UNIT- 3 GENETICS AND EVOLUTION

Theories of evolution

1. Lamarck theory of inheritance of acquired characters

a. It was explained by **Jean Baptiste de Lamarck** (1774-1829), a French Biologist in his book Philosophic Zoologique.

b. It deals with the influence of environment on organisms, use or disuse of organs and the inheritance of acquired characters.

c. According to him, whenever the environment of certain organisms undergoes some changes, it forces the organisms to use certain organs more and put certain other organs to disuse.

d. The organs that are used more will increase in size and those not used continously will degenerate.

e. Such characters that are developed during the life time of an organismm are called acquired characters.

f. eg: Elongation of neck and forelimbs in giraffe (use), absence of limbs in snakes (disuse).

Objections of Lamarck theory of inheritance:

- The main objection for Lamarkism came from **Augustus Welsmann.** He conducted decaudalisation experiments on mice and proposed that the bodies of all the living organisms possess somatoplasm and germplasm. If any change occurs in the somatoplasm, it will not be transferred to the next generation but if any change occurs in the germplasm, it will be inherited to the next generation.
- e.g. 1) Well-developed muscles of athletes are not inherited to their children.

2) Making perforations to pinna for wearing ornaments has been in practice in India for the past several centuries. However no girl cild is born with ready-made perforations in their pinna.

Neo-Lamarkism:

- The contributions of Lamarck paved the way for the gradual acceptance of the concept of biological evolution and stimulated many further studies in this field later, it was modified to make it more acceptable.
- They considered that adaptions are universal. Organisms acquire new structures due to their adaptations to the changed environmental conditions. They argue that external conditions stimulate the somatic cells to produce certain 'secretions' which reach the sex cells through the blood. Such variations can be inherited by the offspring. **Paul Kammarer**observed the development of normal eyes and skin colour in the cave dwelling salamander, when exposed to daylight. The somatic character was passed on to the next generation.

2. Charles Darwin theory of natural selection:

1. It is an evolutionary theory proposed by Charles Robert Darwin (1809-1882), an English Naturalist.

2. Darwin's theory of natural selection does not explain what exactly evolution is, but explains how evolution might have occurred in nature. He believed that evolution is a gradual, rather than a sudden biological event. His theory was based on several facts, observations and inferences. They are:

i. Prodigality of production (or) over production: Every organisms tends to increase its population in large proportions. For example, *Paramecium* divides by binary fission at the rate of three to four times a day. At this rate, the volume of all the Paramecia equals to

10,000 times that of thhe earth at the end of the 9000th generation. **Salmon** fishproduces 28 million eggs and **starfish** one million eggs ina season. If all of them hatch and the larvae grow to reproductive age, all the seas will be filled with them in a few generations.

ii. Constancy in population: However, such an abnormal increase is not noticed in the population of any species in nature as the offspring die in large numbers before reaching the reproductive age. It is true that the food and the other sources do not increase in the same proportion as that of the population.

iii. Struggle for existence: As the food sources are limited, severe competition exists among the members of a population. Darwin called it struggle for existence which is of three types as: i) Intraspecific struggle, ii) Interspecific struggle and iii) Struggle with the environment.

iv. Universal occurence of variations: No two oranismms are exactly similar. They show variations. Even the offspring og the same parents are different.

v. Natural selection: According to Darwin, the individuals possessing harmful variations are reproductively less successful. The organismms with beneficial variations would increase the ability to reproduce and leave more fertile offspring.

vi. Origin of species: Darwin concluded that 'the struggle for existence resulting in the survival of the fittest' allows successsive generation to by Nature accumulate from generation to generation.

Objections to Darwinism:

- It failed to explain the mechanism by which variations occur. Thus Darwin faced the criticism 'DARWINISM EXPLAINS THE SURVIVAL OF THE FITTEST BUT NOT THE ARRIVAL OF THE FITTEST'.

- It could not explain the occurrence of vestigial organs, over specialisation of some organs like large tusks in extinct mammoths, oversized antlers in the extinct Irish deer, etc...
- It focused on small, fluctuating variations which are mostly non-heritable.
- It did not distinguish between sommatic and germinal variations.
- Darfwin did not consider the importance of macro-variations and considered them 'sports of nature'.

Neo-Darwinism:

According to this theory, five basic factors are involved in the process of organic evolution. They are as follows:

i. Gene mutations: Changes in the structure of a gene (DNA molecule) are called gene mutations or point mutations.

ii. Chromosomal mutations: Changes in the structure of chromosomes are called choromosommal mutations. They also bring about variations in the phenotype of organisms which lead to the occurrence of variations in the offspring.

iii. Genetic recombination:Recombinations of genes due to crossing over during meiosis are also responsible for bringing about genetic variability among the individuals of heritable variations.

iv. Natural Selection: Natural selection does not produce any genetic changes but once genetic changes occurred, it favouts some genetic changes while rejecting others. Hence it is considered the driving force of evolution.

v. Reproductive isolation: The absence of gene exchange between populations is called the reproductivve isolation. It plays a great role in giving rise to new species and preserving the species integrity.

3. Mutation theory of Hugo de Vries:

i. It was proposed by **Hugo de Vries**, a Dutch botaist who coined the term 'mutation'.

ii. Mutations are sudden, random inheritable changes that occur in organisms.

iii. He found four different forms in *Oenotheralamarckiana*(commonly called 'evening primose')such as *O*. brevistylis-small style, *O*. levifolia-smooth leaves, *O*.gigas-the giant form, *O*. nanella-the dwarf form (mutant varieties).

Salient features of Mutation theory:

i. Mutations occur from time to time in naturally breeding populations.

ii. They are discontinous and are not accumulated over generations.

iii. They are full-fledged, and so there are no 'intermediate forms'.

iv. They are subjected to Natural Selection.

Evidences for Biological Evolution

The theories that explain the evolution are hypothetical. In fact, it is not possible for anybody to observe even a single change in favour of evolution that occurs in the body of oragnisms as our life span is too short to notice such slow changes. Hence scientists collected evidences from different branches of biology. Some of them are:

a) Evidence from Palaeontology:

- Palaentology is the study of prehistoric life through fossils. Fossils are the remnants of plants or animals that were preserved in the layers of the earth and have been excavated from the soil. They are of various types like moulds, casts, petrifications, coprolites,actual remains of animals preserved in ice, etc...

- They support the idia that life has gradually evolved on the earth. The biologists and palaentologists have found the fossils of many transitional forms which link all the major groups of vertebrates. e.g. Euthenopteron between fishes and amphibians, Seymouria between amphibians and reptiles, etc...
- A complete fossil record of the various stages in the evolution of horse is available. It indicates that evolution is a gradual process and not a sudden creation of species.

b) Evidences from embyology:

- The study of the formation and early development of an organism is called embryology.
- **Ernst Haeckel** is considered he **'Father of Embryology'** and **Von Baer** is considered the **Father of Modern Embrology.**
- When the emvryos of different animals are observed, we find a fundamental similarity which tells us that there is a relationship among the animals.
- Embryology provides evidences from follows:

i) The observation of Von Baer: Von Baer studies the embryology of fish, salamnder, tortoise, chick and man. He observed that the early embryos of the above animals resemble each other closely in some basic/fundamental features. However these embryos differ in the final stages due to the formation of specialized characters. It indicates that the above animals have a common ancestor.

ii) Sequence of the developmental stages: The life of all the multicellular organisms begins with a single called stage, the zygote. It undergoes cleavages to produce the first stage embryo, the **morula**, which develops into a single layered second stage embryo, the **blastula**. It

develops into the third stage embryo, the **gastrula**, which finally develops iinto adult.

During this process, the zygote represents the unicellular stage, morula and blastula stages represent the colonial protozoan stages, whereas the gastrula stage represents the cnidarian stage. The development of the embryos of different organisms differs after the gastrula stage. This sequence of embryos shows that every multicellular organism passes through the above stages representing their common ancestry.

iii) Biogenetic Law or **Theory of recapitulation:** It was proposed by Ernst Haeckel. It states that ontogeny repeats phylogeny which means the development history of an organism repeats the evolutionary history of its ancestor, e.g. 1) Tadpole larva of frog resembles fish both externally and internally. 2) Caterpillar larva of butterfly recapitulates its closest ancestor, the annelid in body form. 3) In the embryos of birds and mammals, the heart is initially two chambered as in fish, then three chambered as in amphibians, and after that an incompletely divided four chambered heart as in reptiles and finally four chambered.

c) Evidences from comparative anatomy: When we compare the anatomy oof different animmals, we find somme similarities among them. For example, the fore limbs of different vertebrates are similar in origin and internal structure. All these indicate that there is a relationship among the organisms. These relationships can be studied under as follows:

i) Homologous organs: The organs which have similar structure and origin but not necessarily the same function are called homologous organs.

