



ZO(N)-202 & ZO(N)-202L

B.Sc. IV Semester
GENETICS, TAXONOMY & EVOLUTION



DEPARTMENT OF ZOOLOGY
SCHOOL OF SCIENCES
UTTARAKHAND OPEN UNIVERSITY

ZO(N)-202 & ZO(N)-202L

Genetics, Taxonomy & Evolution



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Content

Course: Genetics, Taxonomy & Evolution
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Unit number	Block and Unit title	Page number
	Block 1: Genetics	
1	Mendalism and Elements of heredity: Elements of heredity and variation, Mendel's principles of heredity, linkage (Coupling and repulsion), crossing-over (mechanism, theories and importance). Chromosomal mapping (Three point cross).	1-31
2	Chromosomal Mutation: Classification, Translocation, Inversion, Deletion, Duplication, Euploidy, Aneuploidy and Polysomy.	32-50
3	Genetic interaction: Gene structure and function.	51-67
4	Human genetics: Recessive inherited disorder, dominant inherited disorder, inborn errors.	68-90
	Block 2: Taxonomic Concept	
5	Taxonomy and Systematics: Introduction to taxonomy and its relationship with systematics. Importance and applications of biosystematics.	91-107
6	Zoological Nomenclature: International Code of Zoological Nomenclature, Bionomial and Trinomial components of classification.	108-124
7	Kinds of taxonomic characters and classification: Taxonomic characters: Morphological, Embryological, Cytogenetical, Biochemical and Numerical. Components of classification and Linnaean hierarchy.	125-140
8	Concepts of Species: Concept of species and speciation and potential modes of speciation.	141-157
	Block 3: Evolution	
9	Origin of life: Special creation theory, theories of spontaneous generation, cosmozoic theory, theory of chemical evolution and spontaneous origin of life at molecular level.	158-168
10	Concept of organic evolution: Concept of organic evolution: evidences from paleontology (types of fossils and determination of age of rocks and fossils), taxonomy, comparative anatomy, comparative embryology, physiology and biochemistry and cytology.	169-183
11	Theory of organic evolution: Theories of organic evolution: Lamarckism, Darwinism, Mutation theory and modern synthetic theory.	184-208
12	Evolutionary concept: Modern evolutionary Concept and details of micro, macro and mega evolution	209-218

13	Zoogeographical realms: Major Zoo-geographical realms and distribution patterns of animals in different zoogeographical realms. Biogeographical regions in India.	219-236
	Block 4: ZO(N)-202L	
14	Permanent slide preparation Permanent preparations / Minor dissections of the following: Protozoa: Paramecium, Porifera: Sponge spicules and gemmules. Coelenterata: Obelia colony, Obelia medusa, Arthropoda: Mouth parts of honey bee, butterfly, cockroach and grasshopper.	237-236
15	Cytological study a. Study of mitosis and meiosis using available material. b. Study of permanent slides showing stages of cell division, giant chromosome, , mitochondria, Golgi body etc	263-312
16	Genetics experiment Experimentation on Mendelian and non – Mendelian inheritance, study of mutants of Drosophila through charts/photographs	313-349

UNIT I: MENDELISM AND ELEMENTS OF HEREDITY

Contents

- 1.1 Objectives
- 1.2 Introduction
- 1.3 Elements of heredity and variation
 - 1.3.1 - Mendel's principles of heredity
 - 1.3.2 - Linkage
 - 1.3.3 - Coupling and repulsion
- 1.4 Crossing over
 - 1.4.1 - Mechanism of crossing over
 - 1.4.2 - Theories of crossing over
 - 1.4.3 - Importance of crossing over
- 1.5 Chromosomal mapping
 - 1.5.1 - Three point cross
- 1.6 Summary
- 1.7 Self-assessment questions
- 1.8 Terminal Questions

1.1 Objectives:-

Genetics is the study of **genes**, **genetic variation**, and **heredity** in living organisms. It is generally considered as a field of **Biology**, but intersects frequently with many other **Life sciences** and is strongly linked with the study of **information systems**. The father of genetics is “**Gregor Johan Mendel**” a late 19th-century scientist and **Augustinian friar**. **Mendel** studied “**trait inheritance**” patterns in the way traits are handed down from parents to offspring. He observed that organisms (**Pea plants**, *Pisum Sativum*) inherit traits by way of discrete “**units of inheritance**.” This term, still used today, is a somewhat ambiguous definition of what is referred to as a **gene**.

However, in this chapter you will be able to understand the heredity and variation, Mendel’s laws, linkage, crossing over and chromosomal mapping etc.

1.2 Introduction:-

The term **Genetics** was firstly introduced by “**William Bateson**” in **1906**. It has been derived from the Greek word **Gene** - which means “**to become**” or “**to grow into**”. Therefore, Genetics may be defined as “**the science of coming into being**” or study of heredity is called Genetics. **Genetics is the science of inheritance and variation.**

Hence, genetics may be redefined as the science that deals with the structure, organization, transmission and function of genes, and the origin of variation in them.

However, heredity may be defined “**as the transmission of traits from one generation to the following generations**”. It is the tendency on the part of the offspring to reproduce to characters of the parents.

The resemblance between individuals related by descent may be close, but it is never complete. **An offspring is never an exact copy of its parents.** Variation in heredity is observed in sexually produced offspring but not in asexually produced clones unless a mutation occurs. It is due to variations that each individual is unique in it and can be readily distinguished from another.

Heredity and variations which go side by side are the basis of **evolution**. The branch of science that deals with the facts and laws of **heredity** and inherited variations is known as genetics. Genetics is the science which tries to explain why living things resemble with their parents, and yet differ from them.

1.3 Elements of Heredity and Variation:-

The Austrian monk “**Gregor John Mendel**” is considered as the Father of “**Modern Genetics**”. **Mendel** made experiments on garden pea plants, species of *Lathyrus* (*Pisum sativum*). He started his work in **1856** and continued it up to **1863**. He presented or published the first report of his work in **1865** in the Annual Proceedings of “**Brunn Society for the Study of Natural Science**”.

His original paper “*Versuche Uber Pflanzenhybriden*” (Experiments on plant hybridization) was published in the Proceedings of the Society in **1866**. Mendel's work was, however, ignored at that time. This was perhaps because of the following reasons:

1. He published his work in an **obscure Journal**.
2. Scientists failed to notice his work because at that time the scientist world was busy in the controversy arisen by **Darwin's** theory of “**Origin of Species**”.
3. His ideas were ahead of his time as ignorance prevailed in that period about the **cytological basis of heredity**.

Mendel's published work remained unattained for about **34 years**. It received attention in **1900** when the same findings were independently rediscovered by three scientists namely **Hugo de Vries** from Holland, **Carl Correns** from Germany and **Von Tschermak** from Austria.

Mendel worked how characters are transmitted from one generation to following and how genes act together to control variable traits (**variations**) such as length, height, coat color, flower color etc. They developed the central concept of **genetics**. According to this concept, heredity is controlled by a large number of genes that are located on the chromosomes. These are called “**heredity vehicles**”.

During **1930s** beginning was made to apply **biochemical** and **biophysical** methods for the study of chemical nature of the gene. This led to a new branch of genetics “**Molecular Biology**”. This new approach led to the concept that genes are units of “**Biological Information**”.

Because of close association and interdependence between genetics and molecular biology the term “**Molecular genetics**” is now used. It is that branch of science that is concerned with the study of all aspects of the gene.

Mendel made Crosses between different varieties of a **garden pea**. He crossed these varieties which had contrasting traits or characters. In his simpler experiments, he crossed two plants differing in one character only, such a cross was called **monohybrid cross** and the hybrids thus produced called **monohybrids**. In more advanced experiments he crossed two plants differing in

two characters. Such a cross was called **dihybrid** cross and the resultant hybrids were known as **dihybrids**.

1.3.1 Mendel's Principles of Heredity:-

Since ancient times people knew that parental characters were inherited by the offspring, but did not know the mechanism of inheritance. Various beliefs were in vogue due to superstitions. It was given to **Johann Gregor Mendel** to explain the mechanism of inheritance with mathematical precisions.

Johan Gregor Mendel was a priest in the monastery of **Brunn**, a small village in **Austria** (now Czechoslovakia). He conducted a number of experiments with pea plant in the kitchen garden of the parish. He observed several contrasting characters in pea plants such as a tall variety and a dwarf variety, yellow seeds and green seeds, round seeds and wrinkled seeds etc.

These characters were handed down from generation to generation because the pea plants **are self-pollinated**. The seven contrasting characters that were taken into account by **Mendel** are as follow:

- | | |
|--------------------|---------------------------|
| 1. Seed shape | - Round or wrinkled |
| 2. Cotyledon color | - Yellow or green |
| 3. Seed coat color | - Colored or white |
| 4. Pod shape | - Inflated or constricted |
| 5. Pod color | - Green or yellow |
| 6. Flower position | - Axial or terminal |
| 7. Height | - Tall or dwarf |

Based on the observations of his experiments on garden pea, **Mendel** drew some important conclusions. These conclusions are known as **Mendel's Laws of Inheritance**. These are as follows:

1. Law of Dominance
2. Law of Segregation
3. Law of Independent Assortment
4. Law of Recombination

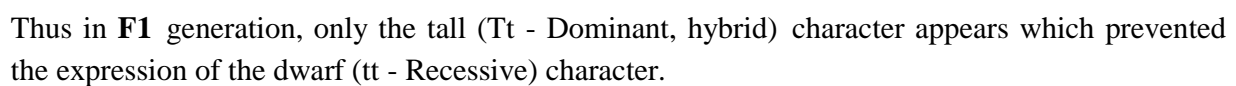
1. LAW OF DOMINANCE

In **monohybrid crosses**, he observed that **F1** offspring or monohybrids show characters or traits of only one parent. It simply indicates that out of two contrasting characters only one appears in

"One character or factor prevents the expression of other".

Example: Mendel crossed pure tall plants with the pure dwarf plants. The seeds thus obtained were sown which gave rise to tall plants:

Tall x Dwarf - Parent generation (P1)



After the observing the results of **F1** generation, Mendel experimented further and self-fertilized the flowers of **F1** plants or generation. The seeds thus obtained from these flowers were sown and developed into plants (**F2**). Mendel noted that all these plants were similar to the original plants i.e. **P1** generation and **F1** plants. They were found to be in a ratio of **3 : 1** (3 plants showing dominant character and 1 showing recessive character). This led Mendel to formulate his second law which is called as "**Law of Segregation**: or "**Law of Purity of Gametes**". It states that:

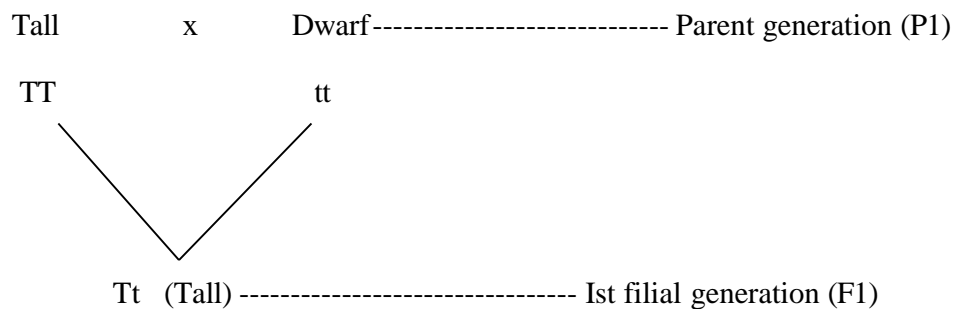
"The hybrids or heterozygote of F1 generation contain two contrasting characters of dominant and recessive nature. These characters do not mix with each other but segregate

or separate at the time of gamete formation in such a manner that each gamete receives only one character either dominant or recessive".

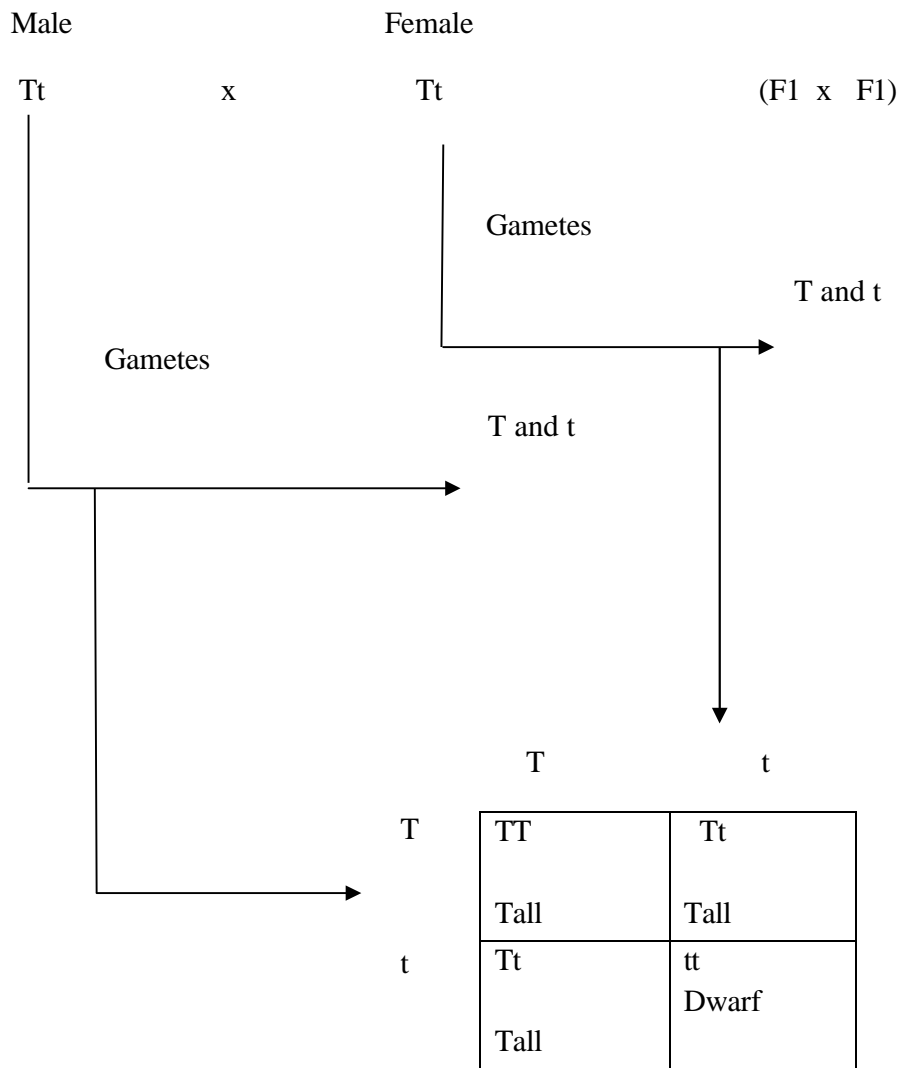
This law is also called as the law of purity of gametes because the gametes contain only one character and are pure for it.

Example: This law may be explained with an example of a garden pea. The hybrid of **F1** produced by the crossing of a **homozygous** (pure) tall plant and a homozygous (pure) dwarf plant was tall. The flowers of this tall plant on self-fertilization produced seeds which in **F2** generation developed into tall and dwarf plants in the ratio of **3:1**. Actually, Mendel obtained **787** tall and **277** dwarf plants. Their ratio is approximately **3:1**.

Cross - I



Cross - II



3 Tall : 1 Dwarf

1 TT : 2 Tt : 1 tt

Homozygous : Heterozygous : Homozygous

(Pure) (Hybrid) (Pure)

Tall Tall Dwarf

3. Law of Independent Assortment:-

After **monohybrid experiments Mendel** tried dihybrid crosses. For this **Mendel** crossed plants that differed in two characters. He crossed pea plant having yellow round seeds with the plant having green wrinkled seeds. In **F1** generation he obtained dihybrid which had yellow and round seeds (dominant hybrid).

But when he self-fertilized the plants developed by these seeds of **F1**, he did not find **3 : 1** ratio as was found in the monohybrid experiments. But in **F2** generation he found four types of seeds in the ratio of **9 : 3 : 3 : 1**. Out of four types of seeds, two types of seeds were like the original parents (**P1** generation) but two types quite new. They neither resembled the parents nor the hybrid of **F1**. By the observations of the dihybrid experiments, **Mendel** formulated his third law of **Independent Assortment**.

Mendel explained that the two characters (seed color and seed shape) are not tied together but they remain independent of each other. The round shape of the seed is not always associated with the yellow color; however, it may remain associated with the green color also. Consequent on these findings the law states that:

"The factors (now genes) for different pairs of contrasting characters segregate or assort independently of each other at the time of gametogenesis in F1 hybrid without affecting or diluting each other".

Example: When a pea plant having yellow and rounded seeds were crossed with the other having green and wrinkled seeds, in **F1** generation all the hybrid plants produced yellow and rounded seeds. When these seeds were sown, the plants developed, which were self-fertilized. After self-fertilization the plants produced **4 types** of seeds which appeared in the ratio of **9: 3: 3: 1**.

1. Yellow round - 9
2. Yellow wrinkled - 3
3. Green round - 3
4. Green wrinkled - 1

CROSS - I

Yellow Round

Green Wrinkled

YYRR

x

yyrr-----P1

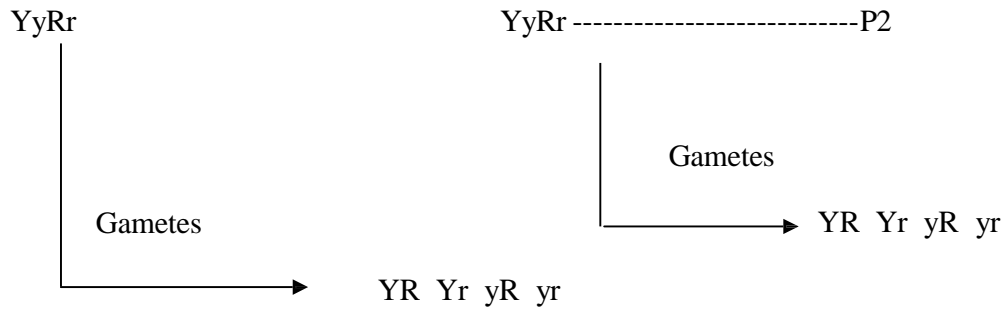
YyRr -----F1

Yellow Round (Dihybrid)

CROSS - II

Yellow Round (Male)

Yellow Round (Female)



		Female Gametes			
		YR	Yr	yR	yr
Male Gametes	YR	YYRR	YYRr	YyRR	YyRr
	Yr	YYRr	YYrr	YyRr	Yyrr
	yR	YyRR	YyRr	yyRR	yyRr
	yr	YyRr	Yyrr	yyRr	yyrr

The **phenotypic** ratio will be: 9 Yellow Round: 3 Yellow Wrinkled: 3 Green Round: 1 Green Wrinkled.

The **genotypic** ratio will be : 1 YYRR : 2YYRr : 1YYrr : 2 Yy RR : 4 YyRr 2 Yyrr : 1yyRR : 2 yyRr : 1yyrr

4. LAW OF RECOMBINATION

It is the **fourth** law of inheritance which states: "Various stable characters appearing in several generations of organisms are determined by an association (combination) of different heredity factors (genes). These factors undergo reshuffling according to the law of chance. This reshuffling produces as many combinations as are **mathematically possible**. Various possible combinations will result in the expression of various stable characters".

1.3.2 Linkage:-

In **1906 Bateson** and **Punnet** reported the first exception to **Mendel's law** of independent assortment. Although **Mendel** studied seven contrasting characters, all of these showed **independent assortment** during gamete formation. Thus, he was successful in formulating the law of independent assortment. The reason for this fact was that the alleles for Mendel's seven pair of characters are present in different **homologous** pairs of chromosomes.

It was, later on, found that independent assortment of **genes** does not take place always because a large number of genes located on the chromosomes are tied together (linked together) and they pass together from generation to generation. This tendency of genes to pass on to the next generation in groups is known as **linkage**. The phenomenon of linkage was discovered by **T. H. Morgan** in **1911** in *Drosophila melanogaster*. **T. H. Morgan** also proposed the "**Theory of Linkage**" in **1911**.

According to the "**Chromosome Theory of Inheritance**" proposed by **Walters S. Sutton** (**1903**), the genes are located in the chromosomes. Each pair of chromosome contains several genes. The genes located on the same chromosome cannot assort independently, rather these tend to **be inherited together**. This phenomenon of inheritance of genes together and to retain their parental combination even in the offsprings is known as **linkage**.

Thus, the linkage may be defined as **the tendency or nature of genes in the same chromosome to remain together during the process of inheritance**. The genes located on the same chromosome and being inherited is known as **linked genes**, and the characters controlled by these genes are **linked characters**. According to **T. H. Morgan**, the degree or intensity with which two genes are linked together is known as **linkage value**. The linkage value depends upon the distance between the linked genes on the same chromosome.

All the genes which are located on the single chromosome constitute a **linkage group**. The total number of linkage groups in an organism is equal to the number of chromosome pairs. For example, there are 4 linkage groups in *Drosophila melanogaster*, 23 pairs in man and 7 in sweet pea.

TYPES OF LINKAGE

The linkage is found in all animals and plants. There are **two types** of linkage:

1. Complete linkage
2. Incomplete linkage

1. COMPLETE LINKAGE

Complete linkage is exhibited when the genes for a particular character are present very close to one another. It is due to non- break in the gene combination situated on a chromosome. Such cases in which **linked genes** are transmitted together to the offsprings only in their original or parental combination for two or more or several generations exhibit complete linkage. In such cases, the linked genes do not separate to form the new or non-parental combinations. This phenomenon is very **rare** in nature.

Example: The best example of complete linkage is **male Drosophila**. When a Drosophila fly with grey body and long wings is crossed with one having a black body and vestigial wings, all the **F1** offspring's produced are having grey bodies and long wings because grey color is dominant over black color and long wings are dominant over vestigial wings.

However, if a male from **F1** generation is back crossed with a double recessive black vestigial female, we should expect **four kinds** of offspring's in equal number as the result of **independent assortment**. But there are only **two types** of offspring's which resemble the two **grandparents**.

The results indicate that grey body character is inherited together with the vestigial wings. It means that these genes are linked together. Similarly, black body character is associated with the long wings. In the above example, the offspring's exhibit only the parental combinations on characters, since any **non-parental combinations** are not found.

Ultimately it may be concluded that the **genes for grey body and long wings are linked together and thus show complete linkage**.

Male

Grey body Long winged

GL/GL

GL

X

GL/gl 3

Grey body Long-winged

Female

Black body Vestigial winged

gl/gl--

--P1

 $_gl$

- Gametes

- F1

CROSS -II

F1 Individual

GL/gl

X

Recessive Individual

gl/gl

Gametes

Gametes

GL, gl

gl, gl

Gametes

Gametes

gl

gl

GL

GL/gl

GL/gl

gl

gl/gl

gl/gl

50% Grey Long and 50% Black Vestigial

Incomplete linkage occurs when the genes for different characters are separated at the time of gamete formation due to breaking and exchange of chromosomes pieces during **meiosis**. However, in the majority of cases, the homologous chromosomes undergo breakage and reunion during **gametogenesis**.

Example: The incomplete linkage is exhibited by female *Drosophila*, tomato, maize, pea, poultry and man etc. In sweet pea, blue flower color and long pollen character exhibit incomplete linkage. When a **sweet pea** variety with blue flowers (B) and long pollen (L) is crossed with another variety having red flowers (b) and round pollen (l), F1 individuals (BbLl) produced blue flowers and long pollen. These F1 individuals when crossed with plants having red flowers and round pollen (bbll) we find the test cross frequency as shown below;

Blue flowers and Long pollen

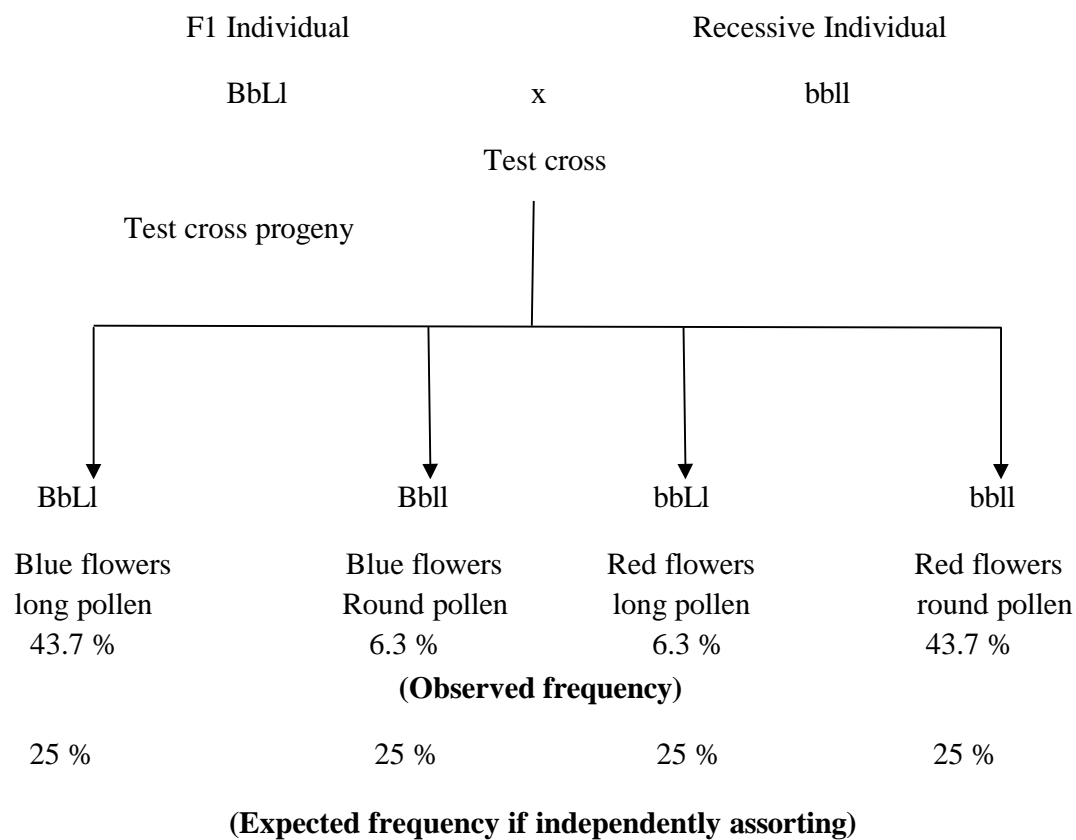
Red flowers and Round pollen

BbLl x bbl----- P1

BL bl----- Gametes

BbLl-----F1

Blue flowers long pollen
(Hybrid)



1.3.3 Coupling and Repulsion:-

The process of linkage was first described by **Bateson** and **Punnet in 1906** in pea plant, and describes it as **coupling**. They found that the results of dihybrid cross in sweet pea, *Lathyrus odoratus*, involving color and shape of pollen grains, do not agree with the law of independent assortment. The results obtained are shown in the table below:

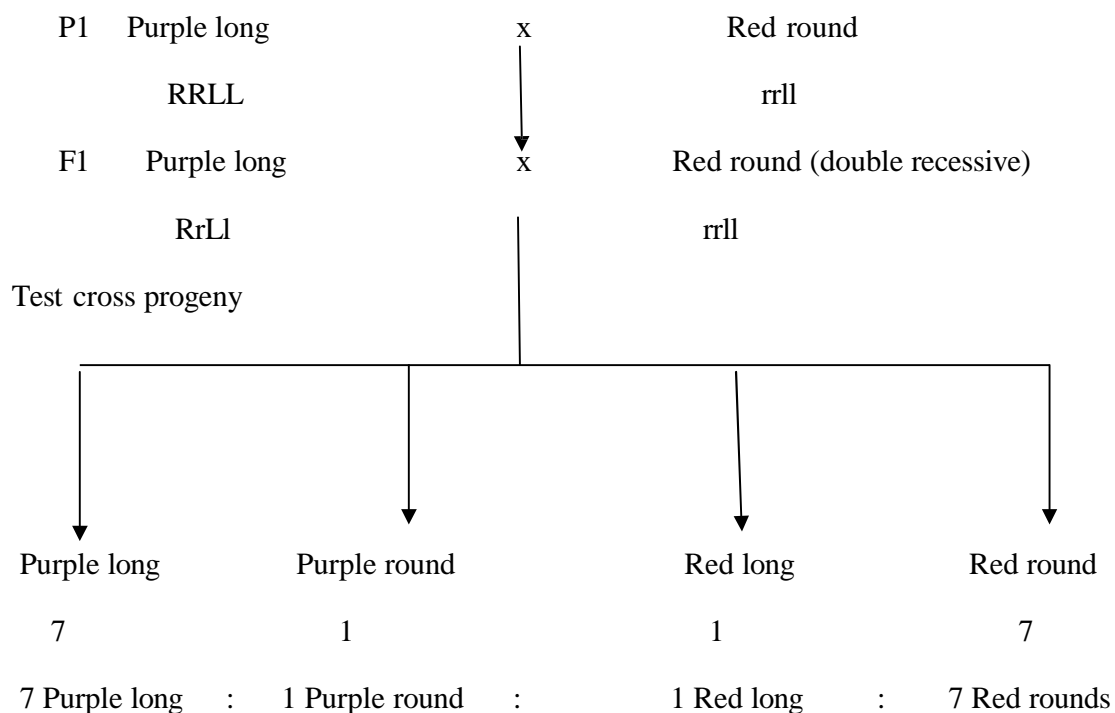
CROSS - I

P1 Purple flower, long pollen x Red flower, round pollen

F1 **Purple flower, long pollen**

Phenotype	Number	Ratio
P2 Purple long	296	11
Purple round	29	1
Red long	27	1
Red round	85	3

When these **F1**, purple, long (heterozygous) hybrid were crossed with the double recessive red and round (homozygous) individuals (test cross) failed to produce expected **1 : 1 : 1 : 1** ratio in **F2** generation. These actually produced following four combinations in the ratio of **7 : 1 : 1 : 7**.



The above results of the test cross indicate that the parental combinations are seven times more numerous than the non -parental combinations. **Bateson and Punnet** ultimately concluded that:

"The alleles coming from the same parent tend to enter the same gamete and to be inherited together (genetic coupling). Similarly, the same genes coming from two different parents, tend to enter different gametes and to be inherited separately and independently (**repulsion**)".

1.4 Crossing Over:-

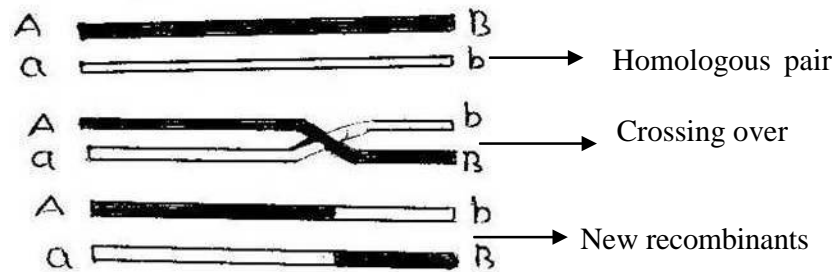
According to **Morgan** genes are located on the chromosomes in a **linear fashion**. These genes are linked to a specific point of a chromosome and inherited from one generation to next generation with **parental combinations**. But sometimes **non-parental combinations** also appear. It means linked genes do not always stay together and they are separated between a homologous pair of chromosomes during **meiosis**.

As a result, **reciprocal exchange** of genes is take place between the homologous pair of chromosomes during chiasma formation in the process of meiosis. This reciprocal process of exchange of genes during **chiasma formation** in meiosis **Morgan** described as **crossing over**.

Hence, crossing over may be defined as "**the recombination of linked genes by interchanging of corresponding segments between the non-sister chromatids of a homologous pair of chromosomes**". The chromosomes which take part in the process of crossing over, are called **cross overs** while other as **non-crossovers**.

In other words "**crossing over is a process, which involves an interchange of corresponding segments between non-sister chromatids of homologous chromosomes, resulting in a recombination of genes. The chromatids after such interchanges of chromosomal parts are known as cross overs**".

Crossing over is a highly precise process as the two chromatids exchange exactly equivalent segment and except in very rare instances neither they lose nor gain any gene. At each chiasma, the **two non-sister chromatids** exchange their section so that the chromosomes carried in the gametes are new as they carry genes which were originally not located in them.



Diagrammatic interpretation of crossing over
Between two chromatids

Fig.1.1

Types of Crossing Overs:-

The types of crossing over depend upon the number of **chiasmata** present in the chromosomes. However, the following three types of crossing over have been recognized depending on the number of chiasmata.

1. Single crossing over
2. Double crossing over
3. Multiple crossing overs

1.4.1 Mechanism of Crossing Over:-

Regarding mechanism of crossing over, a widely accepted model was proposed by **Whitehouse** and **Hasting in 1965**, called **Whitehouse model**. According to this model whole process of crossing over may be divided into four distinct steps:

- A. Synapsis
- B. Duplication
- C. Crossing over
- D. Terminalization

A. SYNAPSIS

During the **zygotene** substage of prophase - I of meiosis the homologous chromosomes (paternal and maternal) come too close each other and starts to pairing. This pairing is the point- to- point like the closing of a **zip** called **synapsis**. First pairing starts at the centromere region and on to the arms. The pairing is very precise and occurs due to the mutual attraction between non-sister chromatids of the pairing homologous chromosomes.

As the results, the pairs of homologous chromosomes are called **bivalents**. Synapsis is an event of great importance because it is the basis of heredity and variations.

B. DUPLICATION

Duplication of chromosomes is followed by synapsis in which bivalent nature of chromosomes changes into **tetravalent**. The phenomenon of duplication is always taking place in **pachytene** substage of prophase - I of meiosis. During this substage each chromosome is become much thickened and longitudinally divided into two chromatids. As each chromosome now has two chromatids, called **bivalents** and the pair of the homologous chromosome containing four chromatids called **tetrad condition**.

C. CROSSING OVER

During the process of crossing over, internal chromatids also called as **non-sister chromatids** of a homologous pair of the chromosome, first break at corresponding points. This breakage of **chromatids** is usually taken place due to the activity of a nuclear enzyme called **endonuclease**.

Now, broken segments of each chromatid connect with opposite broken chromatids in such a way, that both of these non-sister chromatids cross each other at a certain point and became exchange. The fusion or attachment or connection of broken segment with opposite chromatids is taken place due to the action of an enzyme called **ligase**.

Both broken segments cross each other at a certain point by making a cross (**x**) like structure called **chiasma** (Plural Chiasmata) and this phenomenon usually described as chiasma formation. In a real sense, this chiasma formation is called **crossing over** because, at this point, heredity material in the form of broken segments is becoming an exchange.

As a result, **chiasma formation** (crossing over) could take place at **several points** between non-sister chromatids of a pair of the homologous chromosome and several chiasmata could also be formed. During the process of crossing over a little amount (about **0.3 %** of total genome) of **DNA**, synthesization is taken place, which is used in the repairing of broken chromatids of a pair of homologous chromosome.

D. TERMINALIZATION

Crossing over is followed by **terminalization**. After the completion of crossing over, the force of attraction during synapsis between non-sister chromatids became **weak** and the start to **repeal** each other. This repulsion or separation of chromatids start from the centromere towards the ends and the chiasmata also move in **zipper-like** fashion towards the ends. This movement of chiasmata is called **terminalization**.

Ultimately, as a result in the last substage of prophase -I of meiosis, chromatids became condensed, thickened and now became separate into a separate chromosome.

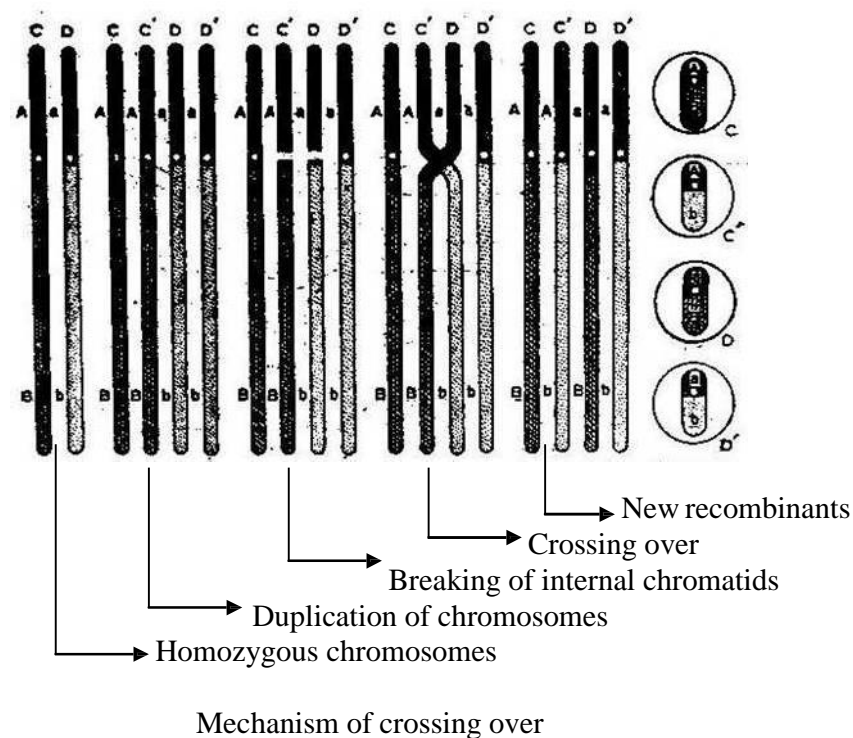


Fig.1.2 Mechanism of Crossing Over

1.4.2 Theories of Crossing Over:-

Various **geneticists** proposed different theories of crossing over. However, some important theories regarding mechanism of crossing over are as following.

A. Classical theory

- B. Chiasma type theory
- C. Copy choice theory
- D. Break and exchange theory

A. CLASSICAL THEORY

The Classical theory proposed by **L.W. Sharp in 1932** in his book namely "**Introduction to cytology**". According to this theory, there is no formation of chiasmata at the meeting point of non - sister chromatids where crossing over takes place. It means **chiasmata are not the result of crossing over** but it is the result of **breakage and rejoining at points of overlap**. But in actual condition there is the formation of chiasmata take place. In this view, adjacent loops have equational and reductional separation of chromatids.

B. CHIASMA TYPE THEORY

This theory was proposed by **F. A. Jansens in 1909** and further extended by **J. Belling** and **C. D. Darlington**. Chiasma type theory was the just **reverse** of classical theory. However, according to this theory, chiasma formation is the result of crossing over. First of all, non-sister chromatids break at their corresponding point. After breaking they reunite and formed chiasma.

Therefore, it can be said that **chiasmata are the result of crossing over where genetic material exchange** and formation of new recombinants are taken place. In this theory, there is reduction in the loop on either side of a chiasma. Thus it may be concluded that chiasma is the result of crossing over.

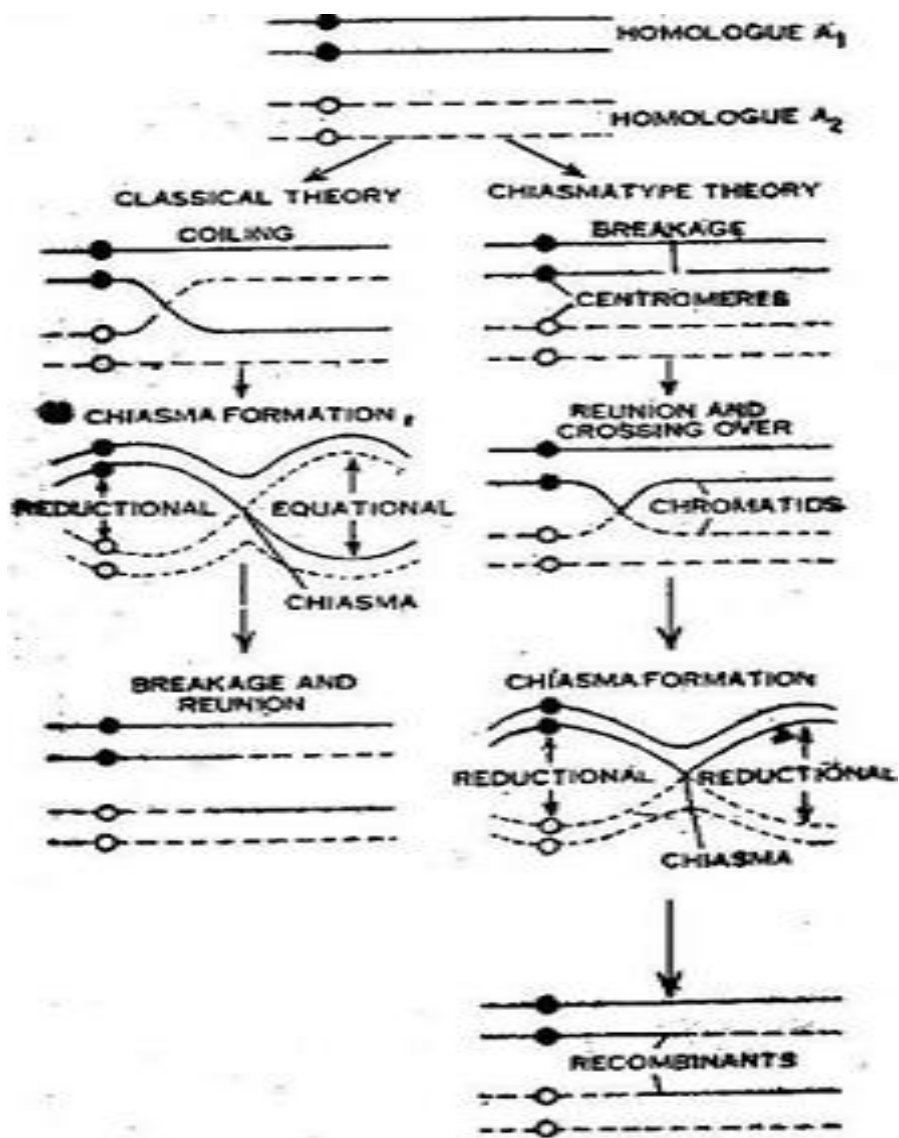


Fig.1.3 Diagrammatic representation of classical and Chiasma type theory

C. COPY CHOICE THEORY

This theory was advanced by **Belling in 1932** for the explanation of crossing over. According to him crossing over is not the result of breakage and reunion of chromatids but it is the result of following two steps:

1. Formation of new genes (duplication)
2. Formation of new connections between the formed new genes (i.e. formation of new chromatids)

Parental genes are situated on **paternal** and **maternal** chromatids act as templates upon which new genes are synthesized and then gene interconnections are developed in the form of a thread. During the synthesis of new genes in front of the pre-existing genes and connections between them, the chromatids of homologous chromosomes intercross, resulting in the formation of chiasma at that point. Thus, new chains of genes will take part in crossing over and represented as a cross over gametes.

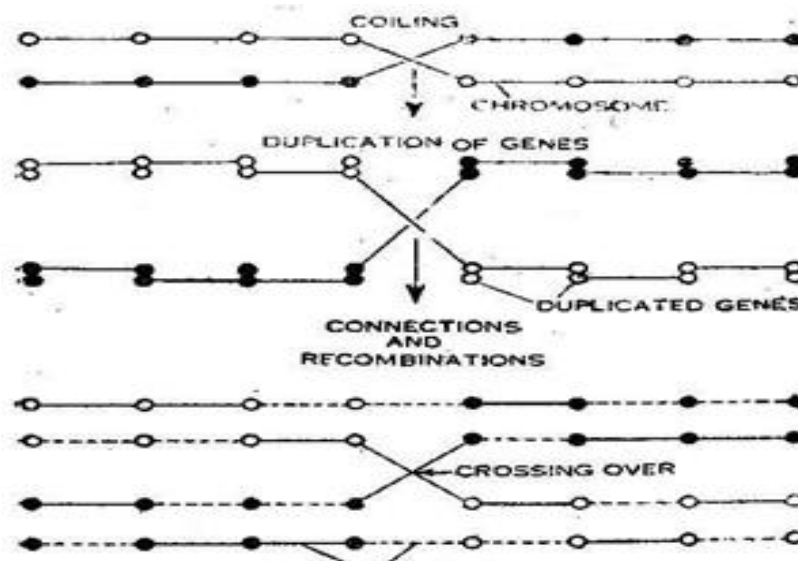


Fig.1.4 Copy Choice Theory

D. BREAK AND EXCHANGE THEORY

This theory was proposed by **Muller** and the **widely accepted theory** regarding mechanism of crossing over. According to this theory, bivalent chromosome became duplicated in **pachytene** substage of prophase I of meiosis I. Now each bivalent containing four chromatids called a **tetrad**. In tetrad condition internal chromatids or non-sister chromatids of a paternal and maternal break at their **corresponding point** by the activity of **endonuclease** enzyme and reunite with the opposite segments due to **ligase** enzyme.

In this process, there is the formation of the cross (x) like structure between non-sister chromatids called chiasmata and here genetic material became an exchange. During this process, there is the formation of a little amount of **DNA (0.3 % of total genome)** is taking place, which is used in the repairing of broken parts of chromatids.

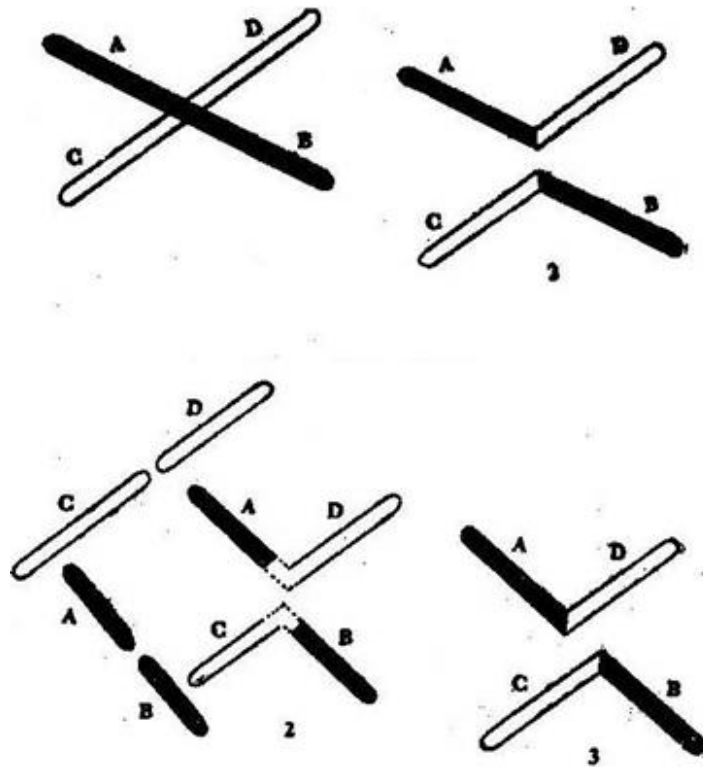


Fig.1.5 Break & Exchange theory

1.4.3 Importance of Crossing Over:-

The phenomenon of crossing over is observed in all groups of organisms from man to virus. It has following genetic importance or significance:

1. The **frequency of crossing over** is of great use in the construction of linkage maps for **genetic maps** of chromosomes.
2. The phenomenon of crossing over provides direct evidence for **linear arrangements of genes** in the chromosomes.
3. The process of crossing over provides the nature and working mechanism of genes.

4. Crossing over is responsible for new **genetical recombination** (variations) which are the raw material of organic evolution.
5. Crossing over plays a very important role in plants and animals **breeding**.
6. Crossing over provides an **inexhaustible store** of genetic variability in sexually reproducing organisms.
7. Crossing over helps in the development of **new characters**.
8. Crossing over also provides new **gene combinations**.

1.5 Chromosomal Mapping:-

The **representation** in the figure of the relative position of genes on the chromosome is called **chromosome map** and the process of identifying gene **loci** is called **mapping**. The chromosome map is based on two important assumptions:

1. That gene are arranged in a linear fashion and
2. That the percentage of crossing over (recombination frequencies) between the two genes is an index of their distance apart.

The relationship between the crossover frequency and the distance between loci was first suggested in **1913** by **A. H. Sturtevant**. Thus the chromosome map is a condensed **graphic representation** of the relative distance between the linked genes, expressed in percentage of recombination among the genes in one linkage group. The distance between genes can be expressed in "**map unit**", where one map unit is defined as **1** percent recombination.

Therefore, the chromosomal mapping may be defined as "**The chromosome maps are condensed graphic representations of the relative distances, expressed in percentage of recombination, among the genes in one linkage group, consequently located on a single chromosome**".

1.5.1 Three Point Cross:-

Bridges and Olbrycht proposed linkage in genes **ec**, **sc** and **cv** in *Drosophila*.

Gene **ec** represent rough (echinus) eyes

Gene **sc** represents scute (few bristles missing)

Gene **cv** represent cross veinless wings

These genes are present on **x-chromosome** and are recessive. When flies with all these three genes were mated with normal flies, all **F2** were normal **phenotypically** but **genotypically** the females had one x-chromosome with gene **ec+** and the allele's **sc** and **cv** and the other x-chromosome with allele **ec** and genes **sc+** and **cv+**.

When these female of **F1** were crossed with the male having all the three recessive genes (i.e. **ec**, **sc** and **cv**) the **F2** offsprings were markedly different from the expected ratio. If these genes were not linked, the offspring's of **F2** generation according to the law of **independent assortment** must have been of eight different types, all in equal numbers. But in actual experiment the result was different:

Noncross over's

- | | | | |
|---------------------------------------|--------------------|---|-----|
| 1. Nonscute, echinus, cross veined | (sc+ ec cv+) - 40% | } | 80% |
| 2. Scute, nonechinous, cross veinless | (sc ec+ cv) - 40% | | |

Single cross overs

- | | | | |
|---------------------------------------|---------------------|---|------|
| 1. Scute, echinus, cross veined | (sc ec cv+) - 3% | } | 7.6% |
| 2. Nonscute, nonechinus, cross veined | (sc+ ec+ cv) - 4.6% | | |

Double cross overs

- | | | | |
|---------------------------------------|----------------------|---|-------|
| 1. Nonscute, nonechinus, cross veined | (sc+ ec+ cv+) - 0.1% | } | 0.15% |
| 2. Scute, echinus, cross veined | (sc ec cv) - 0.05% | | |

From the above results, the frequencies of recombination or crossing over between the genes scute (sc), echinus (ec) and cross veinless (cv) are:

sc - ec - 7.6%

ec - cv - 9.7%

sc - cv - 17.3%

Therefore, it means these genes are arranged in a line and are separated in the case where crossing over has occurred.

1.6 Summary:-

1. Genetics is the study of genes, genetic variation, and heredity.
2. Gregor Johan Mendel is known as the father of modern genetics.
3. Gene is known as the unit of inheritance.
4. Term genetics firstly coined by Bateson in 1906.
5. Heredity is the science of transmission of genetic character or traits from one generation to next generation.
5. Heredity is also known as heredity vehicle.
6. Heredity is also known as the basis of evolution.
7. Mendel used 7 pairs of contrasting characters in his experiments.
8. Law of segregation is also known as the law of purity of gametes.
9. Phenotypic and genotypic ratio is found in the law of segregation are 3 : 1 and 1 : 2 : 1 respectively.
10. 9 : 3 : 3 : 1 ratio is found in the law of independent assortment.
11. Mendel used factor term at the place of the gene.
12. T. H. Morgan in 1911 proposed theory of linkage.
13. Walters S. Sutton proposed chromosome theory of inheritance in 1903.
14. Complete linkage is rarely found in nature.
15. The incomplete linkage is very common and found in all organisms from virus to a man.
16. Coupling and repulsion are two phase of linkage.
17. Genes are situated on the chromosome in a linear fashion.
18. The chromosome which takes part in crossing over is called crossover while other as non-crossovers.
19. Chiasma formation is the result of crossing over.

20. Breakage of chromatids at their corresponding points is taking place due to the activity of endonuclease enzyme.

21. The linkage may be defined as **the tendency or nature of genes in the same chromosome to remain together during the process of inheritance.**

22. The genes located on the same chromosome and being inherited is known as **linked genes**, and the characters controlled by these genes are **linked characters**.

23. There is 0.3 % of DNA of total genome is synthesized in the process of crossing over.

24. Chromosome map is a condensed graphic representation of the relative distance between the linked genes.

1.7 Self Assessment Questions:-

1. The scientific study of heredity, variations and the environmental factors responsible for them is known as
 - a) Physiology
 - b) Genetics
 - c) Evolution
 - d) Ecology

Ans - (b)

2. The mechanism of transmission of characters, resemblances as well as differences from the parental generation to the offspring is called
 - a) Conversion
 - b) Heredity
 - c) Both a and b
 - d) None of the above

Ans - (b)

3. The term gene (unit of heredity) was coined by
 - a) Hugo De Vries
 - b) Wilhelm Johannsen
 - c) Darwin
 - d) Gregor Johan Mendel

Ans - (b)

4. Alternative form of a gene is called
- a) Bivalent
 - b) Allele
 - c) Diploid
 - d) None of the above

Ans - (b)

5. Monohybrid cross refers
- a) The cross between 2 parents differing in a single contrasting characters
 - b) The cross of F1 hybrid with its parents
 - c) The cross between 2 parents differing in two pairs of contrasting character
 - d) None of the above

Ans - (a)

6. Phenotypic ratio of F2 generation in incomplete dominance
- a) 1:2:1
 - b) 3:1
 - c) 9:3:3:1
 - d) None of the above

Ans -(a)

7. A genetic phenomena, in which both the alleles (dominant and recessive) are equally expressed in the Hybrid
- a) Incomplete dominance
 - b) Co-dominance
 - c) Dominance
 - d) None of the above

Ans - (b)

8. The phenomena of independent assortment refers to
- a) Expression at the same stage of development
 - b) Unlinked transmissions of genes in crosses resulting from being located on different chromosomes

- c) Association of an RNA and a protein implying related function
- d) Independent location of genes from each other in an interphase cell

Ans -(b)

9. Law of segregation also refers to

- a) Law of purity of characters
- b) Law of complete purity
- c) Low of purity of gametes
- d) None of the above

Ans - (c)

10. A cross that involves the analysis of two independent traits is

- a) Monohybrid cross
- b) Dihybrid cross
- c) Digametic cross
- d) Both b and c

Ans - (b)

11. Which of the following is a mismatch?

- a) Phenotypic F2 ratio of 1:2:1 codominance
- b) Genotypic F2 ratio of 1:2:1 complete dominance
- c) Genotypic F2 ratio of 1:2:1 codominance
- d) Phenotypic F2 ratio of 3:1 partial dominance

Ans - (d)

12. A phenotype that is not genetically controlled but looks like genetically controlled one is called

- a) Phenotype
- b) Phenocopy
- c) Both a and b
- d) None of the above

Ans - (b)

14. The cross of a F1 hybrid with its recessive parent is known as

- a) Back cross
- b) Test cross
- c) Hybrid cross
- d) Monohybrid cross

Ans - (b)

15. Mendel's law of genetics was rediscovered by scientist

- a) Hugo De Vries, Carl Correns, Bateson
- b) Hugo De Vries, Carl Correns, Erich Tschermak
- c) Carl Correns, Bateson, Darwin
- d) Johansen, Robert Brown, Tschermak

Ans - (b)

1.8 Terminal Questions:-

- 1- Write a short note on law of dominance
- 2- Write a short note on coupling and repulsion
- 3- What is the importance of crossing over
- 4- Write a short note on chromosome maps
- 5- What are the Mendel's Principles of Heredity? Elaborate.
- 6- Write a detail account on linkage
- 7- Write a detail account on crossing over

1.9 References:-

- Griffiths, Anthony J. F.; Miller, Jeffrey H.; Suzuki, David T.; Lewontin, Richard C.; Gelbart, eds. (2000). "Genetics and the Organism" An Introduction to Genetic Analysis (7th ed.). New York: W. H. Freeman.

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- Griffiths, Anthony J. F.; Miller, Jeffrey H.; Suzuki, David T.; Lewontin, Richard C.; Gelbart, eds. (2000). "Nature of Crossing Over". An Introduction to Genetic Analysis (7th ed.). New York: W. H. Freeman.

1.10 Suggested Readings:-

1. Beale, G. Knowels. J. 1978. Extranuclear Genetics. Oxford & IBH Publ. New Delhi.
2. Dobzhanski, T. H. 1951. Genetics and Origin of Species, 3rd edn. Columbia Univ. Press, New York.
3. Bruce Alberts; Dennis Bray; Karen Hopkin; Alexander Johnson; Julian Lewis; Martin Raff; Keith Roberts; Peter Walter (2013). Essential Cell Biology, 4th Edition. Garland Science.
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UNIT 2: CHROMOSOMAL MUTATION

Contents

- 2.1 Objectives
- 2.2 Introduction
- 2.3 Classification of chromosomes
 - 2.3.1 Translocation
 - 2.3.2 Inversion
 - 2.3.3 Deletion
- 2.4 Duplication of chromosomes
 - 2.4.1 Euploidy
 - 2.4.2 Aneuploidy
 - 2.4.3 Polysomy
- 2.5 Summary
- 2.6 Self-assessment questions
- 2.7 References
- 2.8 Suggested readings
- 2.9 Terminal questions

2.1 Objectives:-

Chromosomes are filamentous bodies which are typically present in the **nucleus** and which become visible during **cell division**. They are the carriers of the genes or **units of heredity**. **Chromosomal mutation** represents the structural change in the chromosome which appears **phenotypically**. However, in this chapter, you will be able to read about mutation, chromosomal mutation, Translocation, Inversion, Deletion, Duplication, Euploidy, Aneuploidy, and Polysomy etc.

2.2 Introduction:-

During the cell divisions, the **chromatin network** of the **interphase** nucleus condenses to form thick rod like structures known as **chromosomes**. The name chromosome was given by **Waldeyer in 1888**. The chromosomes play an important role in transmission of **heredity characters** from one generation to another. Thus chromosomes can be defined as:

"The individualized protoplasmic units present in definite number, capable of self-reproduction, maintaining their individuality, morphology and physiological properties throughout, play an important role in heredity".

The term **mutation** was first introduced by a well-known Dutch Botanist **Hugo de Vries in 1902** while working on a plant **Evening primrose** (*Oenothera lamarckiana*). He used word mutation for **spontaneous inheritable changes which occur suddenly and alter the phenotype of an organism**.

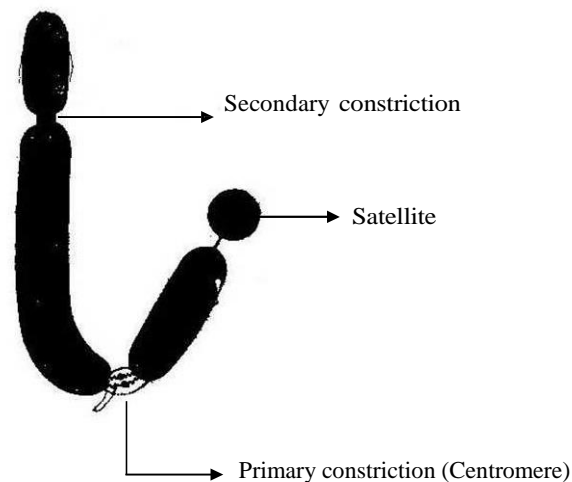
He performed several experiments on plant evening primrose up to eight years continuously and concluded that suddenly occurred spontaneous inheritable changes in plants and animals are the principle cause of **"origin of new species"**.

However, in present condition mutation may be defined as **"large spontaneous inheritable sudden changes in the genotype which alter the phenotype of an individual"**. These mutations may be spontaneous or induced mutation.

2.3 Classification of chromosomes:-

Chromosome number varies from species to species. However, it is constant for a particular number. Size is constant for every species. It ranges from **0.1μ to 30 μ** in length. The shape varies at different phases of cell division. They may be rod-shaped **twisted or spiral, curved or filamentous**. Each chromosome is comprised of following parts:

1. Pellicle and Matrix
2. Chromonemata
3. Centromere
4. Chromomeres
5. Satellite bodies.



Morphology of Chromosome

Fig.2.1 morphology of Chromosome

However, chromosomes may be classified in the following category on the basis of centromeres present.

1. **Monocentric:** The chromosomes which have only **one centromere** are called monocentric.
2. **Dicentric:** The chromosomes having **two centromeres** are termed as dicentric chromosomes.
3. **Polycentric:** In these, the chromosomes possess **many centromeres**.
4. **Acentric:** In these, the chromosomes **lack centromeres**.

Like number, the **position of centromeres** also varies. Depending upon the position of the centromere the chromosomes are of following types:

1. **Telocentric:** When the centromere is situated on the **proximal end or terminal end**, it is called telocentric. This type of chromosome is rod-like.

2. Acrocentric: When the centromere occupies **subterminal end**, it is called acrocentric. It is also rod-like in which one arm is very much smaller than the other.

3. Submetacentric: In **submetacentric** chromosome, the unequal in length giving J or L shaped appearance to the chromosome.

4. Metacentric: In metacentric chromosomes, the centromere lies **exactly in the center** of the chromosome. These chromosomes are V-shaped having equal arms.

5. Diffused or Non-Located: In diffused chromosomes, the centromere is in **distinct** and remains **diffused throughout** the length of the chromosome.

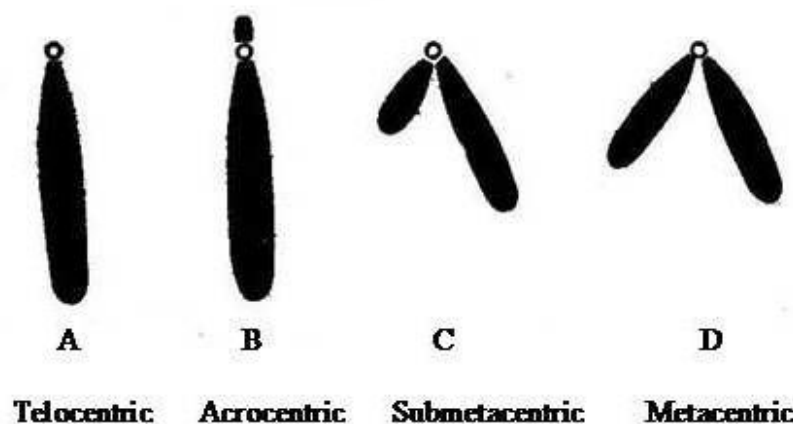


Fig.2.2 Classification of Chromosome on the basis Centromeric position

Chromosomal Mutation

Mutation is a heritable change in the structure of a gene or chromosome or a change in chromosome number. Accordingly, mutations are of three types:

1. Gene mutation
2. Chromosomal mutation
3. Genomic mutations or polyploidy

Hereditary characters are due to the effects of genes. Sometimes, a slight slip occurs in the replication of genes and this change in gene duplication is known as **gene mutation** or **point mutation**. The structural changes in the chromosomes which appear **phenotypically** are called **chromosomal mutations** or **chromosomal aberrations**.

Chromosomal mutations are inheritable and commonly occur in plants and animals. In this type of mutations, changes do not occur in the number of chromosomes but involve the **changes in the sequence of genes** which are located on the chromosomes. These changes were first analyzed by **H. J. Muller (1928)** in *Drosophila* and by **Barbara Mc Clintock (1930)** in *Zea*.

Genomic mutations involve **variations in chromosome number** of a whole genome. Variations in chromosome number (heteroploidy) are mainly of two types namely **euploidy** and **aneuploidy**.

2.3.1 Translocation:-

A **segment** of the chromosome with several genes may get cut off and then get attached to a different non-homologous chromosome is called **translocation**. Therefore, a change in the position and sequence of genes takes place but not in number. **It results in a change in the sequence and position of genes but not their quantity.**

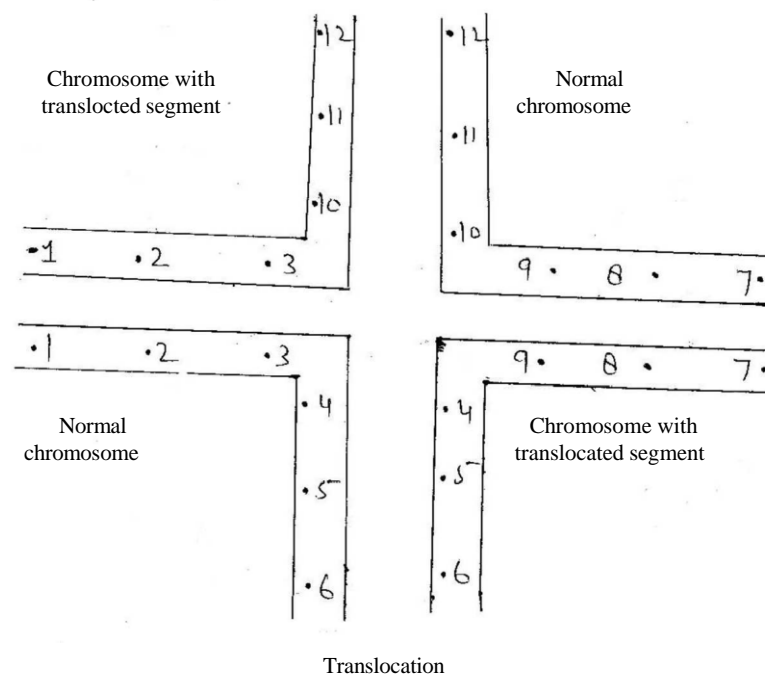


Fig.2.3 Translocation

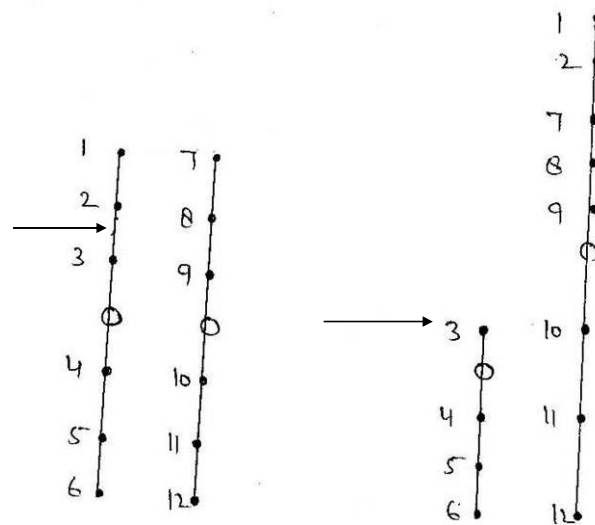
Translocation may be of following three types:

1. Simple translocation
2. Shift translocation
3. Reciprocal translocation

1. SIMPLE TRANSLOCATION

In this type of translocation **chromosome break at a single point** and broken part get attached to one end of another **non-homologous chromosome**.

For example, in below chromosome part 1 and 2 get attached to another non-homologous chromosome.



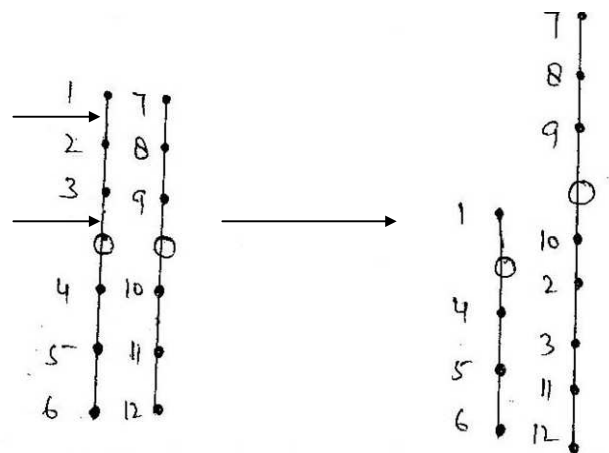
Simple translocation

Fig.2.4 Simple translocation

2. SHIFT TRANSLOCATION

In this type of translocation chromosome first break at **two points** and broken part gets **inserted** at any point of the non-homologous chromosome.

In present example first chromosome break at two point and broken part 2 and 3 get inserted in another non-homologous chromosome between 10 and 11 points.



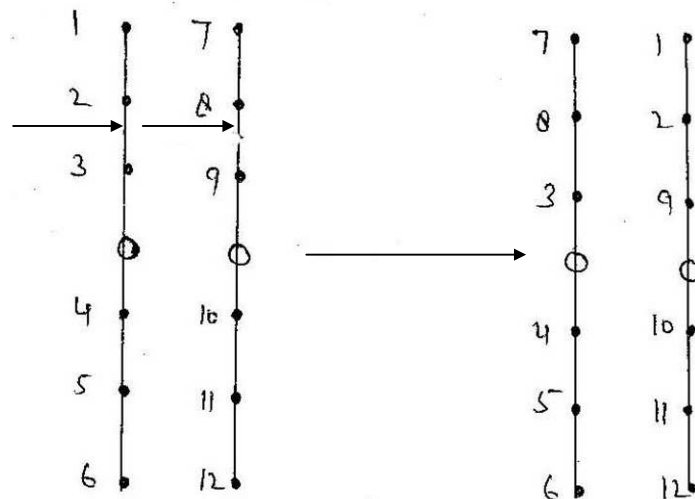
Shift translocation

Fig 2.5 Shift Translocation

3. RECIPROCAL TRANSLOCATION

In this type of translocation, there is **no gain or loss of any part of the chromosome**. Broken part of each non-homologous chromosome becomes exchange. This type of translocation is most frequently found during meiosis.

For example, between two chromosomes 1, 2 and 7, 8 part of both chromosome become break and get attached at the terminal end of both chromosomes respectively. Thus there is no loss or gain in between both chromosomes.



Reciprocal translocation

Fig 2.6 Reciprocal translocation

2.3.2 Inversion:-

When a **part or segment of the chromosome** containing genes rotates on its own axis by **180 degrees** called an **inversion**. **Breakage** and **reunion** both are essential for inversion. Sometimes, a chromosome may break at two points and then become reunited at the same point in a reverse order.

For example, In chromosome A part 4, and 3 is inverted. During the pairing of chromosomes in zygotene substage of meiosis -I, chromosome a is become inverted at this point at 180 degrees as represented in C chromosome, while other B chromosomes in the form of D chromosome become curved at this portion.

During pairing, **repulsion** occurs at the part where the **genes do not match**. This is also called as "**position effect**". Thus, as a result, there is neither a gain nor loss in the genes but a rearrangement of the sequence of the gene take place. Inversion may be of following two types:

A. Paracentric inversion

B. Pericentric inversion

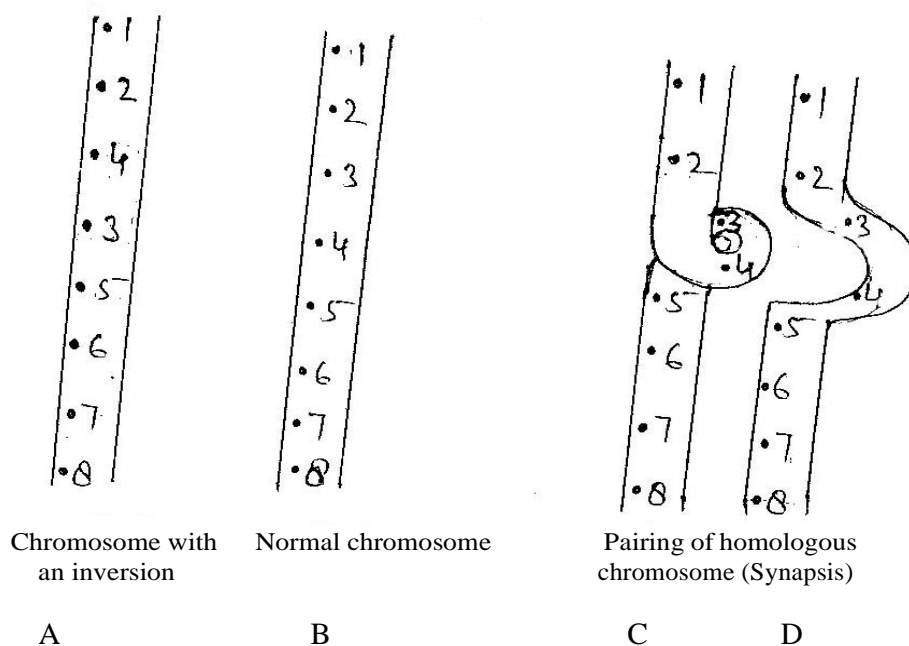


Fig.2.7 Inversion in chromosomes

A. Paracentric inversion: In this type of inversion, the **centromere** is located outside the **inversion loop**. When a cross over occurs within the loop, one product contains a

centromere and the other does not. **At anaphase**, this results in an abnormal chromosomal '**bridge**' and a loss of an entire chromosomal section.

B. Pericentric inversion: Here centromere is located inside the **inversion loop**. When a cross over between two chromatids occurs within the inversion loop, in the resulting chromatids there are some genes in double number while others are missing. Due to this **imbalance**, the cell is not viable.

Thus, if the normal sequence of genes in a chromosome is A B C D E F G, The sequence in **paracentric** and **pericentric inversions** will be A B C D G F E and A E D C B F G respectively.

Inversion has been useful in establishing and maintaining **heterozygous condition** because in inversion heterozygotes crossing – over is suppressed and only parental progeny is produced. **Recessive lethal** can be of added advantage because **heterozygotes** for them are viable but **homozygotes non-viable**.

2.3.3 Deletion:-

When any part or section of a chromosome which containing either one gene or block of genes, is being **lost** called **deletion or deficiency**. Various type of **structural changes** are occurring during **meiosis** (reduction division) and these changes may be recognized in the pairing of homologous chromosomes (**synapsis**) in zygotene substage of prophase –I of meiosis.

When a chromatid breaks at **two places** and the end portion fuse leaving out the central point, called **intercalary deletion**, while terminal, called as **deficiency or deletion**. When a deletion occurs in one member of a homologous pair, the members will become unequal in length.

Genic balance is usually disturbed due to deletion and this affects the **phenotype**. Deletion can be recognized by distortions of chromosomes during meiotic pairing of homologous chromosomes. Due to a terminal deletion one of the paired chromosomes appears to be much longer than the other, whereas due to **intercalary deletion**, the normal chromosome forms a loop near the deficient region of its homolog as an only identical regions pair with each other.

In present A chromosome part 3, 4 is becoming deleted and during the synapsis, chromosome B becomes curved.

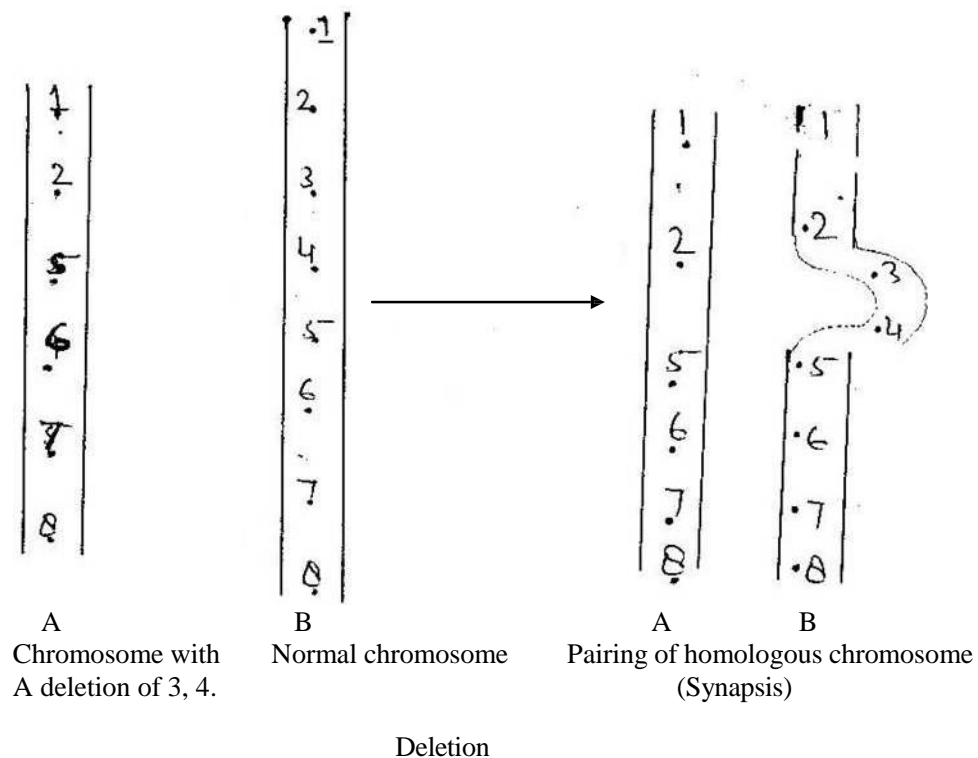


Fig.2.8 Deletion

2.4 Duplication of Chromosomes:-

The presence of a **part of a chromosome** in excess of the normal complement is known as **duplication**. A deleted part or segment of broken part or section of a chromosome attaches itself to a normal homologous or non-homologous chromosome in the presence of a centromere it behaves like an **independent chromosome** and gets included in an otherwise normal nucleus.

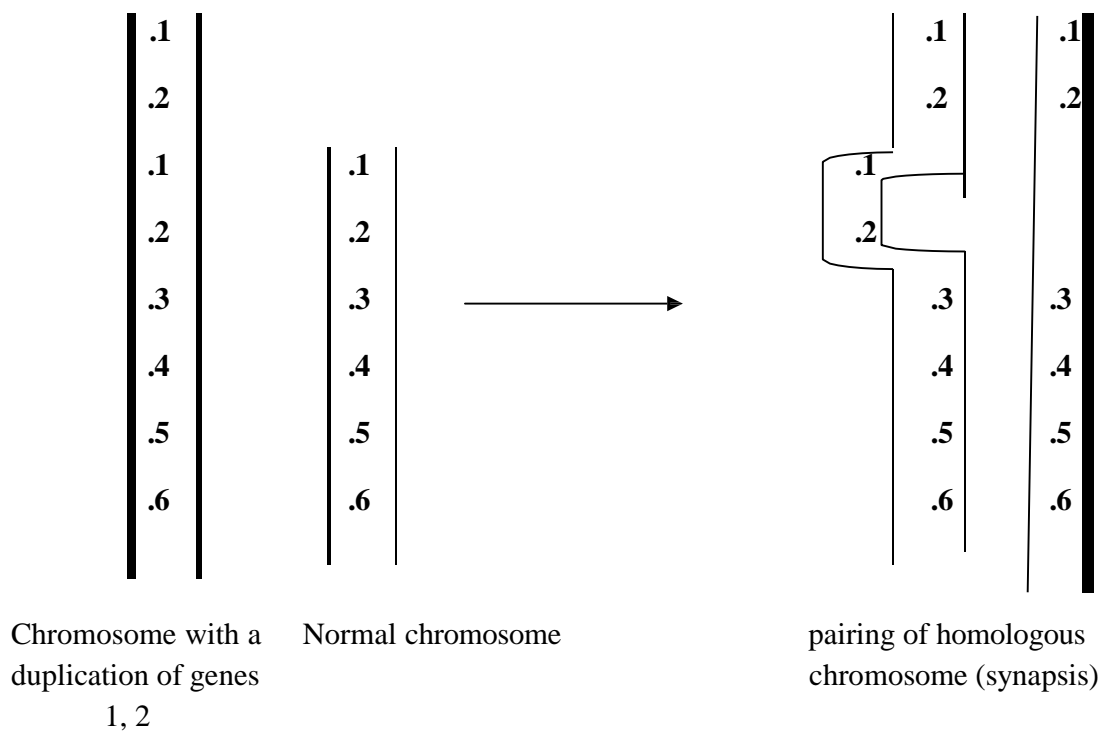


Fig.2.9 Duplication

Depending on the mode of joining of the duplicated region to a chromosome or its independent existence, duplication can be of the following types:

- A. Extra Chromosomal:** In the presence of a centromere, the duplicated part of the chromosome may behave as **an independent chromosome**.
- B. Tandem:** in this case, the duplicated region is situated just by the side of the normal corresponding section of the chromosome and the sequences of genes are the same in the normal and duplicated regions.
- C. Reverse Tandem:** in this case, the sequence of genes in the duplicated section of a chromosome is just the reverse of the normal sequence.
- D. Displaced:** here, the duplicated section is not adjacent to the normal section.
- E. Transposed:** in this case, the duplicated section is attached to a non-homologous chromosome.

like deletions, duplications also result into unequal or looped out configuration at the time of pairing of homologous chromosomes.

