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TOUCH n PASS

सर्वश्रेष्ठ परीक्षा पथ-प्रदर्शक

B.Sc. First Year (Certificate Course) Semester-I

ZOOLOGY

Cytology, Genetics and Infectious Diseases

SYLLABUS

UNIT-I **Structure and Function of Cell Organelles I in Animal cell :** Plasma membrane : chemical structure—lipids and proteins, Endomembrane system : protein targeting and sorting, transport, endocytosis, exocytosis

Introduction to all national and international Biologists (Zoologists) who have contributed/contributing to Zoological and Life Sciences as a mark of tribute to ancient and modern biology will be included as part of the Continuous Internal Evaluation (CIE)

Susruta, Charak, Patanjali, Varahamihira, Prof. H.G. Khorana, Prof. G.N. Ramachandran, Prof. Salim Ali, Prof. J.P. Thaplliyal, Prof. Lalji Singh, Prof. M.K. Chandrashekharan, Prof. R. Mishra-to be discussed with the topics being dealt.

UNIT-II **Structure and Function of Cell Organelles II in Animal cell :** Cytoskeleton : microtubules, microfilaments, intermediate filaments, Mitochondria : Structure, oxidative phosphorylation; electron transport system, Peroxisome and ribosome : structure and function

UNIT-III **Nucleus and Chromatin Structure :** Structure and function of nucleus in eukaryotes, Chemical structure and base composition of DNA and RNA, DNA supercoiling, chromatin organization, structure of chromosomes, Types of DNA and RNA

UNIT-IV **Cell cycle, Cell Division and Cell Signaling :** Cell division : mitosis and meiosis, Introduction to Cell cycle and its regulation, apoptosis. Signal transduction : intracellular signaling and cell surface receptors, via G-protein linked receptors; Cell-cell interaction : cell adhesion molecules, cellular junctions

UNIT-V **Mendelism and Sex Determination :** Basic principles of heredity : Mendel's laws, monohybrid and dihybrid crosses, Complete and Incomplete Dominance, Clinical expressions : Penetrance and expressivity, Genic Sex-Determining Systems, Environmental Sex Determination, Sex Determination with example of Drosophila, Sex-linked characteristics and Dosage compensation

UNIT-VI **Extensions of Mendelism, Genes and Environment :** Extensions of Mendelism : Multiple Alleles, Gene Interaction, The Interaction Between Sex and Heredity : Sex-Influenced and Sex-Limited Characteristics, Cytoplasmic Inheritance, Genetic Maternal Effects, Interaction Between Genes and Environment : Environmental Effects on Gene Expression, Inheritance of Continuous Characteristics

UNIT-VII **Human Chromosomes and Patterns of Inheritance :** Human karyotype, Chromosomal anomalies : Structural and numerical aberrations with examples, Pedigree analysis, Patterns of inheritance : autosomal dominant, autosomal recessive, X-linked recessive, X-linked dominant

UNIT-VIII **Infectious Diseases :** Introduction to pathogenic organisms : viruses, bacteria, fungi, protozoa, and worms; Structure, life cycle, pathogenicity, including diseases, causes, symptoms and control of common parasites : Trypanosoma, Giardia and Wuchereria

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Registered Office
Vidya Lok, Baghpat Road, T.P. Nagar,
Meerut, Uttar Pradesh (NCR) 250 002
Phone : 0121-2513177, 2513277
www.vidyauniversitypress.com

Editing & Writing
Research and Development Cell

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UNIT-I

Structure and Function of Cell Organelles-I in Animal Cell

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Who discovered the cell?

Ans. The cell was discovered by Robert Hooke.

Q.2. Who proposed the cell theory?

Ans. Cell theory was proposed by M.J. Schleiden and Theodore Schwann.

Q.3. Who gave the unit membrane concept?

Ans. Robertson.

Q.4. Define plasmodesmata.

Ans. **Plasmodesmata** are connections between two cell walls that are interrupted by small pores having fine threads of cytoplasm.

Q.5. Write a note on the structure of spectrin.

Ans. Spectrin is a cytoskeletal protein that lines the intracellular side of the plasma membrane in eukaryotic cells. Spectrin forms pentagonal or hexagonal arrangements, forming a scaffold and playing an important role in maintenance of plasma membrane integrity and cytoskeletal structure.

Q.6. Write about structure and function of glycocalyx.

Ans. Glycocalyx is a highly charged layer of membrane bound biological macromolecules attached to a cell membrane. This layer functions as a barrier between a cell and its surrounding. Most glycocalyx associated proteins are transmembrane that can be linked to the cytoskeletion.

Q.7. What do you understand by membrane skeleton?

Ans. The membrane skeleton is a specialized part of the cytoskeleton that is in close proximity to the cell membrane with a protein composition and structure that differ from that of the bulk cytoskeleton.

Q.8. What is the function of cell membrane in human RBCs?

Ans. Far from being just a casing, homever, the red cell membrane plays a critical rote in maintaining cellular functions in the only non-nucleated cell in the body (the cellular organelles are lost during red cell maturation). Thus, the red cell membrane accomplishes vital tasks in the transport of salts and nutrients.

Q.9. Write the concept of unit membrane in brief.

Ans. The unit membrane concept that says that all membranes have an underlying bilayer composed of phospholipids was originally proposed by Denielli and Davson in 1935. This concept extends to amide variety of membranes from different organisms, each of which has a specific function.

Q.10. Give a difference between animal cell and plant cell juctions.

Ans. There are some differences in the ways that plant and animal cells do this plasmodesmata are junctions between plant cells, whereas animal cell contacts include tight junctions, gap junctions, and desmosomes.

Q.11. Write a difference between gap and tight juction.

Ans. A gap junction refers to a linkage of two adjacent cells consisting of a system of channels extending across a gap from one cell to the other allowing the passage while tight junction refers to a specialized connection of two adjacent animal cell membranes, such that, space usually lysing between them is absent.

Q.12. Define plasmodesmata.

Ans. **Plasmodesmata** are intercellular organelles found only in plant and algal cells. The plasmodesmata consist of pores or channels, lying between individual plant cells, and connect the symplastic space in the plant.

Q.13. What is the role of ER in protein synthesis?

Ans. The ER is the port of entry of the protein secretory pathway. The ER is the compartment where newly synthesised polypeptides fold, where many multimeric proteins assemble and where glycoproteins acquire their asparagine linked glycans.

Q.14. What are the main functions of endoplasmic reticulum?

Ans. The endoplasmic reticulum (ER) serves important functions particularly in the synthesis, folding, modification and transport of proteins.

Q.15. How does rough ER differ from smoth ER?

Ans. Smooth ER is derived from rough endoplasmic reticulum by sacrificing the ribosome, whereas rough ER originates from the nuclear membrane.

The main difference between the SER and RER is the presence of ribosomes, as SER does not consist of ribosomes but RER consist of ribosomes.

Q.16. What is the most important scientific contribution of Sushruta?

Ans. Sushruta's great contribution was in the fields of Rhinoplasty (plastic surgery) and ophthalmic surgery (removal of cataracts).

Q.17. Who is called the father of 'Green Revolution' in India?

Ans. **Mankombu Sambasivan Swaminathan** (born 7 August, 1925) is an Indian geneticist and administrator, known for his role in India's Green Revolution, so that he is also known as the "Father of Green Revolution in India".

Swaminathan developed high-yielding varieties (HYN) of wheat and later, promoted sustainable developed which he called, the 'evergreen revolution'.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Write a short note on the concept of cell theory and its extension.

Ans. **Cell** is the fundamental unit of structure and function is all living beings and is capable of independent existence of performing all life activities.

Cell theory

The **Cell theory** or cell docrine was jointly proposed by **German botanist, M.J. Schleiden** for plants and by German anatomist, **Theodor Schwann** (1839) for animals. It states that the bodies of all organisms are made up of cells and their products. Therefore, cells are the units of both structure and function of living organisms.

Extension of Cell Theory

Schleiden, in 1838 concluded that cells are the ultimate units forming the structures of all plant tissues. At the same time, **Schwann** in 1839 proposed that bodies of animals are composed of cells and their products. He defined the cell as a membrane-bound, nucleus-containing structure.

In 1855, **Rudolf Virchow** osberved that new cells develop by the division of pre-existing cells. This led to the development of **theory of cell lineage** which suggests that all living beings have evolved from the same common ancestor and that same cell line is continuing till date from the beginning of life. Soon thereafter, in 1866, **Haeckel** established that nucleus stores and transmits hereditary traits.

Fundamental Features of Cell Theory

The cell theory is one of the **fundamental generalisation of biology.** It states that :

1. All living organisms are made up of minute cells and their products; i.e., Cells are the funadmental units of structure and function in all living organisms.
2. Cells are the smallest entities that can perform all life activities: Cells are physiological units of all living organisms.
3. Cells are hereditary units (these maintain continuity through the hereditary material.
4. New cells originate from the pre-existing cells only : ***Omnis Cellula-a-cellulae.***
5. Cell is the smallest unit of life. All activities of living organisms are the outcome of the activities and interactions of its constituent cells.

Significances of Cell Theory

Modern concept of cell theory emphasizes the structural and functional relationship among the diverse living forms from bacteria to man.

All cells irrespective of their function and position have a nucleus embedded in the cytoplasm and are bounded by cell membrane **(unity in structural plan)** and the same metabolic processes occur in all the cells primitive or specialized **(unity of function).**

This implies that all the living things have originated from the same primitive ancestral type that originated about two billion year ago.

Q.2. What are the exceptions of cell theory?

Ans.

Exceptions to Cell Theory

The cell theory is one of the fundamental generalization of biology and has universal application except for the following exceptions :

1. **Protozoans** are not cellular. They are **acellular,** *i.e.,* their body is not divisible into cells.
2. **Viruses** are an exception to cell theory. They are made up of proteins and one of the nucleic acids, *i.e.,* DNA or RNA. They lack protoplasm, the essential part of the cell.
3. **Bacteria** and **blue green algae** (prokaryotes) lack well-organised nucleus. Nuclear membrane, nucleolus and nucleoplasm are absent. The nucleic acid (DNA) alone forms the chromosome and lies in direct contact with cytoplasm. Basic proteins associated with nucleic acid are absent in bacteria.
4. The protoplasm of cells of skin surface is replaced by nonliving keratin during maturation.
5. **Coenocytic hyphae** of *Rhizopus* (a fungus), and cells of *Vaucheria* (an alga) are multinucleate.
6. Certain cells like RBCs are enucleate. They lose their nuclei during maturation but have a life span of 120 days.

Q.3. Describe the structure of bacteriorhodopsin in brief.

Ans.

Structure of Bacteriorhodospin

A Seven Helices Transmembrane Segments (TMS) Protein, in a Proton Pump : *Halobacterium halobium* represents a class of bacteria, which live in salt-water pools exposed to large amount of sunlight. They have evolved a variety of light activated proteins including bacteriorhodospin, which is a light activated proton pump. **Bacteriorhodopsin** molecule

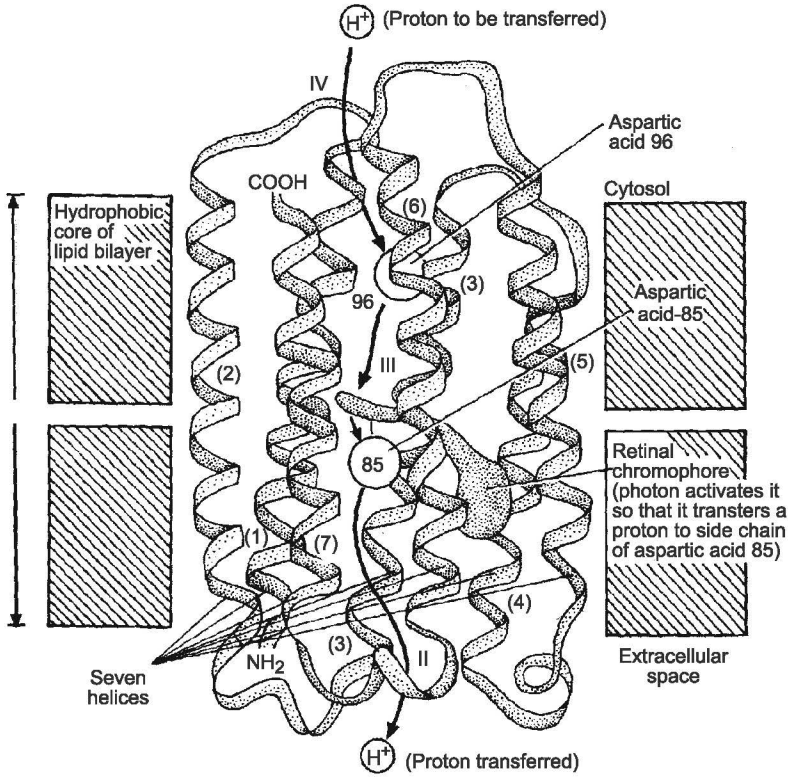


Fig. : Three-dimensional structure of a bacteriorhodopsin molecule-note the presence of seven transmembrane alpha-helices (1-7) and the path of protons during light activation.

contains a light absorbing group or **chromophore** (called **retinal**), which gives the protein its purple colour. In bright light, several hundred protons can be pumped every second by one bacteriorhodopsin molecule. This establishes a proton gradient across the membrane facilitating the production of ATP by another protein, thus providing energy to the bacterial cell.

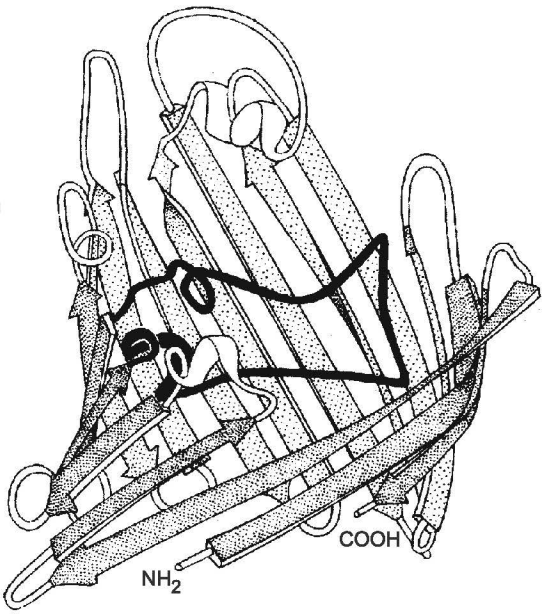
Bacteriorhodopsin is a multipass transmembrane protein having **seven transmembrane α-helices,** which are closely packed, each helix containing about 25 amino acids. The light receptor rhodopsin in vertebrates and many cell surface receptor proteins resemble bacteriorhodopsin, in having seven transmembrane α helices and therefore sometimes described as **seven sisters proteins.**

Q.4. Write a short note on the structure of porins.

Ans. **Structure of Porins (Transmembrane Proteins)**

Although generally, in the transmembrane proteins, the segments traversing the membrane are α-helices, they can also be present as β-**sheets** (β-barrel). Porins represent one such protein found in the outer membrane (which surrounds plasma membrane) of many bacteria like *E. coli*, and allow selected hydrophilic solutes (upto 600 daltons) to pass across this outer lipid bilayer.

Proteins in the outer membrane of mitochondria and chloroplasts also resemble porins in having β-sheet instead of α-helix, as their transmembrane segments. In 1990, three dimensional structure of a porin from ***Rhodobacter capsulatus*** was determined by X-ray crystallography. It is a trimer, each monomer forming a β-barrel, and forms a pore while traversing the lipid bilayer. The β-barrel is formed from a **16-stranded antiparallel β-sheet,** curved to form a cylindrical structure. Polar side chains line the aqueous channel and the non-polar side chains project outside to interact with the surface of the hydrophobic lipid bilayer.



a porin protein consisting of 16 stranded antiparallel transmembrane β barrel

Fig. : Three dimensional structure of a porin trimer of *Rhodobacter capsulatus*

Q.5. Describe the structure of liposomes in brief.

Ans. **Liposomes**

Liposomes were first described by British haematologist, **Alec D. Bangham,** in 1961 while studying lipids under electron microscope. The term was derived from two Greek words : ***Lipos*** = fat + ***soma*** = body and indicates their lipid nature.

Liposomes are spherical, microscopic fluid-filled pouches having aqueous interior. Their walls are made of phospholipid bilayer identical to the phospholipid bilayer of cell membrane with hydrophilic head ends of phospholipid molecules in contact with aqueous medium and hydrophobic tails directed in the middle of bilayer.

Types of Liposomes

Three types of liposomes can be artifically prepared *in vitro* :

- Multilamellar vesicles (MLV)
- Small unilamellar vesicles (SUV)
- Large unilamellar vesicles (LUV)

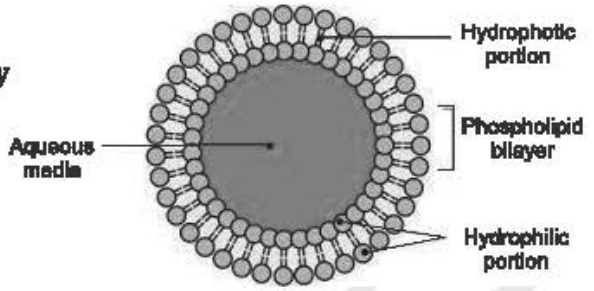


Fig. : A liposome

Application of Liposomes

1. Liposomes are used to deliver certain vaccines, enzymes or drugs like insulin or cancer drugs. There are following methods of drug delivery to target cells by liposomes.
   (i) by direct fusion of liposomes with the lipid bilayer of cell membrane.
   (ii) by diffusion from the liposomal interior into the cell.
   (iii) by endocytosis of liposomes by macrophages so that these from phagosomes and release contents inside macrophages.
   (iv) by ligand endocytosis.
2. Anticancer drugs such as Doxil, Daunoxome Camptothecin are currently being marketed in liposomes to ascertain targeted drug delivery to the cancer cells only. Thus healthy body cells are protected from drug's toxicity and common side effects like nausea and hair loss are eliminated or much reduced.
3. Liposomes are used in cosmetic industry for local and target delivery of specific chemical cosmetics and for long lasting effect of the ingredients due to slow release from liposomes. Moreover, liposomes have good moisturing quality.
4. Liposomes are used in gene therapy to deliver normal gene in the concerned cell and replace the defective disease causing gene.

Q.6. Explain differences between extrinsic and intrinsic proteins.

Ans.

Difference between Extrinsic and Intrinsic Proteins

S.No.	Intrinsic Proteins	Extrinsic Proteins
1.	They are embedded in the plasma membrane partially or completely.	They are present on the surface of the plasma membrane.
2.	They constitute 70% of the total membrane proteins.	They constitute 30% of the total membrane proteins.
3.	They are more hydrophobic and less hydrophilic.	They are more hydrophilic and less hydrophobic.
4.	They function as carrier proteins, enzymes, permeases. **Examples :** Glycophorin, Rhodospin.	They function as receptors, antigens. **Example :** Erythrocyte, spectrin.

Q.7. Explain differences between endocytosis and exocytosis.

Ans.

Endocytosis and Exocytosis

Some molecules or particles are just too large to pass through the plasma membrane or to move through a transport protein. Therefore, cells use two other processes to move these macromolecules into or out of the cell. There two processes are called (i) **endocytosis** and (ii) **excytosis.** Both processes are **active transport** processes, requiring energy.

Differences between Endocytosis and Exocytosis

Endocytosis is the process of capturing a substance or particle from outside the cell by engulfing it with the cell membrane. The membrane folds over the substance and it becomes completely enclosed by the membrane. At this point a membrane-bound sac, or vesicle pinches off and moves the substance into the cytosol. There are two main kinds of endocytosis : **(i) Phagocytosis** *(cellular eating)* occurs when the dissolved materials enter the cell. The plasma membrane engulfs the solid material, forming a phagocytic vesicle. **(ii) Pinocytosis** (*cellular drinking*) occurs when the plasma membrane folds inward to form a channel allowing dissolved substances to enter the cell. When the channel is closed, the liquid is encircled within a pinocytic vesicle.

Exocytosis is the process, where vesicles fuse with the plasma membrane and release their contents to the outside of the cell. Exocytosis occurs when a cell produces substances for export, such as protein, or when the cell is getting rid of a waste product or a toxin. Newly made membrane proteins and membrane lipids are also moved on top the plasma membrane by exocytosis.

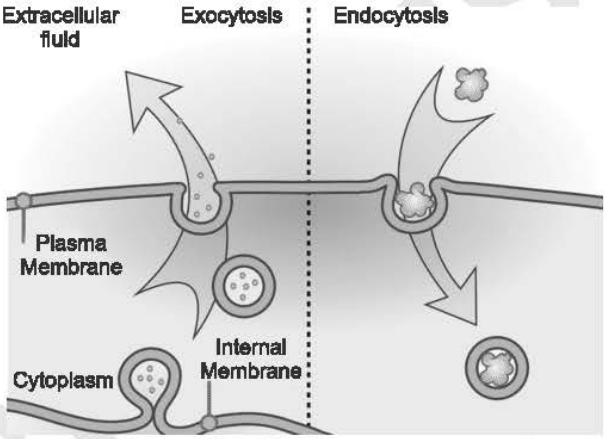


Fig. : Two types of vesicle transport : exocytosis and endocytosis.

Q.8. Write a short note on the Passive transport of ions by diffusion.

Ans.

Passive Transport of Ions by Diffusion

According to **Donnan** (1911), if a cell having non-diffusible negative charge inside is placed in KCl solution, K^+ ions diffuse into the cell by both concentration and electrical gradient and Cl^- ions will diffuse out by concentration gradient but repelled by electrical gradient.

Donnan Equilibrium for KCl is :

$$E = RT \text{ in } \frac{[K^+ \text{ in}]}{[K^+ \text{out}]} = RT \text{ in } \frac{[Cl^- \text{out}]}{[Cl^- \text{in}]}$$

Since ions are charged particles, their diffusion depends on concentration gradient along with electrical gradient, i.e., along its electrochemical gradient through pores in the plasma membrane.

1. **Ionic Transport Through Charged Pores in the Membrane :** Ionic interchanges across the membrane takes place through electrically charged pores. Some of these pores are charged positively or negatively. Hydrophilic ions like Na^+, Cl^- and HCO_3^- diffuse through these charged protein channels or pores. But only a minute fraction of its surface is used for ionic interchange.
2. **Facilitated Transport of Ions :** Transport of Cl^- and bicarbonate anions across the erythrocyte membrane is facilitated by an anion exchange protein, called **band-3 protein.** In majority of other cells, plasma membrane is impermeable to Cl^- anions.

Q.9. What is GERL? Discuss its significance.

Ans. GERL : The GERL system is a complex found in the cell that involves the Golgi apparatus, ER and lysosomes. The GERL system performs biological process like synthesizing and/or recycling of materials, endocytosis and removal of waste through exocytosis.

Significances of GERL

The GERL complex is a system to carry out the following cellular processes :

(i) Endocytosis of external material
(ii) Exocytosis of internal substances
(iii) Synthesizing or recycling of materials and removal of wastes.

Q.10. What are caveolae? Discuss their role in potocytosis and transduction.

Ans. Caveolae : Tiny pits or caves present on the cell surface formed by the fusion of small vesicles coated with protein **caveolin** are called **caveolae**. These were discovered in 1950s and were named so by **Yamada**. These are associated with :

1. **Potocytosis** *i.e.*, taking into the cell interior small molecules like vitamins.
2. **Transcytosis** *i.e.*, transfer of molecules from the blood through endothelial cells to the other side.
3. **Signal transduction** which is transmission of stimuli or signals from outside into the cell.

Q.11. Write a brief account of scientific contribution of Charak and Dr. Ramdeo Mishra.

Ans.

Charak

Charak is considered to be the father of ancient Indian science of medicine. He was the **Raj Vaidya** (royal doctor) in the court of **Kanishka**. His Charak Samhita is a remarkable book on medicine. It has the description of a large number of diseases and gives methods of identifying their causes as well as the method of their treatment. He was the first to talk about digestion, metabolism and immunity as important for health science. In **Charak Samhita**, more stress has been laid on removing the cause of disease rather than simply treating the illness. Charak also knew the fundamentals of Genetics. It is fascinating to learn that thousands of year back, medical science was such an advanced stage in India.

Dr. Ramdeo Mishra

Professor Ramdeo Mishra did his Ph.D. in Environmental Science from the University of Leeds in 1937. After this, he was appointed to the Department of Botany of Banaras Hindu University, where he did world-class work in environmental science. He was awarded several prestigious awards including Fellowship in Indian National Academy and World Academy of Arts and Sciences and Sanjay Gandhi for Environment and Ecology; Due to his efforts, the Government of India established the National Committee for Environmental Planning and Coordination (1972), which paved the way for the Ministry of Environment and Forests to be established (1984).

Ramdeo Mishra (1908-1998) is considered as Father of Indian Ecology simply because he had contributed remarkably in the field of Ecology among his contemporaries in respect to Indian context. R. Mishra was the person who laid the strong foundation of Ecology in India and not

only he just laid it but also he contributed alot to strengthen the development of Ecology further and thus aided it to flourish progressively over the upcoming decades. He helped in shaping Ecology as a major discipline for teaching as well as for Research in traditional departments in India in many ways.

Q.12. Write the names and functions of different cell organelles.

Ans. Name and Functions of Different Cell Organelles

Cell Components	Functions
Plasma membrane	Protection of cell cytoplasm. Control of substances entering and coming out of the cell.
Endoplasmic reticulum	Provides an increased surface area for the metabolic activities. Synthesis of steroids. Concentration of products of synthetic activities of the cell.
Mitochondria	Act as power houses of cell, release energy by the oxidation of food.
Golgi complex	Produces secretions. Provides surface for the synthetic reactions and the concentration and chemical modification of substances.
Centrosome	Plays an important role in the formation of spindle during cell-division.
Lysosomes	Store enzymes for the digestion of cellular components and brings about digestion of proteins and carbohydrates, etc. Bring about digestion of foreign substances entering the cell.
Ribosomes	Act as factories of the cell and synthesis of proteins from amino acids.
Plastids	In the presence of light, green plastids of chloroplasts manufacture carbohydrate from water and carbon dioxide. Chromoplasts give different colours to the structures in which these are present.
Nuclear membrane	Protects the nucleus. Regulates the passage of substances entering and leaving the nucleus.
Nucleolus	Stores ribosomal RNA and controls synthesis of ribosomes and proteins.
Chromatin or nuclear material	Controls nuclear metabolism and cell metabolism. Stores hereditary information. Inherits characters from parents to offspring.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. Describe the structure of cell with the help of a diagram. Discuss the differences between a plant cell and animal cell.

Ans. All animals and plants consist of certain structural units. The structural unit called cell, is now known as the unit of life and the concept that the cell is basic unit of life is known as the cell theory. The surface are of a cell effects its ability to exchange materials with the environment and puts a check on the cell volume. For this reason, the cells that are metabolically more active are small in size.

The eukaryotic cells are the true cells which occur in plants (from algae to angiosperms) and animals from protozoa to mammals. The eukaryotic cells are typically composed of plasma membrane, cytoplasm and its organelles that is mitochondria, endoplasmic reticulum, ribosomes, golgi apparatus and a true nucleus.

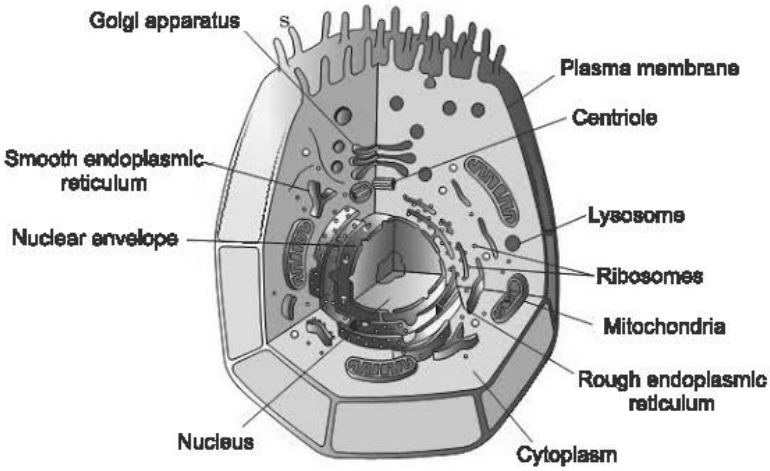


Fig. : Animal (Eukaryotic) cell

A typical animal cell consists of :

1. Plasma membrane or cell membrane,
2. Nucleus and
3. Cytosome or cytoplasm

1. Plasma Membrane or Cell Membrane

Structure : The cytosome of the cell is bounded by an invisible exceptionally thin lipo-protein covering, called **plasma membrane.** It is formed of double layer of lipid molecules sandwitched between two layers of proteins. It is perforated by minute **pores** that are concerned with the material entering or leaving the cell and allow the passage of molecules of specific sizes to and fro. The plasma membrane acts as **selectively permeable.**

Functions of Cell Membrane

(i) Plasma membrane provides mechanical support and definite external form to the cell.

(ii) It forms a part of cell's living machinery because everything that enters or leaves the cell must pass through it.

(iii) It exerts selective influence on the substances entering or leaving the cell. It provides individuality to the cells.

(iv) Helps in the recognition of own cell types and formation of cell aggregates in a tissue.

(v) Permits accumulation of certain substances or ions even more than in the surrounding medium and well regulated reactions. Surrounding the plasma membrane, there may be a tough non-living and supporting membrane or **cell wall.** It is lacking in animal cells, but is thick and formed of cellulose in plants.

2. Nucleus

Structure : The nucleus occurs as a central body in **eukaryotic cells.** Commonly, it is oval, spherical or discoidal, but may be lobed or long and ribbon-like. It may be a single body formed of several separate pieces or in the form of diffused chromatin granules in the cell

substance. The vesicular nucleus is surrounded by a distinct **nuclear envelope** enclosing the nuclear sap or the **nucleoplasm**. In prokaryotic cells the nuclear deoxyribonucleic acid (DNA) is not separated from the cytoplasm by nuclear membrane.

(i) The **nuclear envelope** is similar to plasma membrane in structure and function except that it is formed of two membranes separated by **perinuclear space**. The nucleoplasm is a clear and transparent homogeneous liquid of variable consistency. It contains **nuclear reticulum** and **nucleolus**.

(ii) The **nuclear reticulum** is a fine network of chromatin threads, beaded with coarser chromatin granules. During cell division, chromatin condenses into threads or rods known as **chromosomes**. These are important hereditary vehicles. The chemical constituents of chromatin are nucleoproteins which resolve into four major molecules : a low molecular weight protein **histone**, a complex **deoxyribonucleic acid** (DNA) and **ribonucleic acid** (RNA). The two nucleic acids are compounds of five-atom sugar (pentose), phosphoric acid and four bases. DNA controls all the cell activities, contains all the genetic informations in coded form and controls the formation of RNA, which in turn transports information for the synthesis of specific protein.

Functions of Nucleus

(i) The **nucleus** controls metabolic activities of the cell. It takes an active part during cell division.

(ii) It transmits hereditary informations from generation to generation.

3. Cytosol or Cytoplasm

The interior of the cell is filled with a colourless, transluscent liquid of variable consistency. It is known as **cytoplasm**. It consists of sub-microscopic fibrils, endomembrane system, membrane-bound organelles and non-membranous organelles, structureless ground substance or cytoplasmic matrix. The various components of cytoplasm are :

(i) Cytoplasmic Fibrils and Cytoskeleton : The cytoskeleton of cell is formed of thin, thick and intermediate **filaments** and **microtubules**. These form a network. The cytoskeletal filaments remain interconnected by a network of fine thread-like **microtrabecular lattice**. This lattice also interconnects many membranous organelles.

(ii) Endomembrane System : The endomembrane system of cytoplasm includes : **endoplasmic reticulum, Golgi complex** and **nuclear envelope**. The membranous organelles are mitochondria, **chloroplasts, lysosomes** and **peroxisomes**.

Functions of the Cell

(i) Plasma membrane is formed of cellulose.

(ii) The nucleus controls the metabolic activites. Nucleus takes an active part during cell division.

(iii) The nucleus transmits hereditary information from one generation to next generation.

Differences between a Plant and Animal Cell

S.No.	Plant Cell	Animal Cell
1.	A cellulose cell wall is present surrounding the plasma membrane.	Cell wall is absent. The limiting membrane of the cell is plasma membrane.
2.	These have a large vacuole filled with cell sap.	Vacuoles may be absent, if present they are very small in size.
3.	Centrosome occurs in motile cells of lower plants.	It is present in all animal cells.
4.	Mitochondria are fewer and their cristae are tubular.	Mitochondria are many and with plate-like cristae.
5.	Plant cells contain plastids.	Do not contain plastids.
6.	Nucleus is generally pushed to one side in the peripheral cytoplasm by sap vacuole.	Nucleus is usually located in the centre.
7.	Plant cells do not burst if placed in hypotonic solution due to the presence of cell wall.	Animal cells usually burst if placed in hypotonic solution unless and untill they possess contractile vacuoles.
8.	Lysosomes absent.	Lysosomes present.
9.	Centrioles are absent except in lower plants.	Centrioles are present.
10.	Cytoskeleton does not contain intermediate fibres.	Cytoskeleton contains intermediate fibres.
11.	Crystals of inorganic substances may occur in the cells.	Do not occur in animal cells.
12.	Reserve food is in the form of starch and fat.	Reserve food is in the form of glycogen and fat.
13.	Glyoxysomes may be present.	Absent.
14.	Plant cells are able to synthesise all the amino acids, vitamins and coenzymes needed by them.	Animal cells cannot synthesize all the amino acids, vitamins and coenzymes needed by them.

Q.2. Give a details account of fluid mosaic model of plasma membrane.

Ans.

Fluid Mosaic Model of Plasma Membrane

Fluid mosaic model of biomembrane structure was proposed by **Singer** and **Nicolson** (1972). According to model, concept, biomembranes have **quasifluid structure.** The phospho-lipid molecules form a rather continuous bilayer that forms the structural framework of plasma membrane. The protein molecules are arranged as extrinsic proteins on the surface of lipid bilayer and as integral or intrinsic proteins that penetrate the quasifluid lipid bilayer partially and wholly.

Salient Features of Fluid Mosaic Model

According to fluid mosaic model all biomembranes can be described as 'proteins icebergs' embedded in a sea of lipids.

1. The lipid bilayer is formed of a double layer of phospholipid molecules.
2. Lipids form a fluid crystalline bilayer. This provides flexibility to the membrane and permits transitional movements of lipid and intrinsic protein molecules.
3. The arrangement of phospholipids in biomembranes forms a water-resistant barrier. Through such a membrane lipid soluble substances can pass through readily. For this

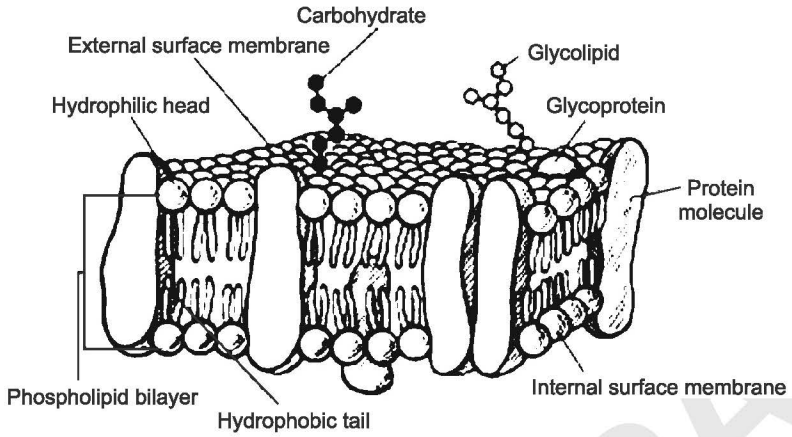


Fig. : Fluid-mosaic model of plasma membrane showing extrinsic and intrinsic proteins.

reason, plasma membrane is permeable to lipid soluble substances, but not for water soluble substances.

