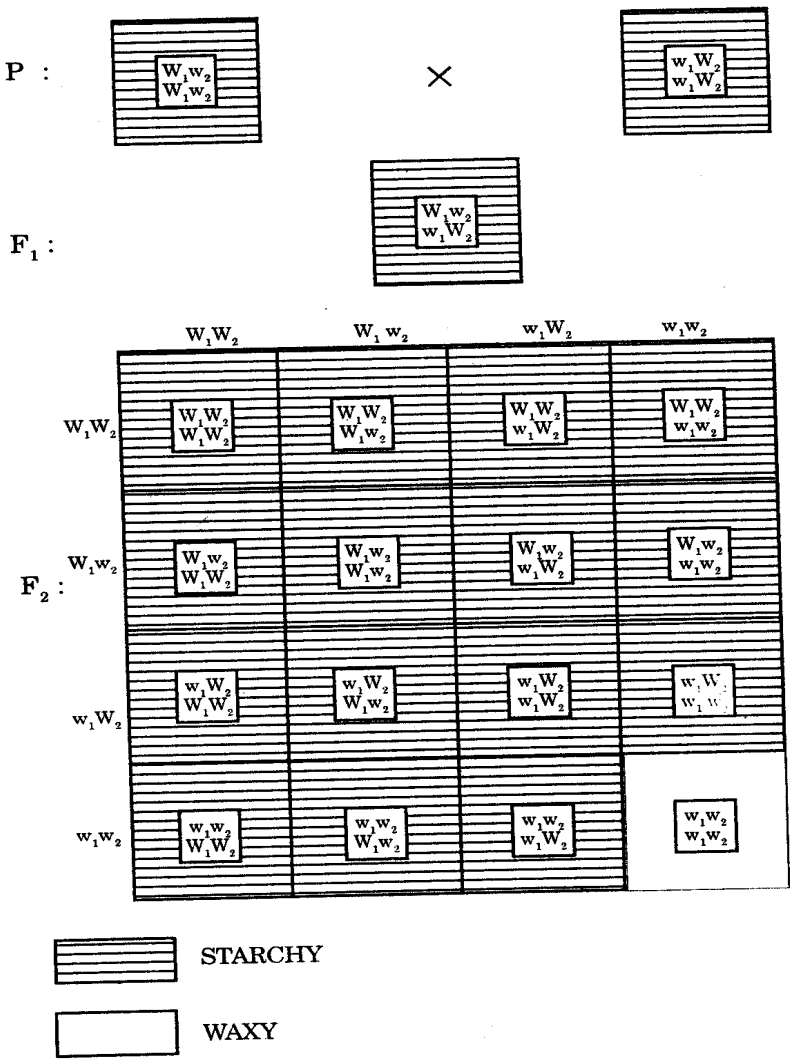
In a cross between a plant with starchy grains and a plant with waxy grains, the F_1 is starchy and the F_2 shows a segregation of 3 starchy : 1 waxy.

In a cross between a second plant with starchy grains and a plant with waxy grains, the F_1 is again found to be starchy and the F2 again shows a segregation of 3 starchy : 1 waxy.

In a cross between the first plant with starchy grains and the second plant with starchy grains, the F_1 is found to be starchy but the F_2 shows a segregation of 15 starchy : 1 waxy.

Starchy grain is evidently due to the presence of a dominant gene Wx, (for the sake of simplicity, denoted as W_1) or another dominant gene Wx_2 (for the sake of simplicity, denoted as W_2) or both. When both the dominant genes are absent, the grain is waxy.

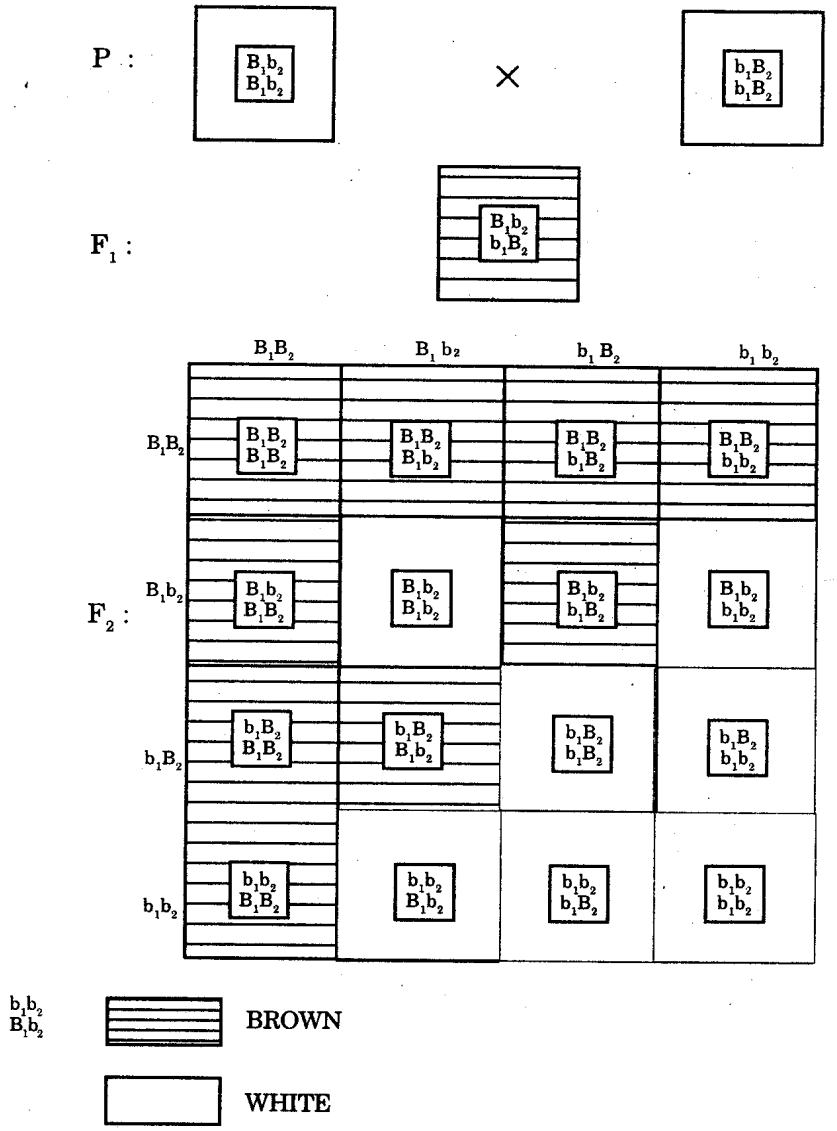
Duplicate genes are two pairs of alleles, either alone or together, producing the same effect. They are identical genes but are situated on two different pairs of chromosome. Each gene is dominate to its allele but does not add to the effect of the other. It is conventional to designate two such genes by the same letter followed by different numerical subscripts, as W_1 and W_2.



Checkerboard showing the 15 : 1 ratio in Sorghum

4. Duplicate recessive epistasis (9:7)

In *Sorghum*, plants with white grains, when self fertilised, produce progeny all of which have white grains, i.e., they breed true. When however, two true-breeding white grained plants are artificially crossed, the F1 is not white but brown. In the F_2, brown and white appear in the proportion of 9 : 7.



Checkerboard showing the 9 : 7 ratio in *Sorghum*

Since the white-grained plants are pure-breeding, they are homozygous but they cannot have identical genotypes, because the two white-grained parents, when crossed, give rise to brown grained progeny. The two white-grained parents are therefore homozygous but with different genotypes.

The brown colour of the grains is presumably due to the presence of two dominant genes, B_1 and B_2. The two white-grained parents are therefore homozygous for one or other of these two dominant genes but because they have different genotypes (due to the fact that they give rise to brown-grained progeny on hybridisation) one white-grained parent is assumed to be $B_1B_1b_2b_2$ and the other white-grained parent to be $b_1b_1B_2B_2$. One parent is white because it lacks B_2 and the other parent is white because it lacks B_1 but a cross between two such white-grained parents produces offspring with both the dominant genes B_1 and B_2 and consequently, brown grains. These genes responsible for the brown colour of grains are called complementary genes.

Two non-allelic dominant genes that act together to produce a phenotype different from that produced by the homozygous recessive of the one or the other or both are called complementary genes.

Complementary genes are differentiated from one another by addicting different alphabetic subscripts to the same letter, e.g., B_m and B_n but, for the sake of simplicity, the symbols B1 and B2 have been adopted in the present discussion.

The expectations in the F_2 from a cross between two plants with white grains are shown.

The expectations in the F_2 from a cross between white-grained Sorghum plants.

F_2			F_3
Phenotype	Genotype	Ratio	
Brown	$\mathbf{B_1B_1B_2B_2}$	1	All brown
	$\mathbf{B_1B_1B_2b_2}$	2	3 brown ($\mathbf{B_1B_2}$) : 1 white ($\mathbf{B_1b_2}$)
	$\mathbf{B_1b_1B_2B_2}$	2	3 brown ($\mathbf{B_1B_2}$) ; 1 white ($\mathbf{b_1B_2}$)
	$\mathbf{B_1b_1B_2b_2}$	4	9 brown : 7 white
White	$\mathbf{B_1B_1b_2b_2}$	1	All white
	$\mathbf{B_1b_1b_2b_2}$	2	All white (3 $\mathbf{B_1\ b_2}$ **:** 1 $\mathbf{b_1\ b_2}$)
	$\mathbf{B_1b_1B_2B_2}$	1	All white
	$\mathbf{b_1b_1B_2b_2}$	2	All white (3 $\mathbf{b_1\ B_2}$ **:** 1 $\mathbf{b_1\ b_2}$)
	$\mathbf{b_1b_1b_2b_2}$	1	All white

Complementary genes in sweet peas

Bateson and Punnett discovered that the F_1 of a cross between two white flowered strains of the Emily Henderson sweet pea, Lathyrus odoratus, was purple flowered. When the F_1 plants were self-fertilised, they produced offspring consisting of about nine-sixteenths purple flowered plants and seven-sixteenths white flowered ones.

Both parents were found to be true breeding and therefore homozygous. The were phenotypically identical in every respect but genotypically different, as otherwise, the F1 would have been white flowered.

It was therefore conclude that the purple colour results from an interaction between two different dominant genes, on e from one white flowered parent and the other from the other white flowered parent and the white colour is due to the absence of either or both of these dominant genes.

P	White	X	White
	$P_1P_1p_2p_2$		$p_1p_1P_2P_2$
F_1		Purple	
		$P_1p_1P_2p_2$	
F_2	9 P_1 – P_2 -	= 9 purple	
	3 P_1 – p_2 p_2 3 p_1 p_1 P_2 – 1 p_1 p_1 P_2 P_2	= 7 White	

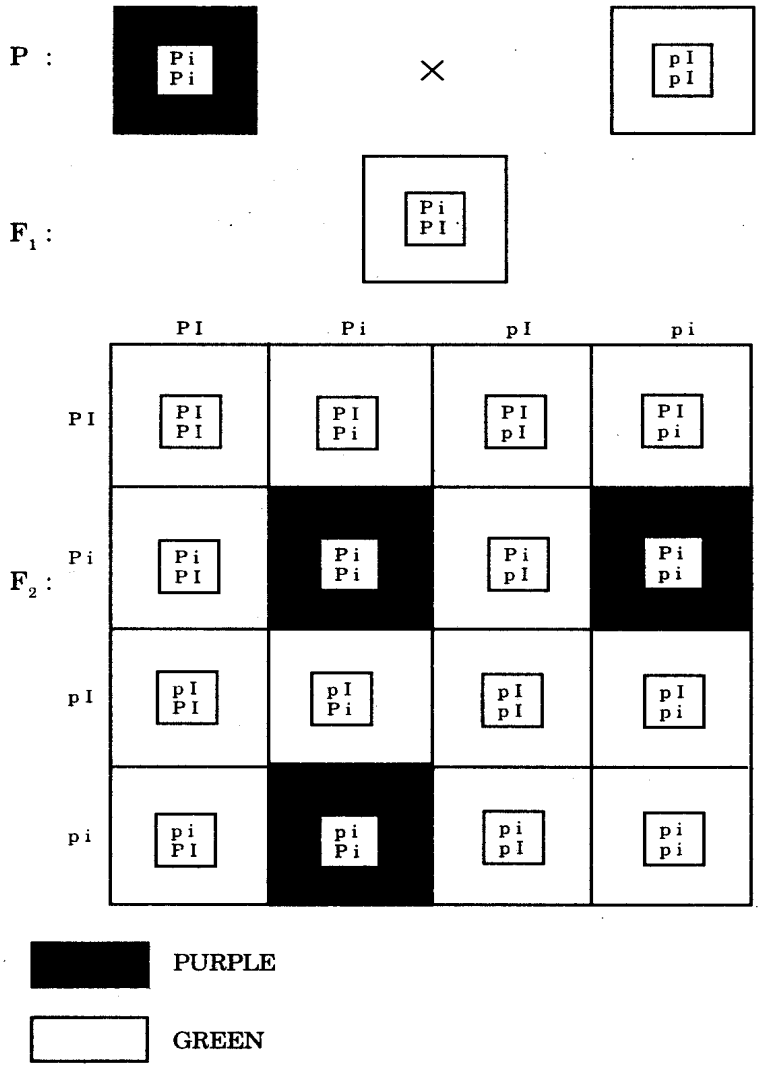
5. Dominant and Recessive epistasis (13 : 3)

In crosses between Sorghum plants with purple node and plants with green node, the F_1 hybrids are with purple node. In the F_2 there is a segregation into 3 purple : 1 green. The gene for purple node P_j (denoted for convenience as P) is therefore dominant over that for green p (denoted as p).

In a certain cross between a Sorghum plant with purple node and one with green node, the F1 is with green node. Since purple is dominant over green, the F_1 is excepted to be purple but it is observed to be green. The gene for purple node is unable to express itself probably because of the presence of another gene. This gene is called an inhibitory gene and is represented by I. this gene is capable of inhibiting the production of purple colour in plants with P.

Among the F_2, 13 are green and 3 are purple. This is because plants are purple only if they possess the gene for purple colour P in the absence of the inhibitory gene. In the presence of the inhibitory gene I, plants with the gene for purple P are unable to exhibit the purple colour and are only green. Plants which do not have the gene for purple colour are green whether they have the inhibitory gene or not.

The inhibitory gene I has thus no phenotypic expression of its own and its presence can be judged only by its effect on the gene for purple P.



Checkerboard showing the 13 : 3 ratio in Sorghum.

The expectations in the F_2 and F_3 from a cross between a plant with purple node and a plant with green node are shown.

Inheritance of colour of node in Great Millet.

F_2			F_3
Genotype	Ratio	Phenotype	
PPII	1	Green	All green
PPIi	2	Green	3 green (PI) : 1 purple (Pi)
PpII	2	Green	All green (3 PI : 1 pI)
PpIi	4	Green	13 green : 3 purple
Ppii	1	Purple	All purple
Ppii	2	Purple	3 purple (Pi) : 1 green (pi)
ppII	1	Green	All green
ppIi	2	Green	All green (3 pI : 1 pi)
ppii	1	Green	All green

Inhibitor genes in Poultry

If white Leghorns are crossed with white Wyandottes, the F_1 is white but the F_2 segregates into 13 white : 3 coloured.

White Leghorns (CCII) have a gene C for production of colour but they are white because they have in addition a gene I which inhibits the expression of colour. White Wyandottes (ccii) are white because they lack the gene can produce colour.

Colour is produced only when the gene C for colour is found in the absence of the gene I which inhibits the expression of colour.

An inhibitory gene is one that has no phenotype of its own but which prevents the expression of a non-allelic dominant gene.

Thus in the case of dominant and recessive epistasis, the dominant allele of one gene in homozygous or heterozygous condition and the homozygous recessive allele of another gene produce the same effect.

The inhibitory gene is also called Recessive suppressor.

6. Duplicate genes with cumulative effect (9 : 6 : 1)

In barley, plants with light purple grains breed true.

In a cross between a plant with light purple grains and a plant with white grains, the F_1 is light purple and the F_2 shows a segregation of 3 light purple : 1 white.

In a cross between a second plant with light purple grains and a plant with white grains, the F_1 is light purple and the F_2 again shows a segregation of 3 light purple : 1 white.

In cross between a first plant with light purple grains and the second plant with light purple grins, the F_1 is found to be dark purple. The F_2 segregation is 9 dark purple : 6 light purple : 1 white.

Light purple colour of the grain is evidently due to the presence of a dominant gene P_1 or another dominant gene P_2. The two non-alleliuc dominant genes P_1 and P_2 possess an additive effect and the colour of the grain is dark purple when the gene P_1 and P_2 are present together. When both the dominant genes are absent, the colour of the grain in white.

P	Light purple	x	Light purple
	$\mathbf{P_1P_1p_2p_2}$		$\mathbf{p_1p_1P_2P_2}$

F_1	Dark purple
	$\mathbf{P_1p_1P_2p_2}$

Genotypic ratio in the F_2

1 **P_1 P_1**	1 **P_2 P_2**	=	1 **P_1 P_1 P_2 P_2**	Dark
	2 **P_2 p_2**	=	2 **P_1 P_1 P_2 p_2**	Dark
	1 **p_2 p_2**	=	1 **P_1 P_1 p_2 p_2**	Light
2 **P_1 p_1**	1 **P_2 P_2**	=	2 **P_1 p_1 P_2 P_2**	Dark
	2 **P_2 p_2**	=	4 **P_1 p_1 P_2 p_2**	Dark
	1 **p_2 p_2**	=	2 **P_1 p_1 p_2 p_2**	Light
1 **p_1 p_1**	1 **P_2 P_2**	=	1 **p_1 p_1 P_2 P_2**	Dark
	2 **P_2 p_2**	=	2 **p_1 p_1 P_2 p_2**	Dark
	1 **p_2 p_2**	=	1 **p_1 p_1 p_2 p_2**	Light

Phenotypic ratio in the F_2

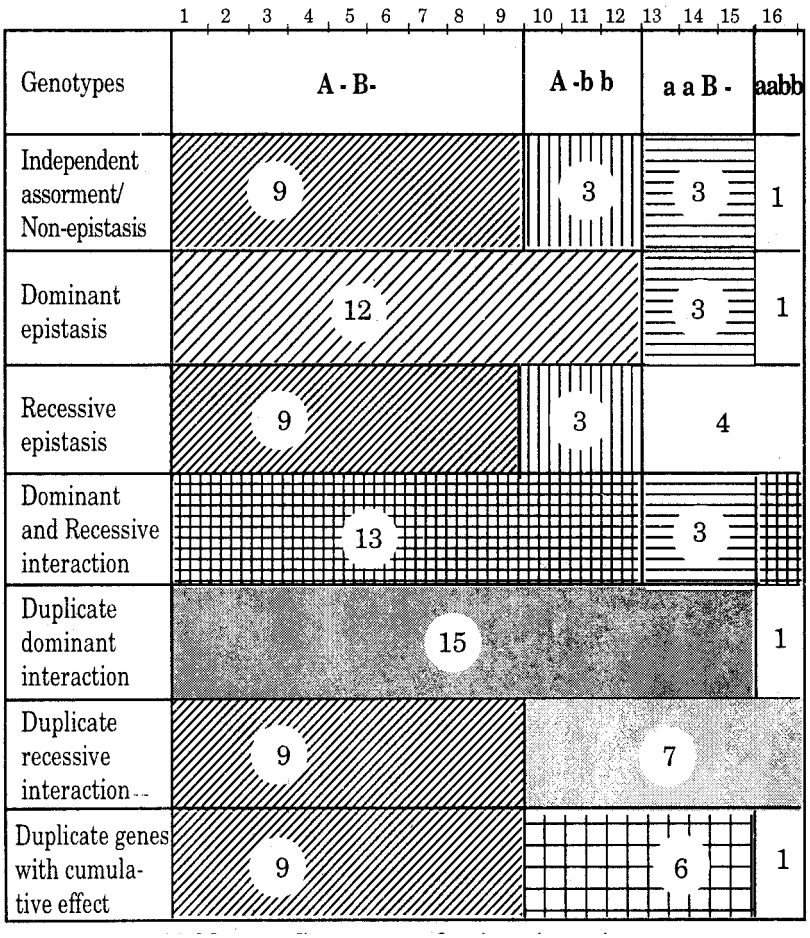
3 **P_1**	3 **P_2**	=	9 **P_1 P_2**	Dark
	1 **p_2**	=	3 **P_1 p_2**	Light
1 **p1**	3 **P_2**	=	3 **p_1 P_2**	Light
	1 **p_2**	=	1 **p_1 p_2**	White.

The inheritance of grain colour in wheat has been shown by Nilsson – Ehle to be similar to that in barley except that, in wheat, the genes are incompletely dominant over their alleles.

Thus the additive or cumulative action of the dominant alleles of two non-allelic genes causes the full expression of a phenotype, as distinctly different from the presence of the dominant allele of any one of the genes.

Summary of epistatic ratios

The epistatic ratios can be summarised as under:



Summary of epistatic ratios.

Lecture IX

Multiple alleles – characteristic features, study of blood group, coat colour in rabbits and self incompatibility in plants.

Multiple Alleles

So far, it has been observed that a given phenotypic trait of an individual depends on a single pair of genes, each of which occupies a specific position called the gene locus, on a homologous chromosome. Moreover, a particular gene has been found to occur in two alternative forms. For example, a gene (L) for length of *Drosophila* wings may occur in two alternative forms: a gene (L+) for normal development of wings and a gene (L") for vestigial wings. Because, most flies have normally developed wings, so, it can easily, be concluded that gene L+ is the original form of gene from which the other form of gene (L") might have originated by certain mutational event at sometime in past. The gene L+ for normal development of wings is called the normal or wild type allele of the gene L and usually symbolized as L+, while the mutated gene L" for vestigial wings is called L" reduced type or mutant allele of gene L. A fly with normal wings, thus has two wild type alleles (L+L+) and the vestigial wings fly has two mutant alleles (L"L"). Both of these allelic forms (L+ and L") of gene L occur at corresponding positions on genetically identical (homologous) chromosomes of same or different individual.

But, now there are ample evidences that a gene for a particular character, besides occurring in two alternative forms or alleles may occur in several alternative forms or alleles. All the variants or alleles of a given gene are supposed to be originated by mutation of a single wild type gene. Out of several allelic forms of a gene, a given locus may bear any one allele, so that, a diploid individual possesses any two alleles of the allelic series. When any of the three or more allelic forms of a gene occupy the same locus in a given pair of homologous chromosomes they are said to constitute a series of multiple alleles. In other words, all the mutant forms of a single wild type gene constitute a series of multiple alleles.

Blood Group

Multiple allelism also occur in man. The blood group inheritance in man can be better understood by learning following aspects:

Multiple Allelic inheritance of A, B, AB and O Blood Types

Bernstein (1925) proposed that inheritance of A, B, AB and O blood types of man is determined by a series of three allelomorphic genes. The gene controlling blood types has been labeled as I (after the name of immune traits) or L (after the name of discoverer, Landsteiner). The L gene exists in three different allelic forms : L^A, L^B and L^O. The first two alleles produce characteristic antigens on the surface of red blood cells or erythrocytes. Thus L^A alleles specifies A antigen, L^B allele B antigen and L^O allele specifies no antigen.

Reference: PSV: 163-170

The pedigree analysis has shown that alleles, L^A and L^B have dominance over allele L^O. Likewise, the pedigree analysis of A and B parents revealed that children have both A and B antigens and so it was concluded that the alleles L^A and L^B have co-dominant relationship between them. The dominance hierarchy of this allelic-series can symbolized as $L^A = L^B > L^O$.

Further, studies have shown that the antigen A is heterogeneous and may have four uncommon subgroups as A^1, A^2, A^3, etc. The B antigen thus, may occur in atleast three other allelic forms, viz., L^{A}_1, L^{A}_2 and L^{A}_3. Pedigree analysis have shown that L^{A}_1 allele is dominant over L^{A}_2 and L^{A}_3 alleles; the L^{A}_2 allele is dominant to L^{A}_3 allele and L^{A}_3 allele is recessive to L^{A}_2 and L^{A}_1 alleles. Now, the dominance hierarchy $L^A = L^B > L^O$ can be better represented as follows:

$$[(L^{A}_1 > L^{A}_2 > L^{A}_3) = L^B] > L^O$$

The series of multiple alleles of gene 'L' may produce 15 genotypes and 8 phenotypes, as have been tabulated in following table.

Phenotypes and genotypes of multiple alleles of gene

Phenotype	Genotype
A1	$L^{A}_1L^{A}_1$, $L^{A}_1L^{A}_2$, $L^{A}_1L^{A}_3$, $L^{A}_1 L^O$
A2	$L^{A}_2L^{A}_2$, $L^{A}_2L^{A}_3$, $L^{A}_2L^O$
A3	$L^{A}_3L^{A}_3$, $L^{A}_3 L^O$
A1B	$L^{A}_1L^B$
A2B	$L^{A}_2L^B$
A3B	$L^{A}_3L^B$
B	L^BL^B, L^BL^O
O	L^OL^O

Characters of Multiple Alleles

The most important and distinguishing features of multiple alleles are summarized below:

1. Multiple alleles of a series always occupy the same locus in the chromosome.
2. Because, all the alleles of multiple series occupy the same locus in chromosome, therefore, no crossing-over occurs within the alleles of a same multiple allele series.
3. Multiple alleles always influence the same character.
4. The wild type allele is nearly always dominant, while the other mutant alleles in the series may show dominance or there may be an intermediate phenotypic effect.
5. When any two of the mutant multiple alleles are crossed, the phenotype is mutant type and not the wild type.

Symbolism for Multiple Alleles

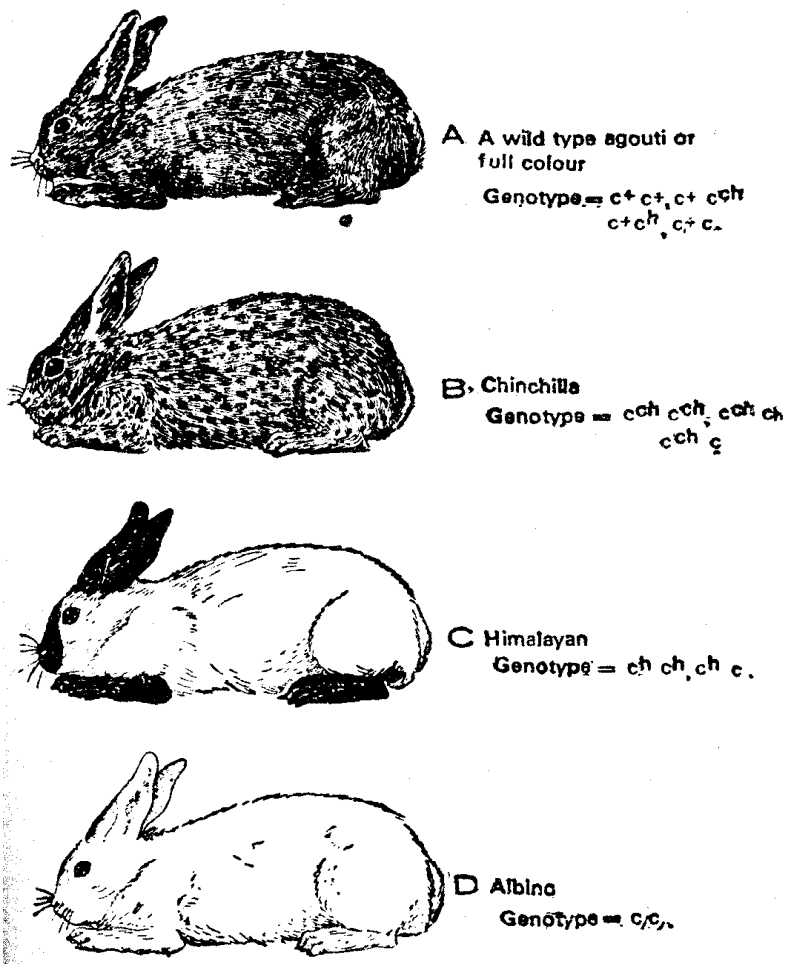
The dominance hierarchy is defined at the beginning of each problem involving multiple alleles. A capital letter is commonly used to designate the allele which is dominant to all other alleles in the series. The corresponding lower case letter designates the allele which is recessive to all others in the series. Other alleles which are intermediate in their degree of dominance between these two extremes, are usually assigned the lower case letter with some suitable super script.

Examples of Mulitple Allelism

Some of the characteristic cases of multiple alleles have been studied in rabbits, guinea pigs, mice, *Drosophila*, man and certain plants, such as *Nicotiana*.

Coat colour in Rabbits

The coat of rabbit may have different colours as described below.