2.4.1 Euploidy:-

Variations that involve an entire set of chromosomes are known as **euploidy**. Euploids have one or more complete genomes, which may be identical with or distinct from each other. In the diploid state, two copies of the same genome are present in the somatic cells; it is represented as **2x**. Euploid variations are designated with reference to the **diploid (2x)** state. Some important euploid types are as following:

A. Monoploidy: Monoploids have a single basic set of chromosomes, e.g. $2n = X = 7$ in barley and $2n = X = 10$ in corn. Haploid, on the other hand, represent individuals with half the somatic chromosome number found in the normal individual. In **haploids**, each chromosome is represented only once due to which there is no **zygotene pairing** and all the chromosomes appear as univalent on a metaphase plate at **meiosis -I**.

During **anaphase - I**, each chromosome moves independently of the other and goes to either of the two poles. Haploids may originate:

1. Due to the **parthenogenetic** or **androgenic** development of gametes.
2. Due to chromosome loss in **hybrid embryos** and
3. By **pollen culture**.

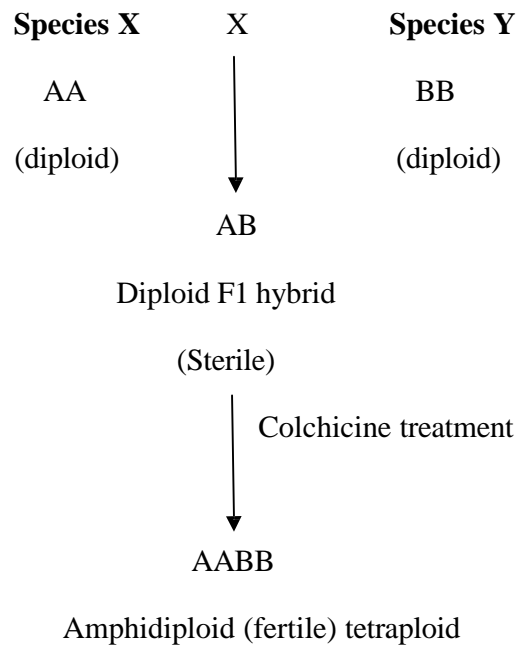
The most important use of **haploids** is in the production of homozygous diploids.

B. Polyploidy: In **polyploids**, each chromosome is represented by **more than two homologs**. Failure of normal mitotic divisions results into nuclei with increased sets of chromosomes.

Depending on whether polyploids are produced by the multiplication of chromosome sets that are initially derived from a single species or from two different species, they are of two types, **autopolyploids**, and **allopolyploids**.

1. Autopolyploidy: Autopolyploids are those **polyploids** which have the same basic set of chromosomes multiplied. For instance, if a diploid species has two similar sets of chromosomes or genomes (**AA**), an autotriploid will have three similar **genomes (AAA)** and an autotetraploid four such **genomes (AAAA)**.

2. Allopolyploidy: Polyploidy resulting from the doubling of chromosome number in a **F1** hybrid derived from two quite different species is known as **allopolyploidy**. Allopolyploidy brings two different sets of chromosomes in **F1** hybrid. Suppose **A** represent a set of chromosomes (genome) in species **X**, and **B** another genome in species **Y**. The **F1** will then have one **A** genome and another **B** genome. The doubling of chromosomes in the **F1** hybrid (**AB**) will give rise to a tetraploid with two **A** and two **B** genomes. Such polyploid is called **allopolyploid** or **amphidiploid**.



2.4.2 Aneuploidy:-

It is the presence of a chromosome number which is different than the multiple of **basic chromosome number**. This type of variation involves one or a few chromosomes but not the entire set. It is either due to loss of one or more chromosomes or due to the addition of one or more chromosomes to complete chromosome complement. **Aneuploidy** is of the following types:

1. Monosomy
2. Nullisomy
3. Trisomy
4. Tetrasomy

1. Monosomy

Monosomics represent the loss of a single chromosome from the diploid set, and they have the chromosome complement **2n -1**. Since monosomies lack one complete chromosome, such aberration creates major imbalance and cannot be tolerated in diploids.

2. Nullisomy

Nullisomics lack a single pair of the homologous chromosome; have the chromosome complement $2n - 2$.

3. Trisomy

Trisomies are those organisms that have an extra chromosome ($2n + 1$) which is homologous to one of the chromosomes of the complement. They are specifically useful in locating genes on a specific chromosome.

4. Tetrasomy

Tetrasomy are those organisms which have an extra pair of homologous chromosomes and have the chromosome complement $2n + 2$.

2.4.3 Polysomy:-

Polysomy is a condition found in many species, including **fungi, plants, insects, and mammals**, in which an organism has at least one more chromosome than normal, i.e., there may be three or more copies of the chromosome rather than the expected two copies.

Most **eukaryotic species** are **diploid**, meaning they have two sets of chromosomes, whereas **prokaryotes** are **haploid**, containing a single chromosome in each cell. **Aneuploids** possess chromosome numbers that are not exact multiples of the haploid number and **polysomy** is a type of **aneuploidy**. A **karyotype** is the set of chromosomes in an organism and the suffix - **somy** is used to name aneuploid karyotypes. This is not to be confused with the suffix - **ploidy**, referring to the number of complete sets of chromosomes.

Polysomy is usually caused by **non - disjunction** (the failure of a pair of the homologous chromosome to separate) during **meiosis**, but may also be due to a translocation mutation (a chromosome abnormality caused by rearrangement of parts between non-homologous chromosomes). **Polysomy** is found in **many diseases**, including **Down syndrome** in humans where affected individuals possess three copies (trisomy) of chromosomes.

Polysomic inheritance occurs during meiosis when chiasmata form between more than two homologous partners, producing multivalent chromosomes. **Autopolyploids** may show **polysomic inheritance** of all the linkage groups, and their fertility may be reduced due to unbalanced chromosome numbers in the gametes. In **tetrasomic inheritance**, four copies of a linkage group rather than two (**tetrasomy**) assort two-by-two.

TYPES OF POLYSOMY:-

Polysomy types are categorized based on the number of extra chromosomes in each set, noted as a diploid ($2n$) with an extra chromosome of various numbers. For example, a polysomy with three chromosomes is called a trisomy, a polysomy with four chromosomes is called tetrasomy, etc.

2.5 Summary:-

1. Chromatin network is found in the interphase of cell division.
2. The term Chromosome was first coined by Waldeyer in 1888.
3. Chromosome plays a very important role in the transmission of heredity characters from one generation to next.
4. Each individual of a species contains a definite number of chromosomes.
5. Term mutation was given by a Dutch scientist Hugo de Vries in 1902.
6. Hugo de Vries used evening primrose for his study on mutation.
7. Mutations are spontaneous inheritable changes which occur suddenly and alter the phenotype of an organism.
8. Spontaneous inheritable changes in plants and animals are the principle cause of the origin of new species.
9. Spontaneous inheritable sudden changes in genotype alter the phenotype of an individual.
10. The mutation may be in the form of gene mutation, chromosomal mutation, and genomic mutation.
11. In inversion inverted part of chromosome rotate by 180 degrees.
12. During pairing, repulsion occurs at the part where the genes do not match, is called position effect.
14. Loss of chromosome segment of any size, down to a part of a single gene.
15. Having a chromosome complement that is an exact multiple of the haploid complement.
16. Inversion is a type of chromosomal aberration in which two breaks take place in a chromosome and the fragment between breaks rotates 180 degrees before rejoining.

17. Paracentric inversion is a type of chromosomal aberration that occupies within one arm of a chromosome and does not span the centromere.

18. Pericentric inversion is a type of chromosomal aberration that involves both the arms of the chromosome, thus spanning the centromere.

2.6 Self Assessment Questions:-

1. Numerical changes in chromosome number are referred to as:

- (a) Change in ploidy (b) Hypoploidy
(c) Hyperploid (d) None of above

Ans - (a)

2. Variation in a number of copies of the genome or complete sets of chromosomes in a cell or organism refers to:

- (a) Euploidy (b) Aneuploidy
(c) Hyperploidy (d) Hypoploidy

Ans - (a)

3. In monosomic individual the number of chromosomes is:

- (a) $2n+2$ (b) $2n-1$
(c) $2n+1+1$ (d) $2n-2$

Ans - (b)

4. Down syndrome usually caused by an extra copy of chromosome:

- (a) 21 (b) 8
(c) 18 (d) 13

Ans - (a)

5. Edward's syndrome caused by trisomy of which chromosome:

- (a) 21 (b) 13
(c) 18 (d) 9

Ans - (c)

6. In a trisomic individual the number of chromosomes is

(a) $2n-1$ (b) $2n+3$

(c) $2n+2$ (d) $2n+1$ **Ans - (d)**

7. If a garden pea has 14 chromosomes in its diploid complement, how many double trisomic could theoretically exist:

(a) 6 (b) 21

(c) 16 (d) 9 **Ans - (b)**

8. A person with Klinefelter syndrome is considered a:

(a) Monosomic (b) Triploid

(c) Trisomic (d) Deletion heterozygote **Ans - (c)**

9. A mechanism that can cause a gene to move from one linkage group to another is:

(a) Translocation (b) Inversion

(c) Crossing over (d) Duplication **Ans - (a)**

10. Which of the following syndrome is an example of sex chromosomal?

(a) Turner syndrome (b) Down syndrome

(c) Patau's syndrome (d) Edward's syndrome **Ans - (a)**

11. A deletion involving two breaks in a chromosome called:

(a) Terminal deletion (b) Centric deletion

(c) Interstitial deletion (d) Both b and c **Ans - (c)**

12. If 2 breaks occur in one chromosome and region between the break rotates 180 degree before rejoining with the two end fragments, than chromosomal mutation called:

(a) Translocation (b) Inversion

(c) Duplication (d) None of the above **Ans - (b)**

13. Which type of chromosomal mutation does not change the overall amount of genetic material:

- (a) Deletion (b) Duplication
(c) Inversion (d) Both a and c **Ans - (c)**

14. Position effect is the result of:

- (a) Mutation (b) Inversion
(c) Transversion (d) Deletions **Ans - (b)**

15. People with Klinefelter syndrome have 47 chromosomes including 3 sex chromosome (XXY). What is the term to describe the aberration that occurs during meiosis that results in abnormal chromosome numbers?

- (a) Crossing over (b) Non-disjunction
(c) Independent assortment (d) Pairing of homologous chromosomes

Ans - (b)

16. The karyotype designation 47, XX, +13 designates which of the following:

- (a) Female with trisomy 13 (b) Female with 13 extra chromosomes
(c) Female with monosomy 13 (d) Female with extra genetic materials on chromosome 13

Ans - (a)

17. Match the following:

- | | |
|---------------------------|-----------------------------------|
| P- Down's syndrome | 1- an additional sex chromosome |
| Q- Cri-du chat syndrome | 2- loss of a part of chromosome |
| R- Klinefelter's syndrome | 3- absence of sex chromosome |
| S- Turner's syndrome | 4- presence of 1 extra chromosome |

- (a) P-4, Q-2, R-1, S-3
(b) P-4, Q-2, R-3, S-1
(c) P-4, Q-1, R-2, S-3
(d) P-3, Q-4, R-1, S-2 **Ans - (a)**

2.7 Terminal Questions:-

- 1- Define chromosome?
- 2- Write a short note on euploidy?
- 3- What is Trisomy?
- 4- Classify the chromosomes on the basis of centromeric positions?
- 5- Write a detail account on mutations with special emphasis on chromosomal mutation?
- 6- Describe various types of chromosomal duplications with examples?

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UNIT 3: GENETIC INTERACTION

Contents

- 3.1 Objectives
- 3.2 Introduction
- 3.3 Structure of gene and functions
 - 3.3.1 Gene structure
 - 3.3.2 Functions of genes
- 3.4 Summary
- 3.5 Self-assessment questions
- 3.6 References
- 3.7 Suggested readings
- 3.8 Terminal questions

3.1 Objectives:-

Genes are the **hereditary units** responsible for the transfer of **genetic characters** from the one generation to the next, located in the chromosomes in a **linear fashion**. The gene is to genetics what the atom is to chemistry. It is also said that genes are like catalysts which bring about reactions without being **changed or consumed**. One chromosome carries a **number of genes**. In the last chapter, you had read about the **chromosomes** and **chromosomal mutations**. In this chapter, you will read about the **genes, roles or functions of genes, the structure of genes and modern gene concept**.

3.2 Introduction:-

Mendel assumed in his experiments the presence of "**unit determiners**" responsible for hereditary characters. These unit determiners are now referred to as "**genes**". The term gene was introduced by **Wilhelm Johannsen in 1909**. The gene is that specific area of the chromosome which determines a particular character.

After the rediscovery of **Mendel's laws in 1900**, **Walter S. Sutton (1902)** pronounced **chromosome theory of heredity**, according to which the chromosomes are the carriers of hereditary particles or **determiners (genes)**. During the second decade of this century, many concepts of genes were established by **Thomas Hunt Morgan, A. H. Sturtevant, C. B. Bridges, and H. G. Muller** on *Drosophila*.

Their results were in accord with the chromosome theory put forward by **Sutton**. So it was finally established that the genes, **controlling hereditary characters are carried on the chromosomes, act as vehicles to carry these genes from one generation to next**.

3.3 Structure of Gene and Functions:-

Based on classic concept following definitions of genes were suggested by various scientists:

A. GENE, THE UNIT OF FUNCTION: According to this definition gene is the **smallest unit** of a chromosome and as well as of physiological activity.

B. GENE, THE UNIT OF MUTATION: According to it the gene is **the smallest unit**, capable of undergoing mutation.

Morgan (1925) defined the gene "as a particle in the chromosome which is distinguishable from other particles either by crossing over or mutation.

C. GENE, THE UNIT OF TRANSMISSION: According to **Castle**, the gene is the smallest particle of chromatin capable of **self-duplication** and is the ultimate unit of heredity.

Based upon its subdivisions the **gene** or **cistron** may be defined "as the functional unit segment of DNA consisting of several subunits (or nucleotide pairs) called mutons or recons"

Thus, the **gene** is the **smallest segment** of the chromosome whose activity can produce a definite effect. So, the **phenotype** is the physiological effect of the gene. But it is not always correct because sometimes functional effectiveness of a gene depends upon other **neighboring genes** and there might be overlapping regions of gene function.

CLASSIC CONCEPT OF GENE:-

The **gene concept** was introduced by **Sutton**. The **theory of gene**, formulated by **T. H. Morgan**, is a summary of the *information about characters genes, chromosomes, linkage and crossing over. A lot about the nature of a gene is now established which leads to the **classic concept of the gene**. The essential features of the modern concept of genes are as following:

1. **Inheritance** of biparental *i.e.* both male and female parents contribute **equally** in the inheritance of characters to the next generation
2. Genes determine the **physical** as well as **physiological** characteristics. These are transmitted from parents to the offspring's generation after generation.
3. Characters of an individual are determined by **paired genes** situated in a definite number of chromosome pairs or **linkage groups**.
4. Genes are situated in the chromosome in a **linear fashion** like the arrangement of **beads** on a string.
5. Several genes are present in each chromosome; all such genes of the same chromosomes are called as **linked genes**.
6. In man about **40,000 genes** are known to be located on **23 pairs of chromosomes** (46 chromosomes).
7. Each gene occupies a **specific position** on a specific chromosome. This position is known as a **locus** (pl. loci).
8. At **mutation**, the members of each pair of genes separate so that either of the gametes possesses only one gene of that kind.

9. Pairs of genes held in different chromosomes or **linkage groups** are assorted independently.
10. A single gene may occur in **several forms** or in **several functional states**. The forms other than normal are known as **alleles**.
11. Many genes have only **two alleles**; one of them is normal and another one is its mutant.
12. Only those genes are known which have their **alternative alleles**.
13. The **alleles** may be related as **dominant or recessive** but not always.
14. Genes lie in a **linear order** in their chromosomes and other **remains constant** until and unless **crossing over** or **mutation** takes place.
15. Gene in one chromosome may be shifted to another of the same homologous pair. It may be either due to **crossing over** or due to **translocation**.
16. Some genes **mutate** more than once and have more than **two alleles**. These are known as **multiple alleles**.
17. The genes may undergo a sudden change in expression due to change in its composition. The changed gene is known as **mutant gene** and the phenomenon of change is known as **mutation**.
18. Rarely genes from one chromosome may be exchanged or transferred to another chromosome which may be its homologous counterpart (**crossing over**) or non - homologous (**translocation**).
19. Genes **duplicate themselves** very accurately. The phenomenon is known as **replication**. Self-duplication of genes leads to chromosomal duplication.
20. Two or more pair of genes may interact to produce to produce a trait (**interaction of genes**).
21. Inbreeding leads to **homozygosity** and out breeding to **heterozygosity** and hybrid vigor.
22. Genes express themselves by producing **enzymes** which are **proteins**. It means each gene synthesizes a particular protein which acts as an enzyme and brings about an appropriate change.

3.3.1 Gene Structure:-

The structure of a gene may be studied in the following headings:

A. LOCATION OF GENE: According to **Demerec (1939)** genes are located on the chromosome along its **entire length** in a **linear fashion**. The chromosomal threads are alike both chemically and physically, side branches at right angles which are given out from the

chromosome and these bear **genes**. The genes on the one branch may be alike or unlike, both **physiologically** and **chemically**.

B. GENES AND GENOME: Organisms possess a **definite number of the chromosome**, and no doubt, the number **varies** from species to species. The number becomes half during **gametogenesis**. The total number of chromosomes found in gametes constitutes **one genome**. Thus, the genome can be expressed as the total sum of genes present on the **haploid set** of chromosomes. Diploid organisms never contain more than **two genomes** while haploid organisms are never more than **one genome**.

C. GENE SIZE: The genes are very **fine structure** and these are too difficult to be measured **directly**. The size consideration of gene implies that it has certain definite limits. Furthermore, when the gene is said to be **functional** and **behavioral** in structure, it is difficult to measure its size directly. According to **Muller (1947)** four genes which are located in a particular length of salivary gland chromosome, had a mean length of **1250Å**. **Pontecorvo** measured the genes in *Aspergillus nidulans* which come to **4500Å**.

Lea noted the **genes volume** between **0.003 to 0.005 millimicrons**. It is also assumed that any alteration with such volume leads to **mutation** and other methods may cause mutation or may alter the **mutation rate**, **gene size** etc. But actual gene size depends upon the **DNA molecule**. According to **Burnham (1962)**, the four giant chromosomes of *Drosophila melanogaster* contain **about 5149 bands and they range in length 0.05 microns to 0.5 microns**.

D. SHAPE OF GENES: **Stanley** and **Stizznski** made a deep study regarding the shape of the gene. **Stanley** studied the **viral structure** and noticed the genes as **rod-like bodies**. This assumption suggests that the gene is a **dot-like structure**, and if it is true, it will present minimum surface area to be lit up by the X-rays during X-rays mutation. And if the gene is **rod-like**, that may break down by X-rays biting causing **structural change**.

More accurately, **Watson, Crick and Wilkins (1962)** gave the **double helical structure** of a **DNA molecule** where two long strands are **coiled spirally** around an axis and are interconnected by several transverse bands.

E. NUMBER OF GENES: **Gowen** and **Gay** have calculated the number of genes present in chromosomes. According to them, one set of the chromosome in *Drosophila* contains about **10,000 to 15,000 genes**, while in **man** the number of **genes per chromosome set is 90,000**. The total number of genes per cell is **100,000**, while **in man, it is 300,000**.

The lowly organized forms of life such as **Bacteriophages** are said to contain about **100 different genes**. In the **bacteria**, the gene number is said to be present between **1,000 to 3,000**. In *E. coli*, the chromosome is about **1mm long** and contains about **400 genes** and about **4,000,000 base pairs**. Some **fishes** and **amphibians** contain about **10 to 20 times** more gene than the **man**.

F. GENE STABILITY: Chromosomes are **quite stable** and continuously transmitted from one generation to the next. The stability of a gene can be studied in terms of the **half life** of a gene, that is, the time elapsing for a **50% probability** that a particular gene will mutate or more clearly, the time in which **50%** of the gene would be expected to mutate.

Stability of the gene can be influenced by **ionizing radiations** and **chemicals**, which can alter the **rate of mutations**. **Giles** reported the breakage of one chromosome out of every **5000 per cell** generation in *Tradescantia*. The **average half-life** which is about **10 to 10⁴ years** has, however, been reported in *Drosophila* (**Muller 1950**).

G. CHEMICAL COMPOSITION OF GENE: Chemically gene contains **deoxyribonucleic acid (DNA)** as the primary component. DNA is a long and mostly **double stranded** molecule the **two strands** of which are **coiled helically around each other** forming a structure like a **spiral staircase**, popularly known as a **double helix**. Chemically each strand of DNA is **polynucleotide chain** made up of repeating units called **nucleotides**.

Each **DNA nucleotide** is a complex combination of **phosphoric acid**, a **deoxyribose sugar**, and one of the four **nitrogenous bases**, i.e. adenine, guanine, cytosine, and thymine. Out of these four, adenine and guanine are **purines** and cytosine and thymine are called **pyrimidines**. Therefore, there are four different types of nucleotides possible in DNA: **adenylic acid, guanylic acid, thymidylic acid and cytidylic acid**. Each nucleotide is a separate entity connected to each other through **chemical bonds** between the sugar and phosphoric acid components.

No restriction is imposed on the sequence and number of nucleotides within the DNA molecule. A gene, which is a **stretch of DNA**, can have any number of nucleotides in any order. However, a particular number and order of nucleotides constitute a **particular gene**. Cells with similar origin have DNA of similar composition.

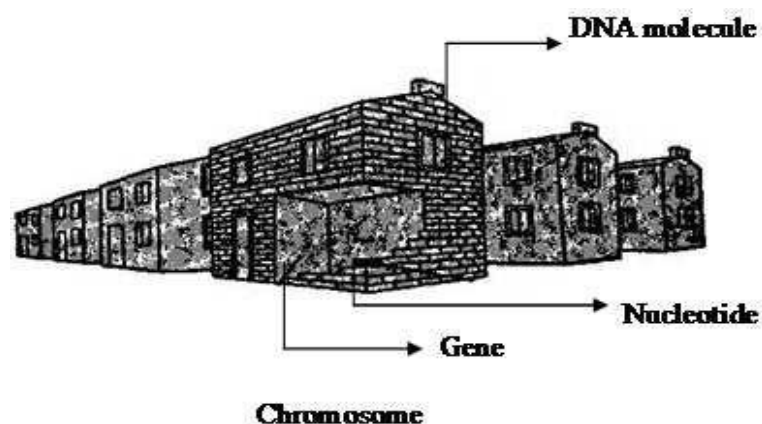


Fig.3.1 Relation between chromosome & Gene

MOLECULAR STRUCTURE OF GENE

Chemically, a gene is formed of DNA, but what length of DNA constitute a gene, has been explained by the relationship between different genetic phenomena and DNA molecule. **Seymour Benzer** postulated the following **three new terms** or functional units to correlate the physiological aspects of the gene:

1. Recon
2. Muton
3. Cistron

1. RECON: It is the **smallest unit** of DNA, capable of undergoing **crossing over and recombination**. A recon may be as small as **one nucleotide pair** in DNA. Crossing over may take place **between two recons** of a chromosome but never within a recon. A recon consists of **one nucleotide pair in DNA** and one nucleotide in **mRNA**. Recon and muton are usually **identical and indistinguishable**.

2. MUTON: It is also **smallest unit of DNA** which could undergo **mutation**. In its smallest expression, it represents a change in a pair of nucleotides. Any change in a base of **triplet** will modify the message carried by the **codon**. So, the gene as a unit of mutation is smaller than a cistron -consisting of fewer nucleotides only. It consists of any one pair **of nucleotides**, which, when changed is able to produce different **phenotypic effects**.

The difference between **normal hemoglobin A** and persons with either **sickle cell trait** or **sickle - cell anemia** is the result of a change of one nucleotide pair from **AT** to **TA** in DNA. Thus, a single nucleotide in messenger **RNA (mRNA)** spells the difference **between Hb-A and Hb-S**. Haemoglobin **G** could be derived from hemoglobin **A** by the change of an **AT** pair in the **seventh triplet** for the **beta cistron** to a **GC pair**. Thus, this base pair in the DNA or the single base in the complementary messenger **RNA codon**, comprise the muton.

3. CISTRON: It is the **largest segment** or subunit of the gene. It represents that segment of DNA which has **codons** for the formation of **one polypeptide chain**. Therefore, in cistron, the number of nucleotides is **3 times** the number of **total amino acids** present in a polypeptide chain. For example, in hemoglobin, there are two polypeptide chains (**K and B**) having **141** and **146 amino acids** respectively. A cistron responsible for **K** chain possesses **141 x 3 = 423 nucleotides** and for **B** chain **146 x 3 = 438 nucleotides**. Cistron is the unit of function. **It is the gene in the real sense**.

It is important to note that each term - **cistron, muton, and recon** has an operational definition that is **functional, mutational and recombinational** respectively. This is the **modern gene concept**. However, the term **muton** and **recon** are now less frequently used because they are known to represent the **same thing** in the physical term. According to **Strickberger**, "the present view is that the smallest recombinational unit (recon) is probably a single nucleotide which is

probably the smallest mutational unit (muton). Even **cistron** is also not popularly used and the term gene is preferred.

Beside above mentioned terms, some more terms are introduced, which are as follows:

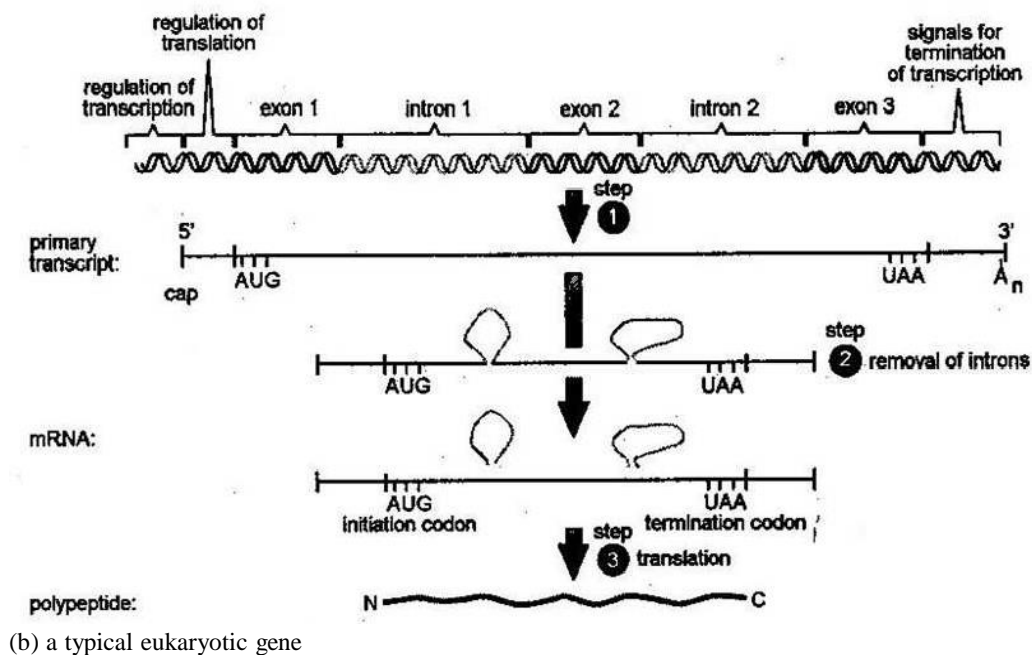
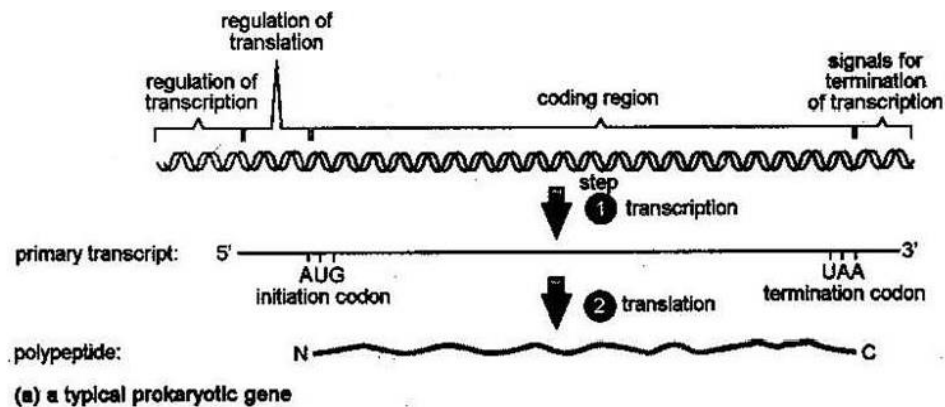
I. COMPLON: It is the unit of complementation, which is **fundamentally similar** to the cistron. It **controls** any change in the polypeptide chain.

II. REPLICON: It is the unit of **replication**. For example, the chromosome of bacteria.

III. CODON: A triplet of messenger which helps in **translation** during protein synthesis.

IV. OPERON: Several related **enzymes** forming a segment of DNA molecule which regulates a **specific function**.

V. TRANSCRIPTION: Several related **enzymes** forming of DNA molecule and as a whole **transcribes the messages**.



Gene structure

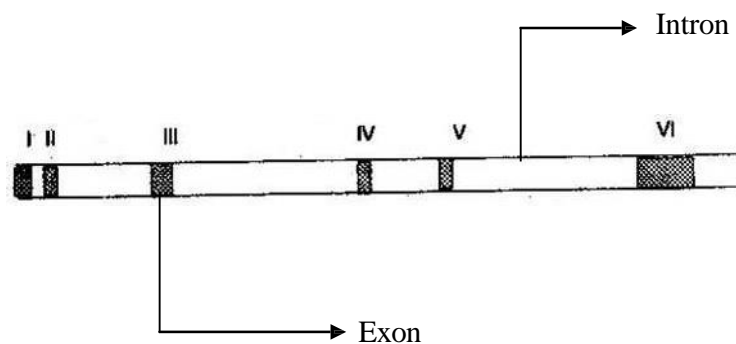
Fig.3.2 Gene structure

DISCONTINUOUS GENES (EXONS AND INTRONS)

In 1977, it was discovered that the biological information carried by some genes are not continuous. It is split into several distinct units separated by regions of noncoding DNA. Such genes are called discontinuous genes or split genes or mosaic genes. The sections containing biological information are called exons and the intervening noncoding sequence is referred to as introns.

INTRONS: Introns are present in the genes of eukaryotes and their viruses. But these are absent in genes of prokaryotes and their viruses. These are present in archaeobacteria but are absent in mitochondrial genome.

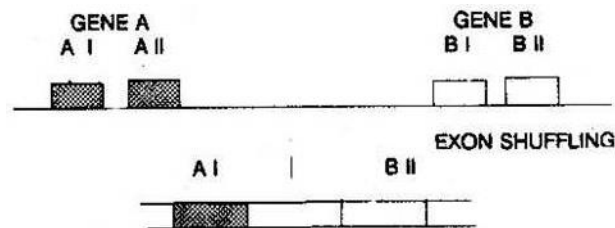
A single gene may contain no introns or may have as many as 52 as found in the mammalian α -collagen gene.



Discontinuous gene with exon and intron

Fig.3.3 Discontinuous gene with exon & intron

According to Gilbert hypothesis, during evolution, exons from different discontinuous genes can be shuffled forming new combinations of biological information. The shuffling may produce new functional proteins.



New gene
Origin of new genes (Gilbert hypothesis)

Fig 3.4 Origin of New Gene

3.3.2 Functions of Gene:-

Genes play a **vital role** in the cellular economy. They are said to be the **fundamental units of heredity** related to:

1. The basic **architecture** of the cell.
 2. Direct or indirect **control of the metabolic** activities of the cell.
 3. **Reproduction** of the cell.
 4. **Protein synthesis**.
 5. **Enzyme formation**.
 6. Due to innate capacity to **mutate**, genes also control the direction of the **evolutionary process**.
 7. Gene has a unique characteristic that its capacity of **self -replication**.
 8. It produces an exact **Photostat copy of itself**.
 9. Genes also act as physiological agent exerting its influence through the medium of **enzymes and hormones**.
-

10. Genes regulate the **biochemical activities** of the cell.
11. Genes are able to **replicate and be inherited** by the progeny faithfully.
12. Genes are **susceptible** to an occasional change by way of **mutation** and such a change should be stably inherited.
13. The sequence of amino acids in a polypeptide chain is determined by the genes, which contain the information in the form of genetic code.
14. The genetic code is the triplet sequence of nitrogenous bases in mRNA molecule, which it has copied from the DNA molecule.
15. Genes are able to carry all the information necessary to program the functions of a cell.

3.4 Summary:-

1. The amino acid is an organic compound containing both amino ($-\text{NH}_2$) and carboxyl ($-\text{COOH}$) groups.
2. The hydrogen bonding of complementary purine and pyrimidine bases is called base pairing.
3. Base sequence is a specific order of purine and pyrimidine base in a polynucleotide chain,
4. Cistron is a genetic unit that carries information for the synthesis of a single enzyme or protein molecule.
5. A sequence of three nucleotide bases (in mRNA) that code for an amino acid for the initiation or termination of a polypeptide chain.
6. Deoxyribonuclease is an enzyme that catalyzes the hydrolysis of DNA to nucleotides.
7. Deoxyribonucleic acid (DNA) is the carrier of genetic information: a type of nucleic acid occurring in the cells, containing phosphoric acid, 2-Deoxyribose sugar, adenine, guanine, cytosine, and thymine.
8. Deoxyribose is a type of sugar that having five carbon sugar atoms and one oxygen atom less than the present sugar, ribose; a component of DNA.
9. Exon is a portion of DNA which codes for the final RNA.
10. Gene is a segment of a chromosome, definable in operational terms as the repository of a unit of genetic information.

11. The genetic information in the nucleotide sequences in DNA represented by a four-letter alphabet that makes up a vocabulary of 64 three - nucleotide sequence or codon is known as genetic code.
12. The genome is a complete set of genetic material.
13. The intron is a noncoding segment of a gene.
14. Locus refers the site on a chromosome occupied by a gene.
15. Genotype is the particular set of genes present in an organism and its cells.
16. Mutant is an organism with changed or new genes.
17. Muton is the smallest unit of genetic material capable of undergoing mutation.
18. A nucleotide is a compound formed from one molecule each of a sugar (pentose), phosphoric acid, and a purine or pyrimidine base.
19. A linear sequence of nucleotide is known as polynucleotide,
20. Production of a strand of DNA from the original is called replication.

3.5 Self Assessment Questions:-

1. In a gene interaction the gene that masks the expression of another gene is termed as:

- (a) Epistatic gene (b) Hypostatic gene
(c) Both (a) and (b) (d) None of these

Ans: (a) Epistatic gene

2. Allelic gene interaction is exhibited by which of these:

- a) Incomplete dominance b) Co dominance
(c) Lethal genes (d) All of these

Ans: (d) All of these

3. Examples of non-allelic gene interaction are:

- a) Complementary genes (b) Supplementary genes
(c) Modifier genes (d) All of these

Ans. (a) Complementary genes

4. Effect of a single gene on one or more than one characteristic is known as:

- a) Lethal gene (b) Hypostatic gene
- b) Pleiotropy (d) None of these

Ans. (c) Pleiotropy

5. Segregation of alleles occurs at the time of:

- a) Cleavage (b) Meiosis
- © Fertilization (d) Crossing over

Ans. (d) crossing over

6. Genes that reduce the viability of individual or cause its death of bearer are called:

- a) Complementary genes (b) Supplementary genes
- (c) Lethal genes (d) none of these

Ans. (c) Lethal genes

7. The ratio of 9:3:4 is obtained instead of 9:3:3:1 under the condition when there is gene interaction involving:

- a) Supplementary genes (b) Complementary genes
- (c) Both (a) and (b) (d) None of these

Ans. (a) Supplementary genes

8. Instead of a usual Mendelian ratio of 9:3:3:1 often ratio of 9:7 is obtained in some of the crosses which are possibly due to the interaction of:

- a) Modifier genes (b) Complementary genes
- (C) Supplementary genes (d) All of these

Ans. (b) Complementary genes

9. Mating of closely related individuals is called:

- a) Inbreeding (b) Outbreeding
- (c) Atavism (d) None of these

Ans. (a)

10. Which of these workers coined the terms 'homozygous' and 'heterozygous' that we use very commonly in genetics:

- a) Bateson (b) Saunders
- © Both (a) and (b) (d) None of these

Ans. (c) Both (a) and (b)

11. Who formulated the chromosomal theory of inheritance?

- a) Sutton (b) Morgan
- (c) Bateson (d) Johannsen

Ans. (a) Sutton

12. Which of these is a heterozygous condition?

- a) RR (b) Rr
- (c) rr (d) RRrr

Ans. (b) Rr

13. A complete set of chromosomes, which is inherited as a unit from one parent is called:

- a) Genotype (b) Genome
- b) (c) Gamete (d) Gene

Ans. (b) Genome

14. A heterozygous individual which carries the unexpressed recessive gene for a sex-linked character is known as:

- a) Carrier (b) Mutant
- (c) Variant (d) None of these

Ans. (a) Carrier

15. Genetic complement is called:

- a) Genotype (b) Phenotype
- b) (c) Alleles (d) None of these

Ans. (c) Alleles

3.6 Terminal Questions:-

- 1- What is an allele?
- 2- Write a short note on Mutton?
- 3- What is the chemical composition of Gene?
- 4- Describe the structure and function of Gene?
- 5- Write a detail account on classic concept of Gene?

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3.8 Suggested Readings:-

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UNIT 4: HUMAN GENETICS

Contents

- 4.1 Objectives
- 4.2 Introduction
- 4.3 Recessive inherited disorder
- 4.4 Dominant inherited disorder
- 4.5 Inborn errors
- 4.6 Summary
- 4.7 Self-assessment questions
- 4.8 References
- 4.9 Suggested reading
- 4.10 Terminal questions

4.1 Objectives:-

Disorder or **disease** is any deviation from or interruption of the **normal structure** and **function** of any part of the body. It is expressed by a characteristic set of signs and symptoms and in most instances, the **aetiology**, **pathology** and **prognosis** are known. Human beings are prone to a wide variety of **diseases**. In this chapter, you will read about various types of **genetic disorders** also known as **genetic diseases** including recessive inherited disorder, dominantly inherited the disorder and Inborn errors etc.

4.2 Introduction:-

The **diseases** which are transferred from generation to generation are called **genetic disorders** or **genetically transmitted diseases**. They are congenital i.e. abnormal conditions are present at birth. These disorders may be due to **incompatible genes** or **abnormalities** in the **structure or number of chromosomes**. They may be inherited or arise a new due to **mutation**.

The **basic principles** of inheritance as applicable to all other living organism are usually applicable to man. The branch of genetics which deals with the inheritance of characters in man is known as Human **Genetics**. **Sir Archibald Garrod**, a British physician was the pioneer of human genetics. He in **1901** pointed out that certain **inborn errors of metabolism** like phenylketonuria, alkaptonuria etc. are controlled by genes and are inherited in simple **Mendelian fashion**.

4.3 Recessive Inherited Disorder:-

Autosomal recessive diseases are genetic diseases that are passed to a **child** through both parent's chromosomes. Each person inherits **23 chromosomes** from each parent and so has **23 pairs of chromosomes**. Each chromosome contains genes. One or both of the chromosomes in a pair may contain a **changed (mutated) gene** that could cause a **genetic disease**. In an autosomal recessive disease, both chromosomes in a pair must have a changed gene for the person to have the disease. If only one chromosome has a changed gene, the person is a **carrier** and does not have symptoms.

However, if **both parents** carry the gene change, there is a:

- **25% chance** in each pregnancy that their child will inherit the changed gene from each parent (two genes) and have the disease.

- **50% chance** in each pregnancy that their child will receive one changed gene and be a carrier.
- **25% chance** in each pregnancy that their child will not receive the changed gene and be neither a carrier nor have the disease.

If only **one parent** carries the gene change, there is a **50% chance** in each pregnancy that the child will:

- Receive the changed gene and be a **carrier**.
- Not receive the changed gene and be **neither a carrier nor have the disease**.

If neither parent carries the gene change, the child will not have this type of disease.

Recessively inherited disorders may be **two types**:

A. Autosomal recessive disorders

B. Sex-linked recessive disorders

A. AUTOSOMAL RECESSIVE DISORDERS

Genes come in pairs. One gene in each pair comes from the **mother** and the other gene comes from the **father**. Recessive inheritance means both genes in a pair must be **abnormal** to cause disease. People with only one defective gene in the pair are called **carriers**. These people are most often not affected with the condition. However, **they can pass the abnormal gene to their children**.

These include **disorders** the genes for which are present on autosomes and are recessive to their **alleles**. These disorders are expressed only in a **homozygous condition** like the typical **Mendelian recessive disorders**. Major autosomal disorders are as follows:

1. Albinism
2. Tay - Sachs disease
3. Cystic fibrosis
4. Sickle cell anemia
5. Phenylketonuria
6. Autosomal recessive polycystic kidney disease (ARPKD)

1. ALBINISM

It is the condition where **skin** and **hair** of the whole body appear colorless due to total or nearly total absence of **pigmentation** (melanin pigment). It is an autosomal recessive disorder against the normally pigmented **skin, eyes, and hairs**. The skin is very light and hair whitish yellow and eyes appear pinkish. Albinos have poor vision, sensitive to sunlight and **prone to skin cancer**. **One out of 20,000 individuals is albino**.

An **albino** is thus always **homozygous** for the character. The absence of the **enzyme melanocyte tyrosinase** leads to failure of melanin formation from **tyrosine**, and the person develops albinism. Albinos although **leads a normal life**, have a strong aversion against exposure to the sun.

Albinism results from a **recessive mutant** of the **normal gene**. It develops only when an individual possesses both genes in recessive condition one from each parent. It means both the parents of an albino child are **heterozygous** for this pair of genes. The recessive disorder tends to appear only in **the siblings**, not in their parents. This can be understood from the study of family pedigree as shown below:

Autosomal recessive is one of the several ways that a trait, disorder, or disease can be passed down through families. An autosomal recessive disorder means **two copies of an abnormal gene must be present in order for the disease or trait to develop**.

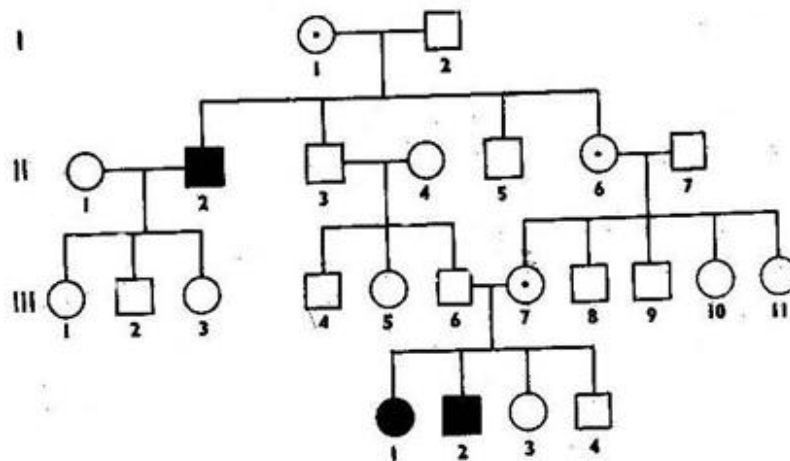


Fig.4.1 Pedigree of recessively inherited disorder

2. TAY-SACHS DISEASE

Tay-Sachs disease is a fatal disorder in **children** (usually by age 5) that causes a progressive degeneration of the **central nervous system**. It is caused by the absence of an **enzyme** called **hexosaminidase A** (or hex A). Without **hex A**, a **fatty substance** builds up on the nerve cells in the body, particularly the brain. Means **the oxidation** of the **lipid sphingomyelin**, which usually takes place in normal individual, does not take place.

The **enzyme hexosaminidase A** is required for **lipid metabolism** and without it, the nervous system **degenerates**. The affected infant becomes **blind, paralyzed** and **mentally deficient** and invariably dies within a very short time. The process begins early in pregnancy when the baby is developing. It is not apparent until several months after the birth. To date, **there is no cure** for **Tay-Sachs**. About **1 in 27 persons** of European Ashkenazi Jewish ancestry carries the Tay-Sachs gene.

3. CYSTIC FIBROSIS

Cystic fibrosis (CF) is a common, inherited, **single-gene disorder** in **Caucasians**. People with CF produce mucus that is abnormally **thick and sticky** that can damage body organs. The **mucus interrupts** the function of vital organs especially the **lungs** and leads to **chronic infections**. CF also involves the **pancreas** and causes decreased absorption of essential nutrients and **reproductive system** damage. With improved treatment and management of the disease, affected people may live well into **adulthood**. Ultimately, death most often occurs from **respiratory failure**. Other people with variants of **CF** may have only lung involvement, **sinusitis or infertility**.

Although, **Cystic fibrosis** is a **hereditary disease**, symptoms of this disease normally appear during the **first few weeks of life** and rapidly become severe. There is an abnormality of secretion of the **exocrine gland**. The mucous gland of lungs instead produces thick, **sticky sputum** that **clogs** and **dilates** the **air passages**. This leads to **severe breathing difficulties and respiratory infections**. The pancreas degenerate and the resulting **lack of pancreatic digestive juices** means that not enough **fat** is absorbed from the intestine.

Abnormality of sweat glands increases chloride content of the sweat. The affected child has a **grossly swollen abdomen** and the rest of his body is **thin and wasted**.

4. SICKLE CELL ANAEMIA

Sickle cell anemia is another common, **inherited, single-gene disorder** in African-Americans. About 1 in 500 African-American babies is born with **sickle cell anemia**. About 1 in 12 African-American people carries the gene for this disease. Sickle cell disease involves the **red blood cells** or **hemoglobin** and their ability to **carry oxygen**.

Normal hemoglobin cells are **smooth, round and flexible**, like the letter "O." They can easily move through the **vessels** in our bodies. Sick cells are **stiff and sticky**. When they lose their **oxygen**, they form into the shape of a **sickle**, or the letter "C". These sick cells tend to **cluster** together and can't easily move through the **blood vessels**. The cluster causes a **blockage** and stops the movement of healthy, normal, **oxygen-carrying blood**. This blockage is what causes the painful and damaging complications of **sickle cell disease**.

Sickle cells live only for about **15 days**. Normal hemoglobin cells can live up to **120 days**. Sick cells risk being destroyed by the **spleen** because of their shape and stiffness. The spleen helps filter the **blood of infections**. Sick cells get "**stuck**" in this filter and die. Due to the decreased number of **hemoglobin cells** circulating in the body, a person with sick cell is **chronically anemic**.

The **spleen** also suffers damage from the **sickle cells**, which block the healthy oxygen-carrying cells. After **repeated blockages**, the spleen becomes very small and does not work properly. Without a functioning spleen, these people are more at risk for **infections**. Infants and young children are at risk for life-threatening infections. Treatment includes prompt emergency care for fevers and infections, appropriate vaccinations, penicillin and **management of anemia**.

5. PHENYLKETONURIA

Phenylketonuria (PKU) is a **rare genetic condition** that causes an **amino acid** called **phenylalanine** to build up in the body. Amino acids are the **building blocks of protein**. Phenylalanine is found in all proteins and some **artificial sweeteners**. Our body uses an **enzyme** called **phenylalanine hydroxylase** to convert **phenylalanine** into **tyrosine**, a non-essential amino acid. Our body needs **tyrosine** to create **neurotransmitters**, such as **epinephrine, norepinephrine and dopamine**.

PKU is caused by a defect in the **gene** that helps create **phenylalanine hydroxylase**. When this enzyme is missing, the body is unable to break down **phenylalanine**. This causes a buildup of **phenylalanine** in the body. Early diagnosis and treatment can help relieve symptoms of **PKU** and **prevent brain damage**.

Thus, the patient is **mentally deficient**. The mental deficiency is due to the fact that excess **phenyl pyruvic acid** depresses **pyruvate dehydrogenase**. The latter is responsible for the conversion of **pyruvic acid** of the brain to **acetyl Co-A**, and as such ultimately **Kreb's tricarboxylic acid cycle** in brain suffers in this disease.

PKU symptoms can range from **mild to severe**. The most severe form of this disorder is known as **classic PKU**. An infant with classic PKU may appear normal for the first few months of their

life. If the baby isn't treated for **PKU** during this time, they'll start to develop the following symptoms:

- Seizures.
- Tremors or trembling and shaking.
- Stunted growth.
- Hyperactivity.
- Skin conditions, such as eczema.
- A musty odor of their breath, skin, or urine.

6. AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE

Autosomal recessive polycystic kidney disease (ARPKD) is a significant **hereditary renal disease** in that appears in **childhood**. The single **gene mutation** called "**PKHD1**" is fully responsible for the disease presentation of **ARPKD**. This **PKHD1** is located on the human chromosome region **6p21.1-6p12.2**. It is also one of the **largest genes** in the genome as it occupies approximately **450 kb of DNA**, and contains at least **86 exons**.

Although autosomal recessive polycystic kidney disease (ARPKD), is a rare inherited childhood condition, where the development of the kidneys and liver is abnormal. Over time, either one of these organs may fail. The condition often causes serious problems soon after **birth**, although less severe cases may not become obvious until a child is older.

ARPKD can cause a wide range of problems, including:

- **Underdeveloped lungs**, which can cause severe **breathing** difficulties soon after birth.
- High blood pressure (**hypertension**).
- Excessive **peeing** and **thirst**.
- Problems with **blood flow** in the liver, which can lead to **serious internal bleeding**.
- A progressive **loss of kidney function**, known as **chronic kidney disease (CKD)**.

Even though **ARPKD is rare**, it's one of the most common **kidney problems** to affect young children. It's estimated that around **1 in 20,000 babies** is born with the condition. Both boys and girls are affected equally.

ARPKD is caused by a genetic fault that disrupts normal development of the **kidneys** and **liver**. In particular, the growth and development of the small tubes that make up the kidneys are affected, causing **bulges** and **cysts** (fluid-filled sacs) to develop within them. Over time, the cysts cause the kidneys to become enlarged and scarred (**fibrosis**), resulting in the deterioration of overall **kidney function**.

Similar problems also affect the small tubes (**bile ducts**) that allow bile (a digestive fluid) to flow out of the **liver**. The bile ducts may develop abnormally and **cysts** may grow inside them. The liver can also become scarred over time.

ARPKD is caused by a **genetic alteration** in the gene **PKHD1**, which in most cases is passed on to a child by their parents. If both parents carry a faulty version of this gene, there's a one in four (**25%**) chance of each child they have **developing ARPKD**. The way **ARPKD** is inherited is different from a more common type of kidney disease called **autosomal dominant polycystic kidney disease (ADPKD)**, which usually doesn't cause significantly reduced kidney function until **adulthood**. **ADPKD** can be inherited if only one **parent carries** one of the genetic faults responsible for the condition.

There's currently no cure for ARPKD, but various treatments can help manage the wide range of problems it can cause.

B. SEX-LINKED DISORDERS

Sex-linked recessive inheritance is a mode of **inheritance** in which a **mutation** in a gene on the **X - chromosome** causes the **phenotype** to be expressed in males (who are necessarily **hemizygous** for the gene mutation because they have one **X** and one **Y-chromosome** and in females who are **homozygous** for the **gene mutation**.

X-linked inheritance means that the gene causing the trait or the disorder is located on the **X chromosome**. Females have **two X chromosomes**, while males have one **X** and one **Y chromosome**. Carrier females who have only one copy of the mutation do not usually express the phenotype, although differences in **X- chromosome** inactivation can lead to varying degrees of clinical expression in carrier females since some cells will express one **X- allele** and some will express the other. The current estimate of **sequenced X-linked** genes is **499** and the total including vaguely defined traits is **983**.

However, some important **sex-linked disorders** are as following:

1. Hemophilia
2. Color blindness
3. Muscular dystrophy

1. HEMOPHILIA

Hemophilia is an uncommon **hereditary bleeding disorder** which primarily affects male but is transmitted by females. **Haemophilia** also spelled **hemophilia**, is a mostly inherited genetic disorder that impairs the body's ability to make **blood clots**, a process needed to **stop bleeding**. This results in people **bleeding longer** after an injury, easy bruising, and an increased risk of **bleeding inside joints or the brain**. Those with mild disease may only have symptoms after an accident or during surgery. Bleeding into a **joint** can result in **permanent damage** while bleeding in the brain can result in **long term headaches, seizures, or a decreased level of consciousness**.

There are two main types of hemophilia:-

1. Hemophilia A - occurs due to not enough **clotting factor VIII**.

2. Hemophilia B - occurs due to not enough clotting factor IX.

They are **typically inherited** from one's parents through an **X- chromosome** with a **nonfunctional gene**. Rarely a new mutation may occur during early development or hemophilia may develop later in life due to **antibodies** forming against a **clotting factor**.

Other types include **hemophilia C**, which occurs due to not enough **factor XI**, and **parahaemophilia**, which occurs due to not enough **factor V**. Acquired hemophilia is associated with **cancers, autoimmune disorders, and pregnancy**. Diagnosis is by testing the blood for its ability to clot and its levels of clotting factors.

Prevention may occur by removing an **egg, fertilizing** it and testing the **embryo** before transferring it to the **uterus**. Treatment is by replacing the missing **blood clotting factors**. This may be done on a **regular** basis or **during bleeding episodes**. Replacement may take place at **home or in the hospital**. The clotting factors are made either from **human blood** or by **recombinant methods**. Up to **20%** of people develop **antibodies** to the clotting factors which makes treatment more difficult. The medication **desmopressin** may be used in those with mild hemophilia A. Studies of **gene therapy** are in early human trials.

Hemophilia A affects about 1 in 5,000–10,000, while **hemophilia B** affects about 1 in 40,000, males at birth. As **hemophilia A and B** are **X- linked recessive disorders**, females are very **rarely severely** affected. Some females with a nonfunctional gene on one of the **X - chromosomes** may be mildly symptomatic. **Hemophilia C** occurs equally in both sexes and is mostly found in Ashkenazi Jews. In the 1800s hemophilia was common within the **royal families** of Europe.

2. COLOR BLINDNESS

Difficulty in **distinguishing** between colors, particularly **red** and **green**, is an inherited defect. **Color blindness**, also known as **color vision deficiency**, is the decreased ability to see color or differences in color. Color blindness can make some **educational activities** difficult. Buying **fruit, picking clothing, and reading traffic lights can be more challenging**, for example. Problems, however, are generally minor and most people **adapt**. People with total color blindness may also have decreased **visual acuity** and be uncomfortable in bright environments.

The most **common cause** of color blindness is an inherited fault in the development of one or more of the three sets of color-sensing **cones** in the **eye**. Males are more likely to be color blind than females as the genes responsible for the most common forms of color blindness are on the **X - chromosome**. As females have **two X chromosomes**, a defect in one is typically compensated for by the other, while males only have one **X chromosome**. Color blindness can also result from **physical or chemical damage** to the eye, optic nerve or parts of the brain.

There is no cure for color blindness. Diagnosis may allow a person's teacher to change their method of teaching to accommodate the decreased ability to recognize color. Special lenses may help people with **red-green color blindness** when under bright conditions. There are also **mobile apps** that can help people identify colors.

Red-green color blindness is the most **common form**, followed by **blue-yellow color blindness** and **total color blindness**. Red-green color blindness affects up to **8%** of males and **0.5%** of females of Northern European descent. The ability to see color also decreases in old age. Being color blind may make people **ineligible for certain jobs** in certain countries. This may include **pilot, train driver** and **armed forces**.

3. MUSCULAR DYSTROPHY:-

Muscular dystrophy is a gradual **wasting disease** affecting various groups of **muscles** or it may also be said that "**a muscular dystrophy is a group of disorders characterized by a progressive loss of muscle mass and consequent loss of strength**". The most common form of muscular dystrophy is **Duchenne muscular dystrophy** which typically affects **young boys**, but other variations can strike in **adulthood**.

Muscular dystrophy is caused by **mutations** on the **X chromosome**. Each version of muscular dystrophy is due to a different set of **mutations**, but all prevent the body from producing **dystrophin**. **Dystrophin** is a **protein** essential for **building and repairing muscles**. **Duchenne muscular dystrophy** is caused by specific mutations in the gene that encodes the **cytoskeletal protein dystrophin**. **Dystrophin** makes up just **0.002 percent** of the total proteins in striated muscle, but it is an essential molecule for the general functioning of muscles.

Dystrophin is part of an incredibly complex group of **proteins** that allow muscles to work correctly. The protein helps anchor various components within muscle cells together and links them all to the **sarcolemma** - the outer membrane. If **dystrophin** is absent or deformed, this process does not work correctly, and disruptions occur in the **outer membrane**. This weakens the muscles and can also actively damage the muscle cells themselves.

In Duchenne muscular dystrophy, dystrophin is almost totally absent; the less **dystrophin** that is produced, the worse the symptoms and **etiology** of the disease. In **Becker muscular dystrophy**, there is a reduction in the amount or size of the **dystrophin protein**. The gene coding for **dystrophin** is the **largest** known gene in humans. More than **1,000 mutations** in this gene have been identified in **Duchenne and Becker muscular dystrophy**.

Currently, **there is no cure for muscular dystrophy**, but certain physical and medical treatments can improve symptoms and slow the disease's progression. However, some important facts about the muscular dystrophy are as following:

1. Muscular dystrophy is a collection of **muscle -wasting conditions**.

2. Duchenne muscular dystrophy is the most **common type**.
3. A lack of a protein called **dystrophin** is the main cause of muscular dystrophy.
4. **Gene therapies** are currently being trailed to combat the disease.
5. There is currently **no cure for muscular dystrophy**.

Muscular dystrophy is a muscle-wasting disease whose predominant forms may affect up to 1 in every 5,000 males. The condition is caused by a **genetic mutation** that interferes with the production of muscle proteins necessary to build and maintain healthy muscles. The **disease is genetic**, and consequently, a history of muscular dystrophy in the family increases the chance of an individual developing the disease.

TYPES OF MUSCULAR DYSTROPHY

There are a number of muscular dystrophy types which are as following:

- **DUCHENNE MUSCULAR DYSTROPHY:** The most **common form** of the illness. Symptoms normally start before a child's third **birthday**; they are generally wheelchair-bound by 12 and die of **respiratory failure** by their early-to-mid-twenties.
- **BECKER MUSCULAR DYSTROPHY:** Similar symptoms to **Duchenne** but with a later onset and slower progression; death usually occurs in the mid-forties.
- **MYOTONIC (Steinert's disease):** The **myotonic** form is the most common adult-onset form. It is characterized by an inability to relax a muscle once it has contracted. The muscles of the face and neck are often affected first. Symptoms also include **cataracts, sleepiness, and arrhythmia**.
- **CONGENITAL:** This type can be obvious from birth or before the age of **2**. It affects girls and boys. Some forms progress slowly whereas others can move swiftly and cause significant **impairment**.
- **FACIOSCAPULOHUMERAL (FSHD):** Onset can be at almost any age but is most commonly seen during **teenage years**. The muscular weakness often begins in the face and shoulders. People with **FSHD** may sleep with their eyes slightly open and have trouble fully closing their **eyelids**. When an individual with **FSHD** raises their arms, their shoulder blades protrude like wings.
- **LIMB-GIRDLE:** This variant begins in **childhood or teenage years** and first affects the **shoulder and hip muscles**. Individuals with the limb-girdle muscular dystrophy might have trouble raising the front part of the foot, making tripping a common problem.
- **OCULOPHARYNGEAL MUSCULAR DYSTROPHY:** Onset is between the ages of 40 and 70. **Eyelids, throat, and face** are first affected, followed by the shoulder and pelvis.

SYMPTOMS OF MUSCULAR DYSTROPHY:-

Below are the symptoms of **Duchenne muscular dystrophy**, the most common form of the disease. The symptoms of **Becker muscular dystrophy** are similar but tend start in the mid-twenties or later are milder and progress more slowly.

Initial symptoms

- A waddling gait.
- Pain and stiffness in the muscles.
- The difficulty with running and jumping.
- Walking on toes.
- Difficulty sitting up or standing.
- Learning disabilities, such as developing speech later than usual.

Later symptoms

- Inability to walk.
- A shortening of muscles and tendons, further limiting movement.
- Breathing problems can become so severe that assisted breathing is necessary.
- The curvature of the spine can be caused if muscles are not strong enough to support its structure.
- The muscles of the heart can be weakened, leading to cardiac problems.
- Difficulty swallowing - this can cause aspiration pneumonia, and a feeding tube is sometimes necessary



Fig.4.2 Sign of Muscular dystrophy

4.4 Dominant Inherited Disorder:-

This category includes those **disorders** the **genes** for which are dominant over their **alleles**. Such disorders are inherited like typical **Mendelian dominant characters**. An essential feature of a dominantly inherited disorder is that it is always present in at least **one parent** in each ancestral generation. Short stature, **polydactyly**, **opalescent teeth**, **Huntington's chorea** and **achondroplastic dwarfism** are some important dominant defective traits.

1. Polydactyl:-

The disorder arising from the presence of an **extra digit** on the hand or foot is called **polydactyl**. This **physical disorder** is dominant in man. There are cases on record where this disorder skips a few generations and then reappear again. In humans/animals this condition can present itself on one or both **hands**. The extra digit is usually a small **piece of soft tissue** that can be removed.

Occasionally, it contains bone without joints; rarely may it be a complete functioning digit. The extra digit is most common on the **ulnar** (little finger) side of the hand, less common on the **radial** (thumb) side, and very rarely within the middle three digits. These are respectively known as **postaxial** (little finger), **preaxial** (thumb), and **central** (ring, middle, index fingers) **polydactyl**. The extra digit is most commonly an abnormal fork in an existing digit, or it may rarely originate at the wrist as a normal digit does.

The incidence of **congenital deformities** in newborns is approximately **2%** and **10%** of these deformities involve the upper extremity. Congenital anomalies of the limb can be classified into seven categories, proposed by **Frantz** and **O'Rahilly** and modified by **Swanson**, based on the embryonic failure causing the clinical presentation. These categories are a **failure of formation**

of parts, failure of differentiation, duplication, overgrowth, undergrowth, and congenital constriction band syndrome and generalized **skeletal abnormalities**.

Polydactyl belongs to the category of **duplication**. Because there is an association between **polydactyl** and **several syndromes**, children with a congenital upper extremity deformity should be examined by a geneticist for other congenital anomalies. This should also be done if a syndrome is suspected or if more than two or three generations of the family are affected.

Research has shown that the majority of **congenital anomalies** occur during the **4-week embryologic** period of rapid limb development. **Polydactyl** has been associated with **39 genetic mutations**. **Polydactyl** can be divided into three major types, which are discussed below.

Ulnar or Postaxial Polydactyl

This is the most **common situation** in which the extra digit is on the **ulnar** side of the hand, thus the side of the little finger. This can also be called **postaxial polydactyly**. It can manifest itself very subtly, for instance only as a **nubbin** on the **ulnar** side of the little finger, or very distinctly, as a fully developed finger. Most commonly, the extra finger is **rudimentary**, consisting of an end phalanx with a nail and connected to the hand with a small skin pedicle.

Radial or Preaxial Polydactyl:-

This is a **less common** situation, which affects the side of the hand towards the thumb. **Radial polydactyly** refers to the presence of an extra digit (or extra digits) on the radial side of the hand. It is most frequent in **Indian populations** and it is the second most common congenital hand disorder.

Central Polydactyl:-

This is a very **rare situation**, in which the extra digit is on the ring, middle or index finger. Of these fingers, the index finger is most often affected, whereas the ring finger is rarely affected. This type of **polydactyl** can be associated with **syndactyly**, cleft hand, and several syndromes. **Polysyndactyly** presents various degrees of **syndactyly** affecting fingers three and four.

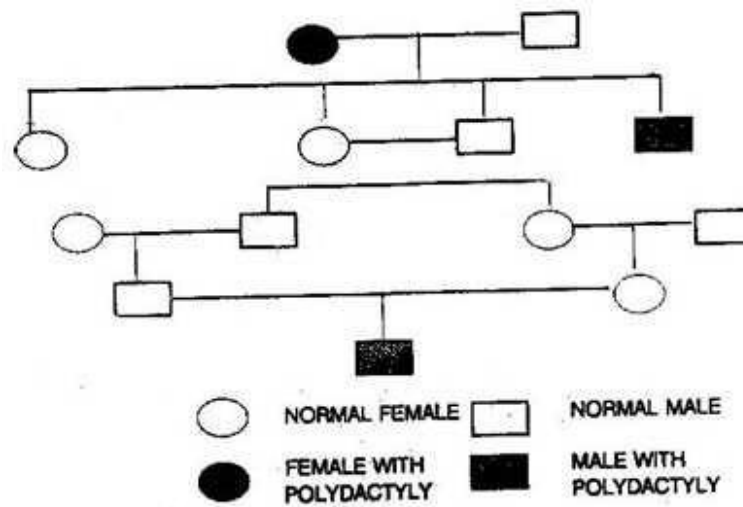


Fig.4.3 Family pedigree of polydactyl in man

2. Huntington's Chorea:-

It is an uncommon inherited disease which causes **gradual mental deterioration** and eventually results in death. **Huntington's disease (HD)**, also known as **Huntington's chorea**, is an **inherited** that results in the death of brain cells. The earliest symptoms are often subtle problems with mood or mental abilities. A general lack of coordination and an **unsteady gait** often follow.

As the disease advances, uncoordinated, jerky body movements become more apparent. Physical abilities gradually worsen until coordinated movement becomes difficult and the person is **unable to talk**. Mental abilities generally decline into **dementia**. The specific symptoms vary somewhat between people.

Symptoms usually begin between **30 and 50 years** of age, but can start at any age. The disease may develop earlier in life in each **successive generation**. About **8%** of cases start before the age of **20 years** and typically present with symptoms more similar to **Parkinson's disease**. People with **HD** often underestimate the degree of their problems.

HD is typically inherited from a person's parents, with **10%** of cases due to a new mutation. The disease is caused by an **autosomal dominant mutation** in either of an individual's two copies of a gene called **Huntingtin**. This means a child of an affected person typically has a **50% chance** of inheriting the disease. The **Huntingtin gene** provides the genetic information for a protein that is also called "**huntingtin**".

3. A Chondroplastic Dwarfism:-

It is also an **inherited dwarfism** that affects the **long bones** of the body which do not grow to normal size, although in every other way the affected individuals are normal. The trait is **dominant** and is exhibited in **every generation**. It does not appear to be sex-linked, as both female and male dwarfs are seen. **Achondroplasia** is a common cause of **dwarfism**. It occurs as a sporadic mutation in approximately **80% of cases** (associated with advanced paternal age) or it may be inherited as an **autosomal dominant genetic disorder**.

People with **achondroplasia** have **short stature**, with an average adult **height of 131 centimeters (52 inches)** for males and **123 centimeters (48 inches)** for females. **Achondroplastic** adults are known to be as short as **62.8 cm (24.7 in)**. If both parents of a child have **achondroplasia** and both parents pass on the mutant gene, then it is very unlikely that the homozygous child will live past a few months of its life. The prevalence is approximately **1 in 25,000**.

Achondroplasia is caused by a mutation in **fibroblast growth factor receptor 3 (FGFR3)**. In normal development, **FGFR3** has a negative regulatory effect on bone growth. In **achondroplasia**, the mutated form of the receptor is constitutively active and this leads to **severely shortened bones**. The effect is **genetically dominant**, with one mutant copy of the **FGFR3 gene** being sufficient to cause **achondroplasia**, while two copies of the mutant gene are invariably **fatal** (recessive lethal) before or shortly after birth (**known as a lethal allele**).

SYMPTOMS

- Disproportionate **dwarfism**.
- Shortening of the **proximal limbs** (called rhizomelic shortening).
- Short fingers and toes with **trident hands**.
- Large head with prominent forehead **frontal bossing**.
- Small **midface** with a flattened nasal bridge.
- Spinal **kyphosis** (convex curvature) or **lordosis** (concave curvature).
- **Varus** (bowleg) or **valgus** (knock-knee) deformities.
- Frequently have ear infections (due to **Eustachian tube blockages**), sleep apnea (which can be **central or obstructive**).

4.5 Inborn Errors:-

Inborn errors of metabolism form a large class of **genetic diseases** involving **congenital disorders**. The majority are due to defects of **single genes** that code for enzymes that facilitate the conversion of various substances (substrate) into others product. In most of the disorders, problems arise due to the accumulation of substances which are toxic or interfere with normal

function, or to the effects of reduced ability to synthesize essential compounds. Inborn errors of metabolism are now often referred to as **congenital metabolic diseases** or **inherited metabolic diseases**.

The term **inborn error of metabolism**, was coined by a British physician, **Archibald Garrod (1857–1936)**, in **1908**. He is known for work that prefigured the "**one gene -one enzyme**", based on his studies on the nature and inheritance of **alkaptonuria**.

Traditionally the inherited metabolic diseases were classified as **disorders of carbohydrate metabolism, amino acids metabolism, organic acid metabolism, or lysosomal storage diseases**. In recent decades, hundreds of newly inherited disorders of metabolism have been discovered and the categories have proliferated.

Symptoms:-

Because of the **enormous number** of these diseases and a wide range of systems affected, nearly every "**presenting complaint**" to a doctor may have a congenital metabolic disease as a possible cause, especially in **childhood**. The following are examples of potential manifestations affecting each of the major organ systems.

- Growth failure, failure to thrive, weight loss.
- Ambiguous genitalia, delayed puberty, precocious puberty.
- Development delay, seizures, dementia, encephalopathy, strokes.
- Deafness, blindness, pain amnesia.
- Skin rash, abnormal pigmentation, lack of pigmentation, excessive hair growth, lumps, and bumps
- Dental abnormalities
- Immunodeficiency, low platelets count, low red blood cell count, enlarged spleen, enlarged lymph nodes.
- Many forms of cancer.
- Recurrent vomiting, diarrhea, abdominal pain.
- Excessive urination, kidney failure, dehydration, edema.
- Low blood pressure, heart failure, enlarged heart, hypertension, myocardial infarction.
- Liver enlargement, jaundice, liver failure.
- Unusual facial features, congenital malformations.
- Excessive breathing (hyperventilation), respiratory failure.
- Abnormal behavior, depression, psychosis.
- Joint pain, muscle weakness, cramps.
- Hypothyroidism, adrenal insufficiency, hypogonadism, diabetes mellitus.

4.6 Summary:-

1. The diseases which are transferred from generation to generation are called genetic disorders or genetically transmitted diseases.
2. A genetic disorder may be due to incompatible genes or abnormalities in the structure or number of chromosomes.
3. The branch of genetics which deals with the inheritance of genetic characters in human beings is known as human genetics.
3. A British physician Sir Archibald Garrod was the pioneer of human genetics.
4. Autosomal recessive disorder is genetic disease that passed to the child through both parents.
5. XX-chromosomes are found in a female while XY-chromosome is found in male in the case of human beings.
6. Albinism is a genetic disorder in which melanin pigment does not form in the body.
7. Disease albino is being produced due to the absence of enzyme melanocyte tyrosinase leads to failure of melanin pigment from tyrosine.
8. Tay-Sachs disease is a fatal disorder in children that cause a progressive degeneration of Central Nervous System (CNS).
9. Tay-Sachs disorder takes place due to the absence of an enzyme called hexosaminidase A, generally known as 'hex A'.
10. Cystic fibrosis is a type of genetic disorder in which mucus becomes thick and sticky.
11. Sickle cell anemia is a common inherited single gene disorder in which smooth, round and flexible RBCs just appear like letter "O" change in stiff and sticky appear like letter "C".
12. Sickle cell loses their oxygen and just change the shape like letter "C".
13. Sickle cell's life span is 15 days, while RBCs live 120 days.
14. Formation of phenylalanine is a type of amino acid cause of phenylketonuria, a rare genetic disorder.

15. Autosomal recessive polycystic kidney disease (ARPKD) is a hereditary renal disease caused by PKHD1.

16. Hemophilia is a genetic disorder in which blood does not clot.

17. Hemophilia may be hemophilia A, hemophilia B, parahemophilia and hemophilia C.

18. Color blindness is a type of genetic disorder in which individually is not able to distinguish between red and green color.

19. Muscular dystrophy is a genetic disorder in which progressive loss of muscle mass and consequent loss of strength take place.

20. Huntington's disease (HD) also known as Huntington's chorea is an inherited disease which damages the brain cells.

4.7 Self Assessment Questions:-

1. Down's syndrome is characterized by:

- a) 19 trisomy
- b) 21 trisomy
- c) Only one X chromosome
- d) Two X and one Y chromosome

Ans. b) 21 trisomy

2. Which of the following is known as a royal disease?

- a) Sickle cell anemia
- b) Hemophilia
- c) Alzheimer's disease
- d) Color blindness

Ans. b) Hemophilia

3. Patau's syndrome occurs due to:

- a) Trisomy of 13th chromosome
- b) Trisomy of 18th chromosome
- c) Trisomy of 21st chromosome
- d) Trisomy of 22nd chromosome

Ans. a) Trisomy of 13th chromosome

4. The most important example of point mutation is found in a disease called?

- a) Thalassemia
- b) Night blindness
- c) Sickle cell anemia
- d) Down's syndrome

Ans. c) Sickle cell anemia

5. The syndrome in which individual somatic cell contains three sex chromosome XXX is called:

- a) Down's syndrome
- b) Super female
- c) Turner's syndrome
- d) Klinefelter's syndrome

Ans. b) Super female

6. A man has enlarged breasts, sparse hair on body and sex complement as XXY. He suffers from:

- a) Down's syndrome
- b) Turner's syndrome
- c) Edward's syndrome
- d) Klinefelter's syndrome

Ans. d) Klinefelter's syndrome

7. In a family, father is having a disease and mother is normal. The disease is inherited to only daughters and not to the sons. What type of disease is this?

- a) Sex-linked dominant
- b) Sex-linked recessive
- c) Autosomal recessive
- d) Autosomal dominant

Ans. a) Sex-linked dominant

8. In man, which type of the following genotypes and phenotypes may be the correct result of aneuploidy in sex chromosome?

- a) 22 pairs + Y females
- b) 22 pairs + XX female

- c) 22 pairs + XXY female
- d) 22 pairs + XXXY female

Ans. c) 22 pairs + XXY female

9. A woman with one gene for hemophilia and one gene for color blindness on one of the X chromosomes marries a normal man. How will the progeny be?

- a) Hemophilic and color blind daughters
- b) All sons and daughters are hemophilic and color blind
- c) 50% hemophilic and color blind sons and 50% normal sons
- d) 50% hemophilic and color blind sons and 50% color blind daughters

Ans. c) 50% hemophilic and color blind sons and 50% normal sons

10. Type of genotype in individual having blood group B will be

- a) $I^A I^O$
- b) $I^A I^B$
- c) $I^O I^O$
- d) $I^B I^O$

Ans. d) $I^B I^O$

11. If a gene product in species A is 90% similar to gene product in species B. Such genes are termed as:

- a) Orthologous
- b) Paralogous
- c) Allologous
- d) Perillogous

Ans. a) Orthologous

12. The probable inheritance is if the inheritance of a disease to next generation is only possible through females:

- a) Sex-linked
- b) Mendelian
- c) Organellar
- d) Autosomal

Ans c) Organellar

13. Genetic disorder *Xeroderma pigmentosum* is due to an error in:

- a) Base excision repair mechanism
- b) Direct repair mechanism
- c) Nucleotide excision repair mechanism
- d) DNA replication mechanism

Ans. c) Nucleotide excision repair mechanism

14. A mother of blood group O has a group O child, the father could be of blood group:

- a) A or B
- b) AB only
- c) O only
- d) A or B or O

Ans. d) A or B or O

15. A heterozygous individual which carries the unexpressed recessive gene for a sex-linked character is known as:

- a) Carrier
- b) Mutant
- c) Variant
- d) None of these

Ans. a) Carrier

4.8 Terminal Questions:-

- 1- Write a short note on Sickle cell anemia?
- 2- Write a short note on Phenylketonuria?
- 3- What is muscular dystrophy?
- 4- Describe in detail about Autosomal recessive disorders with suitable examples?
- 5- Write a detail account on various Sex-linked recessive disorders?

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UNIT 5: TAXONOMY AND SYSTEMATICS

Contents

- 5.1 Objectives
- 5.2 Introduction
- 5.3 Introduction to taxonomy and its relationship with systematic
- 5.4 Importance and application of biosystematics
- 5.5 Summary
- 5.6 Self Assessment Questions
- 5.7 Terminal Questions

5.1 Objectives:-

As **more** and more data regarding life forms come to light, **taxonomy** becomes more and more refined. To understand diversity a system of taxonomy is required. Taxonomy allows understanding diversity better. This chapter will deal with how living beings are named, grouped and classified and some important features of these groups.

About **2.0 million** types of animals and about **1.0 million** types of plants have already been described and named and many new forms are discovered every year. Animals and plants vary greatly in their forms, structures and mode of life. To identify an organism of known characters from the vast number of organisms is simply impossible. The number and **diversity** of living organisms are so enormous that it is very difficult to study without classifying them into certain groups or categories. Thus taxonomy allows us to identify and recognize organisms. Hence, in this chapter, you would learn about the role of taxonomy in the classifying the **organisms** and its relationship with **systematic**.

5.2 Introduction:-

Initially, an attempt was made to classify plants and animals on their habitat, distribution (air, land, and water), beneficial and harmful basis. **Aristotle (384-332 B.C.)** who is known as "**Father of Zoology**" classified animals on the basis of their morphology and categorized into three groups namely as:

- I. Vermes
- II. Insecta
- III. Vertebrata

He classified a total of **520 species** of animals in his book "**Histiria Animalium**". Due to his unique tremendous contribution in **Zoology**, he is also known as: "**Father of Biological Classification**".

John Ray (1627-1705) was first coined the term "**species**" and described **18000 plants** in his book entitled "**Historia Generalis Plantarum**" which was published in three volumes between **1686 to 1704**. He was the first person who made a differentiation between **genus and species**.

In this series, **Theophrastus (370-385 B. C.)**, who was the student of **Plato** and **Aristotle** known as "**Father of Botany**" classified **480 plants** into four groups into his famous book "**Historia Plantarum**". These four groups as follows:

- I. Trees

II. Shrubs

III. Under shrubs

IV. Herbs

However, it was followed by a Swedish Naturalist **Carolus Linnaeus (1707-1778)**, who used "**Binomial Nomenclature**" system of classification instead of using common name of plants and animals both in his famous book entitled "**Systema Naturae**". He listed **9378** species of plants and animals in his book which had published in **1735**. Because of using a scientific system of classification firstly, Carolus Linnaeus has been crowned with the title of "**Father of Modern Taxonomy**".

5.3 Introduction to Taxonomy and its Relationship with Systematic:-

TAXONOMY: (Gk., Taxis -arrangement, nomos - law). The term taxonomy was first coined by **A. P. de Candolle in 1813**. Taxonomy may be defined as the branch of science which deals with the **identification, nomenclature and classification** of any plant or animal all over the world is called taxonomy.