The evolutionary pattern that describes the occurrence of similarity in origin and internal structure is called homology. Such organs show adaptive radiation, hence divergent evolution, e.g. the appendages

of vertebrates such as the flipprees of whale, wings of bat, forelimbs of horse, paw of cat and hand of man, have a common pattern in the arrangement of bones even though their external form and functions may vary to suit their mode of life.

ii) Analogous organs: The organs which have dissimilar structure and origin but perform the same function are called the analogous organs. Analogous organs suggest convergent evolution, e.g. wings of a butterfly and wings of a bird.

iii) Vestigial organs: The organs which were functional in the ancestors but non-functional and reduced in the descendants are called vestigial organs. Presence of vestigial organs is the most convincing evidence in favour of organic evolution and also supports the concept of disuse proposedby Lamarck, e.g. Hind limbs in python, hind limbs and pelvic girdle in whale, wings of flightless birds, etc...

iv) Atavistic organs: Sudden appearance of some vestigial organs in a better developed condition as in the case of the tailed human baby is called atavism. Such organs are called atavistic organs they strongly support the concept of organic evolution.

v) Connecting links: The organisms which possess the characters of two different groups between which they are transitional are called connecting links. They clearly explain the path of evolution, e.g. Peripatus between annelida and arthropoda, prototherians between reptilian and mammalia, etc...

d) Evidences from cell and molecular biology: The field of cell and molecular biology provides the most detailed and convincing evidence in favour of biological evolution. They are studied under three headings as follows:

i) Fundamental unity of life: In all the living organisms, the structural and functional units are the cells. Every eukaryotic cell contains all the kinds of cells organelles such as Golgi complex, mitochondria, E.R.,

ribosomes, lysosomes, nucleus, chromosomes, D.N.A. and R.N.A. In all the living organisms, mitochondria help in energy production and storage, ribosomes help in protein synthesis; DNA has the same four types of nucleotides; all the proteins are synthesized from different combinations of the same 20 types of amino acids, the genetic code is virtually the same everywhere; different types of biochemical substances such as enzymes, hormones, respiratory pigments, etc…, and different types of biochemical reactions that occur in all the living organisms are the same, indicating some relationship among all the organisms on the earth.

ii) Blood precipitation tests: They are also called serological tests and were first conducted by H.F. Nuttal. He first injected a small amount of human serum into a rabbit. As our serum proteins are foreign to rabbit, antihuman antibodies are produced in that rabbit. The serum of the rabbit was collected, and it is called anti human serum. When it is mixed with the serums of an anthropoid ape, a monkey and a dog in separate test tubes, within a short time, a thick precipitate is formed with the serum of the anthropoid ape, moderate precipitate with that of the monkey and no precipitate with that of the dog. It indicates that the anthropoid ape is more closely related to man than to monkey and dog.

iii) Biochemical recapitulations: Animals recapitulate the biochemical aspects of their ancestors, e.g. a) An adult frog excretes urea but its tadpole excretes ammonia as the fishes do. b) Even the mammalian embryo first excretes ammonia then uric acid and finally urea.

MENDEL'S LAW OF INHERITANCE

The Austrian Monk GREGOR JOHANN MENDEL (1822-1884) was the first to explain the mechanism involved in transmission of characters from parents to offspring from generation to generation. From his hybridization experiment with garden pea(PisumSativum), he concluded

that the unit of inheritance is particulate in nature and called it as ‘factor’. He also proposed two universally accepted laws of inheritance : (i) The law of segregation and (ii) The law of Independent Assortment. The two laws of Mendel , especially the law of segregation, formed the basis of modern genetics. He is therefore, considered as the ‘father of genetics’.

MENDEL

Mendel was born on July 22,1822 in Heinzendorf, a village near Brunn in Austria (now Brno in Zechoslovakia). He graduated in 1840 and was admitted to Augustinian monastery on his 25th birthday. In 1849 Mendel became a temporary teacher in a preparatory school in Znaim. From 1851 to 1853, he studied mathematics and natural sciences in the University of Vienna. Here, he could not get success and returned to Brunn as a science teacher. In 1854, Mendel again took up the job of a teacher and continued there for fourteen years. In addition he worked as a priest in the local church. He lived in a house located within the premises of the church, During the period, he had time for his experiments on plant hybridization with the garden pea (PisumSativum). Mendel continued his experimental work over a period of 8 years for 1956 to 1964.The results of these experiments were published in the form of paper entitled – ‘Experiments in Plant Hybridization’ in the Annual proceedings of the Natural History Society of Brunn in 1866. In this paper ,Mendel proposed some basic generic principles

MENDEL’S EXPERIMENTS

1. Selection Of Material Mendel found edible pea (PisumSativum) as the best material for his hybridization experiments. The pea plant has various contrasting characters among its different varities such as : (i)stem may be tall or short; (ii)cotyledons may be green or yellow; (iii)seed may be round or wrinkled; (iv)seed coat maybe coloured or colourless; (v)unripe pods maybe green or yellow;

(vi)ripe pods may be inflated or constricted among the seeds; (vii)flowers may be axillary or terminal positions. Besides these contrasting characters, the pea plants is a very satisfactory material for the hybridization experiments due to the following advantages. (i)It was an annual plant. Its short life cycle makes it possible to study several generations within a period. (ii)It has perfect bisexual flowers and the flowers are predominately selfpollinating. (iii)Because of self-fertilization plants are homozygous. It is ,therefore, easy to get pure lines for several generations. (iv)Flowers are relatively large. Therefore, emasculation (removal of anthers from female flowers) and cross pollination is quite easy. This allows easy artificial hybridization. (v)Pea seeds are large and present no problem in germination.

2. Crossing Technique Mendel studied the inheritance of one character at a time, unlike his predecessors who has considered the organism as a whole.He carried out experiments to F2 and F3 generations and maintained the statistical records of all the experiments and analysed them carefully. He selected genetically pure plants as a parent plants. For this, he adopted self-fertilization technique. For instance, to get pure character for tallness, he self-fertilized a tall pea for many generations till the resulted offsprings always produced only tall plants. Mendel cross pollinated these two varieties of pea plants which were differing in apair of contrasting characters viz. tallness and dwarfness of the stem. To prevent self-fertilization, the anthers were removed from the female parent(seed plant) before their dehiscence. This is known as Emasculation. Then he collected pollen from male parent and dusted on the feathery stigmas or emasculated female flowers. This is known as crossing or Hybridization. These cross pollinated flowers were enclosed in separate bags to avoid further deposition of pollen from other resources. For each of the seven pairs of characters plants with one alternative as male. Reciprocal crosses were also made ,i.e, each of this crosses was made in two ways, depending on which phenotype is used as male or female. Example: Tall() x Dwarf()

Dwarf() x Tall() The population obtained as a result of crossing parents is called F1 generation. The progeny of F1 plants was then obtained by self-fertilization. It forms the F2 generation. Similarly F3, F4 etc. generations can also be obtained.

MONOHYBRID CROSSES A cross between two parents differing in a single pair of contrasting characters is known as monohybrid cross. Mendel self-pollinated pea plants for several generations to get pure homozygous plants.He found that the seven pairs of contrasting characters he had selected were always handed over from parent to offsprings. Seeds from tall plants produced only tall plants and yellow seeds produced plants with yellow seeds.He called them to represent pure lines. In one of his experiments,Mendel crossed a tall plany (about 6') with a dwarf plant (about 1'lenght) . The offspring of such cross were all the tall plants. These plants from the first Filial (F1) generation. When these F1 plants were allowed to selfpollinate, both tall and dwarf plants appeared in 3:1 ratio in F2 generation. This ratio is known as monohybrid phenotypic ratio. This showed him that the character of dwarfness disappeared in first generation (hybrid), but again reappeared in second generation. He self-pollinated the dwarf plants of F2 generation and found only dwarf plants in F3 generation . But when he self-pollinated the tall plants of F2 generation , then he found that one third (1/3) tall plants yielded only tall plants in F3 generation i.e. they are pure or homozygous fro tallness as the original parents. The rest two third (2/3) tall plants produced tall and dwarf plants in the ratio of 3:1. It means, F2 generation consisted of three types of plants (instead of apparent two types): (i)Tall homozygous (pure)-25% (TT) (ii)Tall heterozygous hybrid-50% (Tt) (iii) Dwarf homozygous (pure) – 25% (tt) From this observation, Mendel concluded that F2 ratio is more

accurately considered as 1:2:1 and not 3:1. This ratio is known as monohybrid genotypic ratio.

Explanation:

1. Both male and female parents make equal contribution tp the development of character in their progeny, since the results from reciprocal crosses are identical.

2. A character is produced by a specific gene (factor). Each gene has two alternative forms known as alleles . A gene can be represented by a symbol derived from the name of the character it governs.

3. The gene controlling height of the plant can be written by the latter T from the tallness. Similarly, the allele producing the recessive form of the character can be written by small or lower case letter (e.g. t for dwarfness).

4. Each somatic cell of an organism has two copies of each gene, as the organisms are in diploid condition. An individual may have the two copies of the same allele (e.g. TT or tt).Such an individual is called Homozygous (pure) and the condition is called homozygosity. But when an individual has two different allels of the same gene (i.e. Tt), it is referred to as heterozygous (hybrid) and the condition is called heterozygosity.

5. During gamete formation, the factos or alleles of a pair separate or segregate in such a way that each gamete (haploid) gamete will receive either T or t respectively.

6. When these plants are crossed , the female gamete containing the dominant allele (T) is fertilized by the male gamete containing recessive allele(t) or vice versa. Thus , the F1 generation plants contain both the alleles T and t. In appearance, these plants are tall because, the allele ‘T’ is dominant over the allele ‘ t‘ (Law of Dominance).

7. The factors or alleles present in a hybrid are not contaminated nor mixed with each other. When this plant produces gametes , the alleles segregate or separate in such a way that each gamete receives only one of the two alternative alleles. In other words, the F1 hybrid (Tt) produces two types of gametes : 50% of gametes contain the dominant gene T and the other 50% of gametes contain the recessive gene t. (Law of Segregation). This clearly shows that genes are particulate in nature.

8. The male and female gametes produced by F1 hybrid unite at random to produce four types of zygotes with respect to this gene. They are TT ,Tt, tt in equal proportion. The first three zygotes would develop into tall plants due to presence of T allele , while the fourth (tt) would produce dwarf plants . This would give rise to 3:1 phenotypic ratio by Mendel.

DIHYBRID CROSSES A cross between two parents differing in two pairs of contrasting characters is known as dihybrid cross. The progeny of such cross are called dihybrids . After investigating the behavior of all the seven characters of pea in their F1, F2, F3 generations , Mendel studied the inheritance of two characters at a time . In one of his experiments , Mendel crossed a homozugous pea plant having yellow and round seeds with another pea plants having green and wrinkled seeds. The F1 hybrids were found to have yellow and round seeds. This shows that yellow colour and round shape were dominant and green and wrinkled condition recessive.