Different coat colours in rabbits (after Burns, 1969).

i) Full colour: The coat of the ordinary (wild type) rabbit is referred to as "agouti" or full colour, in which individuals have banded hairs, the portion nearest the skin being gray, succeeded by a yellow band, and finally a black or brown tip. The allele for full colour may be represented by capital letter c^+.

ii) Chinchilla: In some individuals, the coat lacking the yellow pigment and due to the optical effect of black and gray hairs have the appearance of silvery-gray. The allele for chinchilla is represented as, c^{ch}.

iii) Himalyan (Russian): The Himalyan type coat is white except for black extremities (nose, ears, feet and tail). The condition in which black pigmentation is confined to the ears, muzzle, feet and tail, is called acromelanism (Serra, 1965). In Himalyan rabbits eyes remain pigmented. The allele for Himalyan coat is represented by c^h.

iv) Albino: The albino coat totally lacks in pigmentation and the eyes of a albino also remain pink due to lack of pigment in iris of eye. The allele for albino is represented by c.

Crosses of homozygous agouti (c^+c^+) and albino (cc) individuals produce a uniform agouti F_1 ; interbreeding of the F_1 produces and F_2 ratio of 3 agouti : 1 albino. Tow third of F_2 agouti are found to be heterozygous by testcrosses. Thus, it is a case of monohybrid inheritance, with agouti completely dominant to albino. Likewise, crosses between chinchilla and agouti produce all agouti individuals in the F_1 and a 3 agouti : 1 chinchilla ratio in the F_2. Such complete dominance of agouti also occurs on Himalayan. Further crosses, real that c^{ch} allele for chinchilla, though is recessive to c+ allele for agouti coat or skin, is incompletely dominant over Himalayan (c^h) and albino (c) alleles. Likewise, c^h allele for Himalayan coat is recessive to c^+ (agouti) and cch (Chinchilla) but dominates over albino. The results of all these crosses exhibit that c^+ (agouti), c^{ch} (chinchilla), c^h (Himalayan) and c (albino) are allelic to each other and the alleles of this multiple allelic series have following dominance hierarchy : $c^+ > c^{ch} > c^h > c$

P_1 : Agouti (c^+c^+) x Albino (cc)

↓

Agouti

F_1: c^+c

F_2: $1c^+c^+ : 2c^+c : 1cc$

3 Agouti : 1 Albino

A monohybrid cross between agouti and albino rabbits.

P_1 : Agouti (c^+c^+) x Chinchilla ($c^{ch}c^{ch}$)

↓

Agouti

F_1: c^+c^{ch}

F_2: $1c^+c^+ : 2c^+c^{ch} : 1c^{ch}c^{ch}$

3 Agouti : 1 Chinchilla

A monohybrid cross between agouti and chinchilla rabbits.

P_1 : Agouti x Himalayan
c^+c^+ c^hc^h

Agouti
F_1: c^+c^h
F_2: $1c^+c^+ : 2c^+c^h : 1c^hc^h$

3 Agouti : 1 Himalayan

A monohybrid cross between agouti and Himalayan (or Russian) rabbits.

P_1 : Chinchilla x Himalayan
$c^{ch}c^{ch}$ c^hc^h

F_1: light gray
$C^{ch}c^h$
F_2: $1c^{ch}c^{ch} : 2c^{ch}c^h : 1c^hc^h$
1 Chinchilla : 2 Light gray : 1 Himalayan

A monohybrid cross between Chinchilla and himalayan rabbits. Showing incomplete dominance of chinchilla (c^{ch}) on Himalayan (c^h)

P_1 : Chinchilla x Albino
$c^{ch}c^{ch}$ c c

F_1: light gray
$C^{ch}c$
F_2: $1c^{ch}c^{ch} : 2c^{ch}c : 1cc$
1 Chinchilla : 2 Light gray : 1 Albino

A monohybrid cross between Chinchilla and albino rabbits. Showing incomplete dominance of chinchilla (c^{ch}) on albino (c).

P_1 : Himalayan x Albino
c^hc^h c c

Himalayan
F_1: c^hc
F_2: $1c^hc^h : 2c^hc : 1cc$

3 Himalayan : 1 Albino

A monohybrid cross between Himalayan and albino rabbits.

The possible phenotypes and their associated genotypes of this multiple allelic series can be summarized.

The phenotypes and genotypes of multiple allelic series for coat colour in rabbit.

Phenotypes	Genotypes
Full colour (Agouti)	c^+c^+, c^+c^{ch}, c^+c^h, c^+c
Chinchilla	$c^{ch}c^{ch}$
Light gray	$c^{ch}c^h$, $c^{ch}c$
Himalayan	c^hc^h, c^hc
Albino	cc

Self sterility in Nicotiana

multiple alleles have been associated with self-sterility or self-incompatibility in several groups of plants. S elf-sterility is the phenomenon in which the pollen grains from a plant fail to bring about fertilization in the ovules of the same plant. As early as 1764 Kolreuter described self-sterility in tobacco, Nicotiana. In 1925 E. M. East discovered self-sterility in Nicotiana is governed by alleles of multiple allelic series of gene S.

Different alleles of this multiple allelic series were designated as S_1, S_2, S_3, S_4, S_5, etc. None of the cross-fertilizing tobacco plants were homozygous, (i.e., S_1S_1 or S_2S_2) but all plants were heterozygous (e.e., S_1S_2, S_3S_4, S_5S_6, etc.). When crosses were attempted between different S_1S_2 plants, it was observed that pollen tubes did not develop normally, but pollen from S_1S_2 were effective on stigmas of plants with other alleles, for example, S_3S_4.

When crosses were made between seed parents with S_1S_2 and pollen parents with S_2S_3, two kinds of pollen tubes were distinguished. Pollen grains carrying S_2 were not effective, but the pollen grains carrying S_3 were capable of fertilization. Thus, from the cross S_1S_2 x S_2S_3, two kinds of progeny, S_1S_3 and S_2S_3, were produced. From a cross S_1S_2 X S_3S_4, all the pollens were effective and four kinds of progeny resulted : S_1S_3, S_1S_4, S_2S_3, and S_2S_4. Some combinations are summarized.

Genotypes of progenies obtained due to crosses between various self-sterility types of *Nicotiana.*

Seed parents	Pollen parent			
	S_1S_2	S_2S_3	S_3S_4	S_4S_5
S_1S_2		S_3S_2 S_3S_1	S_3S_1 S_3S_2 S_4S_1 S_4S_2	S_4S_1 S_4S_2 S_5S_1 S_5S_2
S_2S_3	S_1S_2 S_1S_3		S_4S_2 S_4S_3	S_4S_2 S_4S_3 S_5S_2 S_5S_3
S_3S_4	S_1S_3 S_1S_4 S_2S_3 S_2S_4	S_2S_3 S_2S		S_5S_2 S_5S_4
S_4S_5	S_1S_4 S_1S_5 S_2S_4 S_2S_5	S_2S_4 S_2S_5 S_3S_4 S_3S_5	S_3S_4 S_3S_5	

Lecture X

Multiple factor hypothesis – Nilsson-Ehle – Wheat kernel colour experiment – Polygenes.

Polygenic inheritance

The inheritance of many of the differentiating characters of plants and animals, for example, yellow and green colour of cotyledons in peas, brown and white colour of grains in Sorghum, can be studied easily because the individuals can be separated into sharply distinct classes by mere observation without resorting to any scale of measurements. Colour of cotyledons in peas, colour of grains in Sorghum, etc., are examples of qualitative characters that show discontinuous variation and are governed by one or two major genes or oligogenes. An understanding of the inheritance of characters like length of earth in corn, yield of grain in rice, yield of milk in dairy cattle is not so easy because the individuals show little differences and can be separated into classes only after making measurements. These characters are examples of quantitative characters that show more or less continuous variation and are governed by a large number of genes called multiple genes or multiple factors or polymeric genes or polygenes.

Nilsson-Ehle's studies on kernel colour in wheat

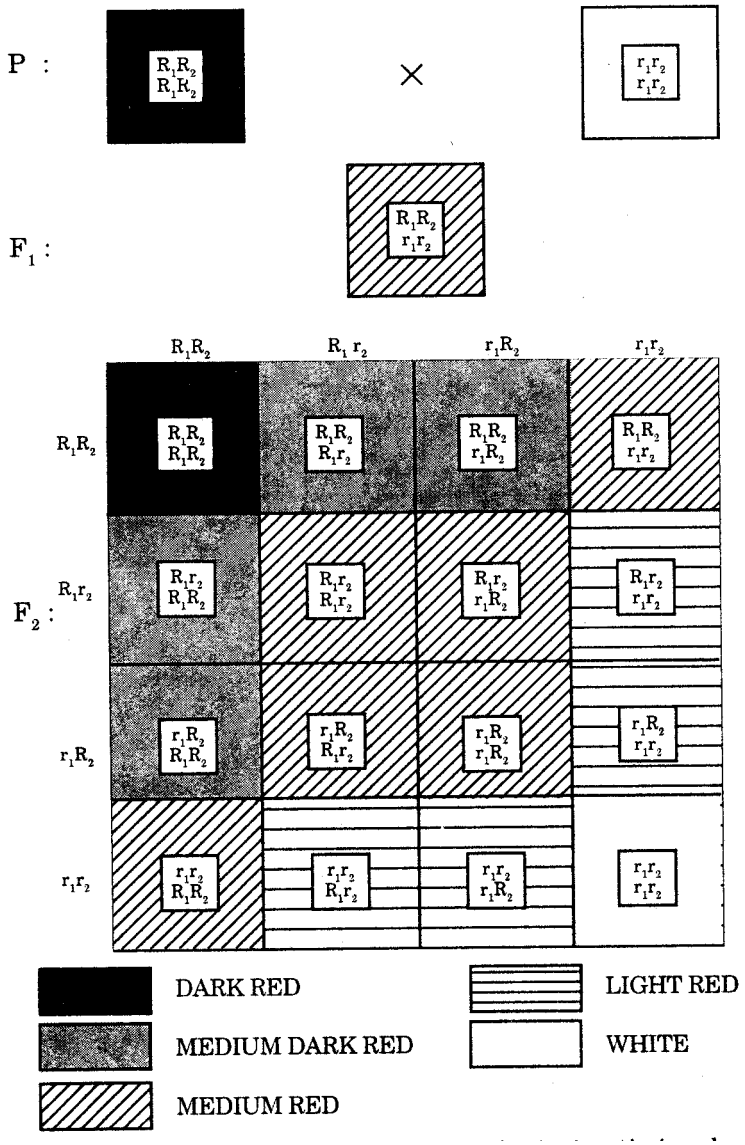
The Swedish geneticist Nilsson-Ehle (1908) effect crosses between different true breeding strains of wheat with red kernels and those with white kernels. In some crosses of red with white, a ratio of 3 red : 1 white was found among the F_2, indicating a single gene difference. Careful examination however revealed that the red colour of the F_1 was not as intense as the red colour of the parent and that in the F_2, some red grains were as dark as those of the parent and others only as dark as those of the F_1.

In some other crosses, a ratio of 15 red : 1 white was found in the F_2, indicating that there are two pairs of genes for red colour and that either or both of these can produce red kernels. Careful examination revealed that all the red kernels were not of the same intensity of colour. It was possible to separate the F_2 into the following classes:

Dark red	1
Medium dark red	4
Medium red	6
Light red	4
White	1

It is evident that red colour is due to two pairs of alleles. Each gene is capable of producing red colour. Each is incompletely dominant over white and is cumulative in its effect. The intensity of the red colour depends upon the number of colour producing genes present. Dark red is due to the presence of four contributing genes for red, medium dark red to three contributing genes, medium red to two contributing genes and light red to one contributing gene.

Reference: PSV: 111-114



Checkerboard showing 1 : 4 : 6 : 4 ; 1 ration in wheat

The genotypes of the F_2 together with their phenotypes are given below:

The F_2 ratio in wheat

Genotype	Genotypic ratio	Phenotype
$\mathbf{R_1R_1R_2R_2}$	1	Dark red
$\mathbf{R_1R_1R_2r_2}$	2	Medium dark red
$\mathbf{R_1r_1R_2R_2}$	2	Medium dark red
$\mathbf{R_1r_1r_2R_2}$	4	Medium red
$\mathbf{R_1R_1r_2r_2}$	1	Medium red
$\mathbf{r_1r_1R_2R_2}$	1	Medium red
$\mathbf{R_1r_1r_2r_2}$	2	Light red
$\mathbf{r_1r_1R_2R_2}$	2	Light red
$\mathbf{r_1r_1r_2r_2}$	1	White

In still other crosses, Nilsson-Ehle found a ratio of 63 red : 1 white in the F_2, a segregation which suggested that three independent pairs of alleles were involved. If the red parent is represented by $\mathbf{R_1R_1R_2R_2R_3R_3}$ and the white by $\mathbf{r_1r_1r_2r_2r_3r_3}$, the F_1, which was essentially uniform but intermediate between the parents in colour, can be represented by $\mathbf{R_1r_1R_2r_2R_3r_3}$. In the F_3 there was a marked increase in the range of colour types. About 1 in 64 of the F_2 was with very deep red kernels and has 6 contribution genes for red ; 6 with deep red kernels have 5 contributing genes; 15 with dark red kernels have 4 contributing genes; 20 with medium dark red kernels have 3 contributing genes; 15 with medium red kernels have 2 contributing genes ; 6 with light red kernels have 1 contributing gene and 1 in 64 was with white kernels and has no contributing genes for the red colour. It was difficult to distinguish these differences in colour as there was a more or less continuous variation among the F_2.

From these studies, Nilsson-Ehle proposed the multiple factor hypothesis for the inheritance of quantitative characters. This assumes that there is a series of independent genes for a given quantitative trait. Dominance is usually incomplete but these genes are cumulative or additive in their effect. Each gene adds something to the strength of expression of the character whereas but intermediate between the two parents. The F_2 shows considerable variability but is intermediate between the two parents, the F_2 mean value being approximately equal to the parental mean and also, the F_1 mean.

Lecture XI

Quantitative Vs Qualitative characters and modifiers.

Quantitative inheritance

Let us suppose that one true-breeding tall plant with a height of 200 cm. has the genotype $T_1T_1T_2T_2$ and a true-breeding short plant with a height of 100 cm. has the genotype $t_1t_1t_2t_2$. Let us also suppose that the environment is so uniform that it is not responsible for variation in height. Let us further suppose that except for the difference in the two loci (i.e., T_1/t_1 and T_2/t_2), the two plants have the same genotype which is responsible for a plant height of 100 cm. The difference in height of 100 cm. between the two plants is due to the four duplicate, cumulative, incompletely dominant genes designated by capital letters, T_1, T_1, T_2 and T_2 (called contributing or active genes), each gene adding 25 cm. to the height of the plant. The alleles designated by small letters t_1, t_1, t_2 and t_2 (called neutral or inert alleles), do not in any manner influence the height of the plant.

The F_1 hybrid would be $T_1\ t_1\ T_2\ t_2$. As the two contributing genes T_1 and T_2 add 25 cm. each to the residual heredity of 100 cm, the F_1 would be 150 cm. high, exactly intermediate between the parents. The F_2 would segregate for plant height and hence would exhibit considerable variability in height.

The F_2 from a cross between plants differing in height

Genotype	Genotypic ratio	Phenotype
$\mathbf{T_1\,T_1\,T_2\,T_2}$	1	200 cm.
$\mathbf{T_1\,T_1\,T_2\,t_2}$	2	175 cm.
$\mathbf{T_1\,t_1\,T_2\,T_2}$	2	175cm.
$\mathbf{T_1\,t_1\,T_2\,t_2}$	4	150 cm.
$\mathbf{T_1\,T_1\,t_2\ t_2}$	1	150 cm.
$\mathbf{t_1\ t_1\,T_2\,T_2}$	1	150 cm.
$\mathbf{T_1\,t_1\,t_2\,t_2}$	2	125 cm.
$\mathbf{t_1\,t_1\,T_2\,t_2}$	2	125 cm.
$\mathbf{t_1\,t_1\,t_2\,t_2}$	1	100 cm

The frequency of each in the F_2 would be as follows:

Frequency distribution in the F_2

Plant height	Frequency	No. of active alleles
200 cm.	1	4
175 cm.	4	3
150 cm.	6	2
125 cm.	4	1
100 cm.	1	0

The mean height of the F_2 plants would be 150 cm., which is equal to the parental mean and also, the F_1 mean.

If instead of four contributing genes, a very large number of genes, each with a very small individual influence, are assumed to be responsible for plant height, the expected hereditary difference between two successive classes in the F_2 is likely to be smaller than even the difference normally due to environment. Where these class differences are very small, the variation in the F_2 population would appear to be continuous and would be typical of quantitative inheritance.

The features of inheritance of quantitative characters are the following:

The individuals of each homozygous parental line have the same genotype and therefore the two lines to which the parents belong would show very little variability within themselves. The phenotypic differences between individuals within a parental line are only due to environment.

All the individuals of the F_1 have the same genotype and, therefore, the F_1 as a whole show very little variability. The F_1 would, however, be intermediate between the parents, the mean of the F_1 being equal to the mean of the two parental values. The phenotypic differences between individuals in the F_1 population are only due to environment.

The F_2 would exhibit considerable variability. Some of the F_2 values would overlap with the values of one parent, some other F_2 values would overlap with the values of the other parent and a large number of the F_2 values would be intermediate between the values of the two parents. This variation in the F_2 is more or less continuous and is largely due to differences in genotype between individuals of the F_2. The F_2 mean would however be equal to the F_1 mean and also, the parental mean.

Quantitative characters are controlled by the joint action of a vary large number of multiple genes which cannot be distinguished from one another because their individuals effects on the phenotype are insignificant in comparison with the fluctuations due o the environment. Multiple genes are usually incompletely dominant duplicate gene with cumulative effects.

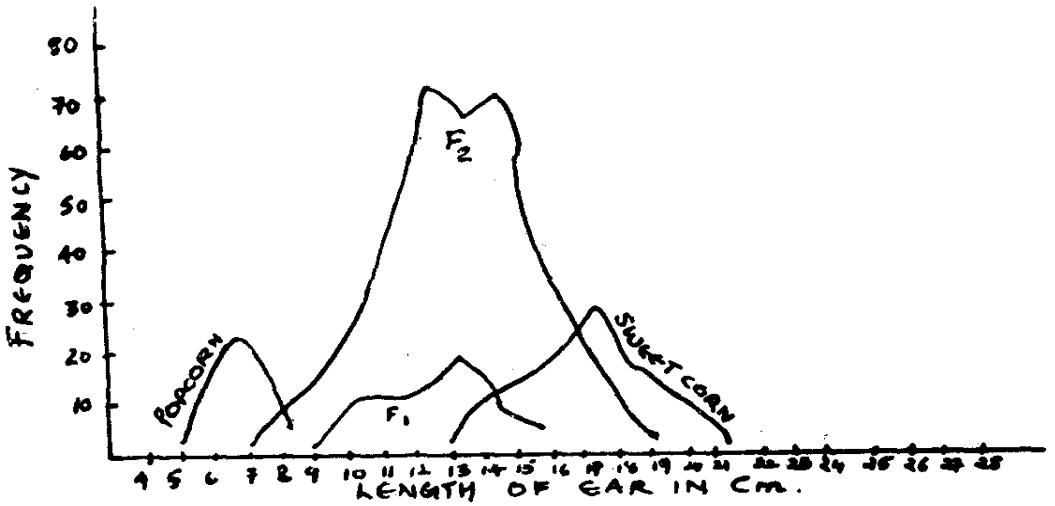
Studies on ear length in corn

Emerson and East (1913) crossed a long eared sweet corn plant from a line having a mean ear length of 16.80 cm. with a shot-eared popcorn plant from a line having a mean ear length of 6.63 cm. within each parental line there was some variability in ear length. As each parental lien was homozygous for genes affecting ear length, this variability could only be due to environment.

The F_1 mean was 12.12 cm. which was intermediate between the two parental lines. The F_1 was however, uniform and the small variation around the mean exhibited by the F1 individuals could be attributed only to the environment a all the F_1 plants were typically alike.

The mean length of ear of the F_2 was 12.89 cm. this is almost equal to the F_1 mean of 12.12 cm. and was approximately intermediate between the mean of the long-eared parent (16.80 cm.) and the mean of the short-eared parent (6.63 cm.)

The facts that the F_1 was uniform but intermediate between the parents and that the F_2 continuous variation but was equal in its mean length of ear to the F_1 mean and the parental mean suggest that length of ear is a quantitative character governed by a number of incompletely dominant genes which have effects similar to one another and which supplement each other.



Frequency curves for length of ear in corn

Modifying genes

A quantitative character is determined by a very large number of genes whose individual effects, though small, are almost equal to one another. There are, however, instances where a certain character is determined fundamentally by one gene but is modified slightly by other genes. The main or major gene gives expression to a character and the minor or modifying genes slightly alters the degree of expression of this character. A modifying gene is one that alters the expression of a major gene but has no effect on the allele of the major gene. The modifiers have very similar but individually small effects and are usually present is such large numbers that they cannot be individually identified.

In the Guernsey breed of dairy cattle, the 'solid' colour (fawn i.e., light yellowish brown) of the coat is due to dominant gene S and the 'spotted' coat (white spotting) is due to its recessive alleles s. A number of these modifying genes influences the intensity of spotting. If a large number of these modifying genes is present in animals with ss, the animals are highly 'spotted'. If only a small number of these modifying genes is present in animals with ss, they are medium 'spotted'. If the modifying genes are absent, animals with ss have only few 'spots'. These modifying genes have no effect in the presence of the gene for 'solid' colour and animals with SS or Ss have solid-colour coats irrespective of the number of modifying genes present.

In *Gossypium barbadense* the presence of petal spot is due to a gene S and the absence of petal spot is due to its recessive allele s. A number of modifying genes increases the intensity of colour in the presence of the gene S.

Quantitative characters are governed by several genes each one with small and cumulative effect. Quantitative characters show a continuous variation and it is not possible to classify them into distinct classes. These characters are considerable affected by the environment.

The important features of polygenic traits are briefly discussed below in contrast with oligogenic or qualitative trait.

1. Variation

The variation is continuous from one extreme to the other in quantitative traits, whereas the variation is discontinuous in case of qualitative characters. Because of continuous variation demarcation into different classes is not possible in case of quantitative traits.

2. Number of genes

All the quantitative characters are governed by several genes with small individual effect. These genes are called as polygenes. Qualitative characters are controlled by one or few genes each having large and easily detectable effect. These genes are known as oligogenes.

3. Effect of single gene

Each gene in quantitative trait has small or minor individual effect and identification or detection of the effect of individual gene is very difficult. Hence quantitative characters are also called as minor gene characters. On the other hand in qualitative characters each gene has major and easily detectable effect. Such traits are called major gene characters or traits.

4. Classification

The classification of quantitative characters into different clearcut groups is not possible because of continuous variation from one extreme to the other. In qualitative characters such grouping is possible because of discrete or discontinuous variation.

5. Gene action

Generally the quantitative traits are governed by additive gene action, but now cases are known where quantitative characters are governed by dominance and epistatic gene action. In case of qualitative traits the gene action is primarily of non-additive type (dominance and epistasis).

6. Environmental effect

The quantitative characters are considerable influenced by the environment. In other words, they are more prone to genotype x environmental interactions. The main effect of the environment is to mask the small differences among different genotypes resulting in continuous variation in the character. When the contribution of environment is 50% the distribution becomes roughly similar to normal curve and with 75% contribution it tends to reach normal distributions. The qualitative traits are not much influenced by the environment. In case of quantitative characters generally the environmental variation ranges from 10 to 50% and even more for some traits, like yield. The high environmental variation results in overlapping of various classes resulting in continuous variation.

7. Metric measurement

It is possible in case of quantitative traits like measurement of size, weight, duration, etc., whereas in case of qualitative traits only the counting of plants with regard to various kinds based on colour and shape is possible. Thus metric measurement is not possible in case of qualitative characters.

8. Transgression

Transgression refers to the phenomenon through which we get variation in F2 or later generation outside the range of both the parents. Transgressive segregants are only possible from the crosses between two plants with mean values for a quantitative trait. Such segregants are not possible in case of qualitative traits.

9. Stability

The quantitative characters are very much sensitive to environmental effects and thus are less stable, whereas the qualitative traits are highly stable to the environmental effects.

10. Field of Genetics

The inheritance of quantitative traits is studied with the help of quantitative genetics or biometrical genetics, whereas the qualitative traits can be studied by Mendelian genetics and population genetics.

11. Heritability

The heritability estimates are generally low for quantitative characters because of high amount of environmental variation. In case of qualitative characters the estimates of heritability are high because there is little difference between the genotype and phenotype of a character.

12. Statistical parameters

Different statistical techniques are used for the study of quantitative and qualitative traits. The inheritance of quantitative characters is studied with the help of mean, variances and covariances, and useful biometrical information is obtained through these estimates from different populations using specific mating schemes and experimental designs. In case of qualitative traits the inheritance is studied with the help of segregation ratios. Such rations are tested with X^2 test to study the significance of difference between the observed and expected values. Thus polygenic and oligogenic traits differ in several aspects (Table 1)

Differences between polygenic and oligogenic traits

	Poly genic traits	Oligogenic traits
1.	Governed by several genes	Governed by few genes
2.	Effects of each gene is not detectable	Effect of each gene is detectable
3.	Usually governed by additive gees	Governed by non-additive genes
4.	Variation is continuous	Variation is discontinuous
5.	Separation into different classes is not possible	Separation into different classes is possible
6.	Highly influenced by environmental	Little influence by environmental factors
7.	Statistical analysis is based on mean, variances and covariance	Statistical analysis is based on frequencies or ratios.

In plant breeding both types of characters showing qualitative and quantitative inheritance have equal economic importance.

Lecture XII

Linkage – coupling and repulsion – experiment of Bateson and Punnett – chromosomal theory of linkage of Morgan – complete and incomplete linkage.

Linkage group

Members of different pairs of alleles undergo independent assortment only because the maternal and paternal members of different pairs of homologous chromosomes are distributed independently to the gametes during meiosis. The number of pairs of alleles, however, exceeds the number of pairs of chromosomes in an organism. Each pair of chromosomes may therefore carry many pairs of alleles. For example, in maize, which has 10 pairs of chromosomes, more than 500 pairs of alleles, have been identified so far. Several of these genes must therefore be located on the same chromosome and will not be assorted independently. Each of these will individually yield F2 ratios expected according to Mendel's Law of Segregation but when studied in groups of two or more, will not yield F2 ratios according to Mendel's Law of Segregation but when studied in groups of two or more, will not yield F2 ratios according to Mendels Law of Independent Assortment. These genes will show linkage, i.e., they will be inherited as a group. A linkage group is a group of genes situated on the same chromosome.

The number of linkage groups will be equal to the haploid number of chromosomes which the species possesses. Thus, maize which has 10 pairs of chromosomes has 10 linkage groups.

Symbols for linked genes

While representing linked genes, the two homologous chromosomes are indicated by two horizontal lines with the genes on one chromosome above the line and genes on the other chromosome below the lines, e.g., $\underline{\underline{\frac{CS}{Cs}}}$.

Some geneticists use a single horizontal line instead of two, e.g., $\frac{CS}{cs}$. Still others use a slanting line in preference to the horizontal line, e.g., CS/cs.

All the genes on a chromosome are said to be linked to one another and belong to the same linkage group. The phenomenon of inheritance of linked gene in same linkage group is called linkage.

Difference in linkage and independent assortment.

Mendel's law of independent assortment is applied only to those genes which are located on separate chromosomes, because, the linked genes of a linkage group (chromosome) inherit together. A dihybrid contains either linked genes or independently assorted genes, can be determined by test crossing it with a double recessive parent. The independently assorted genes give the test cross ratio of 1 : 1 : 1 : 1 and linked genes give the test cross ratio of 1 : 1 as have been illustrated by following examples :

Example I. If genes occur on different chromosomes, they assort indecently and give a test cross ratio of 1 : 1 : 1 : 1 as follows:

P_1 **AABB** x **aabb**

P_1 Gametes : **(AB)** | **(ab)**

↓

Aa Bb

F_1 :

Test cross : **Aa Bb** x **aa bb**

↓ ↓

Gametes : (AB) (Ab) (aB) (ab) (ab)

F_2 : ¼ Aa Bb : ¼ Aa bb : ¼ aa Bb : ¼ aa bb

Or 1 : 1 : 1 : 1 (Test cross ratio).