4. The phospholipid molecules are **amphiphatic** in nature. The term **amphipathy** was coineed by **Hertley** (1936) for those molecules which have both **hydrophobic** (= water heating) or non-polar ends and **hydrophilic** (= water loving) or polar ends.
5. Amphiphatic molecules form liquid crystalline aggregates in a bilayer. The hydrophobic non-polar tails of phospholipids of lipid bilayer adjoin each other. Their hydrophilic polar ends are directed outward towards aqueous phase and are associated with extrinsic proteins.
6. The intrinsic protein molecules are tightly held in position in the lipid bilayer by strong bonds.
7. Proteins in biomembranes are arranged on either surface of lipid bilayer as **extrinsic** or **peripheral proteins** are embeded partially or wholly in the lipid bilayer. These are called **intrinsic** or **transmembrane proteins.**
8. On the other surface of plasma membrane, oligosaccharides are found attached to some integral proteins and form **glycoproteins.** Some oligosaccharides are bound to lipids forming **glycolipids.** These glycoproteins and glycolipids project from the cell surface and form **cell coat** or **glycocalyx.**
9. **Large globular integral protein molecules** project beyond lipid bilayer on both sides. These are called **transmembranes proteins** or **tunnel** or **channel** proteins. These are believed to bind channels that provide passage for water soluble substances. These provide selective permeability to plasma membrane.
10. **Small integral protein molecules** partially penetrate lipid bilayer and are exposed on one surface only.

    (i) Glycoproteins help in recognising own cell types. This can be demonstrated by a simple experiment. Cells from different tissues were mixed and were placed in a culture medium. The cells were found moving and finally those of one kind grouped together. Cell recognition helps in blood grouping, immune response and rejection of transplant.

(ii) Some glycoproteins of plasma membrane act as **cell receptors.** These bind with specific molecules reaching the cell surface and help in the flow of information or material into the cells. For example, some protein receptors bind to adrenalin hormone.

(iii) Lipoproteins act as drug receptors.

Q.3. Describe the structure and functions of membrane protein present in plasma membrane.

Ans.

Plasma Membrane

The plasma membrane is the outer covering of the cytoplasm of the cell. It is ultrathin elastic lining, dynamic and selective transport barrier. Plasma membrane controls the entry of nutrients and exit of waste products.

Plasma membrane is about 7.5 nm thick but its thickness varies in different types of cells and ranges from 6-10 nm. The plasma membrane of intestinal epithelial cells is about 8.5 nm thick and that of RBCs is about 21.5 nm thick.

The ultrastructure of plasma membrane can be studied by isolating it from rest of the cytoplasm. It is easiest to obtain it form RBCs. These cells are treated with hypotonic solution. The cells swell up and rupture releasing its cytoplasmic contents. These haelomolysed cells are called ghosts of RBCS. Their membrane can be used for biochemical and biophysical studies for studying its ultrastructure.

Chemical Composition : The plasma membrane of different organelles are found to contain proteins' lipids and carbohydrates but these are present in different ratio.

Structure of Membrane Proteins

Proteins from main bulk of biomembranes, ranging from 20 to 70%. Proteins separated from plasma membrane of RBCs are collectively known as **tektins.** The variety of forms of proteins found in biomembranes are classified into following two types :

(i) **Transmembrane Proteins :** These extend through the lipid bilayer projecting out on either side of membrane. These occur in following forms :

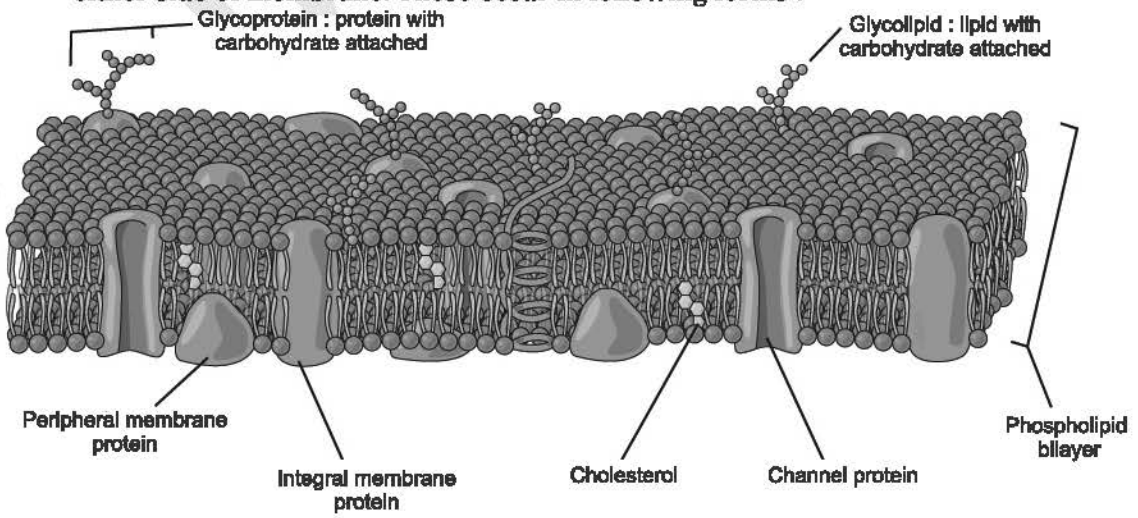


Fig.1 : Association of different types of proteins with lipid bilayer.

(a) **Single pass transmembrane proteins or Bitopic proteins :** These are formed of single polypeptide chains that pass through the lipid bilayer only once. These help in transmitting message from the surface of cell membrane into the cytoplasm, *e.g.,* **glycophorin.**

(b) **Multipass transmembrane proteins or Polytopic proteins :** These proteins possess more than one transmembrane segments which cross through lipid bilayer several times. These proteins form **channels** for the passage of ions and solutes through plasma membrane or may act as **signal receptor proteins,** or as **light sensitive reaction centres.**

(c) **Porins or Barrel proteins :** These occur as β-sheets or β-barrels in the lipid bilayer forming pores or hydrophilic passages in the plasma membrane. These allow hydrophilic solutes to pass across the lipid bilayer.

The transmembrane regions of these proteins are hydrophobic, formed by non-polar amino acids. They interact with the hydrophobic tails of lipid molecules. The parts of trans membrane proteins extending out on both the faces of lipid bilayer are usually hydrophilic and asymmetrical. These may be attached to oligosaccharides, forming **glycoproteins.**

Transmembrane proteins are more than 70% of the membrane proteins. These are of not separated early from the membranes because these are embedded in lipid bilayer and are insoluble in water. Some detergents or organic solvents are needed to separate them from the membrane. Therefore, these are also called **intrinsic proteins.**

The integral proteins remain embedded in the lipid bilayer by three basic types of interactions :

- **ionic interactions** with the polar head groups of lipids.
- **hydrophobic interactions** with the hydrophobic ends of lipids within the lipid bilayer.
- **specific interactions** with defined regions of lipids, glycolipids or oligosaccharides.

Sodium dodecyl sulphate (SDS) is an ionic detergent used for separation of membrane polypeptides.

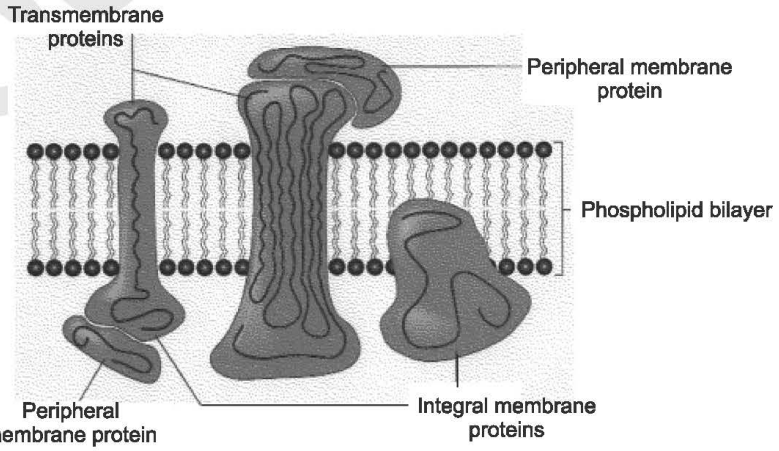


Fig. 2 : Interaction of a typical integral transmembrane protein and a peripheral protein with a lipid bilayer.

The integral proteins occur in following five forms, each carrying out varied functions :

- **Large globular integral protein molecules** project beyond lipid bilayer on both the sides. These are also called **transmembrane** or **channel proteins.** These bind channels or water filled pores that provide passage for water soluble substances and their ions.
- **Small integral protein molecules** partially penetrate the lipid bilayer and are exposed only on one surface.
- **Glycoproteins :** Some oligosaccharides are attached to the integral protein molecules that project on the outer surface of plasma membrane.
- **Carrier proteins :** Some proteins called *permeases* serve for transport of substances across plasma membrane into and out of the cell against concentration gradient (*i.e.,* **Active transport**).
- **Enzymes :** Some membrane proteins act as enzymes. Each membrane carries an assortment of enzymes according to the function it carries out. For example, mitochondrial membranes contain enzymes of electron transport system.

(ii) Peripheral Proteins or Extrinsic Proteins : These are present on both outer and inner surfaces of lipid bilayer and are loosely connected to it either by **covalent** or **non-covalent interactions.** They are hydrophilic or water soluble and are of following types :

(a) **Covalently linked cytosolic extrinsic proteins** located on the cytosolic face of the membrane and attached either by covalently attached fatty acids chains or by prenyl groups.

(b) **Covalently linked non-cytosolic extrinsic proteins** found on the extracellular surface of the lipid bilayer. Each is covalently linked to lipid molecule of the lipid bilayer.

(c) **Non-covalently linked extrinsic proteins** found on either side of lipid bilayer of the membrane and attached to other transmembrane proteins by non-covalent interactions. Thus, these proteins may be cytosolic or extracellular.

Peripheral proteins constitute about 20-30 percent of the total membrane proteins. Some examples of peripheral proteins are : (a) **Spectrin,** (b) **Cytochrome**-C, and (c) **Acetylcholinesterase.** Most peripheral proteins are associated with the inner or cytoplasmic surface of plasma membrane.

The peripheral proteins can be separated by mild treatment such as concentrated salt solution or some chelating agents. These are soluble in aqueous solutions and are usually free of lipids.

Functions of Membrane Proteins

Biological membranes contain three different classes of proteins to carry out different functions :

1. **Enzymes** are catalytic proteins. These form major component of many membranes. More than 30 enzymes have been isolated from plasma membrane. The most common ones are 5'-*nucleotidase,* Mg^{++}, *ATPase,* Na^{+}, K^{+} *activated* Mg^{++} *ATPase, alkaline phosphatase, adenyl cyclase, acid phosphomonoesterase and RNAase.* These **regulate cell activities.**

2. **Structural proteins** form backbone of cell membrane. These provide elasticity and mechanical stability to plasma membrane. These have little catalytic activity.
3. **Membrane transport proteins** or **permeases** serve for the transport of polar molecules of various substances such as ions, monosaccharides, amino acids, nucleotides and certain metabolites across the plasma membrane in and out of the cell. These occur in many forms. Each transport protein is designed to transport a particular class of molecules. These are basically multipass **transmembrane proteins,** projecting on both the sides of lipid bilayer and traversing it multiple times. These are of two types :
   (i) **Carrier proteins** or **carriers** or **transporters** that bind the specific solute to be transported by active transport, *i.e.,* these undergo conformational change in order to transfer the solute across the membrane.
   (ii) **Channel proteins** have pores and form channels that extend across the lipid bilayer. When these pores are open, they allow specific solutes to pass at high flux rates through them across the membrane. The channels can be porins, aquaporins and ion channels.

   Porins are present in some prokaryotes, mitochondria and gap junctions that connect cytoplasms of adjacent cells. These allow passage of solutes and transfer of molecules between adjacent cells.

   Aquaporins allow rapid transport of water across the membrane. These are homoteramer proteins, formed of four identical subunits with each subunit forming a pore.

   Channel proteins are multipass transmembrane proteins that contain a pore region through from one side of the membrane to the other. This pore region is called the **channel pore.**
   - Some channels exist as oligomers of identical or different subunits are formed of two or more subunits, each forming a pore by itself.
   - Some channels are formed of single multipass protein molecule whose transmembrane segments form a pore.
4. **Cell receptors :** Some glycoproteins of plasma membrane act as **cell receptors.** These bind with specific molecules reaching the cell surface and help in the flow of information or material into the cells. For example, some protein receptors bind to adrenalin hormone.
5. **Cell recognition : Glycoproteins** help in recognising own cell types. This can be demonstrated by a simple experiment. Cells from different tissues were mixed and were placed in a culture medium. The cells were found moving and finally cells of one kind grouped together.
6. **Drug receptors :** Lipoproteins act as drug receptors.

Q.4. What are CAMs? Describe different type of CAMs.

Ans.

Cell Adhesion Molecules (CAMs)

Cell adhesion is mediated by **cell adhesion molecules** (CAMs), **substratum adhesion molecules** and **cell junction molecules.** The cell adhesion molecules belong to four different families of membrane proteins. These are four types :

1. Cadherins

Cadherins are a group of glycoproteins that mediate Ca^{2+} dependent cell adhesion. Members of cadherin family are distinguished by the types of cells on which they appear. Although 12 different types of cadherins are known but following three types need mention:

(i) **E-Cadherins** (Epithelial cadherins) or uvomorulins or L-CAM

(ii) **P-Cadherins** (Placental cadherins)

(iii) **N-Cadherins** (Neural cadherins) or A-CAM.

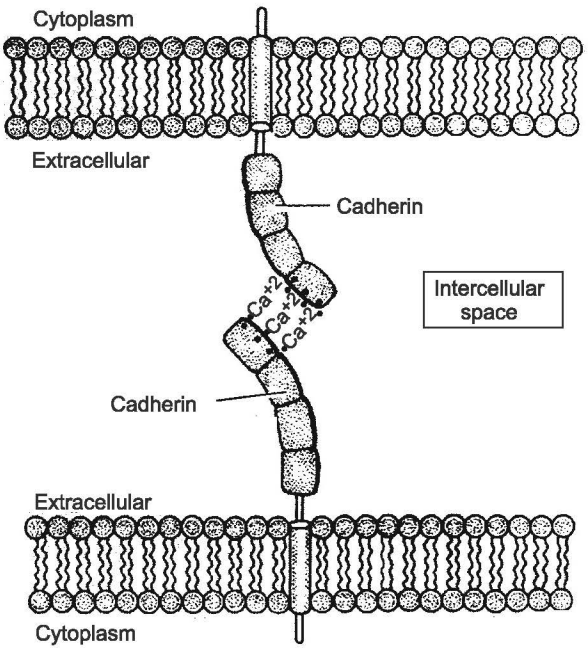


Fig.1 : Cadherin structure and adhesion of two cells through cadherins projecting from their plasma membranes.

Structure : Each cadherin has three major regions :

(a) Extracellular segment : It is the largest segment formed of four domains, each formed by 113 amino acids. It has an NH_2 terminal.

(b) Transmembrane segment : It spans the membrane.

(c) Cytoplasmic segment : It remains within the cell cytoplasm.

2. Selectins

Selectins are a family of integral membrane glycoproteins that recognise and bind to cell specific carbohydrate groups of glycoproteins, projecting from the surface of cells. Selectins also have three parts : (a) a small **cytoplasmic domain,** (b) a **transmembrane domain,** and (c) a large **extracellular domain.** The outermost domain of extracellular segment acts as **lectin** or **carbohydrates recognition domain** (CRD) that binds to the specific carbohydrates groups. There are three types of selectins in vertebrates :

(i) **E-selectin,** which is found on the surface of endothelial cells.

(ii) **P-selectin,** which is expressed on the surface of platelets and endothelial cells.

(iii) **L-selectin,** which is present on all types of leucocytes.

Binding of selectins to their carbohydrate ligands is calcium dependent. Selectins mediate transient interactions between circulating leucocytes and capillary walls to sites of inflammation and clotting.

3. Integrins Cell Receptors for ECM Molecules

Integrins are a class of transmembrane adhesion receptor proteins. These are called integrins because they integrate the extracellular and intracellular scaffolds. They act as **extracellular matrix receptors** (ECM receptors) and adhere to the ECM proteins present on extracellular matrix. These adhesion molecules are formed by **fibronectin** and **vitronectin proteins.**

Each integrin has two non-covalently linked α and β subunits. These form a family of transmembrane α-β heterodimers. There are about 14 different α subunits, 8 different β subunits and 20 different α-β heterodimers.

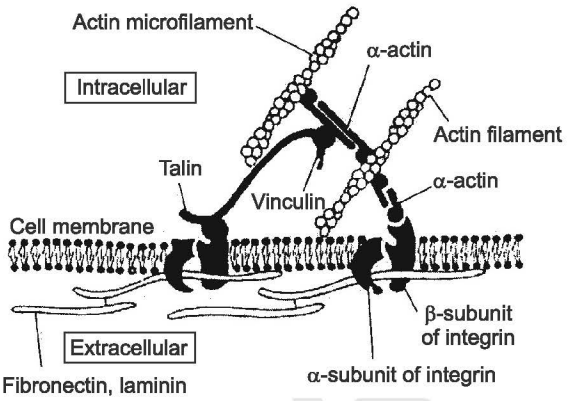


Fig. 2 : Structure and function of integrins in binding to extracellular matrix proteins or cytoskeleton.

4. Immunoglobulins

A group of immunoglobins present on the cell surface act as cell adhesion molecules (CAMs). These form immunoglobulin superfamily of IgSF. These are integral proteins present on the surface of lymphocytes that are involved in various aspects of immune function. Some of these integral proteins mediate **calcium-independent cell-cell adhesion.**

Immunoglobulin-related CAMs were discovered in slime mould-*Dictyostelium.* Most IgSF cell adhesion molecules mediate specific interactions of lymphocytes with macrophages, other lymphocytes and the target cells of the immune response. But neural cell adhesion molecules (NACM) mediate adhesion between non-immune cells.

The CAMs which are found on acitvated endothelial cells and bind to integrins are called intercellular adhesion molecules (ICAMs).

There are at least 20 forms of **NACMs.** Each NACM is formed of following parts :

(a) Extracellular domain : It is folded into five Ig-like domains.

(b) Transmembrane domain spanning the membrane.

(c) Intracellular or cytoplasmic domain : It lies inside the cell cytoplasm which varies in size and is involved in cell signalling or binding to cytoskeleton.

Q.5. What are cell junctions? Describe any two types of cell junctions in animals and plants.

Ans.

Cell Junctions and Their Types

Specialized junctions called cell junctions, are found in all animals tissue between adjoing cells and between cells and extracellular matrix (ECM). In plants, they are described as **plasmodesmata.**

The epithelial cells and cells of cardiac muscle are held together tightly by specialised **adhesive junctions** also called **intercellular junctional complexes.** These are :

1. Desmosomes, 2. Belt Desmosomes, 3. Tight junctions, 4. Gap junctions.

Description of desmosomes and belt desmosomes is given below :

Desmosomes or Macula Adherens

Desmosomes are disc-shaped or button-shaped adhesive junctions. These provide mechanical attachment between adjacent cells, increase rigidity of the tissue and provide attachment for intercellular fibres. These are also called **spot desmosomes.** These have following structural components :

(i) **Intercellular space** between the plasma membranes of adjacent cells. It varies from 30-35 nm.

(ii) **Intercellular core** or **Central stratum** fills the intercellular space. It is formed of specific desmosomal fibres and mucopolysaccharides. It helps in cellular adhesion acting as a cementing substance.

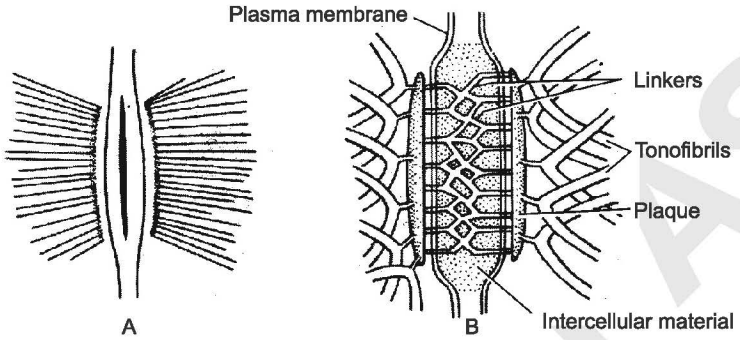


Fig.1 : Structure of a desmosome or macula adherens

(iii) **Intracellular plaque** is discoidal thickening on the cytoplasmic face of each desmosomal plasma membrane. It is about 15-20 nm thick.

(iv) **Tonofilaments** are 10 nm thick or intermediate filaments that run parallel to inner cell surface in bundles and course into and out of the intracellular plaque. These are contractile and formed of keratin. Tonofibrils from cytoskeleton of cell cytoplasm and give it shape and rigidity.

(v) **Transmembrane linkers** are the filaments formed of glycoproteins cadherins. These arise from dense plaques, traverse desmosomal plasma membrane and join the central stratum or intercellular core. Transmembrane linkers bind cells together.

Desmosomes are numerous in cells that are subjected to mechanical stress.

- **Hemidesmosomes :** These are found on the basal surface of some epithelial cells. Their structure is similar to desmosomes but these are represented only by one-half. Their counterpart is usually represented by collagen fibrils.
- **Septate Desmosomes :** These are found in the epithelial cells of invertebrates. These occur in the form of transverse septa disposed parallel in the intercellular space 150-200 Å wide between the plasma membranes of adjacent cells and are continuous.

Belt Desmosomes or Adherens Junctions or Zonula Adherens

These are also called **anchoring junctions** or **intermediary junctions.** Each junction forms an adhesion belt below the tight junction between adjacent columnar cells of intestinal epithelium. Each belt desmosome is formed of two types of fibres :

(i) Actin Microfilaments : These are 6 highly contractile fibres, formed of protein actin. These form a band on the inner surface of plasma membrane and encircle the cell like a belt.

(ii) Intermediate Filaments : These are formed of **actin binding proteins.** These form an interwoven mass and traverse the cell at the level of belt. These also provide attachment to the actin filaments that pass through the core of microvilli.

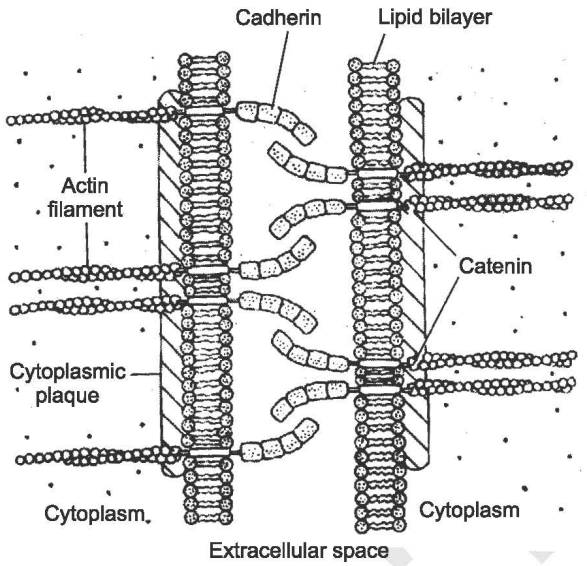


Fig.2 : Structure of zonula adherens or adherens junction

The plasma membrane of adjacent cells at adherens junction are separated by 20-35 nm wide **intercellular space.** This space is occupied by extracellular domain of cadherin molecules from adjacent plasma membranes which are joined by calcium dependent linkages. The intracellular domains of these cadherin molecules are linked to cytoplasmic proteins or to actin filaments of cytoskeleton.

Q.6. What is active transport? Give one example, describe its mechanism and importance in living organisms.

Ans.

Active Transport

Active transport uses specific transport for protein called pumps which use metabolic energy to move against their concentration. Two types of pumps are present in different types of ion or molecules such as calcium pump, proton pump, etc. The movement of molecules can be compared with the uphill movement of water. Energy is required to counteract the force of diffusion and is obtained from ATP. Na^+, K^+, ATPase is an ion pump or cation exchange pump and it participates in active transport of substances.

Example : The ion pumps responsible for maintaining gradients of ions across the plasma membrane ($Na^+ — K^+$ pump) are best examples of active transport driven by ATP. In order to maintain low intracellular concentration of Na^+ ions, the cell extrudes sodium against higher concentration of sodium outside the cell which is about ten times higher than inside concentration of sodium ions. The $Na^+ — K^+$ pump transports Na^+ and K^+ against their electrochemical gradients and uses energy derived from ATP hydrolysis.

Mechanism of Active Transport

1. **Carrier Molecule Mechanism of Active Transport Molecule or Role of Permeases in Active Transport :** It is presumed that a **carrier molecule** which is a component of the plasma membrane picks up the molecule of transport (*i.e.,* the molecule which is to be transferred to the cell) and forms a **carrier-transportant complex.** The carrier

may be protein, lipid or an enzyme. For example, in the active transport of Na^+ ions, Mg^{++} activated ATPase acts as a carrier. Its ATP provides energy for the transport. The sodium ion is picked up from the outside forming a temporary complex which is carried to the opposite side of plasma membrane, to be released inside the cell. The transportant undergoes metabolic changes along with the carrier. The carrier protein is known as **translocase** or **permease.** Certain enzymes are found to assist in this active transport.

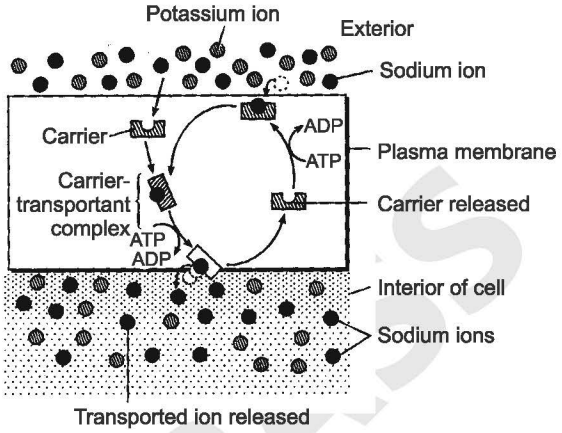


Fig.1 : Process of active transport of Na^+ ions through the plasma membrane when these are in higher concentration inside the cell.

2. **Revolving Door Model for Active Transport : Monod** and **Cohen** have described that during the transport of lactose across the plasma membrane in *E. coli*, the carrier protein has a slot facing outside. The molecule of the substance to be transported fits into this slot. The carrier protein changes its shape as the substance enters the slot and rotates so that the slot comes to lie on the inner side. The substance is released in the cell and protein rotates back to its original form. The energy is spent during the process of rotation.

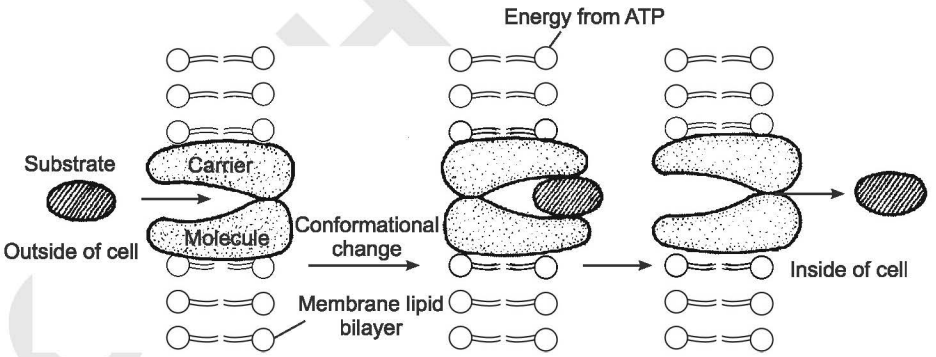


Fig. 2 : Revolving door model of active transport.

New Concept : On ATP molecule exports three Na^+ ions outside the cell in exchange of the import of two K^+ ions inside the cell. The larger sub-unit of Na^+, K^+ ATPase perform the actual function of cation transport. It is believed that the hydrolysis of one ATP molecule some how drives confirmational changes in the Na^+, K^+, ATPase that allow the pump to transport three Na^+ ions out and two K^+ ions inside the cell.

Enzymes for Active Transport

The enzyme ATPase catalyses the hydrolysis of ATP on the intercellular side by utilizing OH^- from the inside and H^+ from the outside. As a result, outer side of the plasma membrane becomes more alkaline and the inner side becomes more acidic.

Importance of Active Transport

Both simple and facilitated diffusion permit passage of a solute across a membrane along concentration gradient. Substances against concentration gradient move by active transport. Therefore, active transport is very essential as it enables the cell in :

(i) The absorption of materials against or independent of concentration gradient, such as :
  (a) **glucose** is absorbed against concentration gradient in the uriniferous tubules,
  (b) absorption of monosaccharides by the intestinal cells is also due to active transport.

(ii) maintaining $Na^+ — K^+$ pump for establishing **membrane potential** by maintaining unequal distribution of ions between inside and outside the cell membrane. It helps in the conduction of nerve impulse in nerve cells and in maintaining a specific level of Na^+ in the blood.

(iii) pumping a solute against the electrochemical gradient.

Q.7. What do you understand by endoplasmic reticulum? Describe its various components. Also discuss about types, structure and functions of endoplasmic reticulum?

Ans.

Endoplasmic Reticulum

The cytoplasmic matrix is transversed by a complex network of interconnecting vacuoles or cavity. These vacuoles are cavities that remain concentrated in the endoplastic protein of the cytoplasm, therefore known as endoplasmic reticulum.

The endoplasmic reticulum is absent in prokaryotes. It occurs in all the eukaryotic cells except erythrocytes (RBCs) of mammals. It is small and undifferentiated in eggs and in undifferentiated embryonic cells. Only a few vacuoles are present in the spermatocytes and muscle cells. In muscle cells it is called **sarcoplasmic reticulum.** But it is highly organised in cells synthesising proteins for export, in cells that are engaged in lipid metabolism and cells involved in the metabolism of steroid hormones, drugs and toxic substances.

Components of ER

The main components of endoplasmic reticulum discussed as below :

1. **Cisternae or Lamellae :** These are elongated, flattened and usually unbranched tubular vesicles arranged in parallel rows. These form successive layers around the nucleus. These are about 40-50 mμ thick. Cisternae are found in the cells actively busy in protein synthesis like the cells of liver, pancreas, and brain.
2. **Tubules :** The tubules are small, smooth-walled branched tubular spaces having a diameter of about 50-190 mμ. These occur in cells that are busy in the synthesis of steroids, cholesterol, glycerides and hormones. These are haphazardly arranged in the cytoplasm of developing spermatids of guinea pig, muscle cells and other non-secretory cells.
3. **Vesicles :** These are rounded, spherical or ovoidal spaces measuring from 35-500 mμ. These occur abundantly in cells busy in the synthesis of protein and can be seen in liver and pancreatic cells.

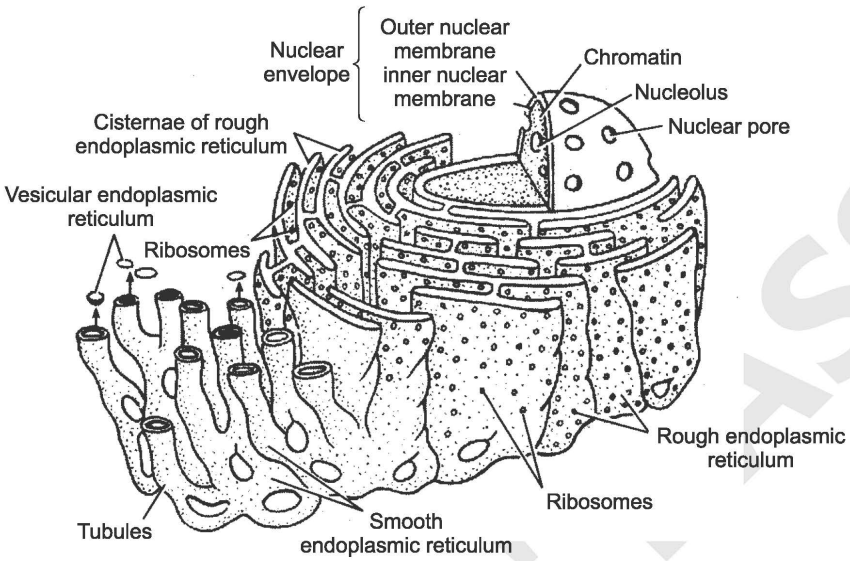


Fig. 1 : Components of ER

Types of Endoplasmic Reticulum

Two types of endoplasmic reticulum have been observed in different types of cell :

1. **Agranular or Smooth Endoplasmic Reticulum (SER) :** The smooth type of endoplasmic reticulum occurs mostly in those cells which are involved in the metabolism of lipids and glycogen. Smooth endoplasmic reticulum occurs in cells that do not synthesise proteins like adipose cells, glycogen storing cells and the muscles cells.
2. **Granular or Rough Endoplasmic Reticulum (RER) :** The granular or rough endoplasmic reticulum is found in those cells which are active in protein synthesis such as pancreatic cells, plasma cells, goblet cells and liver cells.

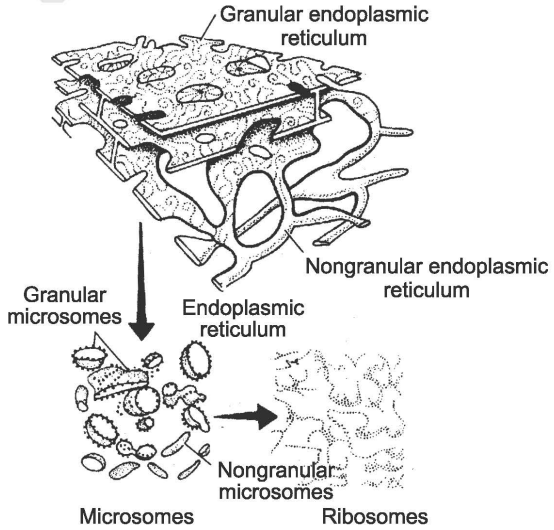


Fig. 2 : Relationship between different components of ER.

Functions of Endoplasmic Reticulum

Following are the main functions of ER :

1. **ATP Synthesis :** ER membranes are the sites of ATP synthesis in the cell. The ATP is used as a source of energy for all the intracellular metabolism and transport of materials.
2. **Glycogen Metabolism :** Agranular ER is related to the withdrawl of glucose from glycogen through the action of glucose-phosphate. This is known as **glucogenolysis.** This is supported by the occurrence of glycogen particles in liver cells in those regions in which agranular ER is well developed.

3. **Mechanical Support :** The vacuolar system of endoplasmic reticulum provides mechanical support to the colloidal matrix of cytoplasm.
4. **Exchange of Materials :** The membranes of vacuolar system maintain osmotic pressure within the cells, isolate the materials synthesised or delivered and regulate exchange between the inner and outer compartments or cell compartments and the cytoplasmic matrix.
5. **Intracellular Transport :** The endoplasmic reticulum acts as a **circulatory system** engaged in import, export and intracellular circulation of various substances through the phenomenon of membrane flow.

   The membrane flow is due to membrane flux. The membrane is synthesised at one point (AA_1) and breaks down at the other point (B). Thus, the membrane flows in the direction A $\rightarrow$ B and the particles attached to the surface of the membrane in this region are incorporated into the cytoplasm. A similar mechanism, but in the reverse direction affects the transport of particles from the cytoplasm to the exterior. The continuity of endoplasmic reticulum either with plasma membrane or with nuclear membrane or with both indicates that membrane flow may be active in this region.
6. **Enzyme Activities and Cellular Metabolism :** Numerous enzymes mainly those involved in the metabolism of steroids (cholesterol and glycerides), phospholipids and hormones (testosterone and progesterone) are associated with the membranes of smooth endoplasmic reticulum. They facilitate free union of enzymes with their substrates.
7. **Synthesis of Lipoproteins :** The smooth endoplasmic reticulum is involved in the synthesis of lipids. In the Golgi membranes, glycerides get associated with the proteins synthesised by the rough surface endoplasmic reticulum and form lipoprotein complexes.
8. **Detoxification :** The smooth surface ER brings about detoxification of many endogenous and exogenous compounds. Prolonged administration of certain drugs produces an increase in the smooth endoplasmic reticulum and the production of specified enzymes.
9. **Synthesis of Exportable Proteins :** Protein synthesis in intimately associated with ribosomes. The ER provides surface for the attachment of ribosomes and facilitates protein synthesis. The synthesised proteins are released and stored in the channels of endoplasmic reticulum and are exported outside the cell.
10. **Segregation of proteins or Protein sorting to different organelles :** Several thousand proteins are synthesised inside the cell, which may be exported out of the cell, incorporated in different intracellular compartments or may be integrated into the endomembranes. The proteins with different fate are segregated in separate chambers such as hydrolytic enzymes are enclosed in lysosomes, and oxidative enzymes in mitochondria.
11. **Biosynthesis of Lipid Bilayers of Cellular Membranes :** The ER membranes produce nearly all the lipids required in the biosynthesis of cellular membranes. These are formed in the cytosolic half of ER lipid bilayer and are organised into a monolayer. A translocator enzyme, *flippase,* catalyses the flipping or transfer phospholipid molecules from this monolayer to luminal side which then organise to form luminal half of lipid bilayer.
12. **Transportation of Message from Genetic Material :** ER provides passage for the genetic material to pass from the nucleus to the various organelles in the cytoplasm, thereby controlling the synthesis of proteins, fats and carbohydrates.