When F1 plants were allowed to cross among themselves , four distinct types of seeds appeared in F2 generation. Two of these were similar to parental combination, while the other two were new combinations. These seeds are – • Yellow rounds - 315 • Yellow wrinkled - 101 • Green Round - 108 • Green Wrinkled - 32 Total - 556

From this data, we can conclude that the four types of seeds appeared in 9:3:3:1 ratio.This ratio is simply a multiple of 3:1 ratio i.e (3:1)^2. So Mendel concluded that this ratio (9:3:3:1) could be obtained only when the two sets of traits (seed colour and shape) inherit independent of each other.

Mendel represented round character of seed by the gene symbol R and wrinkled character by r. Similarly , he designated the yellow character by Y and green by y. Therefore , it was a cross between YYRR and yyrr . The gametes produced are in haploid condition . So, the gametes produced by YYRR are YR and those of yyrr are yr. The F1 offsprings produced by the union of these gametes are double heterozygotes with the genotype YyRr. These plants produced yellow and round seeds , just like the homozygous parent, displaying complete dominance.

Now, the essential point lies in the kinds of gametes produced by the F1 individuals . The allelic pairs Yy and Rr segregate independently of each other during the formation of gametes .Thus , a gamete could carry the allele Y or y and R or r . Whether Y or y will be associated with R or r is a matter of chance . As a result , four types of gametes with two parental an two new combinations i.e., YR, Yr, yR, yr are formed in approximately equal number as shown below.

Thus , recombination of genes takes place at the time of gametes formation in F1 hybrid . This is the principle of independent assortment of genes. The above four types male and female gametes on random mating produce four types of zygotes in ratio of 9:3:3:1 in F2 generation.

MENDEL'S LAW OF INHERITANCE On the basis of the results of his experiments Mendel recognized the phenomenon of the dominance and formulated the following two laws: 1. Law of Segregation 2. Law of Independent Assortment.

1. The phenomenon of dominance : Definition: When two homozygous individuals with one or more sets of contrasting characters at crossed , the characters that appear in the F1 hybrids are called dominant characters and those do not appear in F1 are recessive characters . In other words, a trait or character which appears only in homozygous individual is called a recessive character (e.g. dwarfness). A character which can phenotypically expresses itself in the homozygous as well as heterozygous individual is called dominant character (e.g. tallness). Explanation: In all his hybridization experiments , Mendel observed that when two alternative forms of a character are crossed and brought together , only one form is able to express itself in F1 hybrids i.e, the traits of only one parent was observed . He called the one which appeared in F1 as the dominant character . The other form , which remains masked or unexpressed in the hybrid is called recessive character.

After Mendel , several workers tested the validity of the phenomenon of dominance by performing cross-breeding experiments. They found its wide application in various plants and animals .

2. Law of Segregation :

Definition: Mendel's first law of inheritance is called the law of segregation . This law states that in a heterozygote a dominant and a recessive allele remain together throughout the life (from zygote to the gametogenesis stage). Without contaminating or mixing with each other they finally separate or segregate from each other during gametogenesis. So that each gamete receives only one allele either dominant of recessive . As the gametes are pure for a given character (e.g, Tallness or Dwarfness), this law is also known as Law of purity of gametes . Explanation: The homozygous tall plants possess two genes (factors) TT for tallness and dwarf plants possess two genes tt for smallness . The

gametes formed by tall plant will have a gene ‘T’ union form F1 hybrid which will have the genetic constitution as Tt. This hybrid is apparently tall as T is dominant over t. When this F1 hybrid form gametes, the two genes T and t separate from each other and pass on to separate gametes . As a result, two types of gametes are produced from the heterozygote in equal numbers. 50% of the gametes carry gene ‘t’. The genes (allels of a gene) thus segregate and the gamete can carry only one of the two alternative genes .Therefore , these gametes are pure either for tallness or dwarfness. Hence, the law of segregation is also called as the law of purity of gametes. These gametes during the process of fertilization can unite in three possible combinations, Viz. TT ,Tt and tt to produce two types of individuals in F2 generation 75% individuals have long stems and 25% short stems (Dwarf). The appearance of Dwarf plants in F2 generation indicate that in the F1 hybrid, the allele (t) for dwarfness remains along with allele (T)for Tallness , but does not mix with it or get contaminated by it . These alleles separate or segregate during gametogenesis .

Physical Basis of segregation: The phenomenon of segregation can be easily explained on the basis of behavior of homologous chromosomes during meiosis. As a consequence of segregation, the two alleles of a gene separate and go into different gametes. In F1 hybrid or heterozygote, one of the two alleles of a gene (t) is present in one chromosome , while the allele (t) is present in its homologue. The two homologous chromosomes pair during prophase I and orient at the metaphase plate during metaphase I. At anaphase I , one of the two homologous chromosomes move to one pole, while the other chromosome of this pair move to the opposite pole. Thus , each pole receives only one member of a homologous pair of chromosomes . As a result, one of the two alleles (T) goes to one pole , while the other allele (t) goes to the opposite pole. AT the anaphase II , the two sister chromatids of each chromosome separate and move to opposite poles producing four daughter cells each having a single chromatid from each

homologous pair of chromosomes. Two of these four cells receive sister chromatids from one of the two homologues, while the other two receive sister chromatids from the other homologue .Thus , separation of homologous chromosomes during meiosis result in the segregation of two alleles of a gene.

3. Law of Independent Assortment : Mendel formulated this law from the results of a dihybrid cross . According to his law: "the factors or genes for different pairs of contrasting characters present in a parent assort (separate) independently from one another during gamete production" Thus , any allele of one gene is equally likely to combine with any allele of the other gene and pass into the same gamete. Independent assortment of two genes produces four different types of gametes in equal proportion. A random union among these gametes gives rise to 16 possible zygotes. These zygotes yield a 9:3:3:1 phenotypic ratio, which is known as the typical dihybrid ratio.

Explanation: The mechanism of in assortment can be understood from a dihybridcross . In one of his hybridization experiments , Mendel crossed a homozygous pea plant having yellow round seeds (YYRR) with the homozygous pea plant having green wrinkled seeds (yyrr). The F1 hybrids were found to have yellow round seeds (YyRr). When this F1 hybrids were allowed to cross among themselves, they produced four types of seeds in the ratio of 9:3:3:1 as given below :

- Yellow rounds - 315(9)
- Yellow wrinkled - 101(3)
- Green Round - 108(3)
- Green Wrinkled - 32(1)

From this data , it becomes evident that yellow and green seeds appeared in the ratio of 416:140 i.e., 3:1 . Similarly round and wrinkled seeds appeared in the ratio of 423:133 i.e.; 3:1. Thus, each of the two pairs of alternative characters viz. yellow-green pair and round-wrinkled pair , behave exactly as in a monohybrid cross. This indicates that at the

time of gamete formation, the alleles for colour of seed do not interfere with alleles for shape of seed coat and the two pairs of alleles behave independent of each other. This principle is known as Mendel's law of Independent Assortment. This independent assortment holds good for two or more than two pairs of characters , which are located in different homologous pairs of chromosomes . On the other hand , if the alleles for different characters are present in the same homologous pair of chromosomes, they enter into the same gamete . Thus, law of Independent assortment is not applicable to such cases , where linkage between genes is in operation.

CELL DIVISION

Every organism start its life from a single diploid cell called '**Zygote'.** This zygote undergoes repeated divisions forming a multicellular organism. Cell division is essential for growth and reproduction of organism. **Rudolph Virchow** first pointed out that all new cell arise from pre-existing cells by division. This is called **'cell lineage theory'.**

Cell division is mainly of two types namely

1. Mitosis and 2. Meiosis

1. **Mitosis :** it occurs in the vegetative cells, hence called **'somatic cell division'.** In this division, parent cell produces two daughter cells, identical in all respects, hence also known as '**equational'** (or) '**homeotypic division'**. It mainly helps in the growth of the organism.
2. **Meiosis:** it occurs in reproductive(or)germinal cells. It results in the formation of four daughter cells in which chromosome number is reduced to half, hence called '**reduction division'.** the daughter cells formed are unidentical with that of mother cell. So it is also called '**heterotypic division'.** It mainly helps in reproduction.

MITOSIS (OR) EQUATIONAL DIVISION

Mitotic cell division was first observed in animal cells by,**walter flemming.** Later it was observed in plant cells by **strasburger.** It occurs in vegetative (or) somatic cells. It can be clearly observed in apical meristems of root or shoot