There are about **2.0 million** of species and many are being discovered by taxonomists in all over the world. All of these species may be classified according to the norms of taxonomy and each species may be identified separately by its peculiar characteristics. In the absence of taxonomy, it would be very difficult to recognize, identify and classify plants and animals without committing any mistakes. It means without **systematics** there is no significance of taxonomy and without taxonomy, there is no existence of systematics. **Systematics and taxonomy both are complementary to each other.**

However, the study of taxonomy can be done under the following headings:

- 1. CLASSIFICATION:** Classification may be defined as a system of arrangement of individuals into various categories which exhibit a relationship with each other.
- 2. IDENTIFICATION:** To determine the exact place or position of any plant or animal according to the system of classification (systematics) is called identification.
- 3. NOMENCLATURE:** Nomenclature may be defined as a process of giving a name to plants and animals according to the systematics.
- 4. KEY:** Those distinguishing or diagnostic characters which help in the identification of any plant or animal in the systematics are called key.

Taxonomy is the science of defining groups of biological organisms on the basis of shared characteristics and giving names to those groups. Organisms are grouped together into **taxa** (singular: taxon) and these groups are given a **taxonomic rank**. Groups of a given rank can be aggregated to form a super group of lower rank, thus creating a **taxonomic hierarchy**. The Swedish botanist **Carl Carolus Linnaeus** is regarded as the "**Father of Taxonomy**", as he developed a system known as **Linnean classification** for categorization of organisms and Binomial Nomenclature for naming organisms.

With the advent of such fields of study as **phylogenetics**, **cladistics**, and **systematics**, the Linnaean system has progressed to a system of modern biological classification based on the **evolutionary relationships** between organisms, both living and extinct. An example of a modern classification is the one published in **2015** for all extant taxa (to the level of Order) by **M. Ruggiero and co-workers**.

In the branch of biology, that deals with the framing of laws and principles of classifying the organisms on the basis of their evolutionary relationship. The main aim of the taxonomic study is to assign an appropriate place to an organism in a systematic framework of classification. This framework is called **taxonomic hierarchy**.

In this, the taxonomic groups are arranged in a definite order, from higher to lower categories. Each category is considered a **taxonomic unit** which represents a **taxon**. A natural taxon refers to a group of similar, genetically related individuals having certain characters distinct from those of other groups. For example, all the insects form a taxon.

In classification, the organisms that closely resemble one another are placed in a group; the groups which have similarities are combined together into larger groups and these into still larger ones. These various grouping levels or ranks in classification are known as **categories**. The **taxonomic hierarchy** or hierarchy of categories was first established by **Linnaeus (1758)** in the animal kingdom. The **seven** major categories, in descending order, are:

Kingdom

Phylum

Class

Order

Family
Genus
Species

CATEGORY V/S TAXON

Category	Taxon
1. It is only an abstract term.	1. It is a group of concrete biological objects and is assigned to a category.
2. It represents a rank or level in classification.	2. It represents a group of real organisms.

EXAMPLES

1. The taxon of birds is **Aves** and their category is **Class**.
2. The sponges from the taxon **Porifera** and their category is **Phylum**.
3. *Rosa Indica* is a **taxon** and Species is a **category**.

INTERMEDIATE CATEGORIES

With the discovery of more and more organisms, it becomes difficult to place an organism in the **traditional categories**. Hence, to make the taxonomic position of a species more precise, the categories have been split by prefixing "**super**" or "**sub**" to the existing categories. As they are introduced later on in the hierarchical system, they are called intermediate categories. Thus we have:

Sub-kingdom

Super-division

Super-phylum

Sub-division

Super-class

Sub-class

Sub-order

Sub-family

Genus

Species

SPECIES:-

It occupies a **key position** in taxonomy. It is the **basic unit** for understanding **taxonomy** and **evolution**. A species is defined as "a dynamic, genetically distinct group of organisms, which resemble one another in all essential characters (morphological and reproductive) and interbreed freely in nature to produce fertile offspring's".

For example, mango (*Mangifera indica*), potato (*Solanum tuberosum*) and lion (*Panthera leo*). In this case, indica, tuberosum, and leo are the species of genera *Mangifera*, *Solanum* and *Panthera* respectively.

(The individual of species also represents population of species and they do not breed with individuals of other species).

GENUS:-

It is a group of an assemblage of related species which resemble one another in certain characters. Species, in a genus, usually have many features in common. Such groups of common features are called correlated characters. All the species of a genus are presumed to have evolved from a **common ancestor**.

The genus has a **significant position** in classification. By the rule of binomial nomenclature, a species cannot be named unless it is assigned to a genus. Sometimes a genus may consist of only one existing species.

For example, modern man to the genus *Homo*, such a genus is called monotypic. The other consisting of many species are called polytypic. For example, the genus *Panthera* has a large number of closely related species such as *Panthera leo* (lion), *P. pardus* (leopard) and *P. onca* (jaguar).

FAMILY:-

It is the taxonomic category which contains one or **more related genera**. All the genera of a family have some common features. They are separable from the genera of a related family by some important and characteristics differences. The genera of cats (*Felis*) and leopard (*Panthera*) are included in the family Felidae.

ORDER:-

It is the next higher taxonomic category which includes related families. For example, the families, Canidae, Hyaenidae (hyenas) and Ursidae (bears) are included under the order Carnivora.

CLASS:-

This category includes one or more **related orders**. For example, class Mammalia of animals includes orders of all mammals like Chiroptera (bats), Marsupialia (kangaroos), Rodentia (rodents), Cetacea (whales), Carnivora (carnivores), Primates (apes and man).

DIVISION OR PHYLUM:-

It is formed of one or more related classes. The term **phylum** (Pl. - phyla) is commonly employed for animals while **division** is used for plants. For example, Phylum Chordata of animals includes several classes like Cyclostomata (Lamprey), Chondrichthyes (Cartilaginous fish), Osteichthyes (Bony fish), Amphibia, Reptilia, Aves (Birds) and Mammalia.

KINGDOM:-

It is the **highest taxonomic category**. It includes one or more related divisions or phyla. In the Linnaeus system of classification, all plants are included under kingdom **Plantae** and all animals under the kingdom **Animalia**.

TYPES OF TAXONOMY:-

- 1. α (ALPHA) TAXONOMY:** If taxonomy is concerned with characterization and naming of any species is called alpha-taxonomy.
- 2. β (BETA) TAXONOMY:** If taxonomy is concerned with the arrangement of species according to the law of systematics is called beta taxonomy.
- 3. γ (GAMA) TAXONOMY:** Ultimately when taxonomy is concerned with some biological aspects like taxa, evolutionary rate and trends then it is called gamma taxonomy.

SYSTEMATICS:-

Systematics (Gk., Systema - a system of classification). Term systematics was first described by Carolus **Linnaeus** in his book "**Systema Naturae**". Systematics may be defined as the scientific study of taxonomy which deals identification, nomenclature, and classification of all living individuals and relationships among them are called systematic.

Biological systematics is the study of the diversification of living forms, both past and present, and the relationships among living things through time. It is a modified form of classical systematics (old systematics) which was first used by **Plato and Aristotle**.

The term **New Systematics** (neosystematics or biosystematics) was proposed by **J. Huxley in 1940** to consider some new branches of taxonomy like Morphotaxonomy, Karyotaxonomy, Cytotaxonomy, Experimental Taxonomy, Biochemical Taxonomy, Chemotaxonomy and Numerical Taxonomy etc. Besides this “New Systematics” also deals many aspects of morphology, ecology, biochemistry, physiology, cytology and genetics etc.

Hence, systematic biology is the field that:-

- A.** Provides scientific names for organisms
- B.** Describes them,
- C.** Preserves collections of them,
- D.** Provides classifications for the organisms, keys for their identification and data on their distributions,
- E.** Investigates their evolutionary histories, and
- F.** Considers their environmental adaptations.

This is a field with a long history that in recent years has experienced a notable renaissance, principally with respect to theoretical content. Part of the theoretical material has to do with **evolutionary areas** (topics e and f above), the rest relates especially to the problem of classification. Taxonomy is that part of Systematics concerned with topics **(a) to (d)** above.

Taxonomy, systematic biology, systematics, biosystematics, scientific classification, biological classification, phylogenetics: At various times in history, all these words have had **overlapping meanings** — sometimes the same, sometimes slightly different, but always overlapping and related. However, in modern usage, they can all be **considered synonyms** of each other. For example, Webster's 9th New Collegiate Dictionary of 1987 treats "**classification**", "taxonomy", and "systematics" as synonymous.

Europeans tend to use the terms "**systematics**" and "**biosystematics**" for the field of the study of **biodiversity** as a whole, whereas **North Americans** tend to use "taxonomy" more frequently. However, taxonomy, and in particular alpha taxonomy, is more specifically the identification, description, and naming (i.e. nomenclature) of organisms, while "**classification**" focuses on placing organisms within hierarchical groups that show their relationships to other organisms. All of these biological disciplines can deal both with extinct and with extant organisms.

Systematics uses taxonomy as a primary tool in understanding, as nothing about an organism's relationships with other living things can be understood without it first being properly studied and described in sufficient detail to identify and classify it correctly. Scientific classifications are

aids in recording and reporting information to other scientists and to laymen. The **systematist**, a scientist who specializes in systematics, must, therefore, be able to use existing classification systems or at least know them well enough to skillfully justify not using them.

PRINCIPALS OF SYSTEMATICS

Systematics is the scientific study that attempts to recognize, describe, name and arrange the diverse organisms according to an organized plan based on the unique features of species and groups. It is also called the science of diversity of organisms because it involves a shift from **diversity** to unity through comparison among individuals.

FIELDS OF SYSTEMATICS

The basic **requirements in systematics** are as following:

- I. The arrangement of organisms into groups.
- II. A system for naming the organisms.
- III. Framing the rules for Classification, Nomenclature, and Taxonomy.

UTILITY OF SYSTEMATICS

It provides useful information about the **evolution**, **adaptations** and **diversity** of organisms.

1. It is essential for the study of other branches of life sciences like ecology, cytology and genetics etc.
2. It helps in the identification of crop pests and thus planning in their eradication.
3. It helps in solving the problems of various epidemic diseases throughout the world.
4. It helps in the identification of plants and animals with superior genomes for breeding programs.
5. It helps in the identification of indicator organisms, which provide information about pollution, availability of ground water and minerals etc. in a particular area.
6. It enables us to identify the fossils which give us full clues about the phylogeny of organisms.

However, these terms are often used interchangeably as they are complementary. Biosystematics deals with the variation within a species and its general evolution.

RELATIONSHIP BETWEEN TAXONOMY AND SYSTEMATICS

Taxonomy is concerned with the classification and naming of organisms. Since Darwin's proposal that all organisms on earth share a common ancestor, taxonomists have made sure that

organism that do not share a recent common ancestor are not classified in the same group formally. Taxonomists call groups that have 2 or more separate recent common ancestors "polyphyletic." No taxonomists will knowingly recognize polyphyletic groups.

However, mistakes are sometimes made, even with the best intentions, and sometimes taxonomists do group organisms that are only superficially similar to the same group, resulting in a polyphyletic group. A prime example is Pachydermata, a taxon (group with a name) that is no longer recognized because it is polyphyletic. Pachydermata consisted of thick-skinned, large land mammals like elephants, hippos and rhinos. However, it has been shown that they are only superficially similar because elephants are more closely related to elephant shrews and hippos are most closely related to pigs, cows and whales. The rhinos are in turn more closely related to horses than to the elephants and hippos. Therefore, Pachydermata is no longer recognized as a valid taxon.

Sadly, many practicing taxonomists no longer concern themselves with a number of evolutionary changes that have occurred within or between lineages. Some of them, called cladists, are misguided in their classificatory practice and they recognize such groups as the birds + living reptiles as "Reptilia." Darwinians recognize that birds are distinct from reptiles and classify birds in Aves and living reptiles in Reptilia. Because of these differences in classification philosophy, there is no consensus on the classification of many groups. The result is taxonomic chaos that is going to be around for decades to come.

Systematics is concerned with the evolutionary relationships of organisms. Systematists are concerned with ascertaining which organisms share a recent ancestry with which other organisms. Systematists are also concerned about a number of evolutionary changes that may have occurred within and between lineages.

Systematics is the study of the **units of biodiversity**. Systematics differs from ecology in that the latter is concerned with the interactions of individuals (and therefore species) in a particular time, while the former is concerned with the diversification of **lineages through time**.

Systematics includes the discovery of the basic units of **biodiversity** (species), reconstructing the patterns of relationships of species at successively higher levels, building classifications based on these patterns and naming appropriate **taxa** (taxonomy) and the application of this pattern knowledge to study changes in organisms' features through time. It also includes the building and maintenance of **biodiversity** collections, upon which all the products of systematic studies are based.

Ultimately it may be concluded that taxonomy is the classification and naming of all living things, while, systematics refers to the study of the relationships between these living things as they evolve. The taxonomic hierarchy was devised and published by Swedish scientist Carl Linne in 1735. All branches of systematics, such as Botany, Zoology, Microbiology and

Mycology, are covered under taxonomy. Taxonomy as the science of biological classification is a subdivision of systematics.

5.4 Importance and Application of Biosystematics:-

Systematics is concerned with the evolutionary relationships of organisms. Systematists are concerned with ascertaining which organisms share a recent ancestry with which other organisms. Systematists are also concerned about a number of evolutionary changes that may have occurred within and between lineages. As the **sub-discipline** of biology that investigates relationships of taxa, systematics is the foundation for **comparative biology**. Comparative biology is that type of study that attempts to relate features of one organism, or type of organism, to features in another type of organism. This always is a question of **homology** or sameness due to the common **evolutionary origin**.

In **systematic studies**, we hypothesize homology of features among taxa and then gather data to test these **hypotheses**. This is important because appearance alone is often not a good indicator that features in different taxa are homologous -- many times similar structures will evolve independently in different lineages. If they are **homologous**, we expect that they will share many things because of their common **ancestry**, while if they are not, it is impossible to predict just how similar they will be. Hence, any study that asks why or how about a feature in more than one taxon, and draws comparative conclusions about them, rests on a **systematic foundation**.

APPLICATIONS:-

Biosystematics is playing a very crucial role in this living being world. There are more than **two million species of animals** and about **one million species** of plants, while several more to be discovered yet. These species may be identified and classified with the help of taxonomy according to the rules and regulations of systematics. Otherwise, in the absence of systematics (taxonomy), it would have been very difficult to isolate these species with a particular name.

In addition without knowledge of systematics it would be very difficult to discover new species and also there should not be made any differentiation between them. No **scientific survey** can be made without prior knowledge of systematics. Besides this, there is no being left importance of **civilization** because, civilization and systematics are complementary to each other in the sense that if a man maintains his life (daily routine work) or home systematically, means he is civilized otherwise like animals.

In the same sense or way, there is a vast variety amongst plants and animal species which all are systematically or in the civilization manner are well being arranged only by systematics can be summarized as follows:

1. ROLE OF SYSTEMATICS IN DIVERSITY: It has been already stated that there is a vast variety of plants and animals on the planet. They belong to different **habits** and **habitats**. Several species of plants and animals are to be discovered yet. Systematics or taxonomy provides us different kinds of information about the **ecology** of all these species. **Phylogeny** and evolutionary processes of species could be understood only through systematics.

2. ROLE OF SYSTEMATICS IN APPLIED BIOLOGY: Systematics has been playing a very important role in applied biology. Crops and trees of economic importance are being destroyed at large scale by various types of **pests**. Without knowing the name of such pests, it will be very difficult to eradicate them. Harmful and beneficial plants can be checked with the knowledge of systematics. Several **diseases** of plants and animals can be checked only through the knowledge of systematics.

3. ROLE OF SYSTEMATICS IN PUBLIC HEALTH: Use of many **insecticides** to control the pests is the cause of many health problems in men and their pet animals. Several **diseases** like malaria, filaria, dysentery, dengue fever, Kala - Azar and sleeping sickness are due to cause of **mosquitoes** and **protozoans**. Many water borne, air borne and noise borne diseases are also rapidly spreading. To control these diseases and other health problems, it is essential to correctly identify their sources, vector and control strategy should be planned in such a way that the target source of diseases is being attacked only. It is possible only with the help of systematics.

4. ROLE OF SYSTEMATICS IN PRESERVATION OF WILDLIFE: India is well known for the variety of **wildlife** found here. There are about two million species of animals and one million species of plants. But during the past 50-60 years, wildlife has depleted rapidly due to **indiscriminate killing** of animals and illegal falling of tree and deforestation. Now India faces a crisis of ecological imbalance on a massive scale. During the last three decades alone **95 species** of birds and **37 species** of mammals have been extinct. Today, about a total sum of **200 species** of birds and **100 species** of mammals are facing severe threat of extinction. Systematics identifies such animals and plants and help in the protection of the environment.

5. ROLE OF SYSTEMATICS IN ENVIRONMENT PROTECTION: All living organisms depend upon a balanced environment for their growth and development. All components of the balanced environment are present in a definite ratio but sometime this ratio became disturbed and affects the organism's life. Several undesirable **xenobiotics** and pollutants are entering in our environment continuously by manmade activities.

These activities are the major cause of depletion of our environment. Noise pollution, water pollution, land pollution, air pollution and various chemicals, heavy metals, pesticides, biocides, insecticides, asbestos, fertilizers, antibiotics and detergents are the major cause of environment

depletion. Systematics can play a very important role in the identification of these various kinds of **xenobiotics** properly and could save the depleted environment.

6. ROLE OF SYSTEMATICS IN COMMERCE: Various useful products like honey, silk, and dye are obtained from insects. Many useful varieties can be used for commercial production by **manipulating species**. The production yields can be increased by replacing harmful or neutral germplasm by better quality of germplasm.

The introduction of useful germplasm is possible only through the correct identification of species. Exact identification of harmful pests and their control help in the protection of many medicinal, economic plants and various animals. Many plants of economic importance can be identified for breeding to increase their yields and production of **disease resistant** varieties is possible only through the knowledge of systematics.

7. ROLE OF SYSTEMATICS IN FINDING NEW SPECIES: A long-standing role for systematists is that of going into the field and collecting samples of organisms, then comparing them with known specimens in order to determine whether something significantly different has been found- a new species.

8. ROLE OF SYSTEMATICS IN BIODIVERSITY CONSERVATION: With increasing pressures from a growing world **population** and resulting pressure on **biotic resources**, we now and in the future have to make difficult decisions about what parts of the Earth will be maintained in a “**natural**” state in order to conserve the biodiversity present there. How do we decide, given limited resources, which to protect? If we decide that we want to maximize **biodiversity**, then the phylogenetic patterns produced by systematists give us a way to prioritize areas based upon the diversity they contain.

9. ROLE OF SYSTEMATICS IN DOCUMENTATION: Another crucial role for systematists is that of identification specialists. They are in a unique position to provide this service, with experience and the **necessary tools**. The importance of correct identification cannot be overstated -- when a life, for instance, hangs in the balance depending on whether the plant or mushroom that has been ingested is poisonous or not, this service is critical.

Other types of biological research are essentially valueless if their subjects are misidentified since closely related **taxa** can have very different properties and generalizations must be made carefully. Hence, **documentation** is important so that subsequent investigators can confirm identifications. The only lasting way to document identity is to deposit a **voucher specimen** in an appropriate collection. Studies that do not utilize this service will have less value in the long term because of the impossibility of verifying identification.

10. ROLE OF SYSTEMATICS IN HORTICULTURE AND FLORICULTURE: Knowledge of systematics is essential for horticulture and floriculture also. Several ornamentals

have been introduced due to proper identification and nomenclature. Its knowledge is also required to study the natural resources of areas to know the land potential.

Systematics has its relevance in fisheries and for the study of economically and medicinally important plants. All pharmaceutical studies are based on the work of taxonomists. Taxonomists are being employed at various positions in the museum, colleges, institutions, research institutions and in various public and private organizations. So, it has wide scope and applications as a profession also.

5.5 Summary:-

1. The term taxonomy was first coined by A. P. de Candolle in 1813.
2. Taxonomy is a branch of science which deals identification, nomenclature, and classification.
3. Taxonomy may be categories as alpha, beta and gamma taxonomy.
4. Term systematics was first described by Carolus Linnaeus.
5. Systema Naturae is a famous book of Carolus Linnaeus.
6. Systematics is a scientific study of taxonomy.
7. New systematics or biosystematics or neosystematics was proposed by J. Huxley in 1940.
8. Systematics and taxonomy both are complementary to each other.
9. Aristotle is known as the father of Zoology and father of biological taxonomy due to his tremendous contribution in Zoology.
10. Binomial nomenclature, a system of classification is given by Carolus Linnaeus.
11. Systematics is a modified form of classical systematics or old systematics.
12. Species is the basic unit of taxonomy as well as evolution in both plants and animals.
13. All the organisms should be classified i.e. divided into groups and subgroups.
14. Classification is as old as the power of speech.
15. The method of rearranging and regrouping of organism into various divisions is called classification.

16. Identification means the determination of the correct place of an organism in a previously established plan of classification.
17. Homology establishment helps much in finding the exact position of an organism.
18. The kingdom is the highest category of taxonomic studies.
19. Systematics provides useful information about the evolution, adaptations, and diversity of organisms.
20. Systematics is concerned with the evolutionary relationships of organisms.

5.6 Self Assessment Questions :-

1- The term taxonomy was first coined by

- a- A. P. de Candolle
- b- Carolus Linnaeus
- c- John Ray
- d- Aristotle

Ans- a

2- Taxonomy that is concerned with the arrangement of species according to the law of systematics is called

- a- α (ALPHA) TAXONOMY
- b- β (BETA) TAXONOMY
- c- γ (GAMA) TAXONOMY
- d- all of the above

ans- b

3- refers to the study of the relationships between these living things as they evolve

- a- systematic
- b- taxonomy
- c- classification

d- nomenclature

ans- a

4- The taxon of birds is **Class** and their category is **Aves**. (**True or False**)

Ans- False

5.7 Terminal Questions:-

1. Write the definition of taxonomy?
2. Define species?
3. What is the meaning of Taxa?
4. Describe Taxonomy and its relationship with Systematic in detail?
5. Define Systematic. What are the importance and applications of Systematics?
6. Write a detail account on taxonomic hierarchy?

5.8 References:-

1. Simpson, G. G. 1961. Principles of Taxonomy, Columbia University Press, New York.
2. Griffiths, Anthony J. F.; Miller, Jeffrey H.; Suzuki, David T.; Lewontin, Richard C.; Gelbart, eds. (2000). Selection". An Introduction to Genetic Analysis (7th ed.). New York: W. H. Freeman.
3. Darwin Charles (1859). On the Origin of Species (1st ed.). London: John Murray.

5.9 Suggested Readings:-

1. Simpson, G. G. 1961. Principles of Taxonomy, Columbia University Press,

- New York.
2. Dobzhanski, T. H. 1951. Genetics and Origin of Species, 3rd edn. Columbia Univ. Press, New York.
 3. Arora, B. B. and Sabharwal A. K. 2000. ABC Biology. Modern Publishers, New delhi.

UNIT 6: ZOOLOGICAL NOMENCLATURE

Contents

- 6.1 Objectives
- 6.2 Introduction
- 6.3 International Code of Zoological Nomenclature
- 6.4 Binomial and Trinomial Components of Classification
 - 6.4.1 Binomial Nomenclature
 - 6.4.2 Trinomial Nomenclature
- 6.5 Summary
- 6.6 Self Assessment Questions

6.1 Objectives:-

In the last chapter, you have learned about the **taxonomy** and **systematics** and its relationships between them. About **2 million** types of animals and about **1 million** types of plants have already been described and named and many new forms are discovered every year. Animals and plants vary greatly in their forms, structure and mode of life. To identify an organism of known characters from the vast number of organisms is simply **impossible**. The number and diversity of living organisms are so enormous that it is difficult to study without classifying them into certain **groups** or **categories**. This classification allows us to identify and recognize organisms.

In this chapter, you would learn about the **rules** and **regulation** of nomenclature of animals as well as plants. You will have also learned the **common name** or **vernacular names** and **scientific names** and difference between them. It would also emphasize the importance of scientific names.

6.2 Introduction:-

Nomenclature of any individual either that is animal or plant play a very significant role in the **taxonomy**. There are more than **two million species** of animals and about **one million** species of plant, while several more to be discovered. These species may be identified and classified with the help of nomenclature either it is **zoological** or **botanical nomenclature**. Without nomenclature, it would be very difficult to differentiate them. In this chapter, you will read about the **law of nomenclature**, **binomial and trinomial system of classification**. This chapter will deal with how living beings are named, grouped and classified.

TYPES OF BIOLOGICAL CLASSIFICATION

As more and more data regarding life forms have come to light, classification becomes more and more refined. To understand the **diversity of living beings**, a system of classification is required. Classification allows understanding diversity better. The art of identifying distinction among organisms and placing them into groups that reflect their most significant feature and relationship, morphological and evolutionary and others is called **biological classification**. Organisms have been classified from a different point of view at different times. Three main schemes of classification emerged one after one.

PRACTICAL CLASSIFICATION

BASIS: Organisms were originally classified on the basis of their **utility** to man. They were grouped as the **useful** and **harmful** forms and as **edible** and **inedible** ones. This grouping, though rough, was of immediate and practical use to man. The criteria used in practical classification are arbitrary. For example, animals were recognized as food animals, fur animals, pets, beasts and burden etc., while, plants as crop yielding timber yielding, fiber plants etc. This system of classification based on their value to man, irrespective of their structural similarities is called **practical classification**.

DRAWBACK: It gave **heterogeneous** groups of unrelated organisms. For example, edible animals included shrimps, fishes; chickens and goat though these animals radically differ from one another.

ARTIFICIAL CLASSIFICATION:-

BASIS: The system of classification based on one or few **randomly** selected characters for grouping the organisms is known as **artificial classification**. Such system of classification was in use during the early periods. It is based on few a few **superficial resemblance** rather than **natural** or **evolutionary relationships**. It, therefore, gives only a little information about the groups.

The **Greek naturalists** classified animals according to similarities in their **habitat** and **habit**. The animals were grouped as **aquatic, terrestrial and aerial dwellers** (according to habitat), as **carnivorous** and **herbivorous**. (According to mode of feeding) and as **oviparous** (egg-laying) and **viviparous** (giving birth to young ones) according to the mode of breeding. The plants were classified as **herbs, shrubs and trees**. Aristotle (384 – 322 B. C.) was the first one to adopt this system of classification.

DRAWBACKS: The artificial system of classification is, no doubt easier, but has serious drawbacks:

1. As the characters are randomly picked up, they do not reflect any **phylogenetic relationship**.
2. Unrelated organisms are put together, forming **heterogeneous** groups. For example, birds, bats, and insects are combined together as flying animals. Cacti, Euphorbias, and halophytes are clubbed together as **succulent plants**.
3. Related organisms are placed in different groups. For example, Whales an aquatic and rats a **terrestrial animal**.
4. The characters may change with the change in environment. For example, change of habit in radish (annual and biennial habits).
5. It does not show any **evolutionary relationships**.

NATURAL CLASSIFICATION:-

BASIS: In this system of classification all the important characteristics of the organism that provide information regarding their **natural relationship** are taken into consideration. It, therefore, gives more or detailed information about the groups. The system employs those characters which are relatively constant. The English naturalist, **John Ray (1627-1705)** was the first **systematic** to form the structural similarities as the basis of classification. He used constant and well-defined characters in his classification, thus making systematic a scientific discipline. He published an accurate description of over **18000 plants** in his book *Historia Generalis Plantarum*.

ADVANTAGES: Natural classification is the most logical system of classification and has been **adopted** by all the **biologists**. It is better than artificial classification in many respects:

1. It avoids the **heterogeneous** grouping of unrelated organisms.
2. It indicates **natural relationships** among organisms.
3. It shows **evolutionary or phylogenetic relationships**.

Difference between artificial and natural classification

Artificial Classification	Natural Classification
<ol style="list-style-type: none">1. It is based on one or a few characters.2. It gives meager information about the groups.3. It uses superficial characters such as habitat and habits.4. It gives heterogeneous groups of unrelated organisms.5. It does not reflect phylogenetic relationship of organisms.	<p>It is based on a number of characters.</p> <p>It gives detailed information about the groups.</p> <p>It uses characters pertaining to morphology, anatomy, cytology, ontology, phylogeny, physiology and biochemistry.</p> <p>It gives homogenous groups of related organisms.</p> <p>It depicts the phylogenetic relationship of organisms.</p>

Natural classification considers more evidence than **artificial classification**, including internal as well as external features, similarities of embryo, morphology, anatomy, physiology, biochemistry, cell structure and behavior. Classification is used today are natural and phylogenetic.

PHYLOGENETIC SYSTEM OF CLASSIFICATION:-

Classification based on **evolutionary relationships** of an organism is called **phylogenetic system of classification**. It reflects the true relationships among the organisms. The phylogenetic system was first proposed by **Engler and Prant (1887-1899)**. The concept of fixity of species, prevalent before **Darwin**, changes to a dynamic or over changing one i.e. Species are never **static** and **undergo changes**. Its major source is **fossil record**. This is never complete due to difficulty in formation, discovery, and study of fossils. As and when new fossils are discovered, newer relationships are observed and consequently the **phylogenetic system** is updated.

Thus, like the species, classification is also **dynamic**. In addition to morphological characters, the evolutionary development of groups of organisms, from its origin to the present state, forms the **basis** of classification. From the evolutionary point of view, the presence of fundamental structural similarities in different species is explained on the basis that all the species were derived from a **common ancestor** were related to one another. Thus, the establishment of the theory of evolution puts systematic on new lines.

CLASSIFICATION GUIDELINE:-

To determine the position of an organism in a natural or phylogenetic system of classification, a modern taxonomist uses many principles and criteria. He studies the **similarities** and **differences** in organisms by examining many characteristics. These includes the knowledge of morphology (external features), anatomy (internal structure), cytology (cell structure), physiology (life processes), ontogeny (development of an individual organism), phylogeny (evolutionary history), ethology (behavior), reproductive behavior and biochemistry etc. The main difficulty in the classification is that of sorting the cases of **analogy** or **convergent evolution** i.e. the development of similar adaptations by organisms of different ancestries.

Analogous organs have the same function and are superficially alike but are quite different in fundamental structure and embryonic origin. For example, **insect** and **bird wings**. Both these organs are used for flying in the air, but they are very **different in their structure**. An insect wing is an extension of the integument, whereas, a bird wing is formed of limb bones covered with flesh, skin, and feathers. Another example of analogous organs is pectoral fin of shark and flipper of dolphin.

6.3 International Code of Zoological Nomenclature:-

One of the primary responsibilities of **systematic biology** is the development of our biological **nomenclature** and **classifications**. Nomenclature is not an end to systematics and taxonomy but is a necessity in organizing information about **biodiversity**. Nomenclature functions to provide labels (names) for all **taxa** at all levels in the **hierarchy** of life.

Zoological nomenclature is a language that we use to communicate ideas and information about the **diversity of life**. It is an information retrieval system conveying information about diversity and relationships. In **1898 International Congress of Zoology** organized an **International Commission of Zoological Nomenclature** and suggests some rules and regulations for nomenclature.

These rules were revised in **1948** and **1950** in **International Congress of Zoology** and **International Congress of Botany** respectively. **12th International Congress on Nomenclature in 1975** laid down some general principals in the form of **International Code of Botanical Nomenclature (ICBN)** and **International Code of Zoological Nomenclature (ICZN)** which are as followings:

1. **Binomial and trinomial system of nomenclature** should be adopted.
2. Name of the **genus** should start with **capital letter** followed by **species** with **small letters**.
3. The name of the genus should be a single **word** and difficult, long **should be avoided**.
4. Genus name should be read as a **generic name** followed species as a **specific name**.
5. The scientific name must be derived from **Latin language** only.
6. The scientific name must be always written in **italics or underlined only**.
7. The plants and animals should have **independent** and **different** names.
8. In scientific name first word will be represented by **genus** and second and third (if present) will be represented as **species** and **subspecies** respectively.
9. Within animal kingdom no two genera should have the same name and within the genus, no two species should have the same name.

10. A scientific name must have its **original spellings** and errors must be corrected.
11. The name of author should be written in **Roman script** after the species without comma between them.
12. The scientific name should be too **easy to pronounce**.
13. The scientific name should not have less than **three** and not more than **twelve letters**.
14. The scientific name of plant or animal should be **self-explanatory** in its characters.
15. Every species should have a **generic name**.
16. Other components of taxonomy like phylum, class, order should also start with a **capital letter**.
17. Species should not be identified with its **size**.
18. The name of family should start with capital letter and should be suffix **–IDEA** and subfamily by **INAE**.
19. The generic or specific name first published is the only one recognized. All duplicate names are **synonyms**.
20. The formations of family and subfamily names follow rules which are different in the **Zoological and Botanical Codes**.
21. A name may be based on any part of an animal or a plant, or on any stage of an **Organism's life history**.
22. In case of discovery of different name of same genus and species by different Scientists, the name **first published should be accepted**.

Thus, presently there are four different codes of nomenclature used today.

1. International Code of Zoological Nomenclature (ICZN). 1999.
2. International Code of Botanical Nomenclature (ICBN) 1994.

3. International Code of Nomenclature of Bacteria (ICNB). 1976.
4. International Code of Nomenclature for Cultivated Plants (ICNCP). 1980.

Some General Objectives of Scientific Nomenclature:-

UNIQUENESS: The name of a particular organism gives one immediate access to all of the known information about the particular **taxon**. Every name must be unique because it is the key to the entire literature relating to the species or higher taxon in question. If several names have been given to the same taxon, there must be a clear-cut method whereby it can be determined which of the names has validity.

UNIVERSALITY: Scientific communication would be made very difficult if we had only **vernacular names** for taxa in innumerable languages in order to communicate with each other. To avoid this we have adopted an international agreement for a single language (**Latin**) and a single set of names for biological diversity to be used on a worldwide basis.

STABILITY: As recognition symbols of diversity, names of organisms would lose much of their usefulness if they were changed frequently and arbitrarily.

6.4 Binomial and Trinomial Components of Classification:-

In the system of classification, every individual has identification, for which **nomenclature** is a must. Without nomenclature, it will be very difficult to differentiate it with other individuals. Generally, individuals are classified into **two categories** as **Common or Vernacular Name** and **Scientific Name**.

1. COMMON OR VERNACULAR NAME: A **common name** of a taxon or organism also known as a **vernacular name** in English, is a **name** that is based on the normal or local language in different regions as well as the country. In other words, the locally used name are called **vernacular names**. This kind of **name** is often contrasted with the scientific **name**. Such names are based on the same peculiarity of the organism, e.g. Kanteli (a plant having spines).

For Example, the simple domestic bird is commonly known as **Gauraiya** in India and Pakistan, while in England **House sparrow**, In German **Hausperling**, in Holand **Musch**, and in Japan it is known as **Suzune**.

ADVANTAGES

1. Easy for the local people to follow
2. Easy to learn, speak and write
3. Usually short.

DRAWBACKS

1. The same animal or plant is known by a different common name in **different countries** where different languages are used.
2. A singular vernacular name is often used for **several species**.
3. The common names may be **misleading**. For example, Jellyfish, Silverfish, Starfish are the names of some animals, none of which is a really a fish (except dogfish).
4. The common names lack a **scientific basis** as they do not convey any taxonomic relationship with other organisms.
5. Only those organisms which are **beneficial** or **harmful** to man have been provided these names. Insignificant ones were left out.
6. New common names cannot be introduced nor can the old ones be **changed at will**.

2. SCIENTIFIC NAMES: In view of the shortcomings of common names, another system was called **scientific** or **technical system** has been devised to name the organisms. According to this system, scientific names of organisms are based on agreed **principals** and **criteria** which are acceptable all over the world. The scientific names ensure that only one name is given to an organism and description of the organism should help the other people to arrive at the same name in any part of the world.

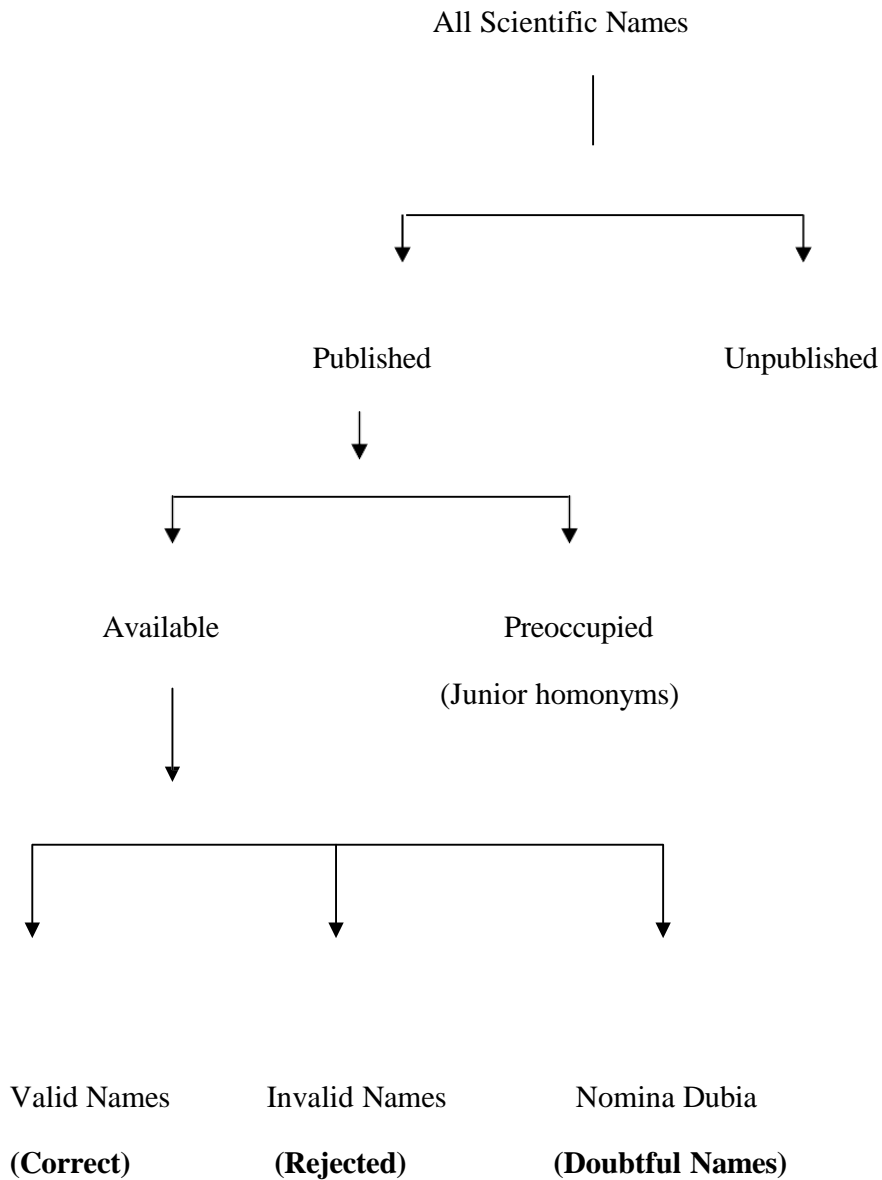
ADVANTAGES

1. They help in **classification**.
2. A newly discovered organism can be **easily described** and **named**.
3. Each kind of organism is given a **single scientific name**.
4. This scientific name eliminates the confusion of **multiple naming**.
5. An incorrect name can be easily set right.
6. They are **universally accepted**.
7. They indicate the relationship of a species with others placed in the **same genus**.
8. They indicate some important **characteristics of the organisms**.
9. The scientific names are often derived from **Latin/ Greek** which are **dead languages**.
10. Hence, there is less possibility of change in the meaning of their words.

RULES FOR SCIENTIFIC NAMES

1. Each organism to be given a **single scientific name**. However, species having subspecies, varieties or races are given a **trinomial name**.
2. The scientific name should be printed in **italics**. (If handwritten or typed, the name is **underlined**).
3. The first (generic) name should always begin with **capital letter**. It is often abbreviated by using only its first initial. For example; *C. familiaris* for *Canis familiaris* (dog).
4. The first letter in a species should always begin with a **small letter**.
5. The names of the division above the genus are not printed in italics. However, they are started with a **capital letter**. For example: The order and the class of humans are written as **Primates** and **Mammalia** respectively.
6. The generic name appears **only once** whereas the specific name may appear **many times**, but each time with a separate genus. For example: *Mangifera indica* and *Tamarindus indicus* are the names of mango and tamarind respectively.

7. Two species belonging to the same genus cannot have the **same specific name**.



Categories of Scientific Names (Modified from Blackwelder, 1967)

SYSTEM OF SCIENTIFIC NAMES:-

Following have been practices of providing scientific names to organisms:

POLYNOMIAL NOMENCLATURE

Polynomial names were in use much before **1750**. In this system, the scientists used to add a series of **descriptive words**. Names become **very lengthy** and **difficult** to remember. For example, the plant **Caryophyllum** has been given the name **Caryophyllum sexatilis folis gramineus umbellatis corymbis**, meaning caryophyllum growing on rocks having grasslike leaves and **umbellate – corymb** arrangement of flowers. This type of naming was called **polynomial nomenclature**.

6.4.1 Binomial Nomenclature:-

The **polynomial system** was quite a trouble or difficult. It also changed from scientist to scientist. Consequently, in **1758**, the system of writing scientific name of plants and animals adopted by **C. Linnaeus** (a Swedish naturalist) is called **Binomial Nomenclature**. This system proved better and ultimately became a common and established.

According to this system, the **scientific name** comprises of two words **genus** (generic name) and **species** (specific name). The generic name is **common** for all the species in a genus, while the specific name is commonly based on some **special** or **definite characters**. Generic names are used to written in **Latin** or **Greek** words usually begin with a **capital letter**, while specific names are always being written in **small letters**. Both generic, as well as specific names, should be written in **italics** or **underlined**.

Sometimes, the scientific name is also written in the **honor** of scientist is followed by a specific name. If the person honored is a man the specific name ends in **“i”**. For example, the earthworm, *Lumbricus friendi* is named after Rev. **H. Friend**. If the person honored is a woman, the specific name ends **“ae”**. Sometimes, the specific name indicates **a locality** as **indica** for India or color as **niger** for black.

All generic and specific epithets have authors, the name(s) of the person(s) who first officially described them in a publication. You will often see scientific names with an author's name following it. This is **often confusing** to non-taxonomists but is really important because it is very

useful in tracing the history of applications of names through time. Scientific names with very similar spellings can usually be distinguished from one another when an author's names are included.

For Example: Indian bird **Gauraiya**, scientific name is *Passer domesticus* and **Dog** is *Canis familiaris* and **Human beings** as *Homo sapiens* Linnaeus.

6.4.2 Trinomial Nomenclature:-

Whenever the system of nomenclature is usually adopted by **three words** called **trinomial nomenclature**. There are some species which contain **subspecies**. Subspecies is generally followed by species and also written in **Latin** word always. These subspecies usually found in the **different region** of the world containing different characteristics.

Thus, For example, the common specific name of **crow** is *Corvus splendens*, but its three species are generally found in India, Burma, and Sri Lanka. **In India**, it is named as *Corvus splendens splendens*, in **Burma** *Corvus splendens insolens*, and in **Sri Lanka**, it is the trinomial nomenclature indicates the **generic, specific and sub-specific name** called *Corvus splendens protegatus*.

Sometimes, the **name of a scientist** is followed by trinomial nomenclature as *Columba livia intermedia* Strickland (Prof. Strickland), *Panthera leo persica* Linn. etc. The scientific names provided are often descriptive and also indicate some important characteristics of the organisms. **For animals**, scientific names are governed by the International Code of Zoological Nomenclature. Only one rank is allowed below the rank of species: subspecies. However, Advantages of using scientific names for an organism are as follows:

- 1.0 The scientific name remains the **same worldwide**, hence is easily recognizable.
- 2.0 The possibility of confusion due to multiple names were given to the same organism in different parts of the world is **eliminated by scientifically** naming the organism.
- 3.0 A relationship between different species of organisms in a particular genus can be **deduced** by scientific names.
- 4.0 It also helps in recognizing or identifying any **new organisms** discovered.
- 5.0 Any incorrect name to a particular organism can be **corrected**.

6.5 Summary

- To study understand **diversity of living beings**, a system of classification is required.
- In ancient time organisms were originally classified on the basis of their utility of man.
- Artificial classification is based on the some randomly selected characters.
- Practical classification is based on their value to man, irrespective of their structural similarities is called **practical classification**.
- John Ray published accurate description of over **18000 plants** in his book *Historia Generalis Plantarum*.
- Natural classification shows **evolutionary or phylogenetic relationships among organisms**.
- Phylogenetic system was first proposed by **Engler and Prant (1887-1899)**.
- **Zoological nomenclature** is a language that we use to communicate ideas and information about the **diversity of life**.
- In **1898 International Congress of Zoology** organized an **International Commission of Zoological Nomenclature** and suggests some rules and regulations for nomenclature.
- **12th International Congress on Nomenclature in 1975** laid down some general principals in the form of **International Code of Botanical Nomenclature (ICBN) and International Code of Zoological Nomenclature (ICZN)**.
- **Binomial and trinomial system of nomenclature** should be adopted.
- International Code of Zoological Nomenclature (ICZN). 1999.
- International Code of Botanical Nomenclature (ICBN) 1994.
- International Code of Nomenclature of Bacteria (ICNB). 1976.
- International Code of Nomenclature for Cultivated Plants (ICNCP). 1980.

- A **common name** of a taxon or organism also known as a **vernacular name** in English, is a **name** that is based on the normal or local language in different regions as well as country.
- Each organism to be given a **single scientific name**.
- Consequently in **1758**, the system of writing scientific name of plants and animals adopted by **C. Linnaeus** (a Swedish naturalist) is called **Binomial Nomenclature**.
- The system of nomenclature is usually adopted by **three words** called **trinomial nomenclature**.
- The trinomial nomenclature indicate **generic, specific and sub specific name**.

6.5 Self assessment Questions:-

- 1- The phylogenetic system was first proposed by
 - a- C. Linnaeus
 - b- Engler and Prant
 - c- Darwin
 - d- Strickland

ans- b
- 2- The main difficulty in the classification is that of sorting the cases of
 - A-Analogy
 - b- Homology
 - c- Physiology
 - d- Ontogeny

ans- a
- 3- Which of following is not a principle of International Code of Zoological Nomenclature (ICZN):
 - a- Binomial and trinomial system of nomenclature should be adopted

- b- Name of the genus should start with capital letter followed by species with small letters
- c- The scientific name must be always written in italics or underlined only.
- d- The scientific name must be derived from Italian language only

Ans- d

- 4- Classification based on of an organism is called phylogenetic system of classification

Ans- evolutionary relationships

- 5- The trinomial nomenclature indicates the generic, specific and name

Ans- sub specific

6.7 Terminal Questions:-

- 1- What is the meaning of phylogenetic system of classification?
- 2- What is the difference between artificial and natural classification?
- 3- Why it is necessary to provide a scientific name to an animal?
- 4- How living beings are named, grouped and classified scientifically?
- 5- Describe various types of Biological classifications?
- 6- Write a detail account on Binomial and Trinomial components of nomenclature?
- 7- Write a essay on ICZN?

6.8 References:-

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6.9 Suggested Readings:-

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UNIT 7: KINDS OF TAXONOMIC CHARACTERS AND CLASSIFICATION

Contents

- 7.1 Objectives
- 7.2 Introduction
- 7.3 Taxonomic characters
 - 7.3.1 Morphology characters
 - 7.3.2 Embryological characters
 - 7.3.3 Biochemical and numerical character
 - 7.3.4 Cytogenetical characters
 - 7.3.5 Geographical characters
 - 7.3.6 Ecological characters
 - 7.3.7 Ethological characters
- 7.4 Classification
 - 7.4.1 Components of classification
 - 7.4.2 Linnaean hierarchy
- 7.5 Summary
- 7.6 Self-assessment questions
- 7.7 References
- 7.8 Suggested readings
- 7.9 Terminal questions

7.1 Objectives:-

In last chapter, you have learned about the **taxonomy and systematics** and its relationships between them. In this chapter, you would learn about the **taxonomic characters** such as morphological, embryological, cytogenetical, biochemical and numerical etc. In this chapter you would be able to answer about the description on classification and its components, binomial, trinomial system of classification, artificial, natural, phylogenetic, modern system of classification and of Linnean hierarchy. The description of the **animal kingdom** has also emphasized. You will also read about taxa, taxon, and ranks.

7.2 Introduction:-

All living organisms **vary** considerably in their forms, structure, and mode of life. To select **an organism** of unknown characters from a **large number of organisms** is almost impossible. They should be classified into **groups** and **subgroups** on the basis of various taxonomic characters described as below. Without a particular method of classification of living beings, it would be very difficult to arrange them. In this sequence, **Linnean hierarchy**, which is a system of classification from higher to lower category, play a very important role in the taxonomy.

7.3 Taxonomic Characters:-

A taxonomic character is any **attribute** of a member of a **taxon** by which it differs or may differ from a member of a different taxon. A characteristic by which members of two taxa agree but differ from members of a third taxon is a **taxonomic character**. However, it is a very important to **taxonomist** or **systematist** to follow the principles and criteria while classifying an organism. While examining the various taxonomic characters, he should keep in mind the various **similarities** and **differences** in the following field also:

Morphology (General external and special characters)

Anatomy (Internal characters)

Cytology (Cell structure)

Physiology (Life processes)

Ontogeny (Development)

Reproduction (Fertilization)

Behavior (Courtship and other ethological isolating mechanisms)

Biochemistry (Physiological processes)

Molecular (Immunology, DNA, RNA sequences, and hybridization, restriction endonuclease analyses, amino acid sequence of proteins)

Ecological (Habitat and hosts, food, seasonal variations, parasites, host reactions)

Geographical (Biogeographic distribution, sympatric – allopatric populations)

7.3.1 Morphological Characters:

Morphology taxonomic character is generally represented by **external features** and some special character as follows:

A. GENERAL EXTERNAL MORPHOLOGY: It includes a **physical appearance** of an animal or its any body parts. For example - plumage of a bird, pelage of mammals, scale counts of fishes and reptiles.

B. SPECIAL STRUCTURES: It is represented by a **special part** of the body, for example – Genitalia. Differences in the genetalic structure have been used to delimit species. It is very effective in insects where the **lock and key** relationship exists between male and female copulatory organs.

7.3.3 Embryological Characters:

Both the soft as well as hard parts of practically all groups of **higher animals** have been used as taxonomic characters. Various **immature** or larval stages, the embryology and sometimes even the eggs may provide taxonomic information *e.g.* the various **sibling species** of the *Anopheles maculipennis* complex were discovered owing to differences in egg structure. The classification of **white flies** is based primarily on the pupae. Comparative studies of embryological characters like cleavage pattern, **blastulation** and **gastrulation** are also useful in certain phyla.

7.3.4 Biochemical and Numerical Characters:

This group of character is **hard** to define. All structures are the products of physiological processes and are thus physiological characters. By physiological characters, one generally means growth constants, temperature tolerances and the various processes studied by a comparative physiologist. These characters cannot be studied in **preserved material**.

The **proteins** of one organism will react more strongly with **antibodies** to the proteins of a closely related organism than to those of one more distantly related. **Sibley** analyzed the egg-white proteins of more than **100 species** of **birds** and was able to establish the relationship among them.

7.3.5 Cytogenetically Characters:

Number of taxonomy is the description of **chromosome** structure, size, shape and number etc. Chromosomes are particularly useful on two different levels. **On the one hand**, they aid in the comparison of closely related species, including **sibling species**. These are often far more different chromosomally than in their external morphology. **On the other hand**, chromosomal patterns are of extreme importance in establishing **phyletic lines**.

7.3.6 Geographical Characters:-

Geographical characters are among the most useful tools for clarifying a confused taxonomic picture and for **testing the taxonomic hypothesis**. The taxonomist is primarily interested in two kinds of geographical characters:

- (1) **General biogeographic patterns** - which are especially useful in the arrangement of higher taxa.
- (2) **The allopatric-sympatric relationship** - which is most helpful in determining whether or not two populations are co-specific or non-specific.

7.3.7 Ecological Characters:

Every species has its own **niche** in nature, differing from its nearest relatives in food preference, breeding season and tolerance to various physical factors, resistance to predators, competitors

and pathogens and in other ecological factors. **For example-** the larvae of both *Drosophila mulleri* and *aldriachi* live simultaneously in the decaying pulp of the fruits of the cactus (*Opuntia lindheimeri*). The two species are markedly specialized in their preference for certain **yeast and bacteria**.

7.3.8 Ethological Characters:

Behavior is one of the most important source of taxonomic characters. They are clearly superior to morphological characters in the study of closely related species.

7.4 Classification:-

Modern science has so far described over **16 lakh species** of living species of living organisms, that span an enormous range in size, from the tiny **viruses** such as those that gives us cold to the whose **whales** and giant **banyan trees**. Our ignorance of these small **creatures** is profound so that most of them are as yet unknown to science. The total number of species exist on earth is believed to lie somewhere between **80 to 120 lakh**.

Hence, the number of living beings including plants and animals are so numerous that is impossible to arrange them in a **systematic order**. All living organisms considerably vary in form, structure, and mode of life. To select an organism of unknown character from a large number of organisms is almost impossible. Therefore, they should be classified into **groups** and **subgroups**. Arranging the organisms in a definite plan, make the study of plants and animals easy.

To arrange things or ideas is the function of highly practical activity called **classification**. However, the **basic or first purpose** of zoological classification is to enable us to keep track of more and more animals represented by many millions of individuals by grouping them into various categories. **The second purpose** of biological classification, one of the more scientific natures, is the discovery of new knowledge. Classification is always the result of **observation of attributes**.

There is **three major system of classification** are as following:

- 1.0 Artificial system of classification
- 2.0 Natural system of classification

3.0 Phylogenetic system of classification

1. ARTIFICIAL SYSTEM OF CLASSIFICATION:

First of all this system was adopted by **Pliny the Elder** (Rome) in the first century **23 -79 A.D.** He described this in his book **Naturalis Historia**. According to this system of classification, The living beings was classified on the basis of some **superficial characters**. The characters were arbitrarily selected for the classification. Pliny classified animals into two groups:

1.0 -Animal that can fly.

2.0- Animals that cannot fly.

2. NATURAL SYSTEM OF CLASSIFICATION:

This system of classification was proposed by **Gorge Bentham** (1800-1844) and **Joseph Dalton Hooker** (1817 – 1911). In this natural system, animals are grouped according to their **basic similarities** into as many groups and sub-groups as their resemblance and differences require. The system is not only based on the **reproductive characters** but structural relationships are also taken into consideration.

This system of classification helps in detecting relationship **affinities** of an organism with another organism also. The system is said to be **better** than the artificial system of classification because it avoids grouping of **heterogeneous** and unrelated organisms.

3. PHYLOGENETIC SYSTEM OF CLASSIFICATION:

This system of classification was proposed by **A. Engler Karl, A. E. Prantl** and **John Hutchinson**. **Engler** and **Prantl** (1884-1930) describe this system in detail in 23 volumes in his book entitled “**Die Naturlichen Familien**”. This system is mainly based on the **evolutionary** and **genetic relationships** of the plants. It enables us to find out the ancestors or derivatives of any taxon.

4. MODERN SYSTEM OF CLASSIFICATION

Well before Linnaeus, plants and animals were considered separate kingdoms. Linnaeus used this as the **top rank**, dividing the physical world into the plant, animal and mineral kingdoms. As advances in **microscopy** made classification of microorganisms possible, the number of kingdoms increased, five and six-kingdom systems being the most common.

Domains are a relatively new grouping. The **three domains** system was first proposed in 1990, but not generally accepted until later. One main characteristic of the three-domain method is the separation of **Archea** and **Bacteria**, previously grouped into the single kingdom Bacteria (a kingdom also sometimes called **Monera**). Consequently, the three domains of life are conceptualized as Archaea, Bacteria, and Eukaryota (comprising the nuclei-bearing eukaryotes). **A small minority of scientists adds Archaea as a sixth kingdom, but do not accept the domain method.**

Thomas Cavalier-Smith, who has published extensively on the classification of **Protists**, has recently proposed that the **Neomura**, the clade that groups together the **Archea** and **Eukarya**, would have evolved from **Bacteria**, more precisely from **Actinobacteria**. His classification of 2004 treats the **Archaeobacteria** as part of a subkingdom of the Kingdom Bacteria, i.e. he **rejects the three-domain system entirely**. **Stefan Luketa** in 2012 proposed a five "**dominion**" system, adding **Prionobiota** (acellular and without nucleic acid) and **Virusobiota** (acellular but with nucleic acid) to the traditional three domains.

7.4.1 Components of Classification :-

Biological classification is a critical component of the taxonomic process. As a result, it informs the user as to what the relatives of the taxon are hypothesized to be. The "**definition**" of a taxon is encapsulated by its description and/or its diagnosis. There are no set rules governing the definition of taxa, but the naming and publication of new taxa are governed by sets of rules. In **Zoology**, the nomenclature for the more commonly used ranks (super family to subspecies), is regulated by the **International Code of Zoological Nomenclature (ICZN Code)**. In the fields of **Botany**, Phycology, and Mycology, the naming of taxa is governed by the **International Code of Nomenclature for Algae, Fungi, and Plants (ICN)**.

A **taxon** may be defined as a unit of classification of organisms which can be recognized to the definite category at any level of classification e.g. fishes, insects, algae, fungi, ferns grasses are taxa. So the taxa are groups of organisms, which can be recognized as a formal unit at any level of **hierarchic classification**. Taxon word relates to a taxonomic group of any rank. However, the initial description of a taxon involves five main requirements:

1. The taxon must be given a name based on the **26 letters** of the Latin alphabet (a binomial for new species, or uninomial for other ranks).
2. The name must be **unique** (i.e. not a homonym).
3. The description must be based on at least **one name-bearing type specimen**.

4. It should include statements about appropriate **attributes** either to describe (define) the taxon, and/or to differentiate it from other taxa.

5. Both codes **deliberately separate** defining the content of a taxon (its circumscription) from defining its name.

These first four requirements must be published in a work that is obtainable in numerous identical copies, as a **permanent scientific record**.

Biological classification uses **taxonomic ranks** called **components of classification**, including among others (in order from most inclusive to least inclusive):

Domain

Kingdom

Phylum

Class

Order

Family

Genus and

Species

In the **modern scientific** age, the available levels also known as **basic levels** or components of classification are rarely enough. Therefore, some additional components or levels of classification are also added to **basic components**. These are as following:

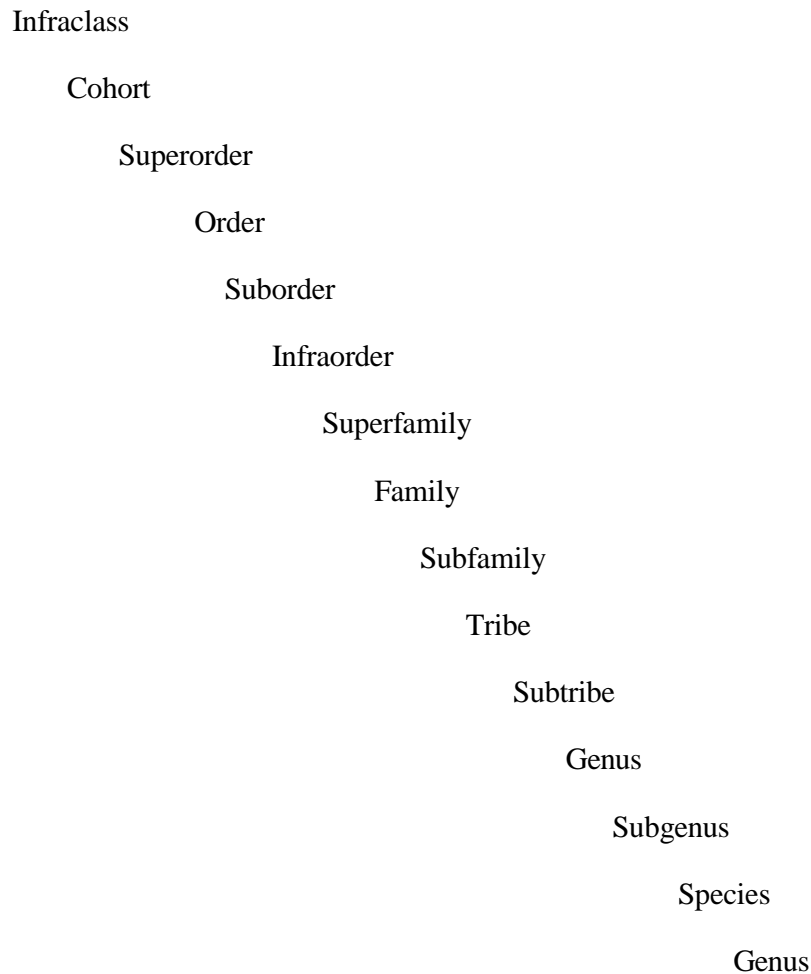
Kingdom

Phylum

Subphylum

Superclass

Class



7.4.2 Linnaean Hierarchy

The hierarchy may be defined as a system of arrangements of taxonomic categories for classification in a logical sequence. Generally, hierarchy means “**a series of a succession of different rank**”. Firstly it is established by **C. Linnaeus** in the animal kingdom, therefore, it is known as a **Linnean hierarchy**. The most peculiar character of the Linnean hierarchy is the **descending sequence of class, genus, species** and variety taxonomic categories. But, subsequently, the Linnaean hierarchy is added by **Phylum** (between kingdom and class) and **Family** (between order and genus).

The Linnaean system of classification consists of a hierarchy of graded taxonomic (named) ranks that are called as **taxa**. Any given **taxon** (singular) may contain several lower taxa, which can be usually distinguished based on certain **common characteristics**. Such lower ranks may, in turn, be divided into a succession of progressively smaller ranks. The lower the rank of a group, the

more similar are the organisms grouped in it. If any two given organisms can be grouped under the same lower rank or taxon, it implies that the two organisms are structurally, functionally, embryologically similar and that they have had a **comparable evolutionary history**.

Within the living world as a whole, the **biggest taxonomic rank is Kingdom**. The next higher rank within a kingdom is the **Phylum** or **Division**. It is customary to use the term Phylum for major groups in the **Animal Kingdom** and the term Division for major groups in the **Plant Kingdom**. The Phylum or Division is a broad grouping of more or less closely related organisms, sharing certain common characteristics.

Each phylum or division has the next taxon called **Class**. The members of each class exhibit certain distinguishing characters that are unique only to them. In the same way, using comparable criteria of similarities and relationships, each class can be divided into orders, each order into families, each family into genera and each genus into species. **Species is normally the basic or fundamental unit of classification**. A **species** is, therefore, the **narrowest taxonomic category** and **kingdom** is the **broadest category** in the Linnaean hierarchy.

Thus, now Linnaean hierarchy containing **seven obligatory taxonomic categories** in **descending order** as following with example of animalia and plantae.

ANIMALS

Kingdom - Animalia
Phylum - Chordata
Class - Mammalia
Order - Primates
Family - Hominidae
Genus - *Homo*
Species - *sapiens*

PLANTS

Kingdom - Plantae
Division - Embryophyta
Class - Dicotyledonae
Order - Sapindales
Family - Anacardiaceae

Genus - *Mangifera*

Species - *indica*

7.5 Summary:-

- All the organisms should be classified i.e. divided into **groups** and **subgroups**.
The method of rearranging and regrouping of organisms into various divisions is called **classification**
- **Aristotle**, who is known as “**Father of Zoology**” first of all classified animals into three, groups namely **verms, insecta and vertebrata**.
- **Aristotle** classified a total of **520 species** of animals in his famous book “**Historia Animalium**”. Due to his unique tremendous contribution in Zoology, he is also known as “**Father of biological taxonomy**”.
- **Theophrastus**, who is known as “**father of Botany**” classified **480 plants** into trees, shrubs, undershrubs and herbs in his book “**Historia Plantarum**”.
- **Carolus Linnaeus** who used “**Binomial Nomenclature**” system of classification instead of using the common name in his famous book “**Systema Naturae**” known as “**Father of Modern Taxonomy**”.
- The unit of classification in both plants and animals is **species**.
- The kingdom is the **highest category** of taxonomic studies.
- The total number of species on earth is believed to lie somewhere between **80 to 120 lakh**.
- The **three domains** system was first proposed in 1990, but not generally accepted until later.
- In Zoology, the nomenclature for the more commonly used ranks is regulated by the International Code of Zoological Nomenclature (**ICZN Code**).
- In the fields of **Botany**, Phycology and Mycology, the naming of taxa is governed by the International Code of Nomenclature for Algae, Fungi, and Plants (**ICN**).
- Generally, hierarchy **means “a series of a succession of different rank”**.
- The most peculiar character of the Linnean hierarchy is the **descending sequence** of class, genus, species and variety taxonomic categories.
- Now Linnaean hierarchy containing **seven obligatory taxonomic categories** in descending order.

7.6 Self Assessment Questions:-

1 According to which Taxonomy is “Theory and practice of classifying organism”

- a. Linnaeus (b) Simpson
- (c) Mayr (d) A. P. de Candolle

Ans .- b

2 Father of taxonomy

- a. Darwin
- b. Mayr
- c. Hippocrates
- d. Linnaeus

Ans - d

3 Who proposed the binomial nomenclature?

- a. Linnaeus
- b. Mayr
- c. Huxley
- d. Darwin

Ans – a

4 Which is not a category?

- a. Aves
- b. Phylum
- c. Class
- d. Genus

Ans – a

5. International code of biological nomenclature applies to

- a. Animals
- b. Plants
- c. Virus

- d. Both a and b

Ans – d

6. Scientific name of animals must be derived from

- a. French
- b. Latin
- c. English
- d. German

Ans – Latin

7. Term Phylum was given by

- a. Linnaeus
- b. Cuvier
- c. Mayr
- d. John ray

Ans – b

8. The existence of two or more names belongs to the same taxon called

- a. Synonymy
- b. Homonymy
- c. Tautonymy
- d. Holonymy

Ans – a

9. Natural system of classification is based on

- a. Phylogeny and morphology
- b. Ontology
- c. Phylogeny
- d. Morphology

Ans – a

10. Linnaeus (1770-1778) published his scheme of classification in his book

- a. Systematics and origin of species
- b. Systema naturae
- c. Origin of species and evolution
- d. None of the above

Ans- b

11. According to binomial nomenclature words used for naming a plant or animal.

- a. Species
- b. Genus
- c. Species and genus
- d. Genus and species

Ans- d

12. The concept of the genus was first proposed by.

- a. Linnaeus
- b. Brunfels
- c. Bentham
- d. Julian Huxley

Ans – b

13. Who is the father of biological classification?

- a. Aristotle
- b. Pluto
- c. Linnaeus
- d. Hippocrates

Ans- Aristotle

14. Important character of Linnaean hierarchy is

- a. Ascending order
- b. Descending order

- c. Both as above
- d. None of them

Ans. – b

15. Presently numbers of category in the Linnaean hierarchy are

- a. 5
- b. 10
- c. 12
- d. 07

Ans. – d

7.7 References:-

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7.8 Suggested Reading:-

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3. Kohli, Rnga, and Lori, *Animal Diversity and Evolution*, Ramesh Book Depot. Jaipur.

7.9 Terminal Questions:-

1. What do you understand by Binomial System of Classification?

2. Explain taxonomic characters?
3. Explain Trinomial System of Classification?
4. What are the different types of classification?
5. Describe the aims and rules of Nomenclature?
6. Describe the Linnaean hierarchy?
7. Describe briefly Linnaean system of classification?
8. Describe taxonomic category?
9. What are taxonomic components?

UNIT 8: CONCEPTS OF SPECIES

Contents

- 8.1 Objectives
- 8.2 Introduction
- 8.3 Species Concept
 - 8.3.1 Concept of Species
 - 8.3.2 Speciation
 - 8.3.3 Potential mode of Speciation
- 8.4 Summary
- 8.5 Self Assessment Questions
- 8.6 References
- 8.7 Suggested Readings
- 8.8 Terminal Questions

8.1 Objectives :-

In this chapter you would learn about species, types of species, who coined the term species, the definition of species, species concept, and nature of species, speciation and potential mode of species. Before taxonomists **philosophers** identify, name and classify organisms according to own interests, and they need to agree on a definition of the concept of species. The definition of species is not as simple as it may seem and has been debated by biologists and philosophers alike for many years.

8.2 Introduction:-

In the last chapter, you have learned about **taxonomic characters** and **binomial, trinomial** systems of classification. In this chapter you would learn about species, and species concept. Species is the most important category in the **taxonomic hierarchy** and it is the **lowest rank** of a group that cannot **interbreed** in nature. Origin of species is the **monumental** and most interesting general treatment of **evolutionary biology**. The pages that follow besides containing explanation due to the theory of **natural selection** contain information about discoveries made since then which relate primarily to the nature and definition of species and the phenomenon of **speciation**.

8.3 Species Concept:-

The term species was first coined by **John Ray** (1628-1705). He described **18000 plants** in his famous book entitled “**Historia Generalis Plantarum**” which was published in **three volumes** from 1686 to 1704. He was the first person who made a differentiation between **genus and species**.

Species is the most important category in the taxonomic hierarchy. It is the **basic unit** of **evolution**. It is a group of individuals in plants, as well as animals which, resemble closely in structure as well as in functions. **Decan Dolle** defines species as a collection of all individuals which resemble with each other, more than they resemble anything else, which can be **mutual fecundation** produce fertile individuals and which reproduce themselves by generations in such a manner that we may form an **analogy** to consider that all have sprung from a single individual. It is a very important unit, not only for taxonomists but also for working in all allied fields of Biology as well.

The species is the **lowest rank** of a group that cannot interbreed in nature. This is what has been called the biological view of the species (as against paleontological, morphological or taxonomic view). One of the best biological definitions of the species has been given by **Mayr**, who says:

“A species consists of a group of populations which replace each other geographically or ecologically and of which neighboring ones intergrade or interbreed whenever they are in contact or which are potentially capable of doing so (with one or more of the populations) in those cases where contact is prevented by geographical or ecological barriers”.

Or shorter

“Species are a group of actually or potentially interbreeding populations which are reproductively isolated from other such groups”. In other words, a **species** is a group of organisms that interbreed and produce fertile offspring only with one another. Some microbiologists and botanists are dissatisfied with the biological **species concept**.

Most **modern biologists** agree that species, unlike higher taxa (genus, family, order, and so on), are **authentic taxonomic units**. In other words, species really do exist in nature and are not merely artificial human constructs. **The term "species" is used simultaneously for the unit of evolution & the unit of classification, a taxonomic category below genus.**

CHARACTERISTICS OF A SPECIES: Species have some important characteristics, which are as following:

1. Each species having a large **gene pool** of which an organism containing a little quantity for a very short period.
2. All organisms of a species resemble each other in all respects.
3. Organisms of a species differ from other groups of organisms.
4. Organisms of a species can interbreed freely and produce **fertile offspring**.
5. Organisms of a species are **reproductively isolated** from other groups of organisms.
6. The organisms of a species containing a common **gene pool** and biochemical properties.
7. All the organisms of a species have a similar **genetic system**.
8. All the organisms of a species show **common ancestry** during organic evolution or speciation.
9. Each organism of a species has the power to produce **new species**.
10. The organisms of a species have the ability of adjustment according to its environment.

8.3.1 Concept of Species:-

Usually the **populations** of the same kind of individuals which are similar in structure, function, behavior and also produced a similar type of offspring called **species**. Although, the species is not only the basic fundamental unit of classification but also basic unit of evolution, while other categories e.g. genus, family, order, class, phylum, and kingdom are the man-made **artificial units**. According to **Simpson**, species may be defined in the form of Biological Species and Genetical Species.

BIOLOGICAL SPECIES: Biological species containing similar cytological, ecological, physiological, behavioral characteristics and able to interbreed with reproductively isolated from such other groups of organisms.

GENETICAL SPECIES: Genetically species is the group of interbreeding populations but reproductively isolated from each other. Mayr (1912) defined species as “a population of interbreeding individuals”.

In 1957 **Mayr** classified as four main species concepts:

- I. Typological species concept
- II. Nominalistic species concept
- III. Genetical species concept
- IV. Evolutionary species concept

I. TYPOLOGICAL SPECIES CONCEPT: Typological species concept is based on **morphology** or **phenotype** of any organism. Species are a type of organism. Species are as many as were created in the beginning by the infinite. According to the **Linnaeus** and other scientists of the eighteenth century, the organism of a species was closely resembled each other in most of the morphological characters. But this species concept has been **rejected** on the basis that the individuals of same species show **some striking differences** like sexual dimorphism, age distribution, different developmental stages, polymorphism and variety etc.

II. NOMINALISTIC SPECIES CONCEPT: According to this concept the species is the result of **man's own creation**. This concept has been rejected on the basis that species is the result of only gradual organic evolution and not the production of human being's mind.

III. GENETICAL SPECIES CONCEPT: According to **Mayr** (1942) the species may have also classified as a population of interbreeding individuals, which are reproductively isolated from each other. Each species has a **large gene pool** while individual having only a small portion of this gene pool for a short period. These individuals are similar in structure, function, behavior and in the production of similar **progeny**. Based on the similarity of **DNA** of individuals or

populations, techniques to compare the similarity of DNA include **DNA-DNA hybridization** and genetic **fingerprinting** is being used.

IV. EVOLUTIONARY SPECIES CONCEPT: A single evolutionary lineage of organisms within which genes can be shared, and that maintains its integrity with respect to other lineages through both time and space. At some point in the evolution of such a group, some members may **diverge** from the main population and evolve into a **subspecies**, a process that may eventually lead to the formation of a **new species** if isolation (geographical or ecological) is maintained. **The process through which species are formed by evolution is called speciation.** A species that gives rise to another species is a **paraphyletic species** or **paraspecies**. An evolutionary species is a single lineage of ancestor-descendant populations which maintains its identity from other such lineages and which has evolutionary tendencies and **historical fate**.

OTHER SPECIES CONCEPTS

ECOLOGICAL SPECIES: A set of organisms adapted to a particular set of resources, called a **niche**, in the environment. According to this concept, populations form the discrete **phenetic clusters** that we recognize as species because the ecological and evolutionary processes controlling how resources are divided up tend to produce those clusters.

REPRODUCTIVE SPECIES: Two organisms have the ability to reproduce naturally fertile offspring of both **sexes**. Organisms that can reproduce but almost always make infertile hybrids of at least one sex, such as a **mule, hinny** or **F1 male cattalo** are not considered to be the same species.

ISOLATION SPECIES: It is a set of actually or potentially interbreeding populations. This is generally a useful formulation for scientists working with living examples of the higher taxa like **mammals, fish, and birds**, but more problematic for an organism that does not reproduce sexually. The results of breeding experiments done in artificial conditions may or may not reflect what would happen if the same organisms encountered each other in the wild, making it difficult to gauge whether or not the results of such experiments are meaningful in reference to natural populations.

COHESION SPECIES CONCEPT: the most inclusive population of individuals having the potential for phenotypic **cohesion** through intrinsic cohesion mechanisms. This is an expansion of the mate-recognition species concept to allow for post-mating isolation mechanisms. No matter whether populations can **hybridize** successfully, they are still distinct cohesion species if the amount of hybridization is insufficient to completely mix their respective gene pools.

SPECIES AS A CATEGORY

Species is a most important **natural feature** of taxonomy. Usually, the individual of the same species is found in all different region of the world. Therefore, a species can be divided into subspecies on the basis of the presence of their individuals. These species differ from main species in some minor **anatomical characteristics** but the major difference may be observed in distinct **diagnostic characteristics**.

In the strict sense, it may be said that individuals of a species may be differentiated on the basis of morphological characters, color, size and shape. Such type of species may be described as **monospecies**, which is the first categorization of any species. The second categorization of any species is the biological species in which the individual of a population have the ability of interbreeding and reproductively isolated from other such groups of organisms. The biological category has the following three **distinct characters**:

- A. Reproductive community
- B. Ecological community
- C. Genetical community

A. REPRODUCTIVE COMMUNITY: The individuals of each species are identified by each other for the purpose of **reproduction** because the individual of one species does not cross breed with individuals of **another species**. Thus, a species behave as a reproductive unit under the biological category.

B. ECOLOGICAL COMMUNITY: The individuals of a species interact with each other and also affect their **environment**. Thus, a species act as an ecological community or unit.

C. GENETICAL COMMUNITY: Each biological species containing a large pool of which an individual having a small portion for a **short period**. There is a large **gene flow** during interbreeding of individuals of same species. Thus, as a result, genetic changes appear in its **progeny**. Therefore, genetical community or unit may also be considered as a **species category**.

KINDS OF SPECIES

Various taxonomists have been defined species according to its **own interests**. Now the definition of species is not restricted only to the taxonomists but cytologists, geneticists, ecologist, biochemist and others define species in different manners. These manners are called kind of species and are as followings:

1. **AGAMO SPECIES:** Asexually reproducing species are called agamo species.
2. **GAMO SPECIES:** Sexually reproducing species are called gamo species.
3. **ALLOPATRIC SPECIES:** The species which are found in different habitat or

Different geographical areas called allopatric species.

4. **ALLOCHRONIC SPECIES:** The species which are found in a different time period called allochronic species.
5. **SYNCHRONIC SPECIES:** The species which are found in the same time period called synchronic species.
6. **POLYTYPE SPECIES:** The species which are containing two or more subspecies called polytype species.
7. **MONOTYPE SPECIES:** The species containing only a single subspecies are called monotype subspecies.
8. **MORPHO SPECIES:** The species which are similar in morphological characters **called morpho species.**
9. **SYMPATRIC SPECIES:** The species which normally inhabit the same **habitat or same geographical area called sympatric species.**
10. **GENETICAL SPECIES:** The population of interbreeding individuals which are reproductively isolated from each other called genetical species.
11. **BIOLOGICAL SPECIES:** The population of interbreeding individuals which are reproductively isolated from one another called biological species.
12. **SIBLING SPECIES:** The group or pair of very closely related species is called sibling species.
13. **TAXONOMIC SPECIES:** The species which have been named under rules of nomenclature are called taxonomic species.
14. **EVOLUTIONARY SPECIES:** The species which shows ancestral relationship during the evolutionary period are called evolutionary species.
15. **PALAEONTOLOGICAL SPECIES:** The species which are found in the form of fossils are called palaeontological species.
16. **NEONTOLOGICAL SPECIES:** The species which are found in a living condition are called neontological species.

8.3.2 Speciation:-

In many respects, **Birds** (Class Aves) are the most highly specialized to **Craniata**. They are adapted for **aerial life**. The organization of existing birds is, singularly uniform and every part of it **modified** in accordance with the unusual environment. They represent less diversity of structure than many single orders of **Fishes, Amphibians** and **Reptiles**.



Fig.8.1 Penguin

Speciation is the evolutionary process by which reproductively isolated biological populations evolve to become distinct species. The biologist **Orator F. Cook** was the first to coin the term '**speciation**' for the splitting of **lineages** or "**cladogenesis**", as opposed to "**anagenesis**" or "**phyletic evolution**" occurring within lineages. **Charles Darwin** was the first to describe the role of "Natural Selection" in **Speciation**.

It is widely appreciated that **sexual selection** could drive speciation in many **clades**, independently of natural selection. However the term "**speciation**", in this context, tends to be used in two different, but not mutually exclusive senses.

The first and most commonly used sense refers to the "**birth**" of **new species**. That is, the splitting of an existing species into two **separate species**, or the budding off of a new species from a parent species, both driven by a biological "**fashion fad**" (a preference for a feature, or features, in one or both sexes, that do not necessarily have any adaptive qualities).

In the second sense, "**speciation**" refers the wide-spread tendency of sexual creatures to be grouped into clearly defined species, rather than forming a continuum of **phenotypes** both in **time and space** - which would be the more obvious or logical consequence of natural selection. This was indeed recognized by **Darwin** as problematic, and included in his "**On the Origin of Species**" (1859), under the heading "**Difficulties with the Theory**". There are several suggestions as to how mate might play a significant role in resolving **Darwin's dilemma**.