13. **Formation of Other Cytomembranes :** Other membranous structures of the cells like nuclear membrane and Golgi complex differentiate from the ER. During cell division, nuclear membrane disintegrates into small vesicles. The vesicles move towards the poles of the spindle in metaphase. Later on, in telephase the fragmented vesicle and the elements of ER migrate and then accumulate around the chromosome groups. The elements thus fuse forming a complete nuclear membrane. The bits of ER participate in the formation of cell-plate also.
14. **Intracellular Impulse Conduction : Portor** (1926) established the possibility of existence of ionic gradients and electric potential across the membranes of endoplasmic reticulum. The sacroplasmic reticulum present in the sacroplasm of the striated muscle cells acts as an intracellular **conducting system** transmitting impulses from the surface membrane or sacrolemma into the deep regions of the muscle fibre. The reticulum of pigmented epithelial cells of retina acts as a **photoreceptor.**

Q.8. Discuss protein sorting and vesicular traffic from endoplasmic reticulum to Golgi.

Ans. In membrane transport, a variety of substances move into and outside the cell or from cytosol to various cell organelles, whereas in vesicular transport only secretory proteins or protein of biosynthetic secretory pathway are transported from one organelle to other or from organelle to outside the cell.

In membrane transport, transportant passes through the plasma membrane either through membrane pores or ionosphores or through protein channels or with the help of transporter proteins. In vesicular transport, the secretory product is enclosed in a membrane, forming **secretory vesicle.** The secretory vesicle moves through the cytosol and ruptures to release contents on the cell surface.

Protein Sorting and Vesicular Traffic from ER to Golgi

The movement of secretory proteins from ER to Golgi and then to the cell surface is believed to follow some specific pathways. The proteins to be transported (the cargo) by vesicles first appear as **pre-Golgi** intermediates (PGI), which move on microtubule tracks to the *cis*-Golgi region. As the cargo moves through the stack of Golgi cisternae, it is modified by **Golgi associated processing enzymes** and reach *trans*-Golgi surface, where sorting of cargo for various destinations takes place. This is described as **directed maturation of Golgi compartments** or **'cisternal progression'.** The transport from ER to Golgi may involve **default pathway** or **controlled** or **signaled pathway** including retention of transport signals.

1. Default Pathway

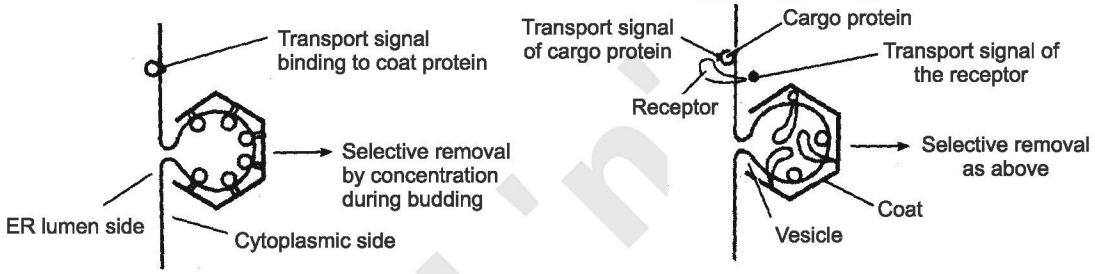
In this pathway, any protein entering ER from cell surface or from cytosol is automatically transported to Golgi and then to the cell surface, unless there are signals either for its retention in a specific compartment or its transport to lysosome or to secretory vesicle. It means such proteins lack both retention or transport signals. The incorrectly assembled or wrongly folded protein molecules are degraded within ER.

2. Controlled or Signaled Pathway

Recently, it has been established that sorting of proteins and their transport from ER to Golgi needs transport signals. Two types of signals control sorting and transport of proteins.

(i) ER Retention Signals : Whether a protein will or will not enter a vesicle budding from ER depends on the absence or presence of retention signal present on the protein. It means proteins with retention signal do not move out of ER. Sometimes, a few molecules of such proteins escape into Golgi or into secretory vesicle due to leakage. But these are brought back to ER. The receptors for ER retention signal are located in Golgi. These receptors attach to the protein molecules carrying retention signal and are transported back from Golgi to ER through vesicular traffic. In this transport, the concentration of cargo is low in the vesicles.

On reaching ER, these complexes separate into retention receptors and protein molecules with retention signal. The receptors return back to Golgi through vesicles, while protein molecules with signal are left in ER. In this mode of transport, the concentration of cargo protein is low in the vesicle in comparison to its concentration in ER. The retention signal on protein *Bip* is a sequence of four amino acids (Lys-Asp-Glu-Leu) and is represented as **KDEL.**



(a) Transport signal binds to coat protein

(b) Transport signal binds to coat protein indirectly through a receptor

Fig. : Transport of membrane bound protein : (a) When transport signal bind to coat protein directly; (b) When transport signal binds to coat protein indirectly through a receptor.

(ii) Sorting Signals : Sorting signals determine whether a protein will enter a secretory vesicle or lysosome or none. The sorting signals include either some peptide domain of 4-25 amino acids or some epitope. A protein might have one or multiple signals, each determining the fate of the protein at one of the successive steps during its transport. Depending upon the sorting signals, there can be following potential fates of a protein during vesicular transport :

(a) In case, the membrane bound cargo protein carries the signal on its cytoplasmic side, its signal binds directly to a coat protein.

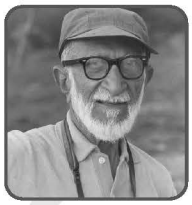
(b) In case, the membrane bound cargo protein carries the signal on its luminal side, the signal binds to a receptor, whose cytoplasmic domain contains a transport signal. This transport signal in turn binds to coat protein.

Q.9. Give important contributions of the following three zoologists of modern India : (i) Salim Ali (ii) Lalji Singh (iii) Har Gobind Khorana.

Ans.

(i) Salim Ali

Salim Moizuddin Abdul Ali (12 November, 1896-20 June, 1987) was an Indian ornithologist and naturalist (study of the behaviour of birds). Sometimes referred to as the *"Birdman of India"*, Salim Ali was the first Indian to conduct systematic bird surveys across India and wrote several bird books that popularized ornithology in India. He became a key figure behind the Bombay


Salim Ali

Natural History Society after 1947 and used his personal influence to garner government support for the organisation, create the Bharatpur bird sanctuary (Keoladeo National Park) and prevent the destruction of what is now the Silent Valley National Park. Along with Sidney Dillon Ripley, he wrote the landmark ten volume *Handbook of the Birds of India and Pakistan,* a second edition of which was completed after his death. He was awarded the Padma Bhushan in 1958 and the Padma Vibhushan in 1976, India's third and second highest civilian honours respectively. Several species of birds, Salim Ali's fruit bat, a couple of bird sanctuaries and institutions have been named after him.

(ii) Lalji Singh


Lalji Singh

Lalji Singh (5 July 1947-10 December 2017) was an Indian scientist who worked in the field of **DNA fingerprinting technology** in India, where he was popularly known as the "Father of Indian DNA fingerprinting". Singh also worked in the areas of **molecular basis of sex determination, wildlife conservation** forensics and **evolution and migration of humans.** In 2004, he received the **Padma Shri** in recognition of his contribution to Indian science and technology.

Lalji Singh founded various institutes and laboratories in India, including the **Centre for DNA Fingerprinting and Diagnostics** in 1995. Laboratory for the Conservation of Endangered Species (LaCONES) in 1998, and Genome Foundation in 2004, aiming to diagnose and treat genetic disorders affecting the Indian population, in particular the under-privileged people residing in rural India.

Singh served as the 25th Vice Chancellor of **Banaras Hindu University (BHU)** and Chairman of Board of Governors of **Indian Institute of Technology (BHU) Varanasi** from August 2011 to August 2014. Before his term as Vice Chancellor of Banaras Hindu University, he also served as director of the **Centre for Cellular and Molecular Biology** (CCMB) from May 1998 to July 2009 and Officer on Special Duty (OSD) of **Centre for DNA Fingerprinting and Diagnostics** (CDFD), Hyderabad, Indian in 1995-1999.

(iii) Har Gobind Khorana


Har Gobind Khorana

Har Gobind Khorana (9 January 1922-9 November 2011) was an **Indian American** biochemist. While on the faculty of the **University of Wisconsin-Madison,** he shared the 1968 **Nobel Prize for Physiology or Medicine** with **Marshall W. Nirenberg** and **Robert W. Holley** for research that showed the order of **nucleotides** in **nucleic acids,** which carry the **genetic code** of the cell and control the cell's synthesis of proteins. Khorana and Nirenberg were also awarded the **Louisa Gross Horwitz Prize** from **Columbia University** in the same year.

Born in **British India,** Khorana served on the faculties of three universities in **North America.** He became a **naturalized citizen** of the **United States** in 1966, and received the **National Medal of Science** in 1987. ❑

UNIT-II

Structure and Functions of Cell Organelles-II in Animal Cell

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. What is rigor mortis and why does it occur?

Ans. **Rigor mortis** is due ot a biochemical change in the muscles that occurs several hours after death, though the time of its onset after death depends on the ambient temperature. Without ATP, myosin molecules adhere to action filaments and the muscles become rigid.

Q.2. What are actin filaments?

Ans. Actin filaments (F-actin) are linear polymers of globur actin (G-actin) subunits and occur as microfilaments in the cytoskeleton and as thin filaments, which are part of the contractile apparatus, in muscle and non-muscle cell (contractile bundles).

Q.3. What do you understand by microtubules?

Ans. Microtubles are polymers of tubulin that form part of the cytoskeleton and provide structure and shape of eukaryotic cells. They are formed by the polymerization of adimer of two glubualr proteins, α and β-tubulin into protofilaments that can then associate laterally to form hollow tube the microtubule.

Q.4. Write about 'sliding filament model' of muscle contraction.

Ans. The sliding filament model of muscle contraction, put forward by hugh Huxley and Iean Hanson in 1954. This theory describes the mechanism that allow muscles to contract. According to this theory, myosin (a motor protein) kind to actin. The myosin then alters its configuration, resulting in a 'stroke' that pulls on the actin filament and causes it to slide across the myosin filament.

Q.5. Describe the location of different enzymes within a mitochondria.

Ans. The enzymes of citric acid cycle are located in the mitochondrial matrix with the exception of succinate dehydrogenase which is bound to the inner mitochondrial membrane as part of complex-II.

Q.6. Describe briefly the origin of mitochondria from free living proteobacteria.

Ans. Intracellular endosymbionts that originally descended from free living prokaryotes have been important in the evolution of eukaryotes by giving rise to two cytoplasmic organelles. Mitochondria arise from α-proteobacteria and chloroplasts arise from cynobacteria.

Q.7. Discuss various functions of Golgi complex in different types of cells.

Ans. Functions of Golgi complex in different types of cells are as follows :

S.No.	Cell Types	Golgi Functions
1.	Goblet cells of intestinal mucosa	Secretion of **mucus** and **zymogens**.
2.	Hepatic cells of liver	Transformation and secretion of lipids.
3.	Follicle cells of thyroid glands	**Prethyroglobulin** (hormone).
4.	Exocrine cells of pancreas	Secretion of **zymogens** (digestive enzymes-proteases, lipases; carbohydrases and nucleases).
5.	Brunner's gland cells of duodenum and ileum	Synthesis and secretion of mucopolysaccharides.
6.	Plasma cells of blood	Immunoglobulins.
7.	Plant cells	Secretion of pectin and cellulose.

Q.8. Write about different types of endosomes.

Ans. Two types of endosomes have been recognised :

(i) early endosomes and (ii) late endosomes.

The former are found beneath the plasm membrane and are believed to give rise to the latter, which are found near the nucleus and nature into lysosomes.

Q.9. Describe different ribosomal subunits in prokaryotes and eukaryotes.

Ans. All prokaryotes have 70S (where, S = Swedberg units) ribosomes white eukarykotes contain larger 80S ribosomes in their cytosol. To 70S ribosome is made up of a 50S and 30S subunits. The 50S subunit contains the 23S and 5S *r*RNA while the 30S subunit contains the 16S *r*RNa.

Q.10. Which diseases are caused due to abnormalities in peroxisomes in humans?

Ans. Peroxisome biogenesis disorders (PBDs) include the Zellmeger syndrome spectrum (PBD-ZSD) and rhizomelic condrodysplasia punctata type-I (RCDP-1). PBD-ZSD represents a continuum of disorders including infantile refsum disease, neonatal adrenoleukodystrophy and Zellmeger syndrome.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Describe different types of intermediate filaments.

Ans. On the basis of morphology and localization within the cell, the intermediate filaments have been grouped into following four main types :

(i) **Keratin filaments** (also known as **tonofil aments, prekeratin** or **cytokeratin**) are the most complex type of intermediate filaments. They are found in epithelial cells, where they are anchored to the cell surface and converge upon **desmosomes**. Mammalian cytokeratins are composed of polypeptides ranging in size from 47,000 to 58,000 daltons. They are α-fibrous proteins, that form bulk of the dead layers or **stratum corneum**.

(ii) **Neurofilaments**, together with microtubules, form the main constituents of axons, dendrites and neuronal perikaryon. They contain three polypeptides ranging in molecular weight from 200,000 to 68,000 daltons.

(iii) **Glial filaments** are found throughout the cytoplasm of astrocytes and are composed of a very acidic protein, 51,000 daltons in molecular weight,

(iv) **Heterogenous filaments** contain different proteins such as **desmin** (consisting of two major polypeptides of 50,000 and 55,000 daltons; found predominantly in smooth, skeletal and cardiac muscles), **vimentin** (mol. wt., 52,000 daltons: found in cells of different origins) and **synemin** (mol. wt. 230,000 daltons; found in skeletal muscles).

Q.2. What is the molecular motor? Also write its types.

Ans.

Molecular Motor

Molecular motors are a large number of motor proteins that operate in conjunction with cytoskeleton and are responsible for cell movements. They are **mechanochemical transducers**, that convert chemical energy (stored ATP) to mechanical energy for moving cellular cargo attached to the motor.

Types of Molecular Motors

A large number of motor proteins are known, which are grouped into three families : **kinesins, dyneins** and **myosins**. The kinesins and dyneins move along microtubular tracks while myosins move along microfilament tracks.

1. **Kinesins :** Kinesin is a large protein. It consists of two heavy chains and is differentiated into **head, stalk** and **tail**. The force generating head binds to the microtubules and the tail binds to the cargo to be transported. In the stalk region, the two heavy chains wrap around each other. Kinesins are associated with the movement of ER derived vesicles, endosomes, lysosomes and secretory granules.

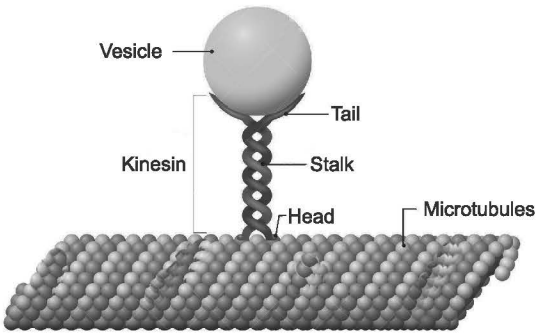


Fig.1 : A. Structure of a kinesin molecule and B. Movement of a kinesin molecule with a vesicle along a microtubular track.

2. **Cytoplasmic Dyneins :** Dynein is a huge protein having a molecular weight over 1 million daltons. It is composed of 9 to 10 polypeptide chains. Each dynein molecule contains two large globular **heads**, a **stalk** and a **base**. The heads act as force-generating engines. Each head is composed of a dynein heavy chain.

   The base is formed of a number of small subunits. These mediate the binding of the motor protein to cargo being transported. Cytoplasmic dyneins are responsible for retrograde movement of cytoplasmic organelles like Golgi vesicles, lysosomes and endosomes towards the cell centre along microtubutes. Kinesins and dyneins move in opposite directions. Most kinesins move forward the plus end and dyneins towards minus end of the microtubules.

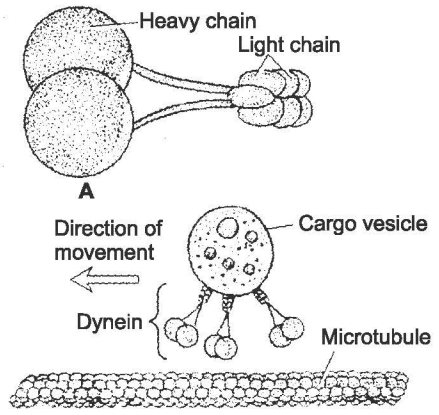


Fig.2 : A. Structure of a cytoplasmic dynein; B. A cargo vesicle moving along microtubule.

Q.3. How are intermediate filaments linked with genetic diseases?

Ans. In several cases, diseases in humans and mice were found to be associated with. disturbances, in the assembly of intermediate filaments (IFs). For instance some skin diseases were found to be associated with mutations in keratins (IF proteins), for which there are 50 different genes in human genome. These keratins have been studied in the mitotically active epidermal keratinocytes of the skin.

The epidermis consists of a stratified epithelium composed of

(i) an inner layer of dividing cells containing 25% of their protein as k 5 and k 14 keratins.

(ii) outer layers of differentiating cells containing k l and k 10 making upto 85% of the protein in mature cells.

Thus, the cells in all these layers possess extensive keratin filament networks. It has been shown that in k 14 null mutations, the cells become fragile and prone to rupturing and degeneration upon mechanical stress. It was also shown that in the absence of k 14, its dimerization partner k 5 was also unstable, so that the keratin network was almost absent.

Mutations in the genes for k 14 and: k 5 keratins were also identified in a human skin disorder called **epidermolysis buliosa complex (EBS)**, which led to disturbances in IFs assembly, although not all cases of EBS are due to such mutations. Similar mutations in genes for k 1 or k 10 resulted in skin disorder called **epidermolytic hyperkeratosis (EH)**, and those in k 9 resulted in **epidermolytic palmoplanar keratoderma (EPPK)**.

Abnormal accumulation and disorganization of **neurofilaments** also results in a number of motor neuron diseases including **amystrophic lateral sclerosis** (ALS), **infantile spinal muscular atrophy** and **hereditary sensory motor neuropathy**. Similarly absence of muscle-specific IF protein **desmin** causes **cardiovascular lesion and skeletal muscle myopathy**, as shown in mice.

Q.4. Describe briefly the transport of protein within mitochondria.

Ans. The mitochondrion contains a distinct set of proteins, but only a few of these proteins are encoded by mitochondrial genome. The proteins encoded by the mitochondrial genome (mtDNA) generally form only some subunits of protein complexes meant for the inner membrane of mitochondria, the other subunits being encoded by the nuclear genome and later transported to mitochondria. Special mechanisms are needed for this transport of proteins and will be briefly discussed.

The precursors of mitochondrial proteins are synthesized in the cytoplasm and after their release from polyribosomes into the cytosol they are taken up by the mitochondria within a minute or two.

These proteins may belong to either the outer membrane, or the inner membrane, or the peri-mitochondrial space or the mitochondrial matrix and the mechanism of transport may differ in each case. While the transport of a protein to outer membrane or to mitochondrial matrix requires a single signal peptide, its transport to the inner membrane or the peri-mitochondrial space also requires a second signal peptide.

Q.5. Describe the salient features of mitochondrial genome.

Ans. **Mitochondrial Genome**

It has been shown earlier that DNA. the genetic material, is found in mitochondria also and is described as mtDNA. This DNA has been shown to be unrelated to nuclear DNA and is

associated with extrachromosomal mitochondrial inheritance. The mitochondrial DNA resembles bacterial DNA and can be ring shaped or linear. In several organisms (particularly in plants including fungi), where mtDNA was earlier (during 1980s) reported to be circular, it was later shown to be linear.

Organisms having linear mtDNA are now known to belong to the following groups : (i) ciliates, (ii) some parasitic protozoa, (iii) algae, (iv) slime molds, (v) oomycetous fungi and (vi) yeasts.

This suggests wide occurrence of linear mitochondrial genome. In these linear mtDNA molecules, mitochondrial telomeres: have also been identified and characterized. The role of these telomeres in mtDNA replication has also been elucidated in some cases. Mitochondrial genome in different eukaryotes varies from 6 kilobases *(Plasmodium)* to 367 kilobases *(Arabidopsis)*.

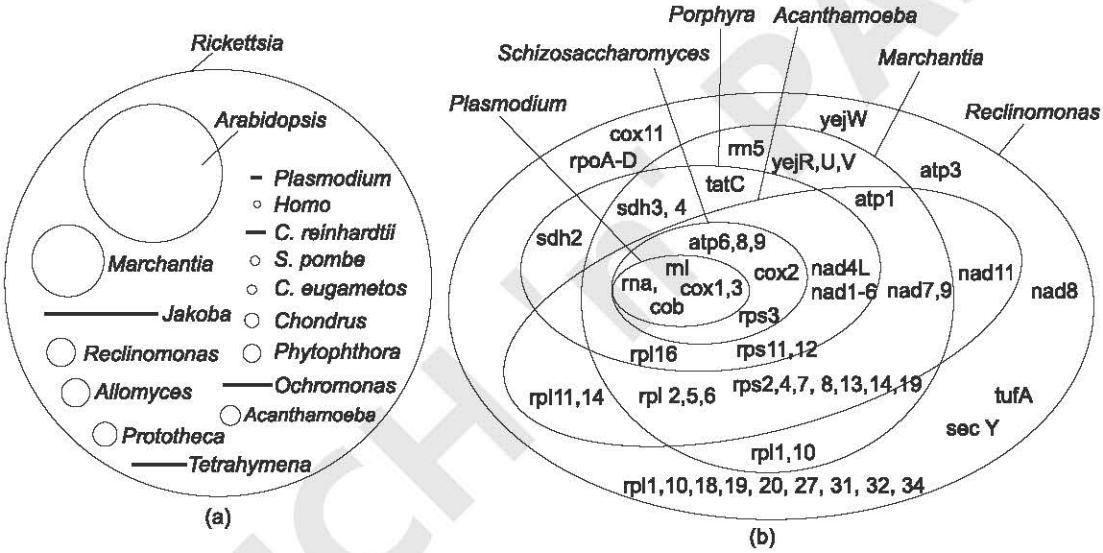


Fig. : Variation in size (a) and gene content and (b) of mitochondrial genomes, compared with an α-Proteobacterial (*Rickettsia*) genome (circles and lines represent circular and linear shapes of genomes).

The gene content of these mitochondrial genomes is rather low relative to that of a eubacterial genome from which they are believed to have been derived through **endosmbiosis**. Only four genes (*cob, coxl* and two rRNA genes) are common to all mtDNAs, and many genes are present in only one or a few unrelated taxa. In mtDNA of *Plasmodium*, even genes for tRNAs are completely absent, which are found in most mtDNAs examined so far.

Complete sequence of mtDNA is now known in many species including human beings and have also been used for drawing phylogenetic conclusions. One of the primitive mtDNA belongs to the heterotrophic flagellate, *Reclinomonas americana*. Complete nucleotide sequence of this mtDNA was published in 1997 and revealed several interesting features, suggesting that this genome is a link between mtDNA of other eukaryotes and the genome of eubacteria.

Q.6. Write brief account of the ATP synthase.

Ans. ATP synthase (F_1F_0ATPase) contains 16 different proteins and is greater than 500kD in size. It consists of two parts (Fig.); (i) F_0 present in the membrane contains the proton channel and (ii) F_1 located on the matrix side of the membrane. F_0 and F_1 are connected by a

stalk consisting of two parallel structures, referred to as a **'rotor'** ($\gamma\varepsilon$) and a **'stator'** ($\alpha\beta_2\delta$) F_1 ATPase can be separated from F_0 and consists of a set of polypeptides in the following stoichiometry : $\alpha(3) : \beta(3) : \gamma(1) : \delta(1) : \varepsilon(1)$. Only β has catalytic site and three β polypeptides at a particular time are found in the following three states: (i) Open β (without ATP); (ii) loose β (ADP+P) and (iii) tight (ATP). This is described as **Boyer's binding exchange mechanism**, according to which energy is needed only for binding of ADP+P or for release of ATP, and not for the formation of ATP. It is also known that $\alpha_3\beta_3$ form a barrel and inside this barrel, γ rotates and facilitates binding of ADP+Pi and release of ATP (**rotational analysis model**).

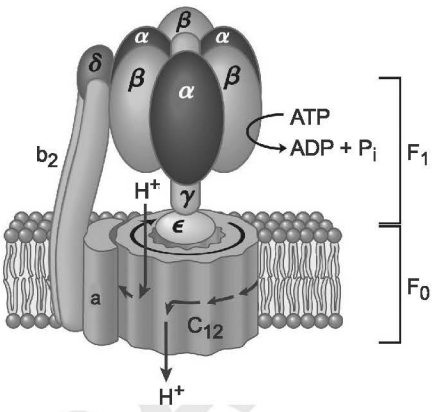


Fig. : Structure of ATP synthase (F_1F_0 ATPase)

Q.7. Describe the chemiosmotic process.

Ans.

Chemiosmotic Coupling Process

The chemiosmotic hypothesis, as proposed in the early 1960s, consisted of four independent postulates. In terms of mitochondrial function, they were as follows :

1. The mitochondrial respiratory chain in the inner membrane is proton translocating; it pumps H^+ out of the matrix space when electrons are transported along the chain.

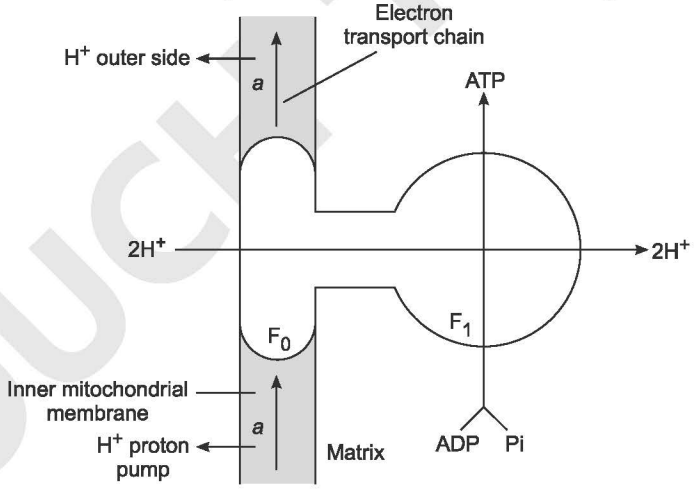


Fig. : Mechanism of oxidative phosphorylation involving the generation of electrochemical proton gradient leading to the synthesis of ATP.

2. The mitochondrial ATP synthase also translocates protons across the inner membrane. Being reversible, it can use the energy of ATP hydrolysis of pump H^+ acorss the membrane, but if a large enough electrochemical proton gradient is present, protons flow in the reverse direction through ATP synthases and drive ATP synthesis.
3. The mitochondrial inner membrane is equipped with a set of carrier proteins that mediate the entry and exit of essential metabolites and selected inorganic ions.
4. The mitochondrial inner membrane is otherwise impermeable to H^+, OH^-, and generally to anions and cations.

Q.8. Describe the biogenesis of ribosome.

Ans.

Biogenesis of Ribosomes

The biogenesis of ribosomes in bacteria (prokaryotic cells) takes place inside the cytoplasm because of the absence of nucleolus. The rRNAs originate from the specific codons of the genome or the ribosomal DNA (rDNA).

In eukaryotic cells the process of biogenesis of ribosomes is complicated. The genes coding for 18S, 5-8S and 28S rRNA are located in the **nucleolar organizer region**. It contains several copies of these rDNA genes which are tandomly repeated in each nucleolar organizer. The rDNA genes are separated by spacer DNA which provides multiple binding sites for specific protein factors. It means 18S, 5.8S and 28S RNAs are synthesised as a single precursor molecule inside the nucleolus as 45S RNA. 5S RNA is synthesised on the chromosomes outside the nucleolus. In Xenopus, 5S RNA genes are located at the telomeres of most chromosomes. The ribosomal proteins are synthesised in the cytoplasm. All these components assemble into ribosomal subunits inside the nucleolus. The subunits are transported to sites of protein synthesis in the cytoplasm.

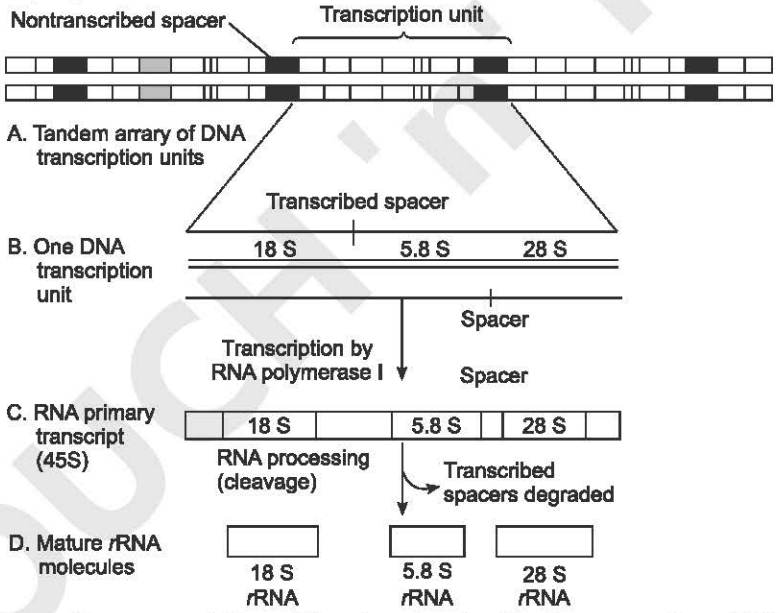


Fig. : Diagrammatic representation of the steps involved in the conversion of 45S nucleolar RNA into 28S and 18S and 5.8S ribosomal RNA.

Q.9. Write differences between EMP, PPP and ED pathways.

Ans.

Differences Between EMP, PPP and ED Pathways

A comparative study of the three systems which operate in respiration would indicate that there is a net gain of 2 ATP molecules in EMP pathway (glycolysis) whereas there is a gain of only one ATP molecule in the other two pathways (Fig.). In all the three pathways it is the oxidation of phosphoglyceraldehyde that leads to the formation of ATP. In glycolysis, one glucose molecule yields two molecules of phosphoglyceraldehyde. In other two pathways, one pair of hydrogen atoms is removed before glucose is split, and, therefore, only one molecule of phosphoglyceraldehyde is formed.

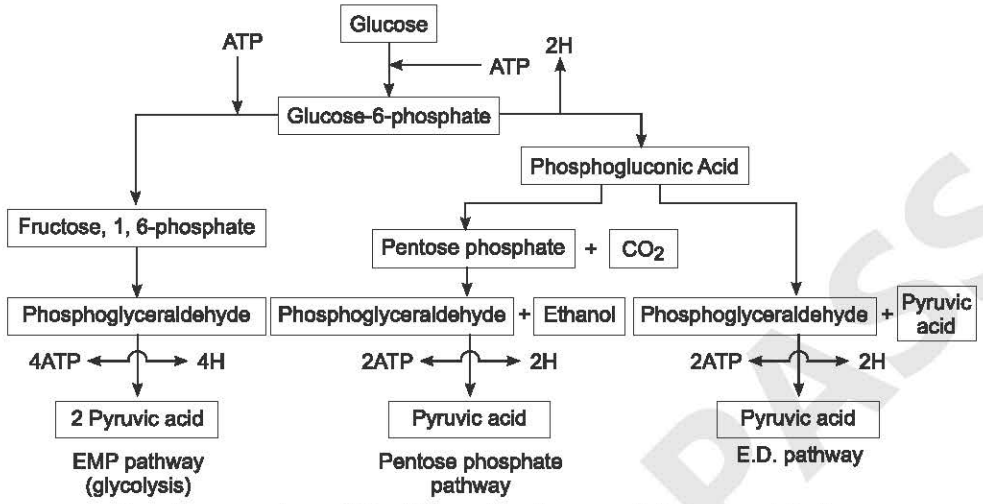


Fig. : A comparison of the different pathways of glucose metabolism.

Q.10. How does glycolysis differ from Krebs cycle?

Ans. **Differences between Glycolysis and Krebs Cycle**

S.No.	Glycolysis	Krebs Cycle
1.	It takes place in the cytoplasm.	It occurs in the mitochondria.
2.	it is a linear pathway.	It is a cyclic pathway.
3.	Substrate used in this pathway is glucose.	Substrate used in this pathway is acetyl coenzyme-A.
4.	End products of glycolysis are two molecules of pyruvic acid, which enter Krebs cycle after oxidative decarboxylation.	End product of Krebs cycle is oxaloacetic acid or oxaloacetate which combines with acetyl CoA to start new Krebs cycle.
5.	It is a common pathway for both aerobic and anaerobic respiration.	If occurs exclusively in aerobic respiration.
6.	Oxygen is not needed during glycolysis.	Oxygen is needed in Krebs cycle.
7.	Generates 4ATP, so there is net gain of 2ATP molecules.	Generates 2GTP molecules from 2 pyruvate molecules.
8.	It consumes 2ATP molecules.	No ATP is consumed.
9.	It does not produce CO_2.	It produces CO_2.
10.	During glycolysis of 1 molecule of glucose 2NAD. H^+ molecules are formed.	During Krebs cycle 6NADH + H^+ and 2$FADH_2$, molecules are formed from two pyruvate molecules.

Q.11. Write a brief account of the lysosomes in plants.

Ans. **Lysosomes In Plants**

Lysosomes similar to those found in animal cells are absent in plant cells, but many membrane bound storage granules are seen to contain hydrolytic enzymes. Following storage granules in plants possess lysosomal enzymes :

1. **Spherosomes :** The spherosomes are spherical particles about. 0.5 to 2.5 μ in diameter. These occur in most plant cells. These originate from ER and contain lipids and proteins. Basically, the spherosomes are associated with the synthesis and storage of lipids. In maize root tip and tobacco endosperm tissue, these are rich in hydrolytic enzymes and are considered to be lysosomes.
2. **Aleurone Grains :** These are also spherical storage particles bounded by unit membrane. These are found in the cells of endosperm and cotyledons of the seeds. These store proteins, specially globulins and phosphate in the form of phytin. In pea seeds the aleurone grains contain a number of hydrolytic enzymes as well. These enzymes are required for the mobilization of stored proteins and phosphates. **Hall** (1974) suggested that these enzymes help to mobilize the reserve food material during germination of seeds.
3. **Vacuoles :** Vacuoles present in plant cells also contain a variety of hydrolytic enzymes and sometimes exhibit polymorphism. These are roughly spherical bodies bounded by a single unit membrane. These are derived from endoplasmic reticulum as small bodies called provacuoles, a number of which fuse to form larger vacuoles.

Q.12. Write short note on polymorphism of lysosomes.

Ans.

Polymorphism of Lysosomes

Lysosomes exhibit considerable degree of polymorphism. It is the result of the association of primary' lysosomes with different materials phagocytised by the cell and the stage or digestion of dissolution of phagocytosed material. At present four types of lysosomes are recognised, of which the first are **primary lysosomes** and other three are **secondary lysosomes**.