Example II. The linked genes do not assort independently, but tend to stay together in the same combinations as they were in the parents. In the following figure, the genes on the left of the slash line (/) are on one chromosome and those on the right are on the homologous chromosome. The linked genes give the test cross ratio of 1 : 1 as follows :

Parents : AB/ab x ab/ab

Gametes : (AB) (ab) (ab) (ab)

F_1 : ½ AB/ab : ½ ab,ab or 1 : 1 (test cross ratio)

Views of classical geneticists on linkage

Mendel could not notice the phenomenon of linkage because fortunately the seven pairs of factors or alleles studied by him in pea were located on seven different pairs of chromosomes. It was noticed and discovered by some other post-Mendelian geneticists who during their genetic investigations came across to linked genes. The evolution of linkage concept took place by the views of following classical geneticists:

Coupling and Repulsion Hypothesis of Bateson and Punnett

Bateson and **Punnett** (1905-1908) formulated the 'hypothesis of coupling and repulsion' to explain the unexpected F_2 results of a dihybrid cross between a homozygous sweet pea (*Lathyrus odoratus*) having a dominant alleles for blue or purple flowers (RR) and long pollen grains (Ro Ro) with another homozygous double recessive plant (rr, roro) with red flowers and round pollen grains. When they test crossed a heterozygous blue or purple long (Rr, Roro) plant with recessive parent (rr, roro), besides getting the 1 : 1 : 1 : 1 test cross ratio, they received phenotypic ratio of 7 : 1 : 1 : 7 as has been illustrated here:

Parent :	Blue or purple long (RR Ro Ro)	x	Red round (rr ro ro)
F_1:	All blue or purple long (Rr Ro ro)		
Test cross :	F_1 blue or purple long (Rr Ro ro)	x	Red round (rr ro ro)

Test cross progeny :	Blue or purple long	=	192
	Red round	=	182
	Blue or purple round	=	23
	Red long	=	30
			427

Test cross ratio: 7 Blue or purple long : 1 Blue or purple round : 1 Red long : 7 Red round or 7 : 1 : 1 : 7.

The 7 : 1 : 1 : 7 test cross ratio clearly indicated that there was a tendency in the dominant alleles (RRo) to pass together to the same gamete. similar was the case with recessive alleles (rro). This tendency of dominant or recessive alleles (rro). This tendency of dominant or recessive alleles to inherit together was explained as 'gametic coupling' by Bateson and Punnett.

Further, when they crossed blue or purple round (RR roro) with red long (rr RoRo), the F_1 hybrids were found to be heterozygous blue or purple long (Rr Roro). The F_1 hybrid when test crossed with recessive (rr roro) parent, the test cross ratio was 1 blue or purple long : 7 red long : 7 blue or purple round : 1 red round, as has been illustrated in following figure :

Parent:	Blue or purple round (RR roro)	x	Red long (rr RoRo)
F1:	All blue or purple long (Rr Ro ro)		
Test cross:	F_1 blue or purple long (Rr Ro ro)	x	P_1 Red round (rr ro ro)

Test cross progeny: 1/16 Blue or purple long (Rr Ro ro) : 7/16 Blue or purple round (Rr ro ro): 7/16 Red long (rr Ro ro): 1/16 Red round (rr ro ro) or 1 : 7 : 7 : 1.

Hence, the two dominant pairs of alleles repelled each other. The tendency of both dominant or both recessive alleles to repel each other, so that the gametes of genotypes of Rro and rRo are formed more frequently, was termed repulsion.

Bateson and Punnett could not explain the exact reasons of coupling and repulsion, and it was **T.H. Morgan** who while performing experiments with *Drosophila*, in 1910, found that coupling or repulsion was not complete. He further suggested that the two genes are found in coupling phase or in repulsion phase, because they are present on

the same chromosome (coupling) or on two different homologues chromosomes (repulsion). Such genes are then called linked genes and the phenomenon of inheritance of linked genes is called linkage by Morgan.

Morgan's views on Linkage

Morgan stated that the pairs of genes of homozygous parent tend to enter in same gametes and to remain together, whereas same genes from heterozygous parents tend to enter in different gametes and remain apart form each other. He further stated that the tendency of linked genes remaining together in original combination is due to their location in same chromosome. The degree or strength of linkage depends upon the distance between the linked genes in the chromosome. Morgan's concept about the linkage developed the theory of linear arrangement of genes in the chromosomes which helped the cytogeneticists in the construction of genetic or linkage maps of chromosomes.

Chromosome theory of Linkage

The chromosome theory of linkage of Morgan and Castle states that:

i) The genes which show linkage, are situated in the same pair of chromosomes.
ii) The linked genes remain arranged in a linear fashion on the chromosome. Each linked gene has a definite and constant order in its arrangement.
iii) The distance between the linked genes determines the degree of strength of linkage. The closely located genes show strong linkage then the widely located genes which show weak linkage.
iv) The linked genes remain in their original combination during the course of inheritance.

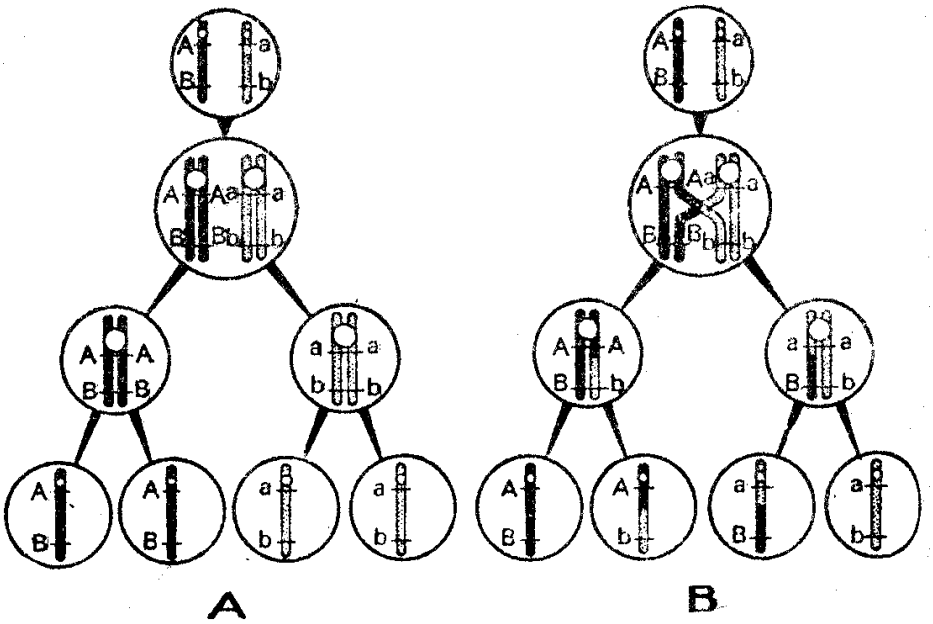
Kinds of Linkage

The phenomenon of linkage is of following two kinds:

1. Complete Linkage

When the linked genes are so closely located in chromosomes that they inherit in same linkage groups for two or more generations in a continuous and regular fashion, then, they are called completely linkage genes and the phenomenon of inheritance of completely linked genes called **complete linkage**.

Example: According to Bridge all the genes of male *Drosophila* remain completely linked. Further, in a mutant strain of *Drosophila,* the genes for bent wings (b^+) and shaven bristles (svn) of the fourth chromosome exhibit complete linkage.



A- Diagram of the segregation of two pairs of allelomorphic genes localized on the same pair of chromosomes without crossing over. The result is two types of gametes, AB and ab. A case of complete linkage. **B-** Diagram of the segregation of two pairs of allomorphic genes on the same chromosome between which crossing over takes place during meiosis, four types of gametes result : AB, Ab, aB and ab. A case of incomplete linkage (after De Robertis *et al.,* 1970).

2. Incomplete Linkage

The linked genes do not always stay together because homologues non-sister chromatics may exchange segments of varying length (which bearing many linked genes) with one another during meiotic prophase, by the process of crossing over. The linked genes which are widely located in chromosomes and have chances of separation by crossing over are called incompletely linked genes and the phenomenon of their inheritance is called incomplete linkage.

Example: Incomplete linkage has been observed in pea, Zea mays (maize), tomato, female (*Drosophila,* Mice, poultry, and man. Here, the examples of linkage have been considered only for Drosophila and Zea mays (maize).

1.Incomplete Linkge in Drosophila

The wild type Drosophila has gray body and long wings (b+v+/ b+v+), where alleles for gray b+ and long v+ dominant over the mutant alleles for black b and vestigial v. when, a gray long fly (bv/bv), the F1 heterozygote is found with gray long phenotype and b+v+/bv genotype. The F1 heterozygote (b+v+/bv) when test crossed with double recessive parent (bv/bv), instead of occurring of two class of phenotypes in the ratio of 1 : 1, occur four classes of phenotypes as shown.

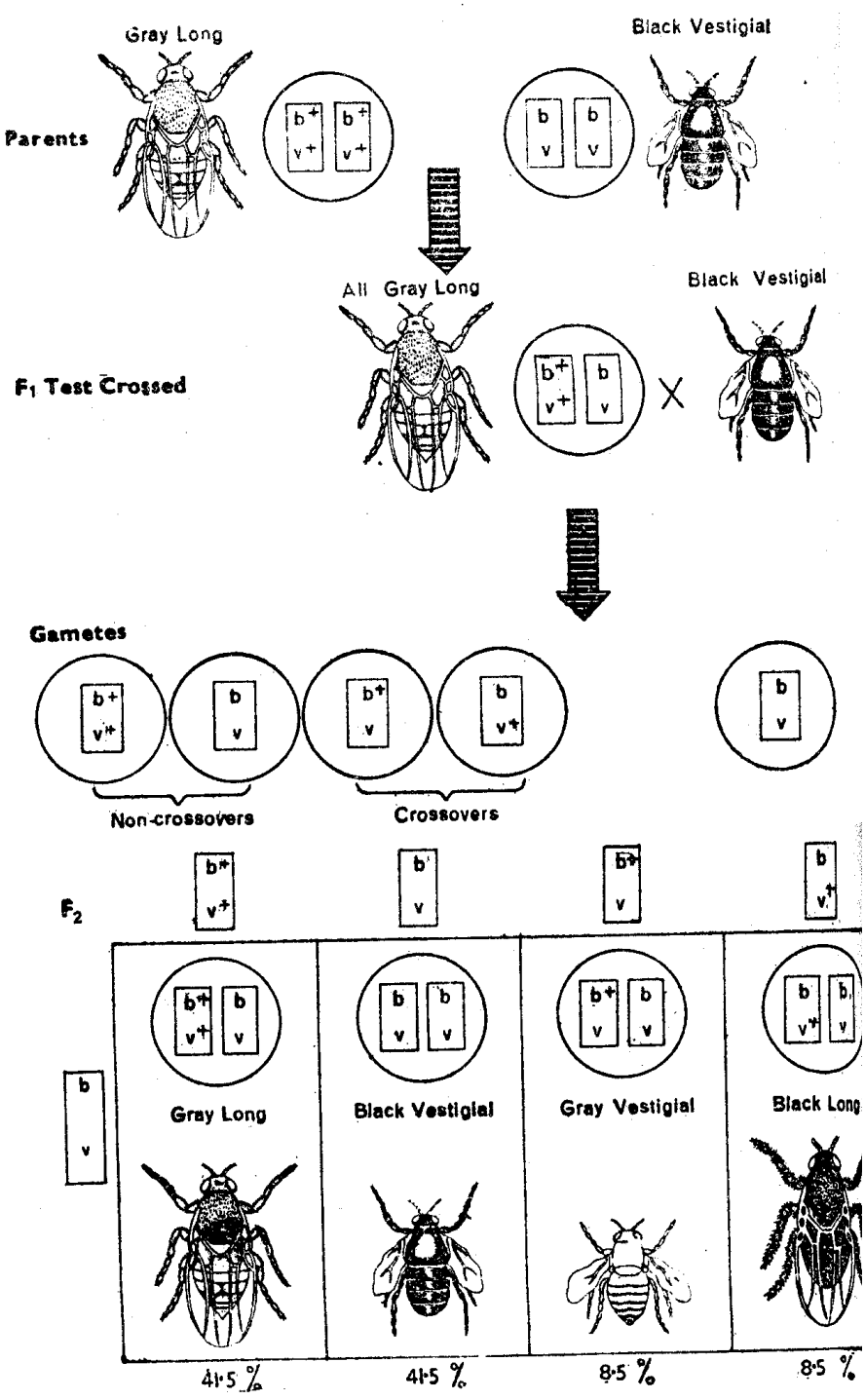


Diagram of a cross involving linkage and crossing over. The genes for vestigial versus normal (long) and black versus gray body in Drosophila are linked, they are located in the same chromosome (after Villee et al., 1973).

The test cross results are clearly showing that parental combinations (gray long and black vestigial) are those expected from complete linkage and appeared in 83% cases. The other two (gray vestigial and black, long) are new combinations and appeared in 17% cases. Thus, in 17% cases crossing over has occurred.

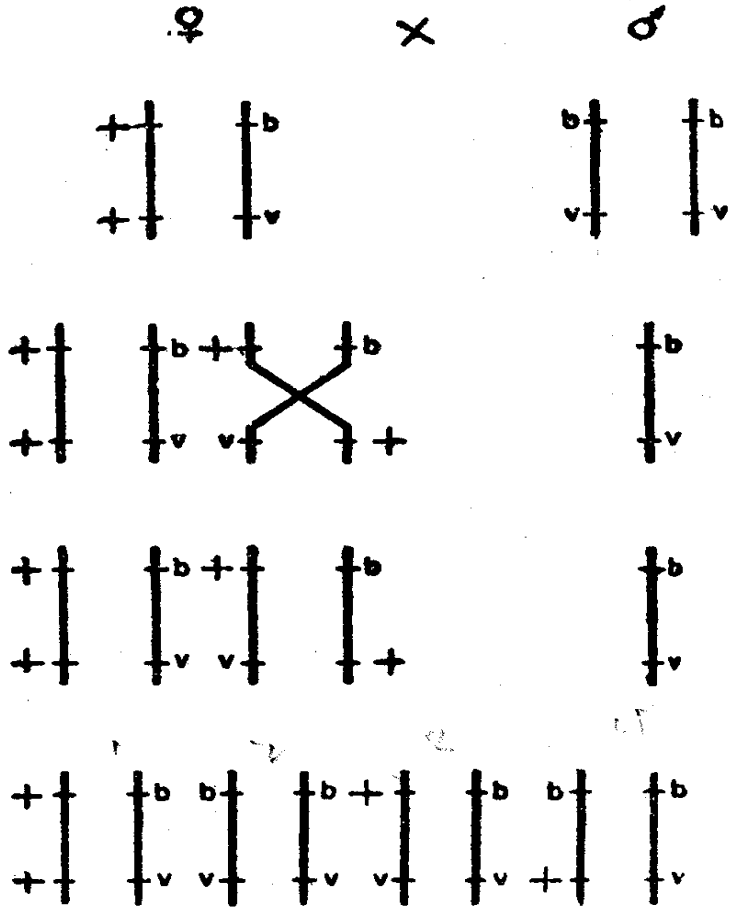


Diagram showing incomplete linkage in linked genes for body colour and wing shape in *Drosophila*. b=black body, v=vestigial wings, +=gray body and +=long wings (after De Robertis *et. al.*, 1970)

2. Incomplete Linkage in Maize

In *Zea mays* (Maize) a case of incomplete linkage between the alleles for colour and shape of the seed has been observed by Hutchison. When a maize plant with seeds having colour and full endosperm (CS/CS) is crossed with another plants having recessive alleles for colourless, shrunken seeds (cs/cs), the F_1 heterozygotes are found with the phenotype of coloured full and genotype of CS/cs. When F_1 hybrid is test crossed with double recessive parent (cs/cs) four classes of descendants are obtained instead of two as showing following figure:

Parents:	Coloured full CS/CS		x		Colourless shrunken cs/cs
F_1 :			Coloured full (CS/cs)		
Test cross:	F_1 coloured full CS/cs		x		Colourless shrunken cs/cs
Test cross results:	Coloured, full CS/CS 48%	Coloured, shrunken Cs/cs 2%		Colourless, full cS/cs 2%	Colourless shrunken cs/cs 48%

The test cross results are clearly showing that parental combination of alleles (eg., CS/CS and cs/cs) are those expected from complete linkage and appear in 96% cases, the other two are new combinations (eg, Cs/cs and cS/cs) and appear in 4% cases. Thus, in 4% cases crossing over have occurred between linkage genes.

Linkage groups

All the linked genes of a chromosome form a linkage group. Because, all the genes of a chromosome have their identical genes (allelomorphs) on the homologous chromosome, is considered as one. The number of linkage groups of a species, thus corresponds with haploid chromosome number of that species.

Example: 1. Drosophila has 4 pairs of chromosomes and 4 linkage groups.
2. Man has 23 pairs of chromosomes and 23 linkage groups.

Significance of Linkage

The phenomenon of linkage has one of the great significance for the living organism that it reduces the possibility of variability in gametes unless crossing over occurs.

Lecture XIII

Crossing over – significance of crossing over – cytological proof for crossing over – Stern's experiment.

Crossing over

In general, (i) certain genes assort randomly to agree with Mendel's law of independent assortment; (ii) other genes do not segregate randomly but are linked. These linked genes tend to be transmitted in unitary groups; (iii) the linked genes do not always "stay together" but are often separated by reciprocal exchanged of genes between chromosomes of a homologous pair to display incomplete linkage. The reciprocal exchange of genes between chromosome of homologous pairs is performed by a process termed as crossing over by Morgan. The process of crossing over can be defined as a process which produces new combinations (recombinations) of genes by interchanging of corresponding segments between non-sister chromatids of homologous chromosomes." The chromatids in which crossing over has occurred have new combinations of genes and are called cross overs. According to its occurrence in the germinal or somatic cells following two types of crossing over have been recognised.

A. Germinal or **meiotic crossing over:** Commonly crossing over occurs only in the germinal cells of reproductive organs during the process of gametogenesis which includes meiosis. This type of crossing over is called germinal or meiotic crossing over. It is universal in its occurrence and has great genetic significance.

B. Somatic or mitotic crossing over: Sometimes crossing over may occur during mitosis of somatic cells. This type of crossing over occur during mitosis of somatic cells. This type of crossing over occurs in rare cases, has no genetic significance and is called somatic or mitotic crossing over.

Mechanism of meiotic crossing over

According to the widely accepted White house model for the crossing over, the whole process of crossing over include following stages, viz., synapsis, duplication of chromosome, crossing over and terminalization (Whitehouse and Hastings, 1965).

1. Synapsis

During zygotene stage, of prophase-I of meiosis occurring in developing sex cells, the homologous chromosomes come close to each other and pairing on Synapsis between the homologous chromosomes (genetically identical chromosomes) takes place. Synapsis is an event of prime importance in meiosis which provides the mechanical basis of heredity and variation. It is started during zygotene when homologous chromosomes are held to make contact with each other at one or more points from which synapsis extends into adjacent regions and it ends or reaches its maximum in pachytene after which the homologs fall apart except the regions of chiasmata. Thus, synapsis is the phase of prolonged and close contact of homologous chromosomes due to attraction between two exactly identical or homologous regions or chromomeres. The resultant pairs of homologous chromosomes are called bivalents.

Causes of synapsis: To explain the question that why do homologous chromosomes, during synapsis, approach each other from a considerable distance and become closely associated, a British cytologist C.D. Darlington in 1937, has advanced the precocity theory that a chromosome must necessarily exist in a double condition, and that pairing of homologous chromosomes is an attempt to satisfy this requirement at a stage when each individual chromosome is single. It has been assumed generally that the force of attraction is electrostatic or chemical in nature (see Wilson and Morrison, 1966).

Recent molecular biological studies of synapsis have supported the precocity theory of synapsis. Hotta, Ito, and Stern (1966) have demonstrated that though main bulk of DNA of the total genome is already synthesized in the premeiotic interphase, but a small amount (approximately 0.3 per cent) of DNA is synthesized during zygotene and pachytene in the pollen mother cells of lily (Lilium). It has been suggested that the shortage of 0.3% DNA in chromosomes during zygotene creates the condition for their homologous pairing. Bogdanov *et al.*, (1968) have investigated similar unsaturated states of chromosomes during zygotene in Gryllus domesticus as there remains a shortage of 0.3% DNA and 25% histone proteins in chromosomes.

However, the exact cause and mechanism of synapsis is still unknown (see Sybenga, 1972). It has been suggested that pairing of relatively condensed chromosomes that cannot be a function of the DNA is regulated by specific loci on the chromosomes, the zygomeres. In some species a few zogomeres might occur in each chromosome, in others many (Sybenga, 1966).

Synaptinemal Complex- Montrose J. Moses (1955) has revealed a highly organized structure of filaments called synaptinemal complex in between the paired chromosomes of zygotene and pachytene stages in crayfish by electron microscopy. Synaptinemal complex has also been observed in a wide variety of species of plants and animal. In electron micrographs the synaptinemal complex appears as three parallel dense lines that lie equally spaced in a plane and flanked by chromatin. The elements of two lateral lines usually appear densest, while the element of central line is of variable prominence. Some fine transverse strands also cross between lateral elements, connecting them with the central element. Chemically, the synaptinemal complex contains DNA and a specific proteinaceous material called synaptinemal complex material.

The synaptinemal complex serves crossing over by facilitating effective synapsis. Robert King (1970) suggested that synaptinemal complex may orient the non-sister chromatids of homologs in a manner to facilitate it enzymatically induced exchanges between their DNA molecules. Comings and Okada (1971) have shown electron microscopically that synapsis occurs at two levels – one at chromosomal level and the other at molecular level. According to them, the synaptinemal complex pulls homologous chromosomes in the approximate association with each other but plays no role in molecular pairing of DNA strand.

2. Duplication of Chromosomes

The synapsis is followed by duplication of chromosomes. During this stages, each homologous chromosome of a bivalent splits longitudinally and form two identical sister chromarids, so that, each bivalent is now, composed of four chromatids. A bivalent having four chromatids is called tetrad.

3. Crossing over

The crossing over occurs in the homologous chromosomes only during the four stranded or tetrad stage. During the process of crossing over, two non-sister chromatids first break at the corresponding points due to the activity of a nuclear enzyme called endonuclease (Stern and Hotta, 1969). Then a segment on one side of each break connects with a segment on the opposite side of the break, so that the two non-sister chromatids cross each other at the point of break and exchange. The fusion of chromosomal segments with that of opposite one takes place due to the action of an enzyme called ligase (Stern and Hotta, 1969). According to the recent findings a little amount of DNA synthesis takes place during the crossing over process and that little amount (about 3% of total genome) of DNA is thought to repair the broken chromosomes. The crossing of two chromatids is called chiasma formation and the resultant cross as chiasma or chiasmata. The crossing over thus, includes the breaking of chromatid segments, their transposition and fusion.

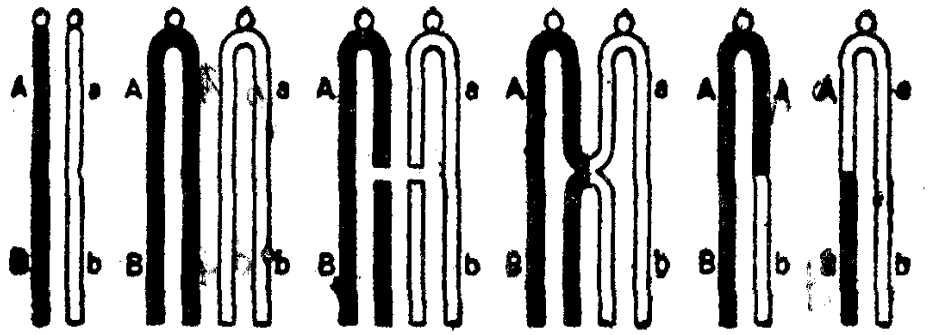


Diagram showing the mechanism of crossing-over.

4. Terminalisation

After the completion of crossing over, the non-sister chromatids start to repel each other because the force of synapsis attraction between them decreases. The chromatids separate progressively from the centromere towards the chiasma and the chiasma itself moves in a zipper fashion towards the end of tetrad. The movement of chiasma is called terminalisation. Due to the terminalisation the homologous chromosomes are separated.

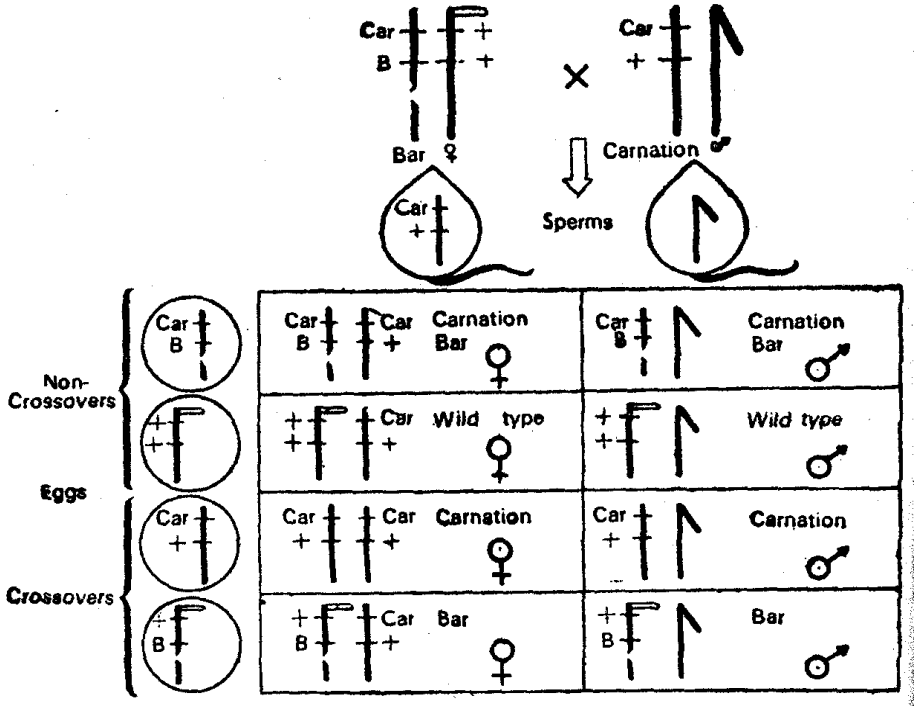
Cytological Proof for crossing over

The examples which have been cited in the section of incomplete linkage (Drosophila and maize) have also furnished good evidence for genetical detection of crossing over, but they did not give any cytologically demonstratable evidence in support of genetical crossing over, because, the homologous chromosomes appeared, on microscopic examination, to be exactly alike. It was impossible to observe whether chromosome blocks had changed places until visible markers of some kind could be incorporated on the chromosomes. The first cytological demonstration of genetic crossing over has been given by Stern (working with Drosophila) and H.B. Creighton and B. McClintock (working with maize) in 1931.

Stern's experiment: A wild type female Drosophila has one recessive gene for round eyes and one dominant gene for red eyes on each of its rod-shaped X chromosome, while, a mutant strain of it, has one mutant dominant gene Bar (B) for narrow eyes and one mutant recessive gene carnation (car) for light red eyes on its X chromosomes. By crossing these two strains, stern obtained a dihybrid having car and B genes on one X chromosome and normal genes (++) on other X chromosome. He made both of the X chromosomes of this heterozygote female aberrant by treating such flies with X-rays. The X chromosome having the genes car and B was broken into two segments, one fragment having both of the genes, while, the other X chromosome having a fragment of the Y chromosome attached to it and contained normal alleles (++). Thus, both aberrant X chromosomes of heterozygote were cytologically detectable. A female heterozygote with the aberrant X chromosomes was mated with a normal male having X chromosome with car + alleles, four classes of eggs (e.g., two type of eggs are cross overs and two types of eggs are non-crossovers) were produced which by fertilization produces following four kinds of females:

1. Carnation bar females with broken X without any fragment of Y.
2. The red round females unbroken X with the attached Y fragment.
3. The carnation round have the unbroken X without an attached Y fragment.
4. The red Bar have the broken X with the attached Y fragment.

Thus, flies in which crossing over was indicated phenotypically showed microscopic evidence of exchanges between homologous chromosomes. The physical or cytological basis of crossing over was thus established.