8.3.3 Potential Modes of Speciation :-

There are **four geographic potential modes of speciation** in nature, based on the extent to which speciating populations are isolated from one another: Speciation may also be induced **artificially**, through animal husbandry, agriculture or laboratory experiments.

All forms of natural speciation have taken place over the course of **evolution**. However, debate persists as to the relative importance of each mechanism in **driving biodiversity**. **One example** of natural speciation is the diversity of the **three – spined Stickleback**, a marine fish that, after the last glacial period, has undergone speciation into new freshwater colonies in isolated **lakes** and **streams**.

Over an estimated **10,000 generations**, the sticklebacks show structural differences that are greater than those seen between different genera of fish including variations in fins, changes in the number or size of their bony plates, variable jaw structure and color differences. However, the natural speciation's are as following:

- A. Allopatric
- B. Peripatric
- C. Parapatric
- D. Sympatric

A. ALLOPATRIC: During allopatric (Greek: allos- "other" + Greek: patra- fatherland") speciation, a population splits into two geographically isolated populations (for example, by habitat fragmentation due to geographical change such as mountain formation).The isolated populations then undergo genotypic and/or phenotypic divergence as:

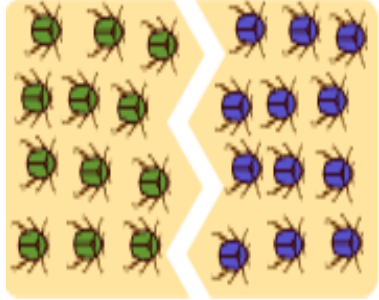
- (a) They become subjected to dissimilar selective pressures.
- (b) They independently undergo genetic drift
- (c) Different mutations arise in the two populations.

When the populations come back into contact, they have evolved such that they are reproductively isolated and are no longer capable of exchanging genes. Island genetically is the term associated with the tendency of small, isolated genetic pools to produce unusual traits. Examples include insular dwarfism and the radical changes among certain famous island chains, as on Komodo.

The Galápagos Islands are particularly famous for their influence on Charles Darwin. During his five weeks there, he heard that Galápagos tortoises could be identified by island, and noticed that finches differed from one island to another, but it was only nine months later that he reflected that such facts could show that species were changeable.


When Charles Darwin returned to England, his speculation on evolution deepened after experts informed him that these were separate species, not just varieties, and famously that other differing Galápagos birds were all species of finches. Though the finches were less important for Darwin, more recent research has shown the birds now known as Darwin's finches to be a classic case of adaptive evolutionary radiation.

Mode of Speciation (Type)	New Species formed from...
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Allopatric (allo = other, patric = place)	Geographically isolated populations	
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C. PERIPATRIC: In peripatric speciation, a **subform** of **allopatric speciation**, new species are formed in isolated, smaller peripheral populations that are prevented from exchanging genes with the **main population**. It is related to the concept of a founder effect since small populations often undergo bottleneck. **Genetic drift** is often proposed to play a significant role in peripatric speciation.

Mode of Speciation (Type)	New Species formed from...
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Peripatric (peripatric = near, patric = place)	A small population isolated at the edge of a larger population	
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C. PARAPATRIC: In parapatric speciation, there is only partial separation of the zones of two diverging populations afforded by **geography**. Individuals of each species may come in contact or cross habitats from time to time, but reduced fitness of the heterozygote leads to selection for behaviors or mechanisms that prevent their **interbreeding**.

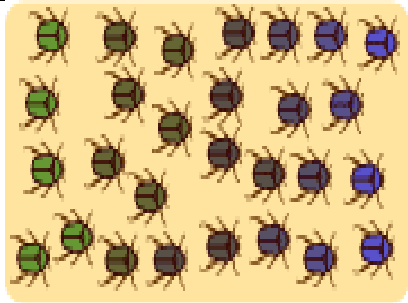
Parapatric speciation is modeled on continuous variation within a "single" connected habitat acting as a source of natural selection rather than the effects of isolation of habitats produced in peripatric and allopatric speciation. Parapatric speciation may be associated with differential **landscape-dependent selection**. Even if there is a gene flow between two populations, strong differential selection may impede assimilation and different species may eventually develop.

Habitat differences may be more important in the development of reproductive isolation than the isolation time. Caucasian rock lizards *Darevskia rudis*, *D. valentini* and *D. portschinskii* all **hybridize** with each other in their **hybrid zone**; however, hybridization is stronger between *D. portschinskii* and *D. rudis*, which separated earlier but live in similar habitats than between *D. valentini* and two other species, which separated later but live in **climatically different habitats**.

Ecologists refer to parapatric and peripatric speciation in terms of **ecological niche**. A niche must be available in order for a new species to be successful.

Mode of Speciation
(Type)

New Species formed from...

Parapatric (para = beside, patric = place)	A continuously distributed population	
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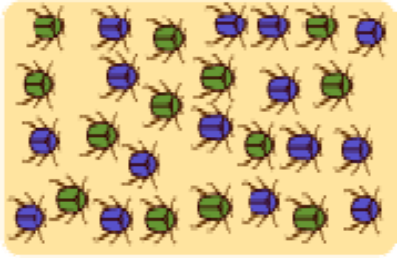
D. Sympatric: sympatric speciation refers to the formation of two or more descendant species from a single ancestral species all occupying the same geographic location. Often-cited examples of sympatric speciation are found in insects that become dependent on different host plants in the same area. however, the existence of sympatric speciation as a mechanism of speciation remains highly debated.

The best-illustrated example of sympatric speciation is that of the **Cichlids of East Africa** inhabiting the **Rift Valley Lakes**, particularly Lake **Victoria**, Lake **Malawi** and Lake **Tanganyika**. There are over **800** described species, and according to estimate, there could be well over **1,600** species in the region. All the species have diversified from a **common ancestral** fish, the Japanese **rice fish** (*Oryzias latipes*) about 113 million years ago. Their evolution is cited as an example of both natural and sexual selection.

Sympatric speciation driven by ecological factors may also account for the extraordinary diversity of **crustaceans** living in the depths of **Siberia's Lake Baikal**.

Budding speciation has been proposed as a particular form of **sympatric speciation**, whereby small groups of individuals become progressively more isolated from the ancestral stock by

breeding preferentially with one another. This type of speciation would be driven by the **conjunction** of various advantages of inbreeding such as the expression of advantageous recessive phenotypes, reducing the **recombination load**, and reducing the cost of sex.

Mode of Speciation (Type)	New Species formed from...	
Sympatric (sym = same, patric = place)	Within the range of the ancestral population	

8.4 Summary:-

- Term species was first coined by John Ray.
- John Ray described 18000 plants in his book “Historia Generalis Plantarum”
- John Ray was the first person who made a differentiation between Genus and Species.
- A species is a basic fundamental unit of classification.
- A species is also a basic unit of evolution.
- A species may be defined as “a group of actively or potentially interbreeding natural populations that are reproductively isolated from other such groups”.
- Each species has a large gene pool.
- An organism contains a little quantity of its gene pool.
- All organism of a species contains a similar genetic system and hereditary materials.
- The species is the lowest rank of a group that cannot interbreed in nature.
- The date on which species name was published may also be added to the scientific name e. g. *Homo sapiens* Linn. 1758.
- Homology establishment helps much in finding the exact position of an organism.
- Simpson defined species in biological and genetical forms.

- Typological species concept is based on **morphology** or **phenotype** of any organism.
- According to the nominalistic concept of species, species is the result of **man's own creation**.
- Each species has a large gene pool while individual having only a small portion of this gene pool for a short period.
- Evolutionary lineage is basically responsible for the evolution of new species.
- The process through which species are formed by evolution is called speciation.
- A set of organisms adapted to a particular set of resources called a **niche**.
- A species behave as a reproductive unit under the biological category.
- **Speciation** is the evolutionary process by which reproductively isolated biological populations evolve to become distinct species.
- **Orator F. Cook** was the first to who coined the term '**speciation**'.
- Charles Darwin was the first to describe the role of Natural Selection in Speciation.
- There are **four geographic potential modes of speciation** in nature

8.5 Self Assessment Questions:-

1. Who introduced the concept of species?

- John ray
- Mayr
- Aristotle
- Huxley

Ans - a

2. Karyotaxonomy is based on

- Number of chromosomes
- Bands founds in chromosomes
- Organic evolution
- Trinomial nomenclature

Ans - a

3. Species which occur at the same time level

- Allochronic species

- b. Synchronic species
- c. Sibling species
- d. Agno species

Ans - b

4. A pair or group of closely related species which reproductively isolated but morphologically identical are

- a. Sibling species
- b. Subspecies
- c. Ecospecies
- d. Variety

Ans - a

5. Species which occupy different areas of distribution are called.

- a. Allopatric
- b. Sympatric
- c. Holotype
- d. Paratype

Ans - a

6. Species do not reproduce sexually but reproduce by parthenogenesis

- a. Panmictic species
- b. Apomictic species
- c. Agno species
- d. None of the above

Ans – b

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8.8 Terminal Questions:-

1. What is speciation? What is a role in nature?
2. What do you understand by species concept?
3. Describe the characteristics of a species?
4. What are the potential modes of speciation?
5. Define species as a category?
6. Write the process of sympatric speciation?

7. Differentiate between the following?
- A. Allopatric and Sympatric species
 - B. Polytypic and Monotypic species.
8. Define species?

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UNIT 9: ORIGIN OF LIFE

Contents

9.1- Objectives

9.2- Introduction

9.3- Special Creation Theory

9.3.1- Special Creation Theory

9.3.2- Theories of Spontaneous generation

9.3.3- Cosmozoic Theory

9.4- Modern concept of origin of life

9.4.1- Chemical evolution

9.4.2- Origin of primary organism

9.5- Summary

9.6- Self-assessment question

9.7- Suggested Readings

9.8- Terminal Questions

9.1 Objectives:-

If there were other life out there in the universe, how similar do you think it would be to life on Earth? Would it use DNA as its genetic material, like you and me? Would it even be made up of cells? We can only speculate about these questions, since we haven't yet found any life forms that hail from off of Earth. But we can think in a more informed way about whether life might exist on other planets (and under what conditions) by considering how life may have arisen right here on our own planet.

In this chapter, we'll examine scientific ideas about the origin of life on Earth. The when of life's origins (3.5 billion years ago or more) is well-supported by fossils and radiometric dating. But the how is much less understood. In comparison to the central dogma or the theory of evolution, hypotheses about life's origins are much more...hypothetical. No one is sure which hypothesis is correct – or if the correct hypothesis is still out there, waiting to be discovered.

9.2 Introduction:-

The term 'evolution' can be defined as “the changes in the genetic composition of a population with the passage of each generation.” The outcome of the evolutionary process is an adaptation of an organism to its environment. The evolution is the property of population and not of individuals. Natural selection is the evolutionary force.

In our solar system, there are seven major planets besides Earth. Neptune, Uranus, Saturn and Jupiter are said to be having clouds like surface but Mercury and Venus lack water and atmosphere. The Mars which distance from earth is about 35,000,000 miles possibly has oxygen, CO₂ and water but with temperature ranging between 10°C to freezing point. Some life has been reported to be present on Mars but still, it is a matter of debate. The life cannot exist all the time on earth because the high temperature and dry climate of the early time of earth would have made life impossible to exist. The world has its own account of the origin of life.

The earth was cast off from some molten and hot gaseous material. This mass later condensed and gradually cooled and decreased in volume. The earth acquired in the course of time a gaseous atmosphere with sufficient pressure to retain water on the surface. The water filled the deep area, which made sea and oceans. The life could appear only after the water and lands had cooled.

9.3 Ancient theories related to Origin of Life:-

9.3.1 Special Creation Theory:-

The biblical story of the creation of world within six days was put forward by Spanish monk father Suarez. He described that the earth and heaven were created on the first day and sky on the second day. The third day the earth surface was dried and ancestors of plant and animals originated. The sun, the moon, and the stars were created on the fourth day. The birds and fishes are created on the fifth day and finally, man and beast were created on the sixth day of creation. In the end of the seventh day, a woman was constructed from the 12th ribbed of the man.

9.3.2 Theories of spontaneous generation:-

This theory is also known as the theory of a-biogenesis. According to this theory, life has originated from the non-living organic material. Anaximander and Anaxagoras believed that life appeared in the small seed which came to earth along with rain water. Aristotle suggested that a number of animals originated in the way discussed above. A number of worms, larvae of bees, larvae of wasps, ticks, flies and many other insects develop from the morning dew or from decaying slime manure, from dry wood, hair, sweat and meat while tapeworms are born in the rotting portion of the body and excreta. Mosquitoes, flies, moths, beetles, fleas, bed-bugs and bees are generated in the slime of well, rivers or sea, in the humus of the fields, in manure, in decaying trees or fruits etc. crab and mollusks were brought to come from the moist soil and decaying slime. Some higher animals have similar origin though in the case of latter his first appearance is in the form of a worm. In fifteen century, it was thought that leaves falling from trees turned into fish if they fell in the water and turned into a bird if they fell on land.

Experimental studies

Redi's experiments

Francesco Redi (1626-1698) was the first to put forth the experimental evidence of the concept of spontaneous generation. He placed the meat or fish in three large jars. One jar was left open, one was covered with gauze and one was covered with a muslin cloth. The meat or fish decayed in all jars and flies were attracted to all. He showed that the white maggots in the meat of the first jar were the larvae of flies and nothing else. He notices that in the second jar, worms did not appear in the meat. However, he noticed the eggs and some developing stages on the wire

gauze. He, therefore, concluded that the decaying substance or soil or mud was only a place or nest for the development of the insects and that the necessary prerequisite for the appearance of the worms was laying of eggs.

Lazzaro Spallanzani disproved the theory of spontaneous generation in 1765. He boiled the meat in the sealed long-necked flask. The broth remained clear for months. No sign of life was recorded. Needham claimed that by boiling, the vital forces necessary for a spontaneous generation had driven out. Then the seal was broken and the broth was exposed to fresh air. On testing the broth, the presence of microbes proved the origin of life from preexisting life.

Louis Pasteur disproved the theory of spontaneous generation in the nineteenth century. He boiled a solution of sugar and yeast for several hours in a swan neck flask and the flask was left unsealed. The solution remained free of microbes because the swan neck flask was shaped so to trap viable microbial particles and to allow only air to enter the flask. After breaking the neck of the flask, he reported the micro-organism in the solution, thus he disproved the concept of spontaneous generation.

9.3.3- Cosmozoic Theory:-

The cosmozoic theory is also known as Panspermia theory. According to this theory, the life is distributed throughout the cosmos in the form of the resistant spores of living forms, the cosmozoa. These reached the earth accidentally from some other planet, and on getting favorable conditions for life these developed into organisms. The cosmozoic theory was proposed by Richter. According to this theory, life came from another planet in the form of celestial bodies and small particles carrying viable germs or spores, which upon reaching on earth accidentally, could develop and initiate panoply of living organisms. Life only changes its form but is never created from dead substances. It has no origin and has always existed. Preyer assumed that life must have existed even at that time when the earth was a mass of molten liquid. According to him, life comes from life and never from dead material.

9.4 Modern concept of origin of life:-

A.I Oparin, published a book named “**The Origin of Life**” in 1939. In addition, several realistic theories have also been offered to explain the origin of the earth and life, but the most widely accepted theory today is known as the Big Bang theory, proposed in 1951. Before twenty billion years the universe was one big ball of neutrons or neutral particles. The movement of these particles becomes greater until the big ball generated the nearly unbelievable amount of heat. The increase in temperature caused a parallel increase in pressure. Finally, the big ball exploded and created the biggest bang ever known. Neutrons were flung everywhere. As the

neutrons moved farther from their point of origin, they began to cool and produce negative charges or electron. The production of electron left behind protons and the attraction of electron to proton created hydrogen. This process continued until the newly formed particles began to aggregate into small balls. Each ball becomes a galaxy; our galaxy is the Milky Way. Within each galaxy, the process continued to form smaller balls, creating the solar system. This ball can be best thought of as clouds of gases, which astronomers call dust clouds. As time passes each dust cloud became cooler. Many dust clouds developed extremely cold temperature that hovered near absolute zero. However, as the particle of the dust cloud showed down and moved closer, heat once again was generated. The heat becomes too intense so as to cause the fusion of hydrogen, forming helium and releasing energy in the form of light and heat. The acceleration of this process caused dust clouds to throw off groups of particles, creating eddies of smaller clouds. The hot and illuminated central masses became the stars of the universe, the less hot eddies of dust radiating around them became a planet. Today these processes continued. Stars and planets are constantly being born throughout the universe while other explode and disappear into oblivion.

Our earth came into existence in five billion years ago. Earth was like other planets. It was at first a very hot molten mass of materials. However, as the mass cooled, hydrogen became the basic building block from which all other elements were made. The core of earth today is still a hot molten ball, volcanic eruptions not only demonstrate the existence of a molten core but also provide a glimpse of what the earth was like much earlier when volcanoes that dotted its surface were continually erupting.

9.4.1 Chemical Evolution:-

From monomers to polymers

First, the amino acids began to accumulate in the oceans and smaller bodies of water and they embedded into proteins and other macromolecules. Sidney Fox of the University of Miami found that heating a dry mixture of amino acids causes the formation of long proteinoids polymers having a molecular weight of more than 10,000. Fox has suggested that such polymerizations took place in volcanic cinder cones and that the proteins formed were then washed into the sea. J.B.S. Haldane and other considered it more likely that the first macromolecules were formed in sea water or pond water rather than formed dried mixtures of monomers. This too has been shown to be possible, for solutions of amino acids will form polypeptides in the presence of hydrogen cyanide even at the suitably low temperatures.

On other hand polymerizing monomers of various types and to wet and dry them, alternately, on the surface of the clay. The historical operation of this mechanism is particularly plausible from geological points of view. According to Miller, in which simple molecules were

formed, many different conditions have been shown to be compatible with the formation of proteins and other polymers.

Microspheres

This is the accumulation of the biological polymers and other compounds into isolated droplets of increasing complexity. There are, in fact, several ways in which such accumulation can be accomplished in the laboratory.

Example: Fox found that his proteinoids have a remarkable tendency to form microspores approximately 2µm in diameter when hot, concentrated solution of the proteinoids is slowly cooled. These microspheres show a double layered boundary resembling a membrane and they swell and shrink as the salt concentrations in the solution is changed. If allowed to stand for several weeks, the microspores absorb more proteinoid material from the solution produce buds and sometimes divide to produce second generation microspheres. Cleavage or division can also be induced by changing pH or adding magnesium chloride. These microspheres should not be taken to be the ancestors of life.

One method for the accumulation of chemical substance into partially organized structures was proposed by the Irish physicist J.D. Bernal. This method involves some clay particles, such particles have electrical charges that attract and bind substances such as protein. Methane, ammonia and water vapour can be subjected to electrical discharge and among the products are spheres, one-quarter of a micrometer in diameter, consisting of mixtures of biological molecules bound to clay-like particles eroded from the glass of the reaction chamber.

Theory of chemical evolution

It is possible that the immediate precursors of the living organism were capsules of chemical reaction similar to coacervate droplets. Some coacervates would enclose reactions that led to the early breakup of the droplets; other would enclose reaction that made them stable. The more stable coacervates would survive longer and could possibly grow at the expense of their surroundings by absorbing chemical substances derived from the remains of the less stable droplets.

If wave action of other chemical forces broke a large coacervate into many small droplets, each of these might be able to absorb the material and grow on its own. This stage of evolution would be purely a matter of chemical competition. Any non-biological catalysts that accelerated the rate of favorable reactions in a given type of coacervate would give it a great advantage over more slowly reaction droplets. Chemical selection, therefore, would favor catalyzed reactions. It is not hard to imagine more and more efficient catalyst would be developed and retained by chemical selection until finally, the evolving system stumbled on to the ultimate improvement of protein like catalysts enzymes.

Oparin postulated the existence of organized metabolizing but non-reproducing systems that he called protobionts. According to this reasoning, the breakthrough that led to truly living organism was the development of reproduction, the ability of a successful chemical system to ensure its survival by duplicating itself. The molecules in which the instructions for duplication are stored in modern living creatures of DNA or RNA. Yet the living unit of life is not just the nucleic acid as a computer without a program, but the DNA or RNA alone can be more live than a program without a computer can also do the calculation. Any simple biological molecules released into today's environment are quickly consumed by already living things. For another, such molecules are no longer accumulating through the mechanism. Earth atmosphere has changed. Oxygen too can oxidize biological molecules. In addition, it gives rise to the ozone that filter ultraviolet from the sunlight falling on the planet. In doing so, it blocks one of the sources of energy once available for promoting chemical reactions. In sum, spontaneous generation is a thing of the past.

Meteorites and extraterrestrial life

The most primitive Precambrian bacteria were probably compared with non-living matter. Their discovery sheds no light on the central question of chemical evolution. Earth is steadily bombarded with showers of meteors presumably the debris the shattered asteroids, and some of this material contains organic molecules also found in living systems. Most meteorites are metallic, but a relatively small number are soft and crumbly, with high carbon content. These soft meteorites are called carbonaceous chondrites, and the meteorites that fell in a shower around Orgueil belong to this category. A variety of hydrocarbons have been found and some of the organic compounds are optical isomers, which are usually associated with synthesis carried out by living organism. Some amino acids found in meteorites are ones not found in organisms on this planet and, hence, cannot be contaminants introduced after the meteorites fell. Spheroids and other organized bodies of some complexity have been reported, but a continuation of the meteorites samples by airborne spores and pollen has confused the issue. In at least on the instance, the organized bodies turned out to be ragweed pollen. Most of the complex organized bodies have proved to be terrestrial contaminants and those that are definitely meteoric in origin are sufficiently simple that they may be natural mineral formation rather than artifacts of life.

The presence of hydrocarbons and other biochemical compounds in the meteorites indicates that at least the first step in molecular evolution for the formation of complex organic compounds can occur spontaneously even in space. These meteorites are not evidenced for life on some shattered planet; they may be evidence for the universality of the organic chemical-rich environment in which life could develop.

Origin of primitive living organism

The coacervates showed some chemical reactions which produced special proteins and enzymes. This led to self-replication of compounds; those processing this property might be

regarded as a free gene. Such a structure is comparable with the free-living virus and is supposed to be, formed of nucleoproteins. Self-replication and mutation of a gene could lead to the formation of gene aggregates. Such gene aggregates may be regarded as independently existing chromosomes. It is believed that some of the smallest bacteria represent such a stage in the evolution. The mutation might be led to the accumulation of metabolites around the chromosomes. The complex so formed represents the exposed nucleus. Some of the bacteria showed this kind of structure. The cytoplasm has been acquired but not separated from the nuclear material as in blue-green algae and in some large bacteria.

Miller's Experiment

Stanley L. Miller proved the important evidence in support of chemical synthesis of life. A mixture of some gasses like ammonia, methane, and hydrocarbon was taken in a special flask. A high-frequency spark by tungsten electrodes was discharged in a constantly circulating mixture of gases for about a week. During the period of the experiment, steam is supplied from the boiling water which mixes with the other gases. The steam thus formed condenses to water through the condenser and flows back to the boiling water flask. After the experiment is completed, the resulting fluid collected in the U-shaped tube and analyzed. The mixture consisted of various acid and amino acids e.g. glycine, alanine, B-alanine and aspartic acid, important for protein synthesis.

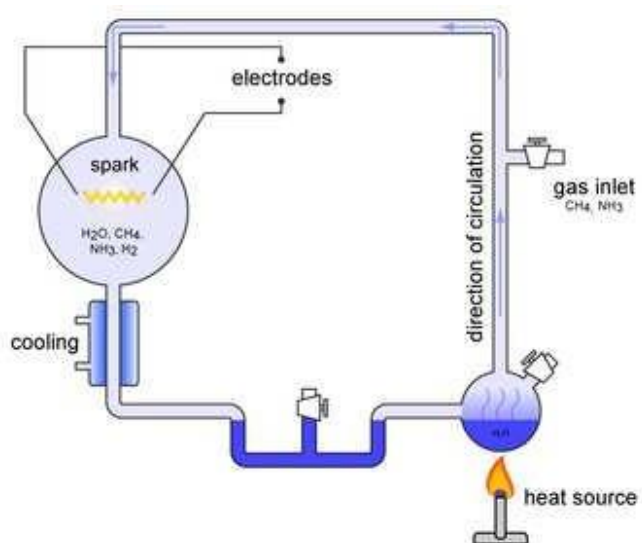


Fig.9.1 Miller–Urey experiment

9.4.2 Origin of primary organism

Oparin's coacervates had common properties and still definite individualism ties and structures. These could grow in size as a result of absorbing substances dissolved in the surrounding water thus, became more complex and created diversity among them, Due to the

presence of some substances like iron, copper, and calcium etc. first rudiments of future enzymes were formed. The primitive organisms were successful coacervates. Blum states that the source of free energy available for the first living thing was the absorption of ultraviolet rays. According to others thought, coacervates utilized energy during fermentation of organic substances absorbed from seawater. The production of energy was done through anaerobic respiration. The sea water provided the necessary raw material for the duplication of nucleoprotein and thus, the amount of it was increased. From nucleoprotein virus-like organisms developed.

Cellular life

Horowitz in 1945 and Orgel in 1976 stated that life originated in its simplest form in the sea. The genes along with proteins developed long chains of nucleoproteins that can be compared with chromosomes. The molecules of nucleoproteins along with organic compounds of sea developed a membranous covering and thus, the cells similar to prokaryotic cell were evolved. This cell has protein and some other organic substance in the colloidal state around DNA molecules but devoid of the nucleus, mitochondria, chloroplast, Golgi apparatus, lysosome and other organelles. These cells were holozoic as far as their nourishment is a concern. They can use the dissolved organic material present in marine water. They used solar energy and synthesized their own food. Now, for the first time, free oxygen was liberated out in the atmosphere.

9.5 Summary

In the end, it can be concluded that the process of evolution is an ever continuing process; it has not stopped but is occurring more rapidly today than in many of the past decades. In the last few hundred –thousand years, hundreds of species of animals and plants have come become extinct and other hundreds arise.

Numbers of simpler and lower animals are aquatic and since the cell and body fluids of all animals contain salts, it is inferred that life began in the ocean. Many biologists believe that life originated in the tidal zone which is rich in oxygen, CO₂, light and minerals and is most suitable for plant and animal growth. The earliest animal remains are all in rocks of marine origin. Various organisms later invaded the freshwater and land.

9.6 Self Assessment Questions:-

- 1- Theory of abiogenesis was put forward by
 - a- Spallanzani
 - b- Van Helmont

- c- F.Redi
 - d- Pasteur
- Ans- b

2- Who disproved the theory of spontaneous generation

- a- Lazzaro Spallanzani
- b- A.I Oparin
- c- Francesco Redi
- d- Anaximander

Ans- a

3. According to which theory, life came from another planet in the form of celestial bodies and small particles carrying viable germs or spores

- a-cosmozoic theory
 - b-spontaneous generation
 - c-chemical evolution
 - d-synthetic theory
- ans- a

4- Coacervates were formed by

- a- polymerisation and aggregation
 - b- DNA
 - c- replication
 - d- polymerisation
- ans- a

5- life originated in its simplest form in the

- a-land
 - b-sky
 - c-underground
 - d-sea
- ans- d

6..... proved the important evidence in support of chemical synthesis of life.

Ans- Stanley L. Miller

9.7 Suggested Reading:-

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2. Rastogi V.B. (1996). Organic Evolution. Kedar Nath Ram Nath, Meerut, U.P.
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9.8 Terminal Questions:-

1. Give a detailed account of the origin of life.
2. Describe the special creation theories in detail.
3. What is the chemical evolution? Write in brief.
4. Describe the spontaneous origin of life at the molecular level.

9.9 References:-

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UNIT 10: CONCEPT OF ORGANIC EVOLUTION

Contents

- 10.1 Objectives
- 10.2 Introduction
- 10.3 Concept of Organic Evolution
 - 10.3.1 Evidence from Paleontology
 - 10.3.2 Types of Fossils
 - 10.3.3 Determination of Age of Rock and Fossils
- 10.4 Taxonomy
 - 10.4.1 Comparative Anatomy
 - 10.4.2 Comparative Embryology
 - 10.4.3 Physiology
 - 10.4.4 Biochemistry
 - 10.4.5 Cytology
- 10.5 Summary
- 10.6 Self-assessment questions
- 10.7 Suggested Readings

10.1 Objective:-

Understand the causes, Process and Consequences of Evolution and theories of Organic evolution.

10.2 Introduction:-

When evolution was busy to seek a plausible explanation for evolution at that time some worker and researcher were trying to collect facts about the evolutionary process. The evidence is physiology, taxonomy, morphology and embryology of living forms and the fossils of previously existing forms. The recent techniques have been helpful in demonstrating the evolution taking place in the laboratory within a short period of only a few years. If an organism with very short life cycles such as fruit fly or bacteria is reared for several generations in the laboratory, new kind of individuals is observed in the progeny. Initially, these individuals differ slightly from their parents, but as they increase in number, differences keep on accumulating and a stage reached when these become so markedly different from their parents that they fail to interbreed with their parents and thus form new species.

Bio-evolution or organic evolution is continuity of life with constant modifications. It means that the living being modifies and adapt according to the ever-changing environmental needs. These modifications keep accumulating in the organism generation, resulting in more complex and better adapted new species.

Principles of organic evolution

1. The new species are always better adapted than their ancestors.
2. Life is capable of changing with the changing environmental conditions. This is called adaptability.
3. The environmental conditions are ever changing.
4. Evolution is a continuous process.
5. All present species had common ancestors at some time of their evolution.
6. Individuals migrate from their place of origin to varied geographical areas and gradually adapt to different sets of environmental conditions.
7. Evolution is a very complex and extremely slow process.
8. Formation of several new species from one individual species is known as divergent evolution.

The name of Charles Darwin is closely associated with the concept of evolution and for many people, Darwinism is itself evolution. Several theories of historical importance were described by Greek philosophers before the birth of Jesus Christ.

According to Thales (624-528 B.C.) propounded the theory of the aquatic or marine origin of life. He considered sea water to be a mother from which all living creatures were originated.

According to Xenophanes (576-480 B.C.), the existence of fossils of marine animals on dry land indicated that the dry land was once under the sea. He suggests that the earth was once under the sea and life has originated in the sea.

Francis Bacon (1561-1626 B.C.) emphasized on variations as being the cause for the origin of new species from old one. He suggested that flying fishes are intermediate between fishes and birds and the bats between birds and quadrupeds. His work influenced the thinking of the successors.

10.3 Concept of organic evolution:-

The contributions made by Lamarck, Darwin, Cuvier, Weismann and Huxley etc. are of great importance, since these provoked real scientific thinking of evolutionary process and their theories are still being helpful.

1. Lamarck theory of inheritance of acquired characters (1744-1829). Lamarck theory emphasizes the influence of environment on the living being. The changes introduced by the environment are acquired by the living being and are inherited to the next generation.
2. Darwin's theory of natural selection (1809-1882) Darwin formulated the theory of "Origin of species by natural selection."
3. Weismann's theory of the continuity of germplasm- the germplasm produces gametes which transmit the characteristics of the parent into offspring.

The modern synthetic theory of evolution has evolved during the last century through an accumulation of facts, which are theoretical conclusions from a number of scientists: Dobzhansky (1937) in his book "Genetics and the origin of species" has emphasized the role of genetic changes in population in the process of evolution.

At present, the synthetic theory of evolution recognizes five basic processes namely gene mutation, changes in chromosomes numbers, genetic recombination, natural selection and reproductive isolation. The three accessory processes also affect the working of the process. These are migration, hybridization, and chance in small populations.

10.3.1 Evidence from Paleontology:-

The paleontology is the study of the ancient life of past geologic ages and it is based on the remains of extinct animals and plant including traces of their existence such as footprints and impressions. Such remains have been found in rocks, mud and snow or in soft sediments are called as fossils. The fossils are the direct evidence of descendants leading stepwise from an ancestor to a descendant species. The evidences in support of evolution so far are of circumstantial nature, but the direct evidence comes from the study of fossils. The fossil means something dug out. Any imprint left by some previous organism in the soft mud, which subsequently hardened or the moulds and cast of entire organisms of the soft animal preserved in some other way manner. Mostly fossils are found in the sedimentary rocks which are formed by the deposition of sand or debris in the bottom of rivers, ponds, lakes and sea. Entire bodies or part of the dead organism become covered by sand deposits. After their burial, most animals rot away without leaving any sign of their existence. Slowly over the centuries, the material of the hard part is replaced molecules by molecules with mineral matter from the surrounding mud. The replacement is sometimes so accurate that even the cellular details can be studied accurately. Within thousand and million years, these layers of mud shrink and harden into rocks.

Definition of fossils

Fossils are only animals and plant which have been dead rather longer than those which dead yesterday (T.H. Huxley).

Significance of fossils

The study of fossils reveals the existence of life in past and illustrate the course of evolution of plants and animals. The fossils records establish the following facts:

1. Fossils are remains of an organism that lived in past. Fossils from different geological strata belong to different genera.
2. The fossils from bottom layers of rock have the simplest organization and become more or more complex in strata lying above them. The simpler forms could match with the unspecialized member of the living phylum.
3. The transitional fossils forms are known between different existing groups from fishes to amphibians, from amphibian to reptiles and from reptiles to birds and mammals.
4. The dominant groups of fossils arose near the close of existing period when great climatic changes were taking place. The groups enjoyed dominance in the next period because of the favorable environmental condition and finally perished by the end of the

period on account of alteration on the climate. These become replaced by some new forms more suited to the changed conditions.

5. The mammals among animals and angiosperms among plants are the most recent products of evolution. Geologist has prepared a time- table which helps in depicting the distribution of animals and plants to geological time. It has been divided into five eras, which are further differentiated into periods. The study of these eras and period depicts the story of the evolution of living beings on earth. It has been estimated that life appeared on this earth about 2700 million years ago. Some names of the era are as: Archeozoic era, Proterozoic era, Palaeozoic era, Mesozoic era and Coenozoic era.

The Mesozoic era is described as the age of reptiles. Mammals and birds diverged from reptiles in Triassic and Jurassic periods respectively. The coenozoic era is the age of mammals.

10.3.2. Types of Fossils

The different types of fossils are can be arranged under the following heads.

1. Actual remains

The recently extinct animals and plants which have been buried by some sort of preserving material constitute the first type under consideration. Such remains have undergone little or no change of the original organic matter into inorganic. Thus we find the complete bodies of great hairy mammoths frozen in the arctic ice. These are so well preserved that dogs have fed upon their flesh. Nearly a thousand species of extinct insects, including many ants, have been obtained practically intact from amber, a form of petrified resin. Innumerable mollusc shells, teeth of sharks, pieces of buried logs, bones of animals buried in asphalt lakes and bogs, have been found in a well-preserved condition.

2. Petrified fossils

The process of petrification involves the replacement, particle for particle, of the organic matter of a dead animals or plant by mineral matter. So, the finer structure is completely preserved that microscopic sections of preserved tissue, especially of the plant, have practically the same appearance as section made from living organisms. Various minerals have been employed in petrification, such as quartz, limestone or iron pyrites.

3. Cast, Moulds and Impression

When the animals and plants are embedded in the hard material, their entire body material may be decayed and dissolved away by the water current under the strata and sometimes percolate by sand or something which become accumulated in that rock moulds. Such moulds and casts have been reported in the rocks of Cambrian age. Only external appearance has been

preserved, as would be the case in making plaster of paris casts. Sometimes tracers of soft-bodied animals have been left upon forming slate or coal that is almost as accurate in detail as a lithograph.

Most remarkable fossils are those found by Professor Charles D. Walcott in the marine oily shale's of British Columbia. A large number of soft-bodied invertebrates of Cambrian age have been found so wonderfully preserved that not only are the external features revealed but sometimes even the details of the internal organ may be seen through the transparent integument.

4. Trails and footprints

Sometimes animals that passed over sand or mud left their foot- print in the sediments and after that when preserved became the fossils in the form of the footprint. Sometimes animals and worm, mollusk left their movement line and formed their trails.

5. Coprolites

When the food particles in the food tract or excretory material become fossilized and are named as coprolites.

Most paleontologists have been able to reconstruct the appearance of extinct animals and also able to from the history of the development of the earth and its life in the form of geological record. They believe that many fossils are quite different from the forms found today and indicate that evolution has taken place. It is also possible, in many cases, to arrange the fossils in a serial order which proves that evolution has taken place through the series. The fossil series of the horse is quite an example of this type. Also, the paleontological history of camel, elephant, and Man are regarded as evidence of evolution.

10.3.3Determination of Age of Rock and Fossils:-

The fossils can be dated in several ways. Carbon 14 method is the most popular method. The stable form of carbon and therefore, the carbon that is most frequently cycled through food chains and webs, is carbon 12. However, part of the carbon that is cycled is carbon14; an unsuitable isotope gives off beta particles which make it radioactive because of its instability, Carbon 14is slowly converted, that is decayed to the next possible stable form, which is nitrogen 14.

The rate of change for Carbon 14 is precisely known as it is exactly 5730 years for half of a specific amount of Carbon 14 to be converted to N. thus the half-life for Carbon 14 is 5730 years. For example, if you only 2 micrograms of carbon 14 in 5730 years you would have only one microgram, in 11460 years you would have 0.5 micrograms.. The ratio of C^{12} to C^{14} in a specific tissue, such as bone, is known for animals alive today. By biochemical assay, the ratio of

C^{12} to C^{14} is measured in fossils as the amount of beta particle emission; the result can be converted into years of age. However, when the specimen being dated is much more than 50,000 years old, the technique loses its accuracy as the original C^{14} is left.

The fission backdating is a new method, like uranium 238 in reverse. If Uranium-238 is placed in an atomic reactor, the explosions that accompany its decay, a series of the etching in the glass can be observed with a microscope. The same process happens naturally in inactive volcanoes. Glass specimens taken from the volcanic sediments show fission tracks that can be counted. The specimen is then placed in an atomic reactor so that the remaining Uranium 238 is used. During its decay a new set of etching is created. The total number of etch lines is proportional to the original amount of Uranium 238 and therefore proportional to the age.

Above mentioned methods of fossil dating are used singly or in any combination, allows paleontologists to determine the age of fossils remains and rocks with reasonable accuracy. Of course, these methods are not perfect, and degree of error is expected. However, an error in a few thousand years or even tens of thousands of years is negligible when considering fossils that are several million years old.

10.4 Taxonomy:-

One of the tenets of the theory of evolution is that all the diverse plant and animal varieties that we observe today have evolved from common ancestral stock. The theory of organic evolution appears most plausible explanation for the occurrence of varied forms of plants and animals on this earth. But the absolute proof in this connection is lacking. If we go through the classification of the organism, we find there is a strong connection (family tree) among the animals as well as plants evolution.

10.4.1 Comparative Anatomy

Morphological studies of various organ systems of vertebrates indicate that these are constructed on the same basic plan. The minor differences seen in some forms are the adaptive modifications to the diverse mode of living. These similarities are known as homology.

I.) Homology and homologous organs

Homology is the similarity between organs of different animals based on common ancestry. Therefore, the homologous organs have a common origin and are built on the same fundamental pattern, but perform varied functions and have a different appearance. Homology is seen in every organ system from fish to man.

1. Homology in limb structure of vertebrates.

The flipper of a seal, wing of a bat, forelimb of a mole, front leg of the horse and the arm of a man look very different and perform different functions, but exhibit the same structural plan. The modification includes shortening or lengthening of bones, variation in shape, reduction in the number of bones or fusion of bones in accordance with the function.

Forelimbs of various animals become evidence that these vertebrates must have had a common ancestor with a prototype of the forelimb.

2. Homology in brain structure

Ranging from fishes to mammals, the brain consists of similar series of parts as olfactory lobes, cerebral hemispheres, optic lobes, cerebellum, and medulla oblongata.

As we progress through the series from fishes to mammals some lobes present gradual enlargement. In fishes, the cerebral hemispheres are even smaller than the optic lobes, but in mammals, there are so much enlarged that they hide the olfactory lobes in front and the optic lobes behind.

3. Homology in the structure of heart

The heart is two-chambered in fishes, consisting of one auricle and one ventricle. The auricle receives blood from entire body and ventricle pumps it to the gills. In amphibians and lower reptiles, the heart is three chambered. There are two auricles and one ventricle. The oxygenated blood from lungs is collected in the left auricle and deoxygenated blood from rest of the body in the right auricle. Thus the oxygenated and deoxygenated blood is stored separately. But it gets mixed in the ventricle while being pumped to the body organs. In higher reptiles, birds and mammals heart are four chambered and the oxygenated and deoxygenated blood is completely separated. This represents a gradual modification in the heart of vertebrate series while the fundamental structure of heart remains same in all the groups.

II.) Serial homology

Several homologies have been observed among invertebrates. All the arthropods have segmented body with an exoskeleton of chitin. The exoskeleton is constructed on the same basic pattern of classes of phylum arthropods. In crustaceans, all the segments of body carry paired jointed appendages. All of them are constructed on a common structural plan, consisting of a basal two segmented portion, the protopodite (coxa and basis), which bears two lateral outgrowths, the exopodite and endopodite. The appendages of various body segments perform different functions and in correlation with that exhibit modification of the basic structural plan. This phenomenon of similarity has been described as serial homology.

III.) Analogy and analogous structure

The analogous organ has the almost similar appearance and performs the same function but these develop in totally different groups on the totally different pattern. For example, the wing of a butterfly, bird, pterodactyls and bat serve the same purpose of uplifting the body in the air, but their basic structure is totally different. The wing of insect is formed of a thin flap of chitin and stiffened by a series of veins. It is operated by muscles attached to its base. In pterodactyl, the wing is an enormous fold of skin supported by an enormously enlarged fourth finger of the forelimb. In the bird, the flight surface is formed by feather attached to the bones of the forelimb. In bat, the wing is formed by a fold of integument (patagium), supported by the elongated and outspread phalanges of last four digits. Similarly, the fins of fishes and Ichthyosaur and the flippers of whale have similar appearance and function but their structure details are totally different. These functional similarities between analogous organ support occurrence of organic evolution.

IV.) Adaptive radiation

The concept of adaptive radiation also provides strong evidence in support of the theory of organic evolution. The adaptive radiation is exhibited by the limb structure in mammals. The limbs in mammals are variously adapted for climbing, flying, running, swimming or burrowing etc. Naturally, these exhibit structural modifications correlated with their mode of working. In the arboreal or tree dwelling forms like sloths and monkeys, limbs are modified for having a powerful grip of the branches. Mammals adapted for flight have their forelimb modified into wings. In aquatic mammals, the limbs get modified into flippers. The size of limb bones is much reduced. The forelimbs of fossorial mammals are modified for digging burrows with short and strong limb bones. In cursorial forms like horses, the limbs are suited for fast running over hard ground.

All the aforesaid limb structures are constructed on the same fundamental pattern and can be derived from the prototype, pentadactyl limb structure. In other words, it could be said that all of them represent evolutionary lines radiating out in various directions from the prototype limb structure. This is known as adaptive radiation which represents the evolution of new forms in several directions from the common ancestral type.

V.) Convergent Evolution or adaptive convergence or parallel evolution

The whale and their relatives, the extinct reptiles and Ichthyosaurs attained fish-like body with their limbs modified into fins or flippers. The similarities are so marked that whale is understood as a fish by laymen. This similar body shape between animals of distantly related groups represents the phenomenon of convergent evolution.

VI.) Vestigial organs or Vestiges

The vestigial or rudimentary organs are the useless remnants of structure or organ which might have been large and functional in the ancestors. These are undersized, degenerate and nonfunctional.

1. Vestigial organs in man:

- a.) Vermiform appendix in man
- b.) Muscles of external ear
- c.) Nictitating membrane
- d.) Vestigial tail vertebrae
- e.) Wisdom teeth

2. Vestigial organ in other animals

- a.) Both whales and pythons have vestiges of bones of hind limbs and pelvic girdle embedded in the flesh of abdomen.
- b.) `Kiwi possesses vestiges of wing supported by tiny replicas of usual bones of bird's wing.
- c.) in horse leg, the splint bones represent the metacarpals of second and fourth digits.
- d.) In the animals living permanently in deep caves, the eyes are rudimentary.

VII.) Evidence from atavism or reversion

Atavism is the reappearance of those ancestral characteristics in an organism of a group; that do not occur normally by the individuals of that group. Such abnormal structure is known as atavistic characters of reversion or atavism. In such cases, abnormal characters appear in the embryo or in adult, which were not present either in the parent or grandparents but in some remote ancestors.

10.4.2 Comparative Embryology:-

Ontogeny is the life history of the individual starting from ovum and phylogeny is the series of adult ancestors of the individual which must have incurred in the evolution of the group of this individual. It means that an individual during its development briefs its ancestral history.

1. Homology in early development

The entire multicellular organism exhibits a common pattern of development. Their development starts from unicellular fertilized egg or zygote. The fertilized egg after repeated cell divisions forms blastula, which finally develops into a two layered gastrula. The outer layer of gastrula represents future ectoderm and inner one future endoderm. The cavity lined by endoderm forms the archenteron, the future digestive tract. The development after gastrula stage becomes modified in different groups of animals.

2. Recapitulation in Human Embryo

The development of man can be taken as an example to illustrate the theory of recapitulation. The fertilized egg may be compared to the single-celled ancestor of all the animals and the blastula to a colonial protozoan of some spherical multicellular form from which might have been the ancestor of all the metazoan. Gastrula represents the coelenterate ancestor and the embryo with the development of mesoderm represents triploblastic stage like a flatworm.

3. Homology in the Embryo

The early embryo in all the vertebrates exhibit remarkable similarity and it is not easy to differentiate a human embryo from the embryo of chick, lizard, frog or fish in early stages. It has also observed that the early embryos of all the individuals are much alike, later those of different classes become recognizable and still later family and species characters become evident i.e. the embryos during their development become progressively more and more different from those of the other animals.

4. Retrogressive metamorphosis

The ascidians tadpole is free swimming and possesses all their chordate characters. On metamorphosis, it changes into sedentary degenerated adult. During metamorphosis, it loses all the chordate features, like notochord, nerve cord and myotomes. This is called retrogressive metamorphosis; it has helped in determining its chordate nature.

5. Neoteny

In some animals, the larva fails to undergo metamorphosis. It develops gonads; attain sexual maturity and starts reproduction. This is called neoteny or paedogenesis e.g. axolotl larva of *Ambystoma*.

10.4.3 Physiology:-

When the physiological processes and the chemical composition of various cells and tissues are considered it is found that there are at least some similarities in different animals showing the relationship between them. This indicates that they have descended from a common ancestor. The physiology of heart, kidney and gonads also show similarities among most of the vertebrates. These similarities explain the idea to common ancestry.

10.4.4 Biochemistry:-

The protoplasm of different organisms is considered it prove to be basically the same. The nucleic acid found in the cells is also similar in all organisms. In the same way, very similar enzymes and hormones are found in many animals. The parts of the central nervous system perform, so far they have been tested approximately the same functions. The chemical reactions involved in the process of respiration are essentially similar in most diverse organisms. Most animals can readily oxidize uric acid to allantoin which is more soluble and as eliminated from the body along with urine, man is unable to do so and, as result, is liable to gout. Serological evidence also proves the remarkable kinship between different animals as shown by Dr. Nuttall. Animals, which on other grounds are closely related, have been found to possess similar blood. Thus it has been experimentally proved that the blood of a horse and ass is similar, so is that of rabbit, hare, man and the anthropoid apes. Serological tests provide a method of measuring the degree of relationship among different animals, and are helpful in establishing their affinities. Chromosomes: it is an essential component of the nucleus in every living cell.. The chromosomes have a fairly constant chemical composition in the living animals being composed of DNA and proteins. The basic unit of DNA is a nucleotide consisting of a molecule of phosphoric acid, one molecule of pentose sugar is deoxyribose and a purine or a pyrimidine as a nitrogenous base. The chemical composition of DNA is basically the same in all living beings except for differences in the sequences of nitrogenous bases. How can such diverse organisms have the same basic fundamental composition? It means all have gradually evolved from some common ancestor.

The hemoglobin is conjugated protein. It is formed of two identical alpha chains and two identical beta chains. Each alpha chain has 141 amino acids and each beta chain has 146 amino acids. B chain of hemoglobin of human and gorilla differ in one amino acid, of human and pig in ten amino acids and of human and horse in 26 amino acids.

The cytochrome is present in all eukaryotic cells. It forms a part of the electron transport system and in all eukaryotes accepts an electron from H^+ ions. It is formed of 104 amino acids.

Insulin: beef insulin is so similar to human insulin that it has been used for the treatment of human diabetes. Even human immune system fails to detect the difference.

10.4.5 Cytology:-

The strong evidence in favor of organic evolution comes from genetics. Source of such cases are hybridization and domestication etc. for example, the mule is hybrid. Mule is the offspring of a jackass (*Equus caballus*). This is an evolutionary dead end and with very rare exceptions, the mule is sterile. But the mule is very strong and hardy. The mule has a different chromosome number and normal gamete formation is prevented. It is clear that these animals can be traced back to a common ancestry and their genetic material are still sufficiently similar but during the course of evolution their chromosomes and genes have diverged so much that they are no longer so similar to allow normal gamete formation.

On the basis of chromosome material, the animals show the relationship among themselves. The chromosomes are chemically composed of nucleoprotein, a combination of protein and nucleic acid. Two kinds of nucleic acid have been found in all species, they are DNA (deoxyribonucleic acid) and RNA (ribonucleic acid). DNA is found to be in the nucleus of cells, while RNA may be found in nucleus and cytoplasm both. In all the cases, except for plant viruses, DNA is the hereditary material while RNA mediates the protein synthesis. Human DNA differs in only 1.8% of its base pairs from chimpanzee DNA and there are no differences between the two in the amino acid sequences for the protein cytochrome C.

10.5 Summary:-

One of the tenets of the theory of evolution is that all the diverse plants and animals' variety that we observe today have evolved from a common ancestral stock. As noted already, life is thought to have arisen on this planet by chance under a special set of physical and chemical conditions that existed in the early stages of the formation of the earth. From this 'early protoplasm', all varieties of organisms evolved gradually by natural selection. If this theory is correct, many basic life processes – release of energy, synthesis of ATP, transfer of genetic information, and so on – should be similar in all organisms. The principle of common ancestry of all organisms is the chemical basis of information transfer. The chemical basis of heredity, DNA was noted as the carrier of heredity in all prokaryotes and eukaryotes, that is, in all organisms on earth. Furthermore, the information encoded in DNA is transcribed in the form of RNA molecules and then translated into the amino acid sequence of proteins. The genetic code that specifies the sequence of amino acids in a polypeptide chain is also identical in all organisms except for some variations in that of mitochondria and chloroplast. Moreover, the enzymes involved in the transfer of this information are similar in all organisms.

10.6 Self Assessment Questions:-

- 1- Evolution is a
- a- continuous process
 - b- discontinuous process
 - c- both
 - d- none of above

Ans- a

- 2- The author of book “Genetics and the origin of species” is

- a- Darwin
- b- Lamark
- c- Cuvier
- d- Dobzhansky

Ans- d

- 3- The half-life of carbon 14 is

- a- 5,730 years.
- b- 6700 years
- c- 2330 years
- d- 8930 years

Ans- a

- 4- Homology is the similarity between organs of different animals based on

Ans- common ancestry

- 5- Which is not a vestigial organs in man:

- a. Vermiform appendix in man
- b. Muscles of external ear
- c. Nictitating membrane
- d. Molar teeth

Ans- d

10.7 Suggested Reading:-

- 1. Colbert, E.H. (1958). Evolution of the vertebrates. Wiley, New York.
- 2. Rastogi, V.B. (1996). Organic Evolution. Kedar Nath Ram Nath, Meerut, U.P.
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10.8 Terminal Questions:-

1. Give a detailed account of organic evolution.
2. Describe the evidence of organic evolution from comparative morphology and comparative anatomy in detail.
3. Give an account of evidence from comparative anatomy supporting the theory of evolution.
4. Describe the biochemical origin of life at the molecular level.
5. What is neoteny?
6. Write short notes on retrogressive metamorphosis.
7. Write an essay on embryological evidence of organic evolution.
8. Give an account of the paleontological evidence of organic evolution.

10.9 References:-

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UNIT 11: THEORY OF ORGANIC EVOLUTION

Contents:-

11.1 Objectives

11.2 Introduction

11.3 Theory of Organic Evolution

11.3.1 Lamarckism

11.3.2 Darwinism

11.3.3 Modern Synthetic Theory or Neo-Darwinism

11.3.4 Mutation Theory

11.4 Summary

11.5 Self-assessment questions

11.6 Terminal Questions

11.1 Objectives:-

Theory of organic evolution, i.e. Lamarckism, Darwinism and the study of Mutation theory and Modern synthetic theory.

11.2 Introduction:-

Lamarck (1744-1829) was a greatest French naturalist who “founded the modern theory of descent”. It was Osborn, who took great pain in the predictions of Lamarck and the former considered the latter “the most prominent figure between Aristotle and Darwin”. At the age of fifty, he became a Professor of Invertebrate Zoology at which he worked so hard that he was considered as a leading authority in that field. Lamarck was the first biologist to postulate a theory of evolution in his famous treatise “Philosophic Zoologique”, in the year 1809. He found that (i) species change under changing external influences, (ii) there is a basic or fundamental unity underlying the diversity of species and (iii) the species are subjected to progressive development.

11.3 Theory of Organic Evolution:-

11.3.1 Lamarckism:-

Lamarckian theory of evolution can be most specifically explained in the following factors which he considered important in evolution.

1. Favorable changes in the environment, soil, food and temperature etc. influence directly the life of plants while indirectly in the case of animals and human being.
2. According to needs new organs originate or modify thus, leading to the appearance of new organs during the life of individuals subjected to the environmental changes.
3. Use and disuse: the proper use of an organ establishes it while no use makes its eventually lost. For example, anterior limbs of birds became capable of sustained flight through use, while hind limbs of whales are lost due to their disuse.

4. Competition: Nature itself balances the number of living things avoiding overcrowding by competition among animals, stronger destroy the weaker. So the smaller multiply rapidly, while larger less rapidly.

5. Acquired characters are transmitted: The advantageous changes resulting from use and disuse and needs are handed down to descending generation thus transmitting the acquired characters attained by parents to offspring.

6. Cross-breeding: The individuals acquiring any peculiar character or defect, on pairing, produce the same character in the offspring. But perpetual crosses between individuals, which have not the same peculiarities of form, result in the disappearance of all the peculiarities acquired in the particular circumstances.

7. Isolation: Animals diversified due to separation by distance. Lamarck expressed this thought in his account of the origin of man from ape and is not applicable to the living thing in general.

Analysis of Lamarck's theory

1st law: Lamarckian first law simply emphasizes on the growth and progress of organisms and the size increases truly due to metabolic activities, though controlled by vital factors. So the first law is accepted.

2nd law: this law is not acceptable because one cannot develop any organ if he or she needs it. It is unbelievable that if we want our eyes on the back of our head just or see behind of us, they can be developed. So, need of any organ does not develop that organ.

3rd law: the third law says that development of organs depends on the use or disuse of that organ. He suggested that variations appear among animals mainly through use or disuse of parts brought about as a result of conscious effort on the part of animals in response to various external stimuli. Such variations according to Lamarck are heritable, being passed on to the offspring during sexual reproduction. A repetition of such efforts by successive generations of offspring under similar, environment conditions will result in the production of new characteristic and thus new species.

According to Lamarck, animals and plants are affected by the environment in which they are living. This can be illustrated by the example of a plant- *Ranunculus aquatilis* commonly called water crowfoot. This plant grows partly immersed water on banks of the streaming water. The part of plant submerged develops filamentous prickles in place of leaves, and the other above water develops rounded and lobed leaves. When this plant grew away from water, it develops leaves on the part that was submerged, but when returned to the water it again develops prickles.

Lamarck gives a number of examples, the most famous amongst them are those of the giraffe and snake.

(i) Giraffe: according to Lamarck the ancestral giraffe was a short-necked form browsing on ground vegetation. When falling in grass or herbage came and they were forced to depend upon the foliage of trees, they had to stretch their neck to reach the higher vegetation. This resulted in a short increase in the neck length and it was transmitted to the next generation. Thus, repeated conscious effort by successive generations to feed on the leaves of trees which grew progressively taller and taller resulted in a continuous increase in the length of the neck eventually leading to the modern long-necked giraffe.

(ii) Snakes: the snakes have been evolved from lizard-like ancestors which were having two pairs of limbs. These ancestors of snakes felt insecure from mammals of that time because the latter were more powerful and enormous in number. To escape from the mammals, the ancestors of the snakes started living in narrow holes or crevices and in thick jungles. To accommodate their body in narrow spaces they could not use their limb that is why the limbs were reduced and finally disappeared, while their body became longer and cylindrical.

(iii) Aquatic birds: Aquatic birds like ducks have been evolved from the terrestrial ancestors. Since they had to go to water due to lack of food. Some structures like a web between the toes developed in them, so that they could live in water easily. The wings were not used for the flying as they were not needed, and later they got reduced.

(iv) Flat fishes. They are flat and bear both the eyes on one side and live at the bottom of the water. During the embryonic stage, their eyes are present laterally, one eye on either side. The body of these fishes is not flat at this stage but later on, both the eyes are shifted to one side and the body becomes flat to withstand the pressure of water.

(v) Flightless birds. The ancestor of these birds was capable to flying,, but due to some environmental factors they had plenty of food and were well protected. So they did not use their wings and that is why the latter become vestigial.

(vi) Deer. The ancestor of deer did not have so much speed in running, but as they needed protection from other animals so they started running due to which present speed was achieved by the deer and consequently their limbs got developed and the body became streamlined.

(vii) Claws of carnivorous mammals. The lions, cats and dogs are well adapted to their carnivorous habits. The claws of these mammals are retractile because they run very fast.

(viii) Skin of man. The use and disuse of organs are best illustrated during the development of man. The skin of the palm and the sole of the feet are generally smooth and velvety in touch at the time of birth, due to constant use the skin gradually becomes thick and rough in the course of time. The presence of vestigial organs in various animals is also explainable on the basis of the use and disuse theory.

The fourth law of Lamarckism doctrine is of many controversies. According to Lamarck, the characters attained during the lifetime of an animal are heritable, which gradually led to evolutionary changes. In other words, the environment acted as the motivating force, which brought about changes in the organism constituting the first step in evolution. This was considered by Lamarck, as “principle of inheritance of acquired characters.” Herbert-Spencer also supported this theory with the example of fish- *Gasterosteus*. These fishes inhabit freshwater, brackish water, and sea. The marine fishes possess 20-30 bony plates on the mid-dorsal line of the trunk, the brackish water forms 3-15 and freshwater possess none of these plates. If a marine species is transferred to fresh water, it gradually tends to lose all the plates and this acquired character is inherited by its offspring. Similarly, if a freshwater form is transferred to a marine environment, in due course of time, it tends to develop the bony plates and this acquired character is transmitted to its progeny. However, many objections were given against Lamarckism. The greatest blow to Lamarckism came from August Weismann.

According to Weismann, the body of an organism consists of somatic cells and the germinal cells. Any change affecting the soma is not heritable and it disappears with the death of the individual. Weismann showed that if we cut the tail of white mice during its lifetime, then the offspring of these rats do possess tail and do not show the absence of the tail indicating that the character is not transmitted. He explained for 22 generation, every time cutting the tails of parental generation and recording the presence of the tail in the progeny. He found not even a single mice developed even a reduce tail.

Pavlov, during the experiments, trained the dog to takes its food on hearing the bell. But this character was not transmitted to the offspring. Instead every dog, in order to develop this character, was needed to be trained similarly. So, this clarifies that the acquired character are not inherited.

11.3.2 Darwinism:-

Charles Darwin was a methodical painstaking English naturalist and he was the first who founded the theory of organic evolution. He was born on February 12, 1809, at Shrewsbury. He was educated at Shrewsbury. The career of Charles Darwin began with his voyage of H.M.S Beagle (December 27, 1831, to October 2, 1836) as the ship’s naturalist. He visited Cape Verde and other Atlantic Islands, New Zealand, Australia, Tasmania, Mauritius and Brazil. He spent about five weeks among the Galapagos Islands and about five years in the voyage. He was very much impressed by the remote animal life.

Charles Darwin married his cousin Emma Wedgwood in 1839. He was blessed with two daughters and five sons. He was very kind to his family, friends and fellow scientists. He was

acknowledged as Great before his death. He died on April 19, 1882 and buried in Westminster Abbey next to Sir Issac Newton.

Evolution before Darwin

The French scientist Georges-Louis Leclerc Comte de Buffon (1707-1788) was among the first to suggest that species undergo changes within the course of time. Buffon believed that these changes took place by the process of degeneration. He suggested that, in addition to the numerous creatures produced by divine creation at the beginning of the world, “there are lesser families conceived by Nature and produced by Time.” Buffon’s hypothesis, although vague as to the way in which changes might occur, did attempt to explain the bewildering variety of creatures in the modern world.

Another early doubter of fixed unchanging species was Erasmus Darwin (1731-1802), Charles Darwin’s grandfather. Erasmus Darwin was a physician, a gentleman naturalist, and a prolific writer, often in verse, on both botany and zoology. He suggested, largely in asides and footnotes, that species have historical connections with one another, that animals may change in response to their environment, and that their offspring may inherit these changes. He maintained, for instance, that a polar bear is an “ordinary” bear that, by living in the Arctic, became modified and passed the modifications along to its cubs. These ideas were never clearly formulated but are interesting because of their possible effects on Charles Darwin, although the latter, born after his grandfather died, did not profess to hold his grandfather’s view in high esteem.

The Earth has a History:-

The person who most influenced Darwin as Charles Lyell (1797-1875), a geologist who was Darwin’s senior by 12 years. One of the books Darwin took with him on his voyage as the first volume of Lyell’s newly published *Principles of Geology* and the second volume was sent to him when he was on the *Beagle*. On the basis of his own observations and those of his predecessors, Lyell opposed the theory of catastrophes. Instead, he produced new evidence in support of Hutton’s early theory of uniformitarianism. According to Lyell, the slow, steady and cumulative effect of natural forces had produced a continuous change in the course of the earth’s history. Since this process is demonstrably slow, its result being barely visible in a single lifetime, it must have been going on for a long time. If the earth had a long continuous history and if no forces other than well known, natural forces were needed to explain the events as they were recorded in the geologic record, might not living organisms have a similar history?

The voyage of the Beagle:-

This was the intellectual equipment with which Charles Darwin set sail from England. As the beagle moved down the Atlantic coast of South America, through the Straits of Magellan, and up the Pacific coast, Darwin traveled the interior, fished, hunted and rode horseback. He explored the rich fossil beds of South America and collected specimens of the many new kinds of plants and animal life he encountered. He was impressed mostly during his long, slow trip down the coast and up again by the constantly changing varieties of organisms he saw. The birds and animals on the west, for example, were very different than those on the east coast, and even as he moved slowly up the western coast, one species would give way to another.

Most interesting to Darwin were the animals and plants that inhabited a small, barren group of islands, the Galapagos, which lie some 950km off the Eastern coast of Ecuador. The Galapagos were named after the islands most striking inhabitants, the tortoises, some of which weigh 100kg or more. Each island has its own type of tortoise; sailors who took tortoises on board as a conventional source of fresh meat on their sea voyages could readily tell which island any particular tortoise had come from. Then there was a group of finch-like birds, 13 species in all that differed from one another in sizes and shape of their bodies and beaks, and particularly in the type of food they ate. In fact, although clearly finches, they had many characteristics seen only in completely different types of birds on the mainland. One finch, for example, feeds by routing insects out of the bark of a tree. It is not fully equipped for this, however, lacking the long tongue with which the woodpecker flicks out insects from under the bark. Instead, the woodpecker finch uses a small stick or cactus spine to pry out the insect loose.

From his knowledge of geology, Darwin knew that these islands, clearly of volcanic origin were much younger than the mainland. Yet the animals and plants of the island were different from those of mainland, and in fact, the inhabitants of different islands in the archipelago differed from one another. Were the living things on each island the product of a separate special creation? “One might really fancy”, Darwin mused at a later date, “that from an original paucity of birds in this archipelago one species had been taken and modified for different ends.” After his return, this problem continued to, in his own words, “haunt” him.

In 1858 Charles Darwin and Alfred Wallace had a paper presented to the Linnaean Society of London in which they used the term evolution to describe the progressive changes in successive generations of living organisms. The theory of evolution is an attractive one for it helps explain two things.

- a) The similarities between related organisms, as being due to their descent from a common ancestor and

- b) The differences between them as being the result of variation inherited from one generation to next.

The theory of evolution by natural selection announced jointly by Darwin and Wallace in 1858, made little impact on the world of science until the publications of Darwin's books "The Origin of Species by Natural Selection" in 1859 in which Darwin expounded the theory. The entire edition was sold out on the day of publication, November 24, 1859. He prevented possible mode of transformation of species natural causes as opposed to the doctrine of the special creation of all the species.

The Data Suggestion Evolution:-

Darwin had a little scientific training when he joined the beagle. The trip itself was his education. It provides him with the facts and experiences that eventually led to the formulation of his theory of evolution. In later years, Darwin recalled those observations that made him question the contemporary theological view that each and every species had been created by the divine power. It is of considerable importance for us to review the data which led to such a view, for evolution is a most abstract idea; one does not look at nature and suspect that evolution has occurred.

These were the observations that made Darwin wonder:

1. The relation of fossil to living species:-

While he was in South America, Darwin collected the remains of stone giant fossil mammals that were covered with armor similar to that of a living armadillo. There are two important features of this observation: First, the living armadillos that Darwin observed on the pampas were obviously different from members of the fossil species. In spite of these differences, the two were clearly of the same general animal type that is both were 'armadillos'. Second, the living and the fossil armadillos occur only in the Western Hemisphere.

The question that ran through Darwin's mind was something probably like this, "Where the two forms, the extinct and the living, created separately, or could the extinct species have been a progenitor of the living? If on the other hand, the two represented separate and independent creations, was it not surprising that two such similar forms should have been created in precisely the same part of the world? He concludes, 'The wonderful relationship in the same continent between the dead and the living will, I do not doubt, hereafter throw more light on the appearance of the organic beings on our earth, and their disappearance from it, than any other class of facts'.

2. Geographical succession of allied species:-

The Beagle made frequent stops along the east coast of South America between central Brazil and southern Argentina. Darwin noticed that in any one locality the individuals of a given species would be identical or nearly so. At the next stopping place of the Beagle, which might be several hundred miles distant, the individuals of this same species would appear homogenous among themselves, yet differ slightly from those in the first locality. With increasing distance between the localities, the divergence in character might be considerable. Darwin observed this type of phenomenon in enough species to be convinced that it was a general rule. Should one conclude that not only was each species created separately but that many slight versions were created, one for each locality?

3. Geographical variation of animals and plants in the “Galapagos Islands”:-

The Galapagos Islands are situated in the Pacific Ocean about 600 miles west of Ecuador. They are volcanic in origin, and in Darwin’s opinion, they were not of great antiquity. For years, the principal visitors were buccaneers and Whalers who came to fill their casks with fresh water and their larders with the giant tortoises. Somewhat more than a dozen islands comprise the archipelago. The main islands of the group are close to one another, each being separated by not more than 30 miles from its nearest neighbor. From these islands, Darwin collected both animals and plants, and when these were studied a number of interesting facts emerged. First, the majority of species that he collected were new to science. Second, most of the species are found only in the Galapagos Islands. Third, although the majority of the species are peculiar to the islands, they are obviously similar to forms inhabiting the American mainland. Fourth, in many instances, a species would be restricted to one island.

To these facts should be added the fifth and most surprising observation of all: namely, Darwin noticed that frequently each island would have its own species of an animal or plant type. Thus, the giant tortoise was found on all the islands, but each island had its own tortoise population that differed slightly from that of every other island. One of the local officials told Darwin ‘that the tortoise differed from the different islands and that he could with certainty tell from which island any one was brought’. Apropos of this, Darwin wrote: I never dreamed that islands, about fifty or sixty miles apart, and most of them in one sight of each other, formed of precisely the same rocks, placed under a climate, rising to a nearly equal height, would be differently tenanted; Not only was this true for tortoises, but for many other animals, and plants also. Thus the plant genus *Scalesia* is restricted to the Galapagos. Six species were found and each was restricted to a single island.

Most Galapagos Islands finches eat seeds they gather from the ground. During times of food shortage, there is severe competition for seeds and many birds die. Birds with different sized bills are most efficient at husking different sized seeds and birds with small bills cannot crack large, hard seeds. The suggestion that competition for food has influenced the evolution of

bill size is supported by differences in bill sizes between populations of finches with and without potential competitors (a) *Geospiza fuliginosa* and *G. fortis* are both small ground feeding finches. (b) When either one is the only finch present on the island, it has a bill size similar to that of the other species (c) When they occur together, however *G. fortis* has larger, and *G. fuliginosa* has smaller bills than either has when it lives alone.

Individuals of *G. fortis* have average or even larger bills on an island lacking the more robust ground finch, *G. magnirostris*.

Some of these differences are probably caused by differences in the kinds of seeds available on the various islands, but competition clearly contributes to the sizes of the bills of these finches. The work of Darwin can be summarized under the following heads:

1. Tendency to rapid increase in number

Organisms produce far more offsprings than those whomever reach maturity. This is the tendency of all living organisms to multiply their number rapidly. For example, one pair of common house flies breeding in April would have by August, if all eggs hatched and all resulting individual lived to reproduce in their turn, 191,010,000,000,000,000,000 descendants. In the case of *Drosophila*, each female lays`200 eggs and the fly completes the life cycle in 10-14 days. Therefore, if the production goes as such, in 40 days there will be about 200,000,000 flies. Another example is taken from *Paramecium*. It multiplies at the rate of 3000 generations in five years, if all the descendants existed, their protoplasm would approximately equal to 10 times the volume of the earth. Similarly, one mosquito may have two hundred billion descendants in one summer. An oyster lays about 1, 14,000,000 eggs in a single spawning. A single *Ascaris* lays about 27 million of eggs in her life span. Among the lower vertebrates where no parental care is given to the young, the potential productivity is necessarily enormous. In Herrings the number of eggs varied from 20,000 to 47,000, in a Cod there may be as many as, 6,000,000 eggs, in Turbot, 9,000,000 and in a Ling, 28,000,000 eggs in a season. One female toad may lay as many as 12,000 eggs. The elephant is the slowest breeder; the one pair normally produces only six off springs in one hundred years. If they are allowed to reproduce, and if all the off springs survive then in a span of 750 years, a single pair will produce 19 million descendants.

2. Survivor's number constant

Although the animals and plants produce great numbers of descendants, yet the number always remains constant, this is because otherwise the food and land will be much less for over population. Similarly to keep the number within reasonable limits curbs and checks are operating. These may be either due to limited food supply, predatory animals, as diseases, as space restriction, as the inanimate environment that includes climate, seasonal changes, drought, flood etc. Thus the numbers of individuals in a species remain more or less the same.

3. Struggle for existence

Struggle for existence is most important check for keeping the numbers constant. Due to the excessive rate of production of organisms, there is an everlasting competition or struggle between the various individuals for food, space and other requirements. Therefore, the population of most species tends to remain more or less constant because of various limitations such as lack of food, living space and breeding spaces etc. For all these requirements, a competition for existence takes place among the individuals of the species and this is what is called as a struggle for existence. There are three types of the struggle for existence, which are as follows:

(a) Intra-specific

This is the struggle among the organisms of single and same species i.e. within the same species because of their requirements like food, shelter, breeding places etc. are similar. Cannibalism (eating the individuals of own species) is the example of intraspecific struggle. Many human wars are also included in this category.

(b) Inter-specific

This is the struggle among organisms of different species i.e. between the different species. For example, the rabbit is preyed upon by a fox, fox by tiger on its turn. In this way a struggle continues between aggressor and a victim.

(c) Extra-specific or Environmental struggle

The environmental factors like extreme cold, heat, heavy rains and earthquakes also play an important role in determining the number of individuals and causing population control.

4. Variation and heredity transmission

Due to everlasting competition, there is a variation of living beings. With the changing conditions, all individuals show at least some and very few changes from each other, and this is the variation. Due to the variations, some individuals would be better adjusted towards the surroundings than the others. Adaptive modifications are caused by the struggle for existence. Darwin considered these variations to be hereditary. Only those variations which are helpful and most suited are transmitted to the next generation while unsuited variations are eliminated.

5. Survival of the fittest or Natural Selection

Darwin suggested that in the struggle for existence only those individuals survive and propagate which adopt the changing variations. This process was named as the “survival of the fittest”. The individuals who lacked favorable variations will not be able to face the condition of life and will perish or fail to reproduce and hence will be eliminated from the population.

According to Darwin, the process would continue to operate in succeeding generations, gradually adapting the animals to their respective environments. When there is a change in the environmental conditions there would also be corresponding changes in the sort of characters that could survive under natural selection.

Darwin's finches: A species in a changing environment or a species migrating to a new environment would in course of time be suitably changed in the new conditions. This is clearly shown by birds known as Darwin finches. There are 13 species of this bird having special adaptations suitable to a particular environment. For example, the birds which fed on insects had long slender beaks by which they could pick small insects from small crevices or pierce them. On the other hand, plant eaters had strong beaks which were helpful in breaking hard nuts. The birds which lived on islands covered with volcanic eruptions had black feathers matching with the surroundings. The birds living in green vegetation have green feathers and those finches inhabiting probably beaches had speckled grey plumage. Darwin, therefore, argued that if a group of individuals of same species survives the conditions of life in a particular area through continued variations along different lines under natural selection, then after many generations each individual will be so different from its ancestors that it will be classified as a new species.

Darwin also cited the example of tiger, leopard, lion and cat that all these animals possess a number of similarities but are different so that they are classified separately. This might be due to the fact that all of them might have diverged from a common ancestor but through variations, competitions and natural selections. They possessed their present position on the taxonomic calendar. Therefore, in the words of Darwin, "it is the nature which decides on the selection or otherwise of particular individuals for continued existence."

6. Origin of New Species

According to Darwin adaptation of survivors to new environments may lead to the formation of new structures and modes of behaviors. Thus, organisms, generation after generation, will show new forms and thus latest forms will be regarded as new species. Thus, modifications in relation to changes of environment will lead to the origin of new species.

Darwin thought that possible new species might have arisen from the old ones with the difference of lines of descent, which produced varieties, incipient species and then species themselves.

Evidence In Favor Of Darwinism:-

There are number of evidences which go in favour of Darwinism. Some of them are given below:

1. Size. In olden days there existed large sized animals which later on faced the scarcity of food, space and the changes in the climate. They are now replaced by the small size and more suited animals.
2. Pedigree. Pedigree of horse, camel, elephant and other animals also support the theory of Darwin
3. Struggle for Existence. Competition or struggle for existence is seen in all organisms.
4. The abundance of Variations. Variations are so abundant in nature that no two individuals of a species are similar, not even the monozygotic twins (they possess some dissimilarity due to their environment).
5. Production of New Varieties of Plants and Animals by Sexual Selection. When a man can produce various new varieties of plants and animals in a short period, nature with its vast resources and a long time at its disposal can easily produce new species by selection.
6. Mimicry and Protective Coloration. They are found in certain animals and are products of natural selection.
7. Correlation between Nectaries of Flowers and Proboscis of insects (Entomophily). The position of nectar in a flower and the length of proboscis in pollinating insects are wonderfully correlated.
8. Pedigrees of some Animals. Pedigrees of horses, camels and elephants also support the Natural Selection Theory.
9. The rate of reproduction. The rate of reproduction is many times higher than the rate of survival in all organisms.

Objectives to Darwinism

In spite of the fact that Darwinism is universally accepted as the factor of evolution, there are many objections to the theory and they are as follows:

1. *Inheritance of small variations.* According to natural selection theory, only useful variations are transmitted to the next generation, but sometimes small variations which are not useful to the possessor, are also inherited. It is beyond understanding that if the appearance of small wings in birds could help them in flying.
2. *Over-Specialization of some organs.* Some organs like tusks of elephants, antlers of deer have developed so much that instead of providing usefulness to the possessor, they often give hindrance to them. This theory cannot explain these facts.

3. *Vestigial organs*. Theory of natural selection does not satisfactorily explain the occurrence of vestigial organs, which are useless and if they were not of any use, they should not have been preserved and further developed by natural selection.

4. *Duration* The geologic time has been too short to give selection opportunity to do its work.

5. *The arrival of the Fittest*. Darwin left us with a very fundamental and important problem, which was not solved by him that “upon what material does natural selection act in the formation of species”. It is true that fittest survive, but what is the origin of fittest?

6. *Discontinuous Variations*. The theory fails to explain the cause of sudden changes in the body. The main drawback of Darwin was a lack of the knowledge of heredity and that is why he could not explain, how the variations are caused.

7. *Selection*. Selection depends on the organisms having a sum total of good and bad characters and not a single character.

8. *Darwinism* does not include the traditional stages in the formation of new species.

9. It is difficult to imagine a reason why variations tending in an infinitesimal degree in any special direction should be preserved.

10. No one has ever observed new species developing from another - this ought to be possible if evolution by natural selection is not in progress.

These are some of the reasons which, on purely rational grounds, appear amply to justify those who decline to pledge their faith in Darwinism in spite of the popularity it enjoys.

11.3.3 Modern Synthetic Theory or Neo-Darwinism:-

Darwinism was generally accepted by biologists in the latter part of the last century. But about 1890, doubts began to be thrown upon it and around 1910, some critics proclaimed the death of Darwinism due to following facts. Darwinism became purely speculative involving selection to explain anything and everything without requiring proof and without providing any explanations. With the discovery of mutations, it is said that hereditary change proceeds by large jumps.

Mendel contributed the force necessary to establish Darwin's concept and its general acceptance. Darwin's natural selection theory contained a number of defects which cannot explain the entire process of evolution. For example, it failed to differentiate acquired characters and inheritable variations. Secondly, natural selection is a limiting and not an initiating force. In

the light of modern developments, Huxley, Haldane, Goldschmidt, Dobzhansky, Fischer and others put forth the theory which supported Darwinism and this is named as Neo- Darwinism.

According to Darwin, evolutionary change in animals is small variations and suitable variations under the force of natural selection survived. This is something true and several important pieces of evidence of natural selection have come to light. For example, if an area of mosquitoes is sprayed with DDT to kill them, mutant forms have been found to have evolved which showed great tolerance to DDT. Thus, Neo-Darwinism has a genetic basis and it lays special emphasis on the occurrence of mutations. Neo-Darwinism only involves the germinal mutations.

Although the genes are highly stable units but undergo mutation which may be caused by physical or chemical changes in the makeup of genes. If a single gene undergoes mutation while the corresponding gene in the other chromosome remains unchanged, the resulting organism will become a hybrid for that particular trait. It has been found that gene mutations can be induced by various extrinsic factors such as X- rays, chemicals etc in every generation by sexual recombination or by mutations inheritable variations may arise.

If the organisms exhibiting such variations survive and reproduce, their genetic make-up will be perpetuated so that ultimately it will spread to many or all the members of the population. Whether such a spreading will take place or not depends on natural selection Natural selection is, therefore, synonymous with differential reproduction and it means that those individuals of a population which leave more offspring than others are more successful.

By various experiments and statistical analysis it has been shown that the gene frequencies in a population will remain constant from generation to generation if (a) mating is a random process, (b) if gene mutations are balanced and (c) if the population is large. This is known as Hardy-Weinberg law (HWL) which states that when a population is in genetic equilibrium, the rate of evolution is zero. When two individuals with mutually stable traits mate the gene, which controls such traits will increase in number through the offspring and spread through populations. Individuals with mutually undesirable traits are not likely to mate and so their genes do not spread through populations. Thus non-random mating in a population means natural selection. As a result, some genes spread more rapidly than others in a population, the gene frequencies will become altered upsetting the HWL equilibrium. This represents evolutionary change, natural selection operating for or against given genes.