1. **Primary Lysosomes or Storage Granules :** These are newly formed lysosomes with hydrolytic enzymes in their original form. Primary lysosomes are also known as **original lysosomes** or **storage granules or inactive lysosomes**.
2. **Heterolysosomes or Digestive Vacuoles or Phagolysosomes :** These lysosomes are formed by the fusion of primary lysosomes with a phagosome or pinosome (a food vacuole containing extracellular substances ingested into the cell by phagocytosis or pinocytosis). Therefore, phagolysosomes contain phagocytised extracellular material and the enzyme contents of lysosome.

   Many primary lysosomes may meet and fuse with secondary lysosome or many secondary lysosomes may also fuse together. Due to hydrolytic action of lysosomal enzymes, phagosome contents are digested and are hydrolysed into products of low molecular weight that pass through the lysosomal membrane and are incorporated into the cell. These lysosomes are also known as *phagolysosomes, heterophagic vacuoles* or *digestive vacuoles.*
3. **Autophagic Vacuoles or Autolysosomes or Cytolysosomes :** Autophagic vacuoles contain lysosomal enzymes and organelles from the cell's own cytoplasm (like mitochondria or portions of rough and smooth ER) in the process of digestion. Autophagic vacuoles bring about autodissolution or autodigestion of cellular organelles. It is of common occurrrence during cell growth and repair. Several autophagic vacuoles are seen in the cells of dedifferentiating tissues and in liver cells during starvation. By this mechanism, cell achieves degradation of its own constituents without irrepairable damage and can remove exhausted or damaged cell components. This process of digesting parts of own cell is known as **autophagy**.

4. **Residual Bodies :** After the process of digestion is completed in secondary lysosomes (phagolysosomes or autolysosomes) some undigested remains are left in them. Such exhausted lysosomes with residues are called **residual bodies** or **telolysosomes** or **dense bodies**. The undigested residues appear as whorls of membranes, grains, amorphous masses or ferritin-like particles. The residual bodies may be extruded from the cell similar to defaecation in *Amoeba* and other unicellular organisms or may accumulate in the cells and may cause ageing.

Q.13. Describe briefly the peroxisome.

Ans.

Peroxisome

The term **peroxisome** was coined by **deDuve** (1969) for those microbodies that are rich in enzymes *peroxidases* and *catalases* and produce hydrogen peroxide during their degradative activity. These are limited by a single membrane and contain fine granular substances that may condense to form an opaque core or nucleoid. The peroxisomes without nucleoid are called microperoxisomes The enzymes present in peroxisomes are: 1. *Uric acid oxidase*, 2. *D-amino acid oxidase* 3. *α-hydrolytic acid oxidase*, 4. *NADH glyoxylate reductase*, 5. *NADP-isocitrate dehydrogenase* and 6. *Catalase*.

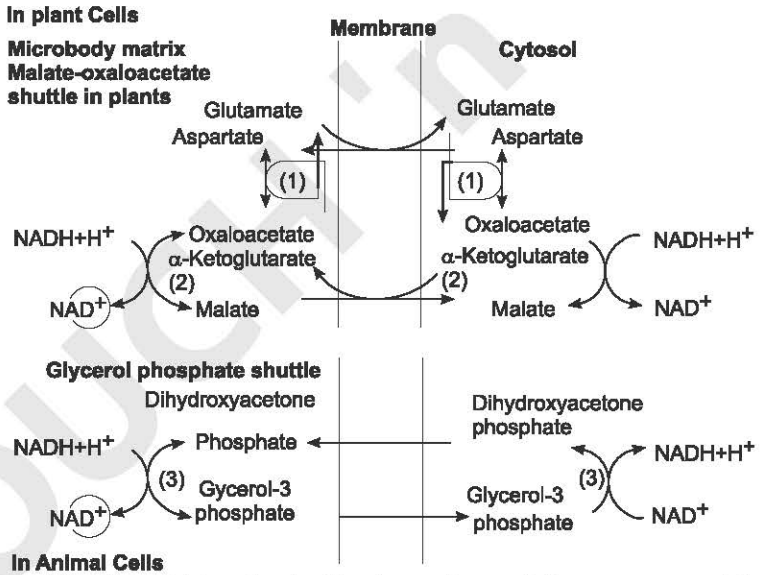


Fig. : The proposed shuttles within microbodies (peroxisomes) that generate oxidized pyrimydine nucleotides. The malate-oxaloacetate shuttle operates in plant peroxisomes and glycerol phosphate shuttle in animal cells.

Formation of Peroxisomes : Peroxisomes generally arise from endoplasmic reticulum. Their enzymes are synthesised by ribosomes and are despatched into the cisternae of ER and from there to the outgrowths which then detach forming peroxisomes.

Functions of Peroxisomes : In green plants peroxisomes carry out photorespiration. The oxidase of peroxisomes transfer hydrogen atoms to molecular oxygen, thereby forming hydrogen peroxide. The H_2O_2 being extremely toxic is degraded into water and oxygen to ***peroxisomal catalase or peroxidase***.

Q.14. Write a brief account of the acrosome development in sperm.

Ans.

Acrosome Development in Sperm

Burgos and **Fawcett** (1966) discovered that the Golgi complex forms **acrosome** during spermiogenesis (maturation of sperm). The acrosome contains hydrolytic enzymes which contribute to the breakdown of protective surfaces of egg.

In early stages of spermiogenesis, Golgi complex is in the form of a spherical body, comprising numerous vesicles or vacuoles in the centre surrounded by several rows of concentrically arranged cisternae. As the development proceeds, the arrangement of cisternae becomes irregular and one or two large coated vacuoles replace the vesicle. Inside each large vacuole there appears a small dense body, **the proacrosomal granule.** If more than one vacuole and proacrosomal granule are left in the end, the vacuole with its granule enlarges in size, migrates towards the anterior pole of nucleus and gets attached to the tip of elongated nucleus. The granule enlarges further and forms the **acrosomal granule.** It forms the core of acrosome. The vacuole loses its liquid contents, spreads over the acrosomal granule and half of the nucleus, and collapses with its wall forming the cap of the spermatozoon.

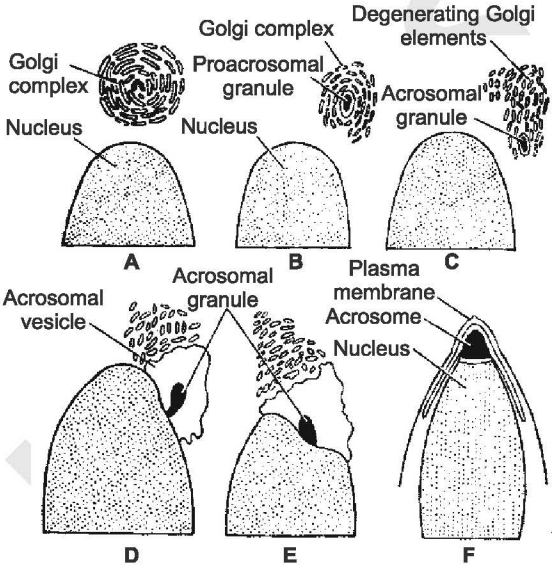


Fig. : Gradual Modification in Golgi Complex to form acrosome during spermiogenesis.

Golgi complex activates mitochondria to produce ATP to be utilised in the respiratory cycle, nervous transmission, and the synthesis of protein and nucleic acids.

All the functions of Golgi complex are closely associated with its secretory function but what is its significance in the nonsecretory cells is still unknown. It is said that Golgi complex also participates in the secretion of fats, elaboration of Nissl granules and metabolism of carbohydrates.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. What do you understand by Cytoskeleton? Write about its functions.

Ans.

Cytoskeleton

An intricate network of protein filaments extends throughout the cytoplasm of all eukaryotic cells. It provides a structural framework for the cell, serves as a scaffold for cell shape and positioning of organelles and is responsible for cell movements. Though, this filamentous network is called **cytoskeleton**, it is much less rigid and permanent. It is a dynamic structure that is continuously reorganized during cell movement and cell division.

Functions of Cytoskeleton

1. **Structural Support:** Cytoskeleton acts as a scaffold, providing structural support that helps maintain the cell shape.

2. **Movement of Substances :** The microfilaments and microfibrils form a part of machinery required for the movement of materials and organelles within the cell, such as movement or transport of vesicles from ER to Golgi to lysosomes, formation of pinocytic or phagocytic vesicles, movement of chromosomes during cell division and movement of neurotransmitter containing vesicles along the length of nerve cell.
3. **Changes in Cell-shape :** Changes in cell shape and movement of cells during embryonic development towards the end of gastrulation is brought about by the contraction of a band of microfilaments. These assemble in the cortical region of cells just beneath the apical cell membrane and their contraction pulls the cell cytoplasm in that direction.
4. **Cell Movement:** Movement of unicellular organisms or cellular locomotion by certain WBCs are dependent on the force generating elements of cytoskeleton.
   (i) **Microtubules and cell movements :** Microtubules form part of mitotic spindle, centrioles and core structure or axoneme of cilia and flagella. Therefore, microtubules are associated with the movement of chromosomes during cell division.
   Microtubules composing axonemes or core of cilia and flagella form the machinery for generating the forces required for locomotion. Cilia and flagella help in swimming of unicellular organisms.
   (ii) **Microfilaments :** These play a key role in all types of contractility and motility within the cell. The contraction of a skeletal muscle fibre results from the sliding of actin-containing thin microfilaments. The contraction and relaxation of muscles help in locomotion and movement.
   (iii) **Non-muscle motility and contractility :** This helps in crawling of cells over a substratum. It is due to the presence of actin fibres. Formation of pseudopodia and amoeboid movement are due to the action of actin microfibrils that are present in the cell cytoplasm.
5. **Internal Frame work :** It forms an internal framework which is responsible for maintaining position of various organelles within the cell.
6. **Signal Transduction :** Cytoskeleton makes contact with the inner surface of plasma membrane and plays a key role in transmitting signals from extracellular environment into the cell interior.
7. **Protein Synthesis :** The translation machinery of the cell remains attached with the cytoskeleton. It means it provides site for anchoring mRNA and ribosomes.

Q.2. What are the main components of cytoskeleton? Describe their structure and functions.

Ans.

Components of Cytoskeleton

The filaments composing cytoskeleton are of following three types :

1. Microfilaments 2. Intermediate filaments 3. Microtubules.

1. Microfilaments

Microfilaments are solid filaments of 7 nm diameter and up to several micrometers long. They are formed of protein **actin**. In presence of ATP, these actin subunits or **G-units**, polymerise in a head to tail fashion to form a flexible filament composed of two strands,of actin molecules wound around each other in a double helix.

Each actin monomer is a globular protein **(G-unit)** formed of 375 amino acids. The filament formed by polymerisation of **G actin** subunits is called **filamentous actin** (Factin). It has distinct polarity with barbed plus end and pointed minus end. The actin polymerisation is **reversible.** The filaments can depolymerise by the dissociation of actin subunits from the ends.

Organisation of Actin Filaments

Individual actin filaments are assembled either into **actin bundles** or into **actin network** by **actin binding domain** (ABD).

1. **Actin Bundles :** In actin bundles the protein filaments are cross-linked by **actin-binding proteins** into closely packed parallel arrays. The actin bundles are of two types and differ in spacing of actin filaments and actin binding proteins :
   (i) **Bundles with closely spaced actin filaments** as in microvilli or other cell surface projections.

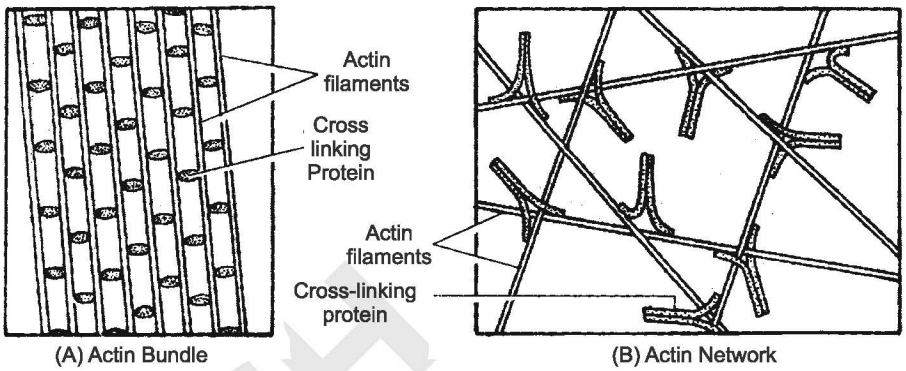


Fig. 1 : Organisation of actin filaments into (A) actin bundle and (B) actin network.

   (ii) **Bundles with widely spaced actin filaments** are called **contractile bundles.** The actin fibres are cross-linked with **α-actinin**. These bundles are more contractile.

2. **Actin Network :** In a network, actin filaments are cross-linked through large and flexible actin binding proteins to form a three dimensional meshwork. It is abundant beneath the plasma membrane and serves following functions :
   (i) Determines cell shape.
   (ii) Allows movement of the cell and change in cell shape.
   (iii) Provides mechanical support.
   (iv) Enables the cell to migrate, engulf particles (phagocytosis) and the cell to divide.

Functions of Actin Filaments

The actin filaments perform following functions :

1. Actin filaments form cytoskeleton of cell. They determine cell shape and provide mechanical support.
2. Actin filaments support permanent protrusions of the cell surface, such as microvilli and stereocilia which help in absorption and hearing respectively.

3. Actin filaments support transient surface protrusion like pseudopodia, filopodia or lamellipodia.
4. Actin bundles attach to the plasma membrane by spectrin fibres and provide cell-cell adhesion and cell-substratum contact.
5. Actin-Myosin filaments help in muscle contraction. Myosin acts as a motor protein that uses energy from ATP and generates force and movement.
6. Contractile assemblies of actin and myosin in non-muscle cells produce a variety of movements and are responsible for cytokinesis during cell division.

2. Intermediate Filaments (IFs)

These are tough, solid, smooth surfaced unbranched filaments having a diameter of approximately 10 nm. These are composed of more than 65 types of proteins. They are intermediate in diameter between microfilaments and microtubules. The different polypeptide subunits of intermediate filaments have a similar arrangement of domains and thus have similar appearance.

Intermediate Filaments : The proteins forming intermediate filaments contain a rod-shaped **central domain** formed of approximately 310 amino acids, the N-terminal **head** and C-terminal **tail**. The central domains of two polypeptides wound around each other in a coiled structure to form the **dimers**. The two dimers then associate in antiparallel fashion to form **tetramers**. The tetramers are associated end-to-end to form **protofilaments**. Approximately eight protofilaments are wound each other to form a rope-like **intermediate filament**.

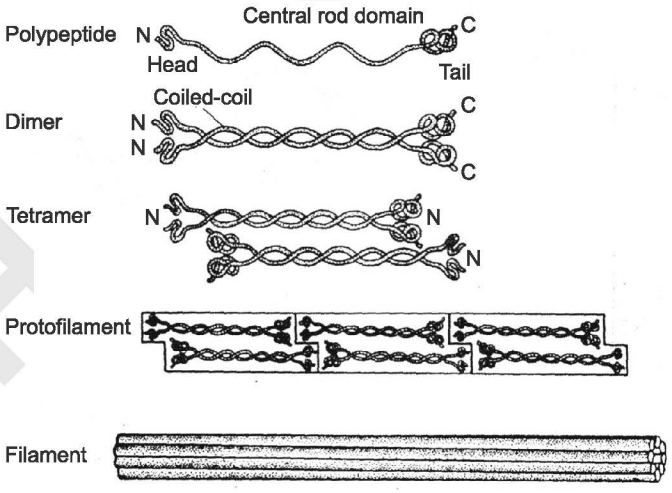


Fig. 2 : Assembly of intermediate filaments.

IFs are highly resistant to tensile force and are more stable to chemical disruption. These are formed of different types of keratin proteins.

Intracellular Organisation of Intermediate Filaments

The intermediate filaments ramify through the cytoplasm and are connected with the microfilaments and microtubules. They are found in a variety of animal cells but are absent from plant cells. They form a ring surrounding the nucleus to the plasma membrane.

Functions of Intermediate Filaments

IFs provide mechanical stability to cells and also perform specialised tissue specific functions as follows :

1. The keratin filaments that attach to the nuclear envelope anchor the nucleus in position within the cell.

2. IFs provide a scaffold that integrates the components of cytoskeleton and organises the internal structure of cell.
3. Neurofilaments present in neurons and their axons provide mechanical support.
4. Desmins in muscle cells connect individual actin-myosin assemblies to one another and to plasma membrane.
5. Keratin filaments in epithelial cells are tightly attached to desmosomes and hemidesmosomes and serve as a mechanical link between adjacent cells.

3. Microtubules

Microtubules are hollow, rigid, cylindrical structures which occur in almost all the eukaryotic cells except mammalian RBCs. These are components of a variety of cell structures. Besides forming skeleton, these are found in the core of cilia and flagella, in the mitotic spindle of dividing cells and in the centrioles and basal bodies.

Structure of Microtubules

Microtubules have a diameter of about 25 nm (i.e., 250 Å) and a wall of approximately 5 nm thickness. These are formed of only one type of protein, called **tubulin**. The wall of microtubules is formed of 13 rows of globular subunits. Each globular subunit consists of a single molecule of protein, **tubulin**, which is a dimer being formed of two globular polypeptides-α **tubulin** and β **tubulin**.

The subunits in the wall of microtubule are arranged in longitudinal rows, called **protofilaments**. The protofilaments are arranged helically around a central axis and are aligned parallel to the long axis.

Microtubules are polar structures. Their one end is called **plus end** and other as the **minus end**. New subunits are added to the microtubules at the plus end. Some **microtubule associated proteins (MAPs)** are required during polymerisation of microtubules. Some MAPs stabilize microtubules by capping their ends, while others disassemble microtubules by depolymerisation at the ends.

Functions of Microtubules

Microtubules carry out following functions :

1. Microtubules act as internal skeleton or scaffold that provides structural support and help maintain the position of cytoplasmic organelles.
2. They form part of machinery that moves materials and organelles from one part of a cell to another.
3. They form components of the machinery responsible for chromosome movement during mitosis and meiosis.
4. They form motile elements of cilia and flagella. Both of them are microtubule based extensions of plasma membrane. Their movements result from the sliding of microtubules due to the action of **dynein motors**.

Q.3. What do you understand by Golgi Complex? Describe the morphology, chemical composition and functions of Golgi Complex.

Ans.

Golgi Complex

Golgi Complex, also called Golgi apparatus, is a differentiated or specialised part of endomembrane system found in the cells of both plants and animals. It is mainly associated with

cell secretion, is often referred as **'traffic police'** of the cell because it plays a key role in sorting out cell's proteins and membrane constituents and in directing them to their proper destinations.

Camillo Golgi (1898) while studying nerve cells of barn owl by special staining with silver impregnation method described a reticular structure. This was named **Golgi Complex** after its discoverer **Golgi. Gatenby** (1917) pointed out similarity between Golgi apparatus of vertebrates and dictyosomes of invertebrates. **Perner** (1958) observed Golgi elements in plants and these were termed as **dictyosomes**.

Morphology of Golgi Complex

The number and size of dictyosomes varies from one cell type to other and also on the metabolic activity of the cell. It increases when the cell is actively secreting material. In plant cells, their number increases during cell division for the formation of cell wall.

Golgi complex is surrounded by a zone of clear cytoplasm which is without ribosomes, mitochondria and glycogen. This is called **zone of exclusion**. Morphologically, the Golgi complex appears as a shallow, saucer-shaped structure consisting of interconnecetd **tubules, vesicles, cisternae** and **Golgian vacuoles**.

1. **Flattened Sacs or Cisternae :** Cisternae form the plate-like central part of Golgi complex. It comprises tubular or flattened fluid-filled compartments about 20Å wide and enclosed by unit membrane. These are stacked in parallel bundles one above the other and are separated by intercisternal space of about 20-30μ (200-300Å). Their number varies from 3-12. The cisternae are somewhat crescentic and are arranged concentrically with convex surface towards the nuclear envelope or ER and concave surface towards the plasma membrane.

   The cisternae in a stack are arranged in a specific order. Those on the convex side are small. This side represents the **forming face** of Golgi apparatus. The transition vesicles and tubules that detach from E.R. fuse here to form new

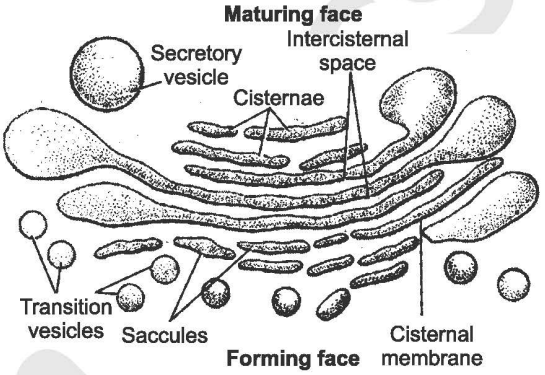


Fig. 1 : Ultra structure of Golgi Complex

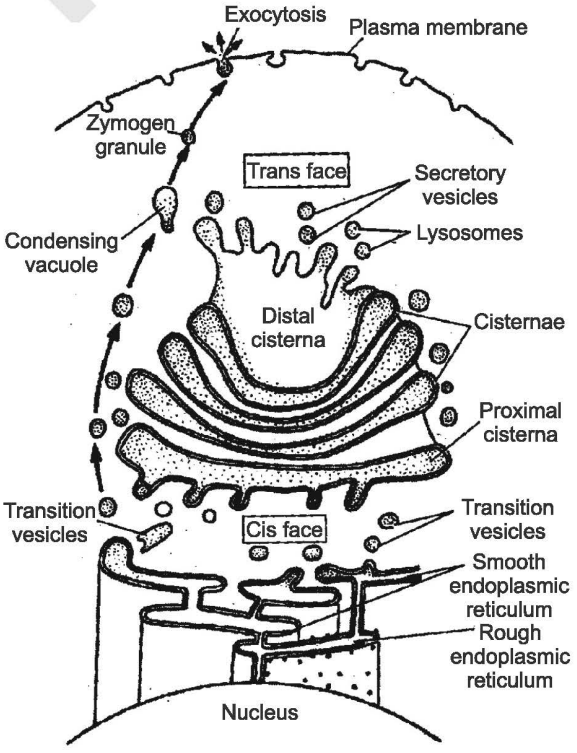


Fig. 2 : Relationship between different components of Golgi complex and their relation with secretion.

cisternae. The opposite concave face of cisternal sack represents the maturing face. It is associated with the secretory vesicles and vacuoles. These are formed by the dilation of edges of cisternae due to accumulation of secretory products formed by the concentration in Golgi cisternae. These vesicles coalesce and finally form large vacuolar structures that are called zymogen granules or lysosomes.

2. **Secretory Vesicles :** These are present on the sides and on the maturing face of Golgi. These are pinched off from the trans face of Golgi cisternae. These contain secretory products of Golgi and are finally converted into zymogen granules or lysosomes.
3. **Transition Vesicles :** These are small drop-like structures about 400A-800Å in diameter. These are associated with the convex forming face of Golgi cisternae and lie next to smooth ER. These develop from the cisternae of ER as bleb and coalesce to form new Golgi cisternae. This region represents **zone of exclusion**, a region of transition from ER to Golgi cisternae.
4. **Coated Vesicles :** In some cell types, the secretory vesicles are coated with bristly layer of protein clathrin. These are called coated vesicles. These are associated with the secretion of highly specialised cell products.
5. **Golgi vacuoles :** These are large rounded sacs present on the maturing face of Golgi. These are formed either by the expanded cisternal or by the fusion of secretory vesicles. The vacuoles are filled with some amorphous or granular substance.

Chemical Composition

Golgi membranes are rich in the following chemicals :

1. **Phospholipids :** Phospholipid composition of Golgi membranes is intermediate between those of endoplasmic membranes and plasma membranes.
2. **Carbohydrates :** Both plant and animal cells have some common carbohydrate components, like **glucosamine, galactose, glucose, mannose** and **fucose**. Plant Golgi lack **sialic acid**, but it occurs in high concentration in rat liver. Some carbohydrates like **xylose** and **arabinose** are present in plant cells only.
3. **Enzymes :** Golgi complex from different plant and animal cells show variations in protein and enzyme contents. Some of the enzymes are ADPase, ATPase, CTPase, NADPH, cytochrome C-reductase, several glycosyl transferases, galactosyl transferase, thiamine pyrophosphate, etc.

Functions of Golgi Complex

The Golgi complex performs many different functions in a variety of cells. However, all of them are associated with secretion activity of the Golgi.

Golgi is associated with transportation of substances. It also carries out post-translation secretion modification of the proteins during glycoprotein synthesis. Some of these given functions have been following :

1. It secretes certain enzymes.
2. Formation of acros somes at the time of spermatogenesis.
3. Activates mitochondria to produce ATP.
4. Absorbs certain inorganic elements like Fe, Cu, Au, Bi and organic compounds including sugar and certain toxic substances.

5. Stimulates endocrine glands to secrete their respective hormones.
6. Its vacuoles and vesicles are able to store protein and lipid.
7. Also forms lysosomes.
8. Helps in the formation of cell wall and produces gum and mucilage as *Drosera*.

Q.4. Write shorte note on EMP pathway.

Ans. **Glycolysis or Embden-Meyerh of Pathway**

Initial steps, known as **glycolysis**, are common in both aerobic and anaerobic respiration. Therefore, glycolysis is called the **common pathway** for both the types of respiration.

Site of Occurrence : Glycolysis occurs in cytoplasm resulting in the break down of glucose into pyruvic acid, a three carbon compound. This process does not require oxygen.

Steps in Glycolysis

During glycolysis, a glucose molecule breaks down into two molecules of **Pyruvic acid** and releases energy which is entrapped in two molecules of ATP. It occurs outside mitochondria in the cytoplasm. It involves several steps, each step catalysed by a specific enzyme. These steps have been worked out by German biochemists, **Embden** and **Meyerhof** and the pathway is also called **Embden-Meyerhof Pathway**.

About 80% to 90% of the glucose metabolized is converted to pyruvic and lactic acids mainly in liver and muscles. During *glycolysis*, it has been shown how glucose-6-phosphate is formed in the muscles from glucose, The next step involves the interconversion of glucose-6-phosphate to fructose-6-phosphate.

1. **Interconversion of glucose-6-phosphate and fructose-6-phosphate :** Glucose-6-phosphate and fructose-6-phosphate are freely interconvertible. The reaction is catalyzed by the enzyme *phosphoglucose isomerase*. At equilibrium, however, glucdse-6-phosphate predominates, having a concentration of twice that of fructose-6 phosphate.

$$\text{Glucose-6- Phosphate} \xrightarrow[\text{phosphate isomerase}]{\text{Glucose}} \text{Fructose-6-Phosphate}$$

$$\text{Fructose-6-Phosphate} \xrightarrow[Mg^{++}, \text{ATP}]{\text{Phosphofructokinase}} \text{Fuctose-1, 6, diphosphate}$$

2. **Conversion of fructose-6-phosphate to fructose-1, 6-diphosphate :** Fructose-6-phosphate is next phosphorylated to form fructose-1, 6-diphosphate by the enzyme *phosphofructokinase* in the presence of Mg^{++} and ATP. The diphosphate is called Harden-Young ester. The reaction is irreversible as the energy exchange is about 4500 calories.
3. **Formation of triose phosphates :** The next step is the splitting of fructose-1, 6-diphosphate by the enzyme *aldolase* to form D-glyceraldehyde-3-phosphate and dihydroxyacetone phosphate. This is a reversible reaction.

$$\text{Fuctose-1 6-diphosphate} \xrightarrow{\text{Aldolase}} \text{Glyceraldehyde-3-phosphate} + \text{Dihydroxyacetone phosphate}$$

The two triose monophosphates are freely interconverted by *triose phosphate isomerase*.

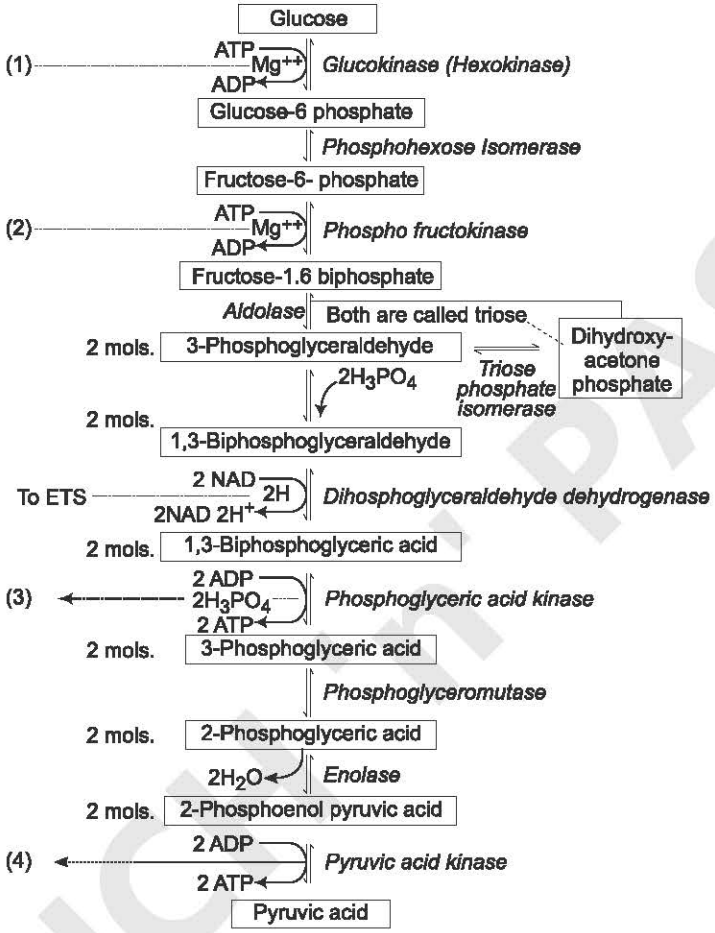


Fig. : Various steps in glycolysis : Steps (1) and (2) represent location of ATP consumption and steps (3) and (4) represent location of ATP generation.

4. **Oxidation of glyceraldehyde 3-phosphate :** The next step in glycolysis is the phosphorylation and oxidation of the two molecules of D-glyceraldehyde-3-phosphate to two molecules of 1, 3-diphosphoglyceric acid, catalyzed by the enzyme *phosphoglyceraldehyde dehydrogenase,* DPN^+ and inorganic phosphate are required. 1, 3-diphosphoglyceric aldehyde is the possible intermediate compound.

$$\text{Glyceraldehyde-3-phosphate} + H_3PO_4 + DPN \rightleftharpoons$$

$$\text{1,3-diphosphoglyceric acid} + DPNH + H^+$$

Transphosphorylation of 1, 3-diphosphoglyceric acid. 1, 3-diphosphoglyceric acid contains a high energy bond; and in the presence of an acceptor, ADP, magnesium ions, and the enzyme phosphoglyceric acid kinase, 3-phosphoglyceric acid and ATP (energy) are formed.

$$\text{1-3 biphosphyoglucerate} + ADP \rightleftharpoons \text{3-phosphoglycerate} + ATP$$

5. **Recovery of phosphate and formation of pyruvic acid :** The next stage involves the recovery of phosphate groups from 3-phosphoglyceric acid, which is converted initially to 2-phosphoglyceric acid by the action of *phosphoglyceromutase.*

$$\text{3, phosphoglyceric acid} \xrightleftharpoons{\text{Phosphoglyceromutase}} \text{2, phosphoglyceric acid}$$

2-phosphoglyceric acid is converted to phosphoenol-pyruvic acid by dehydration in the presence of an enzyme *enolase.* This molecule contains a higher-energy phosphate bond. 2-phosphopyruvic acid then loses its phosphoric acid releasing energy and forming pyruvic acid, which is finally reduced by *lactate dehydrogenase* to form lactic acid.

$$\text{2-phoshoglycerate} \xrightleftharpoons{\text{Enlase}} \text{Phosphoenol pyruvate} + H_2O$$

$$\text{Phosphoenol pyruvate} + \text{ADP} \xrightleftharpoons{\text{Pyruvate kinase}} \text{Pyruvate} + \text{ATP}$$

Significances of Glycolysis

1. Break down of 6-carbon compound (glucose) to two molecules of trioses (pyruvic acid).
2. A total 4 ATP molecules are generated at two different steps. Two ATP are used up during phosphorylation reactions, so there is net gain of 2 ATP molecules during glycolysis of one molecule of glucose.
3. Two molecules of NAD^+ are reduced to two molecules of $NADH^+$ which are subsequently oxidised through electron transport chain to yield 6 molecules of ATP.

Q.5. Write short note on TCA cycle.

Ans. Krebs Cycle or Citric Acid or Tricarboxylic Acid Cycle

The respiratory cycle which takes place in mitochondria in the presence of oxygen is known as **Krebs cycle** after the name of an English biochemist, **Hans A. Krebs** who discovered its various steps in 1937 and received Nobel Prize in 1953 for this work. This cycle is also known as **citric acid cycle** because of the formation of citric acid at the first step. It is called **tricarboxylic acid cycle** (TCA) because many intermediate compounds formed in the cycle have three carboxyl groups. This cycle is confined to the mitochondria.

Steps : TCA cycle involves following steps :

1. **Condensation:** The first reaction of Krebs cycle is the condensation of acetyl CoA with oxaloacetic acid to form **citric acid** in the presence of water and enzyme *citrate synthase.*

$$\text{Acetyl CoA} + \text{Oxaloacetic acid} + H_2O \xrightarrow[\textit{Synthase}]{\textit{Citrate}} \textit{Citric acid} + \text{CoA}$$

2. **Reorganisation:** In the presence of enzyme *aconitase* citric acid (6C) changes into 6-carbon *cis*-aconitic acid by losing one molecule of water.

$$\text{Citric acid} \xrightarrow{\textit{Aconitase}} \textit{Cis}\text{-aconitic acid} + H_2O$$

3. **Reorganisation:** In this step, *cis*-aconitic acid changes to iso-citric acid in the presence of enzyme *aconitase* with the addition of water.

$$\textit{cis}\text{-aconitic acid} + H_2O \xrightarrow{\textit{Aconitase}} \text{Isocitric acid}$$

4. **Dehydrogenation-I and Decarboxylation-I : Stage (a):** This is the first oxidation step of Krebs cycle. In this step, two hydrogen ions (protons) are removed (oxidation) from isocitric acid and oxalosuccinic acid is formed in the presence to enzyme ***isocitrate dehydrogenase***. Hydrogen ions are received by NAD^+ forming $NADH + H^+$.

   Stage (b): In this step, decarboxylation of oxalosuccinic acid takes place in the presence of enzyme *carboxylase* and α-ketoglutaric acid (5C) is formed. During this process CO_2 is released.