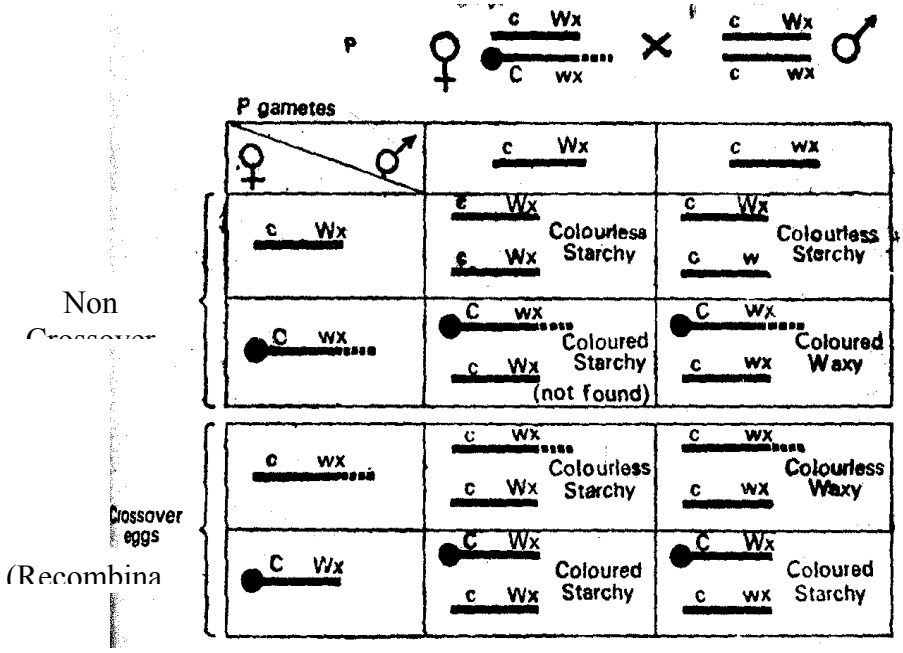
Stern's experiment for the detection of crossing over in Drosophila cytologically (after SRB, Owen and Edger, 1965).

Significance of Crossing over

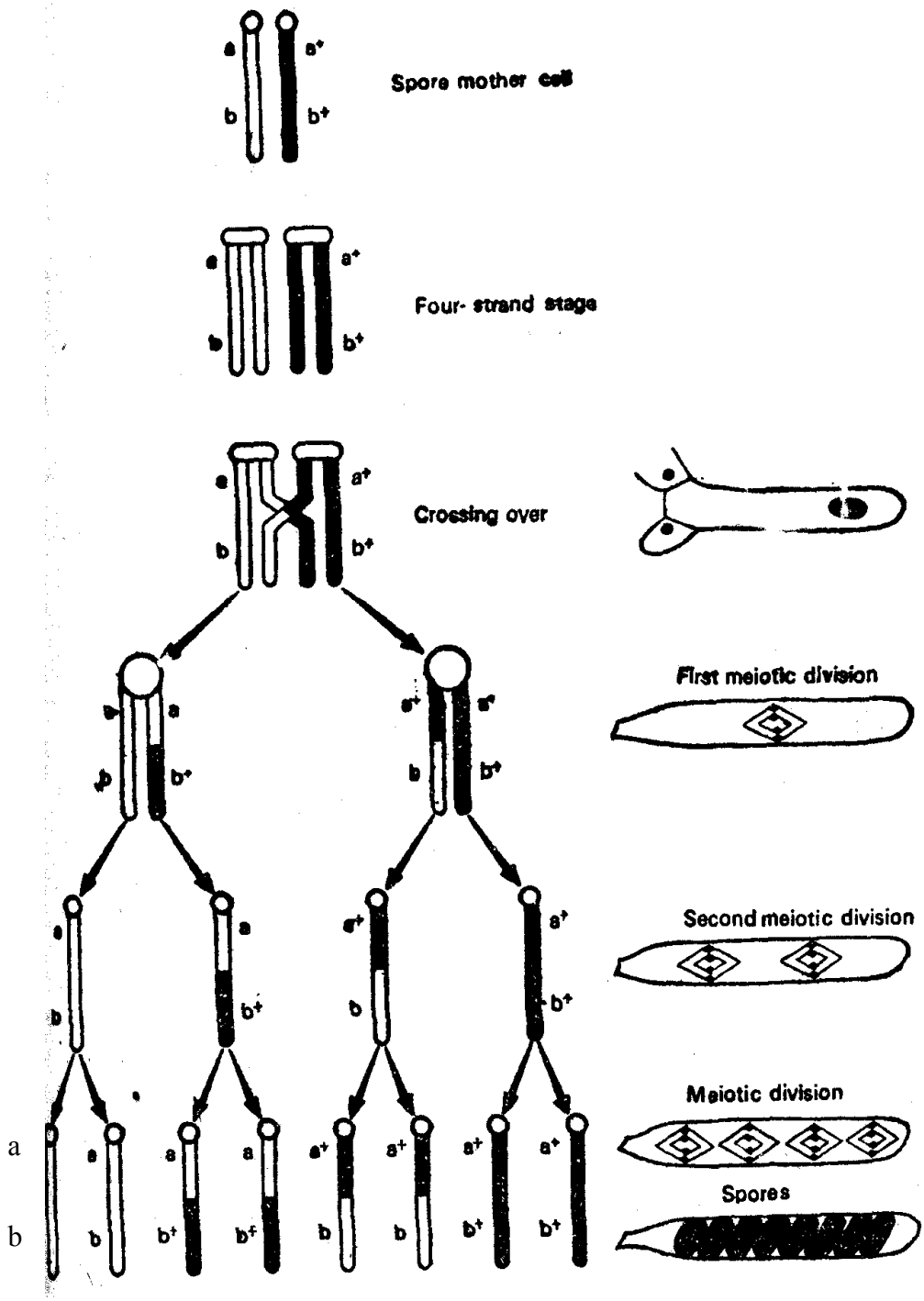
The process of crossing over has following genetical significance:

1. The frequency of crossing over is of great use in constructing genetic maps of he chromosomes.
2. It provides direct evidence for linear arrangement of linked genes in chromosomes.
3. It increases the frequency of genetical variation which are the raw materials of organic evolution.

Creighton and McClintock's experimetn: Creighton and McClintock like Stern made convincing correlation between cytological evidence and gentical resutls of crossing over in maize. They made the use of knob of 9th chromosome of maize which in two different strains might had one allele for either coloured aleurone (C) or colourless aleurone (c) and one allele for either starchy endosperm (Wx) or waxy endosperm (wx). The results of their experiment have been illustrated.



A diagrammatic representation of parallelism between cytological and genetic crossing over showing the phenotypes of aleurone and endosperm of F_1 grains and chromosome morphology and genotypes for microsporocytes produced by F_1 plants (after Burns, 1969).



Crossing over in *Neurospora* at four chromatid stage and crossover and non-crossover products (*i e* , ascospores). (after De Robertis *et al.*, 1970).

Lecture XIV

Strength of linkage and recombination – two point and three point test cross – double cross over, interference and coincidence – genetic map.

Strength of linkage and recombination:
Two point:

The percentage of crossing over between two linked genes is calculated by test crosses in which a F_1 dihybrid is crossed with a double recessive parent. Such crosses because involve crossing over at two points, so called two point test crosses. For example, a dihybrid having the genotype AC/ac is test crossed with a double recessive parent (ac/ac), then among F_2 test cross hybrids we may get 37% dominant genes at both gene loci (AC/ac), 37% recessive genes at both gene loci (ac/ac), 13% dominant gene at first gene locus and recessive genes at both gene loci (ac/ac), 13% dominant gene at first gene locus and recessive at the second gene locus (Ac/ac), and 13% recessive gene at first gene locus and dominant gene at second gene locus (aC/ac). The last two groups (i.e., 13% Ac/ac and 13% aC/ac) were produced by crossover gametes (13 + 13) from the dihybrid and the distance between the loci A and C is estimated to be 26 centimorgans. Because, double crossovers usually do not occur between genes less than 5 centimorgans apart, so for genes further apart, the three point test crosses and used.

Three point test cross:

As three point test cross or trihybrid test cross (involving three genes) gives us information regarding relative distance between these genes, and also shows us the linear order in which these genes should be present on chromosome. Such a three point test cross may be carried out if three points or gene loci on chromosome pair can be identified by marker genes. If, in addition to genes A and C indicated above, a third marker genes B is located in fairly closed proximity in the same linkage group, all three markers may be used together in conducting a more precise analysis of the map distance and the relative position of three points.

Suppose that we testcross trihybrid individuals of genotype ABC/abc and find in the progeny the following:

36% ABC/abc 36% abc/abc	9% Abc/abc 9% aBC/abc	4% ABc/abc 4% abC/abc	1% AbC/abc 1% aBc/abc
72% Parental type	18% Single crossover between A and B. (region I)	8% Single crossover between B and C. (region II)	2% Double crossover

To find the distance A-B we must count all crossovers (both singles and doubles) that occurred in region I = 18% + 2% or = 20% or 20 map units between the loci A and B. To find the distance B-C we must again count all crossover (both singles and doubles) that occurred in region II+8% + 2% = 10% or 10 map units between the loci B and C.

Reference: PSV : 214- 218

The A-C distance is therefore 30 map units when double crossovers are detected in a three point linkage experiment and 26 map units when double crossovers are undetected in the two-point linkage experiment above.

Without the middle marker (B), double crossovers would appear as parental types and hence we underestimate the true map distance (crossover percentage). In this case the 2% double crossovers would appear with the 72% parental types, making a total of 74% parental types and 26% recombinant types. Therefore, for any three linked genes whose distances are known, the amount of detectable crossovers between the two outer markers A and C when the middle marker B is missing ; (A-B crossover percentage) plus (B-C crossover percentage) minus (2X double crossover percentage).

Interference and Coincidence

In most higher organisms it has been found that one chiasma formation reduces the probability of another chiasma formation in an immediately adjacent region of the chromosome, probably because of physical inability of the chromatids to bend back upon themselves within certain minimum distances. The tendency of one crossover to interfere with the other crossover is called interference. The net result of this interference is the observation of fewer double crossover types than would be expected according to map distances the strength of interference varies in different segments of the chromosome and is usually expressed in terms of a coefficient of coincidence, or the ratio between the observed and the expected double crossovers.

$$\text{Coefficient of coincidence} = \frac{\%\text{ of observed double crossovers}}{\%\text{ of expected double crossovers}}$$

The coincidence is the complement of interference, so:

When interference is complete (1.0), no double crossovers will be observed and coincidence becomes zero. When, interference decreases, coincidence increases. Coincidence values ordinarily vary between 0 and 1. Coincidence is generally quire small for short map distance. There is no interference across centromere.

Three Point Test Cross in Maize

The preparation of a linkage map can be exemplified by using an example from maize involving three endosperm characters. These three characters are coloured aleurone (C) versus colourless (c) aleurone, smooth seed (Sh) versus shrunked seed (sh) and normal or non-waxy endosperm (Wx) versus waxy endosperm (wx). J. Sybenga (1972) has cited following results of three point test cross in maize:

Results of a three point test cross in maize and calculation of interference and coincidence. (after Sybenga (1972).

All three factors (genes) involve properties of the seed: the segregation can be read on the cob of selfed F_1 plant.

Parent: P1 c (colourless aleurone)-sh (shrunken seed)-wx (waxy endosperm)
P2 C (coloured aleurone)-Sh (smooth seed)-Wx (normal endosperm)

F1 : c – sh – wx / C – Sh – Wx, test crossed with c – sh – wx / c – sh – wx

Types in testcross with numbers of seeds found (representing gametic ratios):

CShWx	CShwx	CshWx	cShWx	Cshwx	cShwx	cshWx	cshwx	Total
238	672	19	98	107	39	662	2198	6033

Monofactorial segregation: C : c = 3036 : 2997
Sh : sh = 3047 : 2986 Slight shortage of recessives
Wx : wx = 3017 : 3016

Crossing over C-W : Cwx = 672 + 107
cWx = 98 + 662
1539 crossing over $\frac{1539}{6033} \times 100 = 25.51\%$

Crossing-over C-Sh : Csh = 19 + 107
cSh = 39 + 98
263 crossing over $\frac{263}{6033} \times 100 = 4.36\%$

Crossing-over Wx-Sh : Wxsh = 19 + 662
wxSh = 672 + 39
1392 crossing-over $\frac{1392}{6033} \times 100 = 23.07\%$

The greatest crossing-over percentage corresponds with the greatest distance and this must be between the outer two loci. The order, therefore, is C-Sh – Wx. The sum of C-Sh and Sh – Wx is 27.43 which is more than C-Wx estimated directly (25.51). the difference is a result of double crossing-over.

Double crossing-over : Csh Wx = 19
cSh wx = 39
58 percentage $\frac{58}{6033} \times 100 = 0.96\ \%$

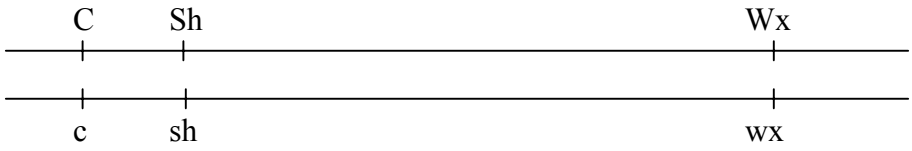
The product of 23.07% and 4.36% = 1.01% is the expected double crossing-over frequency. The difference, is due to interference. The coincidence value C can be calculated as $\frac{0.96}{1.01}$ = 0.95 and the interference equals 1 – 0.95 = 0.05

The distance C-Wx can be estimated directly when double crossing-over is taken into account, ie., the double crossing-over frequencies count twice:

Cwx = 672+107+2 x 19

cWx = 98=662=2 x 19

$$1655 \text{ crossing-over } \frac{1655}{6033} \times 100 = 27.46\%$$



An example of a three-point-test in maize (compare table). The order of three genes c, sh and wx can be given and the distance between them in per cent crossing over: this is the beginning of a genetic map (after Sybenga, 1972).

Lecture XV

Sex determination – chromosomal mechanism of sex determination and its types. Genic balance theory of sex determination of Bridges.

Sex determination and Sex linkage:

Sex differentiation in living organisms into male and female causes morphological, physiological and behavioral differentiation between the two sexes and this phenomenon is called sexual dimorphism. In a large number of species of animals and a small number of species of plants, eggs and sperms are produced by different individuals, *viz.,* females and males respectively. In most of the dioecious organisms, females and males differ visibly in chromosomal constitution. The precise form of the chromosomal differences between the sexes is not the same in different organisms. Four types of sex chromosome mechanism or heterogamesis have been recognised and they are the following:

1. Sex chromosome mechanism

a) Heterogametic Male - XX-XO type

The chromosome theory of sex determination was put forward by McClung (1902), an American zoologist, who observed that the male grasshopper possessed and odd number of chromosomes in contrast to the female which possessed an even number. The proof for this was furnished by Wilson and Stevens (1905) who demonstrated in bugs that chromosome distribution followed a course exactly similar to that of sex distribution.

In the squash bug, *Protenor,* the females have 14 chromosomes and the males have only 13 chromosomes in their somatic cells. In the females, 7 bivalents are formed during meiosis. All the eggs have, therefore, 7 chromosomes each. In the males, one odd, unpaired chromosome and 6 bivalents are seen during meiosis. The unpaired chromosome passes undivided into one of the two daughter cells. Two kinds of sperms in equal numbers are, therefore, formed, one kind with 7 chromosomes each and the other kind with chromosomes each. An egg with 7 chromosomes fertilised by a sperm with 7 chromosomes produces a female with 14 chromosomes and an egg with 7 chromosomes fertilised by a sperm with 6 chromosomes produces a male with 13 chromosomes. The odd chromosome of the male thus determines the sex and hence called the *sex-determiner* or the *sex chromosome* or the 'X' *chromosome*. The other chromosomes which are alike in females and males are called *autosomes.*

In grasshopers and bugs, the female is thus XX and the male is XO (using O to indicate the absence of the X chromosome). Among plants, *Dioscorea sinuata* and *Vallisnaria spiralis* are examples where the female is XX and the male XO.

XX-XY type

In many animals and plants, females and males have the same even number of chromosomes, but, whereas in the females the members of each pair of chromosomes are alike, in the males the members of one pair of chromosomes are dissimilar in size or form.

Reference: DDS: 159-172

In *Drosophila melanogaster* the female has four pairs of chromosomes as follows: (1) a pair of rod-shaped chromosomes, (2) a pair of V-shaped chromosomes, (3) a pair of slightly longer V-shaped chromosomes, and (4) a pair of very short dot-like chromosomes.

In the male *Drosophila,* there is only one rod shaped chromosome, the other member of this pair being inverted J-shaped (i.e., hook-shaped, or like a rod with a bent end). Wilson, who discovered this type of chromosome arrangement in 1905, designated the unlike member of this pair in the male as the 'Y' chromosome and the other member which is like the members of one pair in the female as the 'X chromosome.

All the eggs have one X chromosome and 3 autosomes each. Sperms are, however, of tow kinds; one kind with one X chromosome and 3 autosomes each, and the other kind with one Y chromosome and 3 autosomes each. Any egg fertilised by an X-containing sperm produces a female and any egg fertilised by a Y-containing sperm produces a male.

This type of sex determination in which the female has two X chromosomes and the male one X and the Y chromosome is very widespread, being found in many invertebrates including insects, in some fishes, in mammals including man and in many dioecious plants like *Melandrium album, M. rubrum, Humulus lupulus, rumex angiocarpa, Salix, Smilax, Cannabis* and *Populus.*

In human beings, 4 chromosomes are present in the somatic cells. Females have 22 pairs of autosomes and two X chromosomes. Males have 22 pairs of autosomes and one X chromosome and one very short Y chromosome, considerably smaller than the X chromosome. Each egg carriers 22 autosomes and an X chromosome. Sperms, however, are of two kinds, one kind with 22 autosomes and a Y. The sex of a child is determined at the time of fertilisation by the kind of sperm that happens to meet and penetrate the egg, an X-bearing sperm producing a girl and a Y-bearing one, a boy.

b) Heterogametic Female – ZO-ZZ type

In all the above instances, the female is the homogametic sex because it produces eggs, all of which are alike and the male is the heterogametic sex because it produces two kinds of sperms. But there are instances where the female is the heterogametic sex and the male is the homogametic one.

In a moth, *Talaeoporia,* the females have 59 chromosomes and the males have 60 chromosomes in their somatic cells. The eggs are of two kinds, one kind with 29 chromosomes and the other kind with 30 chromosomes. All the sperms have 30 chromosomes each. On fertilisation, an egg with 29 chromosomes gives rise to a female and an egg with 30 chromosomes gives rise to a male.

To distinguish this from the Protenor type, the sex chromosome found in the male is designated at 'Z'. The female is thus ZO (using O to denote the absence of one Z chromosome) and the male is ZZ.

ZW-ZZ type

In birds, including the domestic fowl, certain insects, fishes and reptiles, the female has an unlike pair of chromosomes, ZW, and forms eggs of two sorts, one with a 'W' chromosome and the other with a 'Z' chromosome. The male has like pairs of chromosomes. On fertilisation, an egg with a W chromosome gives rise to a female and an egg with a Z chromosome gives rise to a male.

Among plants, *Fragaria elatior* is one in which the female is ZW and the male is ZZ.

Balance theory of sex determination

A number of lines of evidence indicate that even in dioecious species, all individuals have genes for both sexes. To quote Bridges, 'Both sexes are due to the simultaneous action of two opposed sets of genes, one set tending to produce the characters called female and the other to produce the characters called male'. Which sex actually develops is decided by the balance, i.e., by the preponderance of the female-determining or of the male-determining genes. The sex chromosomes are merely vehicles of genes which help in tilting the balance in one direction or another.

Support for the balance theory of sex determining comes from the work of Bridges (1921) on *Drosophila*. Bridges observed some females of *Drosophila melanogaster* with three X chromosome and three sets of autosomes (i.e., triploids). When he crossed them with normal (diploid) males, he found that some of the progeny and one or more chromosome less or more than the normal flies (i.e., aneuploids). His results are given below:

	X + A	Y + A
2X + 2A	3X + 3A Female	2X + Y + 3A Intersex
X + A	2X + 2A Female	X + Y + 2A Male
2X + A	3X + 2A Superfemale	2X + Y + 2A Female
X + 2A	2X + 3A Intersex	X + Y +3A Supermale

Bridges found intersex, super females and supermales among the progeny. Intersexes are sterile individuals intermediate between females and males but are different from gynandromorphs which are typically female in certain portions of the body and typically male in others. Superfemales and supermales are sterile individuals which are very weak and very poor in viability.

Flies with two X chromosomes and two sets of autosomes are females but flies with three X chromosomes and the same two sets of autosomes are superfemales.

2X + 2A Female
3X + 2A Superfemale

This shows that in *Drosophla* the X chromosomes carry genes that are predominantly female-determining.

Flies with one X, one Y and two sets of autosomes are normal males but flies with one X, one Y and three sets of autosomes are supermales.

1X + 1Y + 2A	Male
1X + 1Y + 3A	Supermale

This shows that the autosomes carry genes that are predominantly male-determining.

Flies with two X chromosomes and two sets of autosomes are females. So also, flies with two X, one Y and two sets of autosomes are females.

2X +2A	Female
2X + 1Y + 2A	Female

That individuals with two X, one Y and two sets of autosomes are female in spite of the presence of the Y chromosome shows that the Y chromosome plays no positive role in sex determination. That the Y chromosome does not determine maleness is also shown by the fact that flies with one X chromosome and two sets of autosome (i.e., XO flies) are males in spite of the absence of the Y chromosome. They are, however, sterile showing thereby that the Y chromosome contains male fertility genes necessary for the production of fertile male.

Bridges interpreted these results as follows:

Sex in *Drosophila melanogaster* is determined by the X chromosomes as well as by the autosomes, the ratio of the number of X chromosomes to the number of sets of autosomes being the deciding factor. In a normal (diploid) male, the X / A value is 1.00, there being two X chromosomes and two sets of autosomes, and in a normal (diploid) male, the X /A value is 0.50, there being only one X chromosome and two sets of autosomes. Intersexes have an X /A value lying between 0.50 and 1.00. superfemales have an X / A value exceeding 1.00 while supermale have an X / A value less than 0.50. the relationship of chromosomes to sex determination in *Drosophila melanogaster* is shown in Table.

Relationship of chromosomes to sex in *Drosophila*

Chromosome constitution			X / A	Sex
X	Y	A		
3 X		2 A	1.50	Superfemale
3 X		3 A	1.00	Female (triploid)
2 X		2 A	1.00	Female (diploid)
2 X	1 Y	2 A	1.00	Female (diploid)
2 X	1 Y	3 A	0.67	Intersex
2 X		3 A	0.67	Intersex
1 X	1 Y	2 A	0.50	Male
1 X	1 Y	3 A	0.33	supermale

Haplodiploidy

In hymenopterous insect such as ants, bees, sawflies and wasps, the fertilized eggs develop into diploid females and the unfertilized eggs into haploid males. Male haploidy is otherwise termed as arrhenotokous parthenogenesis. Thus haplodiploidy mechanism is found to operate in some organisms in determining sex.

Single gene effect

In certain other organisms such as *Chlamydomonas, Neurospora, Yeast, Asparagus, Drosophila* and several fishes, sex determination is the result of single gene effect.

The Y Chromosome

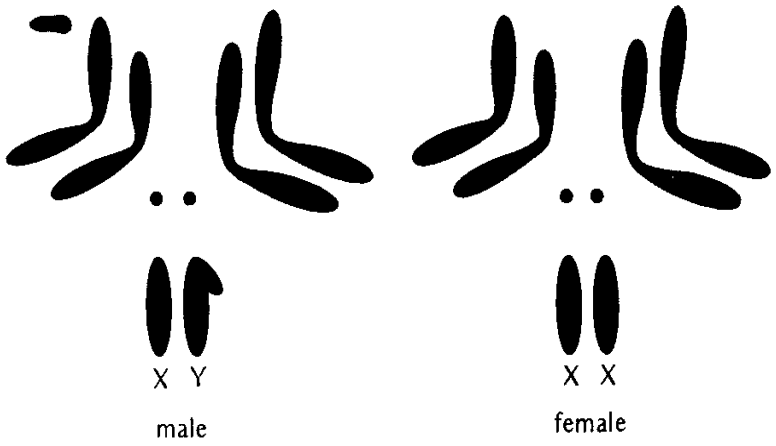
Once different types of sex chromosomes could be distinguished, further investigations by Wilson, Stevens, Montgomery, and others showed a wide variety of chromosomal sex-determining mechanisms. The example of Protenor was essentially the simplest, because only the presence of absence of a single chromosome was the determining factor, and this was consequently called the XO-XX type. The male Protenor, forming two types of gametes, with and without the X chromosome, and designated as the heterogametic sex.. The female, producing gametes all of the same type, was termed homogametic.

It was soon found that the single X chromosome of the male in many species pairs in meiosis with another chromosome, called the Y. Although similar in appearance to the X in some cases, the Y is usually morphologically distinct. In *Drosophila metanogaster*, for example, the Y has a J shape, as compared to the rod-shaped X. the XY-XX system thus results in the same number of chromosomes in both males and females. According to this terminology, if we designate a haploid set of autosomes collectively by the letter A, the male produces two types of sperm, X + A and Y + A, and the female one type, X + A. Fertilization may, therefore, be of two kinds:

(X + A) + (X + A) = XX + 2A = female
sperm egg

(Y + A) + (X + A) = XY + 2A = male
sperm egg

In *Drosophila melanogaster* there are three autosomes in each haploid set, yielding a total diploid number of eight chromosomes in each sex.



Chromosome constitutions (karyotypes) of males and females in *Drosophila melanogaster.*

Lecture XVI

Sex linked inheritance – Cris cross inheritance – Reciprocal difference – Holandric genes – Sex influenced and Sex limited inheritance – Sex determination in plants – Melandrium, papaya and maize.

Sex linkage:

Red Eyed x White eyed

↓

F1 Both sexes – Red eye

↓

F2 Red eye : White eye

3 : 2

F2 Female – Red eye

F2 males - ½ red eye : ½ white eye

White eye x red eye

↓

F1 Females – red eye

Males – white eye

1) F1 females & males were different
2) F1 flies of both sexes from a cross between red eyed female and white eyed male were red eyed.
3) Colour of eyes located in x chromosome.

In a culture of a normal wild type of *Drosophila melanogaster* with red eyes, Morgan found in 1909 a single male individual in which the eyes were white. From this, Morgan established a true breeding strain of white-eyed flies.

Morgan crossed a red-eyed female with a white eyed male and found that all the first generation flies of both sexes were red eyed. When these were bred together and an F_2 generation obtained, it was found that three-fourths of the flies had red eyes, and one fourth had white eyes, indicating that red and white eyes, indicating that red and white eye colours are due to an allelic pair of genes of which red is the dominant. All the F_2 females, however, were red-eyed but of the F_2 males, half were red-eyed and half were white-eyed.

Reference: DDS: 159-172

A reciprocal cross was made between a white-eyed female and a red-eyed male. It was found that among the F_1 offspring, all the females were red-eyed and all the males were white-eyed.

The results were quite unexpected firstly because the phenotypes of the F_1 females and males were different and secondly because all the F_1 flies of both sexes from a cross between a red eyed female and a white-eyed male were red-eyed.

The F_1 females seem to agree with out expectations that the heterozygotes will exhibit the dominant character, *viz.,* red colour of the eye. It is the white-eyed F_1 males that do not agree with our expectations. They do not seem to have received the gene for red eye from their father. They seem to have received only the gene for white eye from their mother.

The female and male flies are similar in having two pairs of V-shaped chromosomes and one pair of dot like chromosomes. Whereas the females have one pair of rod-shaped chromosome, the males have only one rod-shaped chromosome, the other member of the pair being hook-shaped. The different results from the reciprocal crosses could be explained only on the assumption that the gene for colour of the eyes is located on the X chromosome and that the Y chromosome has no gene for colour of the eyes.

Cytological studies revealed that the F_1 males were like the male parent in having one rod-shaped and one hook-shaped chromosomes besides the three pairs of autosomes. The sons must have received the hook-shaped chromosome only form their father. If, therefore, the hook-shaped chromosome of the father carried a gene for red colour of the eye, the sons must have been red-eyed. The sons, however, were white-eyed and it was therefore assumed that the Y chromosome carries no gene for red colour of the eye. The X chromosome of the sons could be received only from their mother. The sons could be white-eyed only if the gene for white eye is located on the X chromosome of the white-eyed mother.

The daughter were like the mother in having two rod-shaped chromosomes besides the three pairs of autosomes. The daughters could not have received more than one X chromosome from the mother. The other X chromosome of the daughters must have been received from the father. The daughters could be red-eyed only if the gene for red eyes is located on the X chromosome of the father.

Because a white-eyed female crossed with a red-eyed male produces red-eyed females and white-eyed males, this method of inheritance is often referred to as criss-cross inheritance.

When the F_1 flies were mated together, the following results were obtained in the F_2.

Red-eyed female	1
White-eyed female	1
Red-eyed male	1
White-eyed male	1

The F_2 consisted of red-eyed and white-eyed individuals in equal numbers in both sexes.

Morgan concluded that the gene for eye colour is located on the X chromosome and that the Y chromosome carries no gene for eye colour.

Sutton and Boveri hypothesised that genes are borne on chromosomes (i.e., the chromosome theory of heredity) but it was Thomas Hunt Morgan (1910) who first associated a particular gene (i.e., the gene for eye colour) with a particular chromosome (i.e., X chromosome) visible in microscopic preparations and showed that the gene for eye colour follows exactly the transmission of the X chromosome. Eighteen other genes in *Drosophila melanogaster* follow the same method of inheritance as white colour of the eye which indicates that these genes are also carried on the X chromosome. As the gene is located on the X chromosome, it is called an X-linked gene. This pattern of inheritance is called sex linkage.