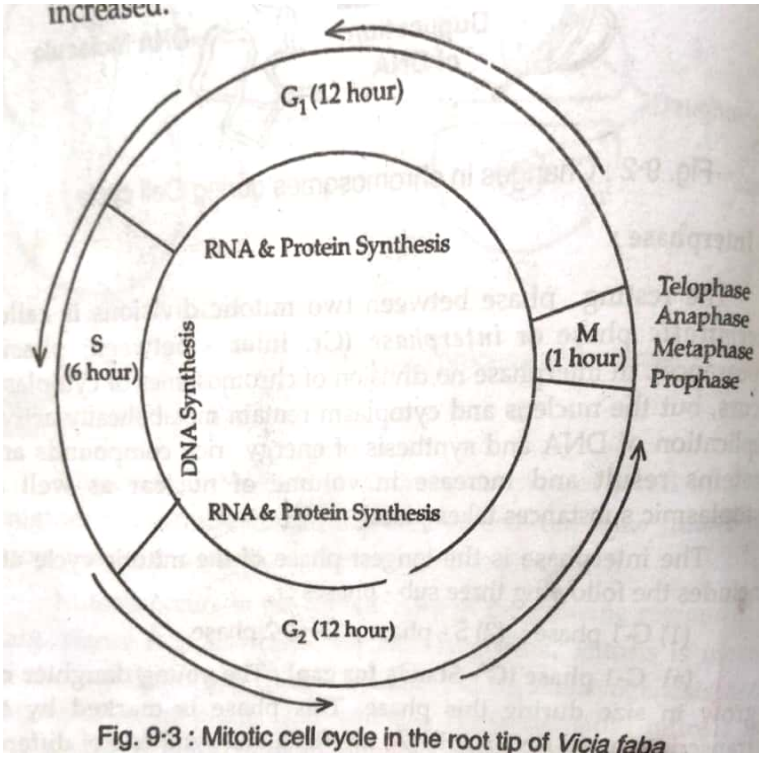
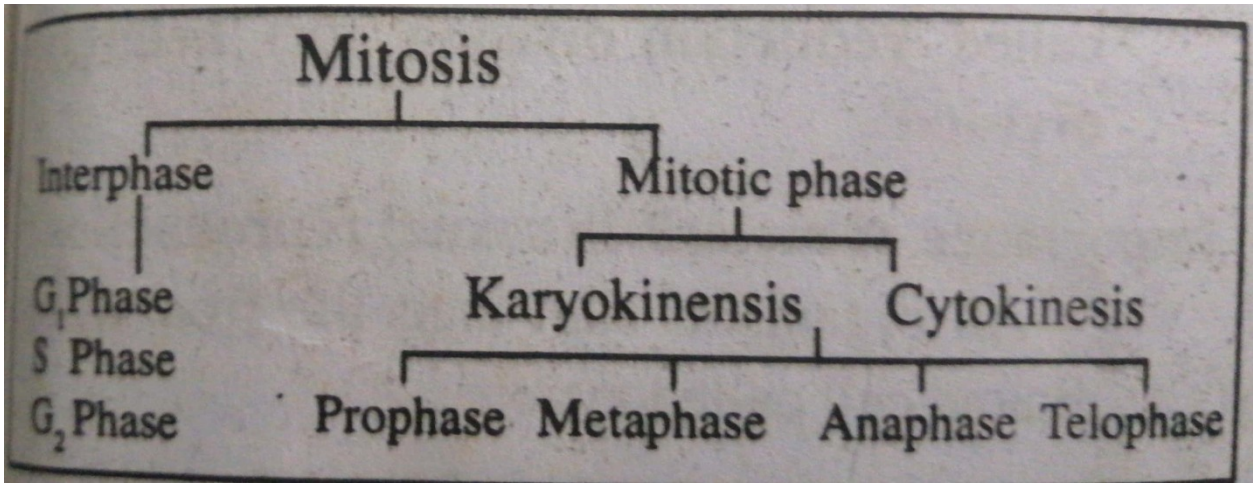


Fig. 9·3 : Mitotic cell cycle in the root tip of *Vicia faba*

STAGES OF CELLCYCLE

Ther are two stages in cell cycle.

A. Interphase B. Mitotic phase



A. **Interphase:**It is the largest phase in the cell. The non dividing stage intervening between two successive cell divisions is called interphase. In this phase no apparent changes occur in the nucleus. So it was previously considered as a resting stage. But later it was observed that a number of biochemical reactions take place by which the cell prepares for the next cell division.

1. **G1 Phase:**in this phase cell enlarges in size. A lot of RNA and protein is synthesized.
2. **Synthetic phase (or) s phase:** it is the most important phase in interphase. During this stage replication of DNA take place. So each chromosome appears to possess two chromatids.
3. **G2 Phase:** during this phase synthesis of RNA and proteins is continued. New cell organelles are synthesized.

Inside the nucleus chromatin appears as slender, elongated threads, called 'Prochromosomes'.

B. **Mitotic phase:** in this division phase, first nucleus divides, followed by the division of cytoplasm. The division of nucleus is called 'karyokinesis' and division of cytoplasm is called 'cytokinesis'.

a. **Kraryokinesis:**the nuclear division innolves many changes. It is divided into four phases, namely

1. Prophase
2. Metaphase
3. Anaphase and

4. Telophase

1. **Prophase:**in the early prophase, prochromosomes are long, thin and slender, prochromosomes become short, thick, distinct, rod shaped chromosomes by a process called '**spiralization'.** Each chromosome splits, longitudinally into two halves called 'chromatids' which remain united at the centromere. Nuclear membrane and nucleolus disappear.

2. **Metaphase :** during metaphase, a bipolar, anastral spindle is formed by the fusion of microtubules. **Spindle apparatus** is formed by types of spindle fibres.

   a. **Continuous fibres:** these are extended from one pole to other.

   b. **Chromosomal fibres:** they originate at one pole and get attached to centromeres of chromosomes.

   c. **Interzonalfibres:** these are small fibres present on either side of equator. The spindle fibres arrange all the chromosomes at the equatorial plane as a plate called '**equatorial plate' (or) 'metaphase plate'.** In the metaphase plate all the centromeres lie in the same plane and arms hang freely in cytoplasm.

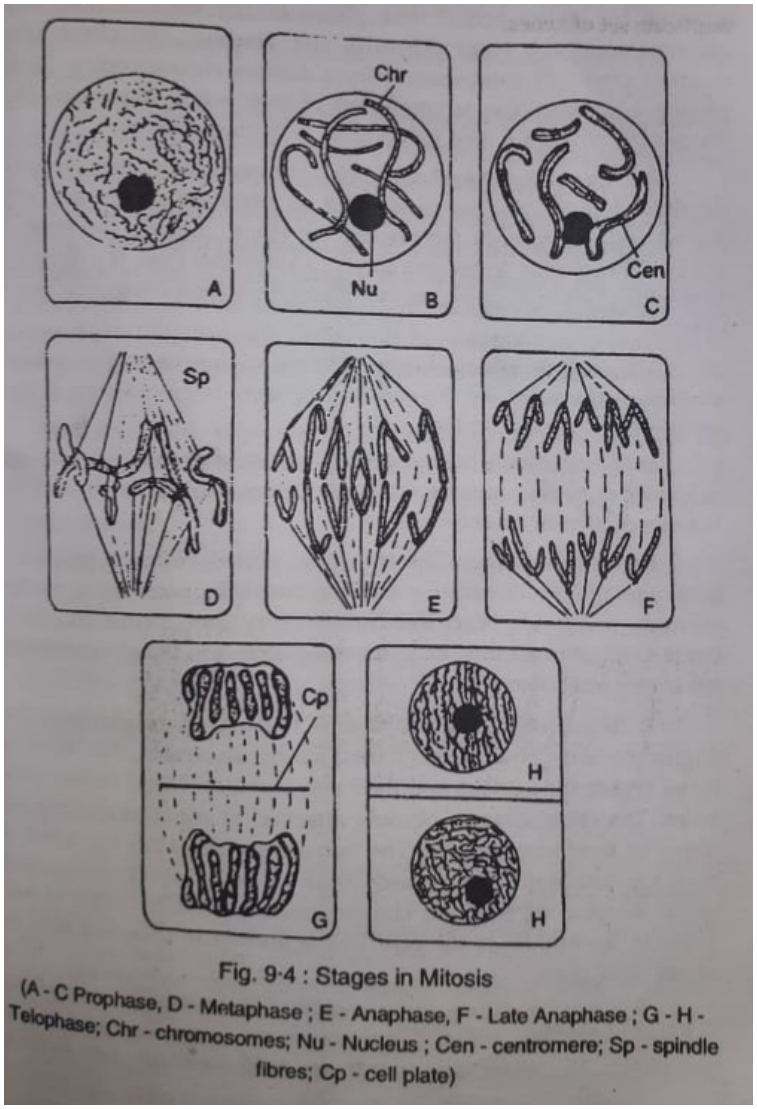


Fig. 9·4 : Stages in Mitosis
(A - C Prophase, D - Metaphase ; E - Anaphase, F - Late Anaphase ; G - H - Telophase; Chr - chromosomes; Nu - Nucleus ; Cen - centromere; Sp - spindle fibres; Cp - cell plate)

3. **Anaphase :** it is characterized by the movement of chromatids. At the beginning, the centromeres divide thus separating the chromatids from each other.now the separated chromatids are called ' **daughter chromosomes'**

Daughter chromosomes move to the poles pulled by spindle fibres. Chromosomes attain different shapes as **V,L,J**(or)**I** .

4. **Telophase :** all the changes are just opposite to that of prophase.

Chromosomes become long and thin by '**despiralisation'.** Nuclear membrane and nucleolus reappear forming two daughter nuclei with same of chromosomes as that of parent cell.

5. **Cytokinesis :**the division of cytoplasm in plant cells occur by cell plate method.

Soon after the formation of daughter nuclei, the spindle fragments unite to form a barrel shaped structure called "**phragmoplast"** at the equator.

The vacuoles of golgicomplex release pectin into **phragmoplast** forming a fluid plate called " **cell plate".** Cell plate grows centrifugally. By undergoing many physical and chemical changes, it gets modified into middle lamellum.

Later primary cell wall is formed by cellulose, hemicelluloses, pectin, thus forming two identical daughter cells.

SIGNIFICANCE OF MITOSIS:

1. It mainly helps in growth of the organism.
2. It helps in conserving the genetic integrity.
3. In unicellular organisims, it helps in reproduction.
4. Healing of wounds.
5. Regeneration of lost parts and grafting experiments.

MEIOSIS (or) REDUCTION DIVISION

Meiosis is a special type of cell division in which the chromosome number is reduced to half.

It occurs only in diploid, reproductive (or) germinal cells. Reduction of chromosome number during meiosis was first identified by **august Weismann** and it was later confirmed by strasburger in plants.

The term meiosis was coined by **J.B.Farmer** and **J.E Moore.**

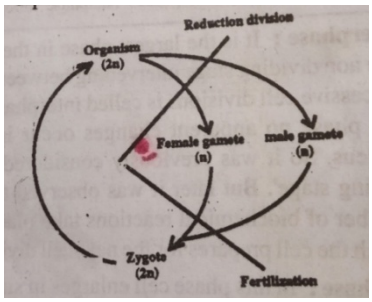
The cells in which meiosis occurs are called '**meiocytes'.**

In angiosperms it occurs in pollen mother cells and megaspore mother cells.

In meiosis a diploid cell produces four haploid unidentical daughter cells. So it is also called ' **reduction division'** (or) ' **heterotypic division'.**

Importance of meiosis in sexual reproduction:

All organisms start their life from a single diploid cell called zygote.