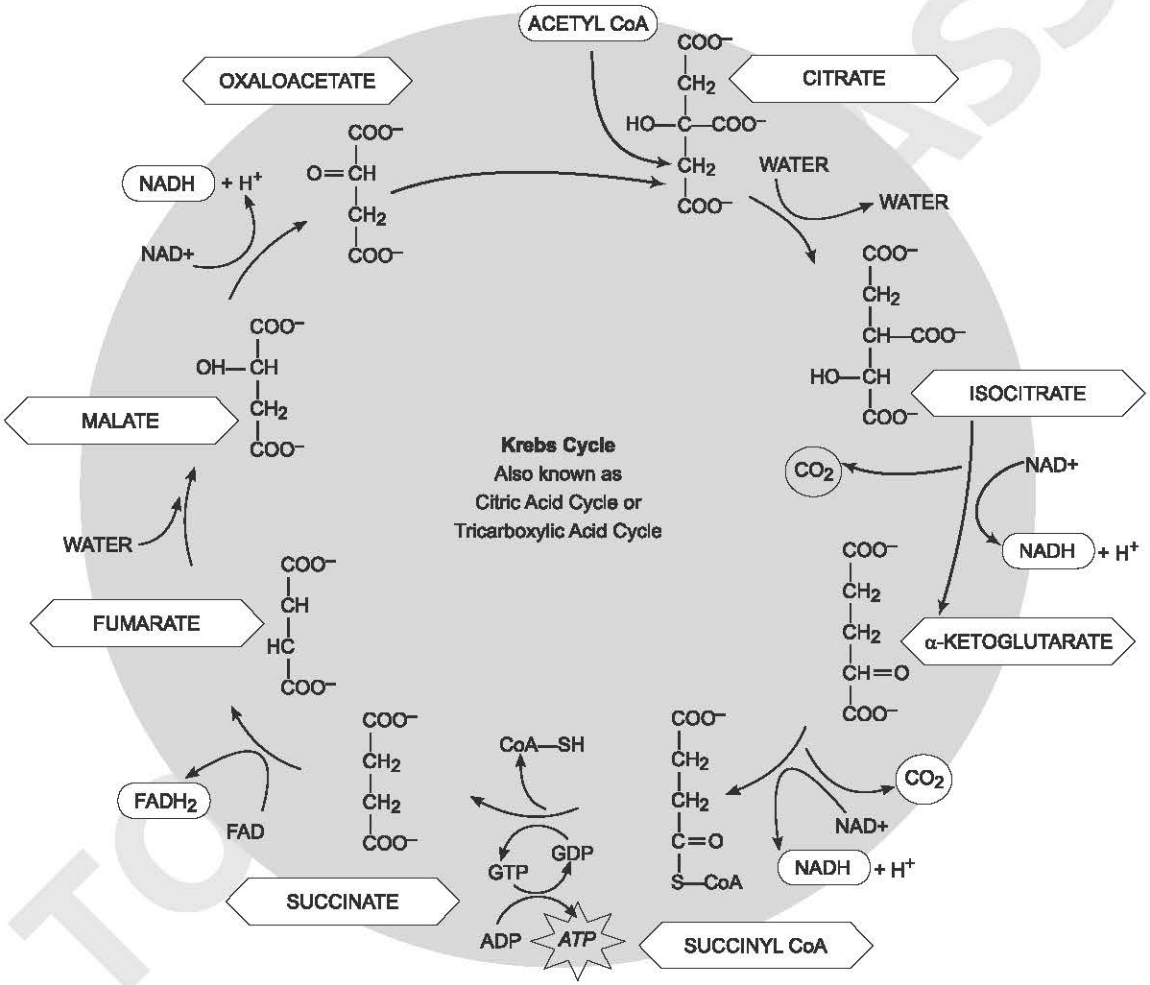


Fig. : The citric acid, or tricarboxylic acid (TCA) cycle.

5. **Dehydrogenation-II and Decarboxylation-II :** This is the second oxidation step of Krebs cycle. This is a two stage process :

   Stage 1. α-ketoglutaric acid reacts with coenzyme-A forming 4-carbon **succinyl coenzyme A**. The reaction is catalysed by α-*ketoglutaric acid* (α-*ketoglutarate*) *dehydrogenase* enzyme. During the reaction one molecule of CO_2 and a pair of hydrogen atoms are released. Hydrogen atoms are taken by NAD^+ and $NADH + H^+$ is formed.

$$\alpha\text{-Ketoglutaric acid} + NAD^+ + \text{Coenzyme A} \xrightarrow[dehydrogenase]{\alpha-ketoglutarate\ acid} \text{Succinyl-CoA} + CO_2 + NADH + H^+$$

Stage 2. Succinyl-CoA is acted upon by enzyme *succinyl thiokinase* or succinate thiokinase to form succinic acid. The reaction releases sufficient energy to form GTP (guanosine triphosphate) from GDP (guanosine diphosphate).

$$\text{Succinyl-CoA} + GDP + H_3PO_4 \xrightarrow[thiokinase]{Succinyl} \text{Succinic acid} + GTP + \text{Coenzyme A}$$

GTP formed in his reaction gives rise to ATP as follows :

$$GTP + ADP \longrightarrow GDP + ATP$$

6. **Dehydrogenation-III:** Succinic acid is dehydrogenated to 4-carbon **fumaric acid** with the action of enzyme *succinic acid dehydrogenase* or *succinate dehydrogenase* and 2 hydrogen atoms are released. Hydrogen atoms received by FAD (flavin adenine dinucleotide) which is reduced to $FADH_2$. It passes hydrogen atoms to ETS.

$$\text{Succinic acid} + FAD \xrightarrow[dehydrogenase]{succinic\ acid} \text{Fumaric acid} + FADH_2$$

7. **Hydration-II:** Fumaric acid changes to 4-carbon malic acid by enzyme *fumarase*.

$$\text{Fumaric acid} + H_2O \xrightarrow{Fumarase} \text{Malic acid}$$

8. **Dehydrogenation-IV:** Finally, malic acid is oxidised to **oxaloacetic acid** in the presence of NAD^+ and enzyme *malic acid dehydrogenase* or *malate dehydrogenase*. During the reaction a pair of hydrogen atoms are released which form $NADH + H^+$ from NAD^+

$$\text{Malic acid} + NAD^+ \xrightarrow[dehydrogenase]{Malic\ acid} \text{Oxaloacetic acid} + NADH + H^+$$

Thus, oxaloacetic acid is regenerated at the end of Krebs cycle, which recombines with acetyl coenzyme A to form citric acid for restarting Krebs cycle.

During one Krebs cycle, complete oxidation of one molecule of Acetyl CoA releases 2 molecules of CO_2. In the four oxidation steps of Krebs cycle, four pairs of hydrogen ions and electrons are removed from intermediates of the cycle. Of these, three are utilised in the reduction of NAD^+ to $NADH + H^+$ and the fourth in the reduction of FAD to $FADH_2$.

Significance of TCA cycle

This cycle serves as a common oxidative pathway for carbohydrates, fats and proteins. The end products of glucose and amino acid changes to acetyl coenzyme A to enter Krebs cycle, whereas β-oxidation of fatty acid produces acetyl CoA as the end product. Some *intermediates of the TCA* cycle are used in synthesising important biomolecules such as glutamate and aspartate.

Q.6. Describe briefly the structure and functions of mitochondria.

Ans.

Mitochondria

It is generally called as the *'Power House'* of the cell. The presence of mitochondria was first described by **Altmann** (1894) and he used the term *bioplast* for it. **Flemming** (1894) called them **'Filial'**. Petzen gave the name **Sarcosome** to this organelle. **Benda** (1897-98) finally coined the term *Mitochondria*.

Shape and size : Shape is variable, it is dependent on the environment or physiological conditions. Generally it is filamentous or swollen at one end to become club shaped or hollow tennis racket-type. According to Green they are 1500Å in length and in width.

The mitochondria can quickly change shape and move around the cell when needed. The mitochondria can reproduce by growing larger and dividing when the cell needed more energy.

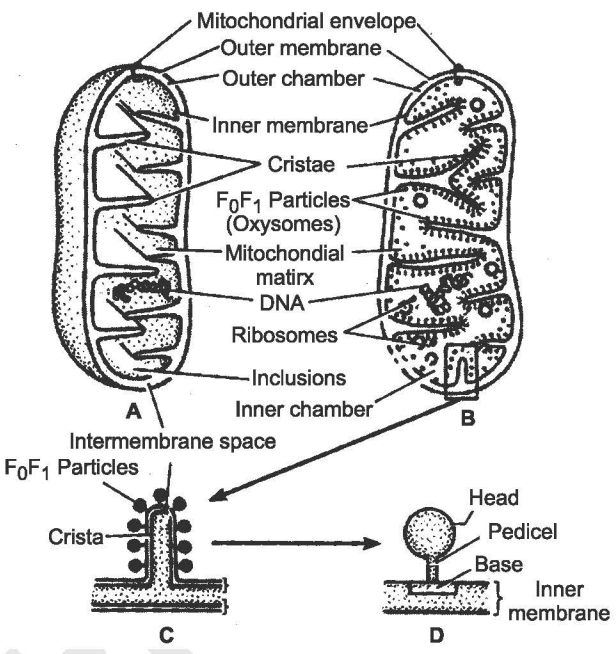


Fig. : Section of mitochondrion showing membranes, chambers and the cristae.

Structure

Mitochondrion has a double layered wrapper with an inner membrane. Each membrane is typical unit membrane being about 50 Å – 70 Å thick. Two membranes are normally 60 Å to 100Å apart. Inner membrane shows a number of inpushings called as the *cristae*. There are two cavities (compartments), outer cavity filled with a fluid rich in coenzymes in between the two membranes and the inner compartment filled with mitochondrial matrix. In this matrix granules of 300 Å – 500 Å are found suspended.

Mitochondrial Chambers :

1. **Outer chamber** is the **intermembranous** or **perichondrial space** between the outer and inner membranes of mitochondria which also extends into the core of crests. It is about 60Å-80Å and is filled with a fluid of low density.

   Because of the highly permeable nature of outer mitochondrial membrane composition of fluid in outer chamber of mitochondria is similar to cytosol.

2. **The inner chamber** is the space enclosed within the inner membrane. It is a wide space and is filled with a homogeneous **mitochondrial matrix.**

Matrix contains high concentration of soluble proteins, some lipids, circular DNA, 70S ribosomes and a few fine filaments or granules. The granules are sites for binding bivalent cations of Mg^{2+} and Ca^{2+}.

According to **Prachy** (1962) these granules are the sites for binding the divalent cations [Ca^{++},Mg^{++}].

Electron microscopy revealed the presence of *oxysomes* outside the outer membrane and inside of the inner membrane. The inner membrane oxysomes comprise of a base, stalk and knob (100 Å). In outer particles stalk is absent.

Chemical Composition

The major constituents of mitochondria are proteins (4/5) and lipids (1/5). The lipid content is composed of 90 per cent phospholipids, (lecithin and cephalin), about 5 per cent free fatty acids and triglycerides. Mitochondria are rich in enzymes related to substrate oxidation, respiration, energy conservation, oxidative phosphorylation and Kreb's cycle. A heterogeneous groups of enzymes related to phospholipid metabolism and fatty acid oxidation are present in outer compartment and outer membrane. Cu^{++}, Mg^{++}, HPO_4^-, HCO_3^-, Cl^-, RNA and DNA (without histone, a protein) are also reported to occur.

Functions of Mitochondria

1. As a power house of the cell is provides energy. Because mitochondria synthesise energy rich compound ATP.
2. Cation accumulation.
3. The oxidation of carbohydrates and fates, large amount of energy is released which is utilised by the mitochondria for synthesis of energy rich compound known as ATP.
4. Formation of RNA and DNA (Mitochondrial DNA and RNA) hence also accounts for the "Cytoplasmic inheritance".
5. The mitochondria are site of oxidation, dehydrogenation, oxidative phosphorylation and respiratory chain of the cell.

Q.7. Describe the structure and functions of ribosomes.

Ans.

Ribosome

Ribosomes, the **'protein factories'** of the cell, are submicroscopic particles formed of ribonucleo-protein. These are found in all the cells. The ribosomes were first observed under electron microscope by **Claude** (1941) and were named **microsomes**. **Robinson** and **Brown** (1953) first noticed them in bean roots and **Palade** (1955) detected them in animal cells and named them **ribosomes**.

Structure of Ribosomes

The ribosome structure is determined by the speed with which they sediment in a centrifugal field. The **'svedberg unit' - 'S'** is used to measure the sedimentation rate. Ribosomes from prokaryotes are small and exist as 70S units and ribosomes from eukaryotes exist as 80S units. A ribosome is formed of two subunits : **a large subunit** and **a small subunit**.

1. Prokaryotic Ribosomes (70S Ribosomes)

Ribosomes of prokaryotes (*E.coli*) have sedimentation coefficient 70S, diameter about 18 nm and 2.8 million daltons particle weight. Their two subunits are **50S** and **30S. Lake** (1985) gave the structural model for prokaryotic ribosome.

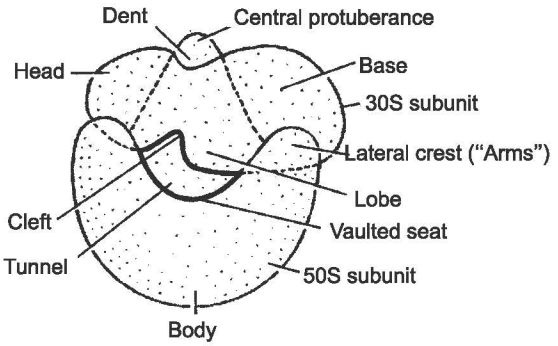


Fig. 1 : Ultrastructure of a prokaryotic ribosome.

Lake's Model of 70S Prokaryotic Ribosomes :

1. **30S subunit :** The small subunit of prokaryotic ribosome is 30S. It is asymmetrical and rod-like. It is partially divided into two lobes by a deep transverse cleft or groove. The smaller segment is called **head** and larger one **base**. A small outgrowth arises from the base segment and is called **platform.**

   The particle weight of 30S subunit is about 1.0 million daltons. It is formed of one molecule of **16S rRNA** and **21 proteins**. 16S rRNA has 1600 nucleotides.

2. **50S subunit :** It is the large subunit. It is more or less spherical and forms the **body.** At its flat anterior end are present three projections-one central projection and two lateral projections.

   The particle weight of 50S subunits is 1.8 million daltons. It is formed of one molecule of **23S rRNA,** one molecule of **5S rRNA** and **34 proteins**. The 23S rRNA has about 3200 nucleotides and 5s rRNA is formed of 120 nucleotides.

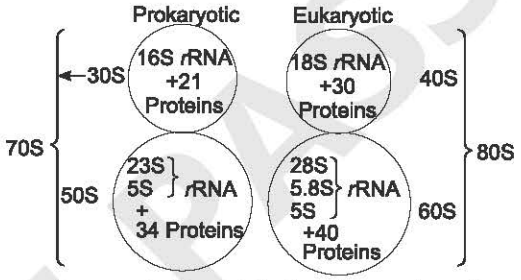


Fig. 2 : Lake's model of prokaryotic and eukaryotic ribosomes.

   In a ribosome, the two subunits are fitted together in such a way that a tunnel is formed between them. During protein synthesis *m*RNA is threaded through this tunnel.

2. Eukaryotic Ribosomes (80S Ribosomes)

The 80S ribosomes have a diameter about 20-22 nm and particle weight 4.2 million daltons. Their two subunits are **60S** and **40S.**

1. The **60S subunit** has a weight about 2.7 million daltons. It contains 28S, 5S and 5.8S **rRNA** and about forty different proteins. It has a broad anterior flat surface and three projections.
2. The **40S subunit** is about 1.5-1.8 million daltons in weight. It has 18 rRNA and 30 different polypeptides.

3. Mitochondrial and Chloroplast Ribosomes

The mitochondrial ribosomes (mitoribosomes) of animals have sedimentation coefficient **55S-60S.** Their large subunit is 40S-45S and small subunit 30S-35S. In fungi **(Ascomycetes)** the mitochondrial ribosomes are 70S-75S with 55S and 30S-40S subunits. In ciliated protozoans these are 80S with each unit of 55S. In higher plants these ribosomes are 77S-80S with subunits 60S and 40S.

Chemical Composition

Ribosomes are formed of approximately equal amount of **RNA** and **proteins.** Proteins form the periphery and RNA lies in the interior remaining interwined within the two subunits.

1. Ribosomal RNA

(i) In prokaryotic ribosomes ribosomal RNA (rNA) occurs in three different forms as 23S rRNA, 16S rRNA and 5S rRNA.

(a) **16S rRNA** lies in 30S subunit. It has a molecular weight 0.6×10^6 daltons and contains about 1,600 nucleotides.

(b) **5S rRNA** also lies in large subunit. It has a molecular weight 3.2×10^4 daltons having 120 nucleotides.

(c) The **23S rRNA** occurs in 50S subunit. It has a molecular weight 1.2 million (*i.e.*, 1.2×10^6) daltons and contains about 3,200 nucleotides.

(ii) **In eukaryotic ribosomes** the ribosomal RNA occurs in four different forms-28S, 18S, 5.8S and 5S.

(a) The **18S ribosomal RNA** is found in 40S subunit and weighs 0.8×10^6 daltons.

(b) **5S** and **5.8S ribosomal RNA** are also found in large subunit and have molecular weight 3.2×10^4 and 5×10^4 daltons.

(c) **28S rRNA** lies in 60S subunit. It has a molecular weight 1.5 – 1.8 million, *i.e.*, $1.5-1.8\times10^6$ daltons.

28S, 5.8S and 18S rRNAs are synthesised in the nucleolus by cleavage of a single precursor RNA, while 5S RNA is synthesised outside nucleolus.

70% of Ribosomal RNA is in the form of double-stranded helical structure due to base-pairing. These helices appear as **'hairpin loops'**. The various ribosomal proteins adhere at specific points on these loops.

2. Ribosomal Proteins

The protein contents of ribosomes are highly complex. Near about 50-55 proteins have been isolated. Of them about 21 proteins occur in small 30S subunit and about 34 proteins are found in the large 50S subunit. These proteins are called **core proteins** or **primary binding proteins**. These specifically and strictly bind to rRNA. When the subunits of ribosomes dissociate into inactive core proteins and RNA, several other proteins are also released from each subunit. These are known as **split proteins (sp)** or **secondary binding proteins.** The split proteins are of SP50 and SP30 types. The core particles are formed of RNA and some proteins. The SP50 and SP30 can be further fractionated into acidic proteins and basic proteins.

SP 50 proteins are indispensable for polypeptide synthesis. 50S proteins bind to tRNA for amino acid incorporation. About 33 different 50S proteins have been identified so far. 30S proteins are of about 20 different types and have special electrophoretic properties. These proteins are coded by different DNA cistrons.

Functions of Ribosomes

Ribosomes are described as the **'protein factories of the cell'** since these are the only site where different components involved in protein synthesis come together. Interaction of tRNA, amino acids and mRNA ensures the linking together of amino acids in a definite sequence to form polypeptide chains and finally the synthesis of protein.

❑

UNIT-III

Nucleus and Chromatin Structure

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Write a note on bacterial and plasmid genome.

Ans. A plasmid is a small, obtain circular DNA molecule found in bacteria and other cells. Plasmids are separate from the bacterial chromosome and replicate independently of it. They generally carry only a small number of gene, notably some associated with antibiotic resistance.

Q.2. Describe the small nucleolar RNAs.

Ans. During the last more than a decade, the list of snoRNAs has grown to over 50, of which, seven snoRNAs (U3, U7, U14, U22, SnR10, SnR30, RNA component of RNAase MRP) are known to be required in the processing of pre-rRNA. Of these seven, U3 snoRNA associates with the nascent pre-rRNA and accompanies it when it passes through the different domains of the nucleolus during its processing. This U3 snoRNA recycles from the granular component (GC) to the dense fibrillar component (DFC) for association with another nascent pre-rRNA.

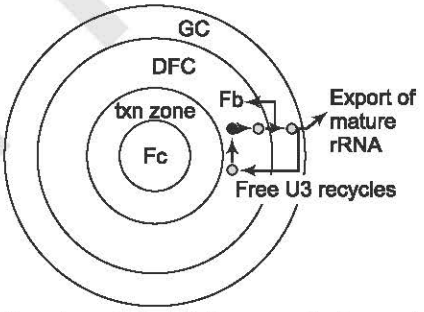


Fig. : A cycle involving association and dissociation of U3 snoRNA with pre-rRNA in the nucleolar regions.

Q.3. Write a short note on satellite.

Ans. The terminal part of a chromosome beyond secondary constriction is called **satellite**. It is attached to the main body of chromosome by a delicate chromatin filament. The satellite may appear as a rounded or elongated knob. It has a constant shape and size for a particular chromosome. The chromosome with satellite is known as **sat-chromosome**.

Q.4. Define the ribozymes.

Ans. Thomas Cech and **Sidney Altman** (1983 & 1985) independently discovered that certain RNAs act as biological catalysts. These are called **ribozymes**. These are associated with the maturation of *hn*RNAs (heterogeneous RNA). Even 235 and 285 ribosomal RNAs of ribosomes are found to catalyse the formation of peptide bonds during synthesis of polypeptide chains in prokaryotic and eukaryotic cells.

Ribozymes act as biological catalysts. These are needed during maturation of heterogeneous nuclear RNAs into functional mRNA molecules. Some ribozymes control peptide formation during synthesis of polypeptide chain.

Q.5. Describe the transformation experiments of Macleod and M. McCarthy.

Ans. **O.T. Avery, C.M. Macleod and M. McCarthy** repeated Griffith's experiments in an in vitro system in order to identify the transforming substance responsible for converting non-virulent into virulent type and reported their results in 1944. Virulence in pneumococcus depends on a polysaccharide capsule which is present in virulent strain S III and is absent in non-virulent strain R II. The cells of non-capsulated type (R II) were treated with an extract of DNA from capsulated strain S III. A few cells of S III type could be isolated from the mixture (Fig.). This phenomenon of transferring characters of one strain to another by using a DNA extract of the former is called **transformation**. When the extract was treated with DNAase (an enzyme which destroys DNA) this transforming ability was lost. Proteases (enzymes which destroy proteins) did not affect the transforming ability. These experiments thus indicated that DNA and not the proteins is the genetic material.

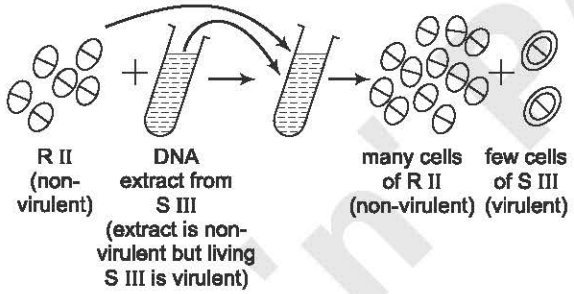


Fig. : Transformation experiment of Avery *et al.* (1944).

Q.6. Name the sugar and the nitrogenous bases present in DNA and RNA.

Ans. The sugar present in DNA is deoxyribose and in RNA there is ribose sugar. The bases adenine, guanine and cytosive are found in both DNA and RNA; thyrmis is found only in DNA and uracil is found only in RNA. The bases are often abbreviated A, G, C, T and U respectively. For convenience the single letters are also used when long sequences of nucleotides are written out.

Q.7. What is caesium chloride density gradient centrifugation?

Ans. Under high centrifugal force, a solution of caesium chloride (CsCl) molecules will dissociate. The heavy Cs^+ atoms will be forced away from the centre towards the outer end of the tube, but will at the same time diffuse back towards the top of the tube, thus forming a shallow density gradient.

Q.8. Define term 'viroid'.

Ans. Viroids are small infectious pathogens. They are composed solely of a short strand of circular, single stranded RNA. Unlike viruses, they have no protein coating. All known viroids are inhabitants of angiosperms, and most cause diseases, whose respective economic importance to humans varies widely.

Q.9. What is antisense RNA?

Ans. **Antisense RNA** is the non-coding strand complementary to a coding sequence of mRNA. A molecule involved in translating genetic instructions into proteins. Antisense RNA hybridixes with and inactivates *m*RNA. Antisense drugs are based on the fact that antisense hybrixes with the inactivates *m*RNA.

Q.10. What is autoradiography?

Ans. **Autoradiography** is the bioanalytical technique used to visualize the distribution of radioactive labelled substance with radioisotope in a biological sample. It is a method by which a radioactive material can be localized within a particular tissue cell, cell organelles or even biomolecules.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Describe the ultrastructure of nucleolus.

Ans.

Ultrastructure of Nucleolus

Nucleolus was first observed by **Fotice Fontana** in 1774, but was formally described by **Wagner** in 1835 from follicles of sheep, and term nucleolus was introduced by **Bowman** in 1848.

Most of cells contain in their nuclei one or more prominent spherical colloidal acidophilic bodies called nucleoli. The size of nucleolus is related with synthetic activity of the cell. The number of nucleoli in the nucleus depends on the species and the number of chromosomes. The position of nucleolus in the nucleus is eccentric. The nucleoli are large in cells that are actively busy in protein synthesis.

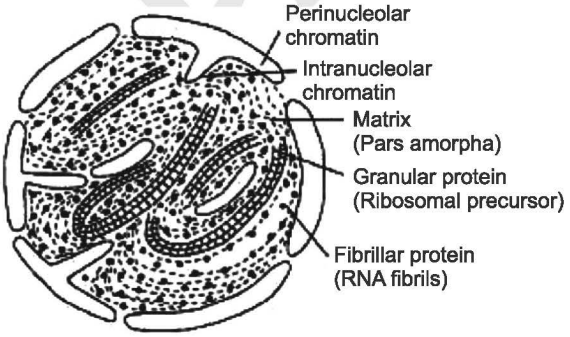


Fig. : Ultrastructure of nucleolus.

Nucleolus consists of following parts :

1. Para amorpha or amorphous matrix,
2. Fibrillar zone,
3. Granular zone,
4. Perinucleolar chromatin.

Fibrillar zone is formed of fibril of rRNA namely 18S, 5.85, and 28S and ribonucleoprotein. Granular zone consists of ribonucleoprotein (RNP) granules. Perinucleolar chromatin formed the chromatin granules.

Chemical Composition of Nucleolus

Nucleolus contains DNA and four types of RNA, 70S type of ribosomal protein. The nucleolus also contains some enzymes such as phosphatase nucleoside, phosphorylase and NAD^+ synthesising enzymes. A ring of DNA is also found which represent hetero-chromatin region of the chromosomes associated with the nucleolus.

Functions of Nucleolus

1. The chromatin is associated with the nucleolus and contains ribosomal gene or RNA that is coded for ribosomal RNA.
2. The ribosomal RNA is synthesised as 28S and 18S RNAs present in the large and small unit of ribosomes of eukaryotic cells.
3. Synthesis of ribosomal RNA (*r*RNA) takes place inside the nucleolus.

4. The two types of *r*RNA's synthesised in nucleolus get associated with the protein that migrate into the nucleolus from the cytoplasm.
5. The ribonucleoprotein molecule represents precursors of ribosomal unit and are seen in the nucleolus as gametes. These precursors come out of the nucleolus into the cytoplasm and join together to form ribosomes.

Q.2. Describe the structure of the Nuclear Pore Complex (NPC) with the help of a suitable diagram.

Ans.

Nuclear Pore Complex (NPC)

A nuclear pore is formed by the fusion of two membranes of the nuclear envelope and is lined with an intricate protein structure called the **nuclear pore complex (NPC)**. The diameter of the entire pore complex is aobut 120 nm. It has an overall mass of some 120 million daltons (Da) and may consist of 100 or more different kinds of polypeptide subunits.

The major component of each nuclear pore complex consists of over 30 differnt pore complex polypeptides. The pore complex as a whole looks like a wheel lying on its side within the nuclear envelope. Two parallel rings, the **cytoplasmic ring** and the **nucleoplasmic ring** are seen in the micrographs. Each ring has a eightfold symmetry. Symmetrically arranged eight spokes extend from each ring to the wheel's hub, which is the **central granule**, now called **transporter**. The transporter carries out import and export of proteins and RNAs.

Thus, the spokes of transporter or the hub are attached to the transporter on the inner side and to the nucleoplasmic and cytoplasmic rings on the outer side. Interspersed between the spokes are aqueous channels 9 nm wide in addition to one channel at the centre of transporter which allow diffusion of proteins and metabolites between the nucleus and the cytoplasm. Proteins extending form the rim into the perinuclear space are thought to help anchor the pore complex to the envelope.

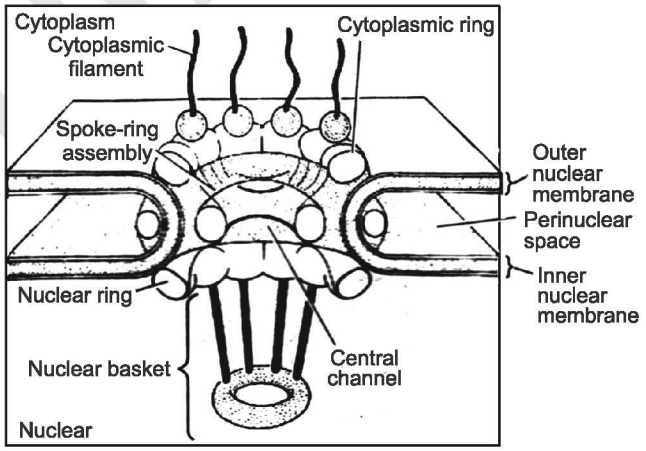


Fig. : Structure of the nuclear pore complex.

On the cytoplasmic side of the pore, thick fibres of 3.3 nm diameter extend into the cytoplasm. On the nuclear side, a large basket-like structure is found which consists of eight filaments, each 100 nm long. These extend from the nucleoplasmic ring of the pore and meet a smaller ring of 60 nm in diameter within the nucleus. The basket plays an important role in RNA export.

Q.3. Write a short note on Balbiani rings.

Ans.

Balbiani Rings

Polytene chromosomes, in addition to bands and interbands, also have special enlargements and extensions of bands into lateral loops or puffs. These large puffs are called **Balbiani rings.**

The puffs form large rings around the chromosome and are presumed to be formed by unwinding or uncoiling of chromonemata (which are otherwise closely folded or coiled in the bands). These then project out into a series of loops. These loops increase thickness or diameter of the chromosome and provide it fuzzy appearance.

Puffs are of following four types :

(a) Stage-specific puffs : These appear at specific stages during development, for example, appearance of puffs during moulting.

(b) Tissue-specific puffs : These appear in the cells of specific tissues at a specific time but not in other types of tissues.

(c) Constitutive puffs : These puffs are active almost all the time in a particular tissue.

(d) Environmentally induced puffs : These appear in a specific tissue after some environmental change. In *Drosophila* and other insects about 80% of puffs are stage specific.

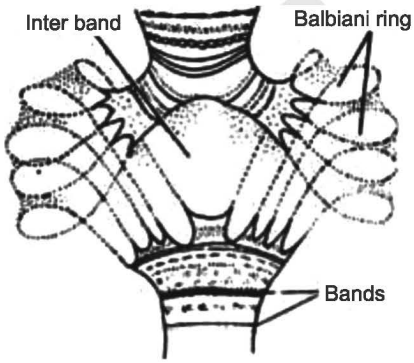


Fig. : Giant chromosome of *Drosophila* showing Balbiani rings.

Control on puff formation : The formation of puffs is controlled by specific genes and takes place at definite time. These are concerned with the synthesis of RNA, because, beside DNA and protein these contain large amount of RNA also. The puffs are chiefly associated with the metabolic activities of the chromosomes because of high concentration of RNA.

Q.4. What is supercoiling of DNA?

Ans. **Supercoiling of DNA** was discovered by **Vinograd** and co-workers (1963). The DNA molecule in viruses and bacteria is twisted upon itself. The supercoiled DNA molecules sediment more rapidly and are more compact. The coiling of DNA could be of following two types :

1. **Negative supercoiling :** When DNA molecule is coiled or twisted in a direction opposite to the direction of twisting of duplex, the two polynucleotide strands tend to unwind. This type of supercoiling that results in underwinding is called negative supercoiling. Negative supercoiling is found in circular DNAs found in mitochondria, viral and bacterial chromosomes.
2. **Positive supercoiling :** When DNA molecule is overwound, it presents positive supercoiling. This is found in eukaryotic chromosomes. It makes eukaryotic DNA to be compacted into a limited space in the nucleus.

Enzymes topoisomerases uncoil or unwind supercoiled DNA in both prokaryotes and eukaryotes. Supercoiling plays key role in allowing DNA to be compacted to as to fit inside the limited space.

Q.5. Write a short note on Lampbrush Chromosome.

Ans.

Lampbrush Chromosome

Chromosomes of a special kind are, found in a variety of primary oocyte nuclei in vertebrates (mainly amphibians) as well as in some invertebrates. These chromosomes, known as lampbrush chromosomes, are found during the prolonged diplotene stage of first meiotic

division in primary oocytes of amphibians, and in spermatocyte nuclei of *Drosophila*. The lampbrush chromosomes are characterized by a remarkable change in structure. The change in structure includes an enormous increase in length. These chromosomes may sometimes become even larger than polynemic giant salivary gland chromosomes. The largest chromosome having a length upto 1 mm has been observed in urodele amphibian. The chromosomes seem to have a chromomeric pattern with loops projecting in pairs from majority of chromomeres.

One to nine loops may arise from a single chromomere. The size of loops varies with an average of 9.5μ in inter-chromomeric fibres. These pairs of loops in these chromosomes give them the characteristic lampbrush appearance. Frequently these loops exhibit a thin axis (which probably consists of one DNA double helix) from which fibres project which are covered with a loop matrix consisting of RNA and protein.

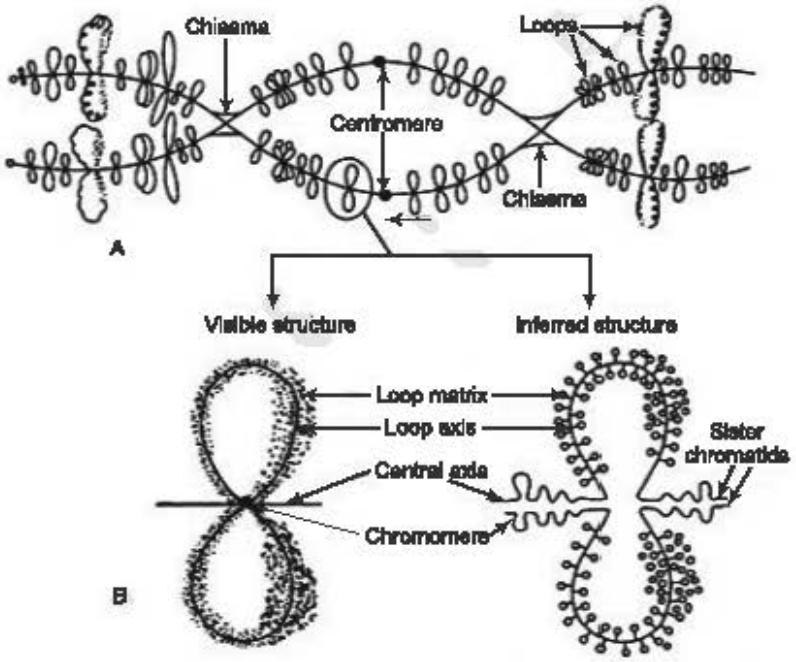


Fig. : Lampbrush chromosomes showing details of structures (redrawn from Lewis & John: Chromosome Marker, 1963).

The number of pairs of loops gradually increases in meiosis till it reaches maximum in diplotene. As meiosis proceeds further, number of loops gradually decreases and the loops ultimately disappear due to disintegration rather than reabsorption back into the chromomere. H. Ris, however, had thought that the loops were integral parts of chromonemata which are extended in the form of major coils. It is also believed that the loops represent the modified chromosome structures at the loci of active genes. It has been observed that if the activity of these genes is stopped by actinomycin D (actinomycin D stops synthesis of RNA on DNA template), the loops will collapse, suggesting that the loops mainly consist of RNA.

Q.6. Differentiate between euchromation and heterochromatin.

Ans. Although, during interphase, the chromatin of the chromosomes spreads out in the form of fine threads of linin but at certain regions, the chromatin remains condensed in the form of darkly stained chromatin mass. These regions are known as the **heterochromatic regions** or **heterochromatin**. The dispersed regions are known as **euchromatic parts** or **euchromatin**. Both euchromatin and heterochromatin are formed of DNA.

1. **Euchromatin :** Euchromatin is transcriptionally active chromatin. Its nucleosomes packing is less condensed and has following specialities—
   (i) Histone H_1 is less tightly bound.
   (ii) Though the four nucleosomal histones are present in normal amounts, they are highly acetylated on lysines near their amino terminus. The acetyl groups are constantly being added to these histones by enzyme histone acetylase and removed by histone deacetylase. (Each acetyl group persists on an average for about 10 minutes only).
   (iii) Active chromatin is highly enriched in a minor variant of histone H2A.
   (iv) Nucleosomes in active chromatin selectively bind two closely related small chromosomal proteins-HMG 14 (high-mobility group) and HMG 17.