Darwin's theory of Natural Selection was accepted to the account of its direct approach and practical nature. The staunch supporter of Darwinism was Wallace, Thomas Henry, Huxley, Ernst Heinrich Haeckel, August Weismann and Mendel etc. But in 1880 doubts started creeping up about its validity and applicability. Many biologists carried out experiments to provide support to Darwinism. These supporters of Darwinism are known as Neo-Darwinians. These have introduced a number of new facts to make the idea of natural selection more conceivable. Some of the experiments conducted in this series are as follows.

1. Whedon's experiments with the shore-crabs of Plymouth sound: Weldon in the experiments with shore-crabs placed a large breakwater near the mouth of Plymouth sound. This slowed the rate of flow of river water and china-clay deposition was increased. This caused the death of numerous crabs. The survivor had slightly narrow frontum and there was a progressive narrowing of the frontum in succeeding generations. This showed that under the changed environmental conditions natural selection operates upon minute fluctuating variations.

2. Cesnola's experiments with mantis: the role of natural selection was illustrated by Cesnola in *Mantis religiosa* by fixing them on plants. Those having color marking harmonious with the plants survived, whereas all others were eaten up by the birds.

3. Polution's and Sander's experiments with butterfly pupae: The survival value of protective coloration was also exhibited by Polution's experiments. The numerous pupae of butterflies with different colors were placed under conditions which favored protective colouration. Some of them were also kept in the non-harmonious background. The protective colouration was found to have a survival value.

4. Davenport's experiment with chicken: Chicken with black, white, barred and checkered color pattern was left in the field. It was found that the chickens with plain colors were killed by hawks while those with barred and checkered color pattern were spared because these were inconspicuous from the surroundings.

In last 25 years, a number of new facts have been added to the knowledge of evolution and the theory of Natural selection has been re-analysed.

The modern theory of Origin of Species or evolution is known as Modern Synthetic Theory of Evolution or Neo-Darwinism. Theodosius Dobzhansky reviewed the Darwinian concept of evolution by Natural Selection in Mendelian populations. In his book 'Genetics and origin of species' (1937), he presented the chromosomal studies of *Drosophila* populations and interrelation among its different species. E.B. Babcock provided botanical support to the 'Neo-Darwinian theory' by studying plant genus *Crepis*. Stebbins (1950) provided an account of 'Variation and Evolution in plants'. It presents a combination of mutation, variations, heredity, isolation and natural selection.

1. Mutation: Any alteration in the chemistry of gene or DNA molecule, which is able to change its effect, is known as gene mutation. Mutation can produce drastic change or may remain insignificant.

2. Variation and heredity: During Darwin's time little was known about the nature of genetic variations caused by reshuffling of genes during sexual reproduction. The phenomenon of meiosis caused a random assortment of genes during synapsis and rearrangement maternal and paternal chromosomes in both kinds of gametes. Such a reassortment of genes, especially in a large population with the large gene pool is one of the basis of appearance of a new organism.

Crossing over of genes during meiosis also adds to the variations and chromosomal variations like inversion and translocation and moreover in the chromosome number (polyploidy) also result in the origin of new species.

4. Natural selection: Natural selection includes all those kinetic forces introduced by physical and biotic factors, which determine how and in what direction an organism is going to change. Natural selection plays no favoritism, but it is obvious that the organism which is more suited for the environmental condition will survive overpowering the force of competition. Thus the natural selection of a creative process which uses the variations and mutations of the raw materials from which better survivals having combinations of better survival value are obtained. How natural selection acts in nature can be exemplified as follows:-

In any physical environment at a given moment, a certain proportion of individuals in the population carry normal genes while others represent the mutants, which have mutant genes combined in such a manner that the individuals carrying them differ from normal parents. If the gene pool of that population achieves stability i.e. there are no more changes in the genotype of individuals of the population, it will exhibit following conditions:-

- i) Mutational equilibrium
- ii) Random Mating
- iii) Equal chances for all genotype to live and reproduce.

But a population is never stable and constant, and changes in its genetic code, chromosomal rearrangement and recombination of genes. Due to the unequal opportunity of mating and inadequate chances of survival in every case, the individuals with changes of survival value survive and perpetuate, while others die off. Thus natural selection due to environmental or biotic factors always exerts a selective influence as a result of which certain mutational changes or variations establish themselves in the line. The process is known as non-random reproduction or differential reproduction.

4. Isolation: Isolation or segregation of individuals of a species into several populations or groups under psychic, physiological or geographical factors is considered to be one of the most important factors responsible for evolution.

Geographical isolation includes physical barriers like mountains, rivers, oceans and long distance, which prevents interbreeding between related forms. Physiological barriers help in maintaining the individuality of the species because these isolations do not permit the interbreeding among the individuals of different species. All these lead to reproductive isolation.

Origin of new species: The population of a species, when representing in different environments, and are separated by some above-mentioned barrier, accumulate different

mutations independently and become morphologically and genetically so different that they become reproductively isolated and form new species.

Difficulties of Neo-Darwinism

Goldschmidt believed that the Neo-Darwinian type of evolution, by the accumulation of micro mutations under the influence of natural selection, is largely restricted to subspecific differentiation within species and that the decisive step in the formation of new species must involve an altogether different genetic process, the systematic mutation. Only a few of the reasons which led him to this conclusion can be indicated here, briefly. If Neo-Darwinian evolution gives rise to new species, then new species should come only if the terminal members of a Rassenkreis and the Rassenkreise of closely related species should blend into one another. But actually, this does not happen. He believed that good species are always separated from their nearest relatives by a bridgeless gap. Controversial cases he believed depend in part upon purely morphological definitions of species which do not take the genetic facts into account. Goldschmidt believed that interbreeding or potentially interbreeding, populations should be treated as a single genetic unit, a species, from an evolutionary point of view, even if other factors may make it advisable for taxonomists to break it up into several species. On this basis, many difficult cases can be resolved in accordance with his ideas.

11.3.4 Mutation Theory:-

Mutation theory is somewhat recent and convincing up to some expectation. It was put forward in 1901 by a Dutch botanist -Hugo De Vries (1848-1935). He was the director of the Botanical Gardens at Amsterdam. His conclusions were based upon careful observations on evening primrose – *Oenothera lamarckiana*. According to his theory, new species arise from pre-existing ones in a single generation by the sudden appearance of marked differences called mutations. Evolution is, thus, a discontinuous and jerky process, rather than a continuous and gradual one as held by Lamarck and Darwin. In other words, there is a jump from one species to another.

The Raw Materials of Evolution

The Dutch botanist Hugo De Vries, one of the three rediscoveries of Mendel's laws, experimented with a number of plants, especially the evening primrose, which grew wild in Holland when he transplanted these into his garden and bred them, he found that unusual forms; differing markedly from the original wild plant, appeared and bred true thereafter. For these sudden changes in the characters of an organism, he coined the name "mutation".

Darwin had referred to such changes but believed that they occurred too rarely to be important in evolution. Countless breeding experiments with plants and animals since 1900 have shown that

such mutations occur constantly and that their effects may be of adaptive value with the development of the gene theory, the term mutation has come to refer to sudden, discontinuous, random changes in the genes and chromosomes, although it is still used to some extent to refer to the new type of plant or animals.

In the plants and animals most widely used in breeding experiments, corn and fruit fly several mutations have been observed in the past fifty years. The fruit fly mutations are tremendously varied, including all shades of body color from yellow through brown and gray to black; red, white brown or purple eyes, peculiarly shaped 'wings and a complete absence of wings, oddly shaped legs and bristles and such extraordinary arrangements as a pair of legs growing from the forehead in place of the antennae. Among domestic animals, mutations are no less common, the six-toed cats of Cape Cod and the short-legged breed of Ancon sheep are two of many examples of the persistence of a single mutation.

Early in the present century, a heated discussion arose as to whether evolution is the result of natural selection or of mutations. As more was learned about heredity, it became clear that natural selection can operate only when there is something to be selected in another word, when mutations present alternate ways of coping with the environment. The evolution of new species, then, involves both mutation and natural selection by differential reproduction.

A similar argument has continued to the present day between the Neo-Darwinists who believe that new species evolve by the gradual accumulation of small mutation, and another group, who believe that new species and genera arise in one step by a macromutation or major change in the genetic system. Such a macromutation producing a major change early in development would result in an adult from, considerably different from its parents into the new species or genus. \.

Many major changes result only in monster which dies almost immediately, though some give rise to what Rich and Goldschmidt of the University of California called "hopeful monsters" forms enabled by their mutation to occupy some new environment. He suggested that the evolution of the extinct ancestral type of bird. Archaeopteryx, into the modern bird, may have occurred in fashion. The archaeopteryx had a long, reptile-like tail. If by a single mutation that tail was greatly shortened, a hopeful monster with the fan-shaped arrangement of feathers might have been the result. The new tail better suited for flying than the old, long one would give its possessors a selection advantage in subsequent evolution. There is, of course, no proof that this is how present birds evolved.

Analogous major skeleton changes to occur as the result of a single mutation. The Manx cat, for example, owes its stubby tail to a mutation in some ancestor, which caused the shortening and fusing of the tail vertebrae. Goldschmidt did not deny the role of the accumulation of small mutations in evolution, geographic races and not to species, genera and the higher taxonomic divisions.

DE VRIES'S EXPERIMENT:-

De-Vries' experimental plant - *evening primrose (Oenothera lamarckiana)* is a plant native of America. It is a biennial plant of about 5-6 feet height. It bears bright yellow flowers at the tips of the branches. The flowers blossom in the evening, hence named evening primrose. During his work in the gardens, De Vries observed not only the original *Oenothera lamarckiana* but also two other varieties which he named as *Oenothera brerisfylis* characterized by short-styled flowers and *Oenothera lamarckiana* characterized by smooth leaves. Out of curiosity, he cultivated the three different plants in his garden and collected 54,343 plants, out of which 837 were different from the original wild parental variety. The markedly different forms were found to breed true. They gave rise to a few still more different plants in each generation. From this, De Vries held that the new types were appearing in evening primrose and that he was actually seeing evolution going on. He called the marked difference 'mutations' or 'spots' and the plants bearing them "mutants". He found that the mutations appeared suddenly and were inherited by the offspring.

As De Vries has pointed out, each mutation may be different from the parent form in only a slight degree for each point although all the points may be different. A unique feature of these mutations is the constancy with which the new form is inherited. It is this fact, not previously fully appreciated, that De Vries work has brought prominently into the foreground. There is another point of great interest in this connection many of these groups that Darwin recognized as varieties correspond to the elementary species of De Vries. These varieties, Darwin thought, 'are the first stages in the formation of species, -and, in fact, cannot be separated from species in most cases. The main difference between the selection theory and the mutation theory is that the one supposes these varieties to arise through a selection of individual variations, the other supposes that they have arisen spontaneously and at once from the original form. The development of these varieties into new species is again supposed, on the Darwinian Theory, to be the result of further selection, on the mutation theory, the result of the appearance of new mutations.

In consequences of this difference in the two theories, it will not be difficult to show that the mutation theory escapes some of the gravest difficulties that the Darwinian theory has encountered. Some of the advantages of the mutation theory may be briefly mentioned here:

1. Since the mutations appear fully formed from the beginning, there is no difficulty in accounting for the incipient stages in the development of an organ, and since the organ may persist, even when it has no value to the race, it may become further developed by later mutations and may come to have finally an important relation to the life of the individual.
2. The new mutations may appear in large numbers, and of the different kinds, those will persist that can get a foothold. On account of the large number of the times that the same mutation appears, the danger of becoming swamped through crossing with the original form will be lessened in proportion to the number of new individuals that arise.

3. If the time of reaching maturity in the new form is different from that in the parent forms, then the new species will be kept from crossing with parent form, and since this new character will be present from the beginning, the new form will have much better chances of surviving than if a difference in time of reaching maturity had to be gradually acquired.
4. The new species that appear may be in some cases already adapted to live in a different environment from that occupied by the parent form, and if so, it will be isolated from the beginning, which will be an advantage in avoiding the bad effects of intercrossing.
5. It is well known that the difference between related species consists largely in differences of unimportant organs, and this is in harmony with the mutation theory but one of the real difficulties of the selection theory.
6. Useless or even slightly injurious characters may appear as mutations, and if they do not seriously affect the perpetuation of the race, they may persist.

Later Investigation or Mutations:-

Since the publication of De Vries's classic investigations a large amount of attention has been paid both by botanists and by zoologists to the subject of mutations. Some of the investigators, notably B.M. Davis, went far toward discrediting the whole of the exceptionally careful work of De Vries by claiming that *Oenothera Lamarckiana* is of hybrid origin. It was pointed out that the form Worked with is a domestic type escaped from cultivation and that there is nowhere in the known world any wild species comparable with it. It is supposed to have been brought to Europe from America many years ago, but there are no such species in America today. Davis claims that he has succeeded in producing, by crossing two American wild species, a hybrid from distinctly resembling *Oenothera Lamarkiana*, and that when inbred this hybrid produces offspring showing various combinations of the two parent species that are not unlike some of the mutants observed by De Vries has also pointed out that the pollen grains of *Oenothera Lamarkiana* exhibit a high percentage of sterility, which he believes to be a stigma of hybridity. The general terms of this type of destructive criticism are to invalidate the whole mutation theory as developed by De Vries and to reduce his mutants to the level of mere mendelian recombination of characters once introduced from two or more parental species.

A large amount of work on the cytology of *Oenothera* by Gates and others has, however, served to show that the mutants of De Vries are more than hybrid segregates. Moreover, the beautiful work of Blakeslee on the Jimson weed (*Datura*) and the work of many other botanists, whose findings are reported by Gates in a contribution quoted below, serve to indicate that the type of evolutionary behavior first observed in *Oenothera* is by no means exceptional, but is probably a common thing at least among plants and may be commoner than we at present know of animals. It may be said by way of anticipation of Gates detailed account that nearly all of the mutations observed in various species of plants may be definitely correlated with observable changes in the

chromosomes of the germ cells, involving changes in number or changes in the arrangement of these nuclear elements.

While botanists buried themselves with their type of mutations, the zoologists, especially T.H Morgan and his able collaborators, were making discoveries of the equal moment in connection with their studies of the mechanism of mendelian heredity in *Drosophila*.

Hundreds of new hereditary types arose, apparently spontaneously, in pure pedigreed stock. Each new type is designated a mutant, and the cause of the changed hereditary condition is not a gross chromosomal change, but an invisible change of a definite point in a definite chromosome, whose cause is unknown but whose location can be exactly determined. Such mutations are known as gene mutations. Like the mutants of *Oenothera*, these *Drosophila* mutants do not differ from the parent species is just one or two characters but in several or many characters. Usually, some one or two characters in any given mutant are especially characteristic, and these serve to give a name to each mutant and make it easier to identify them. Both morphological and physiological characters are involved in these mutants, and every part of the body may be involved. Sometimes the change is so slight as to require an eye sensitized by much training to detect them. It may happen, for example, that two mutants of the eye are so much alike that the human eye is not sufficiently keen to tell them apart, but they may be distinguished by differences in their hereditary behavior. A large percentage of the mutants discovered in *Drosophila* are lethal which means that the change is decidedly for the worse under the prevailing conditions of life and that they render the individual unfit to live.

De Vries regarded the main plants as elementary species and classified them as following:

(a) Progressive species

In this case, one or more new characters are observed which are quite different from the original plants. *O. gigas* is more vigorous than the parent plant and much stouter. With large leaves and flowers.

(b) Retrogressive species

When in mutant there is a loss of one or more characters of the parental nature, the variety is called as retrogressive species. *O. nanella* is a dwarf plant having only one-fourth of the height of the parental plant. A

(c) Degressive species

In this case, one or more, essential characters are lost and due to this, their survival becomes limited. In

Oalbida, the chlorophyll becomes defective hence could not survive.

(d) Inconstant species

These are the mutants which behave just like the parents and occasionally give rise to mutants. *O. lata* bears only pistils in the flowers and hence self-pollination is not possible.

On the observations recorded by De Vries, he suggested that new species arose as a result of large, conspicuous, discontinuous mutations (variations).

Objections to Mutation Theory:-

As per our recent knowledge, it is established that mutations are rare and non-predictable. Therefore, it is doubtful that animals and plant species could appear by mutations.

Moreover, the cases of amazing resemblance of the mimics with their models, harmonization of animal colours with their surroundings and relationship between position of nectaries in flowers and length of proboscis in their insect pollinators cannot be imagined to have developed all of a sudden by mutations because some of the characters are attained during lifetime only by adaptations.

However, De Vries' mutation theory contributes a lot in the field of gene studies.

Evidences Supporting Mutation Theory

Besides objections, there are certain sure points which go in favor of mutation theory. There are definite examples of mutations giving specific characteristics. Some of them are:

- (a) Ancon sheep (a short legged variety) was produced by an ordinary sheep in a single generation in 1891. This mutant is highly beneficial for farmers.
- (b) Hornless cattle from normal were produced in 1889.
- (c) Hairless cats, dogs, and mice were produced from normal parents.

The discovery of mutations in *O. lamarckiana* by Hugo De Vries gave ample ideas to scientists to investigate the nature and behavior of genes.

11.4 Summary:-

To sum up, Darwinism today is very much alive. In certain respects, indeed modern evolutionary theory is more Darwinian than Darwin was himself. Darwin's special contribution to the evolutionary problem was the theory of natural selection, but owing to the rudimentary state of knowledge in certain biological fields was forced to bolster this up with the subsidiary

Lamarckian hypothesis of the inheritance of the effect of use and disuse and modifications produced by the direct agency of environment.

11.5 Self Assessment Questions

1. Mutation is:

- a- Change which affects the parents only and is never inherited.
- b- A factor responsible for plant growth.
- c- Change that is inherited.
- d- A change which affects the offspring's of F₂ generation only.

Ans- c

2. Theory of “Continuity of Germplasm” was propounded by:

- a- Gregor Mendel
- b- Lamarck
- c- August Weismann
- d- Haeckel.

Ans- c

3- “Philosophic Zoologique” is written by

- a- Darwin
- b- Osborn
- c- Haeckel
- d- Lamarck

Ans- d

4- The gene frequencies in a population will remain constant from generation to generation if

- a- mating is a random process
- b- If gene mutations are balanced
- c- If the population is large.
- d- None of the above

ans- d

5- “the proper use of an organ establishes it while no use makes its eventually lost” was said by

- a- Darwin
- b- Weisman
- c- Osborn
- d- Lamark

Ans- d

6- Theory of natural selection is based on

- 1. Use and disuse.
- 2. Inheritance of acquired characters.

3. Struggle for existence and survival of the fittest.

4- Mutations.

Ans- c

11.6 Terminal Questions:-

1. Describes the Lamarckism? Describe it in detail?
2. What is Darwinism? Describe it in detail?
3. Give an account of De Vries' Mutation theory of evolution?
4. What is the Variation? What are their causes?
- 5-What do you know about Weismann's Theory of Continuity of Germplasm? What was its Impact upon Lamarckism?
- 6- What is Neo-Darwinism?
- 7- What is Neo-Lamarckism?
- 8- Briefly, compare Lamarckism and Darwinism?

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UNIT 12: MODERN EVOLUTIONARY CONCEPT AND DETAILS OF MICRO, MACRO, AND MEGA EVOLUTION

Contents

- 12.1 Objectives
- 12.2 Introduction
- 12.3 Evolutionary Concept
 - 12.3.1 Modern Evolutionary Concept
 - 12.3.2 Micro Evolution
 - 12.3.3 Macro Evolution
 - 12.3.4 Mega Evolution
- 12.4 Summary
- 12.5 Self-assessment questions
- 12.6 Suggested Readings
- 12.7 Terminal Questions

12.1 Objective:-

Modern Evolutionary concept and detail of micro, macro and mega evolution and Understand the causes, Process and Consequences of Evolution.

12.2 Introduction:-

The life has been evolving for several billion years, but all observational and experimental studies of evolutionary processes are short term. It is unusual to be able to measure changes in allele frequencies for as long as a century, as we can for industrial melanism. Even in this case, differential survival caused by selective predation by birds has been measured for only a few years. We assume that natural selection has been operated in polluted and unpolluted British woodland in much the same way over the entire one-hundred-year period. This assumption is a safe one because a great deal is known about the history of pollution and about associated vegetation changes in the English Midlands during the last century. Extrapolation over the longer time span and to the less well-known situation is much less certain.

Three processes are central to the long-term evolution of life. One is phyletic evolution: change through time in a lineage of organisms continuously connected genetically from generation to generation. We recognize new species along this lineage, but there has actually been no speciation, just continued, gradual changes. The second is speciation: the splitting of one interbreeding unit into two. The third is extinction: the termination of a lineage. The rates of all three of these processes have changed markedly during geologic time. In a normal course of evolution, a new species is believed to be formed from the pre-existing one by the accumulation of useful and heritable variations or mutation in the course of several successive generations through natural selections. Such heritable variations can be divided into two major categories as micro and macro evolution.

The early earth was very hot, and its surface was molten, explosive eruption of this hot molten sea was common place. Life as we know it would not begin until nearly two billion years later when the surface solidified and water accumulated in basins. But the precursors of life were formed as soon as the surface of the earth began to cool. Hydrogen was common place, and various chemical reactions near the molten surface produced small quantities of oxygen, the oxygen and hydrogen fused to form in the form of steam, which continually rose from the surface of the earth. Other compounds were being formed at this time. Those of importance as precursors to life were the combination of hydrogen and nitrogen to form ammonia, hydrogen, and carbon to form methane gas, and hydrogen and cyanide to form hydrogen cyanide.

As the earth continued to cool, steam rising from its still molten crust began to condense into droplets of water, the most important ingredient to life. At first, it is likely that no water fell to the surface of the earth. Droplets forming in the early atmosphere were converted back to

steam as they fell. When the earth cooled sufficiently, the droplets became rain. The rainy season of our young planets was tens of thousands of years long. Rain brought with it molecules of methane, hydrogen, cyanide and nitrogen, which became dissolved in the first seas. The early ocean is considered to have been a primordial soup that contained all these building blocks of life.

Life did not originate at one specific spot at one specific time in the early oceans of earth, life originated again and again, where ever the necessary precursors accumulated. Various kinds of molecules became concentrated on the bodies of water, and with the energy of lightning volcanic eruptions and ultraviolet radiation, molecules fused together to form macromolecules. The molecules of most important to live were amino acids which fused to form polypeptides and finally protein. Amino acids, as well as proteins, may possess electrical charges, and the attraction of opposite charges is thought to have created increasingly larger molecular aggregates.

12.3 Evolutionary Concept:-

Evolution has been described as the process of gradual modification in the living organism, so as to establish diversity in the world of living being.

12.3.1 Modern Evolutionary Concept:-

Two fundamental patterns could be envisaged in the process of evolution:-

- i) Minor changes in the gene pool of a population from one generation to next, with result, that no new populations are formed, but the descendant population is not genetically identical to its predecessor. This is known as sequential evolution.
- ii) The changes which result in the evolution of new populations, species, families groups or classes. This is known as divergent evolution.

The sequential evolution is, therefore, an example of time without producing new populations. Therefore, the changes occurring on account of evolutionary forces like mutation, variation, natural selection and genetic drift produce only temporary changes which fluctuate at random. For example, in the human population, we find that not even two real sisters or brothers are identical or resemble their parents, yet the changes, do not divide the individuals of a population or race into subcategories. Secondly, these changes are not directional.

The divergent evolution, on the contrary, is an example of directional evolution. The changes occur in a cumulative direction and result in the origin of new populations from the old ones. Therefore, the varied groups of plants and animals either related or unrelated provide an example of divergent evolution. It is the divergent evolution which is more evident. As a matter of fact, the two aspects are rather inseparable. Not even a single population exists, which

exclusively exhibits sequential evolution because all populations diverge in due course of time and split up into the new population. Moreover, the forces responsible for bringing about changes are rather the same in both the cases except that they operate for a very long period and are assisted by additional factors.

Sequential evolution, though help in understanding the operation of various evolutionary forces, does not play any part in the evolution of new species or groups. It is, therefore, the divergent evolution which is seen in fossil records and which actually illustrates the phenomenon of evolution. Goldschmidt has divided evolution into three categories:

- i) Evolution of subspecies or geographic races - Microevolution.
- ii) Evolution of species, genera and so on – Macroevolution.
- iii) Simpson in 1953 has added the term mega evolution for the large-scale evolution of families, orders, classes and phyla.

The sequential evolution is actually microevolution and the divergent evolution in its simplest form i.e. operating at the population level is also nothing but macroevolution only. The fragmentation and development of new populations from the existing population are known as speciation and usually leads to the evolution of new species. Evolutionary changes which are responsible for establishing the taxonomic categories above species level are called macroevolution. It includes adaptive radiation of a population to different new habitats. The mega evolution includes those changes in the organization, which enable the organism to enter into a new major adaptive zone.

12.3.2 Micro-Evolution:-

The evolution, which results from the interaction of the elemental forces of evolution to produce a relatively small change in the population or populations, is known as microevolution. The basic process of microevolution consists of changes in the gene frequencies in a population from one generation to the next. The microevolution, therefore, operates to change the genetic equilibrium in a mendelian population and occurs below the species level.

Micro evolutionary forces- The micromutations or little mutations as described by Goldschmidt are the main sources of producing changes in the gene pool of a population. These are mutations in genes. Genetic recombination or mendelian recombination changes the gene frequency in the gene pool of the population. In addition, genetic drift and natural selection also operate on the populations to change their gene frequency and thereby disturb the genetic equilibrium.

Mechanism of microevolution- The genetic material of the living beings is apt to change. How accurate may be the process of gene duplication or chromosome duplication there

are always some chances of some abnormalities. The changes in the structure or composition of genes are described as gene mutations or little mutations. These may be spontaneous or may be induced by certain chemicals and environmental factors like radiation etc. The recombination of genes by interbreeding also helps in the introduction of new combinations of already existing genes and introduces variations in the genotype of the individuals. Interbreeding helps in the spread of micro-mutations.

The variations introduced in a gene pool of population by mutations and recombination is operated upon by the natural selection. As a result, the offspring of the population are found to be different genetically as well as phenotypically. This changed population or descendant population is the product of microevolution. Changes produced by mutation and recombination may be beneficial or may not be so. But since the changed genotype interacts with the environment, only those changes which increase the rate of reproduction of the organisms directly become more numerous in the population. From this, it could be inferred that variations of adaptive value are preserved and encouraged by natural selection.

Microevolutionary forces operating for a shorter period produce sequential evolution, whereas when continued for generations together result in the evolution of new populations from existing one. The origin of new populations can occur in two different ways:-

- i.) In a succession manner
- ii.) In a divergent manner

The successional microevolution is the evolution within a single population which results in the successional replacement of the pre-existing by the new ones. This could be seen in successive strata of palaeontological series. It leads the micro-evolution to the formation of clines, when characters of a population seem to change gradually across its place of distribution. The formation of clines is an example of gradual changes in response to gradual changes in the climate.

The divergent microevolution results in the splitting of a parental population into two or more new population with the appearance of genetic divergence. Isolation is the additional factors operating to establish genetic divergence in the related populations.

The microevolution is, therefore, a continuous and gradual change in the interbreeding population, which become geographically isolated into local populations. Then each one of these develops small variations, which gradually accumulate to produce large differences in their morphology or physiology so that each such local population becomes markedly different from other and from the parent population. The variation occurs on account of micro mutations and recombination and is favored by natural selection.

12.3.3 Macro Evolution:-

The macroevolution is also known as adaptive radiation. The evolution, which results in the production of new adaptive types through a process of population fragmentation and genetic divergence, is known as macroevolution. It operates above the species level and results in the splitting of the population of a species into several subgroups, each of which exhibits changes in a definite adaptive direction. These changes are known as adaptive trends and the phenomenon as the adaptive radiation or macroevolution. It means macroevolution is actually adaptive radiation.

Mechanism of macroevolution:-

Macroevolution operates above species level and results in the establishment of new genera, families and orders. The changes in the organization occur on account of sudden mutation of large size, which is named macroevolutions or systematic mutations by Goldschmidt. Macroevolution occurs in a group of individuals which have entered a new adaptive zone free of competition. The entire mechanism could be conjectured as follows:

In a new adaptive zone, the number of individuals is far less and the opportunities to avail new habitats are more. Therefore, the intraspecific struggle is roughly nil. Moreover, the new zone will be almost free from the enemies. Therefore, the newly entered populations enter all the available habitats of the adaptive zone and start adapting themselves according to the conditions and need. It means that the one population which had acquired the new zone gets split up into several subpopulations, each of which accumulates mutation and evolves independently but simultaneously in different directions. On account of the different environmental conditions, there is different urge of natural selection and adaptive modifications occur in different directions. Adaptive modifications in each sub-population have a cumulative effect and are, therefore, directional.

Macroevolution, therefore, has following essential features:

1. Macroevolution occurs on account of macromutations.
2. Macroevolution occurs in those populations which have entered or acquired a new adaptive zone.
3. Macroevolution results in evolutionary divergence i.e., the preexisting population divides into several diverging descendent populations by acquiring special adaptations.
4. Macroevolution produces groups of parallel special adaptations among divergent stocks.
5. Macroevolution leads to specialization in a particular direction. As a result, forms with special adaptations become rigidly specialized to narrow adaptive subzones and reach the adaptive peak. This very often leads to overspecialization and finally to the extinction because overspecialized forms are unable to modify when they come to a new adaptive zone.

12.3.4 Mega Evolution:-

Mega evolution has been described as the origin or evolution of new types of biological organization of general adaptation from its predecessor, resulting in the formation of new classes, groups or phyla. Mega evolutionary changes are rare and have occurred only a few times in the evolutionary history of living beings. But the most interesting thing is that all these biological organizations persist without extinction. All the phyla and most of the classes of microorganisms, plants and animals represent their separate organization and are produced as a result of mega-evolution. The origin of amphibians from fishes, the origin of reptiles from amphibians and the origin of birds and mammals from reptiles afford best examples of mega evolution.

Mechanism of mega evolution:-

During mega evolution the organisms of the ancestral stock attempt to enter a new zone, which is uninhabited by these forms and is devoid of competition. These exhibit varied modifications in different directions until one of these has found suitable to the new zone. It means groups of individual of a parental stock develop certain generalized preadaptation which enables them to enter the new zone. Therefore, these make a break-through into the new adaptive zone and start radiating into all the available habitats, thereby developing more specialized adaptations which are known as post adaptations.

The mechanism of mega evolution can be explained by taking the origin of reptiles from amphibians as an example. Amphibians are amphibious creatures which could live in moist places near some source of water. Reptiles evolved as completely terrestrial forms which need not depend on aquatic medium at any stage of their life cycle. At that time the terrestrial zone was unoccupied, devoid of competition and accessible. The principal new general preadaptations which evolved in some of the ancestral amphibians and made the invasion of the terrestrial zone were:-

- i) The development of exoskeleton in the form of scales, plates or scutes which prevented desiccation of the adults.
- ii) The appearance of large cleidoic eggs which enabled the young to develop on land.

Similarly, the origin of birds from some primitive reptiles includes the sudden appearance of wings which enabled the ancestral form to make the invasion of the aerial zone. The fossils of ancestral bird, *Archaeopteryx*, exhibit reptilian characteristics together with wings and feathers. Evolution of mammals can be traced back to a series of fossils synapsid reptiles of the groups therapsids. These developed several mammalian characteristics like the false palate, teeth differentiated into incisors, canines, premolars and molars and the limbs became modified so that the elbows and knees were replaced under the body. But still, these forms were reptiles because teeth were without roots and the quadrate and articular bones did not form the ear ossicles. In therapsid groups, the main preadaptation towards mammalian offshoot was freeing of the quadrate and articular bones from jaw articulation and their conversion into ear ossicle. The particular change served two purposes:-

i) It improved hearing and

ii) Direct articulation of mandible or dentary with the skull strengthening the jaws.

The fossil evidence in favor of mega evolution is relatively rare. The mega evolution, therefore, exhibits following special features:-

1. Mega evolution includes experimentation and exploration of the new zone by the member of the ancestral stock in several divergent lines. This experimentation involves the appearance of new characteristics which may prove suitable for the new zone.
2. Mega evolution operates on individuals which have developed some general adaptations for the new zone.
3. The preadapted group of individuals then crosses the ecological barrier and makes a breakthrough onto the new zone.
4. The breakthrough and shift are always rapid; otherwise, they fail on account of extreme negative selection.
5. The new zone is always ecologically accessible and is devoid of competition.
6. The initial shift is always followed by adaptive radiation which is actually macroevolution.

Macroevolution and megaevolution are highly complex phenomena. These have following characteristics in common.

1. Taking on of the new general adaptations for entering into the new adaptive zone.
2. Invasion of the new zones or subzones within the new adaptive zone by the development of special adaptations.
3. Loss of evolutionary flexibility and channelization into greater and specialization for the ecological conditions of subzones.
4. Reinvasion of the zones and subzones which become partially unoccupied on account of the specialization of the original occupants.
5. Mega evolution is always followed by macroevolution.

12.4 Summary:-

The three levels of evolution i.e. the micro, macro and mega evolution differ to a considerable degree from one another but all are based upon micro evolutionary process and all contribute to adaptation. The elemental forces for the three types of evolution are the same but macro and mega evolution have some additional forces. The macroevolution may be sequential

or divergent but the latter two are always divergent and involve adaptive radiation and divergence. The fossil history of many organism or groups suggests that some species have arisen abruptly and have survived with little change until extinction. For example, in Turkana basin in East Africa 10 of the 13 species of mollusks appeared within the relatively short geological period of 5000 to 50000 years and then remained unchanged for three to five million years. Thus the evolutionary history of some species exhibits abrupt punctuation of rapid speciation in otherwise prolonged periods of unchanging morphology.

12.5 Self Assessment Questions:-

1- Which evolution is also known as adaptive radiation?

- a- Microevolution
- b- Macroevolution
- c- Mega evolution
- d- None of the above

Ans- b

2-operates on individuals which have developed some general adaptations for the new zone.

Ans- Mega evolution

3- The changes which result in the evolution of new populations, species, families groups or classes. This is known as

- a- divergent evolution
- b- sequential evolution
- c- directional evolution
- d- none of above

Ans- a

4- Which type evolution is known as evolution of subspecies or geographic races

- a- Microevolution.
- b- Macroevolution
- c- Mega evolution
- d- All of the above

Ans- a

5- Mega evolutionary changes are

- a- common
- b- rare
- c- rapid
- d- none of above

Ans- b

- 6- The mechanism ofcan be explained by taking the origin of reptiles from amphibians as an example

Ans- mega evolution

12.6 Suggested Reading:-

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2. Rastogi V.B. (1996). Organic Evolution. Kedar Nath Ram Nath, Meerut, U.P.
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12.7 Terminal Questions:-

1. Give a detailed account of evolutionary concept.
2. Describe the mechanism of microevolution in detail.
3. Give an account of macroevolution.
4. Describe the biochemical origin of life at the molecular level.
5. What is mega evolution write in detail?
6. Write a short note on adaptive radiation.
7. Write short notes on sequential evolution.
8. Give an account of the modern evolution.

12.8 References:-

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UNIT 13: ZOOGEOGRAPHICAL REALMS

Contents

- 13.1 Objectives
- 13.2 Introduction
- 13.3 Major Zoogeographical Realms
- 13.4 Distribution Patterns of Animal in Different Geographical Realms
- 13.5 Biogeographic Regions in India
- 13.6 Summary
- 13.7 Self-assessment questions
- 13.8 Suggested Readings
- 13.9 Terminal Questions

13.1 Objectives:

Life occurs in almost all diverse habitats ranging from high mountain peaks more than 20,000 feet to deepest sea bottom up to the depth of about 10,000 meters. A living being can be observed in ponds, pools, ditches, sulphur springs, hot springs, in the devastating cold of the polar region, in the scorching heat of deserts, in dense tropical forests and the wherever existence of life is possible. But all the organisms do not occupy all the possible life supporting areas. Rather they are localized or restricted to a particular area, field country or region. This is known as the range. The range of a species is the area that it occupies of in simple words it is the area of distribution of the species.

13.2 Introduction:-

The above account implies to the distribution of animals in space. There is another aspect of the distribution of animals and that is the distribution of animals in time. It is concerned with the evolutionary history of animals, animal species or groups i.e. when these made their appearance and how long these have existed in their present form.

13.3 Major Zoogeographical Realms:-

The earth surface is occupied by a vast majority of animals and plants but their distribution is not uniform. Each species has its definite range of distribution in which it can thrive the best. Thus the earth surface has been divided into regions, each with a distinctive and characteristic species and genera of animals. A.R. Wallace has divided the earth's surface into six regions. Each has been given the name of the realm and its characteristic organisms constitute the fauna. The entire world has been divided into various regions, which contain specific fauna. It was Sclater in 1857, who first of all divided the earth into following six regions.

1. Palaearctic Region: This includes Europe, temperate Asia, North Africa and Arabia.
2. Ethiopian Region: It includes whole of Africa, Arabia, and South of the tropic of Cancer and Madagascar.
3. Australian Region: This includes Celebes, Lombok, the whole of Australia and the pacific islands.
4. Nearctic Region: It includes Greenland, North America to Northern Mexico.
5. Neotropical Region: Includes South America and Southern Mexico.

6. Indian Region: It includes India, South of Himalayas, China, Borneo, and Jawa.

Six primary zoological regions were established from detailed examples of distribution of chief genera and families of the world. These divisions received the great support from Dr. Gunther who in the Proceeding of Zoological Society for 1858, showed that the geographical distribution of reptiles agreed with it very closely. But, there are apparent objections to this classification on the basis of utilization and distribution of various mammals. Then, in 1876 Alfred Russel Wallace classified the earth into following six regions.

1. Palaearctic Region
2. Ethiopian Region
3. Oriental Region
4. Australian Region
5. Neotropical Region
6. Nearctic Region

Palaearctic Region:-

This is the largest region covering an approximate area of 14, 00,000 sq miles. It includes the whole northern part of the old world i.e. the whole of Europe, the northern part of Africa and Asia, north of Himalayas.

Physical features:-

It shows a wide range of temperature, great variations of rainfall and great diversity in the surface features. The Gulf Stream helps to raise the temperature of Western Europe while the southern portion is sheltered by mountains cutting off the cold winds of the north. The eastern part of Europe is much colder. The highest summer temperature of this area is recorded in northwestern India, Baluchistan, and Arabia.

Most of the northern Palaearctic region is low and flat. Scandinavian and Ural mountains are the only elevated land while low-lying countries are from the Bay of Biscay to Behring. Steppes and desert, with almost no rainfall, extend over the greater part of African and Arabian portions of Palaearctic region.

This region is further divided into four sub-regions:

i) European sub-region: It includes northern and much of central Europe, and extends in the south up to the Pyrenees, the Alps, the Black Sea and the Caucasus range.

ii) Mediterranean sub-region: It includes North Africa to the tropic of cancer, the northern part of the Sahara desert and northern half of Arabia, Persia.

iii) Siberian sub-region: Includes south and Central Asia.

iv) Manchurian sub-region: It consists of Japan, a part of Mongolia and northern China.

Fauna of Palaearctic region:-

Fishes: About 28 genera of freshwater fishes. Paddlefishes, many cyprinids, cobitids, a few catfishes, an umbrid, Dallia, a few mastacembelids. The family Cyprinidae is most dominant.

Amphibia: 16 genera. 8 genera of tailed amphibians- *Proteus*, *Slamendra*, *Hynobius*, *Chiogloss*, *Seiranota*, *Onychodactylus*, *Geotriton* and *Sieboldia*. 8 genera of tailless amphibians are *Bombinator*, *Pelobates*, *Didocus*, *Alytes*, *Pelodytes*, *Discoglossus*, *Laprissa*, and *Latonia*.

Reptiles: True vipers, Pit vipers, *Trigonophis*, *Psammmodromus*, *Hyalosaurus*, *Scincus*, *Ophiomorus* and *Megalochilus*.

Birds: 57 genera. They include grebes, hawks, ducks, quails, grouse, cuckoos, rails, pigeons, owls, goatsuckers, kingfishers, woodpeckers, larks, flycatchers, warblers (*Locustella*), readings (*Conostoma*), and crows (*Pica*).

Mammals: 35 genera. Tailless monkey (*Innus*), *Barbastellus* and Chiroptera (*Plecotus*). 6 genera of insectivore, *Myogale*, *Scaptochirus*, *Anurosorex*, *Scaptonyx*, *Nectogale*, *Uropsilus*. Of carnivore, Raccoon dog of Japan (*Nyctereutes*), Otter (*Lutronektes*), Badger (*Meles*), Panda (*Aeluropus*) and seals (*Pelagius*), camels, deer, antelope, rodents.

Euthiopian Region:-

It includes whole of Africa and Arabia south of the Tropic of Cancer, together with Medagasker and the adjacent islands.

Physical Features

This region is mainly tropical. It is a larger block of rainforests and isolated mountains. This region is further divided into four sub-regions.

i) East African sub-region: Includes Sahara, southern Arabia, north-east Africa and up to Zambesi in the south.

ii) West African sub-region: This includes the whole of the west coast from the south of river Gambia to the Congo.

iii) South African sub-region: This sub-region is bounded by the Kalahari Desert on the northwest and by the Limpopo valley on the northeast and up to Mozambique in the north.

iv) Malagasy Subregion: It includes the mountainous island of Madagascar and its neighboring smaller island.

Fauna of Ethiopian Region:-

Fishes: About 14 genera of freshwater fishes. They include Archaic bichers, lungfishes, many families of isopondyle, many catfishes, some cyprinids, several characins and a few spiny-rayed families.

Amphibia: About 09 genera. Primitive and higher frogs. Family Bufinidae (toads) is peculiar in this region.

Reptiles: 35 genera. Among snakes and *Grayia*, *Leptorhynuhus*, *Rhamnophis*, *Langalia*, *Pythonodepsas*, *Pythons*, a sand boa and typical viperids. There are also *Pelomedusoid* turtles, *Testsudinineland* tortoises and *Emydine*, *Crocodiles*, *Chameleons*, *Skinks*, *Varanus*, *Cordylus*, and *Typhlops*.

Birds: 72 genera. They include hawks, owls, herons, storks plantain-eaters, weaver birds, fruit-thrushes, flycatchers, shrikes, crows, starlings, barbets, cuckoos, rollers, bee-eaters, hornbills, and goatsuckers. Vultures, eagles are also found.

Mammals: 44 families. Among them are otter, shrews, old world monkey, great apes, scaly anteaters, canids, cats, aard-vark, elephants, rhinoceroses, pigs, hippopotamuses, giraffids, bovids, rabbits, rodents, squirrels, spring has, bamboo rats, murids.

Oriental region:-

The Oriental region includes India, Indo-China, South China and the island of Malaya and the islands of Malaya. It is bounded by the Himalayas in the north and the Indian and Pacific Oceans on its other side. But there is no definite physical boundary in southeast corner where the islands, Malaya, Archipelago (Jawa Sumatra) string out until they reach Australia. The big island of Jumandra, Jawa, and Borneo with Philippine group, certainly belong to the oriental region.

Physical features

The climate of the oriental region is mainly tropical in proportion to the extent of its lands. The Oriental region presents a great variety of physical features. The northern portion is composed of plains and desert. More particularly, in the watershed of greater rivers Indus and Ganges, its fauna as a whole shows a great affinity to the Ethiopian region while the desert in it is a debatable and may be regarded as a transitional point between southern portions of India is more luxuriant than north and largely covered with tropical forest. With a series of elevated tracks culminating in the western and eastern ghats.

Finally in the extreme northern portion of this region where the great mountain ranges occur and especially between Bhutan and Yangtiskiang define more temperate condition with an interesting mingling of Palaearctic and oriental type of animals.

Australian Region:-

It includes the whole of Australia, New Zealand, New Guinea, the Malacca and other neighboring islands, and practically the whole of islands is the Pacific Ocean. Its western line is drawn between the islands of Bali and Lombok, then to the east of Celebes or the Philippine islands

Physical features

It is partly tropical and partly south temperate. New Guinea is completely tropical. There are rain forests, grassland, altitudinal vegetation on mountains eucalyptus woods. Northern Australia is also tropical. Southeastern Australia is well watered with eucalyptus woods and some wetter, denser forests on the mountains. Southwestern Australia is wet but it is cut off by desert from the east.

It is further divided into four sub - regions:

- i) Austro-Malayan sub - region: It includes the New Guinea and surrounding islands (such as Aru Island, Mysol, and Waigeon).
- ii) Australian sub – region: It includes the mainland of Australia and Tasmania.
- iii) Polynesian sub – region: This sub – region is composed of islands in the Pacific Ocean (including Sandwich Islands, Marquesas and Society Island, Fiji Island etc.).
- iv) New Zealand sub – region: It includes the mainland of New Zealand and its surrounding islands such as Norfolk Island, Kermadec Islands, Chatham Island and Campbell Island etc.

Fauna of Australian Region:-

Fishes: Freshwater fishes are ceratodontid lungfishes, an osteoglossid and various peripheral fishes.

Amphibia: Only frogs, Leptodactylids, Hylids, Ranids and Brevicipitids.

Reptiles: Among snakes are Pythonidae and Elapidae families peculiar. Other reptiles are trionychid and chelid turtles, crocodiles, geckos, pygopodids, Rhyncocephatids and agamids.

Birds: About 58 families. Peculiar are the paradise birds, pigeons, parrots, Australian Barblers, Honeysucker, scrub birds, flowerpeckers, bell magpies, bower birds, cassowaries, emus, frogmouths, kingfishers, caterpillar shrikes, lyre-birds, mound-makers and shrikes.

Mammals: They are many marsupials, rodents, and bats. *Hypoderma*, *Notopteris*, *Mystacina*. The European rabbit, hare, and fox have been introduced. The wild dogs and pigs were probably brought by prehistoric man. Six families, having about 52 genera of marsupials are distributed. They are Bandicoots, Wombats, Kangaroos and Opossum etc.

Neotropic Region:-

It includes South and Central America and the tropical lowlands of Mexico with Trinidad and West Indies proper. It is joined to the Nearctic region by the Central American isthmus and separated from all other regions by sea.

Physical features

It is mainly tropical but the southern south America continued into the South Temperate Zone. From the west to the east, runs the river Amazon with its hundreds of square miles of evergreen forests.

The Neotropical region is divided into four sub – regions:

- i) Chilian sub – region: It includes the cold and damp forests of Tierra del Fuego, barren plains of Patagonia, treeless Pampas of La Plata in the north.
- ii) Brazilian sub – region: This includes the tropical forest region of South America and its central mass is covered by forest plain of the river Amazon.
- iii) Mexican sub – region: It is composed of land connecting the North and South America. It is mainly mountainous with lowlands on the shores.
- iv) Antillean sub – region: It is composed of West Indies islands such as islands of Cuba, Haiti, Jamaica, Puerto Rico, Angrilla, Grenada, Barbuda, Antigua and other smaller islands.

Fauna of Neotropical Region:-

Fishes: Numerous freshwater fishes are present in this region. Families Polycentridae, Trygonidae, Gymnotidae are present. Catfishes, eels, suckers, nandids, gymnotid are common.

Amphibia: There are caecilians, plethodontid, salamanders, leptodactylids, hylids, Bufo, Pipid frogs, Rhinophrynus present in this region.

Reptiles: In this region, following reptiles are peculiar. Dromicus, Boa, Epicrates, Elaps, Craspedocephalus, Ungalia, Proctotritus, Liolaemus, Celestus, Ameiva, Diploglossus, Phaeodactylus, Crocodiles, Alligators, and Tortoises.

Birds: 23 families. They are Coerebidae, Phytotritidae, Pipridae, Oxyrhyphidae, Dendrocolaptidae, Conopophagidae, Formicariidae, Pterotochidae, Galbulidae, Bucconidae, Momotidae, Todidae, Rhamphartidae, Palamedeidae, Psophiidae, Aramididae, Eurypygididae, Carianidae, Opisthocnidae, and Rheridae.

Mammals: Among mammals peculiar are monkeys (Cebidae), blood sucking bats (Phyllostomidae), Rodents, Sloths (Bradypodidae), armadillos (Dasypodidae), Ant-eaters (Mymecophagidae), carnivores, Marsupials, Sorex and Dicotyles.

Nearctic Region

The region covers the whole of North America and extends south as far as the middle of Mexico. It has Greenland in the east and the Aleutian Islands in the west.

Physical Features:-

It has a great range of temperature. There are large lakes and inland sea in the northeastern portion and ranges of high mountains in the west. Eastern portion is smaller ranges of Appalachian highland. While the central part is of plains, The Southern portion is composed of treeless desert.

The Nearctic region is also divided into four sub – regions:

i) California sub – region: This is the western part of the region and is composed of the country between the Sierra Nevada and the Pacific. It includes Vancouver's island and southern part of British Columbia.

ii) Rocky Mountain sub – region: It includes ranges of mountains extending in the north to the Saskatchewan and in the south up to Rio Grande del Norte, the Gulf of California and Cape St. Lucas.

iii) Alleghany sub – region: This sub – region extends in the west across the river Mississippi, in the south near the Colorado river and to the north up to the boundary between Canada and the United States. It also includes the Nova Scotia and the district between Lake Huron and Ontario.

iv) Canadian sub – region: It includes remaining of North America having pine forest and barren land towards the Arctic Ocean.

Fauna of Nearctic Region:-

Fishes: 24 genera. They include *Huro*, *Brythus*, *Perches*, *Hypodelus*, *Pileoma*, *Noturus*, *Salmon*, *Cyprinids*, *Scaphirhynchus* and *Boleosoma*.

Amphibia: Among amphibians are *Siren*, *Amphiuma*, *Salamanders*, Frogs, toads and *Menobranchus*.

Reptiles: Tortoises, Pythons, *Crotalus*, *Conophis*, *Pituophis*, *Ischnognathus*, *Dinodus*, *Ophiosarus*, *Uta*, *Phrynosoma*, *Uma*, and *Euphryne*.

Birds: 54 genera. They include wood warblers, *Mniotilia*, *Sailia*, *Opororni*, *Helmintherus*, *Harporhynchus*, *Catherpes*, *Chamaea*, Crows, Pigeons, *Hylatomus*, Wading and swimming birds (*Philolea* and *Creagrus*).

Mammals: Monotremes and primates absent. Marsupials (*Virginian*, *Opossum*), armadillo (*Tatusia novencincta*), mountain goat (*Haploceros*), musk ox, rats (*Fiber*), prairie-dogs (*Cynomys*), squirrel (*Tamias*), tree - porcupine (*Erethizon*), Carnivores (dogs, bears, cats, weasels etc.). American Badger (*Taxidea*), Starposed Mole (*Condylura*) and bats (*Antrozous*).

13.4 Distribution Patterns of Animal in Different Geographical Realms:-

Earth originated about five billion years ago. The earth has a polar diameter of about 7900 miles and an equatorial diameter of about 7927 miles. The area of earth's surface is about 197 million square miles, of which about 71 % is covered by seas and oceans. The volume of earth is a little more than 250 billion cubic miles, and its mass has estimated at about 6600 quintillions (6600, 000000000000000000) tons. Life originated on the earth about three billion years ago. Life occurs in almost all diverse habitats ranging from high mountain peaks more than 20,000 feet to deepest sea bottom up to the depth of about 10,000 meters. Living organisms have been reported from ponds, pools, ditches, hot springs, in the devastating cold of polar regions, in deserts, in dense tropical forests and the wherever existence of life as possible. Different organisms inhabit different ecological areas. That is why all living organisms do not occupy all the life supporting areas. Some organisms flourish in one type of areas while others are another

type of area. This is known as the range. Thus the range represents the area of distribution of species.

The distribution of organisms including animals and plants can be studied under following heads:

- i) Distribution in space
- ii) Distribution in time

Distribution of organisms in space

The living organisms distributed unevenly in space including earth's surface i.e. land and water both. This type of distribution can further be classified into:

a) Geographical Distribution

It is also called the surface or horizontal distribution of organisms on land and fresh water in different continents and on different islands. The study of geographical distribution of animals is called Zoogeography.

b) Bathymetric Distribution

It is also called the altitudinal or vertical distribution of organism animals. It deals with the distribution of animals on land and in water. The bathymetric distribution can be studied under the following three subheadings.

- i) Limnobiatic: It deals with the distribution of animals in fresh water sources.
- ii) Holobiatic: It deals with the distribution of animals in the sea.
- iii) Geobiatic: It deals with the distribution of animals on land or terrestrial areas.

Distribution in Time

It is also called the geological distribution or durational distribution. It deals with the distribution of animals in the past earth history. This distribution could be studied only through the fossils.

Pattern of Distribution:-

Animals are found distributed everywhere in the atmosphere. But this distribution is not uniform. Some animals are found in one area but entirely absent from the other. Actually, there is no such place where the animals or plants are not found. The dense tropical forest, the highest mountain, the desert and the Polar Regions, each has its own fauna and flora. Three kinds of distribution of animals have been recognized.

I. Continuous or Cosmopolitan distribution

The animals which are found over a wide and uninterrupted range of surface distribution are the examples of cosmopolitan distribution. Such animals are usually adapted to a wide variety of environmental conditions and possess great powers of movements which enable them to overcome several natural barriers. The green mussel- *Mytilus*, the brine shrimp- *Artemia salina*, rats, bats, hawks, cuckoos are worldwide and represent the extreme of continuous distribution. Birds by their ability to fly over distance have a wider range of distribution and are referred to as eurytopic. The eurytopic animals can thrive in all possible environments. On the other hand, there are some animals which are found in restricted areas and are called stenotopic. These have specialized adaptations to a particular type of environment and are able to colonize in new areas. They are also unable to overcome natural barriers.

II. Discontinuous Distribution

In the case of each of the classes of vertebrates, the center of origin and dispersal was the tropical old world, the largest existing mass. In 1872 Darwin stated that variation would be expected to be greatest in a species having most numerous individual. Later on, this hypothesis was supported by Sir Ronald Fisher in 1930 and F. B. Fore in 1964. According to them, larger the number of individual in a population, greater the number of mutation and recombination genes with the wider variability. On the basis of these descriptions, one can easily understand that there should be some places where a particular group of animals had its birth and then from here it enforced dispersal in all possible directions. Thus it can be concluded that the animals of the neighboring region have some similarities and as the distance of the regions increases the modification in the animals, mode of their life also show great diversity. However, there are some examples where this general rule is not observed. Some members of a group are found in one corner of the earth and the members of the same group are found at the remotest end of this earth and there are absolutely no members of the particular group existing in the region in between.

In such type of distribution where the continuity of the distributed animals is completely broken and no affiliation of one with the other noticeable, the term discontinuous distribution has been applied.

In another way “Geographical discontinuity”, due to the occurrence of the same or related animals in more or less widely separated places, is very common within the main pattern of distribution of vertebrates.

Origin of Discontinuous Distribution:-

According to Philip J. Darlington (1957), the discontinuity in the distribution of animals may be due to:

- i) Reaching the oceanic islands across the water,
- ii) By the submergence of the land mass in between the range, and
- iii) By the extinction of the forms in the intermediate areas.

According to Paul B. Weitz (1966) animals can become discontinuous distributed by the following three ways:

- i) Sweepstake bridges
- ii) Filter bridges
- iii) Corridor bridges

i) Sweepstake Bridge: a sweepstake bridge is the one which depends upon accidental transportation of floating ice, a log of wood clinging mud of the bird's claws or even board of the ship. It provides variously accidental means of species expansion, for example, terrestrial species may cross even extensive water barriers by floating across on uprooted trees or on drifted wood. most small oceanic islands were populated by Sweepstake bridges of this sort. Island contains comparatively few species and subspecies that are present often are quite unique and are found nowhere else. However, they do resemble related species present on the nearest mainland.

ii) Filter Bridge: The main characteristics of a filter bridge are that it does filter out organisms of the connected regions while permitting the passage of other. A filter bridge is a narrow land connection between two continents and has usually existed for the only brief geographical period. Because of its short duration, it would be crossable only by animals and plants capable of migrating fairly rapidly. A good example of a filter bridge is the land connection between North and South America during the late Mesozoic Era. This connection permitted the spread of Marsupial mammals to northward some other animals like anteaters, sloths crossed from North to South America. The land bridge becomes submerged during the Pleistocene, but it reemerged later and formed a continuous connection onward. The bridge allowed many species to cross but it filtered many others. Another major filter bridge was the land connection between Australia and Asia. One more filter bridge is a link between Alaska and Siberia.

iii) A Corridor Bridge: A corridor is board land connection between continents which persist for long geologic periods and allow the free and substantial exchange of species. The best-known corridors are most striking after they no longer exist, that is, after geological events have separated land masses which one were continuous. Simpson has pointed out that New Mexico

and Florida can be regarded as being connected by a corridor at present. The best-known corridor of the past was of the land link between North America and Asia. The same corridor link later became reduced to the filter bridge. During most of the earth history including the whole Mesozoic the land connection between Asia and North America was very extensive. Mesozoic reptiles could cross quite freely and this probably accounts for the present distribution of alligators.

13.5 Biogeographic Regions in India:-

The classification of India is according to biogeographical characteristics. Biogeography is the study of the distribution of species, organisms and ecosystems in geographic space and through geological time. There are ten biogeographic zones in India.

1. Trans-Himalayan zone.
2. Himalayan zone
3. Desert zone.
4. Semiarid zone.
5. Western ghat zone.
6. Deccan plateau zone.
7. Gangetic plain zone.
8. Northeast zone.
9. Coastal zone.
10. Islands present near the shoreline.

1. Trans-Himalayan Region: The Himalayan ranges immediately north of the Great Himalayan range are called the Trans- Himalayas. The Trans-Himalayan region with its sparse vegetation has the richest wild sheep and goat community in the world. The snow leopard is found here, as is the migratory black-necked crane.

2. Himalayan Zone: The Himalayas consist of the youngest and loftiest mountain chains in the world. The Himalayas have attained a unique personality owing to their high altitude, steep gradient, and rich temperate flora. The forests are very dense with extensive growth of grass and evergreen tall trees. Oak, chestnut, conifer, ash, pine, deodar are abundant in the Himalayas.

3. The Desert Zone: There is no vegetation above the snowline. Several interesting animals live in the Himalayan ranges. Chief species include wild sheep, mountain goats, ibex, shrew, and tapir. Panda and snow leopard are also found here.

4. Semi-Arid Areas: Adjoining the desert are the semi-arid areas, a transitional zone between the desert and the denser forests of the Western Ghats. The natural vegetation is thorn forest.

This region is characterized by discontinuous vegetation cover with open areas of bare soil and soil-water deficit throughout the year. Thorny shrubs, grasses, and some bamboos are present in some regions. A few species of xerophytic herbs and some ephemeral herbs are found in this semi-arid tract. Birds, jackals, leopards, eagles, snakes, fox, buffaloes are found in this region.

5. Western Ghats: The Mountains along the west coast of peninsular India are the Western Ghats, which constitute one of the unique biological regions of the world. The Western Ghats extend from the southern tip of the peninsula (8°N) northwards about 1600 km to the mouth of the river Tapti (21°N). The mountains rise to average altitudes between 900 and 1500 m above sea level, intercepting monsoon winds from the southwest and creating a rain shadow in the region to their East.

The varied climate and diverse topography create a wide array of habitats that support unique sets of plant and animal species. Apart from biological diversity, the region boasts of high levels of cultural diversity, as many indigenous people inhabit its forests.

The Western Ghats are amongst the 25 biodiversity hotspots recognized globally. These hills are known for their high levels of endemism expressed at both higher and lower taxonomic levels. Most of the Western Ghat endemic plants are associated with evergreen forests. The region also shares several plant species with Sri Lanka. The higher altitude forests were, if at all, sparsely populated with tribal people. Rice cultivation in the fertile valley proceeded gardens of early commercial crops like areca nut and pepper. The original vegetation of the ill-drained valley bottoms with sluggish streams in elevations below 100m would be often a special formation, the *Myristica* swamp. Expansion of traditional agriculture and the spread of particularly rubber, tea, coffee and forest tree plantations would have wiped out large pockets of primary forests in valleys. The Western Ghats are well known for harboring 14 endemic species of caecilians (i.e., legless amphibians) out of 15 recorded from the region so far.

6. North-West Desert Regions: This region consists of parts of Rajasthan, Kutch, Delhi and parts of Gujarat. The climate is characterized by very hot and dry summer and cold winter. Rainfall is less than 70 cms. The plants are mostly xerophytic. Babul, Kikar, wild palm grows in areas of moderate rainfall. Indian Bustard, a highly endangered bird is found here. Camels, wild asses, foxes, and snakes are found in hot and arid deserts.

7. Deccan Plateau: Beyond the Ghats is Deccan Plateau, a semi-arid region lying in the rain shadow of the Western Ghats. This is the largest unit of the Peninsular Plateau of India. The highlands of the plateau are covered with different types of forests, which provide a large variety of forest products.

8. Gangetic Plain: In the North is the Gangetic plain extending up to the Himalayan foothills. This is the largest unit of the Great Plain of India. Ganga is the main river after whose name this plain is named. The Great Plains cover about 72.4mha area with the Ganga and the Brahmaputra

forming the main drainage axes in the major portion. The thickness of the alluvial sediments varies considerably with its maximum in the Ganga plains. The physiogeographic scenery varies greatly from arid and semi-arid landscapes of the Rajasthan Plains to the humid and per-humid landscapes of the Delta and Assam valley in the east.

Topographic uniformity, except in the arid Western Rajasthan is a common feature throughout these plains. The plain supports some of the highest population densities depending upon the purely agro-based economy in some of these areas. The trees belonging to these forests are teak, sal, shisham, mahua, khair etc.

9. North-East India: North-east India is one of the richest flora regions in the country. It has several species of orchids, bamboos, ferns and other plants. Here the wild relatives of cultivated plants such as banana, mango, citrus, and pepper can be found.

10. Islands and Coastal Zone: The two groups of islands, i.e., the Arabian Sea islands, and Bay Islands differ significantly in origin and physical characteristics. The Arabian Sea Islands (Laccadive, Minicoy, etc.) are the foundered remnants of the old land mass and subsequent coral formations. On the other hand, the Bay Islands lay only about 220 km. Away from the nearest point on the mainland mass and extend about 590 km. With a maximum width of 58 km, the island forests of Lakshadweep in the Arabian Sea have some of the best-preserved evergreen forests of India. Some of the islands are fringed with coral reefs. Many of them are covered with thick forests and some are highly dissected.

India has a coastline extending over 5,500 km. The Indian coasts vary in their characteristics and structures. The west coast is narrow except around the Gulf of Cambay and the Gulf of Kutch. In the extreme south, however, it is somewhat wider along the south Sahyadri. The backwaters are the characteristic features of this coast. The east coast plains, in contrast, are broader due to depositional activities of the east-flowing rivers owing to the change in their base levels.

Extensive deltas of the Mahanadi, Godavari, Krishna and Kaveri are the characteristic features of this coast. Mangrove vegetation is characteristic of estuarine tracts along the coast for instance, at Ratnagiri in Maharashtra. Larger parts of the coastal plains are covered by fertile soils on which different crops are grown. Rice is the main crop of these areas. Coconut trees grow all along the coast.

Fauna of Indian Regions

Fishes: A good number of fishes are found in this region. Osteoglossid, Notopterids, many catfishes, cyprinids, cobitids, homaloptera mandids, pristolepids, anabantids, luciocephalid, mastacembelids, ophiocephalids, and cyprinodonts are present in this region.

Amphibia: 09 families. Nanophys, Cacopus, Tylostrotion, Xenophys, Callula, Toads, tree frogs and true frogs.

Reptiles: 35 families. Dipsas, Pythons, Naja, Trimerusus, Leptotyphlops, Typhlops, sand boas, Homalopsinalis, Sea Snakes, True vipers, Pit vipers, Land tortoises, Crocodilus, Gavialis, many geckos, Chameleons, Skinks and Varanus.

Birds: 71 families. Malcocerus, Garrulax, Sitta, Gallus, Dandrophilla, owls, Hornbills, woodpeckers, Barbets, Cuckoos, Dicrurus, Phyllornis, Munia, Kingfishers Loriculus, Many pigeons, peacocks, and Hoopoes.

Mammals: 35 families. Presbytes, Macacus, felis, Bibos, Antelope, Tarsins, Manis, Loris, Aulurus, Hylobates, Prinodon, Paguna, Simia, Galleopithecus, Hydronys, Gymnura, Mydaus, Elephas, Rhinoceros, Hemigalea, Gymnopus, Melarsus, Indian bears and others (deer, wild pigs, cattle etc.).

13.6 Summary:-

Animal geography or zoogeography is concerned with the distribution of all the animals. There are nearly 10, 00,000 species of animals. It is practically impossible to cover up and study the distribution of each and every species. Most commonly the geography of land and freshwater animals is taken into account, which constitutes just about two percent of the total animal strength. Distribution of animals can be studied at three levels geographical distribution over the whole world, regional distribution in selected segments of the world and local distribution which includes geographical distribution of species in relation to each other and to ecology and evolution. The zoogeography covers the distribution of animals over the whole world.

13.7 Self Assessment Questions:-

1- Palaearctic Region includes

- A-Europe
- B-Asia
- C-South America
- D-North America

Ans- a

2- rats, bats, hawks, cuckoos have

- a- discontinuous distribution
- b- continuous distribution
- c- tandem distribution
- d- none of the above

Ans- b

- 3- The discontinuity in the distribution of animals may be due to:
- a) Reaching the oceanic islands across the water,
 - b) By the submergence of the land mass in between the range, and
 - c) By the extinction of the forms in the intermediate areas.
 - d) all of the above

Ans- d

- 4- The number of Biogeographic region in India
- a- 7
 - b- 8
 - c- 9
 - d-10

Ans- 10

- 5- How many biodiversity hotspots are recognized in the world
- a- 23
 - b- 24
 - c- 25
 - d- 26

Ans- 25

- 6- Which geographical region includes Central and South America
- a-Palaeartic
 - B-Oriental
 - c-Nearctic
 - d-Neotropical

Ans- d

13.8 Suggested Readings and References:-

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13.9 Terminal Questions:-

- 1. Write an essay on the zoogeographical distribution of animals.

2. Describe the principal zoogeographical regions of the world with branches and the name of the fauna of different regions.
3. Write an essay on zoogeographical regions with their characteristic fauna.
4. Give an account of the fauna of Oriental and Ethiopian regions.
5. Write an essay on the bio- geographical regions in India.

13.10 References:-

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UNIT: 14 PERMANENT SLIDE PREPARATIONS

14.1- Objectives

14.2-Introduction

14.3- Method of Microscopic Preparations

14.4. - Methods for slide preparation

14.4.1- Protozoa (Paramecium)

14.4.2- Porifera (Sponge and Gemmules)

14.4.3- Coelenterate (Obelia Colony & Obelia Medusa)

14.4.4- Arthropoda (Mouth parts of Honey Bee, Butterfly, Cockroach and Grasshopper)

14.6- Summary

14.7- Glossary

14.8-References

14.1- Objectives

The study of Permanent slide preparation of Obelia colony: Pharyngeal and septal nephridium of earthworm, parapodia of Nereis and Heteronereis, gill, radula and osphradium of Pila, salivary gland, mouth parts and trachea of cockroach, gills lamina of Uniostacyst and hastate plate of prawn.

14.2-Introduction

A microscope slide is a thin flat piece of glass, typically 75 by 26 mm (3 by 1 inch) and about 1 mm thick, used to hold objects for examination under a microscope. Typically the object is placed or secured ("mounted") on the slide, and then both are inserted together in the microscope for viewing. This arrangement allows several slide-mounted objects to be quickly inserted and removed from the microscope, labeled, transported, and stored in appropriate slide cases or folders. Microscope slides are often used together with a cover slip or cover glass, a smaller and thinner sheet of glass that is placed over the specimen. Slides are held in place on the microscope's stage by slide clips, slide clamps or a cross-table which is used to achieve precise, remote movement of the slide upon the microscope's stage. The following points highlight the seven main processes involved in preparation of permanent slides. The processes are:

1. Killing 2. Fixing and Hardening 3. Staining 4. Dehydration 5. Clearing 6. Mounting 7. Labelling

14.3- Method of Microscopic Preparations

For microscopic studies the specific material (tissue organs or small organism) is mounted on a glass slide. There are two methods of mounting the material on slide.

1. Temporary
2. Permanent

Temporary mounting:-

The temporary mount is prepared either in glycerin, water or normal saline. The material is first washed in tap, then stained and differentiated. Drop of mounting medium (glycerine and water) is placed on center of the slide. The material is then transferred into that drop. It is then covered neatly with a cover slip. The excess of glycerine or water is absorbed by piece of blotting paper. Mount prepared by this method can be used for study only for few hours, after which material loses its original form due to diffusion and other post mortem changes.

Permanent mounting:-

But for the study of microorganisms, smaller animals and histological studies of tissues, an

elaborate technique is employed for making their permanent preparations. These smaller objects are mounted in balsam on a slide. There is a series of processes by which a living organism or its tissue is made fit for microscopic examination in a permanent state. The utility of permanent preparation is that the animal cell or tissue remains as such without undergoing major changes. The permanent preparation includes:

- (1) Killing and narcotization
- (2) Fixing
- (3) Washing
- (4) Staining
- (5) De-staining or removal of excess of stain.
- (6) Clearing or de-alcoholization.
- (7) Mounting on slide.

1. Killing and Narcotization :-

The first step in permanent preparation is killing instantaneously in order to prevent the change in form of the object as it has in living condition and immediately fixing the object. Sometimes killing is preceded by narcotization. The narcotics used are chloroform, menthol, ether, alcohol, acetone, etc. the purpose of narcotization and killing is important as to have the same form and chemically constructed tissue or organisms as it had during its lifetime. In certain cases, for smaller animals killing is heating done by the slide.

2. Fixing :-

Fixing is done with various fixative agents for histological elements. Fixative is essential in every type of microscopic preparations, either for sections or for whole mounts and also in larger specimens. The function of fixation is manifold:

- 1) The tissues become hard and hardening resists further post-mortem changes.
- 2) Fixative agent coagulates and renders insoluble elements of tissues which are dissolved in further processing.
- 3) The fixative agent renders insoluble the various constituent elements of cells, alters their refractive indices and thus makes them optically differentiated under the microscope. Because of Brownian motion there is no possibility of material but we must bear in mind that fixed details are the coagulation artifact of the living structures.

Various fixative agents generally used are absolute alcohol, 90% alcohol plus glycerine, picric acid, corrosive sublimate, formol, osmium tetroxide and nitric acid with or without water. Depending upon the material, corrosive sublimate or alcohol Carnoy's fluid for cytological studies and other fixative for histochemical studies.

3. Washing :-

Washing is essential as by this process the uncombined and excess of fixative agent is removed. The presence of fixative agent in tissues or cells will inhibit good staining. The washing agent depends upon the type of fixative agent used. As alcoholic picric acid in water is removed by 70% alcohol. Formol and corrosive sublimate are washed with water distillate. Sublimate is washed in alcohol.

4. Staining:-

The tissue or cell components are stained in various dyes. The dye makes the tissues distinct in its histological sphere. The various dyes are Orange G, Bordeaux red, Sudan's Congo red, Alizarine oxyquinoline, methylene

blue, neutral red, borax carmine, haematoxylin, picric acid, eosin and Gower's carmine. Mainly two kinds of stains are used.

1. Nuclear stains. Stains the nuclear parts of the cells, such as Delafield's or Ehrlich's haematoxylin.
2. Cytoplasmic stains such as borax carmine, picric acid, eosin, etc., which stain cytoplasm.

For general staining borax carmine is used. Aqueous stains are prepared in water whereas alcoholic stains are prepared in alcohol. When a single stain is used the process is called as simple or single staining. In some cases two stains, i.e., nuclear and cytoplasmic are used and this is called as double staining. Generally single stain is used for whole mounts but for protozoans etc., both cytoplasmic and nuclear stains are used.

Destaining:-

The removal of excess of stain is called as destaining or differentiation. De-staining agents are acid alcohol or acid water. The acid alcohol is used with alcoholic stains while acid water is used with aqueous stains.

5. Dehydration :-

This process is meant for removal of water from the tissues. The dehydration prevents putrefaction or decaying and maintains the same shape and size of tissues or cells. The moisture or water in tissues absorbs various germs of destructive nature so that the tissue may be destroyed, hence the passing the mounting material through various grades of alcohol, such as 30, 50, 70, 90 and 100% alcohols. The tissue is soaked in gradually increasing strengths of alcohol. The lower grades prepared either from 90% or absolute alcohol. The dehydration is carried out in corked or glass-stoppered tubes.

6. De-alcoholization or clearing :-

After dehydration, transparency in tissues is obtained by treating with a clearing agent, which removes alcohol and makes the tissue clear and transparent. The clearing agents are cedar oil, clove oil, xylol and benzol, etc. Xylol is most commonly employed and it makes the tissues hard and brittle. Clove oil is a superior clearing agent especially in the whole mounts. It also possesses higher index of refraction than balsam mounting media.

7. Mounting:-

Mounting forms the end of permanent preparation. The choice of mounting media is not much but they should have the same refractive index as that of the cleared tissue. The refractive index of such a stained, dehydrated and cleared cells is 1.54. Canada balsam or D.P.X has almost the same refractive index. Mounting is an easy process. The tissue is kept over glass slide in a drop of balsam and cover-slip is lowered slightly. After mounting, the excess of balsam on the slide, as generally happens with beginners, should be removed with cotton soaked with the balsam has dried. For much better finishing the edge of the cover-glass may be ringed with a cement such as gold seal or a varnish. The air bubbles present in balsam under cover-glass should be removed by gentle heating.

During all the chemical bathing of tissues, two changes of each reagent are necessary. The time of keeping tissue in various reagents may vary from 5 to 15 minute.

8. Precautions and Instructions:-

1. The articles, such as slide, coverslips and instruments should be perfectly cleaned.
2. The working place should be kept in order.

3. During dehydration, the tissues should be kept in tightly closed cork or glass stoppered tubes. The opened tube will spoil material by absorbing moisture from atmosphere. Even breathing closely with dehydrating tube is undesirable.
4. The change of solution be done very quickly, reducing time of exposure to atmosphere to minimum.
5. The chemicals used once should not be reutilized.
6. The Canada balsam used should be clean, dust-free and not viscous.

14.4. - Methods for slide preparation

14.4.1- Protozoa (Paramecium)

Classification:-

Phylum	Protozoa	→	Unicellular
Sub-Phylum	Ciliophora	→	Ciliary movement in all stages.
Class	Ciliata	→	Cilia present throughout life.
Sub Class	Euciliata	→	Cytopharynx, contractile vacuole, mega and micronucleus present
Order	Holotricha	→	Equal cilia.
Sub-order	Trichostomata	→	Mouth leads in cytopharynx.
Family.....	Paramecidae	→	Oral groove present
Genus.....	Paramecium		

Culture preparation of Paramecium:-

It is found abundantly in the ponds and ditches in decaying vegetation. For culturing paramecia boil 20 grains of wheat plus 20-25 hay steams in 500 cc of distilled water for about 10 minutes. Keep it in dark and cool place for about four days and inoculate it with few paramecia by a micropipette, within little days. The culture will found to contain numerous paramecia.

Examination in living condition:-

Take a clean slide .Through the micropipette put a drop of water from the culture medium of Paramecium Examine the slide under low magnification of compound microscope .Observe the fast moving Paramecia and their cytopharynx.

Many protozoans' move very fast. So, they must be slowed down for proper examination. This is done in three ways:

1. Protozoan's are slowed in 10per methyl cellulose solution. Dissolve 10 gm of methyl cellulose solution50cc of water.Boil , cool and make upto 100 cc .The solution slows down the movement.
2. 2. % sodium carboxymethyl cellulose solution is also good for slowing downh protozoan movement.Boil 2gm of sodium methyl cellulose.Cool.
- 3 .Nickel sulphate acts as anaesthetic .By keeping the animal for 15 min can restrict their movement.

Permanent preparation: -

For the free living and fast moving protozoans, they are first made non motile on a glass slide coated with albumin. Then the small drop of culture containing *Paramecium* is fixed with an equal drop of 1% of Agar solution melted (1gm of Agar in 100 cc of water ditillate) at 45⁰ C. The solution become jelly like. The animal may survive for 30 min. They are fixed with 90% alcohol or by a drop of Schaudinn's fixative.

Pass the slide through descending grade of alcohol 90%, 70%, 50% and 30% and distilled water. Stain both nuclei and cytoplasm by double staining. Stain first with Ehrlich's haemotoxylin. Destain in acid water and wash in tap water. Again dehydrate in ascending grade of alcohol. After 90% alcohol stain in cytoplasmic Eosin. Keep in 100% alcohol, Clear in xylol and mount on D.P.X.

- 1) **Feeding experiment:** As *Paramecium* is a ciliary and selective feeder. The cilia direct the food particles into the cytopharynx or gullet. Its food particles consist of bacterial etc. The food is collected into membranous vesicle which is formed just below the gullet. When the vesicle is filled with food it is detached and is called food vacuole. In *paramecium* food particle is circulated in the body by more or less definite path by slow streaming movement of endoplasm called cyclosis. Digestion and assimilation take place during the journey of food vesicle, First it is alkaline and then acidic and again alkaline.

For observing cyclosis: Take a drop of culture medium of *Paramecia* over a slide. Add a little yeast Congo red in a drop of water. The Congo red is taken into the food vacuole. Observe under low magnification along with the movement of Congo red in Food vacuole.

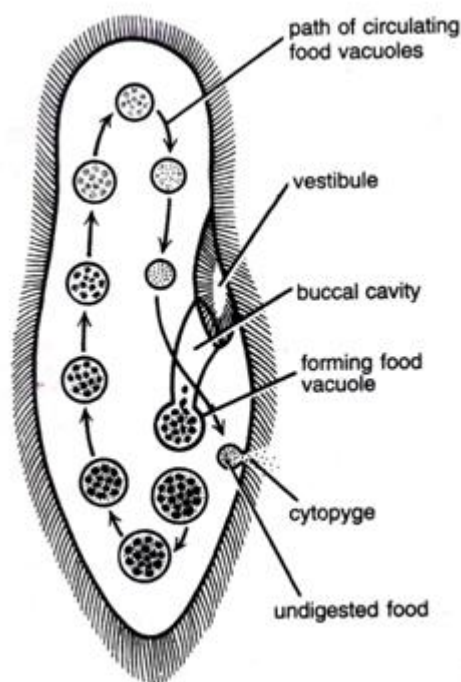


Fig.14.1 *Paramecium* showing Cyclosis

Distribution: It has cosmopolitan distribution.

Habit and Habitat:

Paramecium caudatum is commonly found in freshwater ponds, pools, ditches, streams, lakes, reservoirs and rivers. It is specially found in abundance in stagnant ponds rich in decaying matter, in organic infusions, and in the sewage water.

Paramecium caudatum is a free-living organism and this species is worldwide in distribution.