Just as the eye colour in *Drosophila* whose gene is present only one the X chromosome and not on the Y chromosome, hence called X-linked, there are genes located only on the Y chromosome and its allele absent in the X chromosomes. Such genes are called Y-linked or holandric genes. The gene responsible for hypertrichosis causing hairy pinna (earlobes) in human beings is a Y-linked gene.

There are certain homologous regions on the X and Y chromosomes in which both the alleles of a gene may be present as in the case of bobbed bristles (b) and its allele (b+) for normal bristles. Such genes present both in the X and Y chromosomes are called XY-linked genes. Genes for colour blindness, Xerodermia pigmentosum (Pigmentation on the skin), Retinitis pigmentosa (pigmentation on the eye retina), Nephritis *etc.,* in human beings are XY- linked.

Sex-influenced character

All genes which are carried by the chromosomes are said to be sex-linked. All known sex-linked genes lead to phenotypes which have nothing to do with sex.

Sex-influenced characters, are characters which may be expressed differently in the two sexes even when their genotypes are identical. The mere influence of the sex of the individual may be sufficient to alter the phenotypic expression of a gene. The most common expression of sex influence is that dominance is reversed between the sexes. Genes determining sex influenced characters are borne on autosomes.

A typical example of a sex-influenced character is the presence of horns in sheep. Both sexes of Dorset sheep are always horned while both sexes of Suffolk sheep are always hornless. If Dorset and Suffolk are crossed, the F_1 females are hornless while the F_1 males are horned. On interbreeding the F_1 sheep, and F_2 is obtained in which the females show a ratio of 3 hornless : 1 horned and the males show a ratio of 3 horned : 1 hornless. Presence of horns may therefore be said to be a recessive character in females but a dominant character in males.

P	Horned female		x		Hornless male
	HH				**hh**
F_1			**Hh**		
			Female, hornless		
			Male, horned		
F_2	**1 HH**	**:**	**2Hh**	**:**	**1 hh**
Female	Horned		Hornless		Hornless
Male	Horned		Horned		Hornless

Reciprocal crosses show no differences because the gene is carried by the autosome.

Baldness in human beings is a sex-influenced character which is recessive in females and dominant in males.

Sex-limited character

Sex-limited inheritance is an extreme type of sex influence in which a particular phenotype can be expressed only in one sex. As genes for sex-limited characters are borne on autosomes, all genotypes should occur with identical frequencies in both sexes but the physiological differences between the sexes are such that certain genotypes can be expressed only in one sex. Unlike sex influenced characters in which gene is dominant in one sex and recessive in the other, sex-limited characters are controlled by genes which have no visible influence at all in one sex either as a homozygote or as a heterozygote.

In the yellow clove butterfly, *Colias,* females may be either yellow or white but males are always yellow. White colour in the females depends on a dominant gene W so that females of genotype WW or Ww are white while ww are yellow. Males are yellow irrespective of whether they are WW, Ww or ww. The genotypes of the males can be determined by mating them with yellow females and observing the colour of the female progeny. If all the female progeny are white the male parent in WW. If the female progeny are in the proportion of 1 white : 1 yellow, the male parent is Ww. If all the female progeny are yellow, the male parent is w. Thus white colour is a sex-limited character found only in the females.

In domestic poultry, cock-feathering is a character limited to the male sex. Hen-feathering is due to a dominant gene H and cock-feathering is due to its recessive allele h, but females with genotype hh are hen-feathered. The genotypes and their corresponding phenotypes are as follows :

genotype	Phenotype	
	Female	Male
HH	Hen-feathered	Hen-feathered
Hh	”	”
Hh	”	cock-feathered

Removal of the ovaries in hens with genotype hh results in cock-feathering. This indicates that the female sex hormone inhibits the production of cock-feathering in hens with genotype hh.

Sex reversal

In several species of plants that are normally bisexual, suppression of the male or female structures has been observed in nature. The androecium getting converted into petels in ornamental plants or carpels as in carrot and cabbage or pistils as in maize, papaya and primrose has been observed. When the stamens get converted into rudimentary organs without the pollen sac and pollen, they are called staminodes and a similar conversion of the pistil into nonfunctional rudimentary organ is called the pistillode. The phenomenon in which there is suppression of one sex at the expense of the other is called sex reversal. The sex reversals are mostly due to physiologicl and biochemical alterations involving sex hormones.

In maize, rarely it is observed that the male inflorescence called tassel bears seeds due to sex reversal. The recessive gene ‘ba’ is responsible for barren plants and another recessive gene ‘ts’ is responsible for tassel seed. Sex reversal in maize is due to the genetic constitution of the plants.

Lecture XVII

Cytoplasmic inheritance – its characteristic features – examples for Chloroplast, Mitochondrial, Plasmid and Episomic inheritance.

Cytoplasmic inheritance

Though, the genes of nuclear chromosomes have a significant and key role in the inheritance. Certain experimental evidences suggest the occurrence of certain extranuclear genes or DNA molecules in the cytoplasm of many prokaryotic and eukaryotic cells. These cytoplasmic extra-nuclear genes or DNA molecules of plasmids, mitochondria, chloroplasts, endosymbionts and cellular surfaces have a characteristic pattern of inheritance which does not resemble with that of genes of nuclear chromosomes soma, uniparental, maternal, extra-chromosomal, cytoplasmic and extra-nuclear inheritance.

Comparision of extra-nuclear and nuclear genetic systems

The inheritance of genes of nuclear chromosomes is characterized by the fact that the genes from male and female parents contribute equally to the genetic constitution of the progeny. Therefore, in it, the reciprocal crosses between parents of different homozygous genotypes will yield offsprings of identical phenotype except for sex-linked genes, while in extra-nuclear inheritance, male and female parents tough contribute equally the nuclear genes to the progeny but they do not make equal contributions of extra-nuclear genes to the progeny as pollens or sperms carry little or no cytoplasm or extranuclear genes, while eggs or ova contribute large amount of cytoplasm and many extra-nuclear genes in the form of inactive mRNA, rRNA and tRNA and also in the form of DNA of mitochondria, and chloroplasts.

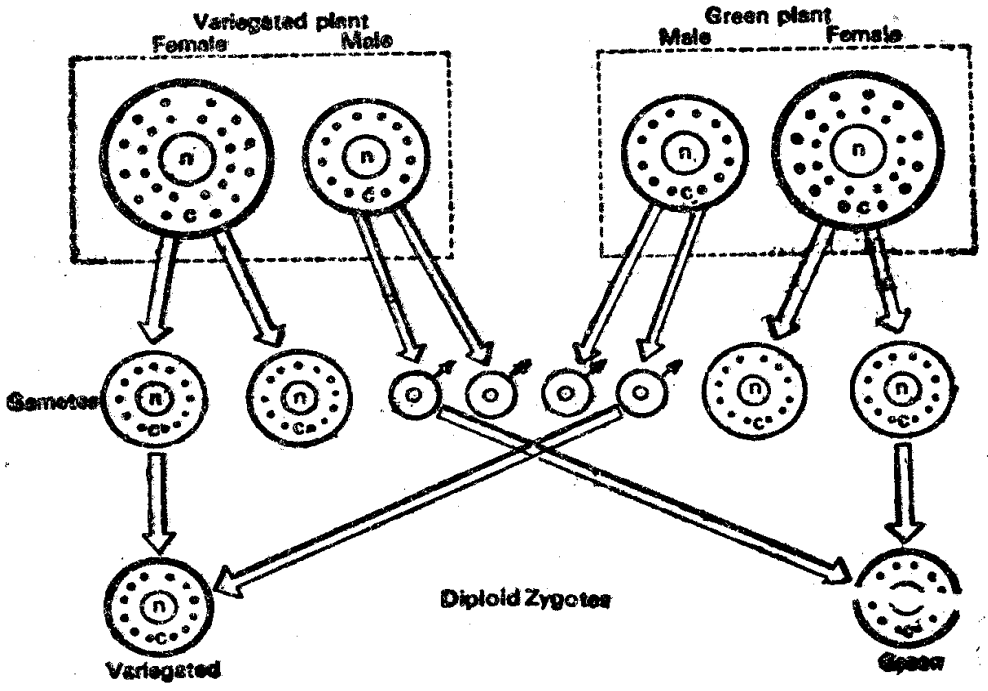
Extra-nuclear inheritance by cellular organelles

Chloroplasts and mitochondria are organells that contain their own DNA and protein-synthesizing apparatus. A widely held theory concerning their origin proposes that they were once infectious endosymbiotic prokaryoties that evolved such a dependence on the gene products of the host that they are no longer able to function autonomously.

This theory has been supported by the fact that the genetic components of these organelles are often similar to those founds in prokaryotes. Chloroplast chromosomes are found to contain more DNA than the average mitochondrial chromosomes, and thus can potentially carry more genetic information. The genetic materials of chloroplasts and mitochondria will be transmitted to offspring almost exclusively via the egg. Maternal inheritance due to chloroplast and mitochondria is well illustrated by following examples:

Reference: PSV: 531-542, DSR:

a) Chloroplasts inheritance in variegated four o' clock plant – The cytoplasmic or extra-nuclear inheritance of colour in plant plastids was first of all described by C. Correns in 1908 in the four o' clock plant, *Mirabilis jalapa*. In contrast to other higher plant, *Mirabilis* contain three types of leaves and parts : (1) Full green leaves or parts having chloroplast, (2) White (pale) leaves and branches having no chloroplast, (3) variegated branches having leucoplast in white (pale) areas and chloroplast in green patches. Because, the chlorophyll pigment of chloroplast is related with photosynthetically prepared food and leucoplasts are incapable to perform photosynthesis, so the white or pale parts of plant survive by receiving synthesis, so the white or pale parts of plant survive by receiving nourishment from green parts. Correns reported that flowers on green branches produced only green offsprings, regardless of the genotype and phenotype of pollen parent and likewise, flowers form the white or pale branches produced only white or pale seedlings regardless of genotypes and phenotype of pollen parents. The plants developing from the white or pale seedlings die because they lack chlorphyll and cannot carry on photosynthesis. Correns further reported that flowers form the variegated branched yielded mixed progeny of green, white (pale) and variegated plants in widely varying ratios.



Diagarammatic illustration of maternal plastids inheritance in a diploid plant such as Mirabilis which has little or no cytoplasm in pollen gametes; n = nucleus and C-cytoplasm, (after Gardner, 1968).

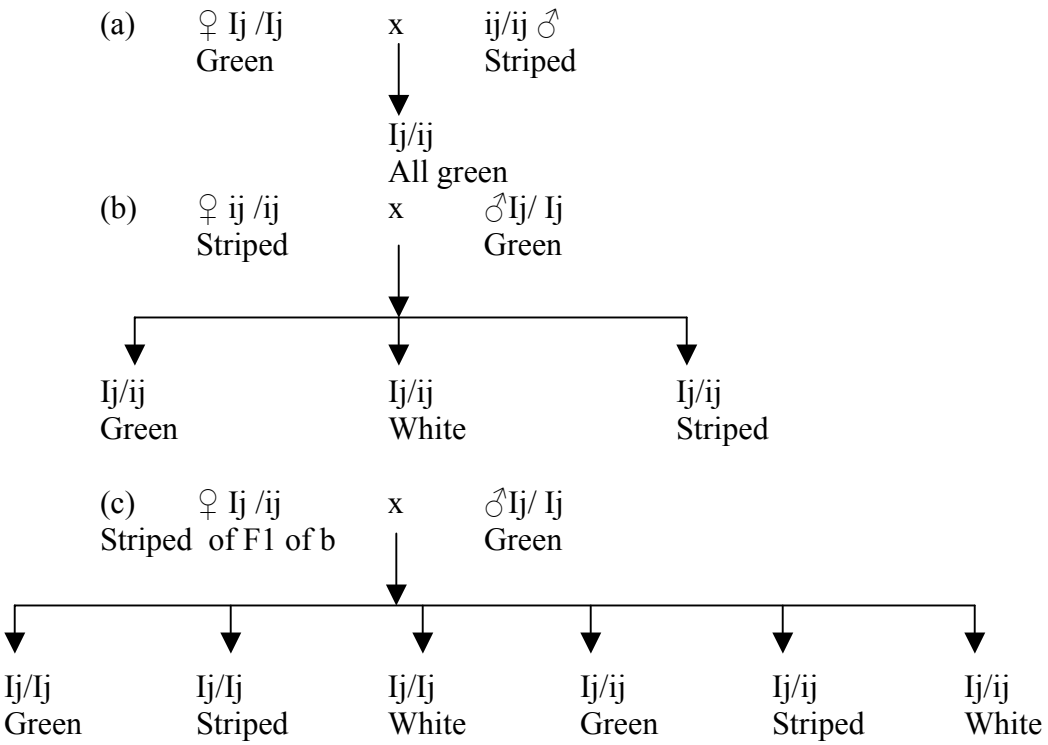
The irregularity of transmission from variegated branches could be understood by considering cytoplasmic gene (plasmogenes) of plastids. A study of the egg during oogenesis in Mirabilis reveals that the ooplasm contains plastids like cytoplasm of other plant cells. If the egg cell is derived from green plant tissues, its ooplasm will contain coloured plastids; if derived from white plant tissues, its ooplasm will contain white plastids; if derived from variegated tissues, its cytoplasm may contain coloured plastids only, white plastids only or a mixture of coloured and white plastids. A study of the pollenogenesis, however, reveals that pollen contains very little cytoplasm which in most cases is devoid of plastids. Without the plastids, the pollen cannot affect this aspect of the offspring's phenotype. In this type of inheritance because maternal cytoplasm has active participation in the determination of the phenotype of future generation, therefore, this is also known as maternal inheritance.

b) Maternal inheritance by iojap gene of corn – Another example from higher plants also suggests the existence of plastid genes controlling plastids integrity. A gene in corn plant called iojap (ij) has been mapped by M. Rhoades (1946) to nuclear chromosomes vii. Plants homozygous for ij are either inviable white seedling or variegated with a characteristic white striping, the phenotype being known s striped. When the variegated plants serve as female sin a cross they give rise to green, white, and striped progeny, regardless of the nuclear genotype of the paternal parent. Thus, if the pollen derives from a normal green Ij/Ij plant as in figure, the resulting progeny will be Ij/ij heterozygotes but many will exhibit abnormal plastid pigmentation : the presence of the "normal" Ij gene has no curative effect. In the reciprocal Ij/Ij♀ x ij/ij ♂ cross on the other hand, the Ij/ij progeny are all normally pigmented.

The iojap trait, thus, exhibits classical maternal inheritance once it has become established n an ij/ij plant. Moreover, once established it becomes independent of the ij gene, as can be demonstrated by crossing F1 Ij/ij variegated females to Ij/Ij normal males. As shown in figure, a mixture of green, striped and white progeny again results, even though some of the striped an white plants now have results, even though some of the striped and white plant snow have and Ij/Ij genotype. Thus the iojap trait, once established, is permanent.

The iojap phenomenon has been explained by two hypotheses. One hypothesis holds that the ij/ij genetic constitution could bring about or permit, frequent mutation sin the chloroplast genome that result in the production of lines of abnormal plastids. Another hypothesis suggests that certain cytoplasmic elements other than chloroplasts mutations come into being or residence in ij/ij cells, are later inheritance in the absence of their "susceptible" or "permissive" genotype, and bring about the bleaching of chloroplasts.

This type of maternal inheritance by plasma genes of chloroplasts has been also studied in many other higher plant such as barley, *Oenothera* sp., rice etc.



a) Cross between green (normal) striped (iojap) plants,
b) Reciprocal of cross (a) C. Cross of F1 striped females (of cross b) normal (green) males.

Maternal inheritance by iojap gene in maize

The egg regularly contributes much more cytoplasm to the next generation than does the sperm. It should therefore be expected that in cases of cytoplsmic inheritance, differences between reciprocal crosses would result.

The proof that a difference in hereditary traits is due to a difference in cytoplasm and not due to a difference in genes necessarily involves the demonstration that the organisms manifesting the different traits have identical genomes. If they have different genotypes, the observed differences might be due to the differences in genes. But if they have identical genotypes the conclusion may be drawn that the cytoplasmic particles are responsible for the differences.

Rhoades (1946) identified the iojap gene (Ij) in maize located in chromosome VII controlling plastid inheritance in the plant. The gene Ij is responsible for the normal green colour of the plant. When normal green plants with Ij Ij are used as females and pollinated by pollen from striped plants with ij ij, the F1 plants with Ij ij are wholly green.

Green	x	Striped
Ij Ij		ij ij

F1 : Green
Ij ij

When striped plants with ij ij are pollinated by pollen from the normal green plants with Ij Ij, the F1 plans, all of which have the same genotype, Ij ij are of three different phenotypes, viz., normal green, striped and white.

Striped	x	Green
ij ij		Ij Ij

F1 : Green, striped or white
Ij ij

When plants with the same genotype Ij ij have different phenotypes, viz., normal green, striped, or white, the differences can be attributed only to differences in plastids.

Cytoplasmic male sterility in maize

In several cases of cytoplasmic inheritance in plants, plastids have been shown to be the vehicles of heredity but in several other cases, cytoplasmic particles other than plastids have been identified as the basis for extranuclear transmission. Among these is a case of male sterility in maize. Most or all of the pollen grains of such male sterile plants are aborted. This character is transmitted only through the female and never by the pollen. When all of the chromosome of the male sterile line were replaced with chromosomes of normal plans, the line still remained male sterile, showing thereby that male sterility is controlled by some agency in the cytoplasm. It was later recognized that cytoplasmic male sterility in maize results from alterations in the hereditary units in the mitochondria (mitochondrial DNA).

Mitochondrial inheritance in Petite mutants of Yeast

Colonies smaller in size than the normal colonies are occasionally observed in the yeast (*Saccharomyces cerevisiae*) when grown on solid medium.. these small colonies are called petite mutants. Petites have slow growth as they grow by clucose fermentation anaerobically and hence inefficient in metabolism as compared to the normal clonies with aerobic metabolism.

Among the petite mutants. There are segregational petites that exhibit normal Mendelian segregation when crossed with wild type and the petite is recessive to wild type. Another class of petite mutant is called natural petite, which on crossing with wild type produces only wild type colones and is thus uniparental in inheritance. The reason for such a differential behaviour seems to be that majority of neutral petites lack most or all mitochondrial DNA responsible for oxidative respiration. The petite phenotype is the result of large deletion in mitochondiral DNA. Thus the mitochondria is found to be responsible for inheritance in respiration defective mutants in yeast.

Cytoplasmic inheritance

Cytoplasmic inheritance is due to the plasmagenes located in cell organelles (plastids and mitochondria). The characteristic features of this inheritance are summarized below.

Characteristics of Cytoplasmic Inheritance

1. Reciprocal Differences
2. Lack of segregation
3. Irregular Segregation in Biparental Inheritance
4. Somatic Segregation.
5. Association with Organelle DNA
6. Nuclear Transplantation.
7. Transfer of Nuclear Genome Through Backcrosses.
8. Mutagenesis.
9. Lack of Chromosomal Location
10. Lack of Association with A parasite, Symbiont or Virus.

Lecture XVIII

DNA, the genetic material – Griffith's experiment – Experiment of Avery, MacLeod and McCarty, confirmation by Hershey and Chase.

Considering the proportion of different constituents of cell, nucleic acid was found to be constant in volume in all the cells as compared to other cellular contents and hence it was inferred to be the hereditary material. There are two types of nucleic acid, the deoxyribonucleic acid (DNA) and the ribonucleic acid (RNA). By staining nucleic acid, Feulgen (1924) found that the DNA was localized in the nucleus, while the RNA was found to occur outside the nucleus in the cytoplasm.

The experiments of Griffith (1928) with the pneumonia bacterium and the interpretation of results by Avery, MacLeod and McCarty (1944) confirmed the DNA as the hereditary material.

Bacterial Transformation

Griffith (1928) worked on the pneumonia causing spherical shaped bacterium, *Diplococcus pneumoniae.* Some of the strains of this bactium have a smooth polysaccharide capsule which causes the disease and hence called virulent S stain. A mutant strain has no capsule and is avirulent or nonpathogenic and is called R strain. In agar medium, the virulent (S) strain produces smooth surfaced colonies, while the avirulent (R) strain produces rough surfaced colonies. There are several types of these two strains, S I, S II, S III, R I, R II, R III *etc.* that differ in the type of antigen they produce. The kind of antigen produced is genetically determined. The S type sometimes mutates to R type but not in the reverse.

Griffith injected the laboratory mice with live R II bacteria and the mice did not get pneumonia as R II is avirulent. When injected with virulent S III, the mice suffered of pneumonia and died. When S III bacteria were heat killed at $65^{O}C$ and then injected into the mice, they did not suffer of the disease and lived. Later, the heat killed S III strain and the live avirulent RII strain were mixed and injected into the mice. Contrary to expectations, the mice suffered of pneumonia and died. On analyzing the blood sample of the affected mice, live S III and live R II bacteria were found in it. This could not be possible due to the mutation of the avirulent R II to virulent types. Evidently, some heat-stable component present in the heat killed, and hence, dead S III strain could have conferred the virulent nature to the live R II strain. Griffith designated this as the 'transforming principle' that transformed the hereditary property of avirulent R II to virulent S III. This phenomenon is called 'Griffith effect' or 'Bacterial transformation'.

Griffith did not understand the cause of bacterial transformation. Avery, MacLeod and McCarty (1944) tested a fraction of the heat killed S III bacteria for the transforming ability. They removed proteins, lipids, polysaccharides and ribonucleic acid from S III extract by a variety of chemical and enzymatic methods without diminishing its ability to transfer R II into S III strain. They found that a cell-free and highly purified DNA extract

Reference: DDS: 268-271

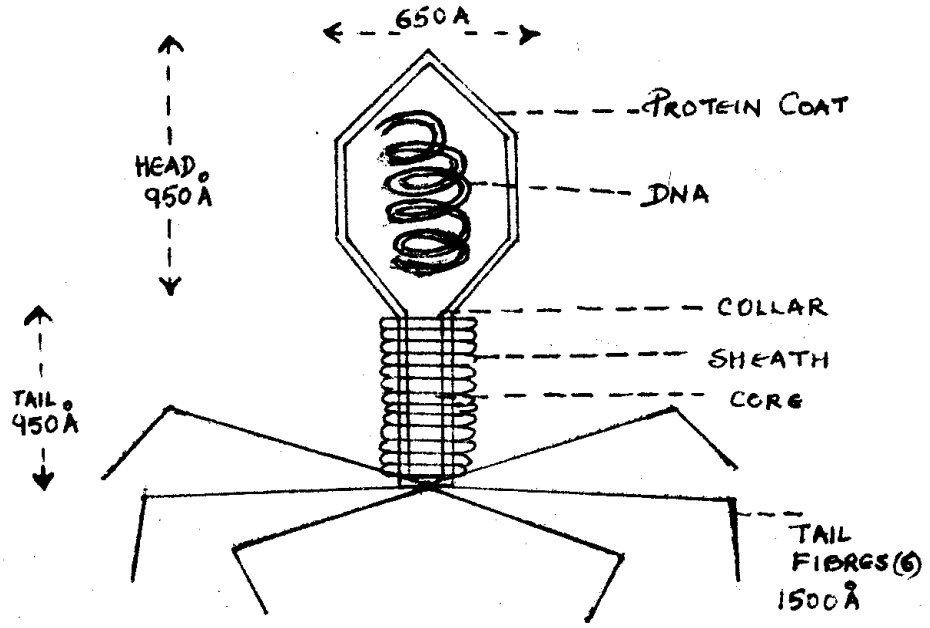
of S III bacteria could bring about transformation of R II into S III and concluded that DNA is the transforming principle and hence the genetic material in bacteria.

Later studies on other bacteria such as *Haemophilus influenzae*, *Bacillus subtilis*, *Escherichia coli* and others revealed that they also undergo transformation.

Transformation is the process of adding a foreign DNA fragment from a donor genome into the genome of a recipient cell. The donor fragment passes through the cell membrane of the recipient cell of the same or different species and becomes incorporated into the genome of the recipient cell through recombination.

DNA as the genetic material in viruses

Hershey and Chase (1952) provided direct proof that DNA is the genetic material in certain bacterial viruses.



Structure of Bacteriophage

Bacteriophage is a virus that infects or feeds on certain specific bacteria. T_2 bacteriophage that infects the colon bacteria, *Escherichia coli* was involved in the studies.

Bacteriophage is electron microscopic. It has a head and a tail. Inside the head, there is a long chain of DNA molecule. The phage attaches itself by its tail to the bacteria and injects the DNA into the bacillus. It dictates the cell to produce many copies of the viral DNA.

Bacteriophages are used in many finer analyses of the genetic material since they are haploid organisms and there is no hiding of mutant effect. As there is no differential sex, there is no need for two different eindividual to unite for reproduction. They multiply enormously and have a short life span. Recombinations and mutations, even if in a very low frequency, could be recognized with relative ease. When a population is raised from a single phage all the descendents will be identical. But occasionally, through errors in copying of genetic material, rare mutants appear and such mutants are called 'copy errors'.

In a chemically defined cultural medium, known quantities of radioactive isotopes of phosphorus P^{32} and sulphur S^{35} were added. *Escherichia coli* were grown in the medium and the labeled *E.coli* cells were used as hosts for unlabelled T_2 bacteriophage. The virus progeny that multiplied inside the bacteria could be traced in the culture medium on lysis (cell wall breakage) of the bacteria.

The viral DNA was labeled with P^{32} and the viral capsid (protein coat) with S^{35}, since DNA contained P and the capsid protein contained S. then the labeled viruses were allowed to infect unlabelled *E. coli* and get multiplied. Later the viruses were separated from the bacterial host cell by agitation and the content of P^{32} and S^{35} of the virus and bacteria was assessed. P^{32} could be traced in the infected bacterial cells. Hershey and Chase inferred that DNA of the virus entered the bacterium and played a role in viral multiplication, whereas the protien of the virus did not play any role in the intercellular replication of the virus. Thus it was established that the genetic material of the virus was DNA.

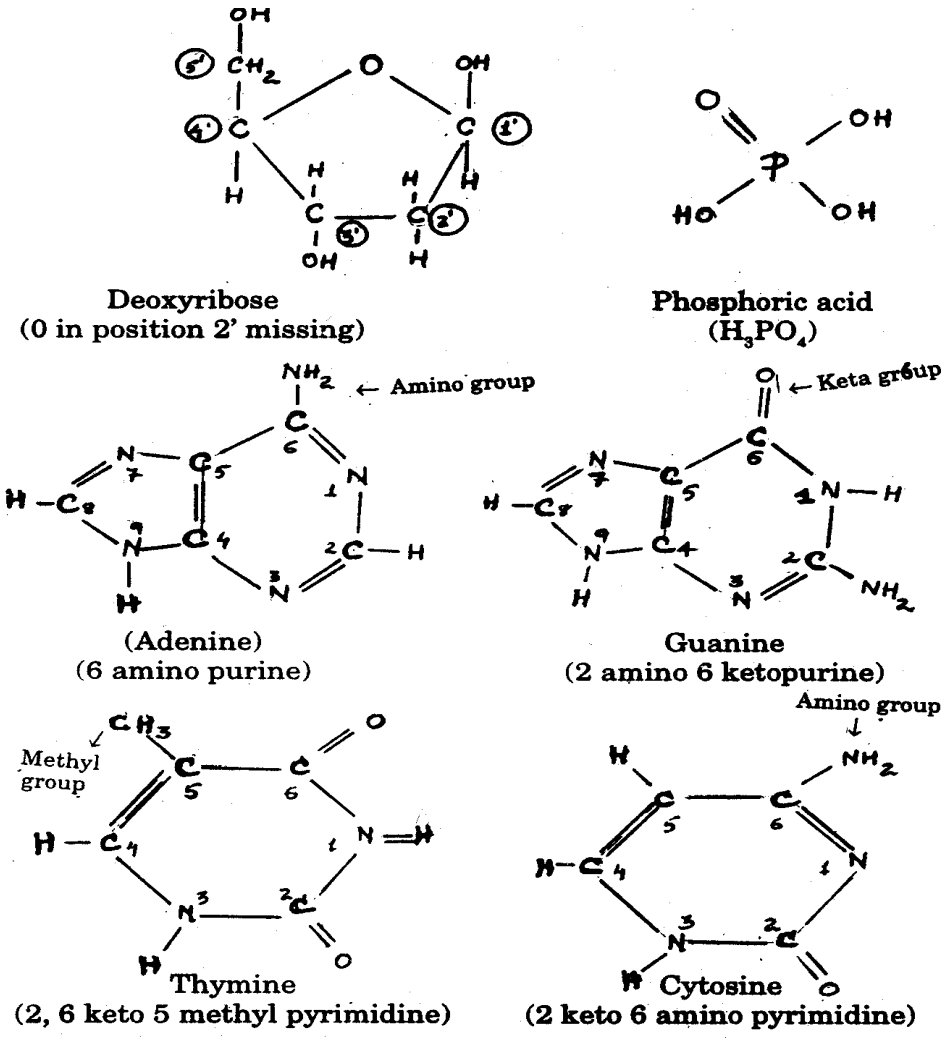
Lecture XIX

Structure of DNA – Watson and Crick model – Semi conservative model of DNA replication, Central dogma.