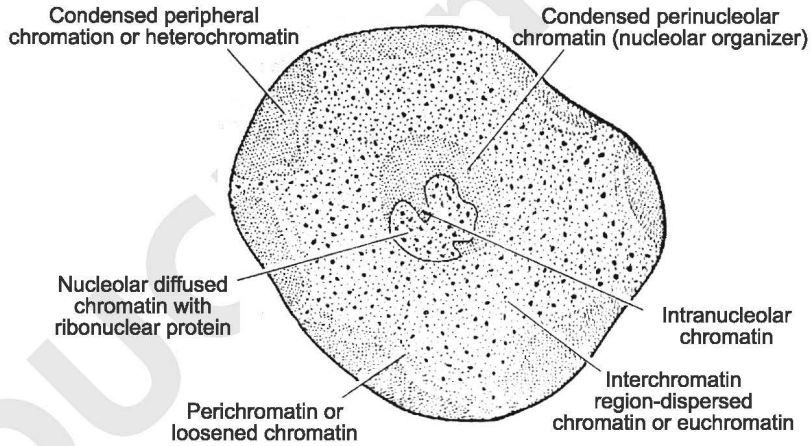


Fig. : Distribution of euchromations and heterochromation in nucleolus.

2. **Heterochromatin : Heitz** (1928) defined heterochromatin as **those regions of chromatin that are transcriptionally inactive and remain condensed in interphase and early prophase and do not uncoil in telophase like rest of chromatin**. In interphase nucleus it appears in the form of darkly stained regions or bodies. The darkness depends upon the degree of coiling of the strands of chromatin.

   Heterochromatin is transcriptionally inactive and is resistant to nuclease digestion. These heterochromatic regions fail to replicate during early cycles of DNA synthesis and replicate after the replication of euchromatic regions.

   Heterochromatin is of two types :

   (i) Facultative Heterochromatin : This represents a temporary state of inactivation of chromatin during interphase in some cell types of an organism but not in others

or inactivation of one chromosome of the pair which becomes partially or totally heterochromatic. For example, in mammals one of the 2X-chromosomes in female somatic cells becomes heterochromatic and forms the **sex-chromatin** or **Barr body** (**Barr** and **Bertram**, 1944). In male somatic cell, there is only one X-chromosome which remains euchromatic (no barr body).

The total amount of facultative heterochromatin is different in different cell types. The embryonic cells have very little heterochromatin, whereas highly specialized cells have a great amount, *i.e.*, as the cells differentiate, progressively more and more genes are packaged in condensed form which are not needed.

(ii) Constitutive Heterochromatin : This type of heterochromatin presents a more permanent feature and is found in both the chromosomes of a pair. It is very often found in the centromeric regions, telomeres, in the regions of nucleolar organizers or as bands in other regions of the chromosomes. It is closely associated with nucleoli in both plants and animals. During interphase, the regions of constitutive heterochromatin aggregate to form **chromocentres.**

Q.7. Describe the account of Yeast Artificial Chromosome (YAC).

Ans.

Yeast Artificial Chromosomes (YACs)

1. Yeast artificial chromosomes (YACs) are genetically engineered chromosomes derived from the DNA of the yeast.
2. It is a human-engineered DNA molecule used to clone DNA sequences in yeast cells.
3. They are the products of a recombinant DNA cloning methodology to isolate and propagate very large segments of DNA in a yeast host.

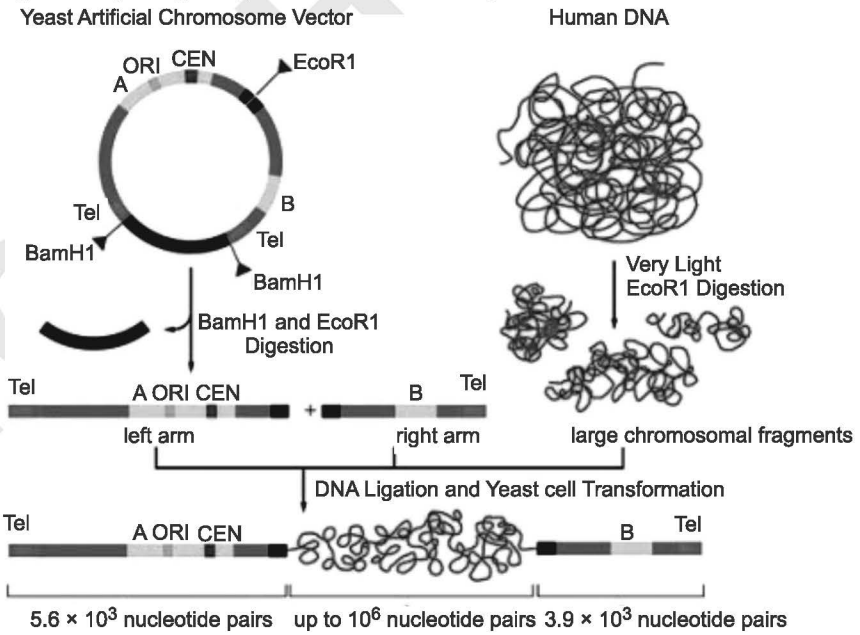


Fig. : Artificial Yeast chromosome with Inserted Human DNA

4. By inserting large fragments of DNA, the inserted sequences can be cloned and physically mapped using a process called chromosome walking.
5. The amount of DNA that can be cloned into a YAC is, on average, from 200 to 500 kb.
6. However, as much as 1 Mb (mega, 106) can be cloned into a YAC.

Structure of Yeast Artificial Chromosomes : A yeast artificial chromosome cloning vector consists of two copies of a yeast telomeric sequence (telomeres are the sequences at the ends of chromosomes), a yeast centromere, a yeast ars (an autonomously replicating sequence where DNA replication begins), and appropriate selectable markers.

Working Principle of Yeast Artificial Chromosomes : The yeast artificial chromosome, which is often shortened to YAC, is an artificially constructed system that can undergo replication. The design of a YAC allows extremely large segments of genetic material to be inserted. Subsequent rounds of replication produce many copies of the inserted sequence, in a genetic procedure known as cloning.

1. The principle is similar to that for plasmids or cosmids.
2. The experimenter introduces some typical elements that are necessary for correct replication.
3. In the case of YACs, the replication origins are the centromeres and telomeres of the yeast chromosomes, which must be inserted into the DNA being cloned.
4. The constructs can be transformed in yeast Spheroplast and are then replicated there. In contrast to the vectors, YACs are not circular; they are made of linear DNA.

Q.8. What do you understand by reverse transcription?

Ans.

Reverse Transcription

In early 1960's, it was shown that some RNA viruses (e.g. Rous Sarcome Virus = RSV), after infecting the host, give rise to DNA as an intermediate step in their multiplication. This above

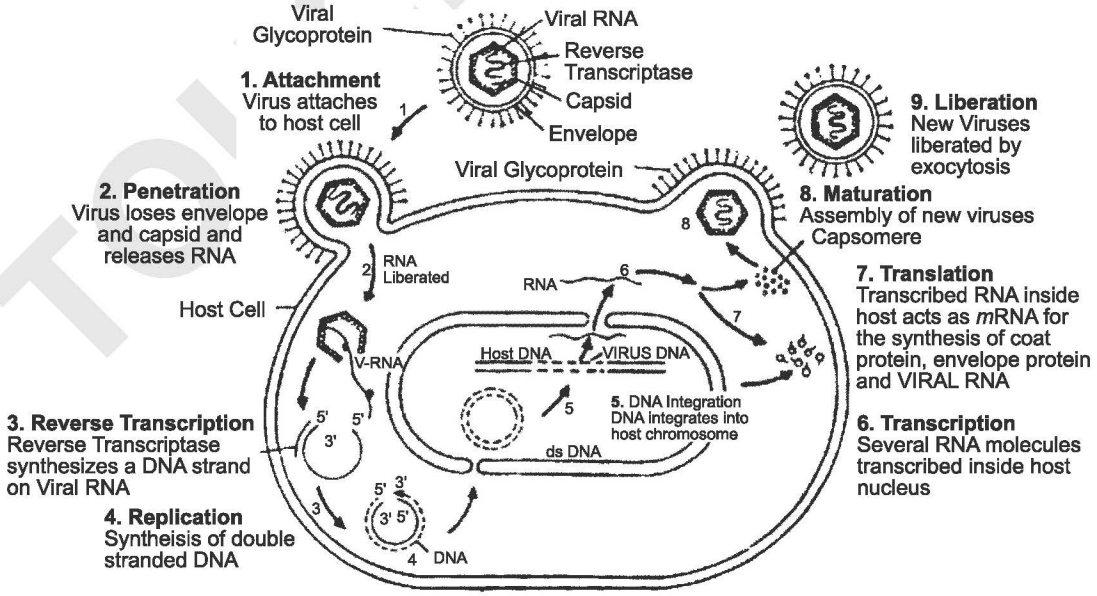


Fig. : Replication of viral particles of retrovirus by reverse transcription inside host cell.

information on RNA directed DNA synthesis was available in 1960's but no enzyme for this activity could be discovered. In 1970, for the first time, disrupted virus particles from **RSV** (used by **H. Temin**) and **MLV** (mouse leukaemia virus; used by **D. Baltimore**) were shown to undertake DNA synthesis, utilizing dNTPs (one of them made radioactive). suggesting that an enzyme existed in the core of virus particle, which could stimulate RNA-directed DNA synthesis.

For this work later, both **Howard Temin** and **David Baltimore** were awarded Nobel Prize. The enzyme was initially called "RNA dependent DNA polymerase" but was later called "RNA directed DNA polymerase", since the purified enzyme was capable of utilizing a variety of templates including synthetic and natural DNA, RNA and RNA-DNA hybrids. The enzyme subsequently became popularly known as **reverse transcriptase**. This enzyme resembles other DNA polymerases and was detected in human tumour cells, initially suggesting its role in cancerous growth of cells. However, later this enzyme was found in the normal cells also, thus reducing the initial excitement about this enzyme being the cause of cancer in humans.

Q.9. Write the differences among A-DNA, B-DNA and Z-DNA.

Ans.

Differences among A-DNA, B-DNA and Z-DNA

S.No.	Characters	A-DNA	B-DNA	Z-DNA
1.	Direction of helix	Right handed	Right handed	Left handed
2.	Appearance	Short and wide	Long and thin	Longer and thinner
3.	Width or diameter of helix	23 Å	20 Å	18 Å
4.	Base pairs per turn	10.9 or 11	10	12
5.	Length of each coil of helix	34 Å	34 Å	45 Å
6.	Distance between adjacent nucleotides	2.7 Å	3.4 Å	3.75 Å
7.	Major groove	Extremely narrow and very deep	Wide and of intermediate depth	Flattened out on helix surface
8.	Minor groove	Very wide and shallow	Narrow and of intermediate depth	Extremely narrow and very deep
9.	Location of axis of helix	Major groove	Through base pairs	Minor groove
10.	Base inclination from helix	13.0	2.0	8.8
11.	Occurrence	Occurs in dehydrated conditions	Occurs in hydrated conditions	Occurs in dehydrated and high salt concentration

Q.10. What is a Klenow fragment?

Ans.

Klenow Fragment

Three different DNA polymerases are known in prokaryotic systems and were isolated from *E. coli*. Of these three enzymes, DNA polymerase I and DNA polymerase II are meant for DNA repair, and DNA polymerase III is meant for actual DNA replication.

DNA polymerase I has two fragments and the larger fragment, called **Klenow fragment**, is mainly involved in removing RNA primers from the leading strand as well as from **Okazaki fragments** of the lagging strand, followed by filling up the gaps thus created. DNA polymerase

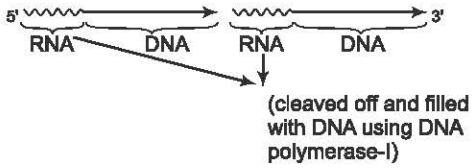


Fig. : Discontinuous DNA replication using RNA primers at 5' ends of segments.

III, has 16 polypeptides involving ten subunits$\{\beta_2\ (\alpha\varepsilon\theta)_2\ \tau_2(\gamma_2\delta_1\delta_1'\chi_1\psi_1)\}$ which are constituted into the following four components :

(i) A polymerase **"core" enzyme** consisting of one molecule each of subunits α, ε and θ, and having the ability to synthesize DNA (two molecules. *i.e.*, $(\alpha\varepsilon\theta)_2$ are used for an initiation complex).

(ii) **A sliding clamp**, consisting of a dimer (β_2) which associates with DNA and forms pre-initiation complex (PIC)

(iii) γ-complex consisting of six polypeptides with five subunits $(\gamma_2\delta_1\delta_1', \chi_1\Psi_1)$; it is also called **matchmaker** because it helps in the loading of sliding clamp (β_2) onto the DNA.

(iv) The τ subunit (used as a dimer τ_2), which is a DNA dependent ATPase of ill defined function. During initiation of DNA synthesis, sliding clamp is first loaded onto DNA in an ATP dependent reaction catalyzed by matchmaker 'γ complex' to form the pre-initiation complex. This is followed by the assembly of the core enzyme with the pre-initiation complex to form the initiation complex in an ATP independent reaction, during which, the matchmaker (γ complex) dissociates. The sliding clamp confers processivity (tendency to remain on a single template rather than to dissociate and reassociate again) to core enzyme and is passively pulled along with core during polymerization. The sliding clamp (β_2) is a ring shaped, head to tail, dimer with a central cavity of sufficient diameter to accommodate duplex DNA. It is a homologue of eukaryotic PCNA

Q.11. What is a primosome?

Ans.

Primosome

In molecular biology, a primosome is a protein complex responsible for creating RNA primers on single stranded DNA during DNA replication. The primosome consists of seven proteins: DnaG primase, DnaB helicase, DnaC helicase assistant, DnaT, PriA, Pri B, and PriC. At each replication fork, the primosome is utilized once on the leading strand of DNA and repeatedly, initiating each Okazaki fragment, on the lagging DNA strand. Initially the complex formed by Pria, PriB, and PriC binds to DNA. Then the DnaB-DnaC helicase complex attaches along with DnaT. This structure is referred to as the pre-primosome.

Finally, DnaG will bind to the pre-primosome forming a complete primosome. The primosome attaches 1-10 RNA nucleotides to the single stranded DNA creating a DNA-RNA hybrid. This sequence of RNA is used as a primer to initiate DNA polymerase III. The RNA bases are ultimately replaced with DNA bases by RNase H nuclease or DNA polymerase I nuclease. DNA Ligase then acts to join the two ends together. Assembly of the Escherichia coli primosome requires six proteins, PriA, PriB, PriC, DnaB, DnaC, and DnaT, acting at a primosome assembly

site on an SSBcoated single-stranded DNA. Assembly is initiated by interactions of PriA and PriB with ssDNA and the pas. PriC, DnaB, DnaC, and DnaT then act on the PriA PriB-DNA complex to yield the primosome.

Q.12. What are Okazaki fragment?

Ans.

Okazaki Fragment

Okazaki fragments are relatively short fragment of DNA synthesized on the lagging strand during DNA replication. At the start of DNA replication, DNA unwinds and the two strands splits in two, forming two "prongs" which resemble a fork (thus, called replication fork). One of the strands goes from 5' to 3' and is called the leading strand; the other strand goes from a 3' to 5' and is called the lagging strand. Unlike the leading strand where DNA can be synthesized continuously the lagging strand is synthesized discontinuously in the form of short fragments called Okazaki fragments that are later connected covalently to form a continuous strand. This is because DNA synthesis can proceed only in one direction – the 5' to 3' direction.

Okazaki fragments are originally discovered by **Reiji Okazaki, Tsuneko Okazaki**, and their colleagues while studying replication of bacteriophage DNA in Escherichia coli in 1968. Word origin named after its discoverers, Reiji Okazaki and his wife, Tsuneko Okazaki, while studying replication of bacteriophage DNA in E. coli in 1968.

Q.13. Describe the clover-leaf model of *t*RNA.

Ans.

Clover-Leaf Model of *t*RNA

According to Clover-Leaf model four different regions or special sites can be recognised in the molecule of tRNA. These are following :

1. **Amino Acid Arm or Acceptor Arm (AA-arm) :** It is double helical and stem-like. It possesses both 5' and 3' ends of the molecule. The 3'terminal always has a base triplet CCA with —OH at the tip. The —COOH group of a specific amino acid joins with the -OH group of adenosine base of CCA in presence of ATP, forming amino acyl tRNA. It is common to all the tRNA molecules. Therefore, 3' end of acceptor arm is called carrier end.

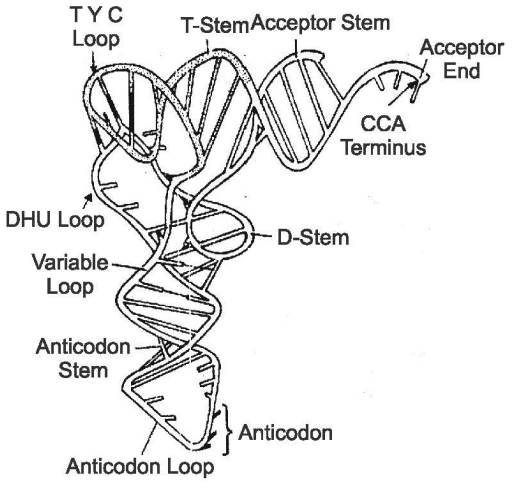


Fig. : Three-dimensional structure of yeast *t*RNA.

2. **Anticodon Arm :** It is loop-like and lies opposite to the AA-arm. It has 3 unpaired ribonucleotides. The nitrogenous bases of these ribonucleotides are complementary to one of the triplet codon of mRNA molecule. Since the base triplet on mRNA chain is called **codon**, its complementary base triplet on tRNA molecule is termed as anticodon. Anticodon reads its appropriate codon on mRNA and temporarily binds to it. Therefore, the terminal end of this arm is called recognition end.
3. **DHU Loop (dihydrouridine loop) :** It is also a loop-like arm. It has enzyme recognition site that binds to specific amino acid activating enzyme which catalyses the union of a specific amino acid to tRNA molecule.

4. **TψC Loop :** It is also a loop-like arm of tRNA with a site for attachment to a ribosome. This site is common to all the molecules of tRNA.

Unusual Base Pairs in tRNA : In addition to the usual bases of RNA (cytosine, guanine, adenine and uracil) each tRNA molecule has several unusual bases. Some of them are **pseudouridine, inosinie acid, methylguanine, methyl aminopurine**, etc.

The presence of these rare nucleotides (unusual) does not affect the pairing of tRNA with mRNA. These probably prevent intramolecular base pairing in the open tRNA loop or help in the recognition of aminoacyl tRNA synthetase enzyme.

tRNA molecules occur in both active and inactive forms. The inactive molecules of tRNA lack the C-C-A sequence of nitrogenous bases at 3' end of the chain either in full or in part. By the addition of this nucleotide sequence an inactive tRNA molecule is activated. The process is governed by cytidine triphosphate (CTP) and adenine triphosphate (ATP).

Q.14. Write a short note on topoisomerases.

Ans. The compactness of DNA inside the nucleus or the nucleoid needs to have a simplified structure for carrying out vital cellular processes. Knotting and unknotting of DNA is precisely controlled by several ubiquitous enzymes, among which DNA topoisomerases maintain the chromosome integrity and fidelity of transmission of genetic materials over generations. Simultaneous release of positive supercoils at the head of replication fork and overwound of tail have been processed by DNA topoisomerases by transiently breaking single or double strand, strand rotation/passage, and religation. Besides maintaining the linking number (Lk), some topoisomerases are also involved in recombination or DNA repair processes. Several families of topoisomerases have been characterized on the basis of their mode of action and sequence homology. Abrogation of topoisomerase function inside cells can have deleterious effect to stall the essential life processes, and targeting the topoisomerase DNA covalent complex or the enzyme alone may open some novel therapeutic aspects against deadly diseases.

DNA topoisomerases are able to solve topological problems resulting from replication, transcription, recombination, and reorganization of the chromatin. Further on, topoisomerases change the state of supercoiling of the DNA and therefore, have great impact on gene activity. The mechanism of topoisomerase action includes the transient formation of an ester bond between a tyrosine residue of the enzyme and the DNA molecule. Later on, the breaks are closed by reformation of the original phosphodiester-bond and the enzyme released from the DNA. In order to decrease gene activity, DNA topoisomerases introduce temporary single-strand breaks (type I) or double-strand breaks (type II) in the phosphate backbone of the DNA.

(i) **Type I topoisomerases** are further classified as type IA and type IB enzymes, depending whether the protein is attached at a 5' or 3'-phosphate, respectively. All human topoisomerases generating double-strand-breaks are designated as type IIA topoisomerases, in order to discriminate them from type IIB enzymes, a recently discovered class with topoisomerase VI from Sulfolobus shibatae as a prototype. Type IIA enzymes act as homodimers in contrast to type IIB enzymes, which are A_2B_2 heterotetramers. Although these latter group of topoisomerases are obviously lacking in mammalian cells, the SPO11 protein, responsible for the introduction of meiotic

double-strand-breaks, has significant similarity to the A subunit. On the other hand, no counterpart for a B subunit is known from mammalian cells.

(ii) **Type II topoisomerases** are associated with ATPase activities which are located either at the N-terminus (type IIA) or on the B subunit (type IIB).

Q.15. Write the difference between DNA and RNA.

Ans.

Differences between DNA and RNA

S.No.	Deoxyribonucleic acid	Ribonucleic acid
1.	DNA is found in the chromosomes of the nucleus and is chiefly concentrated in the nucleus only.	RNA is chiefly concentrated in the cytoplasm although it also occurs in the nucleolus and to some extent in the nucleoplasm and associated with the chromosomes.
2.	The sugar molecule in DNA is deoxyribose.	The sugar molecules in RNA is ribose.
3.	The four nitrogenous bases found in deoxyribonucleic acid are : (a) Adenine, (b) Guanine } Purine bases (c) Cytosine, (d) Thymine } Pyrimidine bases	The four nitrogenous bases found in RNA are : (a) Adenine, (b) Guanine } Purine bases (c) Cytosine, (d) Uracil } Pyrimidine bases
4.	DNA is a double-stranded structure having a helical configuration. The two strands are coiled spirally in opposite directions.	RNA molecules are single-stranded. The strand in some cases (in RNA) is coiled on itself and may be connected by hydrogen bonds.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. Give an account of the structure of a nucleus. Discuss the structure and function of different components of a nucleus.

Ans.

Nucleus

Discovery and Occurrence : The nucleus is the controlling centre of the cell. It directs and controls all the cellular activities. It was first described by **Robert Brown** (1831).

A well organised nucleus with a distinct nuclear membrane is absent in bacteria; viruses and blue-green algae. All other living organisms have a nucleus in their cells; On this basis biologists have separated the living organisms into two groups :

1. Prokaryotes 2. Eukaryotes.

In prokaryotes, the nuclear membrane is absent. The chromatin material (DNA) lies in the cytoplasm. It is known as **prokaryon** or **nucleoid.** In eukaryotes, the nuclear chromatin is isolated from the surrounding cytoplasm by the presence of nuclear membrane.

Structure of Nucleus

The **nucleus** is also known as metabolic nucleus as it controls the metabolic activities of the cell. This nucleus may be spherical; rounded, Spheroidal, cylindrical, prismatic; branched or lobed. The nucleus can be separated into the following parts :

1. Nuclear Envelope or Karyotheca

The nuclear envelope is a double membranous sheath that separates the nuclear material from the cytoplasm. It acts as a dynamic gateway between nucleus and cytoplasm and regulates nucleocytoplasmic interaction.

(a) Nuclear Membranes: The two nuclear membranes of nuclear envelope are separated by a **perinuclear space**. This space is 20 nm in width. The **nuclear membranes** are 7-8 nm thick. The outer membrane has attached ribosomes. At places, it is continuous with the membranes of ER. The nuclear surface of **inner membrane** is coated with filaments and fibres. These form a **fibrous lamina** or **nuclear cortex.**

(b) Nuclear Pores: The nuclear envelope is perforated by pores. At the margin of these pores, the outer and inner nuclear membranes are continuous.

The nuclear pores are 100 nm wide circular channels. These are occluded by some electron dense material. This projects outward in the cytoplasm and inward into the nucleoplasm. These structures are called annuli. The two together form pore complex. Each pore complex has an outer diameter of 90nm, inner diameter of 25 nm, and a wail thickness of 30 nm. The electron dense material consists of fibrous and particulate structures. These are present along the margin of nuclear pore and form annulus.

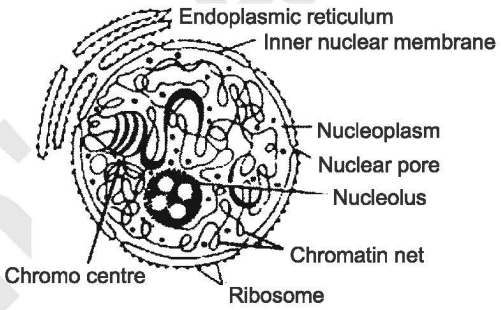


Fig. : Structure of a typical nucleus

The striated muscle fibres and the epidermis of *Ascaris* are syncytial structures. In certain primitive cells, the nuclear chromatin is found scattered in the form of granules and an organised nucleus is lacking.

(c) Annulus: Annulus appears as a ring or cylinder of electron dense material of nuclear pore. It is formed of two sets of eight evenly spaced annular granules and a central granule. One set is arranged on the outer surface and other set on the inner surface of the annulus. The granules may be formed of compact material or fibrillar material. Fine fibres extend from the central granule to peripheral annular granules. Annuli around the pores regulate the exchange of macromolecules in relation to their size.

(d) Fibrous or Nuclear laminar: The inner surface of nuclear envelope is plastered with a fibrous coating, called **nuclear cortex**. Its fibres are proteinaceous and similar to actin polymers. The fibres form funnel-shaped whorls. These funnels communicate with the nuclear pores and most probably direct material towards pore channel to come out of the nucleus.

The nuclear cortex functions as a funnel to direct materials towards pore channels. It also provides a supporting lamella.

2. Nucleoplasm or Nuclear Sap or Karyolymph

It is the transparent ground substance. Chromatin network remains suspended in it. It is a mixture of proteins, large amount of phosphorus and some nucleic acids (RNA). A number of hydrolytic enzymes such as ribonuclease, alkaline photophosphatase and dipeptidase are also found in the nucleoplasm.

3. Nucleolus

Usually, there are two nucleoli in a nucleus of diploid cell and only one in gametes. The size of nucleolus is related with the synthetic activities of the cell. The nucleoli are large in cells that are actively busy in protein synthesis.

Structure: Nucleolus consists of following parts:

(a) **Pars amorpha** or **amorphous matrix**.

(b) **Fibrillar zone** formed of fibrils of rRNA and ribonucleoproteins.

(c) **Granular zone** consists of ribonucleoprotein (RNP) granules. These are precursors to ribosomes.

(d) **Perinucleolar chromatin** formed of chromatin granules. The different zones of nucleus are related in the following manner:

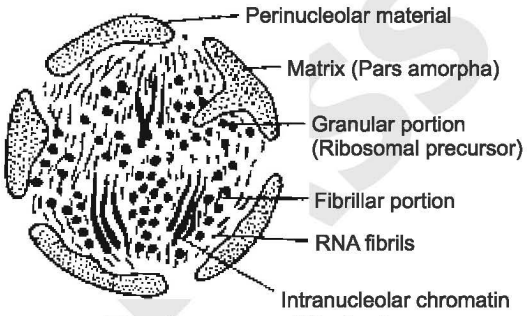


Fig. : Structure of Nucleolus

Nuclear DNA → Fibrillar zone → Granular zone → Cytoplasmic ribosomes.

Nucleolus is mainly composed of RNA and proteins. The proteins are exclusively phosphoproteins, and RNA is similar to ribosomal RNA. A ring of DNA is also found which represents heterochromatin regions of the chromosomes associated with the nucleolus. In addition, .the enzymes like acid phosphatase, nucleoside phosphorylase and DPN synthesising enzymes are found.

4. Nuclear Reticulum or Chromatin Net

Embedded in the nuclear sap is the network of twisted filaments or threads, which are known as **chromonemata** and their network as nuclear reticulum or chromatin net. It contracts and organises into distinct chromosomes during cell division.

Euchromatin and Heterochromation: The cell chromatin can be differentiated into two types. The fine thread-like linen of the chromatin, which stains lightly with basic dyes is called **euchromatin**. At certain regions the chromatin is condensed and darkly stained. This is known as **heterochromatin**. The heterochromatin regions can be seen in the interphase and prophase. It does not unravel in telophase like the remaining euchromatin.

The euchromatin also exhibits different affinities to the dyes. The linen of the chromatin which is in the form of lightly stained'threads is composed of **achromatin** (stains with acidic dyes) and the darkly stained-granules (chromomeres) present on the linen are formed of basichromation or **chromatin proper.**

In some cases, large regions of nucleus take dark stain with basic fuchsin. These are known as chromocentres of karyosomes.

Special sex chromatin bodies or Barr-bodies are found in the periphery of the nucleus. These are more common in mammalian cells and specially females. Their number usually depends upon the number of sets of X-chromosomes. Normally, there is one sex chromatin body for a diploid set of chromosomes.

5. Chromocentres

In the interphase nucleus of certain cells, some areas of considerable size take darker stain than the rest of the chromatin. These darkly stained areas are actually heterochromatic regions of the chromosomes which are pron to premature condensation. There can be only one or many heterochromatic regions of several chromosomes or of all the chromosomes of the nucleus. The chromocentre is well marked in the nuclei of salivary gland cells of *Drosophila*.

Functions of Cell Nucleus

The important functions carried out by a cell nucleus are:

1. Storage of hereditary material, the genes in the form of long and thin DNA (Deoxyribonucleic Acid) strands, referred to as chromation.
2. Storage of proteins and RNA (Ribonucleic Acid) in the nucleolus.
3. Nucleus is a site for transcription in which messenger RNA (mRNA) are produced for protein synthesis.
4. Exchange of hereditary molecules (DNA and RNA) between the nucleus and the rest of the cell.
5. During the cell division, chromatins are arranged into chromosomes in the nucleus.
6. Production of ribosomes (protein factories) in the nucleolus.
7. Selective transportation of regulatory factors and energy molecules through nuclear pores.

As the nucleus regulates the integrity of genes and gene expression, it is also referred to as the control centre of a cell. The nucleus contains all the genetic material of art organism like chromosomes, DNA, genes, etc.

Q.2. Describe the structure, chemical composition and functions of chromosome.

Ans.

Chromosomes

Structure : In non-dividing eukaryotic cells, the genome is nucleoprotein complex, called **chromatin**. It is amorphous and is randomly dispersed in the nuclear matrix as interwoven network of fine chromatin threads. When cell prepares to divide, the chromatin condenses into a species-specific number of well defined **chromosomes**.

Number : The number of chromosomes in the somatic cells of higher animals and plants is known as **diploid** or **somatic** or **zygotic number (2n)**, while in the gametes (sperm and eggs) it is **haploid, gametic** or **reduced (n)**. The number of chromosomes is constant in all the somatic cells of all the individuals of a species. Chromosome number is used in the identification of species and in tracing the relationship within the species.

Shape and Size : The anaphase chromosomes may appear as **rod-shaped, twisted or spiral, curved** or **filamentous**. The chromosomes may be of equal thickness throughout or constricted at places. The shape of chromosomes is usually determined by the type and the position of its centromere. Depending upon the position of centromere, the chromosomes in anaphase may assume the form of **rod, J** or **V**.

Chromosomes differ greatly in size in different organisms, in unlike tissues and to some degree in plants grown in different nutrient solutions. Even the chromosomes of different pairs in the nucleus of the same cell have different sizes. The anaphase chromosomes range from 1 μ to 30 μ in length and 0.22 μ to 2 μ in width.

Chromosomes exhibit cyclic changes in shape and size during cell cycle. In the non-dividing interphase nucleus, the chromosomes form an interwoven network of fine twisted but uncoiled threads of chromatin, and are invisible. During cell division the chromatin threads condense into compact structures by helical coiling. In **prophase** of cell division the chromosomes appear as distinct threads and by **metaphase** and also in **anaphase** these become short, compact bodies having definite shapes and sizes. In anaphase these appear as rod-shaped, V-shaped, L-shaped or J-shaped. The chromosomes are studied and described at this stage. In **telophase** these again uncoil to form the chromatin net.

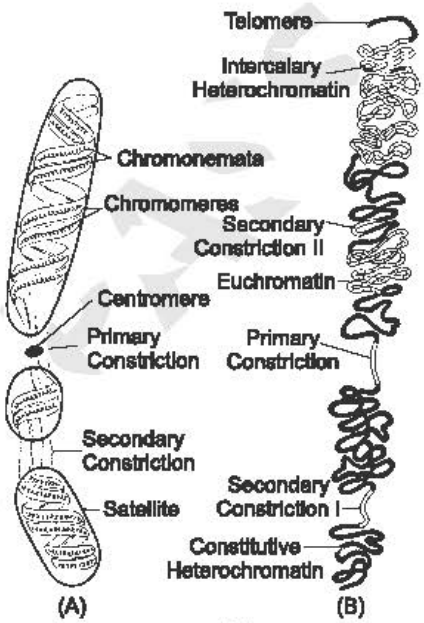


Fig. : Structure of chromosome : A. In Metaphase, B. In Anaphase.

A part of the chromosome is marked by a constriction. It is comparatively narrow than the remaining chromosome. It is known as **primary constriction.** Its position is constant for a given chromosome and forms a feature of identification. The primary constriction divides the chromosome into two **arms.** It shows a faintly positive Feulgen reaction, indicating presence of **DNA** of repetitive type. This **DNA** is called **centromeric heterochromatin.**

Centromere or kinetochore lies in the region of primary constriction. The microtubules of the chromosomal spindle fibres are attached to the centromere. It is a disc-shaped structure, formed of specific DNA sequences and special protain bounded to them. Therefore, centromere is associated with the chromosomal movement during cell division and is therefore, called **Kinetochore.**

Chemical Composition of Chromosome

Each chromosome is formed by the folding and refoldmg chromatin or a nucleoprotein fibre. It is formed of nucleic acids and proteins.

The major chemical components of chromosomes are DNA, RNA, histone proteins and non-histone proteins. Calcium is also present in addition to these constituents.

1. **DNA :** As we know, DNA is the most important of chemical components of chromatin, since, it plays the central role of controlling heredity. Quantitative measurements of DNA have been made in a large number of cases which are reviewed by M.D. Bennett and I.J. Leitch in 1995.

   The most convenient measurement of DNA is picogram (10^{-12} g) which is equivalent to 31 cm of double helical DNA. It has been found that quantity of DNA vanes greatly in cells from different kinds of organisms. The haploid genome of mammals usually contains 1000 times DNA content of bacteria. Other eukaryotes may similarly have 10

to 100 times the bacterial DNA content. It is interesting to note that a human diploid cell has 174 cm (5-6 picograms) of DNA, so that all cells in a human being may have DNA equal to 2.5×10^{10} km (100 g), a length which is equal to 100 times the distance from earth to sun. Similarly a diploid cell of *Trillium* has 37 metres (120 picograms) and that of *Drosophila* salivary glands has 91 metres (293 picograms) of DNA. In comparison of these enormous lengths, the DNA of bacteria measures only 1.1 mm –1.4 mm.

2. **Histones :** There are five fractions of histones, which have been differently designated. H1 histone is most easily removed and so is least tightly bound. This may thus be concerned with holding together a chromosome fibre. H3 and H4 are extremely conserved, having same structure in different species and should thus have a common structural role.

   Histones, isolated from diverse materials showed considerable similarity. It is also assumed that general similarities in histones have been conserved during evolution. This feature alone suggested that these proteins should play a structural role rather than a regulatory role. However, some important experiments involving chromatin reconstitution and other experiments conducted in recent years have established that histones do play a regulatory role. This regulatory role of histones is more of general nature rather than specific and is exercised by repressing the activity of genes.

3. **Non-histones :** The non-histone proteins display more but still limited diversity. In a variety of organisms, number of non-histones can vary from 12 to little more than 20. Heterogeneity of these proteins suggested that these proteins are not as conserved in evolution as histones. These non-histone proteins differ even between different tissues of the same organism suggesting that they regulate the activity of specific genes.

   Chromatin reconstitution experiments described in 1973 by **R.S. Gilmour** and **J. Paul** of Institute for Cancer Research at Glasgow (U.K.), established conclusively for the first time that specific non-histone proteins switch on specific genes. The results of these experiments were later confirmed in a number of cases (**Barrett** *et al.* 1974; **Groner** *et al.* 1975). During 1980s and 1990s, many of the transcription factors have been shown to be non-histone proteins, providing further evidence that non-histone proteins exercise positive control on the activity of specific genes.