Constant chromosome number is maintained during meiosis

Zygote is formed by the fusion of gametes. In the life cycle of diploid organisms the chromosomes number is reduced to half at one stage (or) other. Otherwise at every fertilization the chromosomes number doubles leading to abnormal polyploids causing genetic disorders. So meiosis is most essential in sexually reproducing plants and helps to maintain constant chromosome number in each species

Meiosis-I: actual reduction in chromosome number occurs in meiosis-I, forming two unidentical daughter cells. Hence it is also called ' **reduction'** (or) **'heterotypic division'.**

1. **Prophase-I:**it is the prolonged stage and for the conveninence of study it is divided into 5 sub phases.

a. Leptotene:the chromosomes are long 'thin' slender and show bead like structures called '**chromeres'.**Chromomeres are considered as active genetic centres.
the chromosomes are arranged parallel and well separated but at the end of leptotene, the homologous chromosomes come close together at a point.

b. **Zygotene:**during zygotene the homologous maternal and paternal chromosomes pair with each other. This process is called '**synapsis'.**

Synapsis results in the formation of pairs of chromosomes called '**bivalents'.** Synapsis may be **proterminal,procentric or random.**

Zygotene is also characterizd by enlargement of nucleolus and initiation of spindle formation.

c. **Pachytene:** during pachytene each chromosomes appears to possess two chromatids. So a bivalent has four chromatids called '**pachytene tetrad'.**

In a bivalent two chromatids of a same chromosomes are called ' **sister chromatids'** and chromatids of different chromosomes are called ' **non sister chromatids'.**

In pachytene the non-sister chromatids cross over and exchange their parts mutually at one,two or many places.

Such points where the chromatids physically contact each other are called **chiasmata.** Here the chromosomes appear as 'x'-shaped structures.

During chiasmata, breakage of chromatid arms result dua to '**endonuclease'** enzyme.

The broken chromatid arms exchange mutually with each other and get united due to '**ligase'** enzyme.

The formation of chiasmata leads to the exchange of genetic material and results in the recombination of genetic characters. This process is known as " **crossing over".** This results in origin of new species which leads to evolution.

d. **Diplotene :**in this phase homologous chromosomes repel with each other but remain together at chiasmata.

Condensation, contraction and thickening of chromosomes results.

e. **Diakinesis :**the chiasmata move to the ends of chromosomes. This displacement of chiasmata to the ends is called

terminalisation. Nucleolus disappears. Chromosomes remain apart from each other.

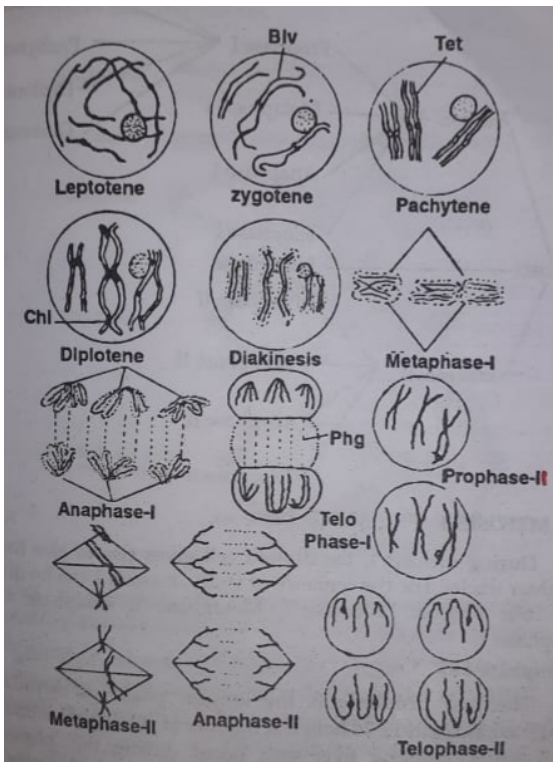
2. **Metaphase I :** a bipolar anastral spindle apparatus is formed. The chromosomes move to the equator and arrange their centromeres directed towards the opposite poles. The two homologous chromosomes of a bivalent remain attached at the telomeres.
3. **Anaphase I :** during anaphase I, chromosome move to respective poles due to contraction of spindle fibres.
This movement of entire chromosome to opposite poles is called 'disjunction' (or) '**seggregation'**. Thus the two genomes are separted resulting in halving of chromosome number and migrate to their respective poles.
4. **Telophase I:**nuclear membrane and nucleolus reappear, thus forming two daughter nuclei each with haploid number of chromosomes.

MEIOSIS II(OR) SECOND MEIOTIC DIVISION

It is similar to mitosis, hence called **equational division.** Each daughter to form two cells,forming four haploid cells. It is divisible into four phases,namely prophase II, metaphase II, anaphase II and telophase II.

1. **Prophase-II :** chromosomes are organized each with two chromatids. Nuclear membrane and nucleolus disappear.
2. **Metaphase-II:** two spindles are formed perpendicular to metaphase-I spindle. Chromosomes are arranged o metaphase plate.
3. **Anaphase-II:**centromeres break and daughter chromosomes move to opposite poles.
4. **Telophase-II:** daughter chromosomes organize into chromatin. Nuclear membrane and nucleolus reappear. Thus at the end of telophase-II, the daughter nuclei with half the number of chromosomes as that of the parent cell are formed.

Cytokinesis:cytokinesis may occur after each nuclear division (or) simultaneously after two nuclear divisions are completed.



SIGNIFICANCE OF MEIOSIS:

1. It helps to maintain constancy in chromosomes number in each species.
2. Due to crossing over variations occur which lead to evolution.
3. It helps in sexual reproduction by forming gametes.

DIFFERENCES BETWEEN MITOSIS AND MESIOSIS

MITOSIS

1. It occurs in both haploid and diploid organisms.
2. It occurs in somatic cells.
3. Nucleus divides once.
4. Daughter cells are identical.
5. Two daughter cells are formed.
6. Prophase is simple.
7. Pairing of chromosomes does not occur.
8. Chiasmata are absent. Crossing over is absent.

9. Centromeres undergo division in anaphase.
10. Daughter chromosomes move to the opposite poles.
11. The chromosome number of daughter nuclei is unchanged.
12. Duration of time is less.

MEIOSIS

1. It occurs only in diploid organisms.
2. It occurs in the reproductive cells.
3. Nucleus divides twice.
4. Daughter cells are unidentical.
5. Four daughter cells are formed.
6. Prophase is complicated and shows five sub stages.
7. Homologous chromosomes pair to form bivalents.
8. Chiasmata are formed. Crossing over occurs between non-sister chromatids.
9. Centromeres do not divide in anaphase-I, but divide in anaphase-II.
10. Bivalents are disjuncted to opposite poles in anaphase-I.
11. The chromosomes number of daughter nuclei is reduced to half.
12. Duration of time is more.

CELL BIOLOGY

7 CHROMOSOMES

7-1 INTRODUCTION

Chromosomes are the rod - shaped, dark - stained bodies located in the nucleus of a cell.

Eukaryotic chromosomes contain DNA and proteins. They are capable of self - replication and play a vital role in heredity, mutation, variation and evolutionary development of the species. Each chromosome has two arms, the shorter one called 'p' arm (from the French, Pekit - small) and the longer 'q' arm (q following the p in the Latin alphabet).

Mitotic metaphase chromosomes are the most suitable for studies on chromosome morphology. In such chromosomes, the following structural features are seen under light microscope : (1) Chromatids, (2) Centromere, (3) Telomere, (4) Secondary constriction and satellite, and (5) Chromomere.

1. **Chromatid :** Each metaphase chromosome consits of two symmetrical structures, called *Chromatids*. Each chromatid contains a single DNA molecule. The two chromatids of a

chro osome are held together at a point called the *centromere*. The two romatids separate from each other during anaphase and more to op osite poles. As a consequence, each chromosome is represented by a ngle chromatid during telophase.

The DNA of each telophase chromosome (composed of a single chro natid) replicates during the synthesis (S) phase of interphase. This produces an identical copy of the chromatid so that during prop hase and metaphase the chromosome is made up of two chromatids. Since the two chromatids of a prophase chromosome are produced through replication, they are referred to as *sister chromatids*. In contrast, the chromatids of homologous chromosomes are reffered to as *non sister chromatids*.