Chemical composition of DNA

DNA is a complex macromolecular or polymeric chemical compound which contains four kinds of monomers (small building blocks) called Deoxyribonucleotides. Each deoxyribonucleotide is made up of 1) a phosphoric acid molecule, biologically called phosphate, discovered by Levene (1910), 2) a pentose sugar called 2-deoxyribose, also discovered by Levene (1910) and 3) four major kinds of nitrogen bases, two heterocyclic and two ringed purines, adenine (A) and guanine (G) and two one ringed pyrimidines, cytosine (C) and thymine (T), discovered by Fischer (1880).

The structure and chemical formulae of the different units are as follows:



Structure and chemical formulae of the components of DNA.

Reference: DDS: 273-277

A nucleotide results from covalent boding of a phosphate and a nitrogen base to the pentose. That part of each nucleotide which contains a nitrogen base and deoxyribose is called Deoxyribonucloside.

The four kinds of deoxyribonuclosides and deoxyribonucleotides are as follows:

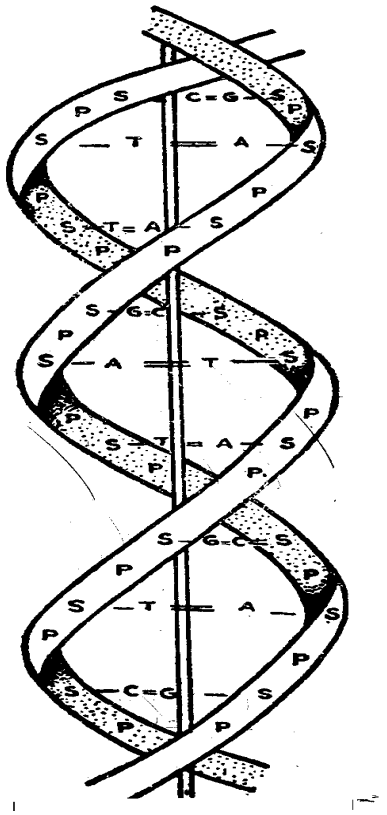
Nucleosides and nucleotides of DNA

Nitorgen base	N base + Deoxyribose = Deoxyribonucleoside	Deoxyribo nucleoside + Phosphoric acid = Deoxyribo nucleotide	Nucleotde
Adenine	Deoxyadenosine	Deoxyadenylic acid	Deoxyadenosine mono – phosphate (damp)
Guanine	Deoxyguanosine	Deoxyguanylic acid	Deoxyguanosine mono phosphate (dGMP)
Thymine	Deoxythymidine	Deoxythymidylic acid	Deoxythymidine mono phosphate (dTMP)
Cytosine	Deoxycytidine	Deoxycytidylic acid	Deoxycytidine mono phosphate (dCMP)

Double helical model of DNA

Based on the findings of Chargaff (1950) that the total amount of purines equalled the total amount of pyrimidines (A + G = T + C), that the amount of adenine equalled the amount of thymine (A = T) and the amount of guanine equaled the amount of cytosine (G = C) and , that the ratio between total purines and total pyrimidines was always not far from one, (A + G) : (T + C) = 1, as well as the crystallographic evidences and X-ray differentiation photographs (Astbury, 1947, Wilkins and Franklin, 1953), the double helical model of DNA was constructed by Watson, an American biologist and Crick, a British physicist in 1953.

The DNA molecule was conceived as a two stranded structure coiled like a rope, and hence called plectonemic, so that if the ends are permitted to revolve freely, the complementary strands could easily separate. The coil was proposed to be helical and conceived to resemble a circular staircase, maintaining the same diameter through out and having a constant width between steps. The steps are connected on either side by a railing.



Watson – Crock model of the DNA molecule

A- adenine G – guanine C – cytosine T – thymine S – sugar P – phosphate

The helix has a diameter of $20A^{O}$ and makes a complete turn at every $34A^{O}$ along its length. The distance between nucleotides is $3.4A^{O}$. Each complete turn has a stack of 10 nucleotides. The helix contains two polynucleotide chains or two stacks of 10 nucleotides each per turn.

Each complementary strand is only half the circular staircase, either side consisting of approximately half the width of the step. Each half step is connected by a railing or backbone. The railing consists of phosphate – sugar linkages which are repeated without change. The half step of one strand extends to meet the half step of the complementary strand. Each half step has either a purine or pyrimidine base. Each step consisting of two half steps is together called base pair.

The fit between the bases is determined by hydrogen bonding. The bonding involves the ability of the H atom with positive charge (H+) to be placed between an O atom with weak negative charge (O-) and a N atom with a light negative charge (N-) from opposite strands. Adenine pairs with thymine with two H bonds (A = T) and guanine with cytosine with three H bonds (G ≡ C). These N bases are connected to each other by deoxyribose and phosphoric acid.

Hydrogen bonds are generally weaker than other chemical bonds. But there are several of them, two between A and T (A = T) and three between G and C (G ≡ C) that give rigidity and stability to the molecules.

Lecture XX, XXI

Gene expression – Protein synthesis – Transcription – role of mRNA, tRNA, rRNA. Genetic code – Translation – formation of polypeptide chain

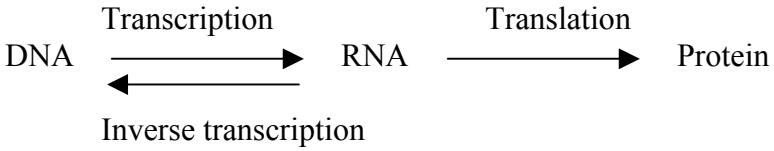
Protein Synthesis
Central Dogma of Molecular biology

The process of protein synthesis involves one of the central dogma of molecular biology, postulated by Crick (1958) according to which genetic information flows nucleic acid to protein.

Protein synthesis involves two steps viz., transcription and translation. Transcription involves a sequential flow of information from DNA to mRNA. This doesnot involve a change of code since DNA and mRNA are complementary. Translation involves a change of code from nucleotide sequences to amino cid sequences.

Generally the flow of information is one way, from DNA to RNA and from RNA to protein.

In certain viruses, the existence of an enzyme 'RNA dependent DNA polymerase' (also called inverse transcriptase) was reported and this enzyme could synthesize DNA from a single stranded RNA template. This finding of Baltimore (1970) and others give rise to the concept of 'central dogma reverse'. According to this, the sequence of information flow is not necessarily from DNA to RNA to protein, but can also take place from RNA to DNA.



Transcription

The process by which the information in the nucleotide sequence of DNA is transferred to a complementary sequence of RNA is known as transcription.

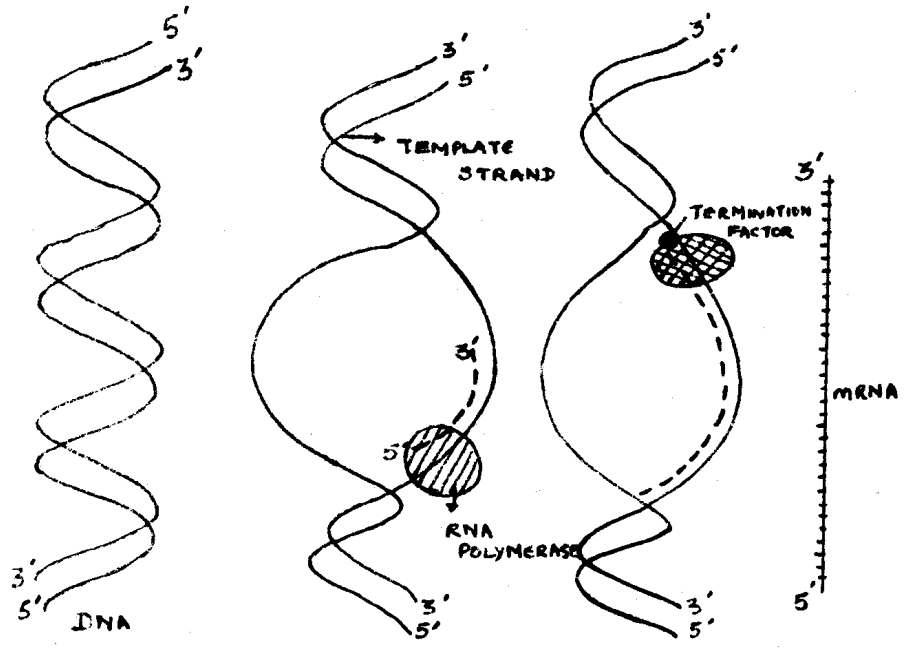
Transcription occurs throughout interphase and continues up to early prophase of cell division. 'DNA dependent RNA polymerase' or 'transcriptase' is the enzyme involved in transcription. The locations of transcription are 1) the nucleolus where genes rRNA are transcribed and 2) the remaining chromatin where mRNA (mRNA) is transcribed.

The system for in vitro RNA synthesis contains 1) ribonucleotide triphosphates (ATP, CTP, GTP and UTP), 2) enzyme RNA polymerase, 3) MG^{++} or Mn^{++} and 4) template DNA. The enzyme links the ribonucleotides together by catalyzing the formation of 3' – 5' phospho diester bonds that pin the nucleotides. Consequently, RNA is synthesized and pyrophosphate is released. The enzyme RNA polymerase acts only in the presence of DNA, against which the correct sequence of ribonucleotides is arranged and they are linked together by the enzyme. That is why the enzyme is known as 'RNA – dependent RNA polymerase'.

$$\begin{matrix} n : rA - P \sim P \sim P\ (ATP) \\ n : rU - P \sim P \sim P\ (UTP) \\ n : rG - P \sim P \sim P\ (GTP) \\ n : rC - P \sim P \sim P\ (CTP) \end{matrix} + \text{Template DNA} \xrightarrow[Mg^{++} \text{ or } Mn^{++}]{\text{RNA Polymerase}}$$

$$\text{Template DNA} + n \left\{ \begin{matrix} r\,A - P \\ r\,U - P \\ r\,G - P \\ r\,C - P \end{matrix} \right. + 4n\ P \sim P$$

The site of transcription in a cistron is called the promotor site. The template strand is called sense strand, while its complementary strand is known as antisense strand. When only one strand of DNA is transcribed for a given region, it is called asymmetrical transcription. When both the strands of the DNA are transcribed, it is known as symmetrical transcription.



Stages in the process of transcription

The details of the transcription process are the following:

The enzyme RNA polymerase attaches itself at the promoter site. The DNA molecule unwinds over a short region. Then the free bases in the template strand of DNA determine the sequence of ribonucleotides in the newly formed mRNA.

The RNA polymerase enzyme joins the nucleotides together to produce RNA transcript. After the transcript become detached the DNA template strand re-forms H-bonds with its complementary strand and rewinds to form the double helix.

RNA polymerase enzyme has five subunits of polypeptide chains viz., α, β, β^1, σ and ω with molecular weight ranging from 10,000 to 160,000. α, β, β^1 and ω subunits form the core of the enzyme and catalyse the linkage of ribose nucleotides by phosphodiester bonds. The σ – factor recognises the start signal in the promotor region of DNA.

Translation

As soon as the mRNA is formed, it leaves the nucleus and reaches the cytoplasm where translation takes place.

Before the process of protein synthesis, the ribosomes occur in dissociated and inactive state. The mRNA binds with 30 S ribosomal subunit in the presence of a protein factor called Initiation Factor (IF). The mRNA carries triplet codons for the synthesis of proteins. Protein synthesis involves mRNA, ribosomes, amino acids and their specific tRNAs.

Translation process involves the following steps:

1. Attachment of mRNA with 30 S ribosomes and formation of polyribosomes

The mRNA transcribed inside the nucleus moves to the cytoplasm and binds itself with 30 S subunit of the ribosome in the presence of Initiation Factor. Then the tRNA present in the cytoplasm bind itself with the first triplet codon 5' – AUG – 3' called the chain initiation codon of mRNA to form the 'Initiation Complex'. Later, the 30 S subunit of ribosome unites with 50 S subunit to form 70 S ribosome, in the presence of Mg ++ ions. The message in the mRNA is not deciphered by one ribosome but many ribosomes are involved in the process and hence they are called polyribosomes.

2. Activation of the amino acids

Amino acids present in the cytoplasm are in a dormant stage. Each amino acid is activated by an activating enzyme called aminoacyl synthetase, beside the energy rich adenosine triphosphate (ATP). The free amino acids react with ATP to produce aminoacyl and pyrophosphate (PP).

Amino acid
+
ATP
+ ⟶ Amino acid ~ AMP – Enzyme (aminoacyl adenylate – enzyme complex) + PP

Amino acyl
Synthetase
enzyme

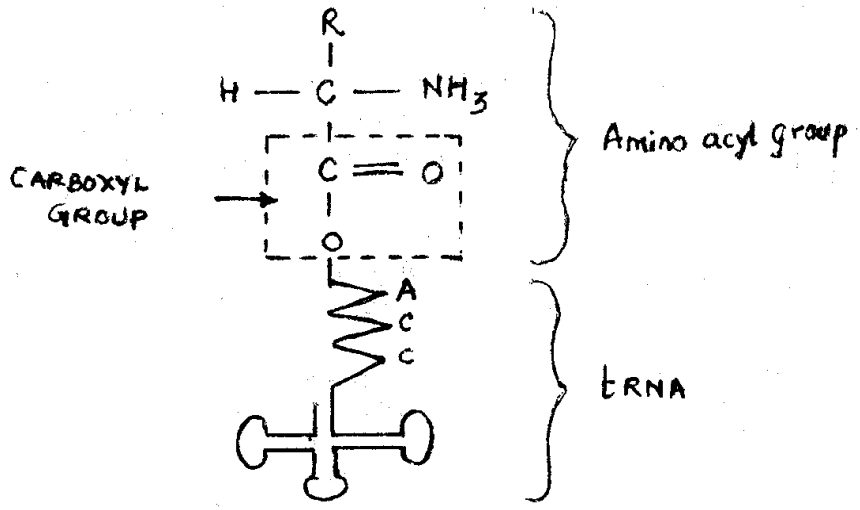
The aminoacyl adenylate enzyme complex bound together by a mono covalent bond attaches itself with the specific tRNA molecule. As the enzyme is specific for specific amino acid, the concerned amino acid gets attached without error.

3. Attachment of activated aamino acid to tRNA

The aminoacyl adenylate remains bound with the enzyme till it is hooked to the tRNA molecule. The dihydrouridine (DHU) loop of tRNA recognizes the synthetase enzyme. Then the amino acid residue of the aminoacyl adenylate is transferred to the amino acid attachment site of tRNA, where it carboxyl group forms linkage with 3 – OH group of the ribose of the terminal adenosine at – CCA end of tRNA.

As a result, adenosine monophosphate (AMP) and the enzyme are released and aminoacyl tRNA is formed. Then the aminoacyl tRNA moves towards the ribosome.

AA ~ AMP – Enzyme + tRNA AA – tRNA + AMP + Enzyme



4. Initiation of the polypeptide chain

In the mRNA, the first triplet codon is AUG at its 5' end. AUG codes for methionine. Hence protein synthesis commences with coding for methionine. The peptide chain formation starts in 5' end and proceeds towards 3' end and this helps in the correct sequence of protein synthesis.

The mRNA moves across the ribosome. A new codon of mRNA is brought in position. A new tRNA charged with specific amino acid is brought in position in such a way that the anticodon of tRNA pairs with the codon of mRNA. The attachment of two

amino acids by polypeptide linkage involves enzymes translocase and peptidyl transferase along with energy rich GTP, and tRNA is released.

This process of movement of mRNA from 5' to 3' direction and addition of aminoacids to polypeptide chain continues till mRNA is no longer translated.

5. Termination of the polypeptide chain

Any one of the three terminating codons in mRNA, viz., UAA, UAG or UGA can signal the termination of chain elongation.

After chain termination, the enzyme peptidyl transferase hydrolyses the ester bond between the chain and tRNA releasing the polypeptide chain, the last tRNA and mRNA.

Thus a polypeptide chain with a specific series of amino acids is formed which results in synthesis of a specific protein that involves in a specific phenotypic expression in the organism.

Genetic Code

In the DNA and RNA, there are four types of nucleotides or bases viz., A, G, T, C and A, G, U, C respectively. If it is assumed that each base codes for one amino acid, then only four amino acids can be coded. If two bases together are responsible for production of one amino acid, then they will code for 42 = 16 amino acids. If three bases together code for an amino acid, then 43 = 64 amino acids could be coded. As the essential amino acids in a biological system are 20 in number, the possibility of one or two bases coding for each amino acid is remote.

Based on studies conducted on B cistron in r" region of T4 bacteriphage having several positive mutants (involving insertion of a nucleotide), negative mutants (involving deletion of anucleotide) and double mutants, Crick and Brunner (1961) suggested that the genetic code might be a triplet code, involving three nucleotide bases to code for an amino acid.

Further investigations by Nirenberg and Mathhaei (1961), Nirenberg (1961), Khorana (1964) and others lead to the construction of a complete genetic code dictionary.

The pattern of genetic code indicates the following:

1. Codons for the aromatic amino acids begin with Uracil

UUU, UUC } Phenyl alanine (Phe)

UAU } Tyrosine (Tyr)

UAC

UGG - Tryptophan (Trp)

2. Codons for amino acids that form amides being with Guanine and Adenine.

GAU, GAU } Asparagin (Asp)

GAA, GAG } Glutamin (Glu)

3. For many of the synonymous codons specifying the same amino acid, the first two bases of the triplet code are constant while the third varies, being less specific.

GCU, GCC, GCA, GCG } Alanine (Ala)

GUU, GUC, GUA, GUG } Valine (Val)

GGU, GGC, GGA, GGG } Glycine (Gly

CUU, CUC, CUA, CUG } Leucine (Leu)

CCU, CCC, CCA, CCG } Proline (Pro)

CGU, CGC, CGA, CGG } Arginine (Arg)

ACU, ACC, ACA, ACG } Threonine (Thr)

UGU, UCC, UCA, UCG } Serine (Ser)

Lecture XXII

Regulation of gene expression – Operon model of Jacob and Monad – Structural genes and Regulator genes

Gene expression refers to manifestation of a phenotypic character by the activity of gene. A gene expresses itself by producing proteins or enzymes. During gene expression, there is flow of genetic information from DNA to protein.

Gene expression involves two steps – transcription and translation. Each adult cell contain full set of genes necessary for the development of an adult animal. However in any cell all the genes will not function to synthesis proteins. Only one or a few genes will be functioning in any adult. The other genes will not function to synthesis proteins. The genes which are not functioning are said to be switched off and the genes which are functioning are said to be switched on. In the cells of islets of langerhans, the genes responsible for the synthesis of insulin are switched on and all other genes are switched off.

Regulation of Gene Expression in Prokaryotes

It is well studied in the bacterium, *Escherichia coli*. *Ta E.coli* and other prokaryotes the gene expression is regulated at two levels. They are i) Regulation of enzyme ii) Regulation of Transcription.

I. Regulation of Enzyme

In *E.coli* certain enzymes are continuously produced regardless of their needs. These are called constitutive enzyme and need no gene regulation. (eg.) enzymes of glycolysis.

Certain other enzymes are synthesized only when the are needed. Such enzymes are called regulated enzymes. They require gene regulation (eg.): Enzymes of lactose metabolism.

The regulation of enzymes is brought about by the following mechanisms.

1. Enzyme induction
2. Enzyme repression
3. Feed back inhibition

1. Enzyme induction

When a substrate is present in the medium, an enzyme is produced by the bacterium to metabolise the substrate. This phenomenon is called as enzyme induction. The substrate which is responsible for the production of enzyme is called inducer and enzyme is called as inducible enzyme. The substrate, inducible enzyme and genes involved in it constitute an inducible system.

The Lactose metabolism in E.coli is an inducible system, when lactose is added to a medium, the enzymes β - galactosidase, β - galactoside permease and thiogalactoside transacetylase are synthesized simultaneously. These three enzymes are involved in the metabolism of lactose. Lactose thus acts as the induces.

2. Enzyme Repression

Certain enzymes are normally synthesized and are present at all time in cell. When the end product is produced in large amount, the enzyme synthesis is stopped. This phenomenon is as gene repression. The enzyme, the synthesis of which is inhibited is called repressible enzyme and end product which inhibits the synthesis of the enzyme is called as co-repressor. The repressible enzyme, the co-repressor and the genes involved constitute repressible system.

This phenomenon of on and off mechanism of genes is called regulation of gene expression.

The regulation of gene expression help the prokaryotes to adjust with environmental changes for (eg.): Changes in the culture medium.

In eukaryotes, the regulation of gene expression facilitates differentiation and also help the animals to respond to hormonal and nervous stimulation.

Histidine pathway is an repressible system.

3. Feed back inhibition.

In feed back inhibition the enzyme activity is inhibited by the end product when it is produced in excess. It is also called as end product inhibition. Here the enzyme synthesis is not inhibited but the enzyme activity is inhibited by the end product.

In *E.coli* isoleucine is synthesis from threonine. This is threonine- isoleucine pathway. When isoleucine is needed for the cell, the threonine- isoleucine pathway continues. If isoleucine is produced in excess, the excess and product inhibits the threonine isoleucine pathway.

II. Regulation of Transcription.

Transcription refers to the synthesis of mRNA from DNA. It is a stage in gene expression. In prokaryotes, the genes are regulated at the level of transcription.

1. Negative regulation, 2. Protein regulation, 3. Auto regulation, 4. Coordinate regulation.

1. Negative regulation: -

In negative regulation, the blocked gene is set free for transcription. An inhibitor present in the cell prevent transcription. The inhibitor is a regulator protein called repressor. The repressor binds with the gene and prevents transcription. The transcription is initiated by another protein called inducer. The inducer binds with the

inhibitor and makes the gene free to initiate transcription. In *E.coli* the synthesis of enzyme proteins in the presence of lactose by the lac operon is due to negative regulation.

In a biosynthesis pathway (anabolism) the end product usually regulates its own synthesis. In the simplest type of negative regulation, absence of the end product usually increases its synthesis and presence of the product decreases its synthesis.

2. Positive regulation: In positive regulation, an effectors molecule (inducer) activates a gene and transcription is initiated. The activator molecule attaches to the initiator site on the gene and transcription is initiated. An inhibitor doesnot inhibit positive regulation.

3. Auto regulation: The protein synthesized by a gene directly inhibits the transcription by the same gene when the protein is produced in high concentration. This protein is translated from a monocistronic mRNA.

4. Coordinate regulation: When several enzymes act in sequences in a single metabolic pathway, the regulation is brought about by the inhibition of synthesis of either all or none of these enzymes. Inhibition of synthesis of all enzymes results from the control of the synthesis of a single polycistronic mRNA molecule encoding all the enzymes. This type of regulation doesnot occur in eukayroties because eukaryotic mRNA is usually mono cistronic.

Operon Hypothesis

Operon is a set of closely linked genes regulating a metabolic pathway in prokaryotes.

The operon hypothesis was put forward by Jacob and Monod in 1961 Nobel prize in 1965.

A bacterium contains thousands of genes. When all the genes are functioning at the same time, the cell will be flooded with enzymes and proteins. This on and off mechanisms was explained by the operon model.

Lac operon

1. Lac operon is a set of genes responsible for the metabolism of lactose in E.Coli. Jacob and Monad in 1961.
2. The lac operon consists of 3 structural genes namely Z, y and a and 3 control genes promoter gene (P), a regulator gene (I) and operator gene (O).
3. The structural genes are responsible for the synthesis of three enzymes namely β – galactosidase by the gene Z, galactoside permease by the gene y, thiogalactoside transacetylase by the gene a.

4. The operator gene is closely linked to the first structural gene Z. when the operator gene is active, the structural genes synthesis enzymes.
5. The activity of the operator gene is decided by a repressor protein synthesized by a regulator protein.
6. When the repressor binds to the operator gene, the operator gene is made nonfunctional. This state of the operator gene is called repressed state and the phenomenon is called repression. During repression, the enzymes are not synthesized.
7. When lactose is introduced into the medium, it diffuse into the cell and binds to the repressor protein to form an in active inducer repressor complex.
8. The in active inducer repressor complex – cannot bind to the operator gene and the operator gene is set free to do the function. This state of the operator gene is called as derepressed state and the phenomenon is called as derepression.
9. When the operator gene is redepressed, the RNA polymerase binds to the promoter gene. It initiates the transcription of structural genes.
10. The transcription of structural genes leads to the synthesis of 3 enzymes namely β – galactoside, galacetose permease and thiogalactoside transcetylase.
11. These three enzymes bring about the metabolism of lactose. B-galactosidase splits lactose unite in to glucose and galactose. Galactoside permease facilitates the entry of lactose into the cell. The function of galactoside transcetylase ins noted known.
12. In the lac operon system, lactose function as an inducer for the synthesis of three enzymes. Hence the lac operon system is called in inducible system.
13. The lac operon is a system of negative regulation. In negative regulation the regulator protein repressor prevents gene transcription.
14. cAMP- CAP* complex is functioning as a regulatory element in lac operon. The cAMP-CAP binds to a base sequence in the DNA of the promoter gene to initiated transcription. Thus the cAMP- CAP complex acts as a positive regulator. Thus lac operon acts independently both positively and negatively. The repressor protein acts as a negative regulator and cAMP- CAP protein acts as a positive regulator.

Lecture XXIII

Split genes, eons and introns – Modern concept of gene – Gene as Cistron, Muton and Recon, Complementation test.

Fine Structure of Gene

The hereditary units which are transmitted from one generation to the other generation are called as genes. A gene is the fundamental biological writ unlike the atom which is the fundamental physical unit. Mendel while explaining the result of his monohybrid and dihybrid crosses, first of all conceived of the genes as particulate units and referred them by various names such as hereditary factors or hereditary elements. But his concept about the gene was entirely hypothetical and he remained ignorant about the physical and chemical nature of the gene.

Even before the rediscovery of Mendel's law in 1900, it was already established that chromosomes have a definite role in the inheritance because it was found that chromosomes were the only link between one generation and the next generation and a diploid chromosome set consists of two morphologically similar sets one is derived form the mother and the other from the father at fertilization. Later on, a parallel behaviour among chromosomes and genes were discovered.

Earlier workers proposed various hypotheses to explain the nature of genes. For instance, De Vries postulated 'one gene one character" hypothesis according to which a particular trait of an individual is controlled by a particular gene. Bateson and Punnett proposed the presence or absence theory. According to them, in a cross, the character which dominates the other has a determiner while the recessive character has no such determiner. But all these theories were discarded by Morgan who proposed the particulate gene theory in 1926. He considered genes as corpuscles which are arranged in a linear order on the chromosomes and appear like beads on a string. Each gene was supposed to be different from all others. The particulate theory of gene was widely accepted and supported by cytological observations. The discovery of DNA molecule, as a carrier of genetic informations has altogether discarded the Morgan's theory.

CLASSICAL DEFINITION OF GENE

A gene is a unit of physiological function that occupies a definite locus in the chromosome and is responsible for a specific phenotypic character. (eg.) vestigial or long wings and white and yellow eyes in *Drosophila*.

A gene is a unit of transmission or segregation because it can be segregated and exchanged at meiosis by way of crossing over.

A gene is a unit of mutation because by a spontaneous or induced change it can give rise to different phenotypic expression.

MODERN DEFINITION OF GENE

After the discovery of DNA, its parallel behaviour with that of chromosomes and proper understanding of most of the molecular phenomena which may interplay in the determination of a phenotypic trait, the gene has been defined as follows:

1. Cistron

The portion of DNA specifying a single polypeptide chain, synonym for the tern gene of physiological function. Cistron was coined by Seymour Benzer. Haemoglobin therefore would require two cistrons fraction globin protein fraction one each for α and β chains. A cistron for α chain has atleast 141 x 3 = 423 nucleotides and the cistron for the β chain 146 x 3 = 438 nucleotides.

2. Muton

There are many positions or sites within a cistron where mutations can occur. Therefore gene as a unit of mutation is smaller, it consists of fewer nucleotides than a cistron. Benzer coined the word muton to that smallest length of DNA capable of mutational change. Different formes of a mutationality defined genes are called as homoalleles.