Functions of Chromosomes

1. Chromosomes control the development and differentiation of different characters of the organism.
2. Changes in the number and structure of chromosomes lead to the changed appearance of different characters.
3. Repititive DNA helps in pairing of homologous chromosomes and crossing over during meiosis.
4. Chromosomes are **'vehicle of hereditary material'.**
5. The chromosomes control the physiological or metabolic activities of the organism by controlling protein synthesis.
6. The heterochromatic region of chromosome participates in the formation of nucleolus.
7. Kinetochore helps in attaching the chromosomes with the spindle fibres during cell division and helps in the movement of chromosomes: during anaphase.

Q.3. What is Watson and Crick's model of DNA structure? How does it explain the biological significances of DNA.

Ans.

Watson and Crick Model of DNA

Watson and **Crick** deduced the double helical structure of DNA and shared Noble Prize in 1962 with Maurice Wilkins. Watson, at present, is Director of Spring Harbor Laboratory. He has worked on RNA synthesis, protein synthesis and role of viruses in cancer. Crick has worked extensively on molecular biology and has contributed in understanding genes and gene expression.

Watson and Crick suggested that in a DNA molecule there are two such polynucleotide chains arranged antiparallel or in opposite directions, i.e., one polynucleotide chain runs in 5 -3' direction, the other in 3'—5'.direction. It means the 3' end of one chain lies beside the 5' end of other. In such a structure the phosphate groups of nucleotides in each polynucleotide chain or strand lie on the outside of the deoxyribose and the nitrogenous bases are directed inward. The nitrogenous bases of the two chains are linked through hydrogen bonds formed between oxygen and nitrogen atoms of the adjacent bases. The unique feature of pairing between bases is :

1. **Purine** (adenine and guanine) pairs with **pyrimidine** (cytosine and thymine), and
2. **Adenine** pairs with **thymine** and **cytosine** pairs with guanine.

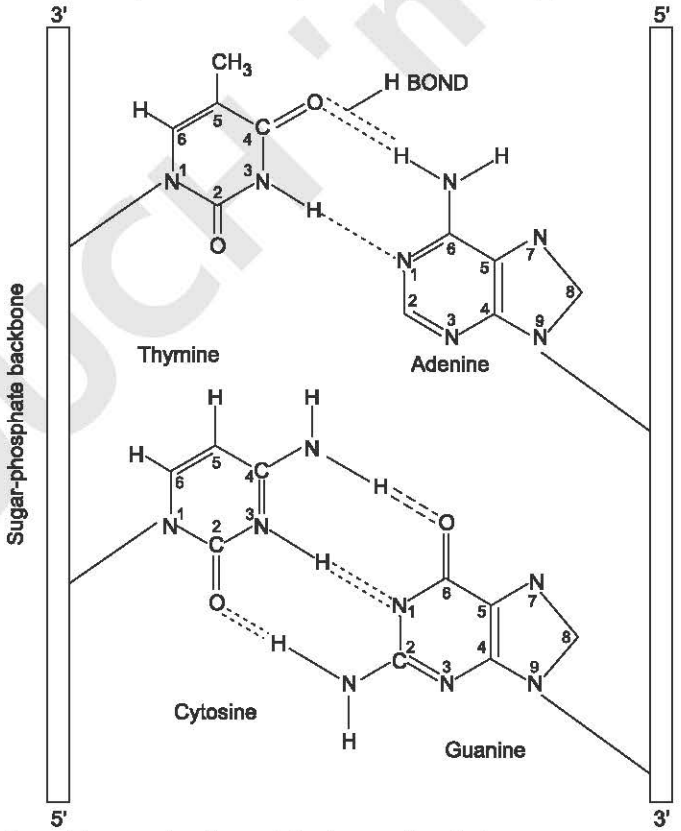


Fig. 1 : Association of base pairs through hydrogen bonds between opposite nucleotides of two polynucleotide chains of DNA.

There are definite reasons for such a specific pairing :

(i) Such pairing forms a perfect match between hydrogen donor and hydrogen acceptor sites on the two molecules. Adenine and thymine share two hydrogen atoms, whereas cytosine and guanine are joined by three hydrogen bonds.

(ii) Such a pairing is further supported by the occurrence of constant diameter of DNA. In a limited area a two-ringed molecule (purine) joins a single-ringed molecule maintaining a constant and roughly equal distance.

A and G pair will be rather too large to fit inside the helix and C and T would appear to be far apart.

Due to this type of base pairing the two strands are **complementary** to each other. It means if a chain has a region with a sequence of nitrogenous bases, **thymine-cytosine-adenine-cytosine-guanine**, then the corresponding region in the complementary chain will have the base sequence **adenine-guanine-thymine-guanine-cytosine.**

DNA consists of two complementary chains twisted around each other forming a **right handed helix.** One turn of helix measures about 3.4Å. It contains 10 paired of nucleotides

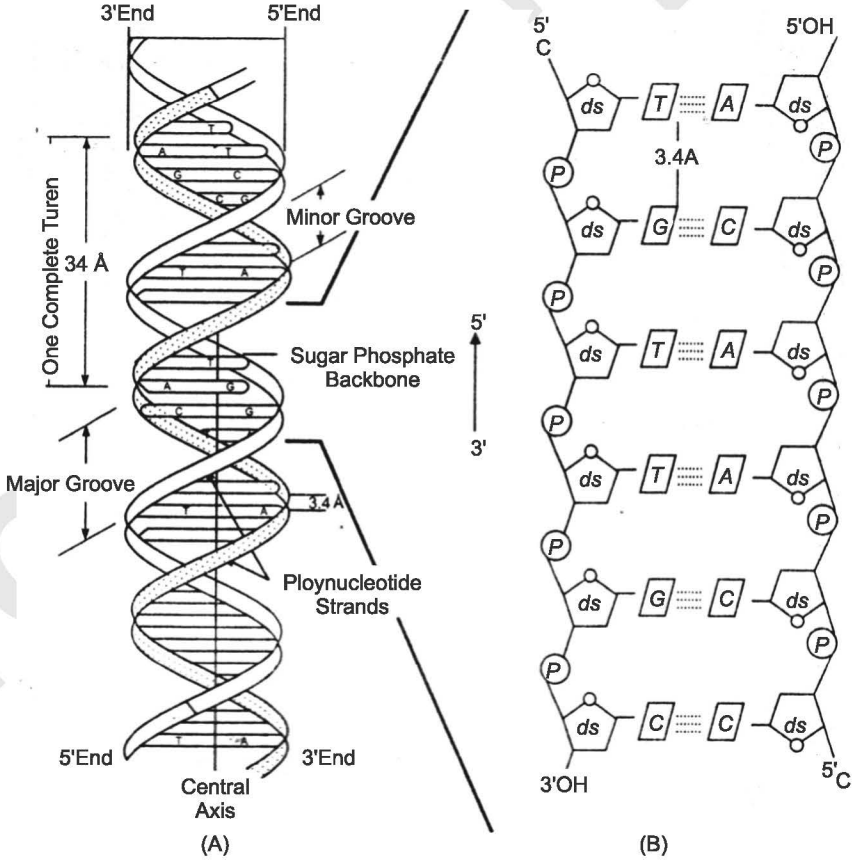


Fig. 2 : Double helical structure of a part of DNA molecule. One left are shown two ploynucleotide chains of DNA twisted spirally in a helical fashion. On the right are shown nucleotides of a strand linked through deoxyribose and phosphoric acid molecules to form polynucleotide chain that joins the nucleotides of antiparallel polynucleotide chain by hydrogen bonds.

placed at regular intervals of 3.4 Å. The diameter of the helix is roughly 20Å. A narrow helical groove and a wide helical groove run along the length of DNA helix. The narrow groove is the distance between the paired molecules while the **wide groove** is the space between successive turns when the pair is wound into a helix.

Biological Significance of DNA

Watson and Crick model of DNA enables the hereditary material to perform following functions :

1. **Information Storage :** Genetic material is expected to act as a repository of information. Its structure must be related to the intimate features of gene-product *i.e.*, the protein. It must be capable of specifying the primary structure of a protein *i.e.*, composition of a polypeptide chain. It means the sequence of bases in the DNA molecules should be able to determine the specific sequence of amino acids in a polypeptide chain. With the discovery of triplet codons and genetic code for different amino acids, the collinearity between codons in DNA and sequence of amino acids in polypeptide chain has been successfully established. Therefore, DNA is like a tape having coded information for the sequence of amino acids in the form of nucleotide sequencing.
2. **Information transfer :** The process of transferring message encoded in DNA and ultimate synthesis of polypeptide chain can be broadly separated into two steps :
   (a) Faithful transfer to information from stationary DNA molecule to a mobile intermediate molecule which can be the liaison between DNA in nucleus) to ribosomes (in cytoplasm) for assembling of amino acids into protein.
   (b) Decoding of the message from mobile carrier into a polypeptide chain.

   Double helical structure of DNA as suggested by Watson and Crick conveniently explains the transfer of information.

   Since the information is coded in the sequence of nitrogenous bases, the same sequence is copied in mRNA. Thus, a complementary strand of mRNA is synthesised along one of the two strands of DNA. This mobile copy of genetic message moves out of the nucleus to the site of protein synthesis (ribosome) in cytoplasm. *t*RNA molecules pick up specific amino acids and align them on *m*RNA in a triple sequence of nitrogenous bases (the codon) recognised by their anticodon region.
3. **Variations :** Genetic material should be capable of undergoing change in its composition. Its structure should be able to undergo mutations so as to produce diversity in the protein-making informtion. Watson and Crick model easily explains the mechanism of appearance of mutations in DNA either by substitution of nitrogenous base pair or by their addition or deletion in the polynucleotide chain.
4. **Self-replication :** The structure of DNA provides the most convenient and the only possible device whereby a particular molecule with particular sequence of base pairs could be duplicated easily. Each strand of double helix of DNA can construct its complementary strand by acting as a template, because the sequence of nitrogenous bases in one strand could easily determine the laying down of bases of the complementary strand.

5. **DNA repair :** DNA molecule is relatively fragile and is easily damaged simply by bending, by shear forces and by local change in pH. Its base sequence changes by chemical agents from environment and also by X-ray and ultraviolet radiations. Still it preserves its integrity after several millions of replications. Actually the double helical structure of DNA presents the most stable structure. It is most suited for the long term preservation of the coded informations. Because the base sequence in the two polynucleotide chains is complementary, the coded informations in a DNA molecule are preserved in duplicate. If anyhow, the sequence of bases on one chain is disturbed, the correct sequence of the other chain may help in correcting the errors that occur in the complementary strand and sends the correct message.

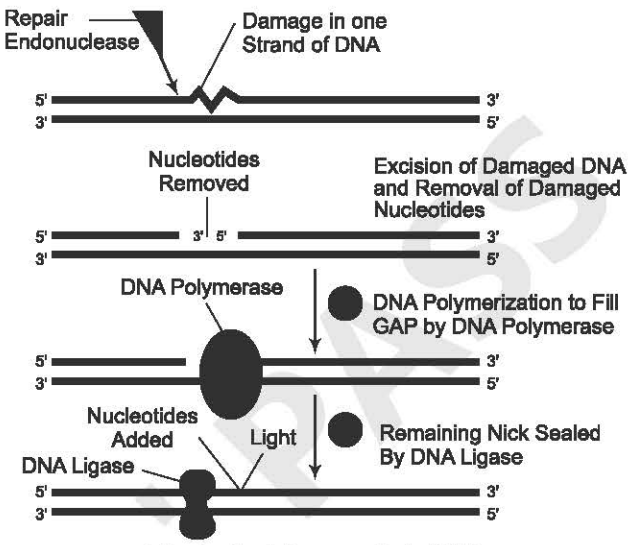


Fig. 3 : Excision repair in DNA

A number of DNA repair systems are found in both prokaryotes and eukaryotes. All these systems use enzymes to excise incorrect nucleotides and replace them with the correct ones. Some of these repair systems are :

(i) **Mismatch repair :** Proof reading activity of DNA polymerase helps in replacing mismatched base pairs left after DNA replication. Due to 3'→ 5' exonuclease activity of DNA polymerase mismatched nitrogenous bases or nucteolides are removed from the newly formed polynucleotide strand, while its 5'→ 3' polymerising activity helps in replacing these with correct nucleotides.

(ii) **Nucleotide excision repair :** This system operates by removing a small section of a DNA strand containing certain types of bulky lesions, such as pyrimidine dimers or nucleotides to which chemical groups are attached.

(iii) **Base excision repair :** This system is used to remove altered nucleotides that produce less distortion of double helix.

Q.4. What is RNA? Discuss its structure and functions. Give a brief account of different type of RNAs found in the cell.

Ans.

RNA

Ribonucleic acid (RNA) is single-stranded nucleic acid found in all living cells. Its different forms are associated with the transmission of information from nucleus into the cytoplasm and with the synthesis of proteins for the regulation of cell activities. However, RNA is the only macromolecule which also functions for the storage of information and also as a catalyst. The catalytic RNAs are called **ribozymes**. RNA is also presumed to have played an important role as an essential chemical intermediate in the evolution of life on this planet.

RNA is found chiefly in the cytoplasm and in the nucleolus. Inside the cytoplasm it occurs freely as well as in the ribosomes. RNA can also be detected from mitochondria, chloroplasts and associated with the eukaryotic chromosomes. In some plant and animal viruses, RNA acts as a hereditary material.

Structure of RNA

The structure of RNA can be easily understood by comparing it with the structure of DNA :

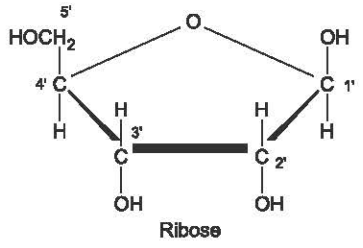


Fig. 1 : A molecule of ribose.

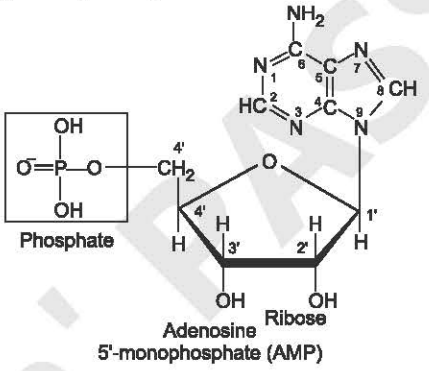


Fig. 2 : A nucleotide of RNA (uridylic acid)

1. More commonly RNA is a single-stranded structure consisting of an unbranched polynucleotide chain, but it is often folded back on itself forming helices. DNA is a double-stranded structure and its two polynucleotide chains are wounded spirally around a main axis.
2. RNA like DNA is formed of several hundreds or thousands of nucleotides arranged in a linear sequence and connected together by 3'–5' phosphodiester bonds.
3. The sugar found in nucleotides of RNA is **ribose**, whereas it is **deoxyribose** in DNA. The nucleotides of RNA are **ribonucleotides**.
4. The four nitrogenous bases found in RNA are adenine, cytosine, guanine and uracil, whereas those in DNA are adenine, cytosine, guanine and thymine. Some unusual nitrogenous bases are also found in RNA. Therefore, in RNA thymine of **DNA is substituted by uracil**.
5. The base composition of RNA does not agree to the A/U=G/C=1.
6. Intramolecular pairing between the nucleotides of single strand of RNA provides stability to RNA. In DNA, nucleotides of two polynucleotide strands pair through hydrogen bonds.

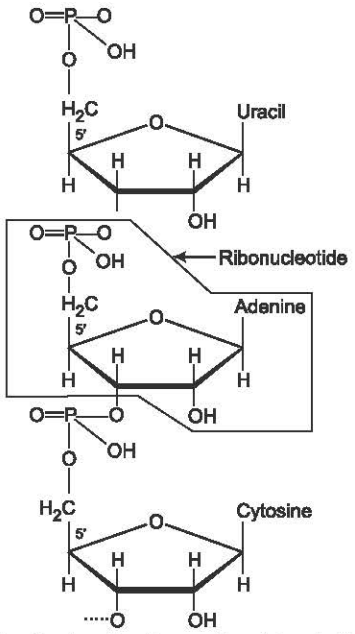


Fig. 3 : A polyribonucleotide chain.

7. DNA is the hereditary material, whereas RNAs are of different types performing different functions during protein synthesis. Of course, in most of the plant viruses and some animal viruses RNA acts as hereditary or genetic material.

Types of RNA

In all other organisms, where DNA is the hereditary material, different types of RNA and non-genetic. The non-genetie RMA is synthesisd from DNA template. In general, three types of RNA have been distinguished :

1. Messenger RNA (*m*RNA) :

Messenger RNA or Nuclear RNA carries genetic information from chromosomal DNA to the cytoplasm, where it acts as a template for protein synthesis. It is complementary to DNA and carries the copy of the same base sequence as found in that part of DNA from which it is copied except that thymine is substituted by uracil. It constitutes about 5% of the total RNA of the cell.

Since, the **'genetic blueprint'** is contained in the nucleus, and the **'work benches'** or **'sites'** of protein synthesis (*i.e.*, ribosomes) are present in the cytoplasm, these RNA molecules serve to transport genetic messages from DNA to ribosomes. The term messenger RNA was used by **Francis Jacob** and **Jacques Monod** (1961). In the cytoplasm, mRNA molecules become attached to ribosomes and act as template for the polymerisation of amino acids into polypeptide chains. The sequence of amino acids in each polypeptide chain is determined by the sequence of codons in its mRNA molecule.

The main part of mRNA is the sequence of nucleotides that codes for a polypeptide. However, a fully processed mRNA molecule has at either end some sequences that are not translated. Because of these sequences the two ends of each mRNA molecule are distinctly marked. Each fully processed mRNA molecule has following parts :

1. A cap of methylated guanine **(G-cap)** at its proximal or 5' end.
2. A **start** or **initiation codon** (AUG or GUG) next to G-cap.
3. A long **coding region.**
4. A termination codon or stop codon (UAA, UAG or UGA) at the end of coding region at the distal end (*i.e.*, 3' end).

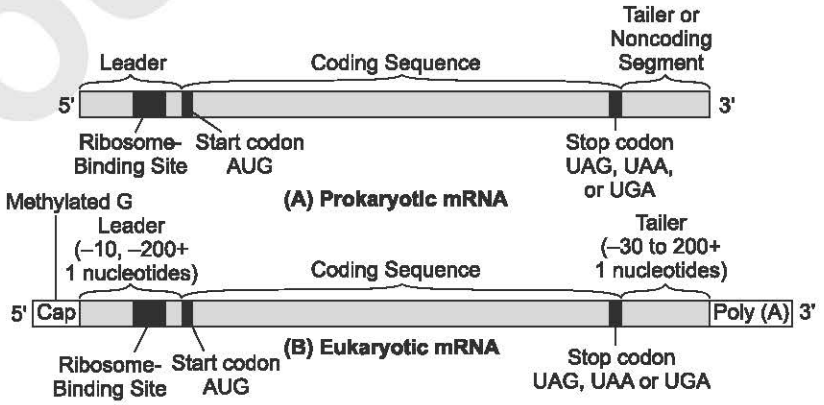


Fig. 4 : A fully processed *m*RNA with marked ends :
A. Prokaryotic *m*RNA; B. Eukaryotic *m*RNA

5. A poly-A tail of many adenine containing nucleotides.
6. A small **nontranslated or noncoding segment** may be present after the G-cap and before poly-A tail. The nontranslated sequence at 5' of *m*RNA is called the **leader**, because it precedes the start or initiation codon. The nontranslated sequence at 3' end is the **tailers** sequence. It follows the termination codon. Though, the leader and tailer sequences are not translated, their presence is essential for the translation of message coded in the coding region.

   The molecules of *m*RNA are heterogeneous because these occur in different sizes having different molecular weight. The heterogeneity defends upon two main factors :

   (i) The size and number of citrons, and

   (ii) The size of the protein molecule.

2. Ribosomal RNA (*r*RNA)

This is also known as insoluble RNA and occurs in the ribosomes which are nucleoprotein. It represents 75% to 80% of the total RNA. The 23 S and 16 S (Swedberg units) RNAs are typical of bacterial ribosomes. The molecular weight is 1.1×10^6 and 0.55×10^6 respectively. In animals the ribosomes are 80 S particles with 60 S and 40 S subunits. These subunits are composed of 28 S and 18 S RNA's. The 50 and 60 S subunits possess another type of rRNA with a sedimentation constant of 5 S. This contains only 115 nucleotides in comparison to about 1000 nucleotides in other types. The smaller type appears to be structural entity of 50 and 60 S ribosomal subunits.

*r*RNA differs in base content from *t*RNA and *m*RNA. It is relatively rich in guanine and cytosine. The base components in *r*RNA of *E. coli* has a molar ratio of adenine 21 : uracil 19 : guanine 36 : cytosine 23.

3. Transfer RNA (*t*RNA)

This represents 10-15% of the total RNA and has a molecular weight of 25000 (4S). *t*RNA is concerned with the binding of specific amino acids during protein synthesis. Therefore there must be at least one tRNA for each amino acid. Due to its small size, *t*RNA has been worked out for the nucleotide sequence. **Holley** (1966) succeeded in determining the primary structure of tRNA specific for the amino acid alanine. At present, it appears that all *t*RNAs are almost the same length (90 nucleotides) and all of them terminate in the sequence cytosine-cytosine-adenine. *t*RNAs from different sources are characterized by certain unusual and methylated purine and pyrimidine bases. Some of them are 6-N-dimethyladenosine, 1-methyladenosine, 6-N-isopentenyl-adenosine, 5-methylcytidine, 6-N-acetylcytidine, 2'-O-methylcytidine, 2-N-methylguanosine and pseudouridine, etc.

*t*RNA molecule consists of two strands of polynucleotides twisted around each other, forming a double helical structure. However, according to **Lake** and **Beeman** (1967) the tRNA molecule possesses three folds of double helix giving a clover leaf-like appearance.

In the *t*RNA molecule, some of the bases, are linked by weak hydrogen bonds while others are unpaired. The C-C-A sequence of nucleotides are concerned with the attachment of

activated amino acids during protein synthesis. At the opposite end of the *t*RNA are three unpaired bases forming the anticodon which recognizes the complementary bases in the *m*RNA. Further, there are two more sites for the recognition of ribosomes and specific amino acid activating enzymes.

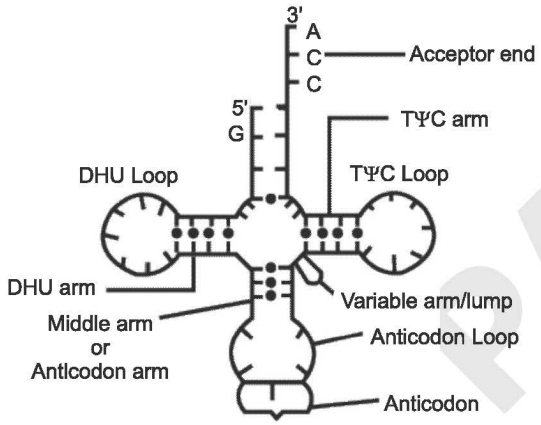


Fig. 5 : Clover leaf model of tRNA molecule from yeast.

Functions of RNA

As has been pointed out earlier, there are a number of different RNAs.

1. The rRNA or insoluble RNA plays an important and delinite role in protein synthesis. However, its exact role is not well understood.
2. The mRNA is complementary to the single strand of a DNA molecule. Soon after its formation, the mRNA leaves the nucleus and attaches itself to the 30 S ribosomes. Thus, it plays an important role in protein biosynthesis.
3. The tRNA also plays very important role in protein synthesis. This RNA recognizes the codons of the mRNA and brings amino acids to the ribosomes.

❑

UNIT-IV

Cell Cycle, Cell Division and Cell Signalling

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Describe the significance of meiosis in alternation of generation.

Ans. In animals, meiosis generates the haploid gametes-sperm and eggs directly. These signal cells fuse to form the zygote which develop into another diploid animal. In most plants meiosis and fertilization divide the life of the organism into two distinct phases or 'generations'.

Q.2. What are cyclin dependent kinase?

Ans. Cyclin-dependent Kinases (CDKs) are protein kinases characterized by needing a separate subunit—a cyclin, that provides domains essential for enzymatic activity. CDKs play important roles in the control of cell division and modulate transcription in response to several extra and intracellular cues.

Q.3. How does synaptonemal complex help in synapsis during meiosis?

Ans. The synaptonemal complex (SC) is a protein structure that forms between homologous chromosomes (two pairs of sister chromatids) during meiosis and is though to mediate synapsis and recombination during meiosis-I in eukaryotes.

Q.4. Describe different DNA damage checkpoints.

Ans. A DNA damage checkpoint is a pause in cell cycle that is induced in response to DNA damage to ensure that the damage is repaired before cell division resumes. Proteins that accumulate at the damage site typically activate the checkpoint and half cell growth at the G./S or G2M checkpoint.

Q.5. Write a short note on bim genes.

Ans. BCL-2 like protein 11 commonly called Bim gene, is a protein that in humans is encoded by the BCL-2L 11 gene.

Q.6. Define the kinetochore.

Ans. A kinetochore is a disc-shaped protein structure associated with duplicated chromatids in eukaryotes cells where the spindle fibres attach during cell division to pull sister chromatids apart.

Q.7. What is the role of P^{53} in cancer?

Ans. P^{53} also known as TP^{53} or tumor protein is a gene that codes for a protein that regulates the cell cycle and hence functions as a tumour suppression. It is very important for cell in multicellular organisms to suppress cancer.

Q.8. Define the microtubules in cell cycle.

Ans. Microtubules are major components of the cytoskeleton. They are found in all eukaryotic cells, and they are involved in mitosis, cell motility, intracellular transport, and maintenance of cell shape. Microtubules are composed of α and β-tubuliln subunits assembled into linear protofilaments.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Define the cytokinesis and explain the formation of cell furrow and cell plate at the end of cell division.

Ans.

Cytokinesis

The process of mitosis is characterised 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. So, division of one nucleus into two is often called **karyokinesis** and is followed by cytokinesis, which divides cytoplasm into two cells and can be brought about in two ways.

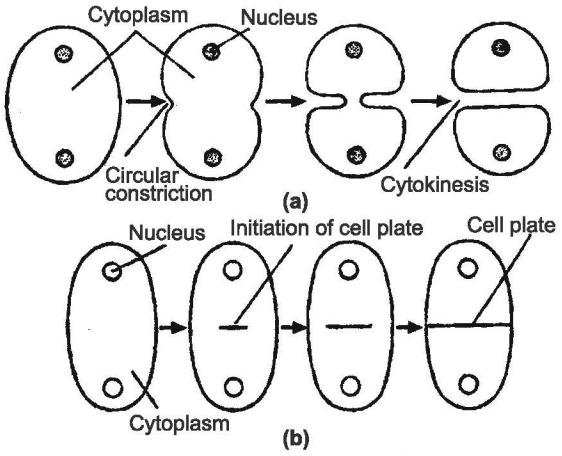


Fig. 1 : Two methods of cytokinesis : (a) Cytokinesis by cell furrow (b) Cytokinesis through formation of cell plate.

1. **Cell furrow :** In case of animals, outer layers are more flexible due to absence of cell wall. In such cases, a circular constriction appears at equator and it converges on all sides finally separating two daughter cells.
2. **Cell plate :** In plant cells, a more rigid cell plate is usually initiated at centre and is completed towards periphery. After the cell plate is laid down, primary walls are deposited on either side. The thick secondary cell-walls of cellulose may be laid down later on.

Q.2. Write a note on checkpoint of cell cycle.

Ans.

Checkpoints in Cell Cycle

During cell cycle, there are two transition points at which the cell can stop its continued progress in case certain conditions for progressive events fail to meet. These transition points are called **check points.** These checkpoints are :

(i) G_1—**S** transition point of G_1**-checkpoint.**

(ii) G_2—**M** transition point or **M-checkpoint.**

Exogenous growth factors influence G_1 cells to enter S-phase through the activity of ***cyclin dependent kinases.*** For example, if a cell is subjected to treatment that damages DNA, the cell cycle is arrested at G_1 stage until the damaged DNA is repaired.

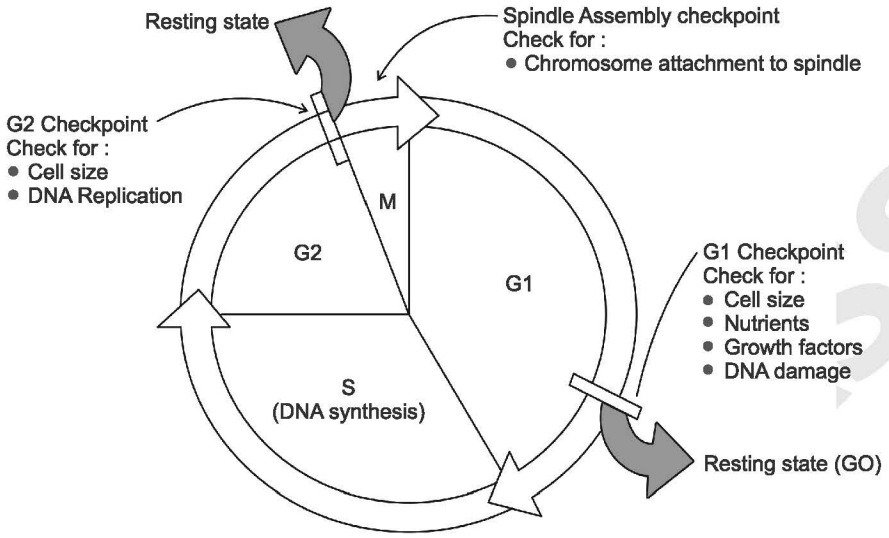


Fig. : Checkpoints in cells cycle.

Arrest of cell cycle at one of these checkpoints is achieved by **inhibitor proteins.** Their synthesis is stimulated by exogenous factors such as cell growth or DNA damage. There is synthesis of a number of such inhibitor proteins in cells that continue to stay in interphase and do not enter mitotic phase. Some of the inhibitor proteins act directly on cdk (-cyclin dependent kinase complex) to inhibit protein kinase activity. Some proteins inhibit DNA replication and still others as transcription factors that stimulate or repress transcription of those genes whose products are needed in cell cycle.

Importance of Checkpoints

Checkpoints provide the cell time to become fully prepared for the division and complete replication of DNA. Unchecked progress of an unprepared cell during cell cycle may lead to its death. Thus cessation of cell cycle is a protective response that keeps the cell from engaging in self destructive activity and ensures formation of normal daughter cells.

Q.3. What are the recombination molecules? Describe their structure and location in relation to their role in meiotic recombination.

Ans. The synaptonemal complex (SC) is though associated with all paired chromosomes at pachytene, its most likely function is perhaps restricted to meiotic synapsis and/or maintenance of the synaptic state. Another class of important structures, associated with paired pachytene chromosomes are **'recombination nodules',** which are believed to be involved in meiotic recombination.

A correspondence between meiotic exchange events and the numbers and locations of recombination nodules has been observed in a variety of materials suggesting the possible role of these structures in recombination.

In *Drosophila* females, two types of recombination nodules, **spherical** (larger in size) and **ellipsoidal** (smaller in size), have been reported. Association of an ellipsodial recombination nodule with a synaptonemal complex (SC) is shown in the given figure.

A number of *Drosophila* mutants, which are defective in recombination, were found to have normal synaptonemal complex, but were found to have changes in the number and morphology of one or both the types of recombination nodules. These observations suggested a positive role of recombination nodules in exchange of chromosome segments during pachytene leading to recombination.

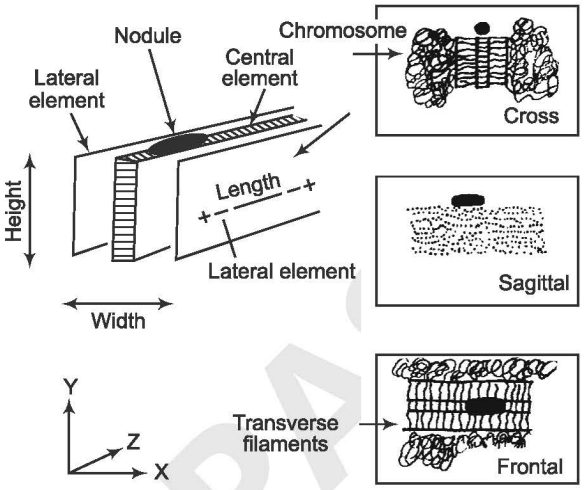


Fig. : An ellipsoidal recombination nodule associated with paired pachytene chromosomes in *Drosopila*.

Although data from many oragnisms firmly establish the correlations between recombination nodules and meiotic recombination (in number/nucleus, number/bivalent arm, distribution on bivalent arms, etc., their exact role in recombination is not clearly understood. A study of mutants defective for recombination suggested, that role of these nodules cannot be trivial.

These nodules in some way perform the following two distinct roles : (i) they help in choice of number and location of recombination sites and (ii) also help in the recombination itself, by performing enzymatic and supporting functions.

A study of mutants in *Drosophila* also suggested similar effects on both the ellipsoidal and spherical nodules, so that both of them look related, although the nature of relationship is not clearly known. According to one hypothesis, ellipsoidal nodules may be precursors of spherical nodules, since they appear and disappear earlier than the spherical nodules, although during some developmental stages (a part of pachytene) both are found.

Q.4. Describe the formation of chiasmata and crossing over.

Ans.

Formation of Chiasmata

The chiasmata are X-shaped figures formed between the homologous chromosomes during diplotene stage of prophase I of meiosis. The views of chiasma formation and behaviour are widely accepted now-a-days. According to **partial chiasma type theory,** proposed by **Janssen** (1909) and **Darlington** (1932), the chiasmata are formed by the breakage and reunion of two nonsister homologous chromatids. But according to **Sax** (1932), the chiasmata formation leads to the breakage of chromatids and their crossing over.

Mechanism of Crossing Over

During meiosis the chromatids break and rejoin and in doing so they exchange parts. This leads to crossing over and exchange of genetic material. According to **precocity theory** put forward by Darlington, during zygotene and pachytene, the undivided homologous chromosomes are relationally coiled about each other and the direction of their coiling is opposite to their internal coiling. The two types of coils are in physical equilibrium. When the chromosomes divide, the equilibrium is disturbed due to the reduction in force of internal coiling. A torsional stress presses each strand, which results in the breakage of one of the

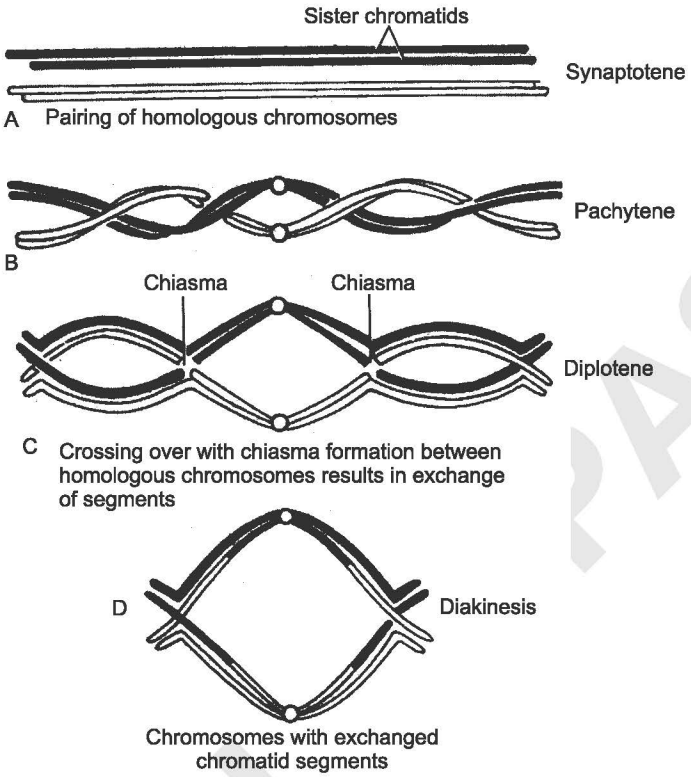


Fig. : Mechanism of chiasmata formation and crossing over.

weaker chromatids. Now the stress is released on the broken chromatid and is transmitted to the other chromatid. A second break then develops in the adjacent nonsister chromatid at the same locus. The broken ends separate and the nonsister chromatid pieces recombine to produce cross-over chromatids. But the serious objection to this theory is that the chromosomes are double even before synapsis. The recent theory of **copy choice** is widely accepted now-a-days which suggests the recombination of nonsister chromatids during DNA synthesis.