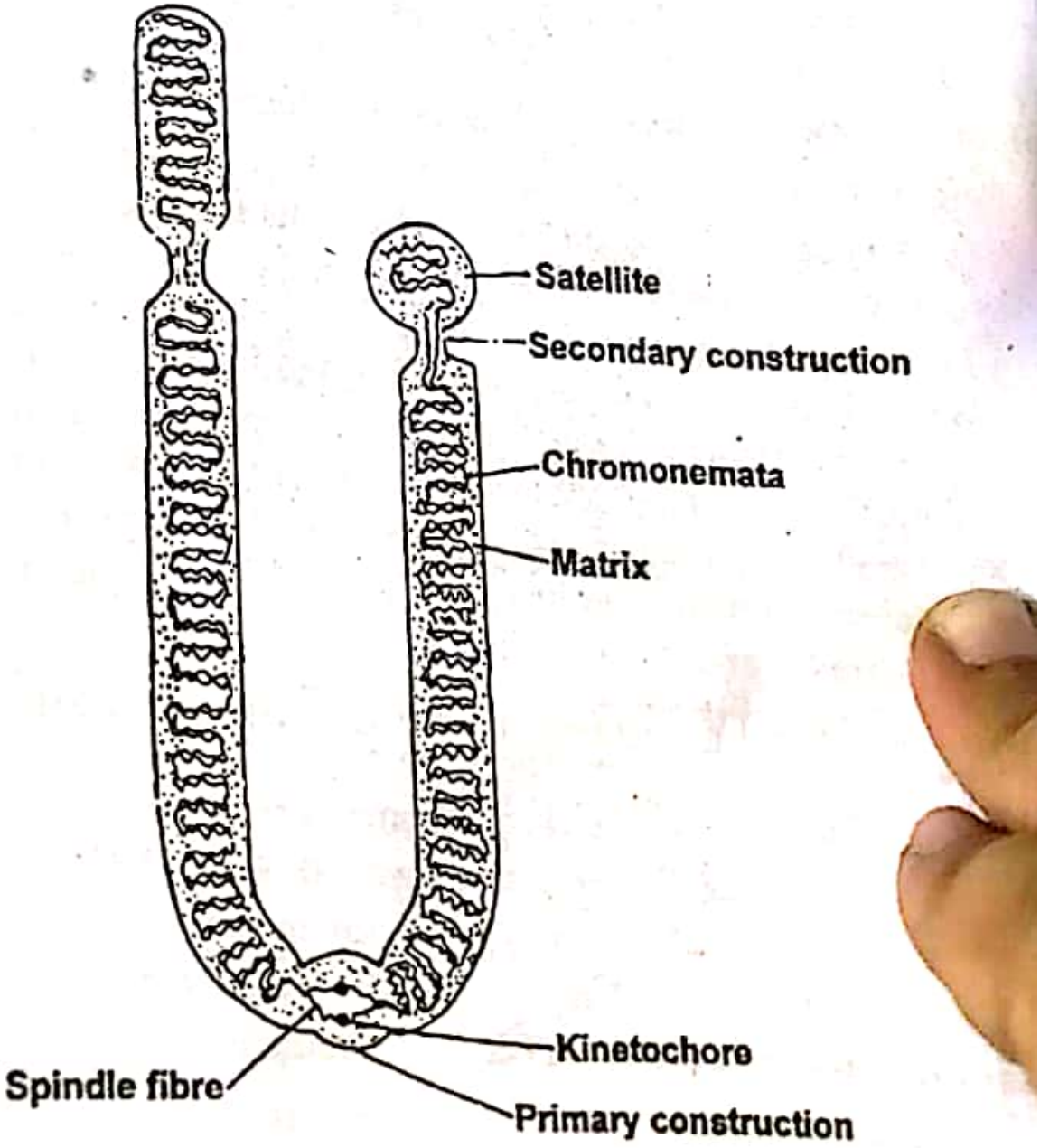


Fig. 7·1 : Structure of a somatic Chromosome at metaphase

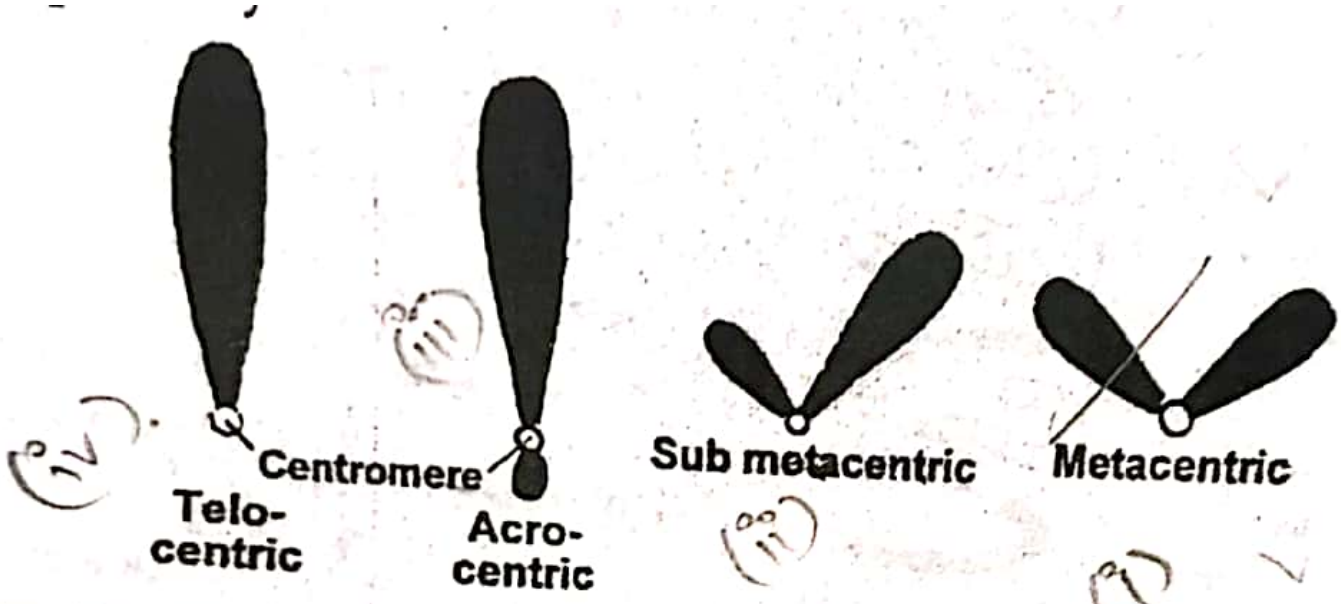


Fig. 7·3 : Types of chromosomes according to the position of the centromere

The position of centromere varies from chromosome to chromosome and it provides different shapes to the latter. They include :

(ii) *Metacentric chromosomes* : The centromere is located in the centre of the chromosome, forming two equal arms. These chromosomes appear as 'V' shaped during anaphase.

(ii) *Sub metacentric* : The centromere is located slightly away from the centre, and thus forming two unequal arms. Such chromosomes appear either as J - or L - shaped during anaphase.

(iii) *Acrocentric* : Centromeres located close to one end of chromosomes are known as sub - terminal, and the chromosomes having them are called *sub - telocentric* or *acrcoentric*. Acrocentric chromosomes may appear either as "J" or 'rod - shaped' during anaphase.

(iv) *Telocentric* : The rod - like chromosomes which have the centromere on the proximal end are known as telocentric chromosomes. Generally, telocentric chromosomes are unstable.

Structure of Centromere : The centromere consists of highly repititive DNA (heterochromatin), with special proteins attached

GENETICS

11

THE GENE

11-1 INTRODUCTION

The hereditary units which are transmitted from one generation to the next generation are called 'genes'. Their presence was first conceived by Mendel in 1865. He called them as *hereditary factors* or *elements*. The term '*gene*' was coined by Johannsen in 1909 for a single unit of heredity occupying a specific position on any chromosome. Initially genes were considered as discrete particles and chromosomes as strings of beads (Morgan, 1911). But the discovery of DNA molecule as the custodian of genetic information has discarded the Morgan's particulate theory of gene.

A gene may be defined as *a segment of DNA which has necessary information for the synthesis of a specific polypeptide or protein or RNA molecule.*

11-2 THE CONCEPT OF GENE

Understanding the structure of gene in terms of four genetic phenomena : transmission, recombination, mutation and gene function is known as *gene concept*. These criteria are interdependent: we can not observe gene function without gene mutation ; while on the other hand, we can not observe gene transmission without gene function. The concept of gene has been changing with the developments in the field of molecular biology and genetics. This changing scenario can be studied under three phases : Classical, Neo classical and Modern.

A Classical concept :

The gene concept was introduced by Sutton and Boveri in 1903. The studies of Morgan, Muller, Bridges, Sturtevant etc. elaborated it.

Thus the segments of a gene include - (i) a transcription unit - which includes the coding sequences (the introns), the flanking sequences (the leader and tailor sequence) and (ii) the regulatory sequences (which flank the transcription unit), which are necessary for its specific function.

11-3 STRUCTURE OF GENE

A gene has a complex structure having a number of different segments often referred to as *elements*. Each element has a specific function to perform during transcription.

A typical gene is made of two regions, the **regulatory region** and the **transcriptional unit.** The regulatory region contains the sequences essential for the transcription of the gene but are not transcribed themselves. **A transcriptional unit is a sequence of DNA transcribed into a single RNA,** starting at the *promoter* and ending at the *terminator*.

1. Genes determine the physical as well as physiological characteristics. They are transmitted as discrete particles from parents to offspring, generation after generation (Mendel, 1865).

2. Chromosomes preserve their individuality during cell division (Sutton and Boveri, 1904).

3. Genes are located on the chromosomes at specific positions in a specific chromosome. This position is known as *locus*.

4. Since the number of genes in each organism is very large in comparison to the number of chromosomes, several genes are located in each chromosome. For example, in man about 40,000 genes are located on 23 pairs of chromosomes.

5. Genes in the chromosome are arranged in a linear order like the beads on a string (Chromosome theory of inheritance; Morgan, 1911).

6. Genes located in the same chromosome are linked together and do not show independent assortment. The number of linkage groups is equal to the haploid number of chromosomes of that species.

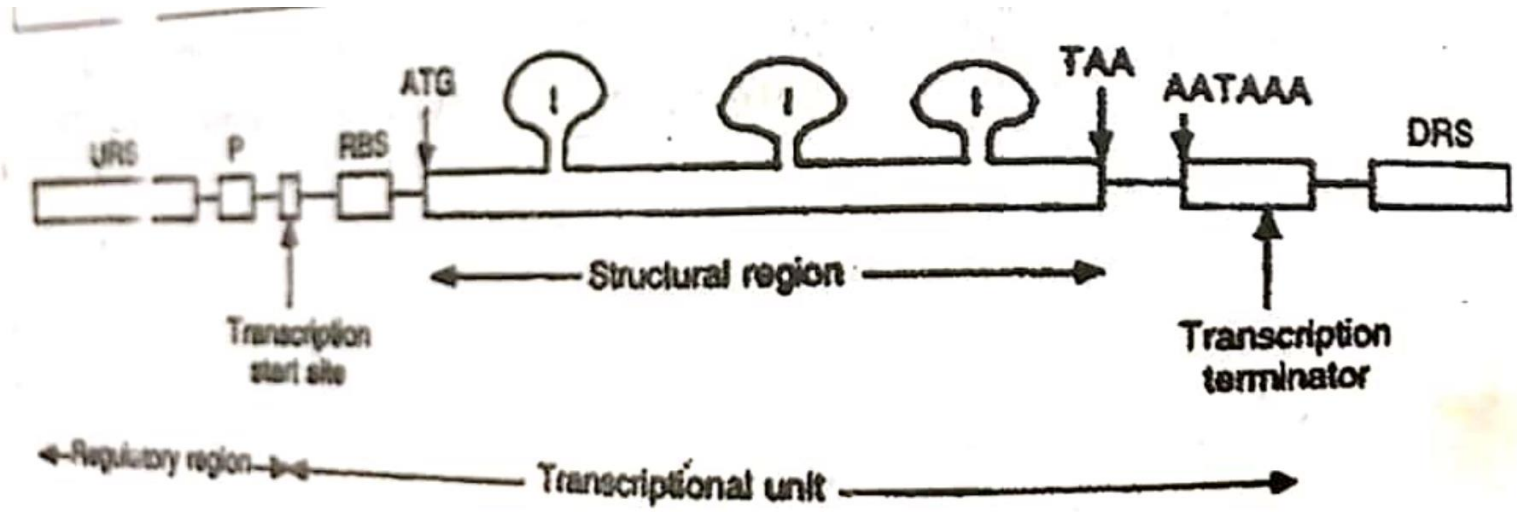
7. A gene controls the development of a specific character through the synthesis of one single enzyme. This is known as one gene - one enzyme hypothesis (Beadle and Tatum 1941).