3. Recon

Sometimes crossing over or recombination occurs in a cistron and this provides still other subdivisional concept of the cistron namely the recon. A recon is the smallest unit of DNA capable of recombination or of being integrated by transformation in bacteria. Recombinationally separable forms of a cistron are called heteroalleles.

"The gene of function is the sequence of nucleotides which specifies the amino acid sequence of a specific polypeptide chain.

SPLIT GENES

There are some genes which are different from normal genes either in terms of their nucleotided sequences or fractions.

Usually a gene has a continuous sequence of nucleotides. Other words, there is no interruption in the nucleotide sequence of a gene. Such nucleotide sequence codes for a particular single polypytede chain. It was observed that the sequence of nucleotides was not continues in case of some genes, the sequence of nucleotides were interrupted by intervening sequences. Such genes with interrupted sequences of nucleotides are referred as split genes or interrupted genes.

Split genes have two types of sequences. Viz. normal sequences and interrupted sequences.

1. Normal sequence: This represents the sequence of nucleotides which are included in the mRNA which is translated from DNA of split gene. These sequences code for a particular polypeptide chain and are known as exons.

2. Interrupted sequences: These are called as introns. These donot code for any peptide chain. Interrupted sequences are not included into mRNA which is transcribed from DNA of split genes. The interrupted sequences are removed from the mRNA during processing of the same. The intervening sequences are discarded in mRNA as they are non coding sequences. The coding sequences or exons are joined by ligase enzymes.

The first case of split gene was reported for ovalbumin gene of chickens. The ovalbumin gene has been reported to consist of seven intervening sequences. Later as it was reported for beta globin genes of mice and rabbits, tRNA genes of yeast and ribosomal genes of Drosophila.

The intervening sequences are determined with the help of R loop techniques. This techniques consists of hybridization between mRMA and DNA of the same gene under ideal condition (ie) at high temperature and high concentration of formide. The mRNA pairs with single strand of DNA. The interfering sequences of DNA make loop in such pairing. The number of loops indicate the number of interrupted sequences and size of loop indicates length of the intervening sequence. These loops can be incurred under electron microscope. The ovalbumin gene has seven interrupted sequences and eight coding sequence (exons).

The intervening sequences are excised during processing to form mature mRNA molecule. Half of the ovalbumin gene is discarded during processing. Split gene have mostly reported in eukaryotes.

Complementation test

If two homologues chromosomes carry mutations with different genes – let one chromosome is mutation in gene p and other in gene Q – and the two chromosomes come to reside in the same cell, one chromosome cell direct the synthesis of a mutant P gene product but a normal Q product and the other will direct the synthesis of normal P but a mutant Q product. That is the cell will come to contain both normal P and normal Q products and thus normal function can occur. In such a case, two mutant chromosomes are said to be complement each other, and it is inferred that the two mutations lie within the boundaries of two different genes. In contrast tow chromosomes carrying mutation in the same gene say P, than neither chromosome will be able to direct the synthesis of a normal P product and even when both chromosomes come to reside in the same cell normal P function cell not be observed. The two mutant chromosomes in this case do not complement each other and it can be inferred that both mutations lie within the boundaries of the same gene.

Complementation: The Production of a normal offspring from a mating between two homozygous affected by similar defect showing that the parentals actually suffered from different genetic lesions.

Complementation test: The introduction of two independently occurred mutation into the same cell for the purpose of determining whether the mutation occurred in the same gene. This test can be accomplished with by mating two homozygous organisms or by somatic cell fusion.

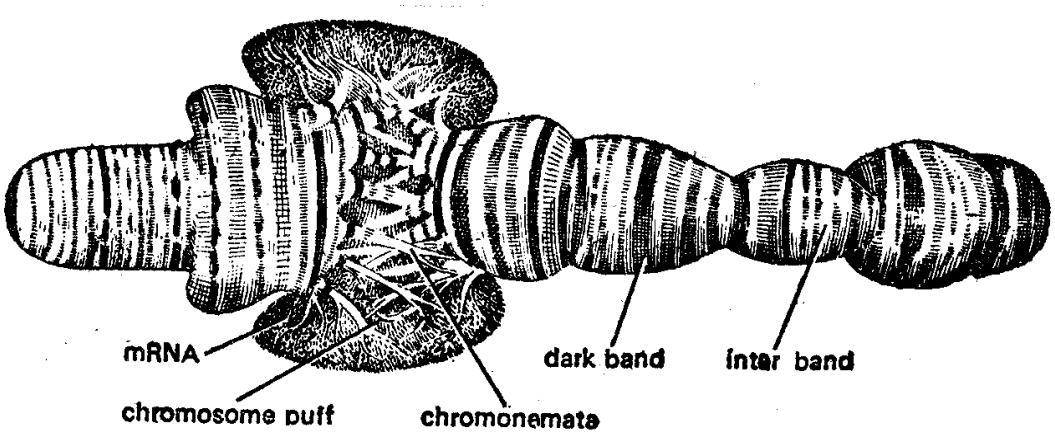
Lecture XXIV

Special chromosomes – Polytene, Lampbrush, B and Iso Chromosomes

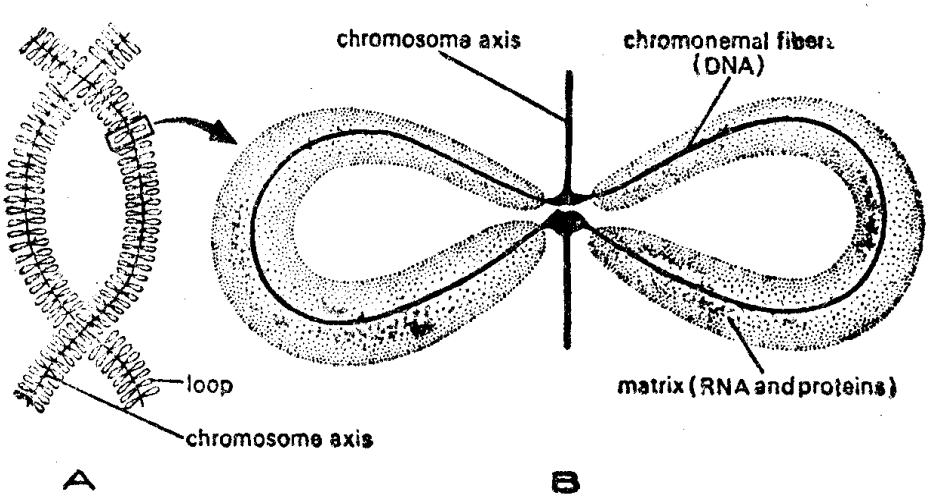
Special Types of Chromosomes

1. Polytene chromosomes – The nuclei of the salivary gland cells of the larvae of *Drosophila* have unusually long and wide chromosomes, 100 or 200 times in size of the chromosomes. Since the salivary gland cells do not divide after the glands are formed, yet their chromosomes replicate several times (a process called endomitosis) and become exceptionally giant – sized to be called polytene chromosomes. The polytene chromosomes of the salivary gland cells of *D. melanogaster* contain 1000 to 2000 chromosomes, which are formed by nine or ten consecutive multiplication cycles and remain associated parallel to each other. Further, the polytene chromosomes have alternating dark and light bands along their length. The dark bands are comparable with the chromomeres of a simple chromosomes and are disc-shaped structures occupying the whole diameter of chromosome. They contain euchromatin. The light bands or inter bands are fibrillar and composed of heterochromatin.

If the polytene chromosomes of dipteran larval salivary glands are examined at several stages of development; it is seen that specific area (sets of bands) enlarge or "puff". Such puffs change location as development proceed, those at specific locations being correlated with particular developmental stages.

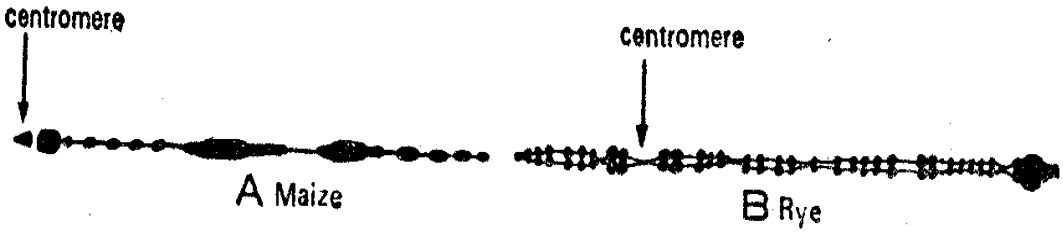


2. Lampbrush Chromosomes – In diplotene stage of meiosis, the yolk-rich oocyte of vertebrates contain the nuclei with many lampbrush-shaped chromosomes of exceptionally large size. The lampbrush chromosomes (discovered by Rucker in 1892) are formed during the active synthesis of mRNA molecules for the future use by the egg during cleavage when no synthesis of mRNA molecules is possible due to active involvement of chromosomes in the mitotic cell division. A lampbrush chromosome contains a main axis whose chromonemal fibres (DNA molecules) gives out lateral loops throughout its length. The loops produce the mRNA molecules of different kinds.



A- At low magnification, B- A loop magnified (after Robertis, *et al.,* 1970).

3. B-Chromosomes – Many plant (maize) and animal (insects and small mammals) species, besides having autosomes (A-chromosomes) and sex-chromosomes posses a special category of chromosomes called B-chromosomes without obvious genetic function. These B-chromosomes usually have a normal structure, are somewhat smaller than the autosomes.

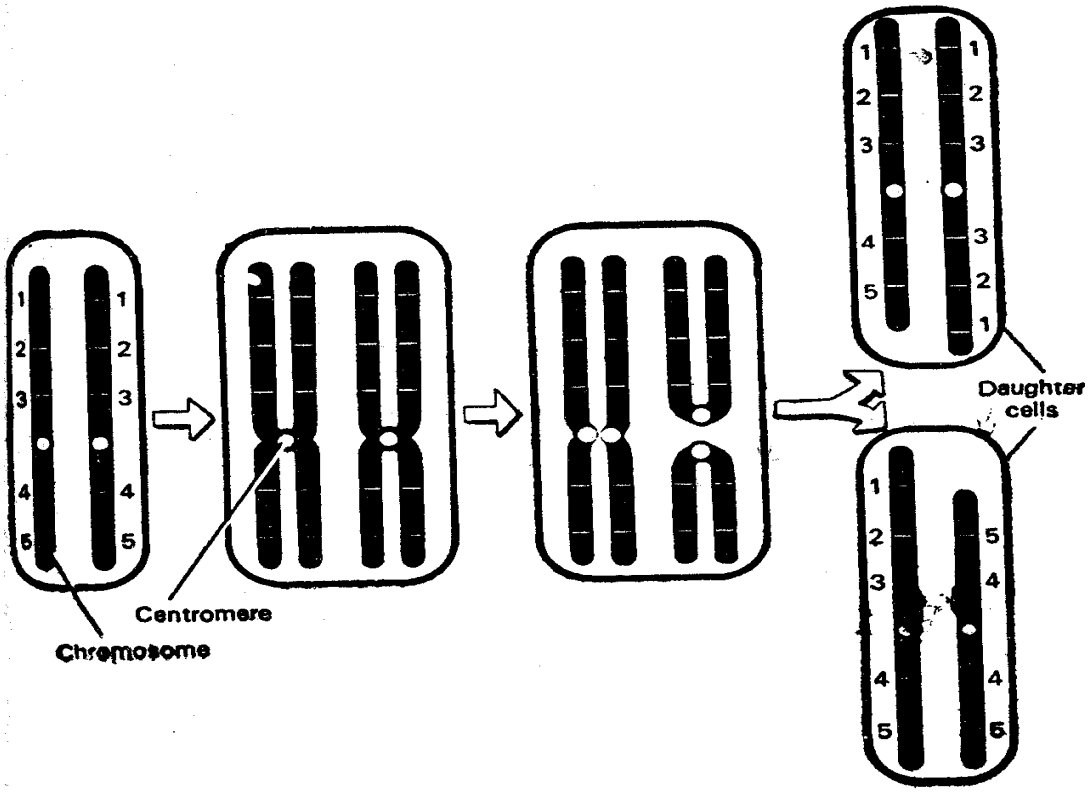


The origin of the B-chromosomes is uncertain. In some animals they may be derivatives of sex chromosomes.

4. ISO Chromosomes- An isochromosome is a chromosome in which both arms are identical. It is thought to arise when a centromere divides in the wrong plane, yielding two daughter chromosomes, each of which carries the informations of one arm only but present twice. The isochromosomes are formed during mitosis and meiosis.

If a gamete having a isochromosome is fertilized by a normal gamete, the zygote will possess an unbalanced karyotype.

In *Drosophila,* the misdivision of centromere of telocentric X chromosome changes that into an "attached-X" isochromosome, In man X-isochromosome causes the disease called gonadal dysgenesis.



Formation of a isochromosome (after Sutton, 1965)

Lecture XXV

Variation in Chromosome Structure – Deletion and Duplication – Genetic and Cytological Implication

Types of chromosomal aberrations:

The chromosomal aberrations may remain confined to a single chromosome or may extend to both of the member of the homologues pair and, therefore, may be of following types:

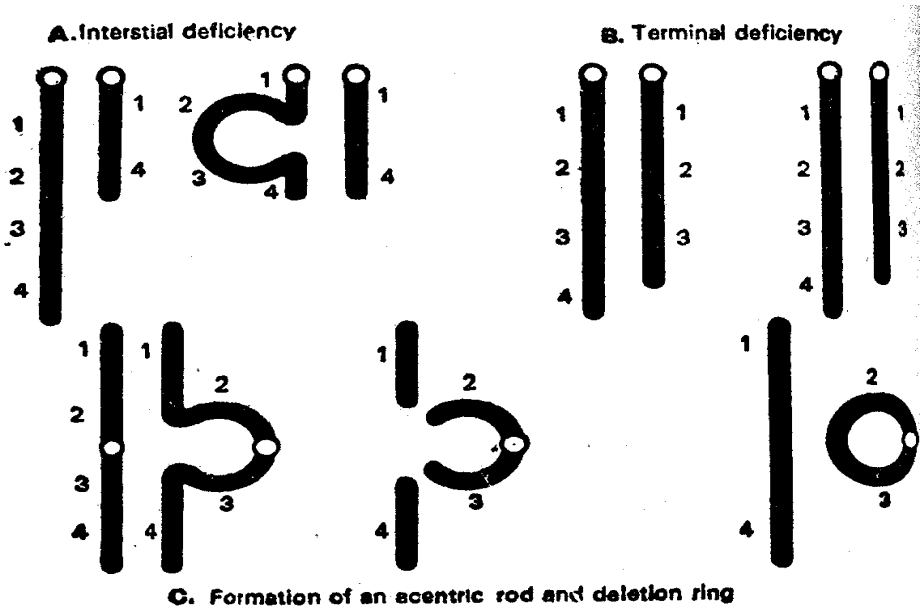
A. Intrachromosomal aberrations,
B. Interchromosomal aberrations.

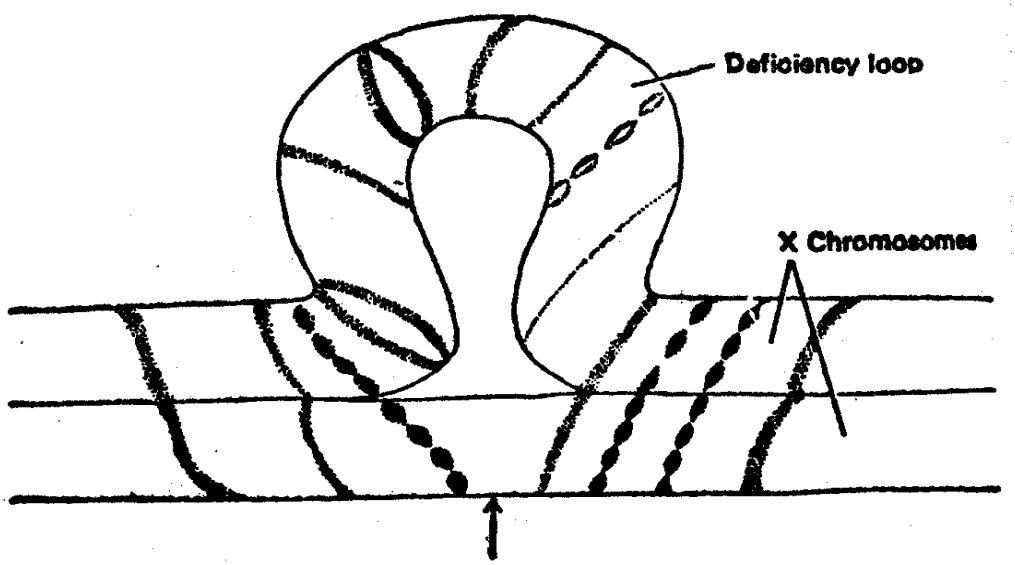
A. Intrachromosomal Aberrations

When aberrations remain confined to a single chromosome of a homologous pair, they are called intrachromosomal or homosomal aberrations.

1. Deficiencies (Deletions)

In deletion or deficiency type intrachormosomal aberration a chromosomal lacks either in an interstitial or terminal chromosomal segment which may include only a single gene or part of a gene. If break occurs near the end of a chromosome, a small piece of the terminal end is lost and thus, terminal deficiency occurs. Sometimes, two breaks may occur at any two points, releasing an intercalary segment which may remain rod-shaped or may become ring shaped, if its broken ends join and fuse. If, this ring-shaped chromosome (called deletion ring) has centromere is persists, but if lacks in that, loses during cell division. The broken ends of original chromosome are fused and has intercalary or intersitial deficiency. If this chromosome has centromere it persists otherwise lost during cell division.





A deficiency loop in the paired X-chromosomes from a salivary gland cell of a *Drosophila* larva heterozygous for notch (after King, 1965).

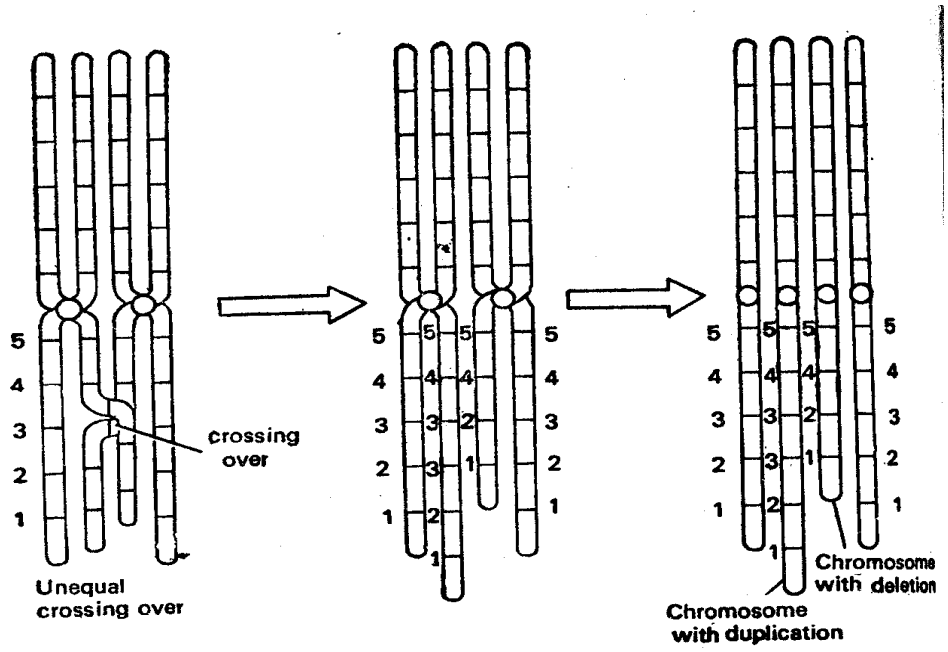
In some organisms terminal deficiencies are more common (ie.e., maize) than in other (i.e., *Drosophila*) which may be related to differences in repair systems or stability of open breaks.

Genetic Significance of Deficiencies

Lethal effect: Organisms with homozygous deficiency usually do not survive to an adult stage because a complete set of genes is lacking.

2. Duplications (Additions)

Duplication occurs when a segment of the chromosome is represented two or more times in a chromosome of a homologous pair. This extra-chromosomal segment may be a free fragment with a centromere or a chromosomal segment of the normal complement. During meiotic pairing the chromosome bearing the duplicated segment forms a loop. Pairing and exchange (crossing over) in inverted and displaced duplications leads to different secondary chromosome structural variants (i.e., chromosomal aberrations) such as reciprocal translocation, inversion, rings, acentric and dicentric chromatids.



Duplication and deletion (after Sutton, 1965).

Genetic significance of Duplications

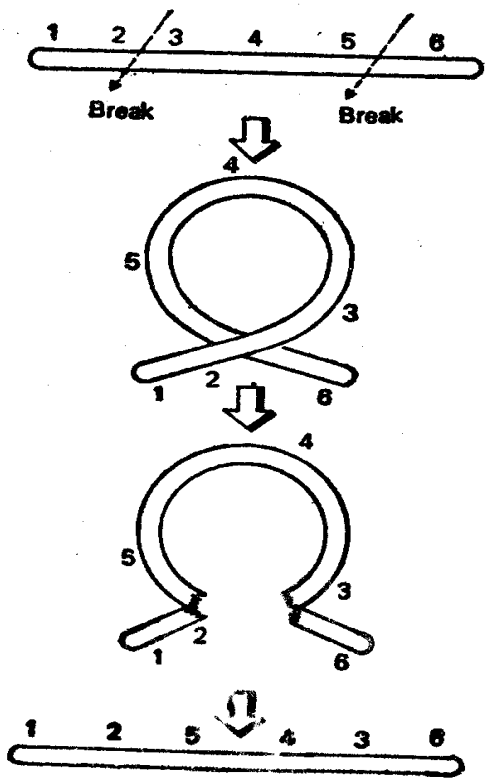
1. The duplications of chromosomes are not deleterious to the organism like the deficiency, but, they usually protect the organism from the effect of a deleterious recessive gene or from an otherwise lethal deletion.
2. some duplications are useful in the evolution of new genetic material. In an organism with duplications, because the old genes can continue to provide for the present requirements of the organism, the superfluous genes may be free to mutate to new forms without a loss in immediate adaptability.
3. Large duplications can reduce the fertility as a result of meiotic complication, and in this way reduce their own probability of survival (Sybenga, 1972).
4. Relocation of chromosomal material without altering its quantity may result in an altered phenotype, this is called position effect.

Lecture XXVI

Inversion and Translocation – Genetic and Cytological Implications

INVERSIONS

An inversion is an intra-chromosomal aberration in which a segment is inverted 180 degrees. For example if a chromosome has segments in the order of 1-2-3-4-5-6 and breaks occur in regions 2-3 and 5-6 and the broken piece (3-4-5-) is reinserted in reverse order, then the inverted chromosome will have segments in order of 1-2-5-4-3-6, such as shown in the figure 21.5:



The origin of an inversion (after Stansfield, 1969).

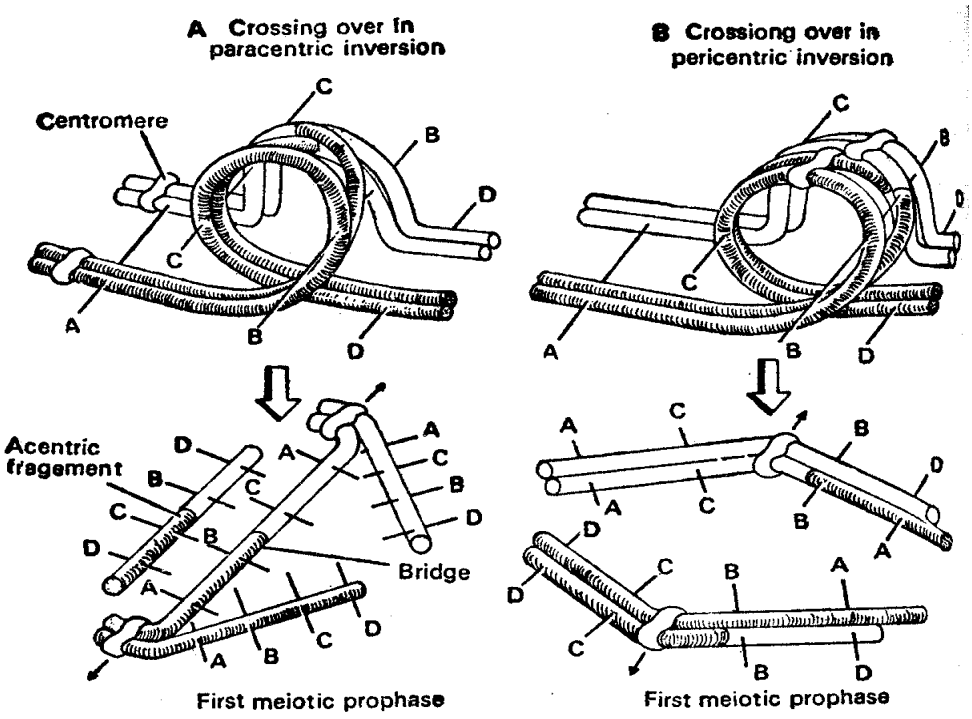
In a diploid organism, when out of two homologous chromosomes one chromosome undergoes the inversion, then, it is called inversion heterozygote. During synapsis of such a homologous pair having inversion heterozygoe, the synapsis configuration attempts to maximize the pairing between homologous regions in the two chromosomes. This is usually accomplished by a characteristic inversion loop in one of the chromosome.

Types of inversions

The inversions are of following types:

i) Pericentric inversions – When the inverted segment of chromosome includes or contains centromere, then such inversions are called heterobranchial or pericentric inversions. If crossing over occurs with in the loop of a pericentric inversion, the resulted chromatids include half on the chromatids with duplications and deficiencies forming nonfunction. The other half of the chromatids form functional gametes: ¼ gametes have normal chromosome order, ¼ gametes have the inverted arrangement.

ii) Paracentric inversions – When the inverted segment includes no centromere and the centromere remains located outside the segment, then such type of inversion is called homobranchial or paracentric inversion. Crossing over within the inverted segment of a paracentric inversion, produces a dicentric chromosome contains two centromeres and forms a bridge from one pole to the other during first meiotic anaphase. When anaphase chromosomes separate towards poles, this bridge breaks somewhere along its length and the resulting fragments contain duplications and/ or deficiencies. The acentric chromosome because lacks in centromere and fails to move to either pole and so, is not included in the meiotic products. Such, breakage-fusion bridge cycles of crossing over of paracentric inversions are most common in maize. The meiotic products includes half non-functional, ¼ functional normal and ¼ functional inverted chromosomes.



Crossing over in paracentric and pericentric inversions
(after SRB, Owen and Edger, 1965)

Genetic significance of inversions

i) Simple inversions do not have primary phenotypic effects other then on chromosome shape. Frequently, however, some DNA at a break point has been damaged and this may result in an observable mutation, often recessive (e.g., c 1B lethal mutation in Drosophila).
ii) Due to inversion a peculiar kind of position effect occurs. The position effect is caused by the transfer of a gene from a euchromatic segment to the vicinity of heterochromatic segment. Heterochromatinization may then extend into a displaced, originally euchromatic region and suppress the transcription of the gene in it.
iii) Normal linear pairing is not possible in inversion heterozygotes. The difficulties encountered with pairing cause a reduction of exchange (crossing over) in and around the inversion.
iv) They maintain heterozygosity from generations to generations.