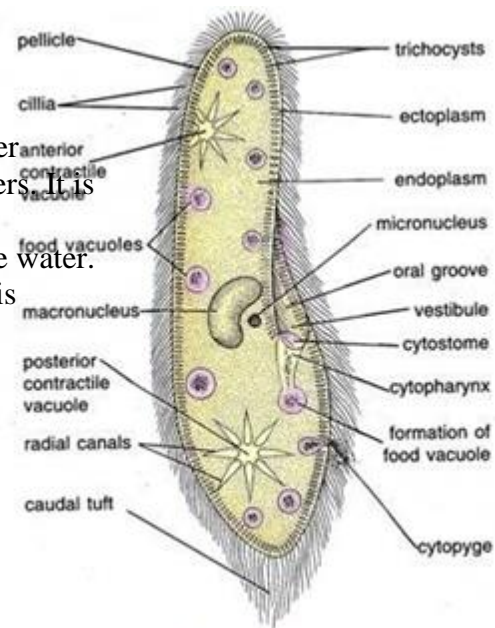


Fig.14.2 PARAMECIUM

Comments:-

1. Commonly called as slipper animalcule, being microscopic, elongated slipper-shaped, cigar-shaped or spindle shaped.
2. Most familiar and extensively studied protozoans.
3. Anterior end is bluntly rounded, while posterior end is pointed.
4. *P. caudatum* measures 80 to 350 microns, while *P. aurelia* 170 to 290 microns.
5. Pellicle covers the body. It is clear, firm and elastic cuticular membrane. Pellicle has series of polygonal or hexagonal depressions for trichocysts.
6. Cilia covers the entire animal. They are hair-like projections of uniform length, except at posterior end where they are longer and at cytopharynx where they form undulating membrane.
7. Infraciliary system consists of basal bodies and kinetodesmata.
8. Cytoplasm contains ecto- and endoplasm. Ectoplasm has myonemes and rod-shaped trichocysts. Endoplasm contains food vacuoles, granules, meganucleus, micronucleus, anterior contractile, posterior contractile vacuole, fat and glycogen.
9. Trichocysts are rod-shaped bodies consisting of lower trichocyst shaft, basal body and projecting cilium. Cilium project through the hexagonal areas. Trichocysts are discharged to anchor with substratum.
10. Reproduction is by binary fission, conjugation, endomixis, hemixis and automixis.
11. Locomotion is ciliary. Nutrition is holozoic and it shows response to light and temperature, etc.

Identification: Since the animal contains slipper-shaped body and 2 contractile vacuoles which are star-shaped and has all above features, hence it is Paramecium.

14.4.2- Porifera (Sponge and Gemmules)

Spicules of Sponges:-

Introduction:-

The body wall of sponges is supported by various minute crystalline and calcareous bodies called as spicules. These are secreted by special mesenchymal cells called scleroblasts.

Spicules provide taxonomic characters and are classified according to the axes and rays called as axon, actine and actinal respectively. These are of two types:

- 1) Megascleres-Support skelton
- 2) Microscleres-Smaller and none supporting.
- 3) These are of following types
 - a) Monaxon-consist of single axis, straight or curved
 - b) Tetraxon-Consist of four rays
 - c) Triaxon-consists of three axes
 - d) Polyaxon-Having several equal rays

Spicules generally support and protect the body and helps in identification classification and metabolism.

Method for Slide preparation:-

For extraction of spicules, boil a small portion of sponge in 15-20% potassium hydroxide solution in a test tube till cells are dissolved. The spicules settle in the bottom. Decant the KOH solution and wash the spicules several times in tap water. Pass the spicules in ascending series of alcohol, 30%, 50%, 70%, 90% and 100% alcohol. Dealccoholize or clear with xylol and mount on a slide after pipetting the spicules. There is no need of staining. Study under the microscope and note different type of spicules as monaxon, triaxon, tetraxon etc.

Comments:

1. Sponge body wall is supported by various minute, crystalline and calcareous bodies called as spicules, which are secreted by special mesenchymal cells called as scleroblasts.
2. Spicules provide taxonomic character and are classified according to the axes and rays, spoken of as axon, actine or actinal respectively.
3. Spicules are of two types: (i) Megascleres or supporting skeleton, (ii) Microscleres small and non supporting. Kinds of Megascleres are as follows;
 - (i) **Manaxon** consists of a single axis, straight or curved. They may be styles, rhabds and tylots.
 - (ii) **Tetraxon** consists of four rays. It also includes triradiate or triactinal spicules.
 - (iii) **Polyaxon** having several equal rays. Amphidisk spicules are found in fresh water sponges. In this type, the rhabdom contains disks at both ends. The arrangement of different types of spicules could be seen in Sycon.
4. **Microscleres** are found throughout the mesenchyme and include spires and asters.
5. Spicules support and protect the body. They are helpful in identification, classification and metabolism.

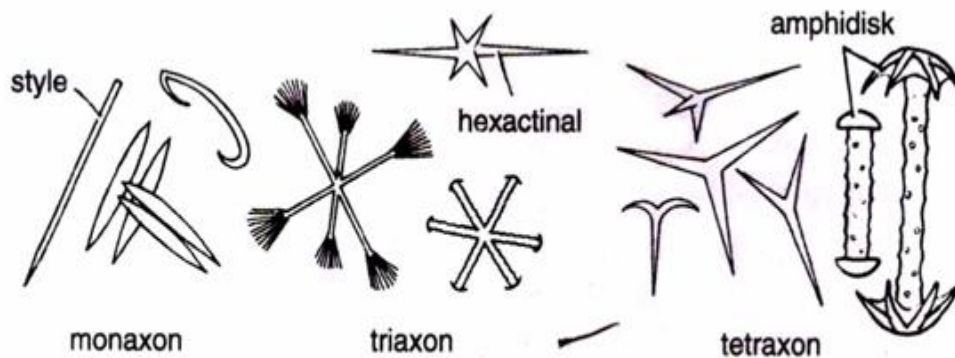


Fig.14.3 Spicules

Identification:-

The clear transparent monaxon or triaxon spicules indicate **Spicule** of sponges.

Gemmules:-

Gemmules are asexual reproductive bodies forming a part of regular life cycle. These are endogenous buds which are diagnostics of Porifers, especially of freshwater and a few marine sponges. Gemmulation or endogenous budding is a peculiar mode of reproduction under unfavourable condition such as excessive cold or drought.

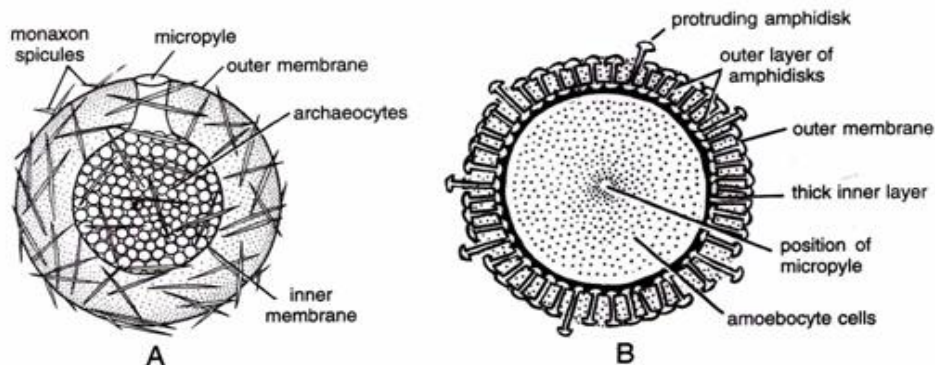


Fig. 11.4. Gemmule of *Ephydatia*. B. Gemmule of *Spongilla*.

Comments:

1. Gemmules are asexual reproductive bodies forming a part of regular life-cycle .
2. Gemmules or endogenous buds are diagnostic of Porifera and especially of fresh-water and a few marine sponges.
3. Gemmulation or endogenous budding is a peculiar mode of reproduction under unfavourable conditions such as excessive cold or draught.
4. Gemmules contain outer and inner membrane.
5. Gemmule is rounded structure formed by the aggregation of archaeocytes into groups accompanied by trophocytes which are impregnated with food particles of glycoproteins or lipoproteins.

6. **Scleroblasts** secrete the **amphidisk spicules**, which form a row in columnar layer between outer and inner membrane
7. Gemmules are resistant to external factors such as freezing and drying. Gemmules of fresh water sponge can be kept for 2 years.
8. They hatch at a temperature of 13-21°C in about 3 days. After hatching, a gemmule gives rise to a young sponge.
9. A full grown gemmule is usually pierced by opening on one side, called a **micropyle**.

Identification: Since the material has micropyle in mature and amphidisk spicules in immature gemmules and has above all features, hence it is Gemmule whole mount.

14.4.3- Coelenterate (Obelia Colony & Obelia Medusa)

OBELIA:-

Obelia is colonial, mainly sedentary hydrozoan zoophyte attached to the seaweed, hills and rocks. It is mostly found in shallow water and also up to approximately 250 ft. deep.

Method for slide preparation:-

Coelenterates are first narcotized in water mixed with menthol crystal or Magnesium sulphate. After decanting the narcotizing liquid, fix the animal by adding drop by drop formalol (commercial preparation). These are then preserved in 70% alcohol or 5% formalin solution.

For making permanent mount, keep the material in 70% alcohol, then stain in borax carmine, if overstain, destain with acid alcohol. Dehydrate in 70%, 90% and 100% alcohol. Clear in xylol or benzene and finally mount on Canada Balsam. Then study under the microscope, draw the diagram, label them and note down the characteristic features.

OBELIA COLONY:-

Classification:

Phylum	Coelenterata-	Tissue grade, diploblastic and acoelomate.
Class	Hydrozoa	- Hydroids: medusa with velum.
Order	Hydroidea	- Polypoid generation well developed
Sub order	Calyptriblastea	- Hydranths have hydrotheca and gonophores with gonotheca.
Genus	Obelia	

Habit and habitat:

Obelia is colonial, marine, sedentary hydrozoan zoophyte, attached to seaweeds, shells and rocks.

Distribution:-

Its range is from the Arctic region to the Gulf of Mexico and the Pacific coast, and from Southern California to Oregon. It is found in shallow water and also up to approximately 250 feet deep.

Comments:-

- 1) It is a dimorphic colony in the form of small seaweed filaments, measuring several cm in height. The filaments may be horizontal and vertical. The colony consists of several parts.

- 2) **Hydrorhiza**: It is basal or horizontal portion called as stolon or rhizostome, which is meant for attachment to substratum. Hydrorhiza gives vertical branches called hydrocaulus.
- 3) **Hydrocaulus** gives alternate branches that terminate into individual zooids called as polyps and medusa.
- 4) **Coenosarc**: Stems and zooids are made of a living hollow, cellular tube called as coenosarcs. It is made up of ectoderm, endoderm and mesogloea.
- 5) Stems and zooids are made up of two components : (i) Outer protective tough, transparent non-cellular covering called as **perisarc** (ii) **mesogloea** (iii) inner living hollow cellular tube called **coenosarcs**.
- 6) Zooids consist of polyp and medusa.
- 7) Medusa grows at the base of polyp-bearing branches and is enclosed in blastostyles. Medusa is composed of upper exumbrellar and lower sub-umbrellar surfaces, manubrium and gonads. Free medusa occurs in the life cycle. It is a reproductive zooid.
- 8) Polyp is a bell-shaped cup made up of lower cub-shaped hydrotheca and upper hypostome. Hypostome is a feeding zooid having circlet of 24 nematocyst bearing tentacles.
- 9) Growth of the colony is sympodial, i.e., each new hydranth arises as bud from the stem, just proximal to the next youngest polyp.
- 10) It reproduces asexually and sexually.

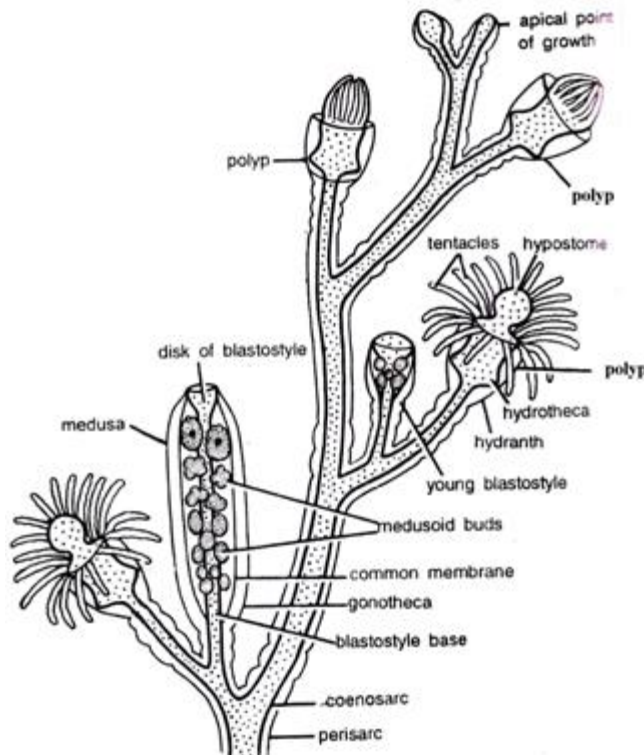


Fig. 14.5 Obelia colony

Identification:

The colony has alternate branches of polyps, blastostyles and all above features, hence it is **Obelia**.

Obelia: Medusa:-

Comments:

- 1) Medusa is a modified zooid for sexual reproduction.

- 2) It is a solitary free-swimming zooid, originating from blastostyles.
- 3) Medusa is umbrella-like and has convex exumbrellar and concave sub-umbrellar surfaces with well defined radial symmetry.
- 4) Umbrellar edge contains radially symmetrical tentacles.
- 5) Base of fully grown tentacle is thickened to tentacular bulb which contains a number of stinging cells.
- 6) In the four radial positions each tentacular bulb contains two otocysts, which are hollow and balancing organs containing calcareous otoliths.
- 7) **Manubrium** hangs from the centre of sub-umbrella, having mouth.
- 8) Mouth communicates with 4 radial canals which join with circular canal lining umbrellar margin which all around contains velum.
- 9) Beneath the radial canals are gonads lying in **Sub-umbrellar** ectoderm.

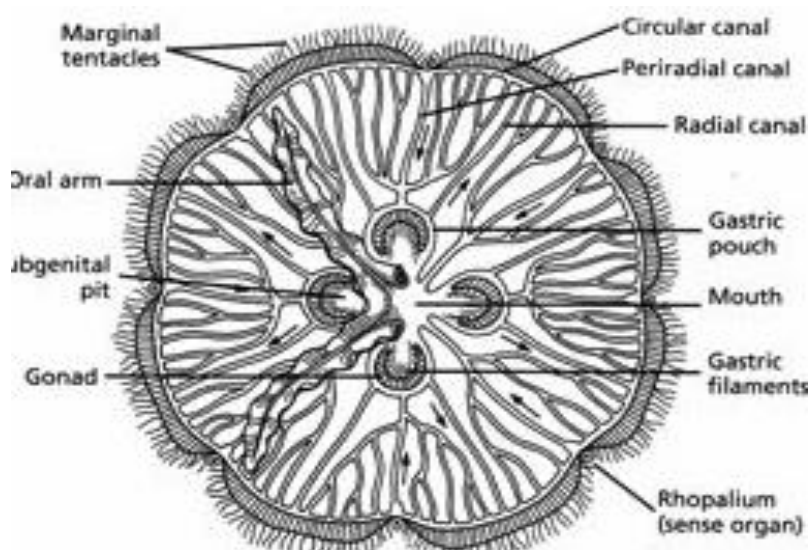


Fig.14.6 Medusa: Obelia

Identification:

Since the mount has circular tentaculated body, 4-radial rounded gonads and all above features, hence it is obelia medusa a very favourite slide-spot.

14.4.4- Arthropoda (Mouth parts of Honey Bee, Butterfly, Cockroach and Grasshopper)

MOUTH PARTS OF INSECTS:-

Insects constitute the largest group of animals in the Animal Kingdom. They have developed different feeding habits as their food differs variously. So for this purpose, they have got certain appendages in their head around the mouth; these appendages together constitute the mouth parts. The mouth parts of insects, therefore, grouped into two main categories; chewing or mandibulate type and sucking or suctorial type

Basically, the mouth parts of insects include a pair of mandibles, a pair of labium or first maxillae and the lower lip represented by and the maxillae and the lower lip represented by fused second pair of maxillae. In chewing type of mouth parts, the mandibles are well developed and the maxillae are simple as

found in Orthopterans like cockroaches and grasshopper. These mouth parts are adapted for cutting or biting and chewing or crushing the food. In suctorial type of mouth parts, the mandibles are vestigial, e.g., lepidopteron or absent, e.g., housefly or blade-like, e.g., honeybee or in the form of piercing needles or stylets, e.g., mosquito. The maxillae, however, exhibit modifications in various ways for piercing and sucking the food.

The mouth parts of insects are, however, classified into following five types:

Chewing type: -

These consist of the labrum forming upper lip, mandibles, first maxillae, second maxillae forming lower lip, hypopharynx and the epipharynx. The labrum is median, somewhat rectangular flap-like. The mandibles are paired and bear toothed edges at their inner surfaces; they work transversely by two sets. The first maxillae are paired and lie one on either side of the head capsule behind the mandibles. Each possesses a five-jointed maxillary palp which is a tactile organ. The first maxillae help in holding the food. The second maxillae are paired but fused to form the lower lip. Its function is to push the masticated food into the mouth. The hypopharynx is a single median tongue-like process at whose base the common salivary duct opens. The epipharynx is a single small membranous piece lying under the labrum and bears taste buds. This type of mouth parts are found in Orthopteran insects like cockroaches, grasshoppers, crickets, etc. These are also found in silver fish, termites, earwigs, beetles, some hymenopterans and in caterpillars of Lepidoptera.

2. Chewing and lapping type. –

This type of mouth parts are modified for collecting the nectar and pollen from flowers and also for moulding the wax, as is found in honeybees. They consist of the labrum, epipharynx, mandibles, first pair of maxillae and second pair of maxillae. The labrum lies below the clypeus, below the labrum is a fleshy epipharynx which is an organ of taste. Mandibles are short, smooth and spatulated, situated on either side of the labrum; used in moulding wax and making the honeycomb. The labium (second pair of maxillae) has reduced paraglossae, the glossae are united and elongated to form the so called retractile tongue, at its tip is a small labellum or honey spoon. The labial palps are elongated. The glossa is used for gathering honey spoon. The labial palps are elongated. The glossa is used for gathering honey and it is an organ of touch and taste. The first pair of maxillae are placed at the sides of labium, they bear small maxillary palps, lacinia is very much reduced but galea are placed at the sides of labium, they bear small maxillary palps, lacinia is very much reduced but galea are elongated and blade-like; The galea and labial palps form a tube enclosing the glossae which moves up and down to collect nectar from flower nectarines. The nectar is sucked up through the tube, so formed, by the pumping action of the pharynx. The labrum and mandibles help in chewing the food.

3. Piercing and sucking type-

This type of mouth parts are adapted for piercing the tissues of animals and plants to suck blood and plant juice, and found in dipteran insects like mosquitoes and hemipteran insects like bugs, aphids, etc. They usually consist of labium, labrum and epipharynx, mandibles, maxillae (1st pair) and hypopharynx.

4. Sponging type:-

This type of mouth parts are adapted for sucking up liquid or semiliquid food and found in houseflies and some other flies. They consist of labrum-epipharynx, maxillae, labium and hypopharynx; mandibles are entirely absent. In fact,

in this type of mouth parts, the labium, i.e., lower lip is well developed and modified to form a long, fleshy and retractile proboscis. The proboscis is divisible into three distinct parts: (i) rostrum or basiproboscis: it is broad, elongated and cone-shaped basal part of proboscis articulated proximally with the head and bears a pair of elongated and cone-shaped basal part of proboscis articulated proximally with the head and bears a pair of unjointed maxillary palps representing the maxillae, (ii) haustellum or mediproboscis; it is the middle part of proboscis bearing a mid-dorsal oral groove and a ventral weakly chitinized plate-like theca or mentum. A duct and closes the grooved of labrum epipharynx form below. The labrum-epipharynx is a long, somewhat firmed and grooved structure covering the oral groove. The food canal or channel is, thus, formed by labium – epipharynx and the hypopharynx and (iii) labella or distiproboscis; it is the distal part of proboscis and consists of two broad, flattened and oval spongy pads having a series of channels pseudotracheae. These open externally by a double row of tiny holes through which liquid food is taken in.

5. Siphoning type:-

This type of mouth parts are adapted wonderfully for sucking flower nectar and fruit juice, found in butterflies and moths belonging to the order Lepidoptera of class- Insecta. They consist of small labrum, coiled proboscis, reduced mandibles and labium. The hypopharynx and epipharynx are not found.

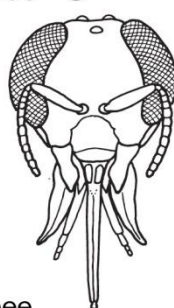
Insect mouthparts

sucking



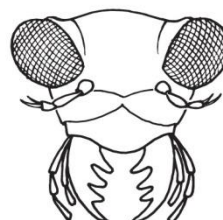
butterfly
(side view)

lapping

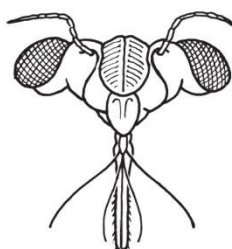


bee
(front view)

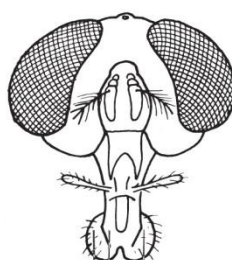
chewing



beetle
(front view)



cicada
(front view)



housefly
(front view)



grasshopper
(side view)

Fig.14.7 Mouth parts of Insects

Slide preparation method:-

For making permanent mount of mouth parts of honey bee, butterfly and cockroach, first cut the head of the above insects. Boil the head in 5% KOH for some time, till the chitin is dissolved. Then wash in water, dehydrate in 30%,50%,70% alcohol. Stain in picro-indigo carmine or acid fuchsin, dehydrate in 90% and absolute alcohol. Clear in xylol or benzene and finally mount in Canada balsam. Study under the microscope draw the diagram and note down the characteristic features,

Butterfly: - Head and Mouth Parts

Comments:-

- 1) Butterfly, belonging to order Lepidoptera, contains siphoning or sucking mouth parts. Head may be examined under binocular microscope for mouth parts.
- 2) Head of butterfly is composed of large compound eyes and antennae. It is broad and contains siphoning type of mouth parts.
- 3) Mouth parts are composed of small labrum in front of clypeus, triangular labium and coiled proboscis.
- 4) Mandibles are absent
- 5) Proboscis is composed of elastic cuticle and greatly elongated galeae of maxillae, grooved internally forming food canal for nectar.
- 6) Proboscis lies in coiled stage, but it immediately uncoils and protrudes in response to a food stimulus, due to rise in blood pressure.
- 7) Labium is triangular and plate-like containing labial palps.
- 8) Other joints of maxillae and maxillary palps are reduced or vestigial.
- 9) Head contains ventral groove for proboscis.

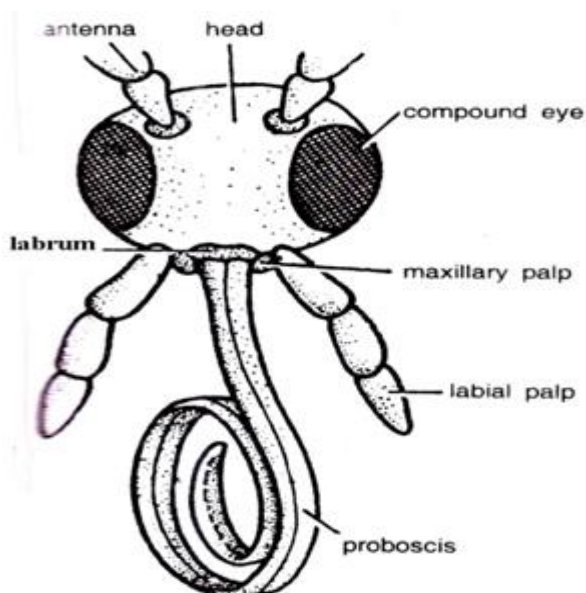


Fig 14.8 Head and mouth parts of butterfly.

Identification:-

Since the mouth contains coiled proboscis, hence it is the mouth parts of butterfly.

APIS: -**Honey – Bee: Mouth Parts of Worker :-****Comments:**

- 1) Honey-bee belonging to the order Hymenoptera contains rasping and lapping mouth parts, adapted for collection of nectar and pollen.
- 2) Head is triangular, containing large compound eyes, 3 ocelli antennae and mouth parts.
- 3) Mouth parts are composed of spoon shaped mandibles, labrum and maxillae devoid of lacinia.
- 4) Mandibles are smooth and spatulate type, food on either side of the labrum.
- 5) It contains vestigial maxillary palps and blade-like galea.
- 6) Labellum is spoon shaped, grooved internally forming a tube and is called as tongue.
- 7) Epipharynx is soft and triangular lying below the labrum. Cardo and stipes are well developed.
- 8) Liquid food taken along tongue is converted into honey in honey-sac by enzymes from salivary glands.
- 9) Prementum contains segmented labial palps, Paraglossae and glossae.
- 10) Honey-bee also moulds waxes in its hive.

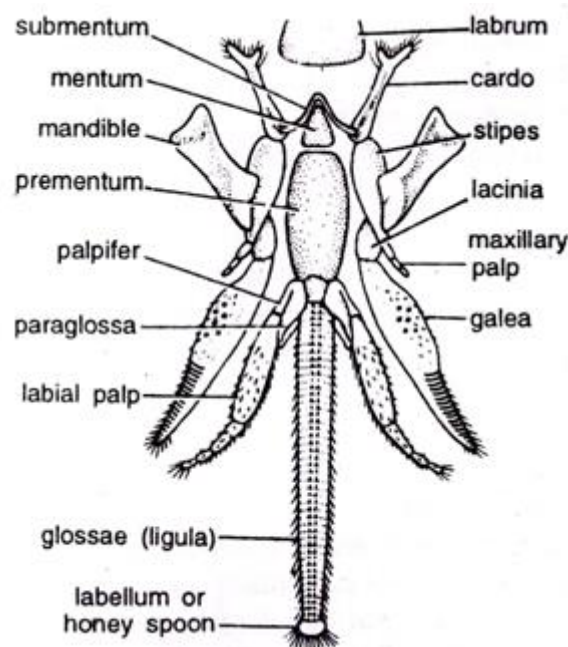


Fig 14.9 Apis Honey-bee Mouth parts of worker.

Identification: -

Since the mouth contains spoon-shaped labellum, hence these are mouth parts of worker, honey-bee.

COCKROACH: HEAD AND MOUTH PARTS

Comments:-

1) Cockroach, belonging to order- Orthoptera, contains chewing mouth parts.

Head is dorso-ventrally elongated and is composed of antennae, large compound eyes and mouth parts.

2) Mouth parts consist of (i) labrum, (ii) mandibles and (iii) maxillae.

3) Labrum protects the mouth. Mandibles are simple and toothed.

4) Maxilla has two part-cardo and stipes. Stipes contains internally lacinia, medially galea and externally maxillary palp.

5) Labium is composed of submentum, postmentum and prementum.

6) Prementum carries glossa internally, Paraglossa medially and palpiger externally.

7) Maxillary and labial palps are tasting organs.

Identification:-

Since the mount shows definitely arranged various parts especially labium maxilla and all above features, hence it is mouth parts of cockroach.

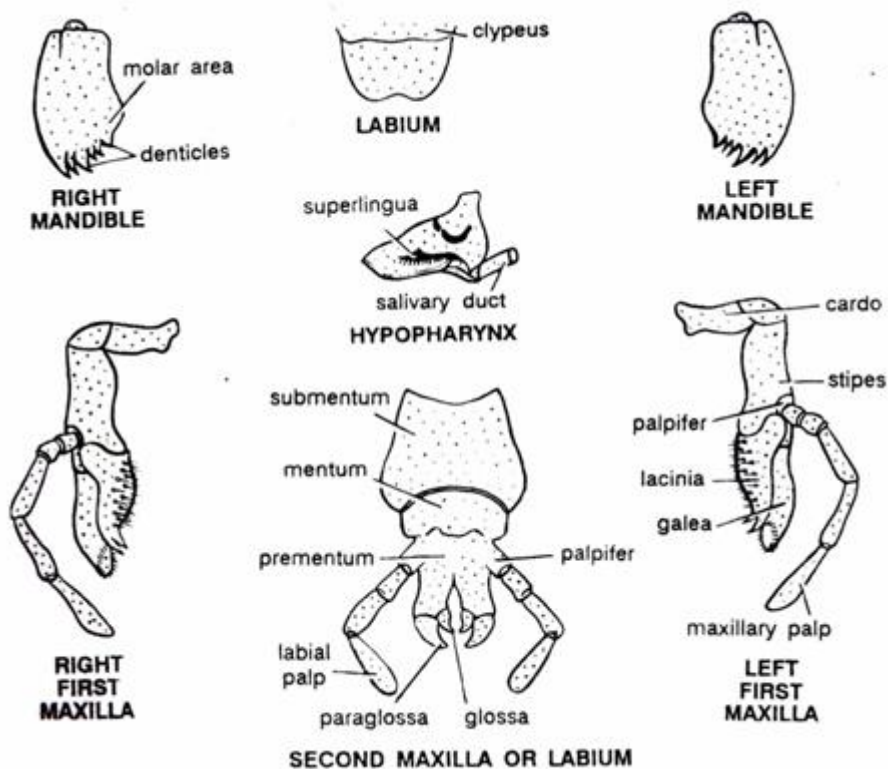


Fig.14.10 Mouth parts of Cockroach

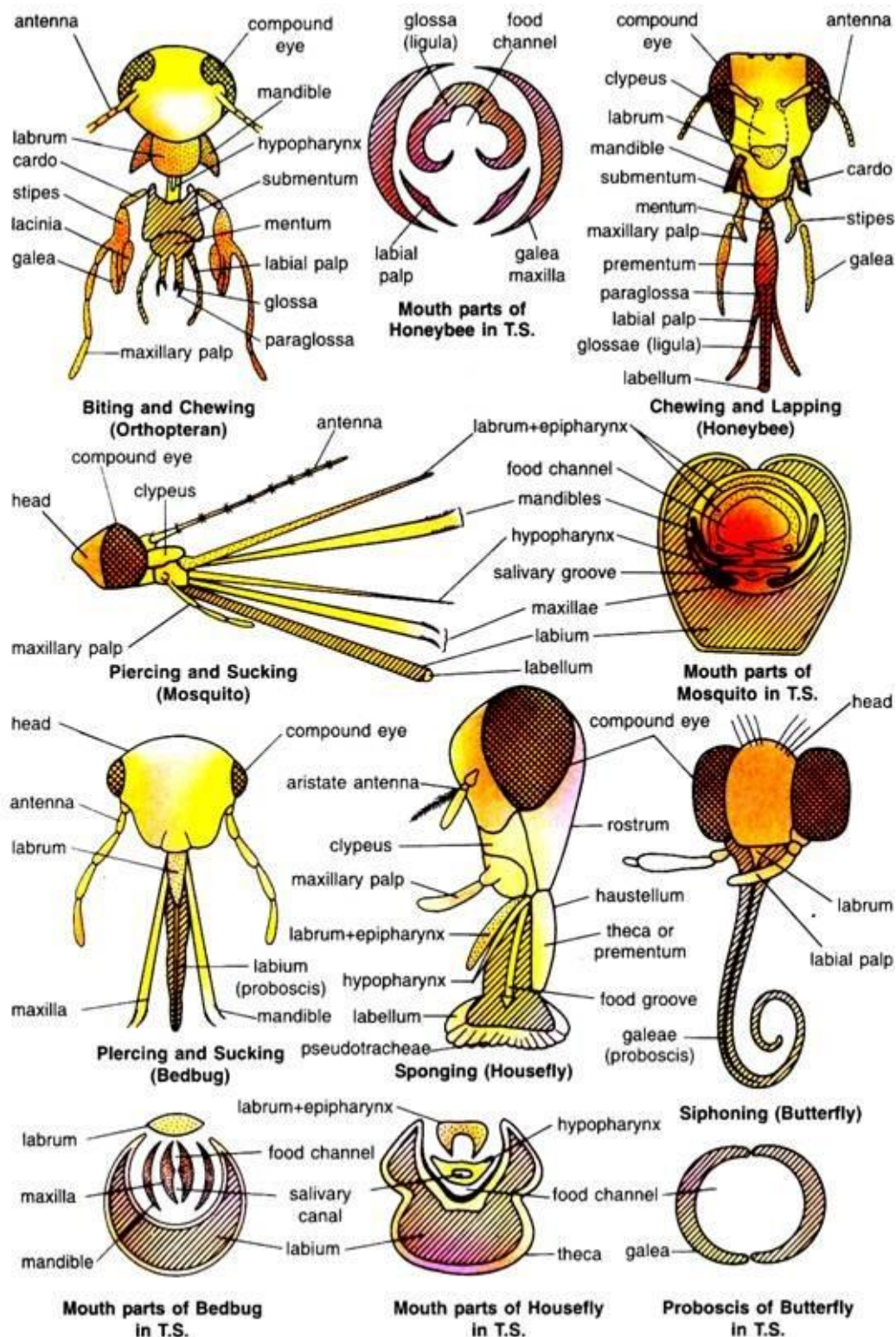


Fig14.11.Mouth parts of insects

APPENDAGES IN COCKROACH:-

(1) Thorax contains 3 pairs of waling legs, which bear dense hairs. Each leg is composed of coxa, trochanter, femur, tibia an 5-jointed tarsus, which ends in a pulvillus (Fig. 147

A. first pairs of legs:

(2) It is found in the prothoracic region; inner surface of **tibia** bears **pollen brush**; **posterior surface of tibia** contains a velar process, which fits into the tarsal notch; and the bristles of the tarsal notch form **antenna comb**.

(3) Anterior edge of the first tarsal segment contains **eye brush** to remove particles from the eyes.

B. Second pair of legs:

(4) It originates from **mesothorax** containing 5 **podomers**.

(5) Inner end of Tibia bears spine-like **pollen spur** for removing pollen from the pollen basket. The outer surface bears **pollen brush**. Terminal part of tarsus contains **pulvillus** and **claw**.

C. Third pair of legs:

(6) It originates from the **metathorax**. The proximal tarsus contains stiff hair, which help in removing the pollen from the body. The tibial podomere is slightly concave and is fringed with long hairs to forming **pollen basket** or **corbicula**.

(7) Distal end of tibia has stiff bristles called as **pecten**. Just below pecten is a plate like structure called **auricle**. Pecten and auricle form **wax pincher** for removing wax from **abdomen**.

(8) Outer surface of tarsus has **pollen brush** while inner surface has **pollen comb** or **scopa**. Terminal segment of tarsus contain **claw** and **pulvillus**.

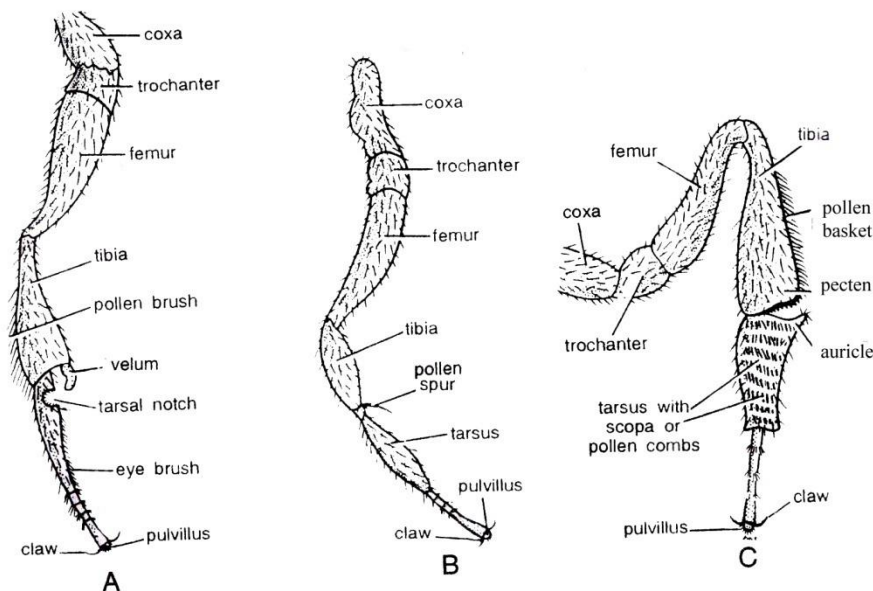


Fig. Legs of honey-bee. **A.** First leg, **B.** Second leg, **C.** Third leg.

HONEY BEE-STING APPARATUS:-

Comments:

- (1) **Sting apparatus** of honey-bee is a modified **ovipositor**, found at the posterior extremity of abdomen in queens and workers)
- (2) It is composed of **sting** or **terebra**, **bulb**, **levering plates** and **glans**.
- (3) Sting is made up of 2 pairs of **gonapophyses** : those of the 8th segment forming **stylets** and of the 9th segment **stylet sheath**, which enclose **poison canal**.
- (4) Distally the stylet sheath and **stylet** contain **pointed spines** or **barbs**.
- (5) Stylet sheath is expanded into the bulb at the base of the sting.
- (6) There are 3 pairs of plates. The anterior one is triangular **fulcral plate**, the **postero-dorsal** is **quadrate plate** and the innermost is **oblong plate** bearing sting palp.
- (7) There are two glands namely **poison gland**, opening into the poison-sac and a small **alkaline gland**, opening into sting bulb. The bite of the **sting** causes burning sensation, pain and swelling of the part concerned.

Identification: Since the mount contains sting and poison gland hence it is sting apparatus of *Apis*.

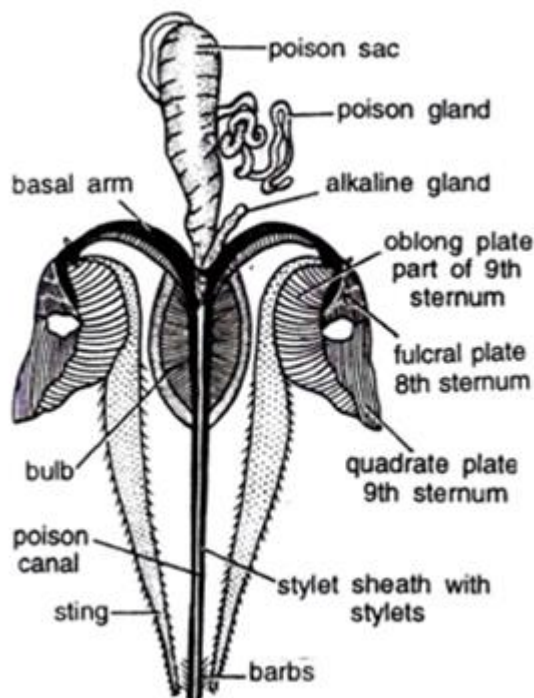


Fig.14.14 sting apparatus of Honey Bee

Minor Dissections:-

Nervous system of cockroach:-

- Procedure:** (i) Take a freshly-killed cockroach for dissection; remove wings, cut off antennae and legs close to their bases
- (ii) Hold cockroach in left hand and cut the lateral membranes between terga and sterna up to the anterior edge of pronotum.

(iii) Lay the insect in the dissecting dish with dorsal side uppermost and pin it in abdominal sterna and coxae of legs. (Another better procedure of fixing cockroach is to float it in petridish containing hard melted wax. Allow it to cool and in due course the animal will be embedded and dissection can be done.) Fix the head by pinning between mandibles by means of fine scissors make a rectangular cut in the head around clypeus and anterior epicranium to expose two cerebral ganglia.

(iv) Make a transverse cut along the posterior edge of the ninth segment (tergum) and gently remove other segment very carefully, so that the underlying organs and tissues are not disturbed.

(v) Uncoil intestine and stretch alimentary canal on one side. Remove fat bodies, tracheae and other muscles to expose internal organs. Study and draw the following parts:

(1) **Heart** :-

13 **chambers** in number (3 thoracic and 10 abdominal narrow chambers). Note inter-segmental alary muscles.

(2) **Alimentary canal** :-

(3) Is divided into three parts:

(a) **Foregut**: It comprises of **mouth, buccal cavity, oesophagus, crop and gizzard**. The buccal cavity, receives the common salivary duct. Crop is meant for storing food. The gizzard has chitinous lining, which is internally produced into six teeth for masticating the food and setae for straining the food.

(b) **Mesenteron or midgut**: It is a narrow duct originating from gizzard and midgut there are 7 to 8 **hepatic** or **mesenteric caeca**. (Their function is to increase the absorptive area).

(c) **Hindgut or proctodaeum**: It includes ileum, colon and rectum. The beginning of ileum is marked by 60-70 fine and long **greenish yellow Malpighian tubules** (excretory in function).

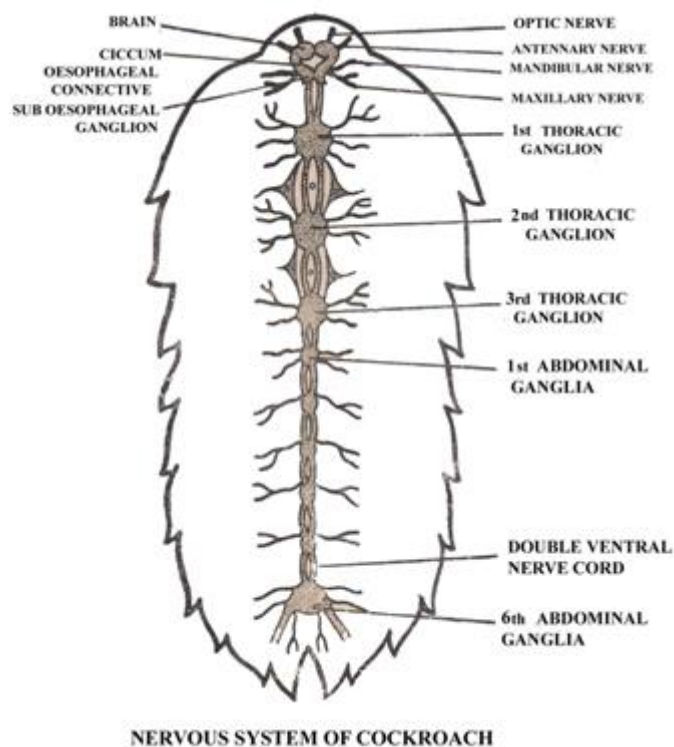


Fig.14.14

Nervous system of Grasshopper:-

Procedure: Take a freshly chloroformed or preserved grasshopper, cut wings and fix the animal with dorsal side upwards. Make incision in pleura and remove the targal sclerites. Remove terga in head region. Now carefully remove the viscera and expose as clearly as possible the entire nervous system. Start from posterior side and gradually trace the nerve cord up to brain. Observe the following parts:

Entire nervous system is divided into 3 parts:-

1. **The central nervous system:** It consists of a dorsal brain or supra-oesophageal ganglia situated above oesophagus between eyes and connected to ventral sub-oesophageal ganglion by circum-oesophageal connectives (Fig. 32).

Sub-oesophageal ganglion is formed by the fusion of mandibular, maxillary and labial ganglia. It gives rise to double ventral nerve cord which extends upto posterior region and shows the following thickenings or ganglia:

- (1). **First thoracic ganglion.**
- (2). **Second thoracic ganglion.**
- (3). **Third thoracic ganglion,** and
- (4). **Five pairs of abdominal ganglia.**

2. **Peripheral nervous system:** The following nerves arise from central nervous system:

- (1) A pair of **optic nerves** originates from optic lobes and supplies to **antennules**.
- (2) **Ocellary nerves:** They innervate ocelli.
- (3) A pair of **antennary nerves** originates from thoracic ganglia.
- (4) **Walking leg nerves.** They originate from thoracic ganglia.
- (5) **Abdominal nerves** arise from abdominal ganglia and supply to various organs.

3. **Sympathetic nervous system:** it includes occipital ganglion, frontal ganglion and ingluvial ganglion, which are associated with brain and control involuntary actions of alimentation, heart ganglion, frontal aorta and genital organs.

Instructions: Draw the diagram of your dissection with the help of the practical book.

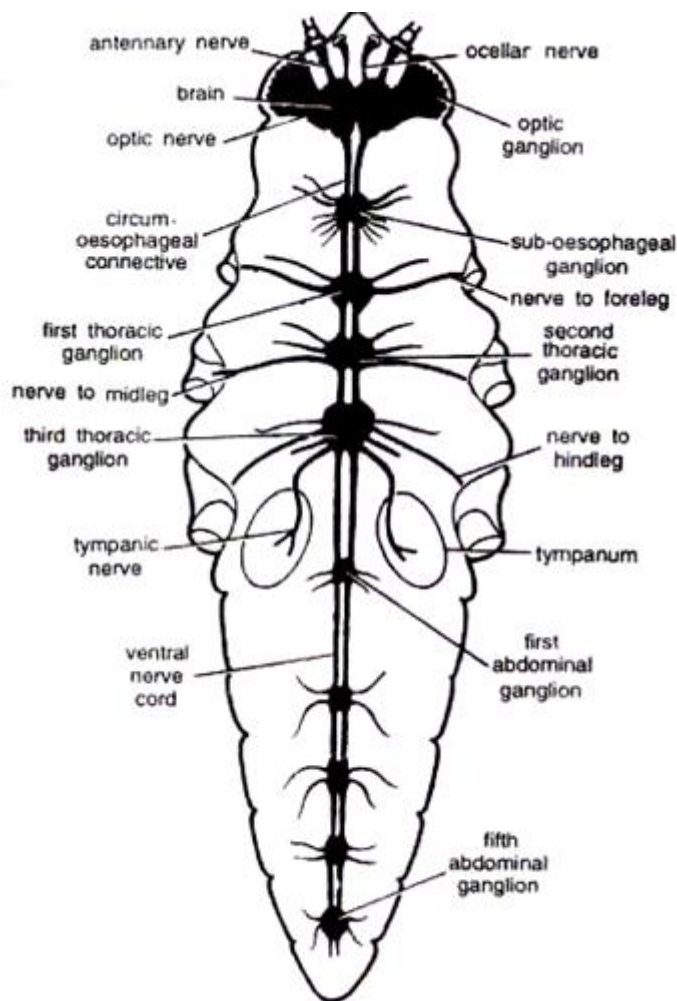


Fig 14.15 Nervous System of Grasshopper

14.7 - Glossary:-

Aboral	Opposite the mouth.
Amoeboid	Cell movements resembling those of the amoeba.
Angstrom	One thousandth of a micron.
Archenteron	Primitive digestive tract of a metazoan embryo, formed during gastrulation.
Autotrophic	Nutrition. Process of nutrition in which an organism manufactures its own food.
Basal disc	Foot of some Cnidaria which is flattened and attaches to a substratum by secretion of a sticky substance.
Binary fission	the type of asexual reproduction by means of which the organism divides into two approximately equal halves.
Buccal	Pertaining to the mouth or oral cavity.
Cnidaria or Coelenterata.	Phylum of animals all possessing cnidoblast structures.
Cnidoblast	Type of cell in which nematocyst is found.

Coelom	The body cavity lined with tissue of mesodermal origin in which the digestive and other organs lie.
Conjugation	A method of sexual reproduction in which two unicellular animals unite, exchange nuclear material and then divide as in the Paramecium.
Contractile vacuole	A space in the cytoplasm of certain species of protozoa where fluids collect before being periodically discharged to the outside.
Ctenophora	Radiate phylum of animals possessing comb- such as comb- jellies.
Cuticle	Thin non-cellular outermost secreted by the underlying epidermis.
Cyst	The stage of an organism where it is enclosed in a resistant wall.
Cytopharynx	Pharynx or gullet of a protozoan such as Paramecium.
Cytostome	Cell mouth, for example in Paramecium.
Diploblastic	Derived from two embryonic germ layers, ectoderm and endoderm.
Enteron	Digestive tract, especially in Cnidaria.
Entoprocta	Pseudocoelomate, sessile phylum with U-shaped intestine, mouth surrounded by circle of ciliated tentacles, and opening within circle.
Extracellular	Outside of the cell or cells.
Exumbrella	Convex, aboral surface of the medusa.
Fission	Asexual method of reproduction by division into two or more approximately equal in size.
Food vacuole	Intra-cellular digestive organelle.
Free-living	Capable of independent existence.
Gastrodermis	Lining of coelenterate digestive cavity.
Gastrulation	Process by which two germ layers, ectoderm and endoderm.
Holophytic	Type of nutrition, found in green plants and in some mastigophores, which involves photosynthesis.
Holozoic	Type of nutrition found in most animals, that involves ingestion and digestion of organic material.
Hydranth	Expand end of a branch of a hydroid colony specialized for vegetative function.
Hydrocaulus	Basal portion of a hydroid colony often branched and root-like used for attachment to substratum.
Hydrotheca	Transparent membrane that extends from the perisarc and surrounds the main part of the hydranth.
Hypostome	Region surrounding the mouth in coelenterates.
Inter-cellular	Between cells.
Intra-cellular	Within cells.
Isogamy	Sexual reproduction involving fusion of two similar gametes but from opposite sexes.
Kinetosome	The basal body of a flagellum or cilium.
Lophophore	Anterior tentacle- bearing area of certain coelomates; serves in food capture.
Mesogloea	Non-cellular jelly-like substance lying between the ectoderm and endoderm in coelenterates.
Metagenesis	Alternation of sexual with an asexual generation in reproduction in the life cycle of a coelenterate such as Obelia.
Myoneme	Type of contractile fibril in certain Protozoa.
Nephridiopore	External opening of an excretory tubule or nephridium.

Pedal	Pertaining to father.
Pellicle	The protective layer on the surface of some protozoans, for example, Paramecium.
Penetrant	Largest type of cnidarians nematocyst, containing a coiled tube and spines, used in prey capture.
Peristome	Region around the mouth of a radially symmetrical animal such as hydra.
Phagocyte	Type of white blood cell that engulfs and digests bacteria and other foreign materials.
Pinocytosis	Cellular drinking or intake of fluid.
Plankton	Floating or drifting aquatic organisms, mostly microscopic.
Plasmasol	Relatively liquid cytoplasm.
Pneumatophore	Air-filled float of siphonophoran hydroids.
Polyp	A tubular coelenterate form.
Prosopyle	One of the surface pores opening into a sponge chamber.
Protozoa	A phylum of acellular animals.
Proximal	Near the point of attachment of an organ.
Pseudocoel	A body cavity not completely lined with mesoderm as found in round worms.
Pseudopodia	Blunt temporary protoplasmic projections found in amoeba or in some ameba like cells.
Schizocoel	The coelom formed by the splitting of embryonic mesoderm.
Sedentary	Staying in one place.
Siliceous	Containing silicon dioxide or silica.
Spicule	One of many solid structures that composed the structural framework of a sponge.
Spongocoel	Paragastric or central cavity of a sponge.
Syngamy	Union of gametes in sexual reproduction forming a zygote.
Taxis	A movement response
Tentacle	A flexible arm like extension from the body of many invertebrates such as hydra. Used in grasping and movement.
Tentaculocyst	Sense organs of some cnidarians.
Triploblastic	Derived from three primary germ layers-ectoderm, mesoderm, and endoderm.
Vestibule	An outer cavity with an entrance to a (usually) larger, deeper cavity.
Zoid	One of the members of a hydroid or siphonophore colony.

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Unit 15: Cytological Study

15.1- Objectives

15.2-Introduction

15.3- Study of Mitosis and Meiosis using available Material

15.4. – Study of permanent Slide

15.4.1- Study of permanent Slide showing stage of cell division

15.4.2- Study of permanent Slide showing stage of giant chromosomes

15.4.3- Study of permanent Slide showing stage of Mitochondria

15.4.4- Study of permanent Slide showing stage of Golgi body

15.6- Summary

15.7- Glossary

15.8-References

15.1- OBJECTIVES:-

To study the Meiosis and describe the chromosomal makeup of a cell using the terms chromosome, sister chromatid, homologous chromosome, diploid, haploid, and tetrad and also recognize the function and products of mitosis and meiosis. Compare and contrast the behaviors of chromosomes in mitosis and meiosis. Recognize when cells are diploid vs. haploid and Predict the DNA content of cells in different phases of mitosis and meiosis stage because meiosis is a specialized and rather complicated type of cell division and we have to recall and describe the phases of the cell cycle co-relate the cell cycle stages to changes in DNA content.

15.2 INTRODUCTION

Meiosis is a specialized and rather complicated type of cell division, occurring only in the diploid reproductive cells and results in the formation of haploid sex-cells of gametes. The gametes, formed as a result of meiosis, possess half the number of chromosomes as found in the parent cells and their chromosome number is represented by **n**, whereas the zygote formed by the fusion (fertilization) of male and female gametes and the cells derived from it are known as **diploid** and their chromosome number is symbolized by **2n**. The two similar chromosomes of diploid cells are known homologous chromosomes or homologous pair.”The chromosomes of a homologous pair are brought together in the zygote by the union of male and female gametes from the parents.

15.3- Study of Mitosis and Meiosis:-

Meiosis occurs in the life cycle of each and every living being whether a plant or an animal, but its period of occurrence varies in different groups. In majority of cases it occurs prior to gamete formation. The cells undergoing meiosis are known as **meiocytes**. In animals, the **meiocytes** are the **primary spermatocytes** and **primary oocytes** while in plants these are represented by **sporocytes**. The relative amounts of RNA and DNA are supposed to initiate meiosis in some way. If the ratio of RNA to DNA is high, the cell will undergo meiosis but if reverse is the case it will lead to mitosis.

15.4. – Study of permanent Slide

Process of Meiosis:-

The process of meiosis is separated into a sequence of events similar to those of mitosis but these events or stages are repeated twice, i.e., in meiosis, two complete cell divisions follow in close sequence, with or without a short interphase between them. The first meiotic division is known as **reduction division or heterotypic division**. In it the diploid parent cell divides into two daughter cells having haploid chromosome number. The second division is known as **homoeotypic division** and it is a simple mitotic division in which the two haploid cells formed as result of heterotypic division divide forming four haploid cells. The two meiotic cell division is further distinguished into phases. These are:-

A. Heterotypic Division or First Meiotic Division or Reduction Division

I FIRST PROPHASE:-

The prophase of first meiotic division is of longer duration and profoundly modified. It is distinguished into following **five** phases or sub stages –

- (a) Leptotene
- (b) Zygotene

- (c) Pachytene
- (d) Diplotene and
- (e) Diakinesis

1. Proleptotene :

The meiocytes or the meiotic cell is comparatively larger in size and possesses a large nucleus. It contains diploid number of chromosomes which form a network.

In the beginning, the movement of centrioles, the formation of astral rays and the gradual condensation of the chromatin material proceed in a similar fashion as in the prophase of mitosis. These preliminary steps constitute proleptotene.

2. Leptotene or Leptonema :

The leptotene stage initiates meiosis. Due to the condensation of chromatin matter the chromosomes appear in diploid number as long, thin and uncoiled threads or slender filaments longitudinally single rather than double as in mitosis.

These threads correspond to the chromonema of the anaphase of mitotic division.

Their arrangement is often irregular but they might exhibit some definite orientation. Each chromosome presents a beaded appearance due to the presence of a longitudinal series of dense, bead-like swelling called **chromomeres**.

The chromomeres **are** of different sizes and occur in definite sequence on each chromosome. The homologous chromosomes display the same sequence of chromosomes.

The DNA and histone synthesis and the chromosomes duplication either starts in this substage or occurs in the later substage but in most cells the duplication is completed by the end of next substage, i.e., **zygotene**.

The nucleolus is well marked and increases in size in leptotene and zygotene.

3. Zygotene or Zygonema

The zygotene commences with the movement of chromosomes. It is affected by the forces of attraction between the two homologous of a chromosome pair. Thus, the chromosomes of a pair approach each other and each chromosome shortly takes a position along the side of its partner to form a bivalent.

The pairing of homologous is known as **synapsis** and is very intimate and precise, the chromomere to chromomere.

Once the pairing has started at some point along the homologues it proceeds from there in zipper-like fashion. This indicates that the homologous chromosomes are not only similar in appearance, but they also carry the same genes in the same sequence.

The pairing may be completed in any of the following methods:

- The two homologues start pairing progresses towards centromere region -**proterminal apis**.
- The pairing may start near the centromere and then progresses towards the ends - **procentric synapis**
- The pairing starts at random either at one point or at many points simultaneously-**random synapsis**.

In organisms with definitely oriented or polarized chromosomes, pairing usually commences at the ends nearest the nuclear membrane and progresses onwards till completion. This peculiar state of orientation, polarization and association is known as bouquet **stage**.

As the pairing proceeds, the chromosomes continue to condense and become shorter and thicker. Two views have come forward to explain the possible initiation of synapsis.

According to precocity theory put forward by **Darlington**, the chromosomes pair due to their singleness. But this theory does not explain the extra synthesis of DNA and chromosomes duplication at leptotene stage.

The **retardation theory** by Sax and others explains that the pairing of homologous is due to the retardation of cessation of metabolic activities of the cell.

At zygotene the nucleolus increases in size and the centrioles move apart initiating the spindle formation.

4. Pachytene Stage or pachynema

With the pairing or synapsis of homologues the nucleus enters the pachytene stage. It represents the stable period in cell division. During this stage the paired chromosomes of bivalent get shortened and thickened due to gradual condensation of chromatin and appear as thick rods of different shapes and sizes, so that the chromosomes are more readily distinguished.

The homologous chromosomes now twist or twin around each other forming relational coils. Each chromosome starts splitting into two sister chromatids by a vertical or longitudinal furrow. As a result the bivalent is now converted into tetrad.

The time of duplication varies in different types of cells. In some it is said to occur in leptotene, while in others in pachytene.

Their relational coiling gets further complicated due to the coiling of two chromatids of each chromosome. This vigorous coiling exerts considerable strain upon the chromosomes. As a result the weaker chromatids break down at points.

These transverse breaks occur in the non-sister chromatids of a pair at corresponding points. The broken ends are then interchanged between the matching chromatids and are attached to their respective remaining portions.

This exchange and recombination of chromosomal parts is known as **crossing over**. Its completion marks the end of pachytene.

5. Diplotene or Diplonema

The separation of homologous chromosomes initiates diplotene. The synaptic forces of attraction

between them lapse due to breakage at one or more points so that the homologous chromosome uncoil and starts separating.

But the separation is none the less incomplete since the homologous are in contact are known as **chiasmata** (sing. **Chiasma**, meaning, **cross**) which present cross-shaped appearance.

The chiasma is the bivalent varies in the same pair of chromosomes and in different cells of the same individual. By the end of diplotene the chiasmata begin to move along the length of chromosomes from the centromere towards the end.

This displacement of chiasmata is termed as *terminalization*. When the **terminalization of chiasmata**. The degree of terminalization is generally expressed as **coefficient of termination** (*T*).

$$T = \frac{\text{Number of terminal chiasmata}}{\text{Total number of chiasmata}}$$

The average number of chiasmata in bivalent is known as frequency of chiasmata 9Fq)

$$\text{Frequency (Fq)} = \frac{\text{Total number of chiasmata}}{\text{Total number of bivalents}}$$

According to Darlington, two types of repelling forces operate on the chromosome at diplotene. One of the forces is electro negatively charged and operates on the surface of the chromosome throughout its length and the other with electropositive charge is localized on the centromere. The former controls the repulsion of the chromosomes and the latter cause's distal movement of the chiasmata.

6. Diakinesis:

The bivalents still contract and get thickened into deeply stained bodies. These migrate to the periphery of the nucleus. The two chromatids of each chromosome become closely oppressed together losing their individual identity.

At the same time the homologues move still apart due to the force of repulsion developed between their centromeres. In doing so the chiasmata move towards the ends.

At this stage the nucleolus and nuclear membrane disappear and the formation of nuclear spindle starts.

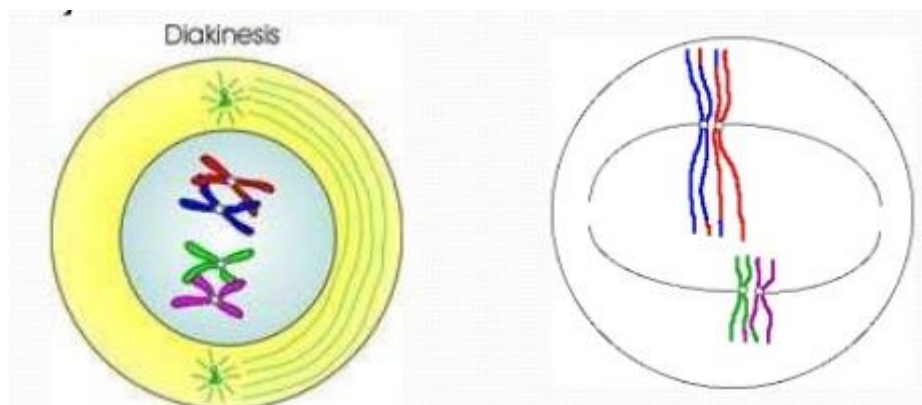


Fig.15.1 Diakinesis

II FIRST METAPHASE (METAPHASE I):-

The metaphase of meiosis is very similar to that of mitosis. At the close of diakinesis the nuclear membrane disappears and the formation of amphaster or achromatic figure or the spindle is completed. In metaphase the bivalents move to the equator.

Later on, they orient themselves on the equator in such a way that their centromeres lie one on either side and equidistant from the equatorial plate.

Their centromeres face the pole of the spindle and the arms are directed towards the equator and rest on the equator.

III FIRST ANAPHASE (ANAPHASE I) :-

During this stage the bivalents move apart towards the opposite poles of the spindle. The tetrad which was having four chromatids now separates into two dyads due to the complete separation of maternal and paternal chromosomes of the bivalent.

Therefore, each separated half consists of two sister chromatids attached together by a common centromere. This process of separation is known as **disjunction** and this involves the separation of those homologous chromosomes which were brought together in the zygote stage.

By this time the two chromatids of a dyad also separate except at the points of centromere, so that they present V-shaped appearance.

IV FIRST TELOPHASE (TELOPHASE I)

The first telophase commences with the formation of nuclear wall around the haploid group of chromosomal dyads which have already reached the poles of the spindle.

The chromosomes elongate and uncoil. Nucleolus is also formed.

The cell cytoplasm also segments into two. Thus two daughter cells are formed, each of which contains haploid number of chromosomes.

V INTERPHASE:-

It is the resting stage of dividing meiocytes and its duration depends upon the species involved. It may be totally absent and the chromosomes of first anaphase directly pass into second prophase omitting the telophase.

In this condition the nuclear material remains unchanged and the nuclear membrane is not formed. If the interphase is present the nucleus assumes its original form by the development of nuclear net and nuclear membrane. But if at all interphase is present it is of a very short duration.

B. Homeotypic Division:-

The second meiotic division is essentially mitosis, occurring independently in both the haploid sister cells. It may follow immediately after first meiotic division or may not occur until much later.

1. Second Prophase or Prophase II

During second prophase the nucleus and nuclear membrane disappear in both the daughter haploid cells and the formation of spindle starts.

The chromatids are coiled and the dyad has X-shaped appearance having chromatids joined by centromere and arms radiating.

2. Second Metaphase or Metaphase II

The second metaphase is of short duration. The chromatids move towards the centre of the spindle and orient on the equator.

Their centromeres touch the equator but the arms radiate out toward poles. Later on, the centromere in each dyad divides into two.

3. Second Anaphase or Anaphase II

The chromatids with their independent centromeres from sister chromosomes and move apart towards the opposite poles of the spindle.

The chromatids of second anaphase are not short and compact bodies like those of first anaphase but are very similar to the chromosomes of anaphase in mitotic division.

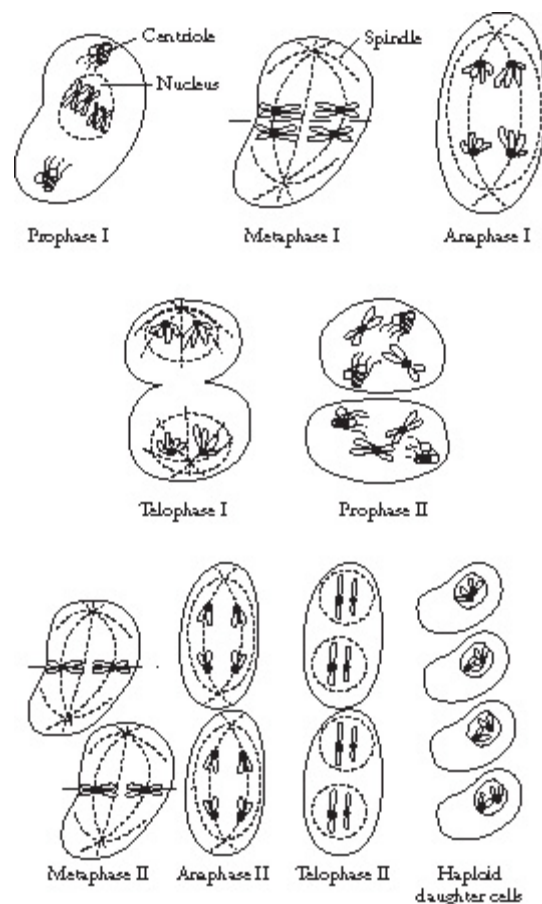


Fig.15.2 Homeotypic Division

4. Second Telophase or Telophase II

The chromosomes at each pole uncoil and thin out to form the nuclear net. Each group gets surrounded by a nuclear membrane. Nucleolus reappears. Thus two nuclei are recognized in each cell.

Significance of Meiosis

The significance of meiosis is threefold:-

1. The meiosis is a logical and necessary part in the life cycle of sexually reproducing animals since it leads to the formation of gametes or sex cells that participates in fertilization. These are haploid cells having only one member of each homologous pair.
2. The meiosis is concomitant of doubling chromosome number due to gametic fusion. The gametes formed as a result of meiosis are haploid and the zygote formed by their fusion is diploid. Thus, it is only means for restoring the chromosome number characteristics of the species.
3. Meiosis results new combination of genetic material. During crossing over, the hereditary factors from male and female parents get mixed due to breakage and exchange of chromatids in pachytene. Thus, the gametes produced are not all alike but with variable combination of genes. The random segregation of chromosomes and the new alignments of genes in them resulting

from crossing over ensure genetic variations in the population. The inherited variability leads to the evolution of organisms.

Cytological Study Exercise:-

(1) - Object: To study the meiosis by using available material.

Requirement: - Living grasshoppers, Chloroform, normal saline, Carnoy's fluid, acetocarmine, slides, cover-slip, blotting paper and microscope.

Procedure: - Take a chloroformed grasshopper and dissect it in normal saline. Take out its testis and fix them in Carnoy's fluid for 2-12 hours. Take a small lobe of testis and stain it in acetocarmine. Put the stained lobe on a clean slide and cover it with a cover slip. Warm the slide over the flame of a spirit lamp and then put a blotting paper over it, press it smoothly by your thumb. Examine the slide under microscope.

Result: The cells of testis lobes are spread out and became distinct. Carefully observe different stages of meiosis under microscope and draw them in practical notebook.

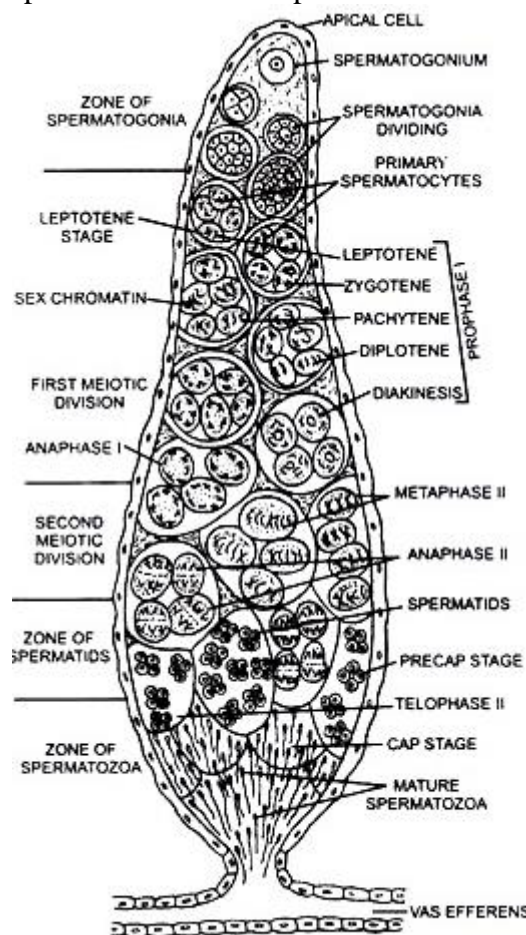


Fig. 15.3 T.S of one follicle of testis of Grasshopper to show the stage of meiosis

Cell Division :-

The process of cell division is found to be essentially the same in all living organisms and the events are chiefly centered in the nucleus. Three types of cell divisions have been distinguished.

- (i) **Amitosis or direct cell division.**
- (ii) **Mitosis or indirect cell division.**
- (iii) **Meiosis or reduction division.**

Amitosis:-

Amitosis or direct type of cell division is characterized by the splitting of the nucleus followed by that of cytoplasm. It is seen in unicellular organism like protozoan's and the cells of foetal membranes.

The beginning is marked by the elongation of the nucleus. Due to the appearance of depression or constriction in the middle line, the nucleus assumes dumb-bell shaped appearance. The depression increases in size and splits the nucleus into two.

Simultaneously, the cell body or the cytoplasm is also constricted into two equal or approximately similar halves. During the process of amitotic cell division there is complete absence of nuclear events and the mechanism is very simple.

Mitosis:-

Objective: (B) To study the Mitosis

Definition:-

Mitosis involves the exact replication of parent cell followed by its division into two daughter cells which are identical and contain the same number of chromosomes as found in the parent cell.

Introduction:-

This nuclear division was first observed by **Straburger (1870)** in plant cell and **Flemming (1882)** in animal cells. Flemming used the **term mitosis (Gr. Mitos, thread)** for this process with reference to the thread-like appearance of chromosomes early in the cell division. An illustrated account of behavior of chromosomes during the period of cell division has been given by **Darlington**. The cell division where chromosomal duplication (i.e. longitudinal splitting of chromosomes) is followed by the nuclear division so that each daughter cell possesses the same number of chromosomes as present in the parent cell.

Mitosis, division of a living cell nucleus (control centre), leading to the production of two offspring or daughter cells, normally with the same genetic information. Mitosis is the standard way that cells multiply. It occurs all the time in the human body and other multi-cellular living things, especially during growth to make more cells, and during maintenance to replace damaged and worn-out cells. In single-celled

organisms, it represents asexual reproduction. In plants, it is the basis of asexual or vegetative reproduction (making cells for sexual reproduction involves another type of cell division). Genes exist as chemical codes on lengths of the chemical deoxyribonucleic acid (DNA) inside the nucleus. During a cell's "resting" period, or interphase, the DNA copies or replicates itself to form two complete sets. Mitosis then occurs in four main stages.

Process of Mitosis

The process of mitosis is characterized by the duplication of chromosomes, their separation into two and then their movement to opposite poles so as to construct two daughter nuclei.

It is followed by the constriction of cytoplasm to form two daughter cells.

The replication and distribution of chromosomes is known as **karyokinesis** while the division of cell cytoplasm and separation into two daughter cells is called **cytokinesis**.

It means cell division can be separated into two categories:

- The nuclear division or **karyokinesis**, and
- The division of the cytoplasm or **cytokinesis**.

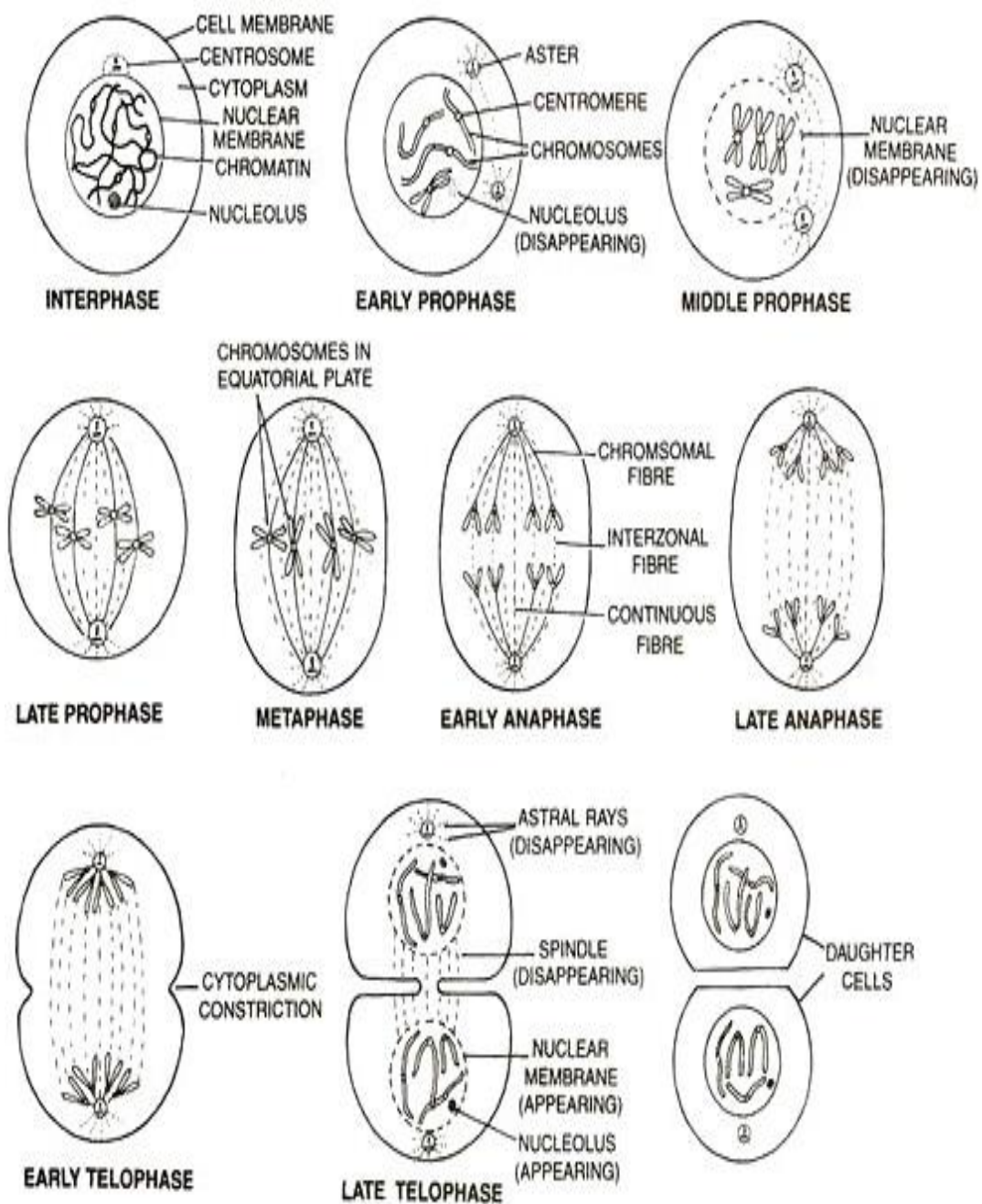


Fig. 15.4 cell division in Animal Cell

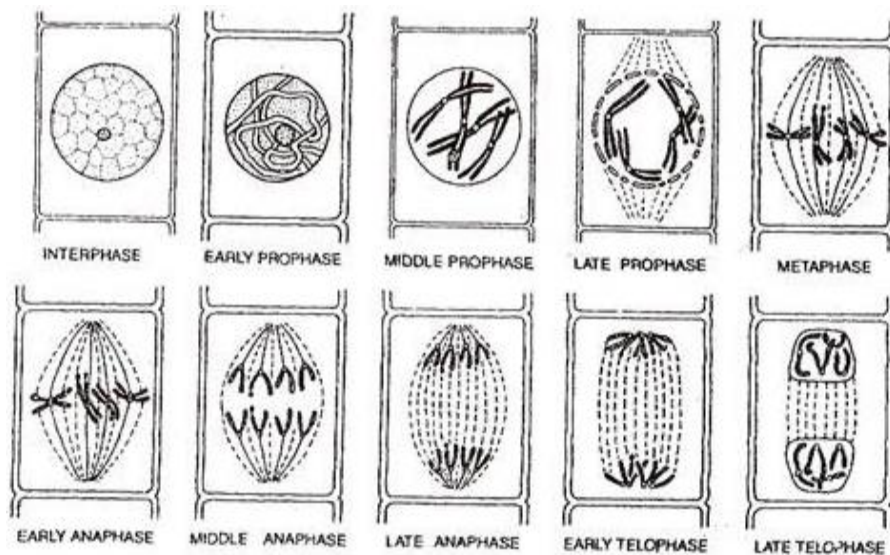


Fig. 15.4 cell division in plant Cell

Karyokinesis:-

The process of karyokinesis includes the division of cell nucleus into two daughter nuclei, It is divided into prophase, metaphase, anaphase and telophase.

1. Prophase

The nuclear division (mitosis) begins with prophase. The important events during this phase are given below-

(A) Nuclear Changes:

- (a) The chromatin materials of nucleus gradually condense into distinct chromatin thread by losing water.
- (b) The chromatin threads coil like cylindrical spring and in so doing they gradually become shorter and thicker and form the chromosomes.
- (c) The proteinous matrix gets deposited around the chromosomes, so that these gradually become shorter and thicker and form chromosomes.
- (d) Each chromosome is already doubled due to the doubling of DNA contents in interphase.

- (e) By the end of prophase the two chromatids of each chromosome become more distinct and each chromosome appears to be splitted up lengthwise.

(B) Cytoplasmic Events:

- (a) The centriole divides into two and then one of the daughter centrioles moves towards the opposite pole.
- (b) Astral rays radiate out from each daughter centriole.

(2) Metaphase

The metaphase is marked by the appearance of spindle and arrangement of chromosomes on the equator of spindle.

- a) The microtubules in the cytoplasm of the cell orient in between the centrioles of the opposite poles and form the spindle. Such a spindle is known as **amphiaster**.
- b) The chromosomes from periphery of the nucleus migrate towards equator of the spindle, lie on the equator and are attached to the chromosomal fibres of the spindle, whereas the arms are orient towards the poles.
- c) Each chromosome becomes more compact and short and its two chromatids separate except at the centromere which has not divided so far.

(3) Anaphase

- (a) The centromere of each chromosome divides and allow the separation of two sister chromatids into two daughter chromosomes.
- (b) The daughter chromosomes move apart and migrate towards opposite poles.
- (c) The movement of chromosomes is governed by the contraction of spindle fibres, the centromere is pulled first towards the pole of the spindle and the arms of chromosomes are dragged behind.
- (d) In anaphase, the arms of daughter chromosomes are directed towards the equator and centromeres towards the poles of the equator.

(4) Telophase

- (a) Chromosomes reach poles of the spindle and form two groups.
- (b) Chromosomes begin to uncoil and form chromatin net.
- (c) The nuclear wall and nucleolus reappear.

Mitotic apparatus or mitotic spindle:-

The mitotic spindle is formed of spindle fibres extending between the two centrioles and the astral rays radiating out from each centriole.

Structure of Spindle Fibres:-

Spindle fibres are formed of microtubules, arranged in parallel bundles. These are about 250-270Å in diameter and with a 50-70 Å thick wall. The number of microtubules composing the spindle of yeast cells (Moor, 1967).

Chemical Composition of Spindle Fibres:-

The spindle fibres represent long chain protein molecules oriented in longitudinal direction between the two poles. The protein chain are linked by bonding of protein monomers by –SH and –S–S bonds. These contain 90% proteins and 5% RNA.

Formation of Spindle Fibres:-

- Spindle fibres are cytoplasmic in origin and about 15% of the cytoplasmic proteins form the spindle.
- The formation of spindle starts in the late prophase and is completed in metaphase.
- Commonly it begins outside the nuclear membrane more or less simultaneously with the disappearance of nuclear membrane.
- During the formation of microtubules of the spindle, the polymerization of protein monomers to form amorphous gel and the formation of secondary bonds through –SH and –S–S groups takes place.
- The process is initiated by the release of RNA from the nucleus.