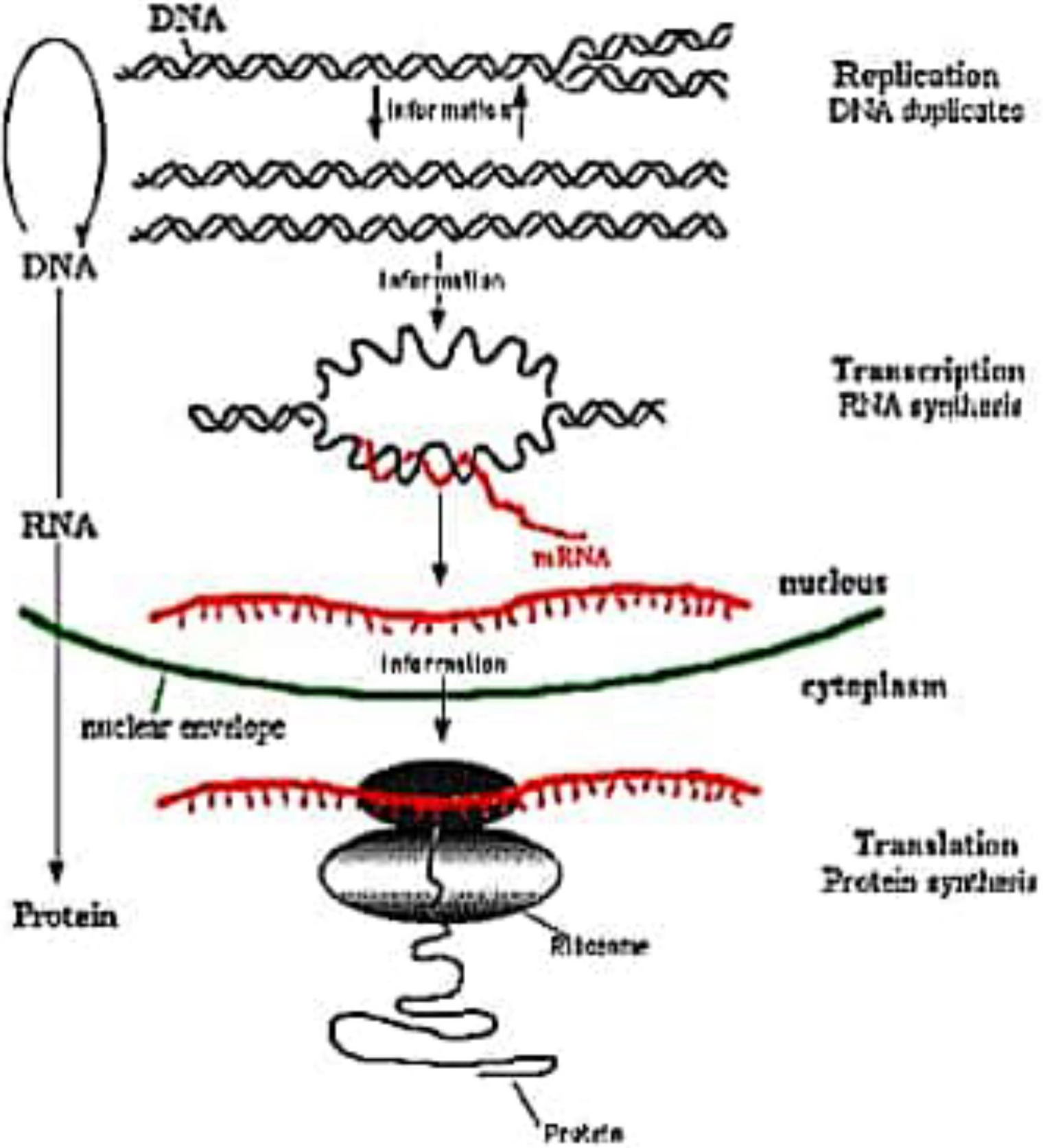
8 Mutation result in the formation of alternative forms a gene. These alternative forms are known as '*alleles*'. Many genes usually have only two alleles, one of them is **normal** (wild) and the other one is a **mutant**. For example, red and white colours of flowers in pea plant are the two alternative expressions of the gene for flower colour.

9. When several mutations occur, various alternative forms of a gene would result and these are known as **multiple alleles**. Whatever may be the number of alleles in a multiple allele series, only two of them are found in an individual, since chromosomes exist in homologous pairs in diploid individuals.

10. Genes from one chromosome may be exchanged or transferred to other chromosomes, which may be its counter part (crossing over), or a non - homologous chromosome (translocation). But different alleles of a gene do not show crossing over or recombination with each other.

11. A gene thus, is regarded as a unit of function, a unit of mutation and a unit of recombination.





The Central Dogma of Molecular Biology

The **central dogma of molecular biology** describes the two-step process, transcription and translation, by which the information in genes flows into proteins: DNA → RNA → protein. Transcription is the synthesis of an RNA copy of a segment of DNA. RNA is synthesized by the enzyme RNA polymerase.

Francis Crick

The central dogma of molecular biology is a phrase by **Francis Crick**, who proposed the double helix structure of DNA. It means that information passes from DNA to proteins via RNA, but proteins cannot pass the information back to DNA. **Crick** first wrote it in 1958, and repeated it in 1970.

Replication, Transcription, and Translation are the **three** main **processes** used by all cells to maintain their genetic information and to convert the genetic information encoded in DNA into gene products, which are either RNAs or proteins, depending on the gene.

CELL BIOLOGY

5 NUCLEIC ACIDS

5-1 INTRODUCTION

Nucleic acids are complex informational macromolecules. They are found in all living cells and viruses. The most common nucleic acids are - *deoxyribonucleic acid* (DNA) and *ribonucleic acid* (RNA). DNA is mainly found in the chromosomes, whereas RNA is present mainly in the cytoplasm and in ribosomes.

Nucleic acids were first isolated in 1868, by a Swiss scientist - Joseph Frederick Meischer from the nuclei of the pus cells on hospital bandages. He thought it to be a phosphorous rich nuclear protein and named it as *nuclein*. Later Altman (1889) coined the term *nucleic acid*.

Both DNA and RNA are large molecules and may occur either n single - or double stranded states.

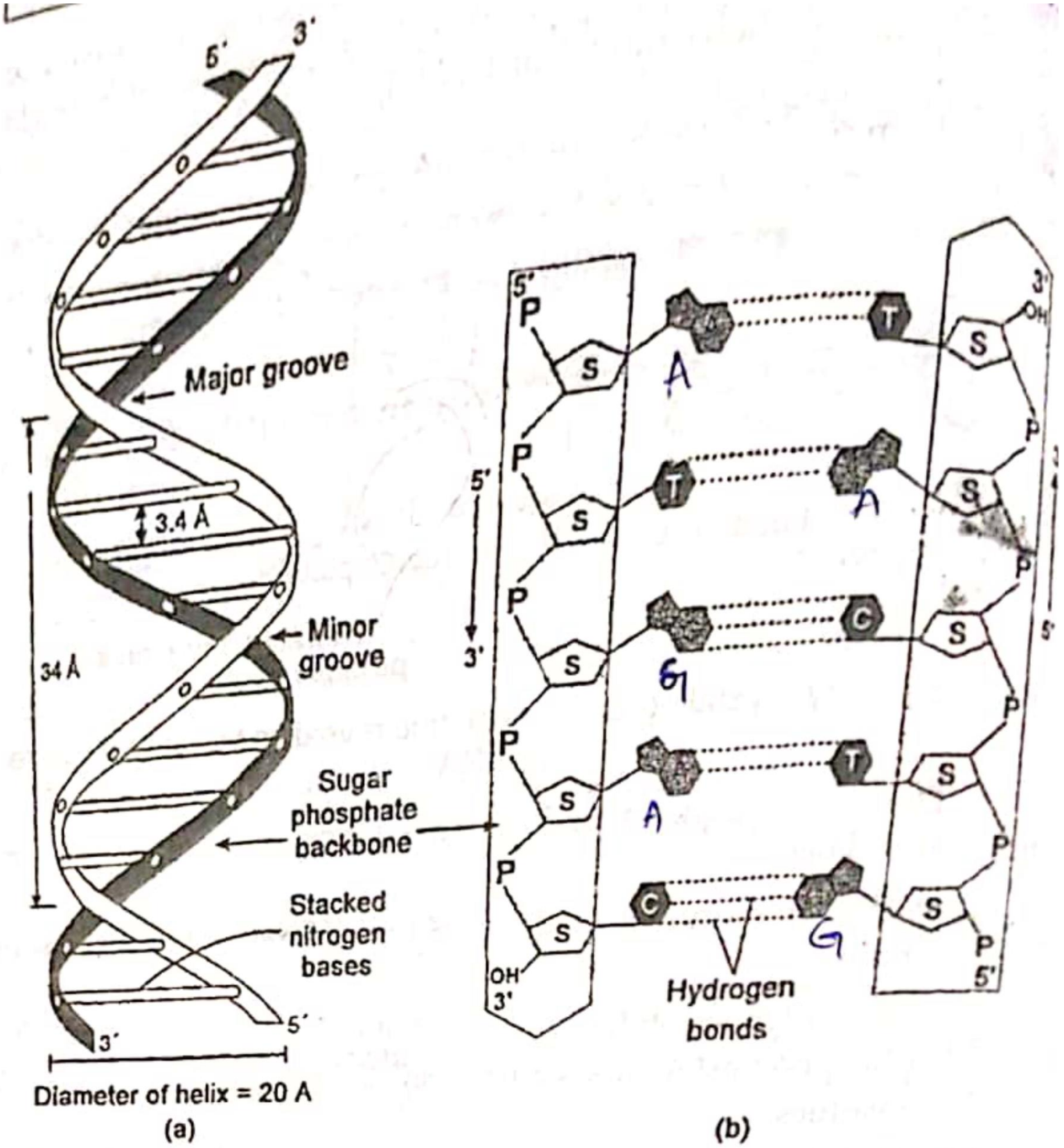


Fig. 5·6 : (A) Watson - Crick double helical model of DNA, (B) Molecular structure of DNA