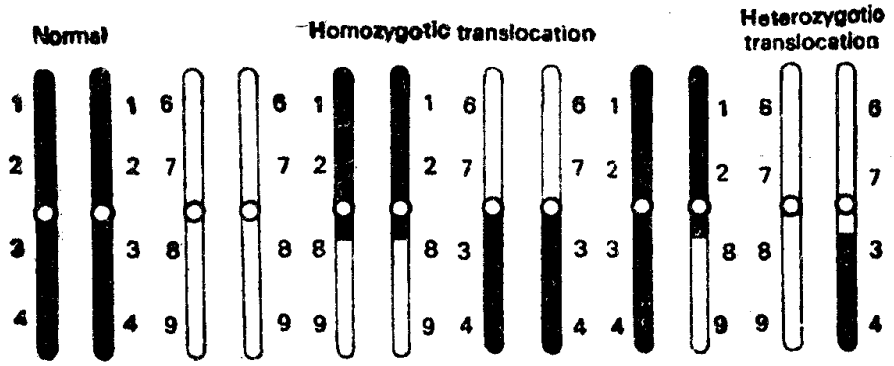
B. Interchromosomal aberrations

When breaks occur in non-homologous chromosomes and resulting fragments are interchanged by both of the non-homologous chromosomes, the inter-chromosomal or heterosomal aberrations occur. The inter-chromosomal aberration is of following type:

TRANSLOCATION

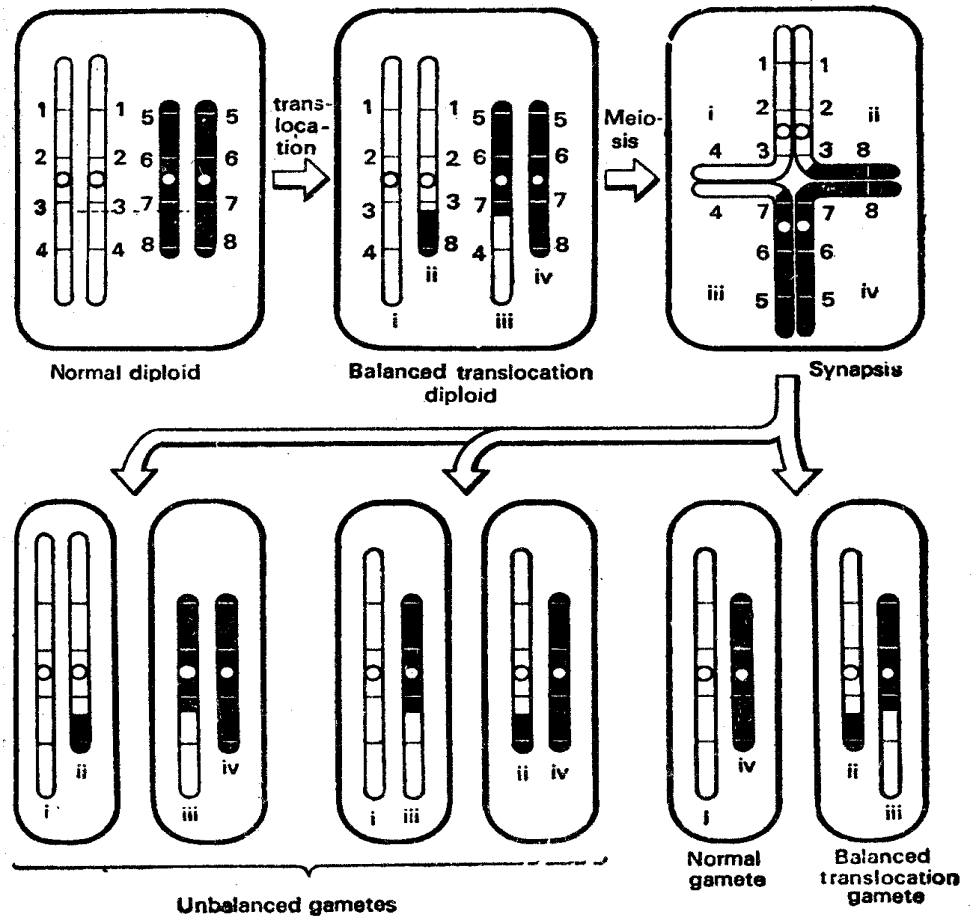
Translocation involves the shifting of a part of one chromosome to another non-homologous chromosome. If two non-homologous chromosomes exchange parts, which need not be of the same size, the result is a reciprocal translocation. The reciprocal translocation may be of following types:

1. **Homozygotic translocation** - In homozygotic translocation normal meiosis occur and cannot be detected cytologically. Genetically they are marked by altered linkage group by the fact that a gene with new neighbours may produce a somewhat different effect in its new location (position effect).



Homozygotic and heterozygotic translocations
(after De Robertis, Saez and Nowinski 1970)

2. **Heterozygotic translocation –** In heterozygotic translocation a considerable degree of meiotic irregularity occur. During meiosis, an individual which is heterozygous for a reciprocal translocation must form a cross-shaped configuration in order to affect pairing of all homologous segments. This cross-shaped configuration often opens out into a ring as chiasmata terminalize. The meiotic products (gametes) are of three types –normal balanced and unbalanced gametes as have been illustrated in following diagram:



Meiosis in heterozygotic translocation (after Sutton 1965).

Genetic significance of Heterozygotic Translocation:

1. The heterozygous translocation produce semi-sterile organisms because between half and two third gametes fail to receive the full complements of genes required for normal development of sex.
2. Some genes which formerly assorted independently, exhibit linkage relationships after translocation has occurred; a single reciprocal translocation will reduce the number of linkage groups by one.

3. The phenotypic expression of a gene may be modified when it is translocated to a new position in the genome (position effect).

Lecture XXVII

Variation in chromosome number – Euploid, Aneuploid – types of euploids

Types of changes in Chromosome Number

The somatic chromosome number of any species, whether diploid or polyploid, is designated as 2*n*,and the chromosome number of gametes is denoted as *n*. An individual carrying the gametic chromosome number, *n,* is known as haploid. A monoploid, on the other hand, has the basic chromosome number, *x.* In a diploid species, *n*= *x*; one *x* constitutes a genome or chromosome complement. The different chromosomes of a single genome are distinct from each other in morphology and/ or gene content and homology; members of a single genome do not show a tendency of pairing with each other. Thus a diploid species has two, a triploid has 3 and a tetraploid has 4 genomes and so on.

Individuals carrying chromosome numbers other than the diploid (2x, and not 2n) number are known as heteroploids, and the situation is known as heteroploidy.

The change in chromosome number may involve one or a few chromosomes of the genome; this is known as aneuploidy.

Heteroploidy that involves one or more complete genomes is known as euploidy.

By definition, therefore, the chromosome numbers of euploids are an exact multiple of the basic chromosome number of the concerned species.

A summary of the terms used to describe heteroploidy (variation in chromosome number)

Term	Type of change	Symbol*
Heteroploid	A change from 2x	-
A. Aneuploid	One or a few chromosomes extra or missing from 2n	2n ± few
Nullisomic	One chromosome pair missing	2n – 2
Monosomic	One chromosome missing	2n – 1
Double monosomic	One chromosome from each of two different chromosome pairs missing	2n – 1 – 1
Trisomic	One chromosome extra	2n + 1
Double trisomic	One chromosome from each of two different chromosome pairs extra	2n + 1 + 1
Tetrasomic	One chromosome pair extra	2n + 2
B. Euploid	Number of genomes or copies of a genome more than two	
Monoploid	One copy of a single genome	*x*
Haploid	Gametic chromosome complement	*n*
Polyploid	More than 2 copies of one genome or 2	

	copies each of 2 or more genomes**	
1. Autoployploid	Genomes identical with each other	3x
Autotriploid	Three copies of one genome	4x
Autotetraploid	Four copies of one genome	5x
Autopentaploid	Five copies of one genome	6x
Autohexaploid	Six copies of one genome	8x
Autooctaploid	Eight copies of one genome	
2. Alloployploid	Two or more distinct genomes	
AABB	(generally each genome has two copies)**	$(2x_1 + 2x_2)$**
Allotetraploid	Two distinct genomes	$(2x_1+ 2x_2 + 2x_3)$**
Allohexaploid	Three distinct genomes	$(2x_1+2x_2+2x_3+2x_4)$**
Allooctaploid	Four distinct genomes	

*2n = Somatic chromosome number (and complement) and n = gametic chromosome number (and complement) of the species, whether diploid or polyploid.
X = The basic chromosome number (and complement) or genomic number.
x_1, x_2, x_3, x_4 = Distinct genomes from different species.
** In general, this condition occurs; other situations may also occur.

Aneuploid individuals from which one chromosome pair is missing (2n – 2) are termed as nullisomics, while those lacking a single chromosome (2n – 1) are known as monosomics. A double monosomic individual has two chromosomes missing, but the two chromosomes belong to two different chromosome pairs (2n – 1 – 1). An individual having one extra chromosome (2n + 1) is known as trisomic, and that having two extra chromosomes each belonging to a different chromosome pair is called double trisomic (2n + 1 +1). When an individual has an extra pair of chromosomes, it is known as tetrasomic (2n + 2). The detailed terminology describing aneuploidy in very complex, The breeder is generally concerned with monosomics and trisomics, and in some situations, with nullisomics and tetrasomics.

In euploids, the chromosome number is an exact multiple of the basic or genomic number. Euploidy is more commonly known as polyploidy. When all the genomes present in a polyploidy species are identical, it is known as autopolyploid and the situation in termed as autopolyploidy. In the case of allopolyploids, two or more distinct genomes are present. Euploids may have 3 (tripoid), 4 (tetraploid), 5 (pentaploid), 6 (hexaploid), 7 (heptaploid), 8 (octaploid) or more genomes making up their somatic chromosome number.

In case of autopolyploidy, they are known as autotriploid autotertaploid, autopentaploid, autohexaploid, autoheptaploid, autooctaploid and so on, while in the case of allopolyploidy they are termed as allotriploid, allotetraploid, allopentaploid, allohexaploid, alloheptaploid, allooctaploid etc.

Amphidiploid is an allopolyploid that has two copies of each genome present in it and, as a consequence, behaves as a diploid during meiosis. A segmental allopolyploid contains two or more genomes, which are identical with each other, except for some minor differences.

Lecture XXVIII

Polyploids – Auto and Allopolyploids

AUTOPOLYPLOIDY

In autopolyploidy are included triploidy, tetraploidy and higher levels of ploidy. Autopolyploids are produced directly or indirectly through chromosome doubling, which is briefly considered as follows.

Origin and production of Doubled Chromosome Numbers

Cells/ individuals having doubled chromosome numbers may originate in one of the following several ways:

(1) Spontaneous,
(2) Due to treatment with physical agents,
(3) Regeneration in vitro,
(4) Colchicine treatment, and
(5) Other chemical agents.

(1) **Spontaneous –** Chromosome doubling occurs occasionally in somatic tissues and unreduced gametes are also produce in low frequencies.

(2) **Physical Agents –** Heat or cold treatments, and X–ray or gamma-ray irradiation may produce polyploids in low frequencies. Tetraploid branches were produced in Datura in response to cold treatment. Exposure of maize (*Z.mays*) plants or ears to a temperature of 38-45^{O}C at the time of the first division of zygote produces 2-5 per cent tetraploid progeny. Heat treatment has been successfully used in barley (*H.vulgare)*, wheat (*T.aestivum*), rye *(S.cereale)* and some other crop species.

(3) **Regeneration in vitro –** Polyploidy is a common feature of the cells cultured *in vitro*. Some of the plants regenerated from callus and suspension cultures may be polyploids. Plants of various ploidy have been regenerated from callus cultures of *Nicotiana, Datura,* rice (*O. sativa)* and several other species.

(4) **Colchicine treatment –** *Colchincine treatment is the most effective and the most widely used treatment for chromosome doubling.* It has been used with great success in a large number of crop species belonging to both dicot and monocot groups. Pure colchicine is $C_{22}H_{25}O_6N$. It blocks spindle formation and thus inhibits the movement of sister chromatids to the opposite poles. The resulting restitution nucleus includes all the chromatids; as a result, the chromosome number of the cell is doubled.

(5) Other chemical agents – several other chemical have polyploidizying effect. Notable among them are, 8- hydroxyquinoline and nitrous oxide. These chemicals are much less effective than colchicine and are not commonly used.

Morphological and Cytological Features of Autopolyploids

Morphological features of polyploids vary to some extent from one species to the other. Some general features are summarised below.

1. Polyploids have larger cell size than diploids. Guard cells of stomata are larger, and the number of stomata per unit area is lower in polyploids than in diploids.
2. Pollen grains of polyploids are generally larger than those of the corresponding diploids.
3. Polyploids are generally slower in growth and later in flowering.
4. Polyploids usually have larger and thicker leaves, and larger flowers and fruits, which are usually less in number than the diploids.
5. Polyploids generally show reduced fertility due to irregularities during meiosis and due to genotypic imbalance.
6. In many cases, autopolyploidy leads to an increase in general vigour and vegetative growth. But in some cases polyploids are smaller and weaker.
7. Different species have different levels of optimum ploidy. For sugarbeet (*Beta vulgaris*), the optimum level is 3x, while for Timothy it is between 8-10x.

Autopolyploid crop species

Common name	Scientific name	Somatic chromosome number (2n) of the cultivated form	Somatic chromosome number of related wild species
Potato	*Solanum tuberosum*	48 (4*x*)	24 (2x) form of *S. tuberosum*
Coffee	*Coffea arabica*		22,66,68
Alfalfa	*Medicago sativa*	44 (4*x*)	14,16,32
Peanut	*Arachis hypogaea*	32 (4*x*)	
Banana	*Musa sapientum (M.paradisiaca)*	40 (4*x*)	22
Sweet potato	*Ipomoea batatas*	33 (3*x*)	-
		90 (6*x*)	

Application of Autopolyploidy in Crop Improvement

Autopolyploidy has found some valuable applications in crop improvement. These are briefly summarised below.

Triploids: Triploids are produced by hybridization between tetraploid and diploid strains. They are generally highly sterile, except in a few cases. This feature is useful in the production of seedless watermelons. In certain species, they may be more vigorous than the normal diploids, ***e.g.,*** in sugarbeets.

Seedless watermelons are grown commercially in Japan. They are produced by crossing tetraploid (4x, used as female) and diploied (x, used as male) lines. The triploid plants do not produce true seeds; almost all the seeds are small, white rudimentary structures like cucumber seeds. But a few normal sized seeds may occur, which are generally empty. For good fruit setting, pollination is essential. For this purpose, diploid lines are planted in the ratio 1 diploid: 5 triploid plants.

Triploid sugarbeets (*B. vulgaris*) produce larger roots and more sugar per unit area than diploids, while tetraploids produce smaller roots lower yields than diploids. Apparently, 3x is the optimum level of poloidy in sugarbeets. Triploid sugarbeet varieties have been grown commercially in Europe and Japan. Seed production of triploid sugarbeet is difficult because the beet flower is small. Triploid seed may by produced in one of the following two ways: (1) using 4x plants as females and 2x as male or (2) using 4x as male and 2x as female. Commercial triploid sugarbeet seed in produced by interplanting 4x and 2x lines in the ratio 3 : 1. Triploid sugarbeet may give 10-15 per cent higher yields than diploids.

A triploid (3x) clone of tea (*Camelia assamica*) has been recently released by the Tea Research Association, India, for cultivation in the Northern parts of the country. The triploid cultivar, TV 29, produces larger shoots and , there by, biomass, yields more cured leaf per unit area and is more tolerant to drought than the available diploid cutlivars

ALLOPOLYPLOIDY

Allopolyploids have genomes from two or more species. Several of our crop plants are allopolyploids. Production of allopolyploids has attracted considerable attention; the aim almost always was the creation of new species. Some success has been obtained as is evident from the emergence of *Triticale* as a new crop species in some areas, and the promise shown by some other allopolyploids, e.g., Raphanobrassica and some allopolyploids of forage grasses.

Some genera, which contain allopolyploid species, and one or more crop species. The crop species themselves may be allopolyploid or diploid. Genera like Triticum, Brassica and Gossypium have both diploid and allopolyploid crop spices.

Scientific name	Common name	Gametic chromosome number (n)	Cultivated / Wild**
A. sativa	Cultivated oats	14	C
B. oleracea	Cabbage	9 (C)	C
B. juncea	Rai, Indian mustard	18	C
	Rape		
B. napus	Asiatic (desi) cotton	19	C
Gossypium arboreum	Asiatic cotton	13 (A_2)	C
G. herbaceum	Wild American cotton		
G. thurberi	Sea island (Egyptian) cotton	13 (A_1)	W
	American upland cotton	13 (D_1)	W
G. barbadense	Cultivated barley		
		26 (A_2D_2)	C
G. hirsutum	Noble canes		
		26 (A_1D_1)	C
Hordeum vulgare	Indian canes		
Saccharum officinarum		7	C
S. barberi	Indan canes		
	Kans (wild canes)	40	C
S. sinense			
S. spontaneum		41,45,46,58,62	C
		58,59	
		20-64	C
			W

* Letters within parentheses denote the symbols used for genomes present in the species.
**C = Cultivated; W = Wild.

Lecture XXVIII

Role of polyploidy in evolution of crops – Wheat, Cotton, Tobacco and Brassica

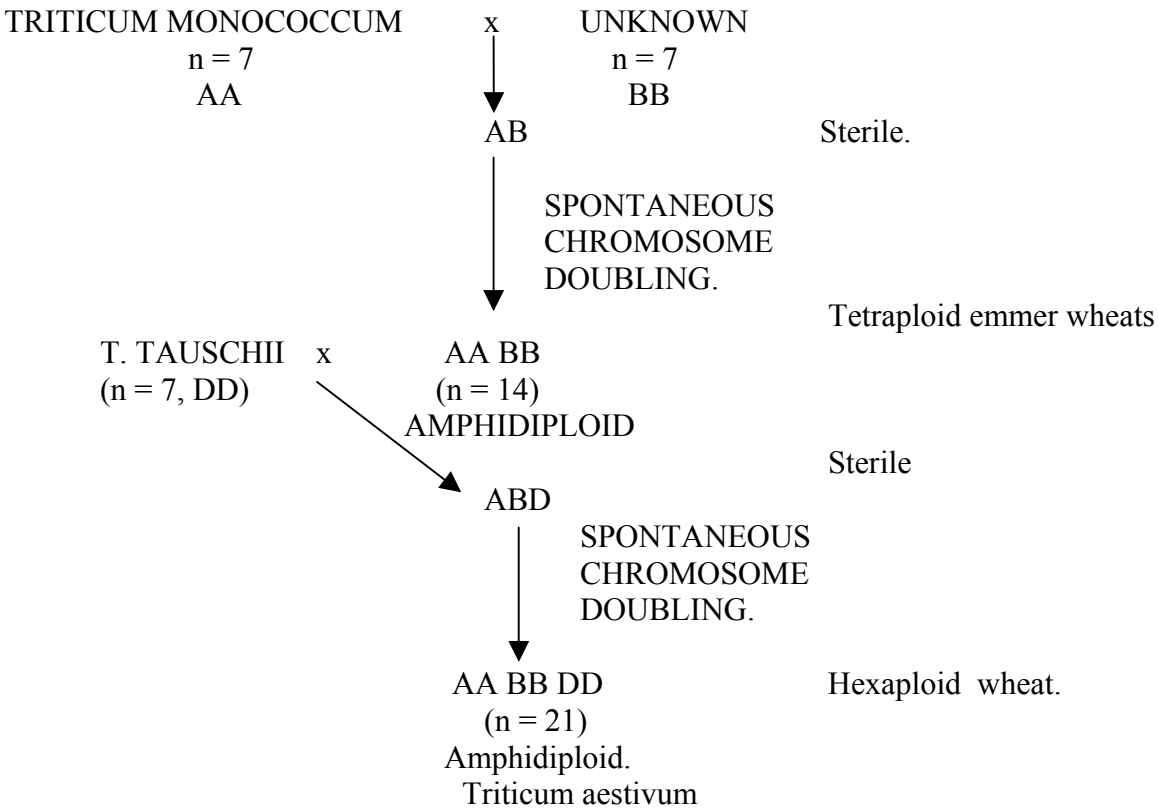
Role of Allopolyploidy in Evolution:

It is estimated that about one-third of the Angiosperms are polyploids, and by far the vast majority of them are allopolyploids.

The identification of parental diploid species is primarily based on pairing between the chromosomes of the diploid and the allopolyploid species. When the chromosomes of a diploid species pair with some of those of the allopolyploid species, homology between chromosomes of the two species is apparent. This homology suggests that the diploid species may be one of the parental species of the allopolyploid.

We shall briefly consider the possible evolutionary history of some important allopolyploid crop species, *viz.,* wheat, tobacco, cotton and Brassica.

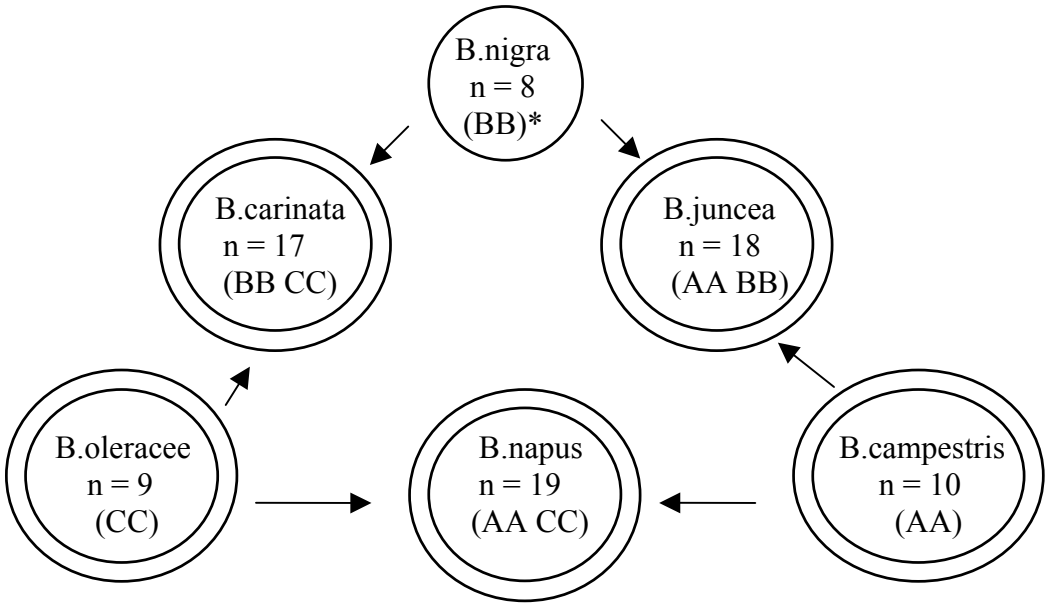
Evolution of Bread Wheat (*Triticum aestivum*). Evolutionary history of wheat has been the most extensively investigated, and is perhaps the least understood. Identity of the diploid species contributing the three genomes (A, B, and D genomes) of *T. aestivum* has been investigated by many workers, more notably by Sears, Kihara and others.



Evolution of Nicotiana tabacum. *N. tabacum* (n = 24) is most likely an amphidiploid from the cross *N. sylvestris* x *N. tomentosa*; both the species are diploid with n = 12. The interspecific hybrids *N. tabacum* x *N. sylvestris* and *N. tabacum* x *N. tomentosa* produce 12 II and 12 I at metaphase I. This indicates a homology between chromosomes of *N. tabacum* and those of *N. sylvestris* and *N. tomentosa*. The amphidiploid from the cross *N. sylvestris* x *N. tomentosa* is similar to *N. tabacum* in many characteristics, which further supports the above conclusion. The species *N. tabacum* has undergone considerable differentiation during its evolutionary history, mostly due to the accumulation of gene mutations and, to some extent, due to the loss of some duplicated segments of the two genomes.

Evolution of *Gossypium hirsutum*. The 9 old World and the 8 New World species of Gossypium have n = 13, but the chromosomes of the New World species are smaller than those of the Old World species. Three other species, *G. hirsutum,* G. barbadense and *G. tomentosum* (wild Hawaii cotton), have n = 26; in these species, 13 chromosomes are relatively larger than the remaining 13. A possible origin of *G. hirsutum* is from the cross between Asiatic cotton *G. arboreum* x *G. thurberi* (American wild cotton), followed by chromosome doubling of the inter specific hybrid. According to a more recent scheme, *G. hirsutum* has originated from the cross *G. herbaceum* var. *africanum* x *G. raimondii,* followed by chromosome doubling of the F_1.

Evolution of Amphidiploid *Brassica* Species*:* The origin of amphidiploid *Brassica* species is presented based on the famous U's Triangle proposed by N. U in 1935. According to this scheme, *B. juncea* (n = 18) is an amphidiploid from *B. nigra* (n =8) x *B. campestris* (n = 10), *B. napus (n* = 19) is an amphidiploid from the cross *B. oleracea (n* = 9) x *B. campestris* (n = 10), and *B. carinata* (n = 17) is an amphidiploid from the cross *B. nigra* (n = 8) x *B. oleracea* (n = 9). The synthetic allopolyploids produced according to the above scheme resemble the natural amphidiploids, cross easily with them, and the hybrids between the synthetic and natural amphidiploids are reasonably fertile.



Leccture XXIX

Types of Aneuploids and their origin

ANEUPLOIDY

Of the various anecuploids, monosomics (in polyploid species, such as, tobacco, wheat and oats) and trisomics [in diploid species, *e.g.,* maize, bajra, tomato, rye, pea, spinach, etc.] are most commonly used in genetic studies.

Nullisomics are viable in a few highly polyploid species only, *e.g.,* wheat and oats; they are not viable even in tobacco. Aneuploids are usually less vigorous than their diploid progenitors. Another characteristic of aneuploids is their high sterility resulting form irregular meiosis.

Monosomics

A monosomic is an individual that lacks one chromosome of the normal complement of somatic cells (2n – 1).

If the lost chromosome is one that is not absolutely essential for the organism, it may survive but if the lost chromosome is one that is very important, it may not live.

Loss of one chromosome in normal diploid plants may result in lethality. Thus, for example, monosomics are inviable in *Datura sp.* Polyploid plants, however, have been found to tolerate the loss of one chromosome. Twenty-four different monosomics, each lacking a single different chromosome of the normal complement, have been isolated in *Nicotiana tabacum* which is a tetraploid with 2n = 48. These 24 monosomics are morphologically distinct from each in haploid wheat (2n = 42), 21 different monosomics have been isolated.

Monosomics produce two kinds of gametes, one kind with n chromosomes and the other kind with n –1 chromosomes. When selfed, monosomics, therefore, produce normal (i.e., disomic), monosomic and nullisomic offspring.

Nullisomics

A nullisomic is an individual that lacks both members of one specific pair of chromosomes (2n-2).

Nullisomics are inviable in some species like *Nicotiana tabacum*, but in other species like *Triticum aestivum*, they are viable. In the Chinese Spring variety of wheat, Sears established 21 nullisomic lines (2n = 40), each lacking a single pair of chromosomes of the normal complement of the somatic cells. Different nullisomics are morphologically different from one another and from the normal Chinese Spring. They are reduced in size and vigour and are highly sterile. On selfing, they produce only nullisomics as their gametes contain only n – 1 (i..e, 20) chromosomes each.

Reference: B.D. Singh – 634-635

Trisomics

A trisomic is an individual with one chromosome more than the normal complement of the somatic cells (2n +1).

In general, an extra chromosome does not produce so striking effect as a missing one. In wheat, trisomics (2n = 43) occur but they are nearly indistinguishable from normal plants (i.e., disomes with 2n = 42. Blakeslee has isolated 12 different trisomics in *Datura sp.* (2n = 24), each having in triplicate a single different chromosome of the normal set. These trisomics differ morphologically from one another and from the diploid form.

Although trisomics give rise to two kinds of gametes, one kind with n chromosomes an the other kind with n + 1 chromosomes, they tend to be somewhat more stable genetically than monosomics.

Tetrasomics

A tetrasomic is an individual with two chromosomes more than the normal complement of the somatic cell (2n +1).

In a normal tetrasomic, two units of the same chromosome will be found besides the normal diploid number. If two different chromosomes (say chromosome No. 1 and chromosome No.2) are present besides the normal diploid number, it is called a double trisomic (2n + 1 + 1). During meiosis a quadrivalent is formed besides the bivalents in a tetrasomic, while two trivalents and two bivalents are formed in a double trisomic.

Tetrasomic produce gametes with n + 1 chromosome and when crossed with normal diploids (2n), they produce high frequency of trisomics.

Origin and Production

Spontaneous. Aneuploids originate spontaneously at a low frequency. The earlier cases of aneuploidy were produced spontaneously in experimental populations. Meiotic irregularities lead to the formation of n + 1 and n- 1 gametes, e.g., in Datura about 0.4 per cent of pollen is likely to be n + 1; these gametes give rise to 2n + 1 and 2n –1, respectively, progeny following fertilization.

Autotriploid Plants. *The best sources of aneuploids are triploid plants.* Distribution of chromosomes at the first meiotic anaphase of triploids is irregular leading to the production of a whole range of aneuploids in the progeny.

Asynaptic And Desynaptic Plants. In asynaptic and desynaptic plants, few to all chromosomes are present as univalents at metaphase I of meiosis. In the progeny of such plants, a relatively high frequency of aneuploids occur.

Translocation Hetrozygotes. A 3 : 1 disjunction of the ring or the chain of four chromosomes in a translocation heterozygote would produce one n + 1 and one n –1 gamete. As a result, a variable frequency of aneuploids are found in the progeny of translocation heterozygotes.

Tetrasomic Plants. Tetrasomic (2n + 2) plants would produce n + 1 gametes in considerable frequencies. Therefore, when they are crossed with normal disomic (2n) plants, they produce a high frequency of trisomics, where possible, tetrasomics may be maintained for the production of trisomics.

Double Trisomy - In a diploid organism when two different chromosomes are represented in triplicate, the double trisomy is resulted. A double trisomic has the chromosomal formula 2n + 1 + 1.