Types of Spindle Fibres : -

The spindle fibres are of the three types-

- **Continuous Fibres:** These extend from one pole of the spindle to the other poles.
- **Chromosomal Fibres:** these fibres extend from pole of the spindle to the centromere of chromosomes. These are also called **kinetochore microtubules**.
- **Interzonal Fibre :** These appear in anaphase and telophase and extend between the centromeres of separating chromatids (daughter chromosomes)

Role of Spindle Fibres: -

Spindle fibres help in the movement of chromosomes from equator to the pole of spindle.

Chromosomal Movement during Cell Division:-

Cell division is characterized by the movement of chromosomes and of a number of other cellular structures. These movements are:

- (1) Movement of spindle poles or centrioles to the opposite sides of the cell during prophase.
- (2) Oscillatory movement of chromosomes to the equator of spindle during prometaphase.
- (3) Movement of chromosomes from the equator of spindle towards poles during anaphase A.
- (4) Elongation of spindle during anaphase B.

Duration of Mitosis:-

- The time required for mitosis differs with species and environment.
- Temperature and nutrition, in particular, are important factors.
- The entire sequence of phases may be completed in 6 minutes to many hours.
- Normally the entire cycle of cell division takes approximately 18 hours, about 45 minutes from prophase to the end of telophase and about 17 hours for the interphase.
- Different phases of mitosis are of different duration.
- Anaphase is the shortest, the prophase and telophase the most prolonged, and the metaphase of intermediate duration.

Mitotic Poisons: -

- There are certain substances that affect the cells in mitosis or prevent them from entering it.
- These are commonly known as mitotic poisons.
- The **colchicine** inhibits spindle formation and holds the cells in metaphase.
- The enzymes ribonuclease is prophase poison.
- Mustard gas fragments and agglutinates the chromosomes.
- Higher concentration of some of these poisons may lead to the immediate death of the cells.

Significance of Mitosis:-

- Mitosis is a significant aspect in the growth of living matter.
- It ensures that the new cytoplasm is accompanied by an appropriate amount of governing nuclear material.
- Individual cells cannot grow indefinitely and their size remains within economical limits with respect to the intake of foodstuffs and their transformation into energy and new cytoplasm.
- As a result of mitosis each new cell receives a set of chromosomes to regulate the activities of the cytoplasm.

- Mitosis ensures a continuous succession of similarity endowed cells, because from one dividing cell two daughter cells with exactly the same number and the same type of chromosomes are formed.
- Thus, no matter how many consecutive cell divisions have taken place, all the cells have an array of chromosomes identical to the parent cell from which they have descended by division.
- Mitotic divisions help not only in the increase of size by cell accumulation but also in replacing the old and damaged tissue by the new cells.
- In plants these do not cease to divide even when the plant is mature but continuously go on cutting new cells from the cambium.

Cytological Exercise:-

The study of cells necessarily involves sophisticated equipments and techniques.

Following is very simple and elementary methods are being described here to study cell division and making preparations of certain cell components.

(1) Objective: To observe the stages of mitosis using onion root tips.

Requirements:

- Onion root tips fixed in Carnoy's fluid.
- Microscope glass slide.
- Cover-slip.
- Acetocarmine.
- Sprit lamp.
- Blotting paper and
- Microscope.

Procedure:

- Take a drop of acetocarmine on a clean microscopic slide and put on it one or two tips.
- Place a cover slip over it and tap it gently by a needle.
- Warm the slide over the flame of a sprit lamp and then put a blotting paper over it, press it smoothly by your thumb.
- Examine the slide under microscope.

Result:-

The cells and their chromosomes are spread out and become distinct. Observe carefully the different stages of mitosis.

Cytological Exercise

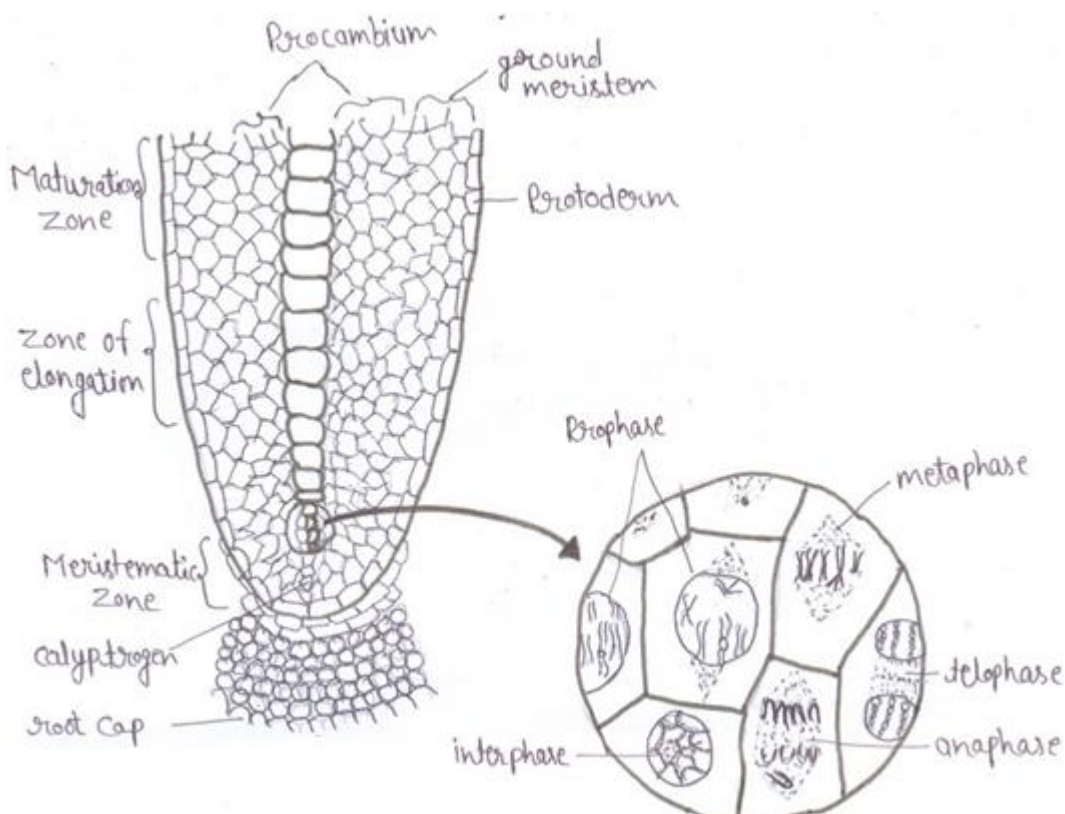


Fig.15.5 onion root tip showing different stage of mitosis

15.4. – Study of permanent Slide:-

What is a cell?

Unicellular organisms are capable of:-

- (i) Independent existence and
- (ii) Performing the essential functions of life.

Anything less than a complete structure of a cell does not ensure independent living. Hence, cell is the fundamental structure and functional unit of all living organisms.

Anton Von Leeuwenhoek first saw and describe a live cell. Robert Brown later discovered the nucleus.

The invention of the microscope and its important leading to the electron microscope revealed all the structural details of the cell.

Introduction:-

During cell division oxidative process are minimum and the deficiency of oxygen has no visible effect on the process and speed of mitosis. It is, therefore, presumed that the dividing cell obtains energy by **glycolysis**.

No doubt prior to division, when DNA synthesis takes place in the nucleus, the oxygen consumption is normal. Swan (1957) has suggested that an energy reservoir is created inside the cell before it enters cell division. Recently, **Allfrey and coworkers** (1975) and later have suggested that nucleus synthesis ATP through oxidative phosphorylation mechanism.

There are several evidences in support of the view that the oxidative processes are maximum in premitotic period and minimum during active mitosis:

- (a) Oxygen consumption is minimum during cell division and deficiency of O₂ or concentration of carbon monoxide has no effect on the process of mitosis.
- (b) Certain glycolytic enzymes (lactic acid dehydrogenase, triphosphate dehydrogenase, aldolase) are present in high concentration in the nucleus, whereas enzymes associated with respiration and oxidative phosphorylation in cytoplasm are absent from the nucleus.
- (c) Mitochondria fragment into granules which completely disappear during mitosis (Agrell, 1955; Chevermont and Fredric, 1952). This reconstitution starts during terminal stages of mitosis.

The body of multicellular organism is formed of different types of cells and tissues. These cannot be studied directly by the microscope. The required tissue is separated from the body and is prepared in a way that it becomes suitable for fixing them properly. The following techniques are often employed for the study of tissues or cells:

1. Teasing or Dissociation

The tissue to be studied is teased either directly in the stain or saline solution with the help of needles on a microscopic slide. It is covered with a cover slip and studied under microscope. The muscular tissue is studied by this method.

2. Smear Technique

Fluid tissue containing cells (blood) or small fragments of tissue such as aspirated bone marrow are smeared on the microscopic slide so that a thin film is formed on the slide. It is then fixed immediately, stained and maintained. The smear technique is very popular in **exfoliate cytology** (study of superficial cells shed from mucous membrane) or in the study of chromosomes and cell division.

3. Sectional Method

In this method the specimen is cut into very thin sections. For this purpose tissue is first fixed by immersion in some fixative and is embedded in paraffin wax or colloidin. Thereafter, is cut into thin sections which are stained and then studied under the microscope.

Cytological Study of Preserved Cells: -

Fixation:

As soon as the living processes of a tissue are grossly disturbed either by death or by the removal of tissue from animal body, some changes begin.

These changes are introduced by the onset of **autolysis (auto digestion)**, by the attack of bacteria and moulds and by drying or due to osmotic effect. Some of these changes can be minimized by fixation.

The fixation is the process that brings about sudden death of the cells or tissues in such a manner that their morphological and chemical composition is retained either by the use of chemicals or by freezing.

Aims and Effects of Fixation:

- (a) Fixation hardens the tissues and gives them a consistent form.
- (b) It prevents autolysis and bacterial decomposition.

- (c) It coagulates the tissue, renders the contents insoluble and prevents loss of easily diffusible substances.
- (d) It avoids cell shrinkage and distortion in form due to postmortem changes.
- (e) Improves the optical differentiation of cell components by changing refractive indices and thus increases their visibility.
- (f) Prepares the tissue for staining.
- (g) It fortifies tissue against the harmful effects of various stages in the preparation of sections.

A. Chemical Fixation: The tissue is fixed by some chemical compounds such as formaldehyde, mercuric chloride, picric acid, chromic acid, osmium tetroxide, acetic acid and ethyl alcohol. These are called fixative.

1. Simple Fixatives

- (a) **Formalin:** 4-10% formalin solution is used for fixing golgi apparatus, mitochondria and enzymes. It fixes and hardens the tissue but causes little or no shrinkage.
- (b) **Mercuric Chloride:** It is an intolerable fixative and is used only in combination with some other fixatives. It hardens and causes shrinkage in the tissue but does not distort it. It precipitates the proteins and fixes lipids.
- (c) **Picric Acid:** it precipitates proteins and nucleoproteins. It produces shrinkage. Commonly it is not used for cytological studies.
- (d) **Chromic Acid:** 0.5-1% chromic acid is used to fix those tissues which are studied for golgi complex and mitochondria. It precipitates all proteins and fixes carbohydrates.
- (e) **Osmium Tetroxide:** 0.5-2% solution of osmium tetroxide is used for fixing cytoplasm, golgi complex, mitochondria and fat. It fixes lipids and causes their blackening. It forms additive compounds with proteins, its penetration is poor but fixation is very nice. It is extensively used for electron microscopy.
- (f) **Potassium dichromate:** 2.5-5% potassium dichromate solution is used in conjunction with some other chemical substance. It renders protein insoluble in water and fixes lipids. It is used for the fixation of chromosomes.
- (g) **Acetic Acid:** Glacial acetic acid is never used alone because of its swelling effect. It is used along with other fixative to counteract their shrinkage effect. It precipitates nucleoproteins but not the cytoplasmic proteins. It destroys golgi complex and mitochondria. It is, therefore, used for the fixation of nucleus and chromosomes.

(h) Ethanol: 70% to absolute alcohol is used as fixative.

2. Compound Fixatives

Since each of the primary fixative listed above has its virtues and defects, none of them is ideal to preserve and allows the observation of every component of the tissues and cells.

As a practice a mixture of two or more reagent is used as a fixative to make use of the special properties of each.

The most essential feature of a fixative should be its quick penetration power. Some of them are mentioned below:

(a) 19% formal saline: It is mixture of formaline and normal saline solution.

(b) Formal alcohol (FAA): A mixture of 10ml formaline, 90 ml of 90% alcohol and 5ml glacial acetic acid is called formal alcohol. It is used as a fixative for polysaccharides and nucleoproteins.

(c) Carnoy's solution: It is mixture of ethanol (absolute alcohol) 60ml and glacial acetic acid 10 ml and 30ml chloroform. It fixes nucleoprotein and chromosomes. It combines the properties of ethanol and acetic acid.

(d) Bouin's fluid: It is mixture of 75 parts picric acid, 25parts formalin and 5 parts glacial acid. It precipitates all proteins, penetrates rapidly and produces little shrinkage. It is used for the histological studies. It fixes chromosomes.

Procedure of Fixation:-

When a piece of tissue is immersed in the fixative, cellular death does not occur instantaneously and "post-mortem" changes due to anoxia, changes in the concentration of hydrogen ions and enzymatic action (autolysis) may occur.

The fixative penetrates the tissue by diffusion in such a way that the most external cells are fixed more rapidly and better than the central ones. Thus, every fixed tissue has a gradient of fixation, progressive dilution with the liquid of the cells.

The rate of penetration of the fixative depends upon the type of protein barrier of precipitation produced at the periphery of the tissue. If the precipitate is very fine as in the case of osmium tetroxide, it forms a barrier preventing further passage of the fixative.

Mechanism of staining:-

It is a well known fact that proteins, certain polysaccharides and nucleic acids have the property of ionization. But the ionization of proteins depends upon pH of the medium.

At pH values above isoelectric point, acid groups become ionized and below isoelectric point, all the basic groups dissociate. Thus, at a pH above isoelectric point, the proteins react with basic dyes and exhibit basophilic property.

The intensity of staining depends upon the degree of acidity or alkalinity of the medium. The basophilic or acidophilic property of cell components also depends on the fixative used.

15.4.1- Study of permanent Slide showing stage of cell division:-

The onion cell which is a typical plant cell has a distinct **cell wall** as its outer boundary and just within it is the **cell membrane**.

The cells of human cheek have an **outer membrane** as the delimiting structure of the cell. Inside each cell is a dense membrane bound structure **called nucleus**. The nucleus contains the **chromosomes** which in turn contain the genetic material, **DNA**. Cells that have membrane bound nuclei are called **eukaryotic** whereas cells that lack of a membrane bound nucleus are **prokaryotic**.

In both prokaryotic and eukaryotic cells, a semi-fluid matrix called cytoplasm occupies the volume of the cell. The cytoplasm is the main area of cellular activities in both the plant and animal cells. Various chemical reactions occur in it to keep the cell in the “living state”.

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

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

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

Cells differ greatly in size, shape and activities. For example, **Mycoplasmas**, the smallest cells, are only 0.3µm in length while bacteria could be 3 to 5 µm. The largest isolated single cell is the egg of an ostrich. Among multicellular organism, human red blood cells are about 7.0 µm in diameter.

Nerve cells are some of the longest cells. Cells also vary greatly in their shape. They may be disc-like, polygonal, columnar, cuboid, thread like, or even irregular. The shape of the cell may vary with the function they perform.

The ability to grow and reproduce is a fundamental property of living organisms. However, growth of single cells is fundamentally limited. As new proteins, nucleic acids, carbohydrates, and lipids are synthesized, their accumulation causes the volume of a cell to increase, forcing the plasma membrane to expand to prevent the cell from bursting.

But cells cannot continue to enlarge indefinitely; as a cell grows larger, there is an accompanying decrease in its surface area/volume ratio and hence in its capacity for effective exchange with the environment.

Therefore, cell growth is generally accompanied by cell division, whereby one cell gives rise to two new daughter cells. (The term daughter is used by convention and does not indicate that cells have gender.)

For single-celled organisms, cell division increases the total number of individuals in a population. In multicellular organism, cell division either increases the number of cells, leading to growth of the organism, or replace cells that have died.

In an adult human, for example about 2 million stem cells in bone marrow divide every second to maintain a constant number of red blood cells in the body.

Although often cell growth and cell division are coupled, there is a notable exception. A fertilized animal egg typically undergoes many divisions without the growth of its cells, dividing the volume of the egg into smaller and smaller parcels. Here as well, however, tight regulation of where and when cells divide is crucial.

When cells grow and divide, the newly formed daughter cells are usually genetic duplicates of the parent cells, containing the same (or virtually the same) DNA sequences.

Therefore, all the genetic information in the nucleus of the parent cell must be duplicated and carefully

distributed to the daughter cells during the division process. In accomplishing this task, a cell passes through a series of discrete stages, collectively known as the **cell cycle**.

15.4.2- Study of permanent Slide showing stage of giant chromosomes

Chromosome is single large DNA molecules and its associated proteins, containing many genes, stores and transmits genetic information. These are popularly known as hereditary vehicle.

Lampbrush Chromosomes

In the oocytic nuclei of those animals which have large yolky eggs, the prophase of first meiotic division is extremely extended.

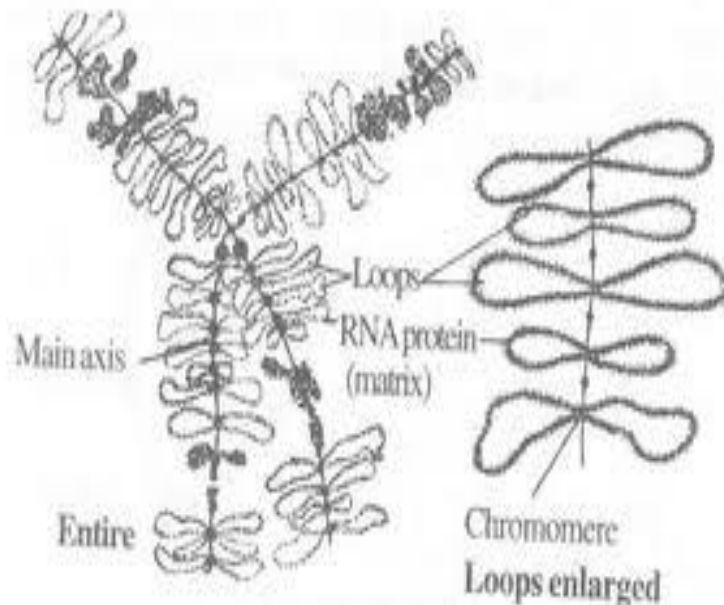


Fig.15.6 Lampbrush *Chromosomes*

During this phase the oocyte grows and synthesizes nutrients for the future embryo. In them, the chromosomes become greatly enlarged and assume unusual configuration. A large number of loops project out from the chromatid axis, giving a lampbrush appearance. Hence, these chromosomes are called **lampbrush chromosomes**.

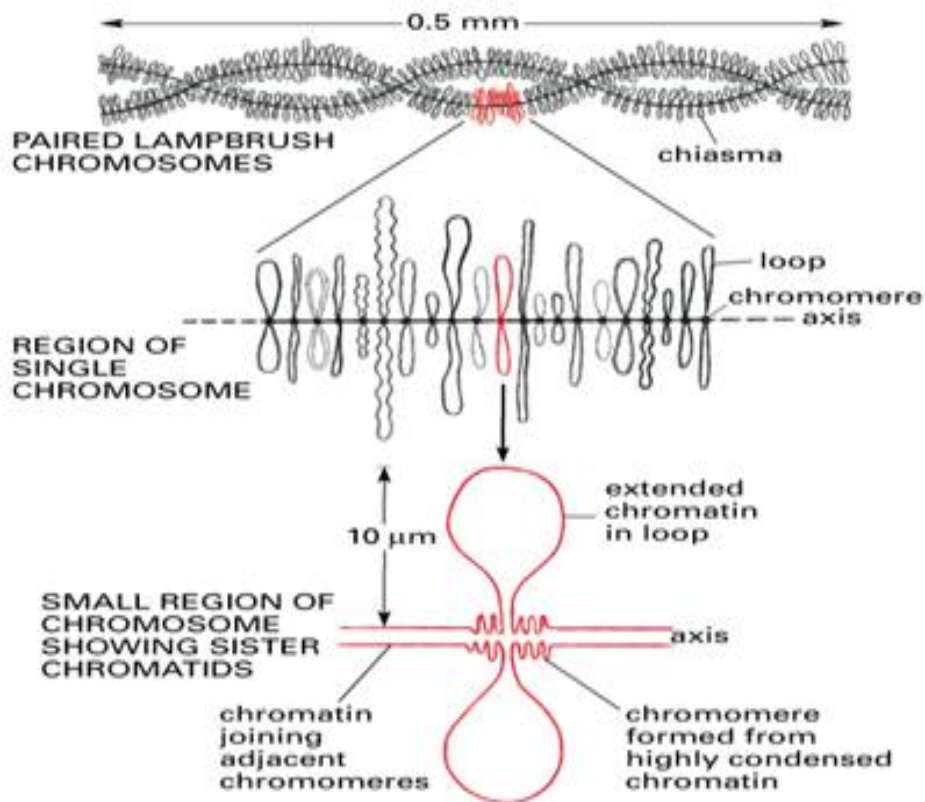


Fig.15.7 Lampbrush chromosome

The lampbrush chromosomes are bivalent each consisting of two chromatids. This persists during the prolonged diplotene phase of first meiotic prophase.

History: -

Lampbrush chromosomes were first observed by **Flemming (1882)** in amphibian oocyte.

A detailed study was made by **J. Rucert (1892)** in the oocytes of sharks.

Occurrence:-

Lampbrush chromosomes are found in the oocytes of insects, sharks, amphibians, reptiles and birds which produce large and yolk eggs.

These have also been found in plants and invertebrates like *Sagitta*, *Sepia* and *Echinaster*.

Size:- Lampbrush chromosomes are enough to be seen under light microscope. These may be as long as 1,000 μm or more and about 20 μ in width.

In salamander oocyte these may attain a length of about 5,900 μ.

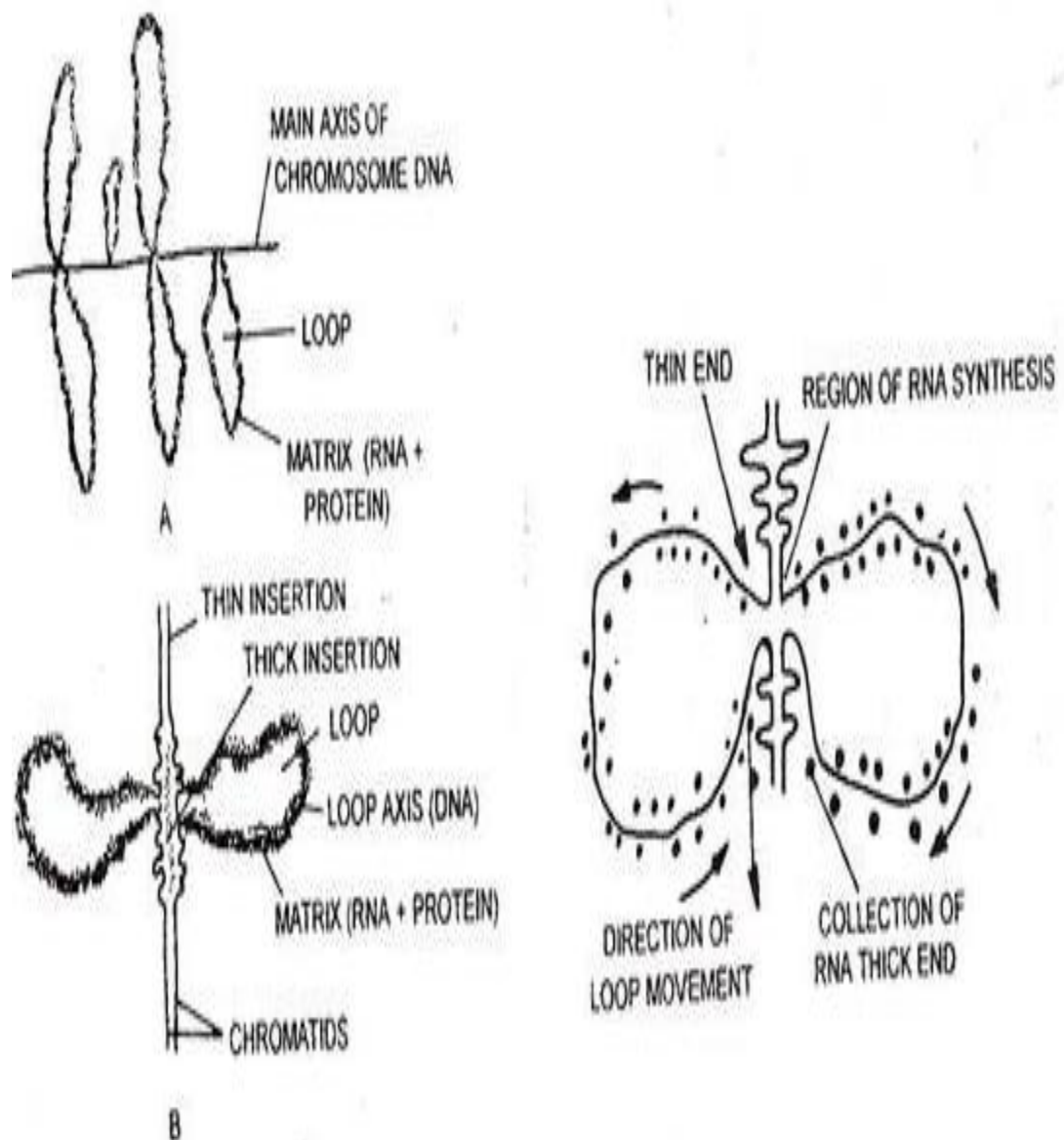


Fig 15.8 Lampbrush (A) Gross structure (B) Enlarged view (C) Synthesis of RNA in a loop of lampbrush chromosome

Structure:- A lampbrush chromosome (in diplotene stage) consist of two homologous chromosome of the pair is formed of two chromatids which lie parallel and form of high density loops which are lightly coloured, and arise on both sides of the chromosomal axis.

The chromosomal axis, the chromomeres and the loop axis all are formed of DNA. The chromomeres are found in pairs, one chromosome on each chromatid.

These are about 0.25 to 2.0 μm in diameter and are spaced about 2 μm from entire to centre along chromatid axis. These probably represent heterochromatic regions where axial filament remains tightly coiled.

The lateral loops arise from the chromomeres either 2 or in multiple of two. These extended on either side of the chromosomal axis about 550 μm and are about 30-50 \AA (3-5nm) in diameter. Each loop consists of an axial fibre formed of DNA.

It is surrounded with the matrix composed of RNA and proteins. This gives fuzzy appearance to lateral loops.

Electron Microscopic Structure:-

Electron microscope studies by Miller and Beaty (1969) on Lampbrush chromosomes of salamander oocyte have shown the presence of dense granules on the loop axis of DNA.

These dense granules represent large molecules of enzyme RNA polymerase. On getting attached to DNA, these initiate RNA synthesis. Arising from these RNA polymerase molecules are seen fine fibrils of RNA.

Each loop is considered to be long operon consisting of a series of identical copies of the same structural genes (cistrons) separated by spacer DNA. Each gene locus probably produces a very long RNA molecule. This interacts with protein to form ribonucleoprotein.

- According to Callan and Liod (1960) a chromosome is the master gene with solenoid which produces several identical copies of its own. These extended out as a lateral loop formed of linear strand of nucleosomes, representing the transcriptionally active stage. These are called Salve gene copies.
- According to spinning out and retraction hypothesis, a chromomere is fully transcribed from end to end by spinning out a transient loop. The new loop material spins out on one side of a chromomere at the end of loop and returns to a condensed stage on the other side after completing the synthesis of RNA.
- These are associated with the rapid synthesis of yolk and protein in the maturing ovum. These disappear by the end of first prophase when chromosomes become thick and more condensed.

Polytene Chromosome:-

Polytene chromosomes are one of the giant chromosomes found in animals and plants. In animals it is found in the salivary glands, Malpighian tubules, the epithelium cell lining of the gut and in the fatty cells of the larvae of certain Diptera.

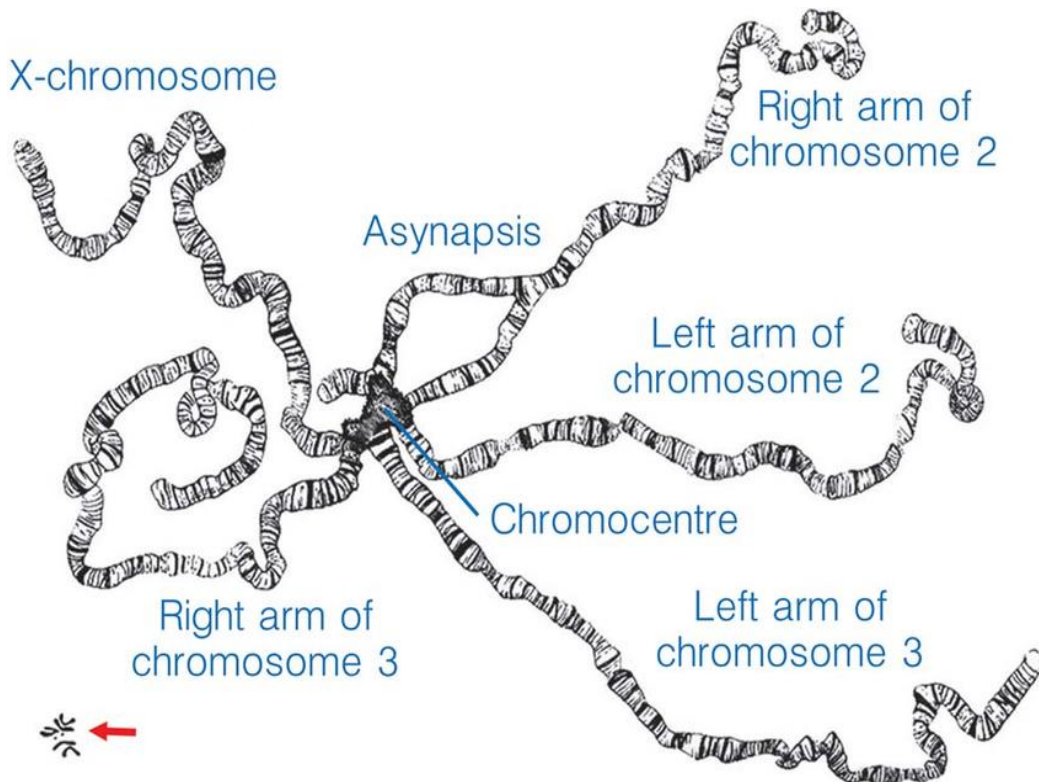


Fig 15.9 Polytene chromosome

The polytene chromosomes of salivary glands in *Drosophila* larvae of certain Diptera

The polytene chromosomes of salivary glands in *Drosophila* larvae can be demonstrated easily in laboratory. It can also be demonstrated in the larvae of Chironomous fly.

Preparation of Polytene Chromosomes from *Drosophila* Larvae:-

Preparation of Culture:-

- Take a small specimen jar. Make a mixture of the pulp of apple, banana and lemon.
- Take 2 gm of moldex add it in 40 ml boiling water.
- Cool it and add in pulp of fruits prepared.
- Place the jar in the culture of *Drosophila* flies, keep it open for 2-3 days during which some of the flies will visit the jar for feeding and lay eggs.
- Now cover the jar with fine muslin cloth. Within a week larvae will appear.

- Observe carefully for 3rd instar larvae which will be white coloured.

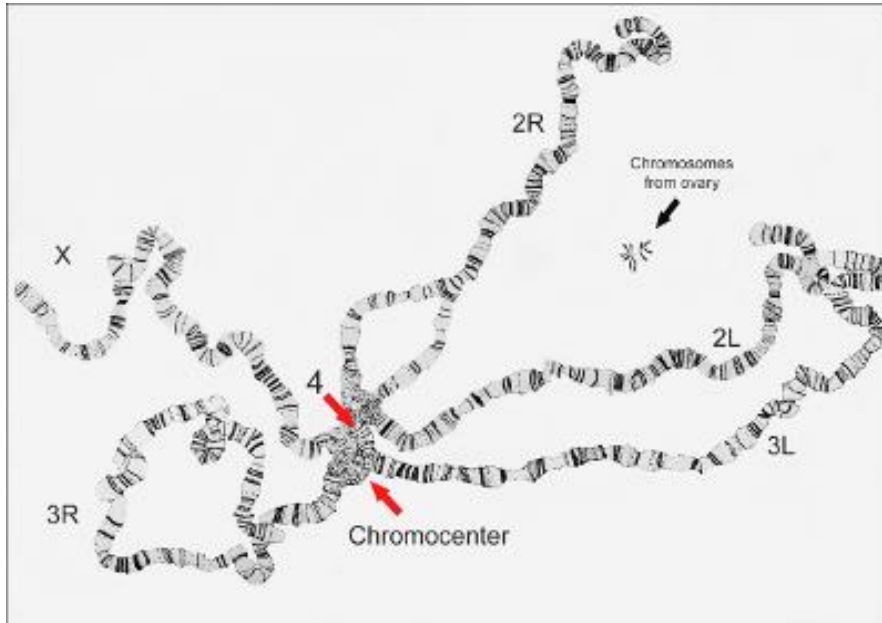


Fig.15.10 Polytene chromosome in the salivary gland of Drosophila

Dissection of 3rd instar larvae for salivary gland: -

- Take a few drop of saline on a clean slide and put the 3rd instlar larvae in it.
- Locate the junction of thorax and abdomen.
- Take two needles, one in each hand. Press the first needle firmly on the posterior end of thorax and other needle at the junction of thorax and abdomen.
- Pull the second needle so that abdomen is separated from head and thorax.
- Then press the thorax with a needle and observe that the salivary glands are seen floating in the saline water on the slide.

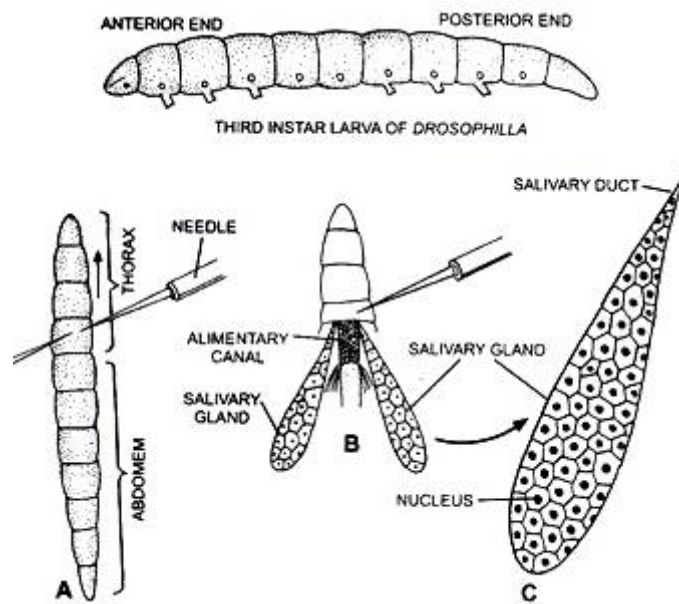


Fig 15.11 Dissection of 3rd instar *Drosophila* larva for salivary gland

Preparation of Slide: -

- Take a clean slide; put a drop of acetocarmine on it.
- Transfer the salivary glands in acetocarmine on slide and cover it with a cover slip.
- Leave it for 10 minutes and then warm it gently and then put a blotting paper over it press it smoothly by your thumb.
- Observe the slide under microscope and for details observe under high power of microscope.

Comments:-

1. These are large-sized, hence, called giant chromosomes.
2. These chromosomes present alternate pattern of dark bands and light inter-bands.
3. The dark bands contain rich amount of DNA and RNA, and composed of much coiled chromonemal thread.
4. The light bands contain rich amount of proteins and little amount of DNA and RNA.
5. A polytene chromosomal is multistranded, it is formed of large number of chromosomal thread or strands.
6. A polytene chromosomes exhibits puffs and Balbiani rings at certain points.
7. The puffs are made of lateral extension of bands of chromosomal strands into side loops.

8. The puffs and Balbiani rings are related with the metabolic activities of the chromosomes.
9. These Chromosomes help in the synthesis of proteins, nucleic acids and formation of nuclear material.
10. These were discovered by Balbiani in 1881.

15.4.3- Study of permanent Slide showing stage of Mitochondria:-

The main components of a typical animal cell are as follows:-

1. Nucleolus
2. Nucleus
3. Ribosome
4. Vesicle
5. Rough endoplasmic reticulum
6. Golgi apparatus (or "Golgi body")
7. Cytoskeleton
8. Smooth endoplasmic reticulum
9. **Mitochondrion**
10. Vacuole
11. Cytosol (fluid that contains organelles, comprising the cytoplasm)
12. Lysosome
13. Centrosome.
14. Cell membrane

The **mitochondrion** (plural **mitochondria**) is a double membrane-bound organelle found in all eukaryotic organisms, although some cells in some organisms may lack them (e.g. Red blood cells). A number of organisms have reduced or transformed their mitochondria into other structures. To date, only one eukaryote is known to have completely lost its **mitochondria**.

The word mitochondrion comes from the Greek, *mitos*, i.e. "thread", and, *chondrion*, i.e. "granule" or "grain-like". Mitochondria have been described as "the powerhouse of the cell" because they generate most of the cell's supply of adenosine triphosphate (ATP), used as a source of chemical energy.

Mitochondria are commonly between 0.75 and 3µm in diameter but vary considerably in size and structure. Unless specifically stained, they are not visible.

In addition to supplying cellular energy, mitochondria are involved in other tasks, such as signaling, cellular differentiation, and cell death, as well as maintaining control of the cell cycle and cell growth. Mitochondrial biogenesis is in turn temporally coordinated with these cellular processes. Mitochondria have been implicated in several human diseases, including mitochondrial disorders, cardiac dysfunction, heart failure and autism.

The number of mitochondria in a cell can vary widely by organism, tissue, and cell type. For instance, red blood cells have no mitochondria, whereas liver cells can have more than 2000. The organelle is composed of compartments that carry out specialized functions.

These compartments or regions include the outer membrane, the inter membrane space, the inner membrane, and the cristae and matrix. Mitochondrial proteins vary depending on the tissue and the species. In humans, 615 distinct types of protein have been identified from cardiac mitochondria, whereas in rats, 940 proteins have been reported. The mitochondrial proteome is thought to be dynamically regulated. Although most of a cell's DNA is contained in the cell nucleus, the mitochondrion has its own independent genome that shows substantial similarity to bacterial genomes.

History:-

The first observations of intracellular structures that probably represented mitochondria were published in the 1840s. Richard Altmann, in 1894, established them as cell organelles and called them "bioblasts". The term "**mitochondria**" was **coined by Carl Benda in 1898**.

Leonor Michaelis discovered that Janus green can be used as a supravital stain for mitochondria in 1900. In 1904, Friedrich Meves, made the first recorded observation of mitochondria in plants in cells of the white waterlily, *Nymphaea alba* and in 1908, along with Claudius Regaud, suggested that they contain proteins and lipids.

Benjamin F. Kingsbury, in 1912, first related them with cell respiration, but almost exclusively based on morphological observations. In 1913, particles from extracts of guinea-pig liver were linked to respiration by Otto Heinrich Warburg, which he called "grana".

Warburg and Heinrich Otto Wieland, who had also postulated a similar particle mechanism, disagreed on the chemical nature of the respiration. It was not until 1925, when David Keilin discovered cytochromes, that the respiratory chain was described.

In 1939, experiments using minced muscle cells demonstrated that cellular respiration using one oxygen atom can form two adenosine triphosphate (ATP) molecules, and, in 1941, the concept of the phosphate bonds of ATP being a form of energy in cellular metabolism was developed by Fritz Albert Lipmann. In the following years, the mechanism behind cellular respiration was further elaborated, although its link to the mitochondria was not known.

The introduction of tissue fractionation by Albert Claude allowed mitochondria to be isolated from other cell fractions and biochemical analysis to be conducted on them alone. In 1946, he concluded that cytochrome oxidase and other enzymes responsible for the respiratory chain were isolated to the mitochondria.

Eugene Kennedy and Albert Lehninger discovered in 1948 that mitochondria are the site of oxidative phosphorylation in eukaryotes. Over time, the fractionation method was further developed, improving the quality of the mitochondria isolated and other elements of cell respiration were determined to occur in the mitochondria.

The first high-resolution electron micrographs appeared in 1952, replacing the Janus Green stains as the preferred way of visualizing the mitochondria. This led to a more detailed analysis of the structure of the mitochondria, including confirmation that they were surrounded by a membrane.

It also showed a second membrane inside the mitochondria that folded up in ridges dividing up the inner chamber and that the size and shape of the mitochondria varied from cell to cell. The popular term "**powerhouse of the cell**" was coined by **Philip Siekevitz in 1957**.

In 1967, it was discovered that mitochondria contained ribosomes. In 1968, methods were developed for mapping the mitochondrial genes, with the genetic and physical map of yeast mitochondrial DNA being completed in 1976.

Origin and evolution:-

There are two hypotheses about the origin of mitochondria, **endosymbiotic and autogenous**. The endosymbiotic hypothesis suggests that mitochondria were **originally prokaryotic** cells, capable of implementing oxidative mechanisms that were not possible for eukaryotic cells; they became endosymbionts living inside the eukaryote.

In the autogenous hypothesis, mitochondria were born by splitting off a portion of DNA from the nucleus of the eukaryotic cell at the time of divergence with the prokaryotes; this DNA portion would have been enclosed by membranes, which could not be crossed by proteins. Since mitochondria have many features in common with bacteria, the most accredited theory at present is endosymbiosis.

A mitochondrion contains DNA, which is organized as several copies of a single, circular chromosome. This mitochondrial chromosome contains genes for redox proteins, such as those of the respiratory chain.

The CoRR hypothesis: CoRR is short form of co-location for redox regulation. CoRR hypothesis proposes that this co-location is required for redox regulation. The mitochondrial genome codes for some RNAs of ribosomes, and the 22 tRNAs necessary for the translation of messenger RNAs into protein. The circular structure is also found in prokaryotes. The proto-mitochondrion was probably closely related to the *Rickettsia*.

However, the exact relationship of the ancestor of mitochondria to the alphaproteobacteria and whether the mitochondrion was formed at the same time or after the nucleus remain controversial.

The ribosomes coded for by the mitochondrial DNA are similar to those from bacteria in size and structure. They closely resemble the bacterial 70S ribosome and not the 80S cytoplasmic ribosomes, which are coded for by nuclear DNA.

The endosymbiotic relationship of mitochondria with their host cells was popularized by Lynn Margulis. The endosymbiotic hypothesis suggests that mitochondria descended from bacteria that somehow survived endocytosis by another cell, and became incorporated into the cytoplasm.

The ability of these bacteria to conduct respiration in host cells that had relied on glycolysis and fermentation would have provided a considerable evolutionary advantage. This symbiotic relationship

probably developed 1.7 to 2 billion years ago.

A few groups of unicellular eukaryotes have only vestigial mitochondria or derived structures: the microsporidians, metamonads, and archamoebae. These groups appear as the most primitive eukaryotes on phylogenetic trees constructed using rRNA information, which once suggested that they appeared before the origin of mitochondria.

However, this is now known to be an artifact of long-branch attraction—they are derived groups and retain genes or organelles derived from mitochondria (e.g., mitosomes and hydrogenosomes).

Structure:-

A mitochondrion has a double membrane; the inner one contains its chemiosmotic apparatus and has deep grooves which increase its surface area. While commonly depicted as an "orange sausage with a blob inside of it" (like it is here), mitochondria can take many shapes and their inter-membrane space is quite thin.

A mitochondrion contains outer and inner membranes composed of phospholipid bilayers and proteins. The two membranes have different properties. Because of this double-membraned organization, there are five distinct parts to a mitochondrion. They are:

1. The outer mitochondrial membrane.
2. The intermembrane space (the space between the outer and inner membranes),
3. The inner mitochondrial membrane,
4. The cristae space (formed by in-folding of the inner membrane), and
5. The matrix (space within the inner membrane).

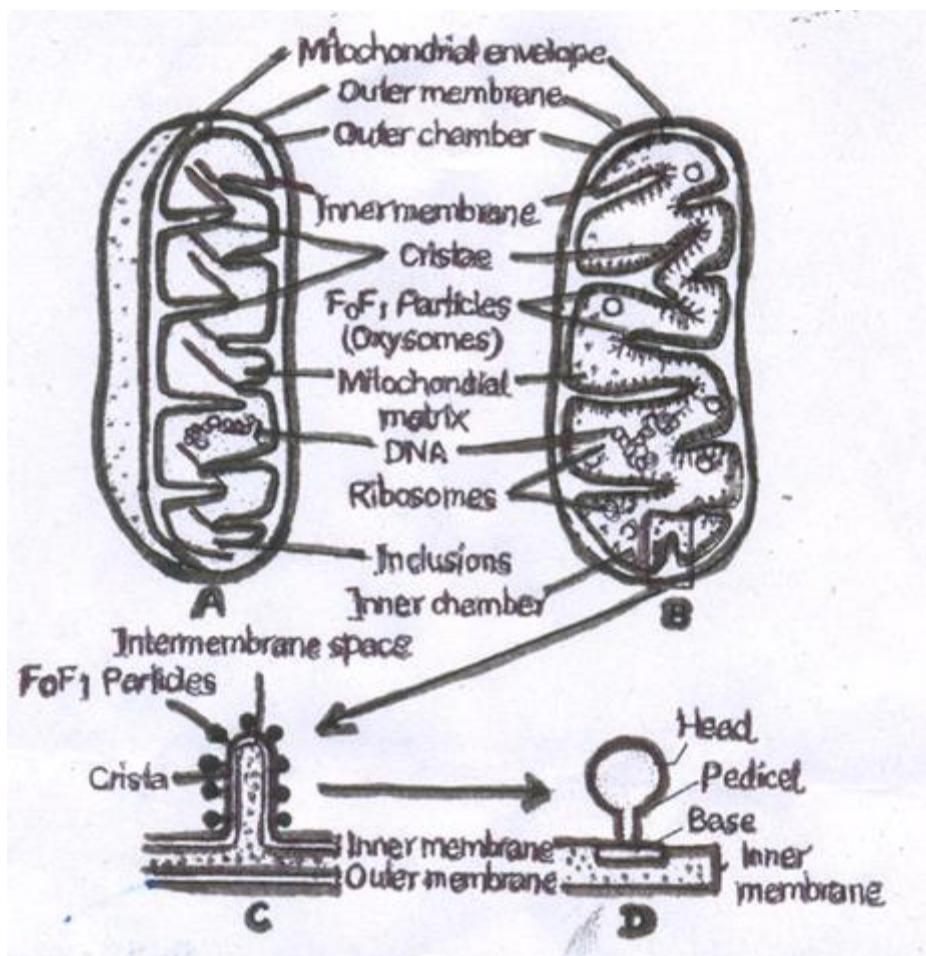


Fig.15.12 Section of Mitochondria showing membrane Chamber and cristae

Mitochondria stripped of their outer membrane are called mitoplasts.

Outer membrane:-

The **outer mitochondrial membrane**, which encloses the entire organelle, is 60 to 75 angstroms (\AA) thick.

It has a protein-to-phospholipid ratio similar to that of the eukaryotic plasma membrane (about 1:1 by weight). It contains large numbers of integral membrane proteins called porins. These porins form channels that allow molecules of 5000 daltons or less in molecular weight to freely diffuse from one side of the membrane to the other.

Larger proteins can enter the mitochondrion if a signaling sequence at their N-terminus binds to a large multisubunit protein called translocase of the outer membrane, which then actively moves them across the membrane.

Mitochondrial pro-proteins are imported through specialised translocation complexes. The outer membrane also contains enzymes involved in such diverse activities as the elongation of fatty acids, oxidation of epinephrine, and the degradation of tryptophan.

These enzymes include monoamine oxidase, rotenone-insensitive NADH-cytochrome c-reductase, kynurenine hydroxylase and fatty acid Co-A ligase. Disruption of the outer membrane permits proteins in the intermembrane space to leak into the cytosol, leading to certain cell death. The mitochondrial outer membrane can associate with the endoplasmic reticulum (ER) membrane, in a structure called MAM (mitochondria-associated ER-membrane).

This is important in the ER-mitochondria calcium signaling and is involved in the transfer of lipids between the ER and mitochondria. Outside the outer membrane there are small (diameter: 60Å) particles named sub-units of Parson.

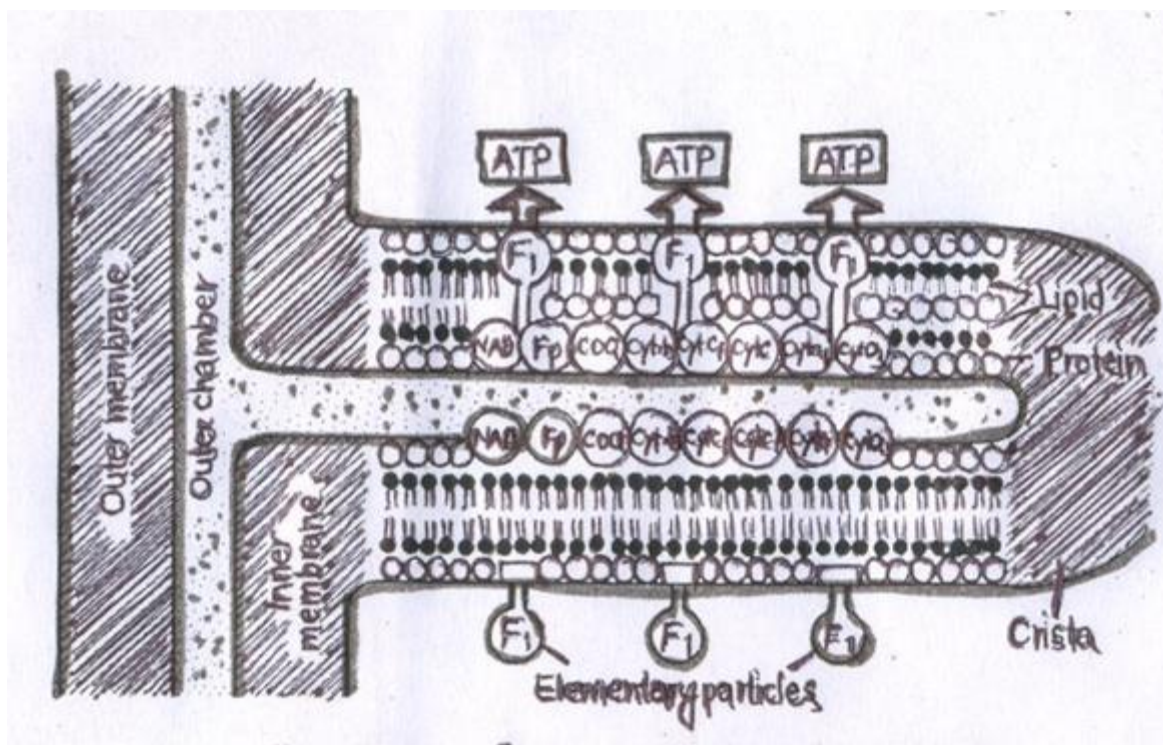


Fig.15.13 Ultrastructure of mitochondrial crest showing F1 particles

Intermembrane space:-

The intermembrane space is the space between the outer membrane and the inner membrane. It is also known as perimitochondrial space. Because the outer membrane is freely permeable to small molecules, the concentrations of small molecules, such as ions and sugars, in the intermembrane space is the same as in the cytosol.

However, large proteins must have a specific signaling sequence to be transported across the outer membrane, so the protein composition of this space is different from the protein composition of the

cytosol. One protein that is localized to the intermembrane space in this way is cytochrome c.

Inner membrane:-

The inner mitochondrial membrane contains proteins with five types of functions:

1. Those that perform the redox reactions of oxidative phosphorylation
2. ATP synthase, which generates ATP in the matrix
3. Specific transport proteins that regulate metabolite passage into and out of the matrix
4. Protein import machinery
5. Mitochondrial fusion and fission protein

It contains more than 151 different polypeptides, and has a very high protein-to-phospholipid ratio (more than 3:1 by weight, which is about 1 protein for 15 phospholipids). The inner membrane is home to around 1/5 of the total protein in a mitochondrion. In addition, the inner membrane is rich in an unusual phospholipid, cardiolipin.

This phospholipid was originally discovered in cow hearts in 1942, and is usually characteristic of mitochondrial and bacterial plasma membranes. Cardiolipin contains four fatty acids rather than two, and may help to make the inner membrane impermeable. Unlike the outer membrane, the inner membrane doesn't contain porins, and is highly impermeable to all molecules. Almost all ions and molecules require special membrane transporters to enter or exit the matrix.

Proteins are ferried into the matrix via the translocase of the inner membrane (TIM) complex or via Oxa1. In addition, there is a membrane potential across the inner membrane, formed by the action of the enzymes of the electron transport chain.

Cristae:-

The inner mitochondrial membrane is compartmentalized into numerous cristae, which expand the surface area of the inner mitochondrial membrane, enhancing its ability to produce ATP. For typical liver mitochondria, the area of the inner membrane is about five times as large as the outer membrane.

This ratio is variable and mitochondria from cells that have a greater demand for ATP, such as muscle cells, contain even more cristae. These folds are studded with small round bodies known as F₁ particles or oxysomes. These are not simple random folds but rather invaginations of the inner membrane, which can affect overall chemiosmotic function.

One recent mathematical modeling study has suggested that the optical properties of the cristae in filamentous mitochondria may affect the generation and propagation of light within the tissue.

Matrix:-

The matrix is the space enclosed by the inner membrane. It contains about 2/3 of the total protein in a mitochondrion. The matrix is important in the production of ATP with the aid of the ATP synthase contained in the inner membrane.

The matrix contains a highly concentrated mixture of hundreds of enzymes, special mitochondrial ribosomes, tRNA, and several copies of the mitochondrial DNA genome. Of the enzymes, the major functions include oxidation of pyruvate and fatty acids, and the citric acid cycle.

Mitochondria have their own genetic material, and the machinery to manufacture their own RNAs and proteins. A published human mitochondrial DNA sequence revealed 16,569 base pairs encoding 37 genes: 22 tRNA, 2 rRNA, and 13 peptide genes.

The 13 mitochondrial peptides in humans are integrated into the inner mitochondrial membrane, along with proteins encoded by genes that reside in the host cell's nucleus.

Mitochondria-associated ER membrane:-

The mitochondria-associated ER membrane (MAM) is another structural element that is increasingly recognized for its critical role in cellular physiology and homeostasis. Once considered a technical snag in cell fractionation techniques, the alleged ER vesicle contaminants that invariably appeared in the mitochondrial fraction have been re-identified as membranous structures derived from the MAM—the interface between mitochondria and the ER. Physical coupling between these two organelles had previously been observed in electron micrographs and has more recently been probed with fluorescence microscopy.

Such studies estimate that at the MAM, which may comprise up to 20% of the mitochondrial outer membrane, the ER and mitochondria are separated by a mere 10–25 nm and held together by protein tethering complexes.

Purified MAM from subcellular fractionation has been shown to be enriched in enzymes involved in phospholipid exchange, in addition to channels associated with Ca^{2+} signaling. These hints of a prominent role for the MAM in the regulation of cellular lipid stores and signal transduction have been borne out, with significant implications for mitochondrial-associated cellular phenomena, as discussed below.

Not only has the MAM provided insight into the mechanistic basis underlying such physiological processes as intrinsic apoptosis and the propagation of calcium signaling, but it also favors a more refined view of the mitochondria.

Though often seen as static, isolated 'powerhouses' hijacked for cellular metabolism through an ancient endosymbiotic event, the evolution of the MAM underscores the extent to which mitochondria have been integrated into overall cellular physiology, with intimate physical and functional coupling to the endomembrane system.

Phospholipids transfer:-

The MAM is enriched in enzymes involved in lipid biosynthesis, such as phosphatidylserine synthase on the ER face and phosphatidylserine decarboxylase on the mitochondrial face. Because mitochondria are dynamic organelles constantly undergoing fission and fusion events, they require a constant and well-regulated supply of phospholipids for membrane integrity.

But mitochondria are not only a destination for the phospholipids they finish synthesis of; rather, this organelle also plays a role in inter-organelle trafficking of the intermediates and products of phospholipid biosynthetic pathways, ceramide and cholesterol metabolism, and glycosphingolipid anabolism.

Such trafficking capacity depends on the MAM, which has been shown to facilitate transfer of lipid intermediates between organelles. In contrast to the standard vesicular mechanism of lipid transfer, evidence indicates that the physical proximity of the ER and mitochondrial membranes at the MAM allows for lipid flipping between opposed bilayers.

Despite this unusual and seemingly energetically unfavorable mechanism, such transport does not require ATP. Instead, in yeast, it has been shown to be dependent on a multiprotein tethering structure termed the ER-mitochondria encounter structure, or ERMES, although it remains unclear whether this structure directly mediates lipid transfer or is required to keep the membranes in sufficiently close proximity to lower the energy barrier for lipid flipping.

The MAM may also be part of the secretory pathway, in addition to its role in intracellular lipid trafficking. In particular, the MAM appears to be an intermediate destination between the rough ER and the Golgi in the pathway that leads to very-low-density lipoprotein, or VLDL, assembly and secretion. The MAM thus serves as a critical metabolic and trafficking hub in lipid metabolism.

Calcium signaling:-

A critical role for the ER in calcium signaling was acknowledged before such a role for the mitochondria was widely accepted, in part because the low affinity of Ca^{2+} channels localized to the outer mitochondrial membrane seemed to fly in the face of this organelle's purported responsiveness to changes in intracellular Ca^{2+} flux.

But the presence of the MAM resolves this apparent contradiction: the close physical association between the two organelles results in Ca^{2+} microdomains at contact points that facilitate efficient Ca^{2+} transmission from the ER to the mitochondria. Transmission occurs in response to so-called " Ca^{2+} puffs" generated by spontaneous clustering and activation of IP3R, a canonical ER membrane Ca^{2+} channel.

The fate of these puffs—in particular, whether they remain restricted to isolated locales or integrated into Ca^{2+} waves for propagation throughout the cell—is determined in large part by MAM dynamics. Although reuptake of Ca^{2+} by the ER (concomitant with its release) modulates the intensity of the puffs, thus insulating mitochondria to a certain degree from high Ca^{2+} exposure, the MAM often serves as a firewall that essentially buffers Ca^{2+} puffs by acting as a sink into which free ions released into the cytosol can be funneled. This Ca^{2+} tunneling occurs through the low-affinity Ca^{2+} receptor VDAC1, which recently has been shown to be physically tethered to the IP3R clusters on the ER membrane and enriched at the MAM.

The ability of mitochondria to serve as a Ca^{2+} sink is a result of the electrochemical gradient generated during oxidative phosphorylation, which makes tunneling of the cation an exergonic process.

Normally, mild calcium influx from cytosol into the mitochondrial matrix causes transient depolarization that is corrected by pumping out protons.

But transmission of Ca^{2+} is not unidirectional; rather, it is a two-way street. The properties of the Ca^{2+} pump SERCA and the channel IP3R present on the ER membrane facilitate feedback regulation coordinated by MAM function. In particular, the clearance of Ca^{2+} by the MAM allows for spatio-temporal patterning of Ca^{2+} signaling because Ca^{2+} alters IP3R activity in a biphasic manner.

SERCA is likewise affected by mitochondrial feedback: uptake of Ca^{2+} by the MAM stimulates ATP production, thus providing energy that enables SERCA to reload the ER with Ca^{2+} for continued Ca^{2+} efflux at the MAM. Thus, the MAM is not a passive buffer for Ca^{2+} puffs; rather it helps modulate further Ca^{2+} signaling through feedback loops that affect ER dynamics.

Regulating ER release of Ca^{2+} at the MAM is especially critical because only a certain window of Ca^{2+} uptake sustains the mitochondria, and consequently the cell, at homeostasis. Sufficient intraorganelle

Ca^{2+} signaling is required to stimulate metabolism by activating dehydrogenase enzymes critical to flux through the citric acid cycle. However, once Ca^{2+} signaling in the mitochondria passes a certain threshold, it stimulates the intrinsic pathway of apoptosis in part by collapsing the mitochondrial membrane potential required for metabolism.

Studies examining the role of pro- and anti-apoptotic factors support this model; for example, the anti-apoptotic factor Bcl-2 has been shown to interact with IP3Rs to reduce Ca^{2+} filling of the ER, leading to reduced efflux at the MAM and preventing collapse of the mitochondrial membrane potential post-apoptotic stimuli. Given the need for such fine regulation of Ca^{2+} signaling, it is perhaps unsurprising that dysregulated mitochondrial Ca^{2+} has been implicated in several neurodegenerative diseases, while the catalogue of tumor suppressors includes a few that are enriched at the MAM.

Molecular basis for tethering:-

Recent advances in the identification of the tethers between the mitochondrial and ER membranes suggest that the scaffolding function of the molecular elements involved is secondary to other, non-structural functions. In yeast, ERMES, a multiprotein complex of interacting ER- and mitochondrial-resident membrane proteins, is required for lipid transfer at the MAM and exemplifies this principle.

One of its components, for example, is also a constituent of the protein complex required for insertion of transmembrane beta-barrel proteins into the lipid bilayer. However, a homologue of the ERMES complex has not yet been identified in mammalian cells.

Other proteins implicated in scaffolding likewise have functions independent of structural tethering at the MAM; for example, ER-resident and mitochondrial-resident mitofusins form heterocomplexes that regulate the number of inter-organelle contact sites, although mitofusins were first identified for their role in fission and fusion events between individual mitochondria.

Glucose-related protein 75 (grp75) is another dual-function protein. In addition to the matrix pool of grp75, a portion serves as a chaperone that physically links the mitochondrial and ER Ca^{2+} channels VDAC and IP3R for efficient Ca^{2+} transmission at the MAM. Another potential tether is Sigma-1R, a non-opioid receptor whose stabilization of ER-resident IP3R may preserve communication at the MAM during the metabolic stress response.

Function:-

The most prominent roles of mitochondria are to produce the energy currency of the cell, ATP (i.e., phosphorylation of ADP), through respiration, and to regulate cellular metabolism.

The central sets of reactions involved in ATP production are collectively known as the citric acid cycle, or the Krebs cycle. However, the mitochondrion has many other functions in addition to the production of ATP.

Energy conversion:-

A dominant role for the mitochondria is the production of ATP, as reflected by the large number of proteins in the inner membrane for this task. This is done by oxidizing the major products of glucose: pyruvate, and NADH, which are produced in the cytosol.

This type of cellular respiration known as aerobic respiration, is dependent on the presence of oxygen. When oxygen is limited, the glycolytic products will be metabolized by anaerobic fermentation, a process that is independent of the mitochondria.

The production of ATP from glucose has an approximately 13-times higher yield during aerobic respiration compared to fermentation. Recently it has been shown that plant mitochondria can produce a limited amount of ATP without oxygen by using the alternate substrate nitrite.

ATP crosses out through the inner membrane with the help of a specific protein, and across the outer membrane via porins. ADP returns via the same route.

Additional functions:-

Mitochondria play a central role in many other metabolic tasks, such as:

- Signaling through mitochondrial reactive oxygen species
- Regulation of the membrane potential
- Apoptosis-programmed cell death
- Calcium signaling (including calcium-evoked apoptosis)
- Regulation of cellular metabolism
- Certain heme synthesis reactions Steroid synthesis.
- Hormonal signaling

Mitochondria are sensitive and responsive to hormones, in part by the action of mitochondrial estrogen

receptors (mtERs). These receptors have been found in various tissues and cell types, including brain and heart

Some mitochondrial functions are performed only in specific types of cells. For example, mitochondria in liver cells contain enzymes that allow them to detoxify ammonia, a waste product of protein metabolism. A mutation in the genes regulating any of these functions can result in mitochondrial diseases.

Mitochondrial diseases:-

Damage and subsequent dysfunction in mitochondria is an important factor in a range of human diseases due to their influence in cell metabolism. Mitochondrial disorders often present themselves as neurological disorders, including autism.

They can also manifest as myopathy, diabetes, multiple endocrinopathy, and a variety of other systemic disorders. Diseases caused by mutation in the mtDNA include Kearns-Sayre syndrome, MELAS syndrome and Leber's hereditary optic neuropathy.

In the vast majority of cases, these diseases are transmitted by a female to her children, as the zygote derives its mitochondria and hence its mtDNA from the ovum. Diseases such as Kearns-Sayre syndrome, Pearson syndrome, and progressive external ophthalmoplegia are thought to be due to large-scale mtDNA rearrangements, whereas other diseases such as MELAS syndrome, Leber's hereditary optic neuropathy, myoclonic epilepsy with ragged red fibers (MERRF), and others are due to point mutations in mtDNA.

In other diseases, defects in nuclear genes lead to dysfunction of mitochondrial proteins. This is the case in Friedreich's ataxia, hereditary spastic paraplegia, and Wilson's disease.

These diseases are inherited in a dominance relationship, as applies to most other genetic diseases. A variety of disorders can be caused by nuclear mutations of oxidative phosphorylation enzymes, such as coenzyme Q10 deficiency and Barth syndrome. Environmental influences may interact with hereditary predispositions and cause mitochondrial disease.

For example, there may be a link between pesticide exposure and the later onset of Parkinson's disease. Other pathologies with etiology involving mitochondrial dysfunction include schizophrenia, bipolar disorder, dementia, Alzheimer's disease, Parkinson's disease, epilepsy, stroke, cardiovascular disease, chronic fatigue syndrome, retinitis pigmentosa, and diabetes mellitus.

Mitochondria-mediated oxidative stress plays a role in cardiomyopathy in Type 2 diabetics. Increased fatty acid delivery to the heart increases fatty acid uptake by cardiomyocytes, resulting in increased fatty acid oxidation in these cells. This process increases the reducing equivalents available to the electron transport chain of the mitochondria, ultimately increasing reactive oxygen species (ROS) production. ROS increases uncoupling proteins (UCPs) and potentiate proton leakage through the adenine nucleotide translocator (ANT), the combination of which uncouples the mitochondria.

Uncoupling then increases oxygen consumption by the mitochondria, compounding the increase in fatty acid oxidation. This creates a vicious cycle of uncoupling; furthermore, even though oxygen consumption increases, ATP synthesis does not increase proportionally because the mitochondrion is uncoupled.

Less ATP availability ultimately results in an energy deficit presenting as reduced cardiac efficiency and contractile dysfunction. To compound the problem, impaired sarcoplasmic reticulum calcium release and reduced mitochondrial reuptake limits peak cytosolic levels of the important signaling ion during muscle contraction.

The decreased intra-mitochondrial calcium concentration increases dehydrogenase activation and ATP synthesis. So in addition to lower ATP synthesis due to fatty acid oxidation, ATP synthesis is impaired by poor calcium signaling as well, causing cardiac problems for diabetics.

15.4.4- Study of permanent Slide showing stage of Golgi body:-

The **Golgi body** also known as the **Golgi complex**, **Golgi apparatus**, or simply the **Golgi**, is an organelle found in most eukaryotic cells. It was identified in 1897 by the Italian scientist Camillo Golgi and named after him in 1898.

Part of the cellular endomembrane system, the Golgi apparatus packages proteins into membrane-bound vesicles inside the cell before the vesicles are sent to their destination. The Golgi apparatus resides at the intersection of the secretory, lysosomal, and endocytic pathways.

It is of particular importance in processing proteins for secretion, containing a set of glycosylation enzymes that attach various sugar monomers to proteins as the proteins move through the apparatus.

Owing to its large size and distinctive structure, the Golgi apparatus was one of the first organelles to be discovered and observed in detail. It was discovered in 1898 by Italian physician Camillo Golgi during an investigation of the nervous system. After first observing it under his microscope, he termed the structure the internal reticular apparatus. Some doubted the discovery at first, arguing that the appearance of the structure was merely an optical illusion created by the observation technique used by Golgi.

With the development of modern microscopes in the 20th century, the discovery was confirmed. Early references to the Golgi referred to it by various names including the "Golgi–Holmgren apparatus", "Golgi–Holmgren ducts", and "Golgi–Kopsch apparatus". The term "Golgi apparatus" was used in 1910 and first appeared in the scientific literature in 1913.

Among eukaryotes, the sub cellular localization of the Golgi apparatus differs. In mammals, a single Golgi apparatus complex is usually located near the cell nucleus, close to the centrosome. Tubular connections are responsible for linking the stacks together. Localization and tubular connections of the Golgi apparatus are dependent on microtubules. If microtubules are experimentally depolymerized, then the Golgi apparatus loses connections and becomes individual stacks throughout the cytoplasm. In yeast, multiple Golgi apparatuses are scattered throughout the cytoplasm.

In plants, Golgi stacks are not concentrated at the centrosomal region and do not form Golgi ribbons. Organization of the plant Golgi depends on actin cables and not microtubules. The common feature among Golgi is that they are adjacent to endoplasmic reticulum (ER) exit sites.

Structure:-

In most eukaryotes, the Golgi apparatus is made up of a series of compartments consisting of two main networks: the cis Golgi network (CGN) and the Trans Golgi network (TGN). The CGN is a collection of fused, flattened membrane-enclosed disks known as cisternae, originating from vesicular clusters that bud off the endoplasmic reticulum.

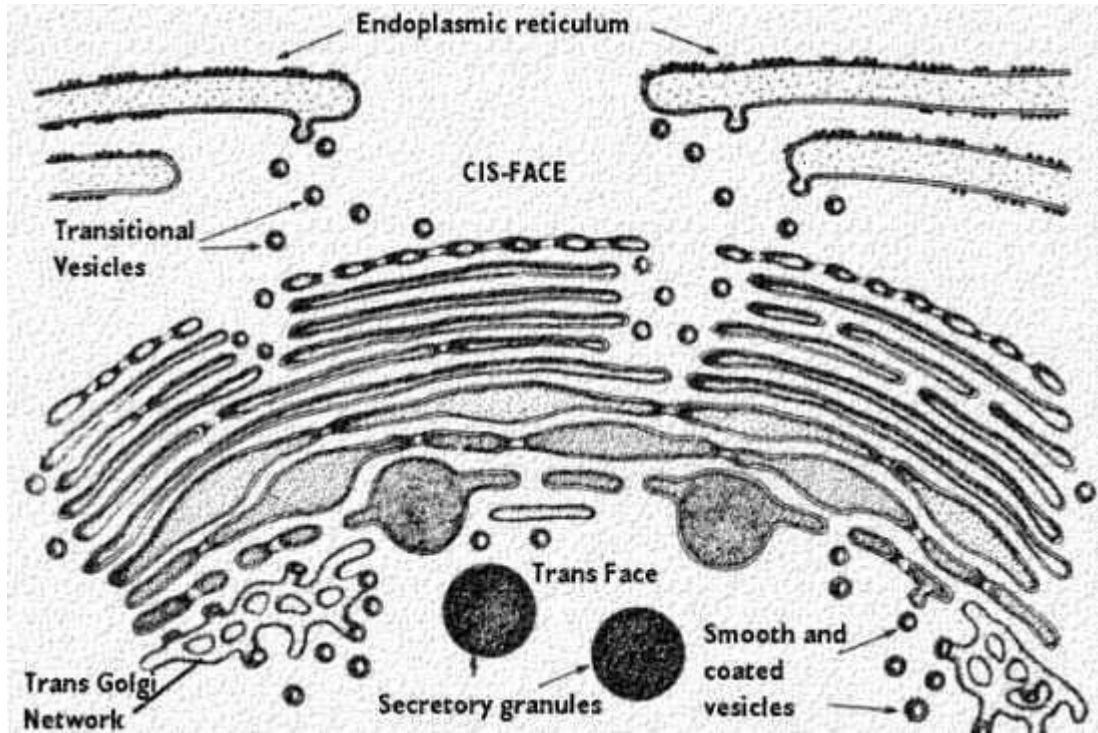


Fig.15.14 Relation between different component of Golgi body & their relation with secretion

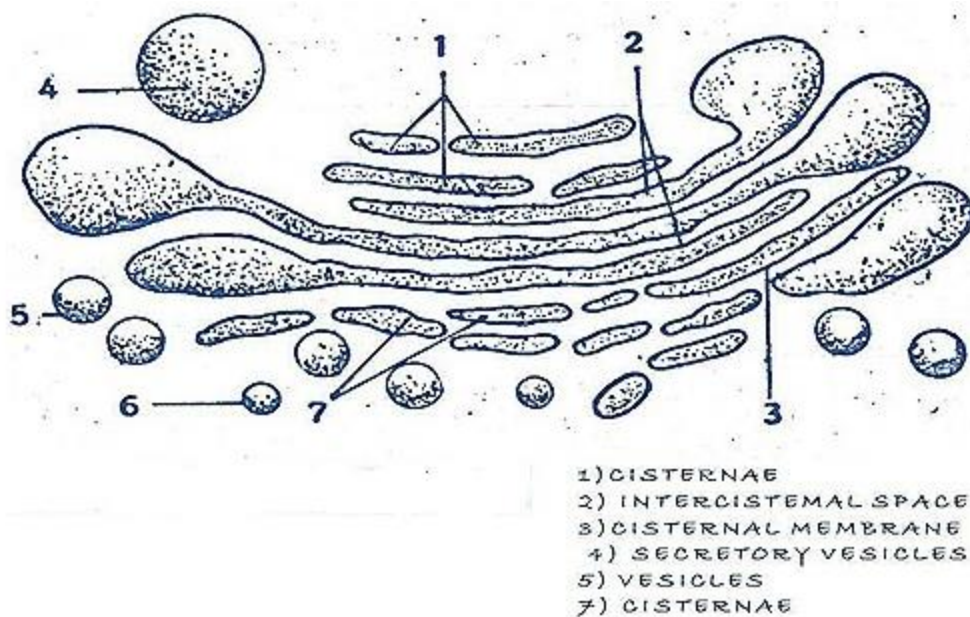


Fig.15.15 Ultrastructure of Golgi complex

A mammalian cell typically contains 40 to 100 stacks. Between four and eight cisternae are usually present in a stack; however, in some protists as many as sixty cisternae have been observed. This collection of cisternae is broken down into cis, medial, and Trans compartments.

The TGN is the final cisternal structure, from which proteins are packaged into vesicles destined to lysosomes, secretory vesicles, or the cell surface. The TGN is usually positioned adjacent to the stacks of the Golgi apparatus, but can also be separate from the stacks. The TGN may act as an early endosome in yeast and plants.

There are structural and organizational differences in the Golgi apparatus among eukaryotes. In some yeasts, Golgi stacking is not observed. *Pichia pastoris* does have stacked Golgi, while *Saccharomyces cerevisiae* does not. In plants, the individual stacks of the Golgi apparatus seem to operate independently.

The Golgi apparatus tends to be larger and more numerous in cells that synthesize and secrete large amounts of substances; for example, the antibody-secreting plasma B cells of the immune system have prominent Golgi complexes.

In all eukaryotes, each cisternal stack has a cis entry face and a trans exit face. These faces are characterized by unique morphology and biochemistry. Within individual stacks are assortments of enzymes responsible for selectively modifying protein cargo.

These modifications influence the fate of the protein. The compartmentalization of the Golgi apparatus is advantageous for separating enzymes, thereby maintaining consecutive and selective processing steps:

enzymes catalyzing early modifications are gathered in the cis face cisternae, and enzymes catalyzing later modifications are found in Trans face cisternae of the Golgi stacks.

Function:-

The Golgi apparatus is a major collection and dispatch station of protein products received from the endoplasmic reticulum (ER). Proteins synthesized in the ER are packaged into vesicles, which then fuse with the Golgi apparatus.

These cargo proteins are modified and destined for secretion via exocytosis or for use in the cell. In this respect, the Golgi can be thought of as similar to a post office: it packages and labels items which it then sends to different parts of the cell or to the extracellular space. The Golgi apparatus is also involved in lipid transport and lysosome formation.

The structure and function of the Golgi apparatus are intimately linked. Individual stacks have different assortments of enzymes, allowing for progressive processing of cargo proteins as they travel from the cis to the trans Golgi face. Enzymatic reactions within the Golgi stacks occur exclusively near its membrane surfaces, where enzymes are anchored.

This feature is in contrast to the ER, which has soluble proteins and enzymes in its lumen. Much of the enzymatic processing is post-translational modification of proteins. For example, phosphorylation of oligosaccharides on lysosomal proteins occurs in the early CGN. Cis cisterna is associated with the removal of mannose residues. Removal of mannose residues and addition of N-acetylglucosamine occur in medial cisternae.

Addition of galactose and sialic acid occurs in the trans cisternae. Sulfation of tyrosines and carbohydrates occurs within the TGN. Other general post-translational modifications of proteins include the addition of carbohydrates (glycosylation) and phosphates (phosphorylation). Protein modifications may form a signal sequence that determines the final destination of the protein. For example, the Golgi apparatus adds a mannose-6-phosphate label to proteins destined for lysosomes.

Another important function of the Golgi apparatus is in the formation of proteoglycans. Enzymes in the Golgi append proteins to glycosaminoglycans, thus creating proteoglycans. Glycosaminoglycans are long unbranched polysaccharide molecules present in the extracellular matrix of animals.

Vesicular transport:-

The vesicles that leave the rough endoplasmic reticulum are transported to the cis face of the Golgi apparatus, where they fuse with the Golgi membrane and empty their contents into the lumen. Once inside the lumen, the molecules are modified, and then sorted for transport to their next destinations.

Those proteins destined for areas of the cell other than either the endoplasmic reticulum or the Golgi apparatus are moved through the Golgi cisternae towards the trans face, to a complex network of membranes and associated vesicles known as the trans-Golgi network (TGN). This area of the Golgi is the point at which proteins are sorted and shipped to their intended destinations by their placement into one of at least three different types of vesicles, depending upon the signal sequence they carry.

Current models of vesicular transport and trafficking:-

Model 1: Anterograde vesicular transport between stable compartments:-

- In this model, the Golgi is viewed as a set of stable compartments that work together. Each compartment has a unique collection of enzymes that work to modify protein cargo. Proteins are delivered from the ER to the *cis* face using COPII-coated vesicles. Cargo then progress toward the *trans* face in COPI-coated vesicles. This model proposes that COPI vesicles move in two directions: anterograde vesicles carry secretory proteins, while retrograde vesicles recycle Golgi-specific trafficking proteins.

Strengths: The model explains observations of compartments, polarized distribution of enzymes, and waves of moving vesicles. It also attempts to explain how Golgi-specific enzymes are recycled.

Weaknesses: Since the amount of COPI vesicles varies drastically among types of cells, this model cannot easily explain high trafficking activity within the Golgi for both small and large cargoes. Additionally, there is no convincing evidence that COPI vesicles move in both the anterograde and retrograde directions.[12]

- This model was widely accepted from the early 1980s until the late 1990s.

Model 2: Cisternal progression/maturation:-

In this model, the fusion of COPII vesicles from the ER begins the formation of the first cis-cisterna of the Golgi stack, which progresses later to become mature TGN cisternae. Once matured, the TGN cisternae dissolves to become secretory vesicles. While this progression occurs, COPI vesicles continually recycle Golgi-specific proteins by delivery from older to younger cisternae. Different recycling patterns may account for the differing biochemistry throughout the Golgi stack. Thus, the compartments within the Golgi are seen as discrete kinetic stages of the maturing Golgi apparatus.

Strengths: The model addresses the existence of Golgi compartments, as well as differing biochemistry within the cisternae, transport of large proteins, transient formation and disintegration of the cisternae, and retrograde mobility of native Golgi proteins, and it can account for the variability seen in the structures of the Golgi.

Weaknesses: This model cannot easily explain the observation of fused Golgi networks, tubular connections among cisternae, and differing kinetics of secretory cargo exit.