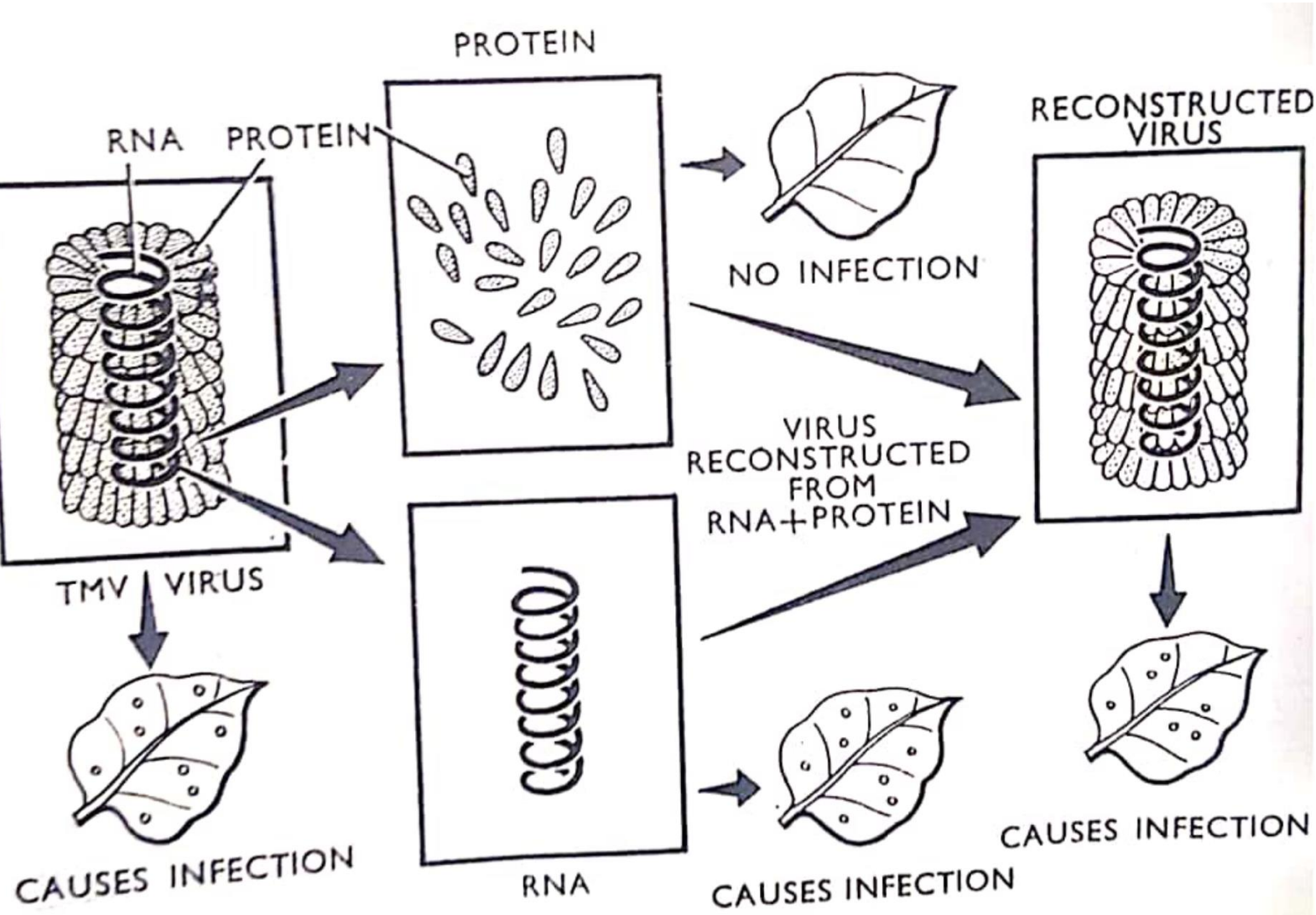


Fig. 18.6. Experiment of Fraenkel–Conrat on TMV showing RNA to be the hereditary material.

NUCLEIC ACID DNA AS GENETIC MATERIAL

Ans. Frederick Griffith (1928) conducted experiments on streptococcus pneumoniae and observed a transformation in bacteria. When streptococcus were grown on a culture plate, some produced smooth shiny colonies (s) while others produced rough colonies (R). Mice injected with 's' shain (mucous coat) die from pneumonia infection but mice injected with R strain do not develop pneumonia.

He injected heat killed 's' strain bacteria to mice, It is healthy. Finally he injected heat killed S and R strains, the mice died. He concluded that the R strain bacteria had been transformed by heat killed 's' strain bacteria. Some transforming principle transferred from heat killed strain to R strain to synthesize a mucous coat and become virulent. This is due to the transfer of genetic material.

2. Avirulent strain (R–II)—The pneumococcus cells of this strain do not produce symptoms of pneumonia. These lack a polysaccharide capsule. Their colonies have irregular appearance and are known as **rough (R)**.

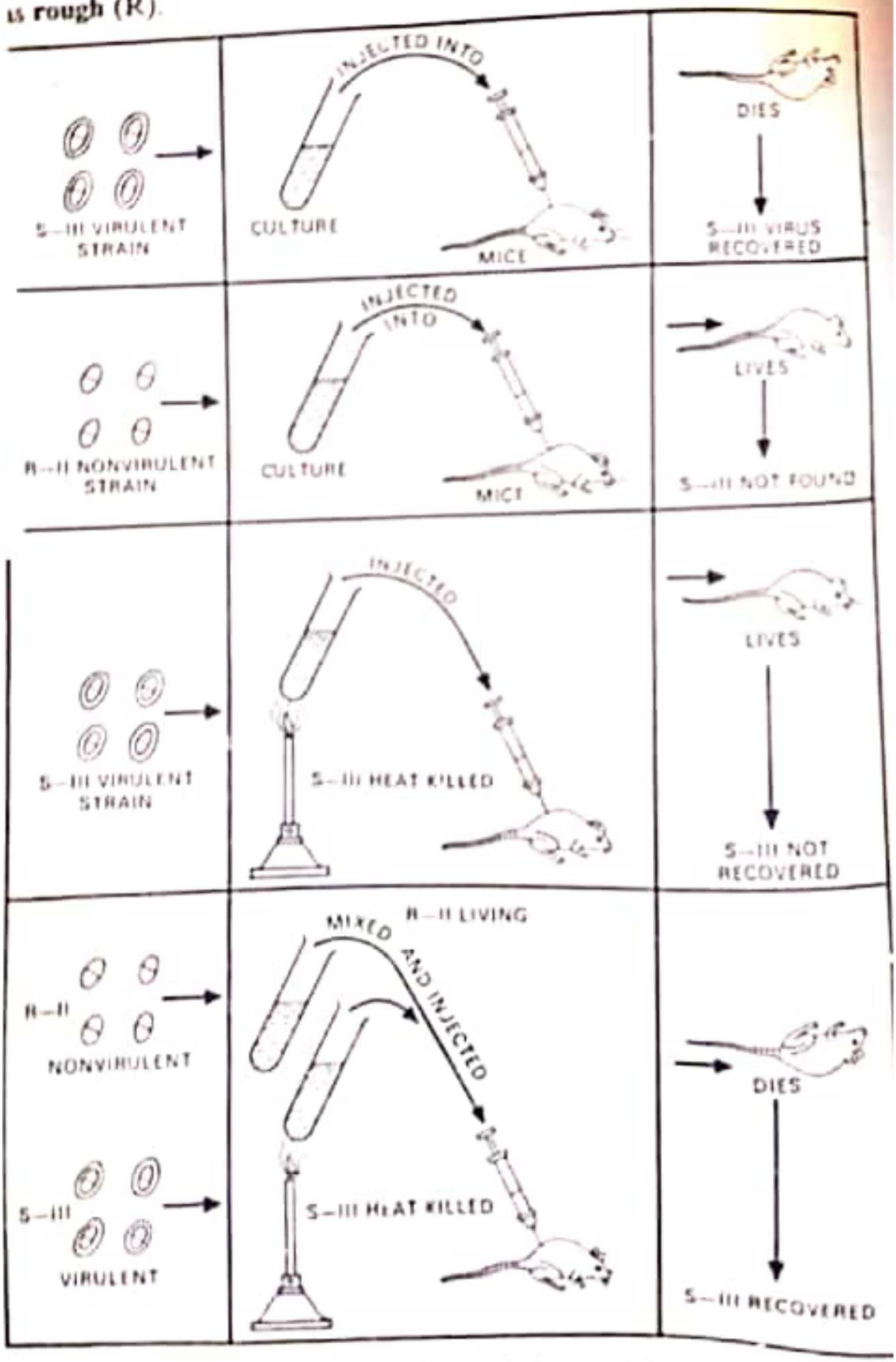


Fig. 18.1. Griffith's experiment to demonstrate genetic transformation in *Pneumococcus* bacteria.