Model 3: Cisternal progression/maturation with heterotypic tubular transport:-

This model is an extension of the cisternal progression/maturation model. It incorporates the existence of tubular connections among the cisternae that form the Golgi ribbon, in which cisternae within a stack are linked. This model posits that the tubules are important for bidirectional traffic in

the ER-Golgi system: they allow for fast anterograde traffic of small cargo and/or the retrograde traffic of native Golgi proteins.

Strengths: This model encompasses the strengths of the cisternal progression/maturation model that also explains rapid trafficking of cargo, and how native Golgi proteins can recycle independently of COPI vesicles.[12]

Weaknesses: This model cannot explain the transport kinetics of large protein cargo, such as collagen. Additionally, tubular connections are not prevalent in plant cells. The roles that these connections have can be attributed to a cell-specific specialization rather than a universal trait. If the membranes are continuous, that suggests the existence of mechanisms that preserve the unique biochemical gradients observed throughout the Golgi apparatus.

Model 4: Rapid partitioning in a mixed Golgi:-

This rapid partitioning model is the most drastic alteration of the traditional vesicular trafficking point of view. Proponents of this model hypothesize that the Golgi works as a single unit, containing domains that function separately in the processing and export of protein cargo. Cargo from the ER moves between these two domains, and randomly exits from any level of the Golgi to their final location. This model is supported by the observation that cargo exits the Golgi in a pattern best described by exponential kinetics. The existence of domains is supported by fluorescence microscopy data.

Strengths: Notably, this model explains the exponential kinetics of cargo exit of both large and small proteins whereas other models cannot.

Weaknesses: This model cannot explain the transport kinetics of large protein cargo, such as collagen. This model falls short on explaining the observation of discrete compartments and polarized biochemistry of the Golgi cisternae. It also does not explain formation and disintegration of the Golgi network, nor the role of COPI vesicles.

Model 5: Stable compartments as cisternal model progenitors:-

This is the most recent model. In this model, the Golgi is seen as a collection of stable compartments defined by Rab (G-protein) GTPases.

Strengths: This model is consistent with numerous observations and encompasses some of the strengths of the cisternal progression/maturation model. Additionally, what is known of the Rab GTPase roles in mammalian endosomes can help predict putative roles within the Golgi. This model is unique in that it can explain the observation of "megavesicle" transport intermediates.

Weaknesses: This model does not explain morphological variations in the Golgi apparatus, nor define a role for COPI vesicles. This model does not apply well for plants, algae, and fungi in which individual Golgi stacks are observed (transfer of domains between stacks is not likely). Additionally, megavesicles are not established to be intra-Golgi transporters.

Though there are multiple models that attempt to explain vesicular traffic throughout the Golgi, no individual model can independently explain all observations of the Golgi apparatus. Currently, the cisternal progression/maturation model is the most accepted among scientists, accommodating many observations across eukaryotes. The other models are still important in framing questions and guiding future experimentation. Among the fundamental unanswered questions are the directionality of COPI vesicles and role of Rab GTPases in modulating protein cargo traffic.

15.6- Summary:-

Cytology, branch of biology concerned with the study of the structure and function of cells as individual units, supplementing histology, which deals with cells as components of tissues. Cytology is concerned with the structure and activities of the various parts of the cell and cell membrane; the mechanism of cell division; the development of sex cells, fertilization, and the formation of the embryo; cell derangements, such as those occurring in cancer; cellular immunity; and the problems of heredity.

Until modern times, cytology was concerned primarily with the microscopic observation of stained dead cells and the correlation of such observations with known physiological phenomena. Recently, new procedures have been introduced by which the living cell can be observed and studied. The phase-contrast microscope provides a means of studying the living cell in action without the use of dyes. Micro dissection, microinjection, and microchemistry furnish methods for drawing off minute amounts of living protoplasm through tubes a half micron in diameter, and subjecting them to analysis.

Cytology is important in modern medicine, especially in the diagnosis of diseases by examination of the cells occurring in the various body fluids. The determination of the number and proportion of the different types of cells in the blood, by a blood count, is important in diagnosing acute infections and other diseases. Variations in the size and shape of the red blood cell indicate the presence of: sickle-cell anemia if the cell is half-moon shaped; pernicious anemia if it is very large; or iron-deficiency anemia if it is very small. The type of disease may also be determined through cytology, as, for example, in distinguishing the various types of meningitis by examination of the cells present in the cerebrospinal fluid.

15.7 –Glossary:-

Active Transport:	The movements of ions or molecules of a substance through the plasma membrane from a solution of low concentration to a solution of high concentration i.e. against electro-chemical gradient. The process needs energy.
Amino Acid	Organic compounds with acidic (-COOH) and amino (-NH ₂), groups; 20 of which, different in organic chain attached to carbon atom, are the structural units of protein macromolecules.
Amitosis	Direct division of nucleus into two, without differentiation of chromosomes and formation of spindle etc.
Anaphase	A stage in nuclear division immediately after metaphase and is followed by telophase. It is characterized by the movement of sets of daughter chromosomes from the equatorial plate towards the opposite poles of the spindle.
Aneuploids	The organism having chromosomes of a set parent in different numbers
Autolysis	Disintegration of cells by the action of their own enzymes
Budding	A mode of asexual reproduction in which new organism develops from the parent

	body in the form of an outgrowth or projection.
Catalase	An enzyme which facilitates conversion of hydrogen peroxide to water and oxygen
Cell division	The process of division of pre-existing (parental) cell into two new daughter cells.
Chromatin	Deeply stained part of the nuclear reticulum mostly of DNA, which condenses into chromosomes during cell division.
Chromomere	Irregular masses of heterochromatin
Coenzyme	Organic compound which activates the enzyme.
Colloid	Substances having particles which range from 1μ to 100μ in size
Conjugation	Temporary association between the organisms of two different strains so as to facilitate nuclear exchange
Cytolysis	Dissolution or disintegration of a cell
Deletion	Loss of segment from a chromosome
Diplotene	A stage in the first prophase of meiosis, in which each of the synaptic chromosomes get doubled by splitting. It comes after pachytene and is followed by diakinesis
Germ cell	As gamete
Gonad	Gamete producing organ
Haploid	Having half the number of chromosomes that are present in the diploid organism. Usually the gametes..
Heterogamy	Darkely stained part of the chromatin in the interphase nucleus which represents the condensed chromatin and results due to failure of its conversion into a nuclear reticulum.
Matrix	Intercellular substances in which animal cells are embedded
Micron	A unit of measurement: $1/1000$ mm usually designated by the Greek letter μ .
Operon	A group of genes that are transcribed into a single length messenger RNA for a single character
Osmosis	The passage of a fluid through a semi-permiable membrane due to osmotic pressure
Pachytene	Midprophase stage of first reduction division (or meiosis) in which the chromosomes are visible as long paired threads
Polar body	Bodies extruded out during oogenesis one after each maturation division
Promotor	A site on a chromosome where RNA-polymerase binds and initiates RNA synthesis
Recombination	The appearance in an individual of alleles for different characters that were not present together in either parent
Spindle	The chromatic figure formed during cell division by the differentiation of cytoplasm into radiating fibres which are disposed in such a manner that these form a spindle figure. The equator of spindle provides surface for the orientation of chromosomes
Tetrad	The group of four chromatids resulting from the pairing of homologous chromosomes and their splitting during 1 st prophase of meiosis
Triploids	Organism having three haploid sets of chromosomes i.e. $3n$
Unit membrane	The membrane formed of two layers of lipid molecules sandwiched between the two layers of protein molecules. It forms the outer boundary of almost all the cell organelles.

15.8: Self assessment Questions

1. Who proposed the cell theory?
 2. Mitochondria are generally called the power houses of cell. Why?
 3. What are the special types of chromosomes?
-

4. What are the difference between mitosis and meiosis?
5. What do you understand by meiotic division?

15.9: References:-

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De Robertes & Robertis: Cell & Molecular Biology, 1987, Lee & Fibiger Philadelplna.
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Brachet.j. and e. Mirsky (1959). The living Cell, Sci.Am. 205, (3) 50.
Freeman, J.A., (1954), Cellular Fine Structure, McGraw-Hill Book Company, New York.
Landley, L.L. (1968). Cell Function, 2nd ed. Van Nostrand Reinhold Company, New York.

15.10: Suggested Readings:-

Cell Biology-Harvey Lodish,
Dr. S.P. Singh, Cytology – Prof. P.K.Gupta

15.11: Terminal Questions:-

1. Describe the structure and function of Mitochondria.
2. What are Lampbrush Chromosomes? Mention its structure and significance.
3. What is cell theory? Give an illustrated account of the structure and function of golgi body.
4. Discuss the methods of preparation and dissection of 3rd instar larvae for salivary gland.
5. Write short notes any two of the following.
(a) Polytene Chromosome (b) Cristae © Karyokinesis

UNIT 16: GENETICAL EXPERIMENT

16.1- Objectives

16.2-Introduction

16.3- Experimentation on Mendelian inheritance.

16.4- Experimentation on non Mendelian inheritance

16.5. – Study of mutant *Drosophila* through chart and photographs

16.6- Genetical Exercise

16.6- Summary

16.7- Glossary

16.8-Self Assessment Question

16.9- References

16.10- Terminal Question

16.1- OBJECTIVES

We will develop the practical understanding on Mendelian and non-Mendelian hereditary experiments. Genetics, scientific study of how physical, biochemical, and behavioral traits are transmitted from parents to their offspring's. The word genetics was coined in 1906 by the British biologist William Bateson.

16.2 INTRODUCTION:-

Genetics deals with the mechanism of heredity and causes of variations in living beings. The heredity or inheritance, in turn, deals with the process of transmission of characters from one generation to another. Several interesting experiments based on the principles and laws of heredity can be conducted in our biological laboratories. However, the exercises of genetics have been planned in the following ways to illustrate some basic principles of heredity. Geneticists determine the mechanisms of inheritance whereby the offspring of sexually reproducing organisms do not exactly resemble their parents, and the differences and similarities between parents and offspring recur from generation to generation in repeated patterns. The investigation of these patterns has led to some of the most exciting discoveries in modern biology. The science of genetics began in 1900, when several plant breeders independently discovered the work of the Austrian monk Gregor Johann Mendel. He published his work in 1866. His work remained unnoticed for decades and gained posthumous recognition as father of modern genetics. Working with garden peas, Mendel described the patterns of inheritance in terms of seven pairs of contrasting traits that appeared in different pea-plant varieties. He observed that the traits were inherited as separate units, each of which was inherited independently of the others. He suggested that each parent has pairs of units but contributes only one unit from each pair to its offspring. The units that Mendel described were later given the name genes. Soon after Mendel's work was rediscovered, scientists realized that the patterns of inheritance he had described paralleled the action of chromosomes in dividing cells, and they proposed that the Mendelian units of inheritance, the genes, are carried by the chromosomes. This led to intensive study of cell division. Every cell comes from the division of a pre-existing cell. All the cells that make up a human being, for example, are derived from the successive divisions of a single cell, the zygote, which is formed by the union of an egg and a sperm.

The great majority of the cells produced by the division of the zygote are, in the composition of their hereditary material, identical to one another and to the zygote itself. Each cell of a higher organism is composed of a jellylike layer of material, the cytoplasm, which contains many small structures. This cytoplasm material surrounds a prominent body called the nucleus. Every nucleus contains a number of minute, threadlike chromosomes. Some relatively simple organisms such as cyan bacteria and bacteria, have no distinct nucleus but do have cytoplasm, which contains one or more chromosomes. Chromosomes vary in size and shape and usually occur in pairs. The members of each pair, called homologues, closely resemble each other. Most cells in the human body contain 23 pairs of chromosomes, whereas most cells of the fruit fly *Drosophila* contain four pairs, and the bacterium *Escherichia coli* has a single chromosome in the form of a ring. Every chromosome in a cell is now known to contain many genes, and each gene is located at a particular site, or locus, on the chromosome. The process of cell division by which a new cell comes to have an identical number of chromosomes as the parent cell is called mitosis. In mitotic division each chromosome divides into two equal parts, and the two parts travel to opposite ends of the cell. After the cell divides, each of the two resulting cells has the same

number of chromosomes and genes as the original cell. Every cell formed in this process thus has the same genetic material. Simple one-celled organisms and some multicellular forms reproduce by mitosis; it is also the process by which complex organisms achieve growth and replace worn-out tissue. Higher organisms that reproduce sexually are formed from the union of two special sex cells known as gametes. Gametes are produced by meiosis, the process by which germ cells divide. It differs from mitosis in one important way: in meiosis a single chromosome from each pair of chromosomes is transmitted from the original cell to each of the new cells.

Thus, each gamete contains half the number of chromosomes that are found in the other body cells. When two gametes unite in fertilization, the resulting cell, called the zygote, contains the full, double set of chromosomes. Half of these chromosomes normally come from one parent and half from the other.

16.3- EXPERIMENTATION ON MENDELIAN INHERITANCE

Mendelian inheritance is inheritance of biological features that follows the laws proposed by Gregor Johann Mendel in 1865 and 1866 and re-discovered in 1900.

It was initially very controversial. When Mendel's theories were integrated with the Boveri–Sutton chromosome theory of inheritance by **Thomas Hunt Morgan in 1915**, they became the core of classical genetics while Ronald Fisher combined them with the theory of natural selection in his 1930 book

The Genetical Theory of Natural Selection, putting evolution onto a mathematical footing and forming the basis for Population genetics and the modern evolutionary synthesis.

The laws of inheritance were derived by Gregor Mendel, a nineteenth-century Austrian monk, and later Prälet, conducting hybridization experiments in garden peas (*Pisum sativum*) he planted in the backyard of the church. Between 1856 and 1863, he cultivated and tested some 5,000 pea plants.

From these experiments, he induced two generalizations which later became known as Mendel's Principles of Heredity or **Mendelian inheritance**.

He described these principles in a two-part paper, *Versuche über Pflanzen-Hybriden*, that he read to the Natural History Society of Brno (Brunn) on 8 February and 8 March 1865, and which was published in 1866.

Mendel's conclusions were largely ignored. Although they were not completely unknown to biologists of the time, they were not seen as generally applicable, even by Mendel himself, who thought they only applied to certain categories of species or traits.

A major block to understanding their significance was the importance attached by 19th-century biologists to the apparent blending of inherited traits in the overall appearance of the progeny, now known to be due to multigame interactions, in contrast to the organ-specific binary characters studied by Mendel.

In 1900, however, his work was "re-discovered" by three European scientists, Hugo de Vries, Carl Correns, and Erich von Tschermak.

The exact nature of the "re-discovery" has been somewhat debated: De Vries published first on the subject, mentioning Mendel in a footnote, while Correns pointed out Mendel's priority after having read De Vries' paper and realizing that he himself did not have priority.

De Vries may not have acknowledged truthfully how much of his knowledge of the laws came from his own work, or came only after reading Mendel's paper.

Later scholars have accused Von Tschermak of not truly understanding the results at all.

Regardless, the "**re-discovery**" made Mendelism an important but controversial theory. Its most vigorous promoter in Europe was William Bateson, who coined the terms "genetics" and "allele" to describe many of its tenets.

The model of heredity was highly contested by other biologists because it implied that heredity was discontinuous, in opposition to the apparently continuous variation observable for many traits.

Many biologists also dismissed the theory because they were not sure it would apply to all species.

However, later work by biologists and statisticians such as Ronald Fisher showed that if multiple Mendelian factors were involved in the expression of an individual trait, they could produce the diverse results observed, and thus showed that Mendelian genetics is compatible with natural selection.

Thomas Hunt Morgan and his assistants later integrated the theoretical model of Mendel with the chromosome theory of inheritance, in which the chromosomes of cells were thought to hold the actual hereditary material, and created what is now known as classical genetics, which was extremely successful and cemented Mendel's place in history.

Mendel's findings allowed scientists such as Fisher and J.B.S. Haldane to predict the expression of traits on the basis of mathematical probabilities.

A large contribution to Mendel's success can be traced to his decision to start his crosses only with plants he demonstrated were true-breeding. He also only measured absolute (binary) characteristics, such as colour, shape, and position of the offspring, rather than quantitative characteristics. He expressed his results numerically and subjected them to statistical analysis.

His method of data analysis and his large sample size gave credibility to his data. He also had the foresight to follow several successive generations (F₂, F₃) of pea plants and record their variations.

Finally, he performed "test crosses" (back-crossing descendants of the initial hybridization to the initial true-breeding lines) to reveal the presence and proportion of recessive characters.

Mendel observed seven traits that are easily recognized and apparently only occur in one of two forms:

1. Flower colour is purple or white
2. Seed colour is yellow or green
3. Flower position is axial or terminal
4. Pod shape is inflated or constricted
5. Stem length is long or short
6. Pod colour is yellow or green
7. Seed shape is round or wrinkled

MENDEL'S LAWS:-

Mendel discovered that, when he crossed purebred white flower and purple flower pea plants (the parental or P generation), the result was not a blend. Rather than being a mix of the two, the offspring (known as the F₁ generation) was purple-flowered.

When Mendel self-fertilized the F₁ generation pea plants, he obtained a purple flower to white flower ratio in the F₂ generation of 3 to 1. The results of this cross are tabulated in the Punnett square to the right.

He then conceived the idea of heredity units, which he called "**factors**". Mendel found that there are alternative forms of factors—now called genes—that account for variations in inherited characteristics. For example, the gene for flower colour in pea plants exists in two forms, one for purple and the other for white.

The alternatives “forms” are now called alleles. For each biological trait, an organism inherits two alleles, one from each parent. These alleles may be the same or different.

An organism that has two identical alleles for a gene is said to be homozygous for that gene (and is called a homozygote). An organism that has two different alleles for a gene is said by **heterozygous** for that gene (and is called a **heterozygote**).

Mendel also hypothesized that allele pairs separate randomly, or segregate, from each other during the production of gametes: egg and sperm.

Because allele pairs separate during gamete production, a sperm or egg carries only one allele for each inherited trait. When sperm and egg unite at fertilization, each contributes its allele, restoring the paired condition in the offspring.

INDEPENDENT ASSORTMENT: -

The genotype of an individual is made up of the many alleles it possesses. An individual's physical appearance, or phenotype, is determined by its alleles as well as by its environment. The presence of an allele does not mean that the trait will be expressed in the individual that possesses it.

If the two alleles of an inherited pair differ (the heterozygous condition), then one determines the organism's appearance and is called the dominant allele; the other has no noticeable effect on the organism's appearance and is called the recessive allele.

Thus, in the example above dominant purple flower allele will hide the phenotypic effects of the recessive white flower allele.

This is known as the Law of Dominance but it is not a transmission law, dominance has to do with the expression of the genotype and not its transmission. The upper case letters are used to represent dominant alleles whereas the lowercase letters are used to represent recessive alleles.

MENDEL'S LAWS OF INHERITANCE:-

1. Law of dominance:

Some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.

2. Law of segregation:

During gamete formation, the alleles for each gene segregate from each other so that each gamete carries only one allele for each gene.

3. Law of independent assortment:

Genes for different traits can segregate independently during the formation of gametes.

In the pea plant example above, the capital "P" represents the dominant allele for purple flowers and lowercase "p" represents the recessive allele for white flowers.

Both parental plants were true-breeding, and one parental variety had two alleles for purple flowers (*PP*) while the other had two alleles for white flowers (*pp*). As a result of fertilization, the F_1 hybrids each inherited one allele for purple flowers and one for white.

All the F_1 hybrids (*Pp*) had purple flowers, because the dominant *P* allele has its full effect in the heterozygote, while the recessive *p* allele has no effect on flower color.

For the F_2 plants, the ratio of plants with purple flowers to those with white flowers (3:1) is called the phenotypic ratio. The genotypic ratio, as seen in the Punnett square, is 1 *PP*: 2 *Pp*: 1 *pp*.

LAW OF DOMINANCE:-

Mendel's Law of Dominance states that recessive alleles will always be masked by dominant alleles.

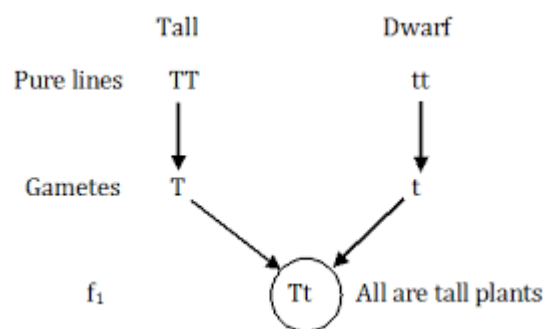
Therefore, a cross between a homozygous dominant and a homozygous recessive will always express the dominant phenotype, while still having a heterozygous genotype.

Law of Dominance can be explained easily with the help of a mono hybrid cross experiment:- In a cross between two organisms pure for any pair (or pairs) of contrasting traits (characters), the character that appears in the F1 generation is called "dominant" and the one which is suppressed (not expressed) is called "recessive." Each character is controlled by a pair of dissimilar factors.

Only one of the characters expresses. The one which expresses in the F1 generation is called Dominant. It is important to note however, that the law of dominance is significant and true but is not universally applicable.

According to the latest revisions, only two of these rules are considered to be laws. The third one is considered as a basic principle but not a genetic law of Mendel.

Example:



Mendelian Trait:-

A Mendelian trait is one that is controlled by a single locus in an inheritance pattern. In such cases, a mutation in a single gene can cause a disease that is inherited according to Mendel's laws.

Examples include sickle-cell anemia, Tay-Sachs disease, cystic fibrosis and xeroderma pigmentosa.

A disease controlled by a single gene contrasts with a multi-factorial disease, like arthritis, which is affected by several loci (and the environment) as well as those diseases inherited in a non-Mendelian fashion.

Law of Segregation of genes:-

The Law of Segregation states that every individual organism contains two alleles for each trait, and that these alleles segregate (separate) during meiosis such that each gamete contains only one of the alleles.

An offspring thus receives a pair of alleles for a trait by inheriting homologous chromosomes from the parent organisms: one allele for each trait from each parent.

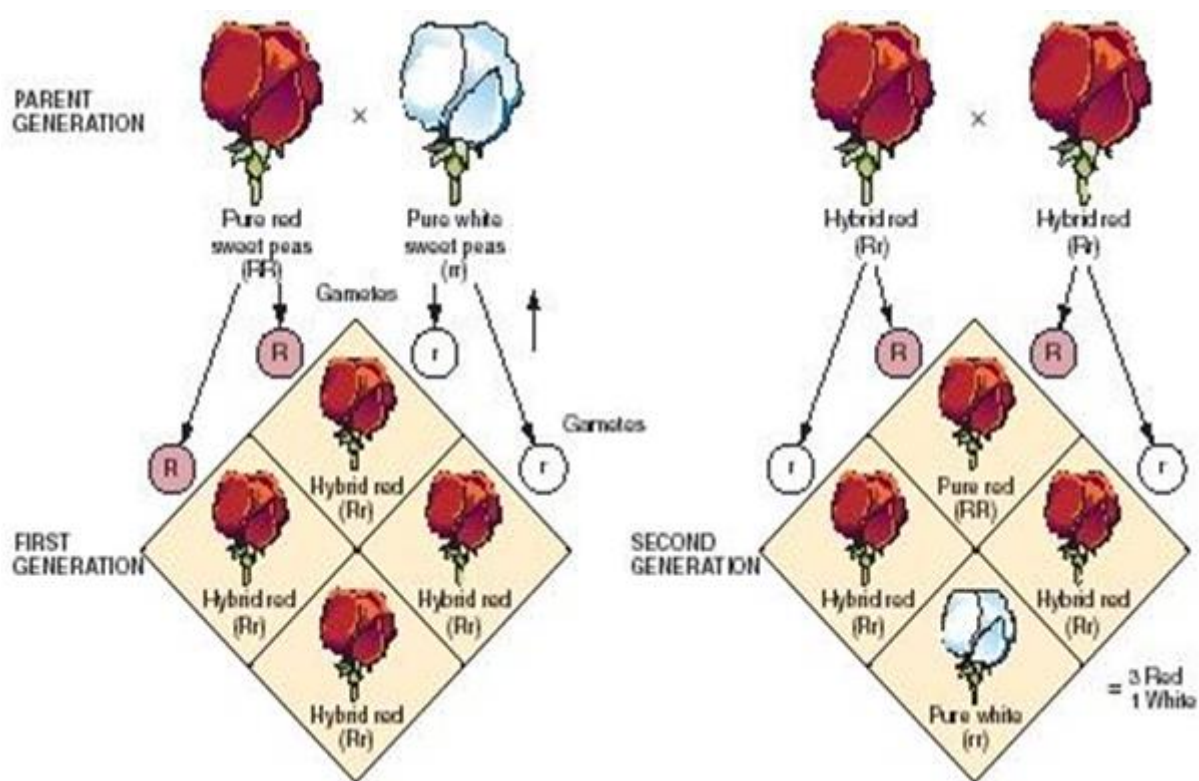


Fig.16.1 Mendel first law: Law of segregation

Molecular proof of this principle was subsequently found through observation of meiosis by two scientists independently, the German botanist Oscar Hertwig in 1876, and the Belgian zoologist Edouard Van Beneden in 1883.

Paternal and maternal chromosomes get separated in meiosis and the alleles with the traits of a character are segregated into two different gametes.

Each parent contributes a single gamete, and thus a single, randomly successful allele copy to their offspring and fertilization.

Explanation:-

Mendel's law of segregation describes what happens to the alleles that make up a gene during formation of gametes.

For example, suppose that a pea plant contains a gene for flower color in which both alleles code for red.

One way to represent that condition is to write RR, which indicates that both alleles (R and R) code for the color red. Another gene might have a different combination of alleles, as in Rr. In this case, the symbol R stands for red color and the r for "not red" or, in this case, white.

Mendel's law of segregation says that the alleles that make up a gene separate from each other, or segregate, during the formation of gametes. That fact can be represented by simple equations, such as:

$$\mathbf{RR \rightarrow R + R \text{ or } Rr \rightarrow R + r}$$

Mendel's second law is called the law of independent assortment. That law refers to the fact that any plant contains many different kinds of genes.

One gene determines flower color, a second gene determines length of stem, and a third gene determines shape of pea pods, and so on.

Mendel discovered that the way in which alleles from different genes separate and then recombine is unconnected to other genes.

That is, suppose that a plant contains genes for color (RR) and for shape of pod (TT). Then Mendel's second law says that the two genes will segregate independently, as:

$$\mathbf{RR \rightarrow R + R \text{ and } TT \rightarrow T + T}$$

Mendel's third law deals with the matter of dominance. Suppose that a gene contains an allele for red color (R) and an allele for white color (r).

What will be the color of the flowers produced on this plant? Mendel's answer was that in every pair of alleles, one is more likely to be expressed than the other. In other words, one allele is dominant and the other allele is recessive.

In the example of an Rr gene, the flowers produced will be red because the allele R is dominant over the allele r.

Law of Independent Assortment:-

The Law of Independent Assortment states that alleles for separate traits are passed independently of one another from parents to offspring.

That is, the biological selection of an allele for one trait has nothing to do with the selection of an allele for any other trait. Mendel found support for this law in his dihybrid cross experiments.

In his monohybrid crosses, an idealized 3:1 ratio between dominant and recessive phenotypes resulted. In dihybrid crosses, however, he found a 9:3:3:1 ratios.

This shows that each of the two alleles is inherited independently from the other, with a 3:1 phenotypic ratio for each.

Independent assortment occurs in eukaryotic organisms during meiotic prophase I, and produces a gamete with a mixture of the organism's chromosomes.

The physical basis of the independent assortment of chromosomes is the random orientation of each bivalent chromosome along the metaphase plate with respect to the other bivalent chromosomes.

Along with crossing over, independent assortment increases genetic diversity by producing novel genetic combinations.

There are many violations of independent assortment due to genetic linkage. Of the 46 chromosomes in a normal diploid human cell, half are maternally derived (from the mother's egg) and half are paternally derived (from the father's sperm).

This occurs as sexual reproduction involves the fusion of two haploid gametes (the egg and sperm) to produce a new organism having the full complement of chromosomes.

During gametogenesis—the production of new gametes by an adult—the normal complement of 46 chromosomes needs to be halved to 23 to ensure that the resulting haploid gamete can join with another gamete to produce a diploid organism.

An error in the number of chromosomes, such as those caused by a diploid gamete joining with a haploid gamete, is termed aneuploidy.

In independent assortment, the chromosomes that result are randomly sorted from all possible maternal and paternal chromosomes.

Because zygotes end up with a random mix instead of a pre-defined "set" from either parent, chromosomes are therefore considered assorted independently.

As such, the zygote can end up with any combination of paternal or maternal chromosomes. Any of the possible variants of a zygote formed from maternal and paternal chromosomes will occur with equal frequency.

For human zygotes, with 23 pairs of chromosomes, the number of possibilities is 2^{23} or 8,388,608 possible combinations.

The zygote will normally end up with 23 chromosomes pairs, but the origin of any particular chromosome will be randomly selected from paternal or maternal chromosomes.

This contributes to the genetic variability of progeny.

To this point we have followed the expression of only one gene. Mendel also performed crosses in which he followed the segregation of two genes.

These experiments formed the basis of his discovery of his second law, the law of independent assortment. First, a few terms are presented.

Dihybrid Cross: -

A cross between two parents that differ by two pairs of alleles (AABB x aabb)

Dihybrid: -

An individual heterozygous for two pairs of alleles (AaBb), again a dihybrid cross is not a cross between two dihybrids. At a dihybrid cross that Mendel performed.

Parental Cross:-

Yellow, Round Seed x Green, Wrinkled Seed

F1 Generation:-

All yellow, rounds

F2 Generation:-

9 Yellow, Round, 3 Yellow, Wrinkled, 3 Green, Round, 1 Green, Wrinkled

Symbol

Seed Color:

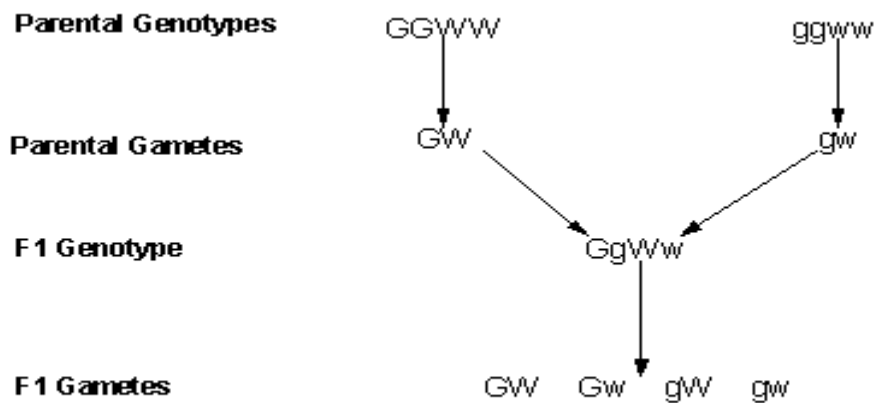
Yellow = G; Green = g

Seed Shape: -

Round = W; Wrinkled = w

The dominance relationship between alleles for each trait was already known to Mendel when he made this cross.

The purpose of the dihybrid cross was to determine if any relationship existed between different allelic pairs.



The Punnett Square for the F₂ cross.

		Female Gametes			
		GW	Gw	gW	gw
Male Gametes	GW	GGWW (Yellow, round)	GGWw (Yellow, round)	GgWW (Yellow, round)	GgWw (Yellow, round)
	Gw	GGWw (Yellow, round)	GGww (Yellow, wrinkled)	GgWw (Yellow, round)	Ggww (Yellow, wrinkled)
	gW	GgWW (Yellow, round)	GgWw (Yellow, round)	ggWW (Green, round)	ggWw (Green, round)
	gw	GgWw (Yellow, round)	Ggww (Yellow, wrinkled)	ggWw (Green, round)	ggww (Green, wrinkled)

The phenotypes and general genotypes from this cross can be represented in the following manner:

Phenotype	General Genotype
9 Yellow, Round Seed	G_W_
3 Yellow, Wrinkled Seed	G_ww
3 Green, Round Seed	ggW_
1 Green, Wrinkled Seed	ggww

The phenotypes and general genotypes from this cross can be represented in the following manner: The results of this experiment led Mendel to formulate his second law.

16.4- EXPERIMENTATION ON NON MENDELIAN INHERITANCE:-

Non-Mendelian inheritance is a general term that refers to any pattern of inheritance in which traits do not segregate in accordance with Mendel's laws.

These laws describe the inheritance of traits linked to single genes on chromosomes in the nucleus.

In Mendelian inheritance, each parent contributes one of two possible alleles for a trait. If the genotypes of both parents in a genetic cross are known, Mendel's laws can be used to determine the distribution of phenotypes expected for the population of offspring.

There are several situations in which the proportions of phenotypes observed in the progeny do not match the predicted values.

Although inheritance of traits in fungi, viruses, and bacteria are all non-Mendelian, the phrase "non-Mendelian inheritance" is usually only used to describe the exceptions which occur in eukaryotic reproduction. Non-Mendelian inheritance plays a role in several disease processes.

The F1 offspring of Mendel's pea crosses always looked like one of the two parental varieties.

In this situation of "complete dominance," the dominant allele had the same phenotypic effect whether present in one or two copies. But for some characteristics, the F1 hybrids have an appearance in between the phenotypes of the two parental varieties.

A cross between two four o'clock (*Mirabilis Jalapa*) plants shows this common exception to Mendel's principles.

Some alleles are neither dominant nor recessive. The F1 generation produced by a cross between red-flowered (RR) and white flowered (WW) *Mirabilis jalapa* plant consists of pink-colored flowers (RW). Which allele is dominant in this case? Neither one.

This third phenotype results from flowers of the heterozygote having less red pigment than the red homozygotes. Cases in which one allele is not completely dominant over another are called incomplete dominance. In incomplete dominance, the heterozygous phenotype lies somewhere between the two homozygous phenotypes.

A similar situation arises from co-dominance, in which the phenotypes produced by both alleles are clearly expressed.

For example, in certain varieties of chicken, the allele for black feathers is codominant with the allele for white feathers. Heterozygous chickens have a color described as "erminette," speckled with black and white feathers.

Unlike the blending of red and white colors in heterozygous four o'clock, black and white colours appear separately in chickens. Many human genes, including one for a protein that controls cholesterol levels in the blood, show co-dominance, too.

People with the heterozygous form of this gene produce two different forms of the protein, each with a different effect on cholesterol levels.

In Mendelian inheritance, genes have only two alleles, such as *a* and *A*. In nature, such genes exist in several different forms and are therefore said to have multiple alleles. A gene with more than two alleles is said to have multiple alleles.

An individual, of course, usually has only two copies of each gene, but many different alleles are often found within a population.

One of the best-known examples is coat colour in rabbits. A rabbit's coat colour is determined by a single gene that has at least four different alleles.

The four known alleles display a pattern of simple dominance that can produce four coat colours. Many other genes have multiple alleles, including the human genes for ABO blood type.

Furthermore, many traits are produced by the interaction of several genes. Traits controlled by two or more genes are said to be polygenic traits. Polygenic means "many genes."

For example, at least three genes are involved in making the reddish-brown pigment in the eyes of fruit flies. Polygenic traits often show a wide range of phenotypes.

The variety of skin colour in humans comes about partly because more than four different genes probably control this trait.

Extra-nuclear inheritance:-

Extranuclear inheritance (also known as cytoplasmic inheritance) is a form of non-Mendelian inheritance first discovered by Carl Correns in 1908.

While working with *Mirabilis jalapa* Correns observed that leaf colour was dependent only on the genotype of the maternal parent. Based on these data, he determined that the trait was transmitted through a character present in the cytoplasm of the ovule.

Later research by Ruth Sager and others identified DNA present in chloroplasts as being responsible for the unusual inheritance pattern observed.

Work on the poky strain of the mold *Neurospora crassa* begun by Mary and Hershel Mitchell ultimately led to the discovery of genetic material in mitochondria as well.

According to the endosymbiont theory, mitochondria and chloroplasts were once free living organisms that were each taken up by a eukaryotic cell. Over time, mitochondria and chloroplasts formed a symbiotic relationship with their eukaryotic hosts.

Although the transfer of a number of genes from these organelles to the nucleus prevents them from living independently, each still possesses genetic material in the form of double stranded DNA.

It is the transmission of this organellar DNA that is responsible for the phenomenon of extranuclear inheritance. Both chloroplasts and mitochondria are present in the cytoplasm of maternal gametes only.

Paternal gametes (sperm for example) do not have cytoplasmic mitochondria. Thus, the phenotype of traits linked to genes found in either chloroplasts or mitochondria are determined exclusively by the maternal parent.

In humans, mitochondrial diseases are a class of diseases, many of which affect the muscles and the eye.

Gene conversion:-

Gene conversion can be one of the major forms of non-Mendelian inheritance. Gene conversion is a reparation process in DNA recombination, by which a piece of DNA sequence information is transferred from one DNA helix (which remains unchanged) to another DNA helix, whose sequence is altered.

This may occur as a mismatch repair between the strands of DNA which are derived from different parents.

Thus the mismatch repair can convert one allele into the other. This phenomenon can be detected through the offspring non-Mendelian ratios, and is frequently observed, e.g., in fungal crosses.

Infectious heredity:-

Another form of non-Mendelian inheritance is known as infectious heredity. Infectious particles such as viruses may infect host cells and continue to reside in the cytoplasm of these cells.

If the presence of these particles results in an altered phenotype, then this phenotype may be subsequently transmitted to progeny.

Because this phenotype is dependent only on the presence of the invader in the host cell's cytoplasm, inheritance will be determined only by the infected status of the maternal parent.

This will result in a uniparental transmission of the trait, just as in extranuclear inheritance.

One of the most well studied examples of infectious heredity is the killer phenomenon exhibited in yeast. Two double-stranded RNA viruses, designated L and M, are responsible for this phenotype.

The L virus codes for the capsid proteins of both viruses, as well as an RNA polymerase. Thus the M virus can only infect cells already harboring L virus particles.

The M viral RNA encodes a toxin which is secreted from the host cell.

It kills susceptible cells growing in close proximity to the host. The M viral RNA also renders the host cell immune to the lethal effects of the toxin.

For a cell to be susceptible it must therefore be either uninfected, or harbor only the L virus.

The L and M viruses are not capable of exiting their host cell through conventional means. They can only transfer from cell to cell when their host undergoes mating.

All progeny of a mating involving a doubly infected yeast cell will also be infected with the L and M viruses. Therefore, the killer phenotype will be passed down to all progeny.

Heritable traits that result from infection with foreign particles have also been identified in *Drosophila*. Wild type flies normally full recover after being anesthetized with carbon dioxide.

Certain lines of flies have been identified that die off after exposure to the compound. This carbon dioxide sensitivity is passed down from mothers to their progeny. This sensitivity is due to infection with σ (Sigma) virus, a rhabdovirus only capable of infecting *Drosophila*.

Although this process is usually associated with viruses, recent research has shown that the *Wolbachia bacterium* is also capable of inserting its genome into that of its host.

Genomic imprinting:-

Genomic imprinting represents yet another example of non-Mendelian inheritance. Just as in conventional inheritance, genes for a given trait are passed down to progeny from both parents.

However, these genes are epigenetically marked before transmission, altering their levels of expression.

These imprints are created before gamete formation and are erased during the creation of germ line cells. Therefore, a new pattern of imprinting can be made with each generation.

Genes are imprinted differently depending on the parental origin of the chromosome that contains them. In mice, the insulin-like growth factor 2 gene undergoes imprinting.

The protein encoded by this gene helps to regulate body size. Mice that possess two functional copies of this gene are larger than those with two mutant copies.

The size of mice that are heterozygous at this locus depends on the parent from which the wild type allele came.

If the functional allele originated from the mother, the offspring will exhibit dwarfism, whereas a paternal allele will generate a normal sized mouse.

This is because the maternal Igf2 gene is imprinted. Imprinting results in the inactivation of the Igf2 gene on the chromosome passed down by the mother.

Imprints are formed due to the differential methylation of paternal and maternal alleles. This results in differing expression between alleles from the two parents. Sites with significant methylation are associated with low levels of gene expression.

Higher gene expression is found at unmethylated sites. In this mode of inheritance, phenotype is determined not only by the specific allele transmitted to the offspring, but also by the sex of the parent that transmitted it.

Mosaicism:-

Individuals who possess cells with genetic differences from the other cells in their body are termed mosaics. These differences can result from mutations that occur in different tissues and at different periods of development.

If a mutation happens in the non-gamete forming tissues, it is characterized as somatic. Germline mutations occur in the egg or sperm cells and can be passed on to offspring.

Mutations that occur early on in development will affect a greater number of cells and can result in an individual that can be identified as a mosaic strictly based on phenotype.

Mosaicism also results from a phenomenon known as X-inactivation. All female mammals have two X chromosomes. To prevent lethal gene dosage problems, one of these chromosomes is inactivated following fertilization.

This process occurs randomly for all of the cells in the organism's body. Because a given female's two X chromosomes will almost certainly differ in their specific pattern of alleles, this will result in differing cell phenotypes depending on which chromosome is silenced.

Calico cats, which are almost all female, demonstrate one of the most commonly observed manifestations of this process.

Trinucleotide repeat disorders:-

Trinucleotide repeat disorders also follow a non-Mendelian pattern of inheritance.

These diseases are all caused by the expansion of microsatellite tandem repeats consisting of a stretch of three nucleotides.

Typically in individuals, the number of repeated units is relatively low. With each successive generation, there is a chance that the number of repeats will expand.

As this occurs, progeny can progress to premutation and ultimately affected status.

Individuals with a number of repeats that falls in the premutation range have a good chance of having affected children.

Those who progress to affected status will exhibit symptoms of their particular disease.

Prominent trinucleotide repeat disorders include Fragile X syndrome and Huntington's disease.

In the case of Fragile X syndrome it is thought that the symptoms result from the increased methylation and accompanying reduced expression of the fragile X mental retardation gene in individuals with a sufficient number of repeats.

16.5 – Study of mutant *Drosophila* through chart and photographs

Drosophila melanogaster is a species of fly (the taxonomic order Diptera) in the family Drosophilidae. The species is known generally as the **common fruit fly** or **vinegar fly**.

Starting with Charles W. Woodworth's proposal of the use of this species as a model organism, *D. melanogaster* continues to be widely used for biological research in studies of genetics, physiology, microbial pathogenesis, and life history evolution.

It is typically used because it is an animal species that is easy to care for, has four pairs of chromosomes, breeds quickly, and lays many eggs. *D. melanogaster* is a common pest in homes, restaurants, and other occupied places where food is served.

Flies belonging to the family Tephritidae are also called "fruit flies". This can cause confusion, especially in Australia and South Africa, where the Mediterranean fruit fly *Ceratitis capitata* is an economic pest.

Physical appearance:-

Wildtype fruit flies are yellow-brown, with brick-red eyes and transverse black rings across the abdomen.

They exhibit sexual dimorphism: females are about 2.5 millimeters (0.098 in) long; males are slightly smaller with darker backs.

Males are easily distinguished from females based on colour differences, with a distinct black patch at the abdomen, less noticeable in recently emerged flies, and the sex combs (a row of dark bristles on the tarsus of the first leg).

Furthermore, males have a cluster of spiky hairs (claspers) surrounding the reproducing parts used to attach to the female during mating.

Lifecycle and reproduction:-

The *D. melanogaster* lifespan is about 30 days at 29 °C (84 °F). It had been recorded that their lifespan can be increased to 3 months.

The developmental period for *D. melanogaster* varies with temperature, as with many ectothermic species.

The shortest development time (egg to adult), 7 days, is achieved at 28 °C (82 °F). Development times increase at higher temperatures (11 days at 30 °C or 86 °F) due to heat stress.

Under ideal conditions, the development time at 25 °C (77 °F) is 8.5 days, at 18 °C (64 °F) it takes 19 days and at 12 °C (54 °F) it takes over 50 days.

Under crowded conditions, development time increases, while the emerging flies are smaller.

Females lay some 400 eggs (embryos), about five at a time, into rotting fruit or other suitable material such as decaying mushrooms and sap fluxes. The eggs, which are about 0.5 mm long, hatch after 12–15 hours (at 25 °C or 77 °F).

The resulting larvae grow for about 4 days (at 25 °C) while molting twice (into second- and third-instar larvae), at about 24 and 48 h after hatching.

During this time, they feed on the microorganisms that decompose the fruit, as well as on the sugar of the fruit itself.

The mother puts feces on the egg sacs to establish the same microbial composition in the larvae's guts which has worked positively for herself.

Then the larvae encapsulate in the puparium and undergo a four-day-long metamorphosis (at 25 °C), after which the adults eclose (emerge).

Females become receptive to courting males at about 8–12 hours after emergence. Specific neuron groups in females have been found to affect copulation behavior and mate choice.

One such group in the abdominal nerve cord allows the female fly to pause her body movements to copulate.

Activation of these neurons induces the female to cease movement and orient herself towards the male to allow for mounting. If the group is inactivated, the female remains in motion and does not copulate. Various chemical signals such as male pheromones often are able to activate the group.

The female fruit fly prefers a shorter duration when it comes to sex. Males, on the other hand, prefer it to last longer.

Males perform a sequence of five behavioral patterns to court females. First, males orient themselves while playing a courtship song by horizontally extending and vibrating their wings.

Soon after, the male positions itself at the rear of the female's abdomen in a low posture to tap and lick the female genitalia.

Finally, the male curls its abdomen and attempts copulation. Females can reject males by moving away, kicking, and extruding their ovipositor. Copulation lasts around 15–20 minutes, during which males transfer a few hundred, very long (1.76 mm) sperm cells in seminal fluid to the female.

Females store the sperm in a tubular receptacle and in two mushroom-shaped spermathecae; sperm from multiple matings compete for fertilization. A last male precedence is believed to exist in which the last male to mate with a female sires about 80% of her offspring. This precedence was found to occur through both displacement and incapacitation.

The displacement is attributed to sperm handling by the female fly as multiple matings are conducted and is most significant during the first 1–2 days after copulation.

Displacement from the seminal receptacle is more significant than displacement from the spermathecae. Incapacitation of first male sperm by second male sperm becomes significant 2–7 days after copulation.

The seminal fluid of the second male is believed to be responsible for this incapacitation mechanism (without removal of first male sperm) which takes effect before fertilization occurs.

The delay in effectiveness of the incapacitation mechanism is believed to be a protective mechanism that prevents a male fly from incapacitating its own sperm should it mate with the same female fly repetitively.

Sensory neurons in the uterus of female *D. melanogaster* respond to a male protein, sex peptide, which is found in sperm. This protein makes the female reluctant to copulate for about 10 days after insemination.

The signal pathway leading to this change in behavior has been determined. The signal is sent to a brain region that is a homolog of the hypothalamus and the hypothalamus then controls sexual behavior and desire

D. melanogaster is often used for life extension studies, such as to identify genes purported to increase lifespan when mutated.

Genetic markers:-

Genetic markers are commonly used in *Drosophila* research, for example within balancer chromosomes or P-element inserts, and most phenotypes are easily identifiable either with the naked eye or under a microscope.

In the list of example common markers below, the allele symbol is followed by the name of the gene affected and a description of its phenotype. (Recessive alleles are in lower case, while dominant alleles are capitalized.)

- Cy¹: Curly; the wings curve away from the body, flight may be somewhat impaired.
- e¹: ebony; black body and wings (heterozygotes are also visibly darker than wild type).
- Sb¹: stubble; bristles are shorter and thicker than wild type.
- w¹: white; eyes lack pigmentation and appear white.
- y¹: yellow; body pigmentation and wings appear yellow. This is the fly analog of albinism.

Drosophila genes are traditionally named after the phenotype they cause when mutated. For example, the absence of a particular gene in *Drosophila* will result in a mutant embryo that does not develop a heart.

Scientists have thus called this gene *tinman*, named after the Oz character of the same name. This system of nomenclature results in a wider range of gene names than in other organisms.

Similarity to humans:-

Study by National Human Genome Research Institute comparing the fruit fly and human genome estimated that about 60% of genes are conserved between the two species.

About 75% of known human disease genes have a recognizable match in the genome of fruit flies, and 50% of fly protein sequences have mammalian homologs.

An online database called Haemophila is available to search for human disease gene homologues in flies and vice versa. *Drosophila* is being used as a genetic model for several human diseases including the neurodegenerative disorders Parkinson's, Huntington's, spinocerebellar ataxia and Alzheimer's disease.

The fly is also being used to study mechanisms underlying aging and oxidative stress, immunity, diabetes, and cancer, as well as drug abuse.

Sex determination:-

Drosophila flies have both X and Y chromosomes, as well as autosomes. Unlike humans, the Y chromosome does not confer maleness; rather, it encodes genes necessary for making sperm.

Sex is instead determined by the ratio of X chromosomes to autosomes. Furthermore, each cell "decides" whether to be male or female independently of the rest of the organism, resulting in the occasional occurrence of gynandromorphs

X Chromosomes Autosomes Ratio of X: A Sex:-

XXXX	AAAA	1	Normal Male
XXX	AAA	1	Normal Female
XXY	AA	1	Normal Female
XXYY	AA	1	Normal Female
XX	AA	1	Normal Female
XY	AA	0.50	Normal Male
X	AA	0.50	Normal Male (STERILE)
XXX	AA	1.50	Meta Female
XXXX	AAA	1.33	Meta Female
XX	AAA	0.66	Intersex
X	AAA	0.33	Meta Male

Three major genes are involved in determination of *Drosophila* sex. These are sex-lethal, sisterless, and deadpan.

Deadpan is an autosomal gene which inhibits sex-lethal, while sisterless is carried on the X chromosome and inhibits the action of *deadpan*.

An AAX cell has twice as much deadpan as sisterless, so sex-lethal will be inhibited, creating a male.

However, an AAXX cell will produce enough sisterless to inhibit the action of deadpan, allowing the sex-lethal gene to be transcribed to create a female.

Later, control by deadpan and sister less disappears and what becomes important is the form of the sex-lethal gene.

A secondary promoter causes transcription in both males and females. Analysis of the cDNA has shown that different forms are expressed in males and females.

Sex-lethal has been shown to affect the splicing of its own mRNA.

In males, the third exon is included which encodes a stop codon, causing a truncated form to be produced.

In the female version, the presence of sex-lethal causes this exon to be missed out; the other seven amino acids are produced as a full peptide chain, again giving a difference between males and females.

Presence or absence of functional sex-lethal proteins now goes on to affect the transcription of another protein known as double sex.

In the absence of sex-lethal, double sex will have the fourth exon removed and be translated up to and including exon 6 (DSX-M[ale]), while in its presence the fourth exon which encodes a stop codon will produce a truncated version of the protein (DSX-Female]).

DSX-F causes transcription of Yolk proteins 1 and 2 in somatic cells, which will be pumped into the oocyte on its production.

STUDY OF MUTANTS OF DROSOPHILA:-

Introduction:-

The fruit flies in this exhibit show just a few of the mutations that occur in natural fruit fly populations.

The genetic instructions to build a fruit fly-or any other organism-are imprinted in its DNA, a long, threadlike molecule packaged in bundles called chromosomes.

Like a phone book made up of different names and addresses, each chromosome consists of many individual sections called genes.

Each gene carries some of the instructions for building one particular characteristic of an organism.

Structure:-

To build a complete organism, many genes must work precisely together. A defect in a gene can cause a change in the building plan for one particular body part-or for the entire organism.

Mutations are neither good nor bad: some may be beneficial for an organism; others may be lethal.

By creating new gene versions, mutations are a driving force for changes in evolution, sometimes leading to new species.

Biologists learn about the proper function of any gene by studying mutations. If a defective gene causes short wings, for instance, scientists know that the healthy version of the gene is responsible for correct wing formation.

Altered wings Structure:-

Normal Fruits Flies: These are normal fruit flies, or "wildtypes." Notice the shape and length of their wings. Now compare them with the other fruit flies here.

Short-Winged Flies: Notice the shortened wings of these flies. Flies with vestigial wings cannot fly: they have a defect in their "vestigial gene," on the second chromosome. These flies have a recessive mutation. Of the pair of vestigial genes carried by each fly (one from each parent), both have to be altered to produce the abnormal wing shape.

If only one is mutated, the healthy version can override the defect

Curly-Winged Flies:-

Notice the curled wings of these flies. They have a defect in their "curly gene," which is on the second chromosome.

Having curled wings is a dominant mutation, which means that only one copy of the gene has to be altered to produce the defect. In fact, if both copies are mutated, the flies do not survive.

Normal Fruit Flies:-

These normal fruit flies, or "wild types," have black-and-tan striped bodies. Compare them with the other fruit flies here.

Yellow Flies:-

Notice that these flies are yellower than normal flies. They have a defect in their "yellow gene," which is on the X chromosome.

Since the yellow gene is needed for producing a fly's normal black pigment, yellow mutant flies cannot produce this pigment.

Ebony Flies:-

Notice that these flies have a dark, almost black, body. They carry a defect in their "ebony gene," on the third chromosome.

Normally, the ebony gene is responsible for building up the tan-colored pigments in the normal fruit fly. If the ebony gene is defective, the black pigments accumulate all over the body.

Animal Models for the Study of Learning and Memory:-

Four animal models have proved particularly useful. The first is perhaps the most surprising: the fruit fly *Drosophila*, long the subject of study by geneticists for the speed with which it breeds and the ease with which mutants could be generated.

Fruit flies will spontaneously fly towards particular odors, but if they receive an electric shock as they approach they can learn to avoid that particular odor.

A series of mutants have been generated that either could not learn to respond by avoidance, or forgot after varying periods of time.

Each class of mutants had a specific biochemical abnormality—the loss of activity of a particular enzyme or of one of the factors required for the synthesis of specific proteins.

Hence it is argued that the missing enzyme or factor is also necessary for learning and memory to occur.

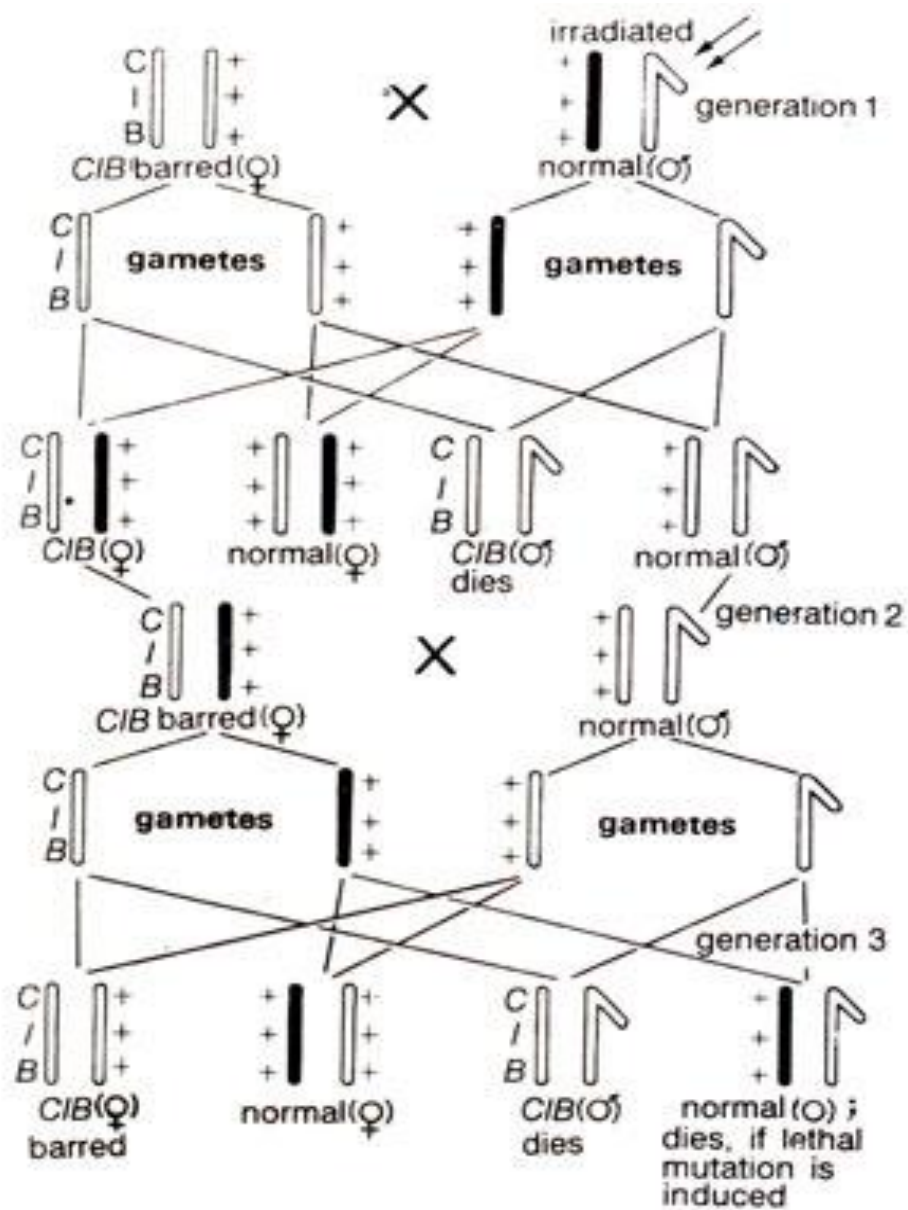


Fig.15.2 Muller's CIB method for detection of sex linked lethal mutations in *Drosophila*.

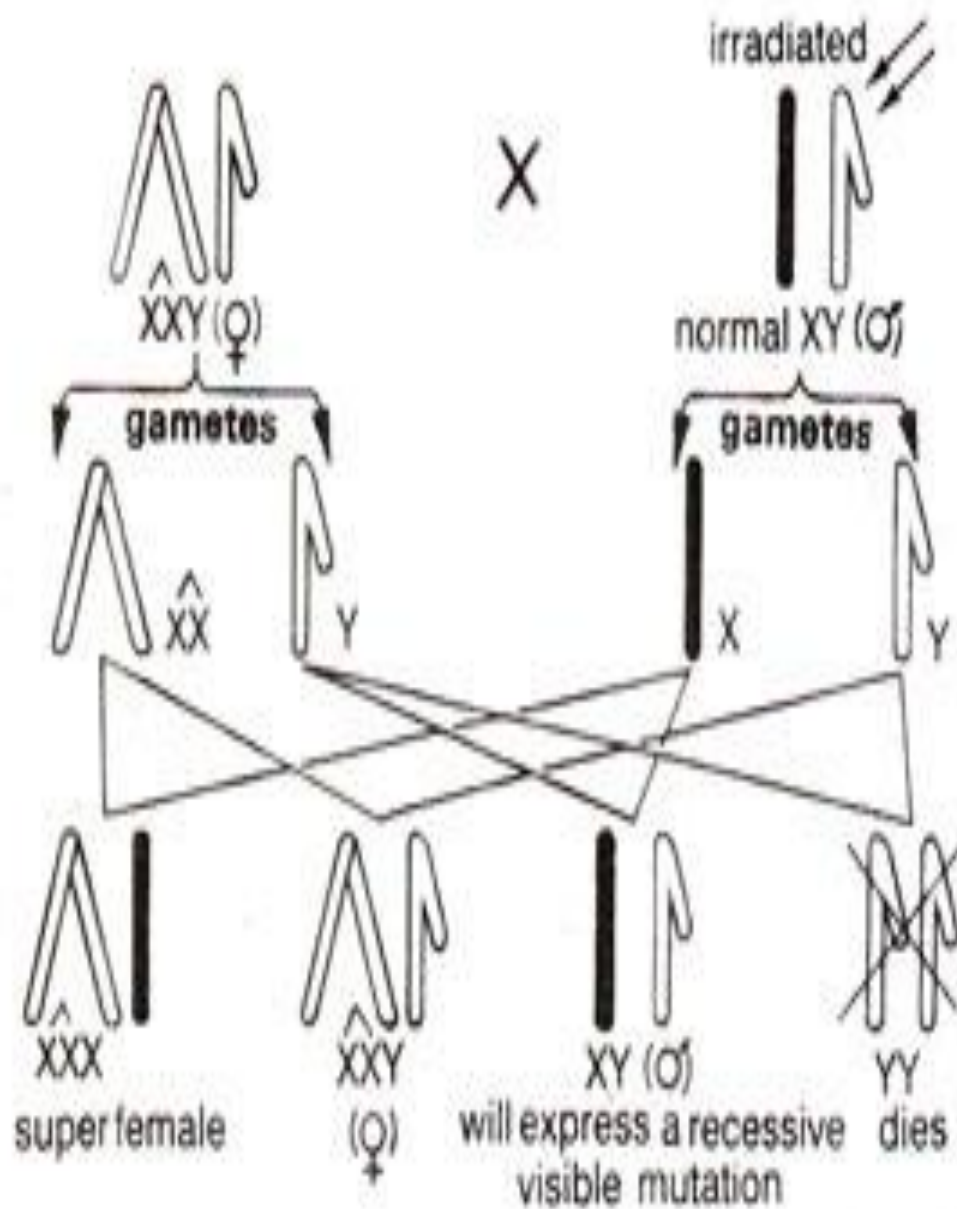


Fig.16.3 Attached X-method for detection of sex linked visible mutations.

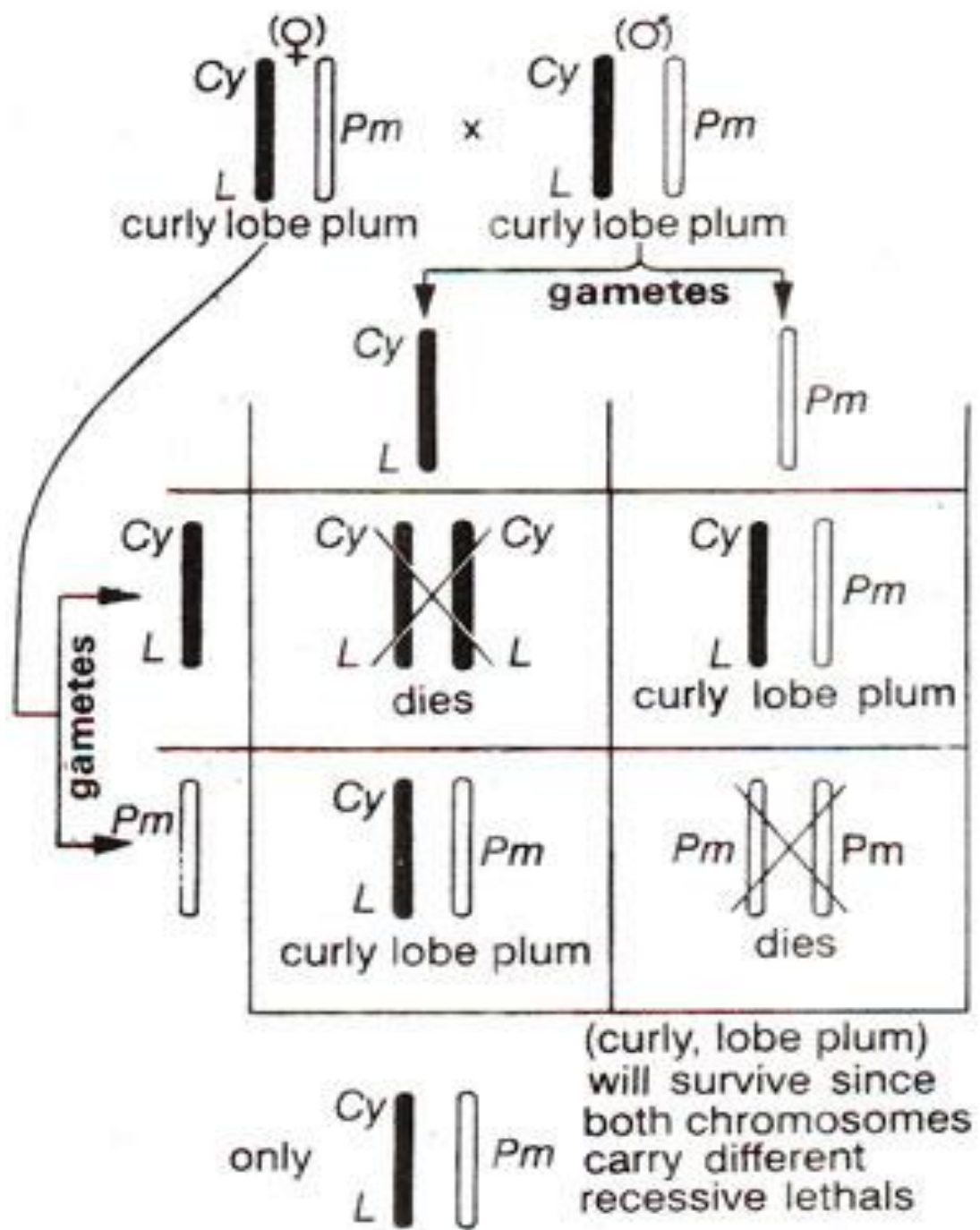


Fig.16.4 Balanced lethal system in *Cy* *L*/*Pm* *Drosophila*

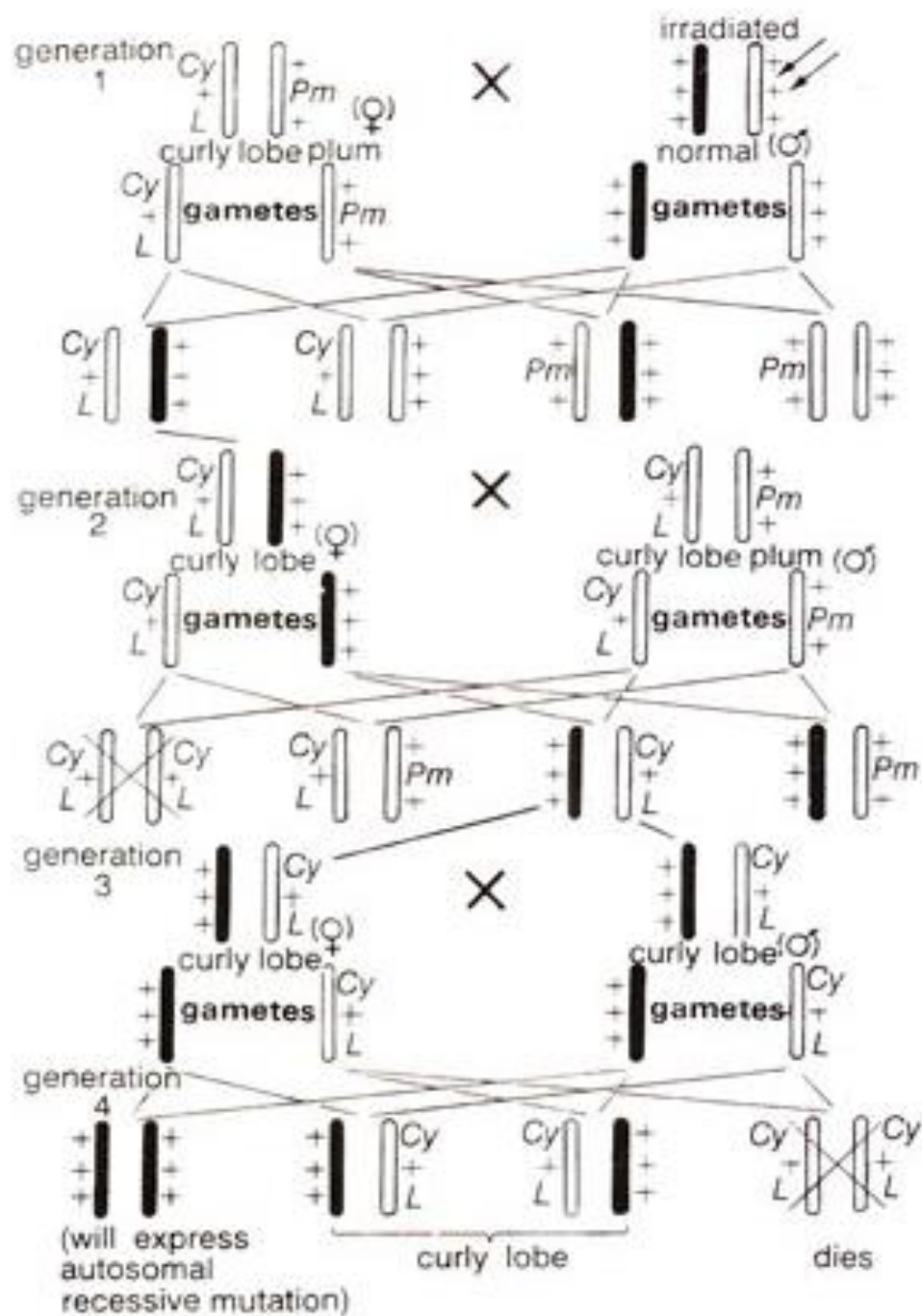


Fig.16.5 Detection of autosomal mutations in *Drosophila*

16.6- GENETICAL EXERCISE

Exercise No 1: -

Objective:

Two grey *Drosophila* flies on breeding produce 152 grey and 49 black off-springs. Give the genotypes of the parents and justify your answer giving reasons. (Given grey is dominant over black).

Observation:

The ratio of the dominant and recessive off-springs in the given problem 152:49 comes to be approximately 3:1.

Hence, the parents of these off-springs will be heterozygous for grey and black traits having genotypes Gg and Gg.

Explanation:-

In case of a monohybrid cross when heterozygous individuals are crossed among themselves, they produce off-springs with dominant and recessive characters in a ratio of 3:1.

Grey	*	Grey
Gg	↓	Gg
↓	↓	↓
Gametes		Gametes
	G	g
Gametes		
G	Gg	Gg
	Grey	Grey
g	gg	gg
	Black	Black

Conclusions: -

The genotypes of the parents will be Gg and Gg. It is a case of simple Mendelian Inheritance showing the phenomenon of dominance and recessive.

Exercise No2: -

Objective: -

Both the parents of a blue-eyed child are brown-eyed. Find out the genotypes of the parents if brown eyes (B) are dominant over blue (b) which is recessive.

Observation: -Since recessive character is only expressed in homozygous condition, hence, the genotype of the child will be **bb**.

The child receives characters from both the parents in equal amount. Therefore, its one **b** has come from one parent and the other **b** from other parents causing his eyes blue.

But in the given problem both the parents are brown-eyed. Therefore, they must have been heterozygous with genotype **Bb** and **Bb**.

Explanation: -

When heterozygous brown-eyed parents cross, they produce brown-eyed and blue-eyed off-springs in a ratio of **3:1**.

The brown-eyed off-springs having genotype **BB** and **Bb**, while blue-eyed with **bb**

Brown Eyed father	*	Brown Eyed father
Bb	↓	Bb
↓	↓	↓
Gametes		Gametes
B		b
Gametes	B	B
B	Gg	Gg
	Grey	Grey
B	gg	gg
	Black	Black

Conclusion:

The genotype of the parents of a blue-eyed child will be **Bb**. It is a case of simple Mendelian Monohybrid Inheritance showing the phenomena of **dominance**, **recessive** and **segregation**.

Exercise No 3:-

Objective:

In garden pea, tall (**T**) is dominant over dwarf (**t**) and red flower colour (**R**) to white (**r**) If pure red tall is crossed with a dwarf white, what will be

- (i) P1 genotypes,
- (ii) the gametes of **P1**,
- (iii) the F1 phenotype and genotype,
- (iv) the gametes of **F1**, and (**V**) **F2** phenotypic ratio.

Point out the genetic principles involved.

Observation:

It appears from the object that it is a case of di-hybrid cross and based on the cross as suggested of Mendelian Inheritance.

Explanation:-

To find out the solution of the questions asked in the object, work out the cross as suggested as pure red tall means homozygous tall plants with red flowers, genotype will be **TTRR**.

Dwarf white means homozygous dwarf plants with white flowers, genotype will be **ttrr**.

It will be homozygous because recessive are expressed only when they are in homozygous condition.

	Phenotype	Tall red	×	Dwarf white
(P1)	Genotype	TT RR	↓	tt rr
	Gametes	TR, TR	↓	tr, tr
(F1)	Genotype	TR tr	↓	TR tr
	Phenotype	Tall red	↓	Tall red
	Gametes	TR, Tr, tR, tr	×	TR, Tr, tR, tr
			↓	

♂ \ ♀	TR	Tr	tR	tr
TR	TRTR Tall Red	TRTr Tall red	TRtR Tall red	TRtr Tall red
Tr	TrTR Tall red	TrTr Tall white	TrtR Tall red	Trtr Tall white
tR	tRTR Tall red	tRTr Tall red	tRtR Dwarf red	tRtr Dwarf red
tr	trTR Tall red	trTr Tall white	trtR Dwarf red	trtr Dwarf white

Phenotypic ratio: 9 Tall red: 3 Tall white: 3 Dwarf red: 1 Dwarf white

Conclusion: (i) P1 genotype are TTRR and ttrr

(ii) The gametes of P1 are TR and tr types

(iii) F1 phenotype is Tall red and genotype is TRtr for all possible offsprings

(iv) The gametes of F1 are TR, Tr and tr, and

(v) The F2 phenotype ratio is 9:3:3:1

The genetic principles involved are the phenomena of dominance and recessive, law of segregation and the Law of Independent Assortment.

Based on Incomplete Dominance:-

Objective:

Explain giving reasons for the occurrence of modified **3:1** phenotypic ratio **1:2:1** in a monohybrid cross.

Observation:

According to the phenomenon of dominance, a monohybrid cross should always result into **F2** off-springs in a phenotypic ratio of **3:1** and their genotypic ratio being **1:2:1**. But sometimes, due to incomplete

dominance, **3:1** phenotypic ratio comes to be **1:2:1**. It means that the phenomenon of dominance shows exceptions.

Explanation:

Let us consider an example to illustrate the modified **1:2:1** phenotypic ratio.

An Andalusian fowl a cross between its homozygous black and white varieties results in blue hybrids.

These blue hybrids when crossed among themselves, the **F₂** generation off-springs are **1 black : 2 blue : 1 white**.

The colour of **F₁** hybrid blue is a blended character, i.e., a colour midway between the two parents.

It means that none of the colour of the parents are either fully dominant or recessive.

Therefore, both the factor expresses them partially resulting into a blending of parental characters.

Such factors are usually referred to as intermediate factors or genes.

♂	♀	B	B
		Bb	Bb
B		Blue	Blue
b		Bb	Bb
		Blue	Blue

Blue Fowl	*	White Fowl
Bb	↓	bb
↓	↓	↓
All Blue FowlsF1		
↓		
Cross- fertilization		

♂	B	b
♀	B	b
B	BB Black	Bb Blue
b	Bb Blue	bb White

Phenotypic ratio = 1 Black: 2 Blue: 1 White

Conclusions: The modified phenotypic **1:2:1** ratio is due to the phenomenon of incomplete dominance. As referred to, it is due to intermediate genes, hence, called intermediate inheritance.

16.6: SUMMERY:-

Genetics, scientific study of how physical, biochemical, and behavioral traits are transmitted from parents to their offspring.

Geneticists determine the mechanisms of inheritance whereby the offspring of sexually reproducing organisms do not exactly resemble their parents, and the differences and similarities between parents and offspring recur from generation to generation in repeated patterns.

The investigation of these patterns has led to some of the most exciting discoveries in modern biology.

Most physical characteristics of humans are influenced by multiple genetic variables as well as by the environment.

Some characteristics, such as height, have a relatively large genetic component. Others, such as body weight, have a relatively large environmental component.

Still other characteristics, such as the blood groups and the antigens involved in the rejection of transplanted organs, appear to involve entirely genetic components; no environmental condition is known to change these characteristics.

The transplantation antigens have recently been extensively studied because of their medical interest. The most important ones are produced by a group of linked genes known as the HLA complex.

This group of genes not only determines whether transplanted organs will be accepted or rejected, it is also involved in the body's resistance to various diseases (including allergies, diabetes, and arthritis).

The identification and study of genes are of great interest to biologists, and are also of medical importance when a particular gene is involved in disease.

The human genome contains approximately 30,000 genes, of which about 4,000 may be associated with disease.

A globally coordinated effort, called the Human Genome Project, was started in 1990 to characterize the entire human genome.

By 2003 a complete sequence map of the human genome had been produced. Its primary goals had been to determine the complete sequence of the 3 billion DNA subunits (bases), generate various genome maps, including the entire nucleotide sequence of the human genome, and identify all human genes, making them accessible for further biological study.

The project was greatly assisted by the ability to clone large fragments of DNA into yeast artificial chromosome vectors for further analysis, the automation of many techniques such as DNA sequencing, and the use of supercomputers in whole-genome shotgun sequencing, for example.

16.7: GLOSSARY:-

Alleles	A pair of gene located at corresponding position on a pair of homologous chromosomes.
Aneuploidy	The action or loss of individual chromosomes leading to abnormal chromosomes constitution
Anticodon	A sequence of three nucleotides on one of the loops of tRNA which is complementary to a codon of mRNA
Bacteriophage	Virus which parasitizes bacteria
Cistron	The smallest unit of DNA that codes for one polypeptide chain. Is synonymous with structural gene
Codon	A sequence of three nucleotides on the mRNA which codes for a particular amino acid
Down's syndrome	Mutant condition resulting from chromosomal trisomy or translocation, characterized by severe mental retardation, also known in the past as mongoloid idiocy
Euchromatin	Lightly stained portions of chromosomes
Genetic Drift	A random change in gene frequency from one generation to another in a population
Genome	The total genetic constitution of an organism
Haploid	The number of chromosomes found in mature gametes of sexually reproducing organism; one half of the species number of chromosomes; referred to as the n-number of chromosomes
Induced mutation	Mutation as a result of manmade factors
Karyotype	A composite picture of an individual's chromosomes, made by taking a photomicrograph of specially prepared cells and then cutting out the chromosomes and matching them
Monosomy	Presence of a single copy of a chromosome in cells which are genetically diploid organism
Muton	The small genetic unit that can mutate

Operator gene	In bacteria, the gene that controls transcription of structural gene
Pleiotrophy	The occurrence of a syndrome of diverse effects, resulting from the mutation of a single genetically determined
Polygeny	The determination of a trait by several pairs of genes with additive effects on the phenotype
Polyploidy	Presence of extra sets of chromosomes
Polysome	Cluster of ribosome formed during protein synthesis
Regulator gene	In bacteria, the gene which produces a repressor substance that binds to the operator gene
Somatic	Pertaining to body cells other than gametes. A somatic mutation is one occurring in a body cell rather than a sex cell
Structural gene	In bacteria, the gene that contains the coding for a specific polypeptide.
Tetrad	A cluster of four chromatids formed by duplicated homologous lying parallel to one another following synapses.
Tolerance	An immunological phenomenon in which an organism does not react to the presence of antigen.
Transcription	The synthesis of new RNA molecules using DNA as a template
Trisomy	Presence of three copies of a chromosome in cells of a normally diploid organism
X-linkage	Linkage of genes on the X-chromosome

16.8: SELF ASSESSMENT QUESTIONS:-

1. Which plant was used by Mendel in his experiment?
 2. Who proposed the Law of Dominance?
 3. Who proposed the hypothesis of one gene one enzyme?
 4. Give an example of Incomplete Dominance.
 5. What is the ration of Dihybrid cross in F₂ generation?
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16.9: REFERENCES:-

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16.10: TERMINAL QUESTIONS:-

1. What do you understand by spontaneous mutations and induced mutations? Discuss variation in mutation rates and frequencies at different loci within an organism.
 2. Discuss Mendels laws of inheritance. Which law do you think is most important and why?
 3. Discuss the pre- Mendelian ideas about the mechanism of inheritance of traits. Why workers before Mendel did not succeed in formulating laws of inheritance?
 4. Discuss sex determination with the help of suitable examples.
 5. Write short notes any two of the following-
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- (a) Dihybrid crosses
- (b) Allele or allelomorph
- © Pure line