Q.5. What is the difference between plant and animal cytokinesis?

Ans. Differences between Plant and Animal Cytokinesis

S.No.	Plant cytokinesis	Animal cytokinesis
1.	It usually occurs by cell plate method.	It takes place by cleavage.
2.	The spindle usually persists during cytokinesis.	The spindle begins to degenerate soon after anaphase.
3.	Central part of spindle grows in size and forms an interdigited complex called phragmoplast.	A mid body of dense fibrous and vesicular material is formed in the middle.
4.	Vesicles derived from Golgi apparatus reach the equator of the phragmoplast and fuse to form cell plate and new cell membranes.	The event is absent in animal cytokinesis.
5.	Cell plate grows centrifugally.	Cleavage progresses centripetally.
6.	The new cell membrane is derived from vesicles of Golgi apparatus.	The new cell membrane is usually derived from endoplasmic reticulum.

Q.6. Write the differences between mitosis and meiosis division.

Ans.

Differences between Mitosis and Meiosis Division

S.No.	Mitosis	Meiosis
1.	It occurs in somatic cells.	It occurs in germ cells.
2.	Nucleus divides only once.	Nucleus divides twice.
3.	Two daughter cells are formed.	Four daughter cells are haploid.
4.	Daughter cells are diploid.	Daughter cells are haploid.
5.	It occurs more frequently.	It occurs less frequently.
6.	Daughter cells form somatic organs.	Daughter cells form gametes.
7.	There is only one prophase, one metaphase, one anaphase and one telophase.	There are two of each phase and five sub-phases in prophase-1.
8.	Number of chromosomes are not changed in the daughter cells.	Number of chromosomes are reduced to half.
9.	Chromosome number doubles at the beginning of each cell division.	Chromosome number is not doubled. It doubles after the end of first meiotic division.
10.	No crossing over in chromosomes.	Crossing over occurs chromosomes.
11.	Equation division.	Reduction division.

Q.7. Describe the role of Cdk inhibitor in the progression of cell cycle with suitable examples.

Ans.

Cdk Inhibitors (CKIs)

The recognising the pre-eminent role of Cdk-cyclin complex in cell division, this complex has also been described as **cell cycle's engine.** The Cdk subunit of this complex is inactive as a protein kinase without the cyclin subunit, which confers basal kinase activity to the Cdk. Both Cdk and cyclin have one or more amino acid residues (threonine or tyrosine, which can be phosphorylated and dephosphorylated (reversible phosphorylation).

The activity of Cdk is also regulated due to binding of inhibitors called cyclin dependent kinase inhibitors (CKIs). Several of the growth inhibiting signals, including the growth inhibitory factor TGF-β act through these cyclin dependent kinase inhibitors (e.g. Sic1; Far1, Rum1, p21, p27, p57, p16, p15, p18, p19). The protein p21 is a universal Cdk inhibitor and binds with Cdsk2, Cdk4 and Cdk6, suggesting that it inhibits progression through all stages of G1/S. The protein p27 has a sequence that is partly related to p21 amnd binds promiscuously to all Cdk-cyclin complexes. Both p21 and p27 bind and render the Cdk-cyclin complex incapable of being activated through phosphorylation by Cak. Another cyclin dependent kinase inhibitor is **rum1,** which also causes re-replication and results in concomitant accumulation of cdc18p.

Interaction of cdc6p, cdc18p and rum1 with Cdks

The proteins, cdc6p in budding yeast and cdc18p in fission yeast are important proteins that remain associated with Cdk and influence the cell cycle through their effect on DNA replication. It has been shown that cdc6p and **origin recognition complex (ORC)** nucleate **MCM proteins** to chromatin and thus help in DNA replication, since MCM proteins are absolutely necessary for the replication to proceed further (MCM = minichromosome maintenance).

Activity of cdc2/cdc13 complex (Cdk) is also influenced by another protein rum1 (replication uncoupled from mitosis), which regulates progression through G1 phase and thus controls

entry into S-phase. Overexpression of rum1, like cdc18p, also allows multiple rounds of replication to occur (endoreduplication leading to origin of polyploid cells), so that the cell fails to enter mitosis. When the gene rum1 is deleted, the cells enter mitosis prematurely suggesting than rum 1 is a Cdk inhibitor at M-phase. In other words deletion of rum1 allows the cells to undergo two successive mitosis without an intervening pre-start G-1 phase (S-phase has been divided into pre-start G1, start and post-start G1). While rum1 inactivates M phase Cdk (cdc2/cdc13), it simultaneously activates cdc18p by dephosphorylating it.

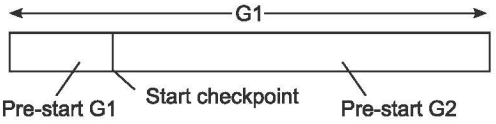


Fig. : G1 phase of the cell cycle in yeasts, showing pre-start G1, start and post-start G1 phases.

cdc18 ⟶ cdc18-p ⟶ cdc18

active (initiates S-phase) | (inactive) (M-phase initiated) | (active) (M-phase inhibited & S-phase initiated)

Q.8. Describe the structure of cytokine receptors.

Ans.

Cytokine Receptors and Nonreceptor Protein-Tyrosine Kinases

The cytokine receptors are similar to receptor protein-tyrosine kinases except that the protein-tyrosine kinase is not an integral part of the cytokine receptor but is associated with it. The cytokine ligand binds to the extracellular N-terminal cytokine binding domain of the receptor. The C terminal cytosolic domain is associated with **nonreceptor protein-tyrosine kinase** which is activated when ligand binds to the N-terminal domain of receptor.

When cytokine ligand attaches with two molecules of cytokine receptors, the nonreceptor protein-tyrosine kinases is first phoshorylated and then it phosphorylate the cytokine receptors. These receptors then send the signal downstream in the cytosol.

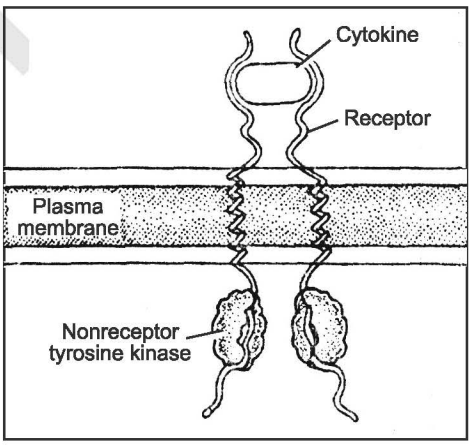


Fig. : Structure of a cytokine receptor and non receptor protein-tyrosine kinase.

Q.9. How are cell cycle and cancer interlinked?

Ans.

Cell Cycle and Cancer Interlinked

During 1980s and 1990s, it became clear that derangements in the cell cycle machinery may actually be responsible for uncontrolled cell growth, characteristic of cancer. Both active oncogenes and mutations for lack of function in any tumour suppressor gene may lead to cancer. Intrinsic defects in the cell cycle machinery may also cause cancer.

1. Cyclins and Cdks in Cancer

Cyclin D1 is one of the eight or more cylins known in mammals. If cyclin D1 is overproduced or produced at a wrong time, it would stimulate inappropriate cell divisions by keeping the

corresponding Cdk (Cdk4, CDk6) on, when it should be turned off. In 1991 the gene for cyclin D1 was found to be on and its amplification and over-expression were shown to cause a variety of cancers (breast, oesophagus, B cell lymphoma, etc.). Similarly, cyclin E and cyclin A ere found to be overexpressed in cancer cell lines. Some Cdks like Cdk4 have also been found to be amplified in some cancer cells.

2. Cell Cycle Inhibitors and Cancer

Loss of activity or function of some cell cycle inhibitors may also lead to cancer. These inhibitors include : (i) p53, which blocks are activity of Cdk2, and other Cdks, (ii) an inhibitor blocking specifically the Cdk4, and (iii) an inhibitor mediating TGF-β inhibitory effects.

3. Tumour Suppressors and Cancer

Two important tumour suppressor genes include the following : (i) p53 and (ii) *Rb* (retinoblastoma) gene. Protein p53 has actually been considered to be so important that it has been described as the **'guardian of the genome'** or a **'watchman',** so that the conditions like DNA damage, hypoxia, oncogene activation and virus infection are sensed by p53, which in its turn either arrests the cell cycle, or cause cancer. p53 is one of the commonest mutated (inactivated) gene in almost 50% of all human cancers.

4. Aneuploid and Cancer

A defect in spindle assembly checkpoint has also been found to cause aneuploidy associated with tumours (reported in 1998). One such defect was a mutation in *hBUB*1 (h stands for human), a gene involved in spindle assembly checkpoint, so that the anaphase separation will be disrupted causing aneuploidy. It is possible that the missing chromosomes may carry tumour suppressor gene(s), whose absence may lead to uncontrolled cell division.

Q.10. Describe structure and function of receptor protein-tyrosine kinases.

Ans.

Receptor Protein-Tyrosine Kinases (RTKs)

These receptors are transmembrane proteins consisting of an **N-terminal** extracellular **ligand binding domain** present on the surface of cell membrane, a signal transmembrane **α-helix,** and a cytosolic **C terminal domain** with **protein-tyrosine kinase enzyme activity.** When ligand binds to the exctracellular domain, the cytosolic kinase domain is activated and causes phosphorylation of itself and the intracellular target protein. These are receptors for most growth factors.

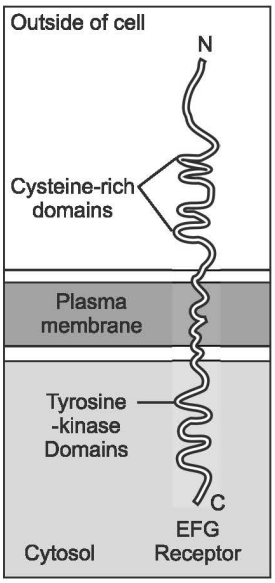


Fig. : Structure of receptor

Functions of RTKs

RTKs are a a sub-class of tyrosine kinase that are involved in cell to cell communication and controlling a wide range of complex biological functions, including cell growth, motility, differentiation and metabolism.

Q.11. Justify the dependence of mitosis on DNA replication and DNA repair. How common is DNA damage and repair within a cell?

Ans.

Dependence of Mitosis on DNA Replication and DNA Repair

Within a cell, DNA damage and DNA replication checkpoints are also available, so that the cells, which suffer either DNA damage or/and a block in DNA replication, exhibit following responses : (i) cell cycle arrest in G1, S and G2; (ii) slowing of DNA replication and (iii) increase in transcription of genes meant for DNA repair and DNA replication. These responses involve **signals, sensors, transducers** and **effectors.**

1. DNA Replication Checkpoint : The Licensing Concept

Important information on the control of DNA replication came from experiments involving cell fusion, among human HeLa cells. On the basis of these cell fusion studies and other studies, a **'licensing factor'** model has been proposed, according to which, after the disassembly of nuclear envelope, the **replication licensing factors (RLFs)** bind to chromatin. This makes the chromosomes competent to replicate. Once the replication is complete, these RLFs would be destroyed thus preventing further found of DNA replication. The MCM proteins also have been implicated in rendering the cells competent for replication and in limiting DNA replication to one per cycle. RLF-M has been shown to contain some MCM proteins. The nucleation of MCM proteins is facilitated by the protein cdc6p/cdc18p, as mentioned in the previous section.

2. DNA Damage Checkpoint

The DNA damage during cell division often leads to arrest of cell division either before DNA replication in G1 (G1 checkpoint) or before mitosis in G2 (G2-M checkpoint). This response to DNA damage, leading to arrest of cell divsion is facilitated by regulation of Cdk-cyclin complexes, using checkpoint proteins. The checkpoints sense the DNA damage and transduce an inhibitory signal.

We know that in yeast, cdc2 protein remains inactive due to phosphorylation and is activated by cdc25 (a phosphatase) through dephosphorylation. At the DNA damage checkpoint, DNA-damage leads to sequestration of cdc25, so that cdc25 will not be available for activation of cdc2-P (tyr^{15}) and the cell division will be arrested at G2-M transition.

Q.12. Write a short note on apoptosis.

Ans.

Apoptosis

Initially, during 1980s, research in cell division cycle suggested that the cell cycle is regulated mainly by the timely synthesis of several regulatory proteins like Cdks, cyclins, CDk inhibitors, Cakes, phosphatases and some unknown non-cyclin proteins. However, later during mid-1990s, there was a major shift in emphasis towards the study of timely destruction of key proteins (proteolysis) during the progress of cell cycle. The proteolysis was also found to be facilitated by the protein **ubiquitin** which forms a **26S proteosome** with the target proteins to be destroyed.

Proteolysis actually involves degradation of the following proteins at three checkpoints; (i) cyclins, (ii) inhibitors of the activity of CDKs and that of its components, (iii) a putative protein

that is a chromosome tether and holds chromatids together, it needs to be degraded before chromatids separate. Three enzymes called E1, E2 and E3 (letter E was used, because these were first defined as eluates from ubiquitin affinity column) are known to be involved in the attachment of ubiquitin-to the protein that needs to be degraded. While E2 remains active throughout the cell cycle, E3 has a temporal control (temporal control means it will be active at a specific time). A large E3 complex known as **cyclosome** or the **anaphase-promoting complex (APC)** is also needed for proteolysis that is essential for anaphase progression in many organisms.

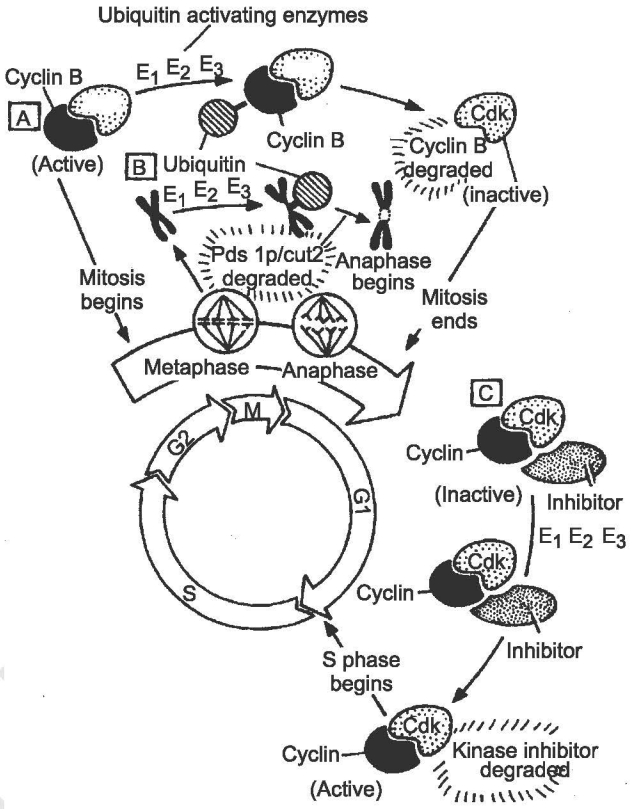


Fig. : Ubiquitination of cell cycle proteins with the help of three enzymes, (E_1, E_2, E_3) as a pre-requisite for degradation.

Apoptotic and Autophagic PCD

Programmed cell death (PCD) can be of the following major types : (i) Apoptosis or Type I cell-death, and (ii) Autophagic or Type II-cell death (cytoplasmic; characterized by the formation of large vacuoles, which eat away organelles in a specific sequence prior to the nucleus being destroyed). Besides these two major types of PCD, other pathways have been descovered, called **"non-apoptotic PCD"** (or "caspase-independent programmed cell-death" or "necrosis-like PCD").

These alternative routes to death are as efficient as apoptosis and can function as either backup mechanisms or the main type of PCD.

Other forms of PCD include (i) **anoikis,** almost identical to apoptosis except in its induction; (ii) **cornification,** a form of cell death exclusive to the eyes; (iii) **excitotoxibity** and (iv) **Wallerian degeneration.** Plant cells undergo particular processes of PCD, which are similar to autophagic cell death. However, some common features of PCD are highly conserved in both plants and metazoa.

Q.13. Write a short note on ion channels.

Ans. Ion-channel-linked Cell Surface Receptors

These cell-surface receptors are each linked to an ion channel, the conductance of which is modulated by the binding of an agonist or antagonist. Ion channels were discovered in 1950, and represent a diverse family of integral membrane proteins (more than 100 types of ion

channels are known), each ion channel being a single channel protein. These ion channels mediate ion fluxes across cellular membranes, and influence a variety of essential biological processes including the following : cell volume regulation, swimming behaviour in unicellular organisms like paramecium, movement of stomatal pores, leaf closing response of mimosa plant, muscle contraction, excorine cell secretion, excitation of neurons for signal transmission, etc.

In many cases, these biological processes depend on fluxes of specific ions that are stimulated and regulated by intracellular and extracellular signals. However, all ion channels are not associated with cell surface receptors, so that when associated with receptors, these are described as ion-channel linked receptors. In these receptors, the receptors perceive the stimulus and then use the associated ion channels for signal transduction through ion flux.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. What do you understand by cell cycle? Describe all those events that occur during interphase of cell cycle that prepare the cell for nuclear division.

Ans.

Cell Cycle

The growth and development of every living organism depends on the growth and multiplication of its constituent cells. In unicellular organisms, cell division is the means of reproduction, which produces two or more new individuals from the mother cell. On the other hand, multicellular organisms develop from a single primordial cell, the **zygote,** which also comes from the preceding cells. **Negali** pointed out that new cells are always formed through the division of pre-existing cells. **Virchow** (1885) supported the above theory and investigated the process of cell division.

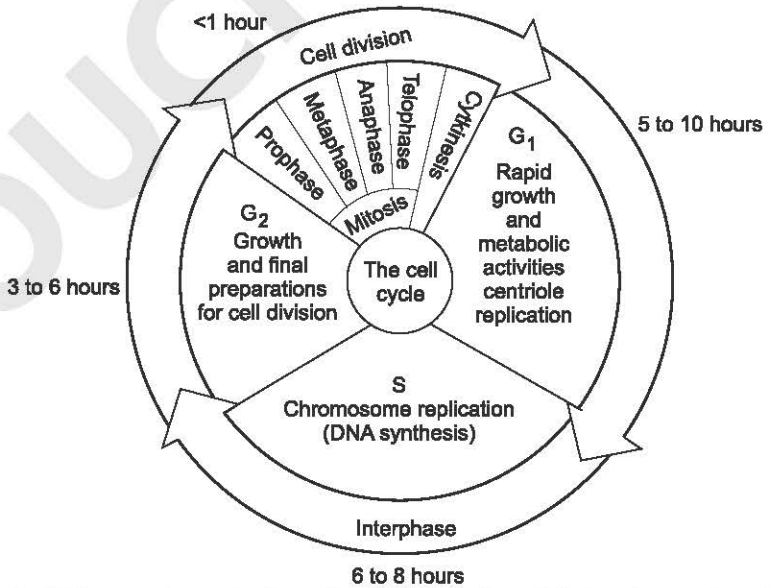


Fig. 1 : Different phases in the cell cycle of a cell and their relative duration.

The life cycle of a cell begins with its formation as daghter cells from mother cell at the end of telophase. These daughter cells are smaller than the parent cell. Moreover, their DNA content is just half of the parent nucleus. These daughter cells grow in size by synthesising new cytoplasmic and nuclear material till the total volume of each cell becomes four times of its original volume and the DNA content gets doubled by the replication of DNA. After this the cell is again ready to undergo division.

Therefore, growth and division occur alternating in a cyclic fashion. This cycle is known as **cell cycle.** It involves two distinct phases :

1. Growth phase or non-dividing phase or interphase
2. Division phase or Mitotic Phase : M-Phase

1. Growth Phase or Interphase

Interphase is the interval between two successive cell divisions. During this period, the cell prepares itself for the next division by synthesising and storing all those substances which are essential for cell division. This period is also called **preparatory phase.** Cells pass major part of their life cycle in this phase. Nondividing cells remain permanently in this stage.

During growth phase, the biosynthetic activities of the cells are at their maximum. Because of this, growth phase is also called **metabolic phase** or **biosynthetic phase.** Based on the difference in biosynthetic activities, growth phase is divided into three substages :

(i) G_1-Phase (First Growth Phase) : During this period the young daughter cell grows in size by synthesising cytoplasm. It synthesises and stores enzymes, mRNA, tRNA, ribosomes and proteins, etc. and also nitrogenous bases needed for DNA replication. During this phase chromosomes are extended into slender fine threads forming an interwoven chromatin network.

The duration of G_1 phase is most variable. It takes about 30-40% time of the complete cell cycle. In mammalian cells whose life cycle is of about 16 hours duration, G_1 phase is of about 5 hours. In non-dividing mammalian cells (for example, lymphocytes of blood or nerve cells) the cell cycle stops at a point in this phase. This period is called G_0-phase. When such a cell prepares to divide, it enters G_1-phase.

(ii) S-substage or Phase of DNA Synthesis : This stage is characterised by (a) the replication of DNA and (b) synthesis of histones (proteins) which are associated with DNA. It takes about 30-50% time of cell cycle. The S-phase in vertebrate cells is about 6-8 hours duration. If DNA replication does not take place within 24-28 hours after division, the cell enters G_0 phase and does not undergo division.

(iii) G_2-substage (Second Growth Phase) : During this phase, the nucleus has double amount of DNA. The nuclear volume increases due to synthesis of ribosomal RNA, messenger RNA and nucleolar RNA. The cells synthesise proteins required during cell division. This phase is only of 10-20% duration of the cell cycle.

2. Division Phase or Mitotic Division

This is the last phase of cell cycle. It is also called **M-phase.** It gives rise to two daughter cells. This phase is divided into prophase, metaphase, anaphase and telophase.

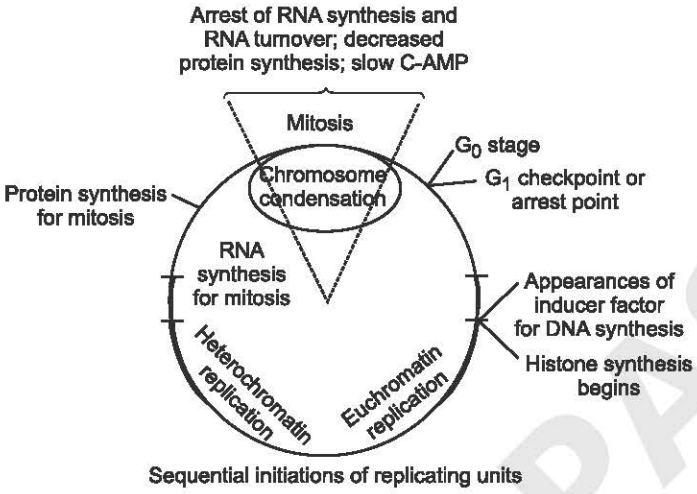


Fig. 2 : Molecular events during different phases of cell cycle.

Q.2. Describe the process of meiosis. What is its significance in sexually reproducing organisms?

Ans.

Meiosis or Meiotic Division

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 (fertilisation) of male and female gametes and the cells derived from it are known as **diploid** and their chromosome number is symbolised by 2*n*. The two similar chromosomes of a diploid cell are known as **'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.

So, Meiosis is a specialised and rather complicated type of a cell division, occurring only in the diploid reproductive cells and results in the formation of haploid **sex-cells** or **gametes.**

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. The cells undergoing meiosis are known as **meiocytes.** In animals, the meiocytes are the **primary spermatocystes** 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.

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 is a simple mitotic division in which the two haploid cells formed as result of heterotypic division divide again forming four daughter cells, each with haploid number of chromosomes. Each of the two meiotic cell divisions is further distinguished into phases. These are—prophase, metaphase, anaphase and telophase.

A. First Meiotic Division or Reduction Division

The following phases are included under the meiotic division :

1. First Prophase

The prophase of first meiotic division is of longer duration and profoundly modified. It is distinguished into following five phases or substages—Leptotene, zygotene, panchytene, diplotene and diakinesis.

(i) Proleptotene : The meiocyte 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.

(ii) 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

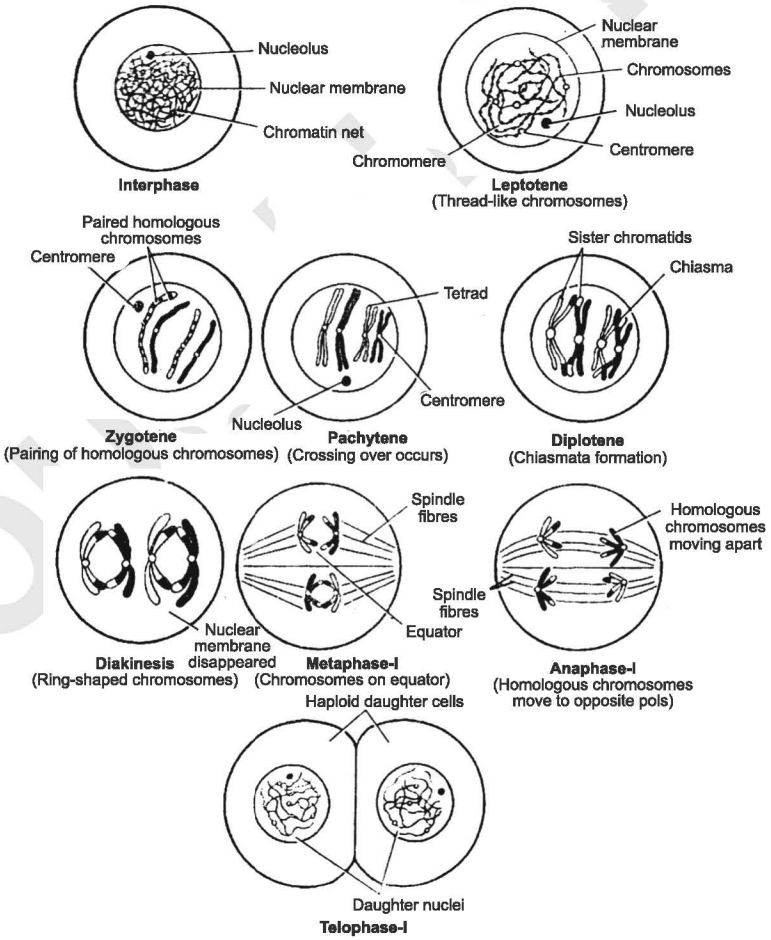


Fig. 1 : Different stages in the first meiotic division.

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 swellings called **chromomeres.** The chromomeres are of different sizes and colour 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.

(iii) Zygotene or Zygonema : The zygotene commences with the movement of chromosomes. It is affected by the forces of attraction between the two homologues 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 homologues 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 :

(a) The two homologues start pairing at the ends and then the pairing porgresses towards centromere region—**proterminal synapsis.**

(b) The pairing may start near the centromere and then progresses towards the ends—**procentric synapsis.**

(c) The pairing starts at random either at one point or at many points simultaneously—**random synapsis.**

In organisms with definitely oriented or polarised chromosomes, pairing usually commences at the ends nearest the nuclear membrane and progresses onwards till completion. This peculiar state of orientation, polarisation 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 pairing of homologues is due to the retardation or cessation of metabolic activities to the cell.

At zygotene the nucleolus increases in size and the centrioles move apart initiating the spindle formation.

(iv) 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 eah other forming relational coils. Each chromosome starts splitting into two sister chromatids by a vertical or longitudinal furrows. 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 nonsister 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.

(v) 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 chromosomes uncoil and start separating. But the separation is none the less incomplete since the homologues are in contact at one or more points where the crossing over has already taken place. These points of contact are known as **chiasmata (sing. chiasma,** meaning **cross**) which present cross-shaped appearance. The chiasma is the morphological equivalent of genetic crossing over. The number of chiasma in a bivalent varies in the same pair of chromosome 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 is complete, the homologous chromosomes are in contact by terminal chiasmata. The degree of terminalization is generally expressed as **coefficient of terminalization (T).**

$$T = \frac{\text{Number of terminal chiasmata}}{\text{Total number of chiasmata}}$$

The average number of chiasmata is bivalent is known as **frequency of chiasmata** (Fq.)

$$\text{Frequency (Fq)} = \frac{\text{Total number of chiasmata}}{\text{Total number of bivalents}}$$

According to **Darlington,** two types of repelling forces operate on the chromosomes at diplotene. One of the forces is electronegatively charged and operates on the surface of the chromosome throughout its length and the other with electropositive charge is localised on the centromere. The former controls the repulsion of the chromosomes and the latter causes distal movement of the chiasmata.

(vi) Diakinesis : The bivalents still contact and get thickened into deeply stained bodies. These migrate to the periphery of the nucleus. The two chromatids of each chromosomes 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.

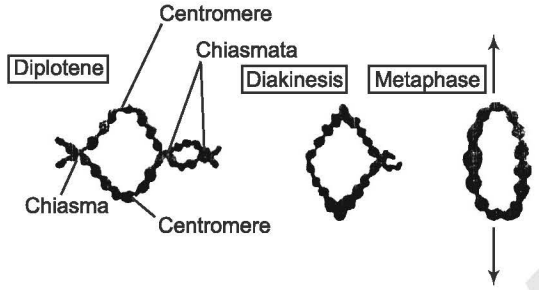


Fig. 2 : Diagram showing terminalisation in homologous chromosomes during diplotene and diakinesis.

At this stage the nucleolus and nuclear membrane disappear and the formation of nuclear spindle starts.

2. 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 amphiaster 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.

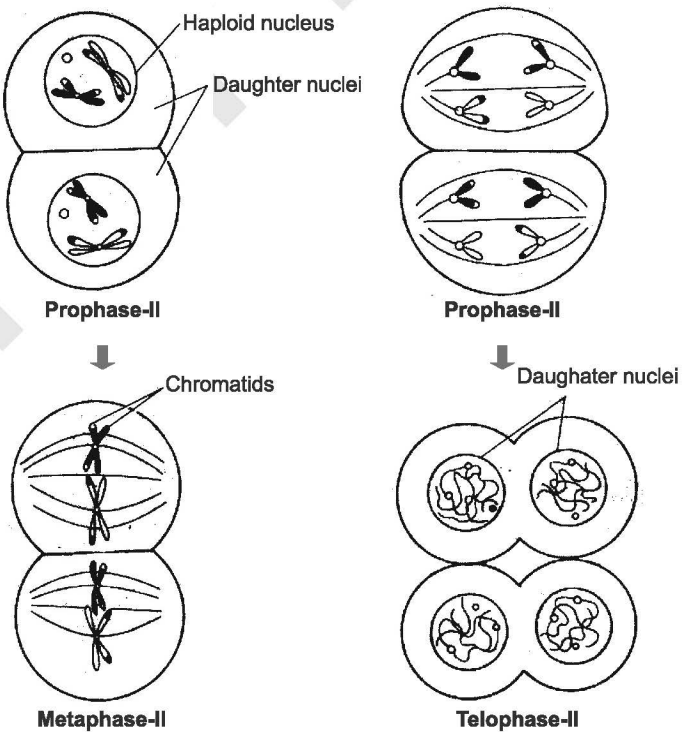


Fig. 3 : Diagrammatic representation of homeotypic division of meiosis.

3. Anaphase-I

During this stage, the bivalents move apart towards the opposites pole 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 point of centromere, so that they present V-shaped appearance.

4. 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.

5. 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. Second Meiotic or Homeotypic Division

The second meiotic division is essentially a meitosis, occuring independently in both the haploid sister cells. It may follow immediately after first meiotic division or may not occur until much later.

1. 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. 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. Anaphase-II

The chromatids with their independent centromeres form 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.

4. Telophase-II

The chromosomes at each pole uncoli and thin out to form the nuclear net. Each group gets surrounded by a nuclear membrane. Nucleolus reappears. Thus two nuclei are recognised in each cell. This is soon followed by cytokinesis and two cells are formed from each haploid daughter cell. Thus, as a result of meiosis four cells are produced, each with a haploid set of chromosomes, *i.e.,* each contains just one member of each homologous pair.

Significance of Meiosis

The significance of meiosis is threefold :

1. 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 the only means for restoring the chromosome number characteristic of the species.

2. 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 fertilisation. These are haploid cells having only one member of each homologous pair.
3. Meiosis results new combinations 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 ensures genetic variations in the population. The inherited variability leads to the evolution of organisms.

Q.3. What is synaptonemal complex (SC)? Describe its structure in detail and discuss its functional role in meiotic chromosome pairing.

Ans.

Synaptonemal Complex

Moses in 1956 first discovered synaptonemal complex (SC), a feature of meiotic prophase. Synaptonemal complex is a tripartite structure usually found between the two paired homologous chromosomes of each bivalent in all animal and plant nuclei undergoing meiosis. This is considered to be a physical structure which is associated with synapsis of homologous chromosomes. Complete synaptonemal complexes are seen at zygonema in the region of pairing. At pachynema these complexes are even more conspicuous.

1. Structure of Synaptonemal Complex

The synaptonemal complex is a configuration in which the 10 nm fibres of the chromosomes are arranged into a superstructure that can be viewed under the electron microscope. It is composed of three parallel, electron dense elements that are separated by less dense areas. The two **lateral elements** seem to be composed of fibres that are slightly wider than 10 nm and are called **synaptomeres.** They vary in structure at different stages of meiotic prophase I within a species. The **central element** is a ladder like configuration in the centre of the SC. In some species it is comparatively more pronounced. The **transverse elements** are electron-dense filaments that interconnect the central element with the lateral elements. The lateral elements may be spaced at a distance ranging from 20 nm to 30 nm to as much as 100 nm to 125 nm. Cytochemical studies have demonstrated that the lateral elements are rich in DNA, RNA and proteins, but that the central element contains mainly RNA, protein and little DNA.

Sometimes synaptonemal complex like structures are also found which are not associated with synapsed chromosomes. They show some similarities with the true

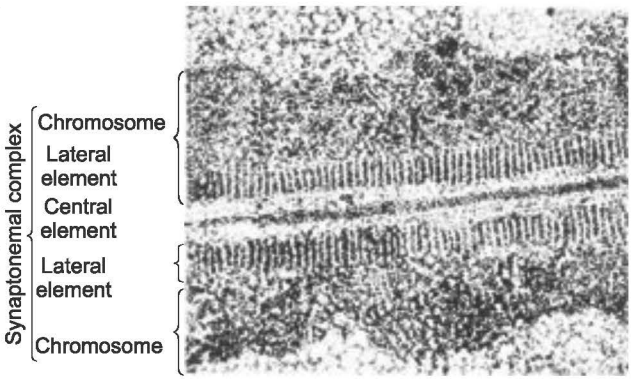


Fig. 1 : A bivalent with a synaptonemal complex showing central element and lateral elements.

synaptonemal complex. The lateral elements of these anomalous complexes may differ in size and density from those in the true synaptonemal complexes of that species. However, the true autosomal synaptonemal complex is always a unit tripartite complex but anomalous complexes are often multiple stacks of alternating lateral elements and central elements.

2. Formation of Synaptonemal Complex and Meiotic Pairing of Chromosomes

In 1970, **King** presented a hypothesis for the formation of synaptonemal complex. The hypothesis is known as **'The Synaptomere Zygosome Hypothesis'**. According to this hypothesis, there are structures called synanptomeres which are coiled polynucleotide segments scattered along the length of a pair of synapsed chromosomes lying in close proximity to one another. Each synaptomere is composed of three segments : A, B and C. The lateral segments of the synaptomeres (A and C) pair with the respective segments of the adjacent synaptomeres. The B segments are directed towards the central element and the sites where the so-called **zygosomes** are attached. Zygosomes are rod shaped sub-units, assembled in the nucleoplasm and are each visualized as protein molecules having a folded head by which they can attach to the central segment (B) of the synaptomere. The tail ends of the zygosomes contain charged sites that are represented by four dots. These charges allow the zygosomes to bind laterally with adjacent zygosomes in a ladder-like fashion.

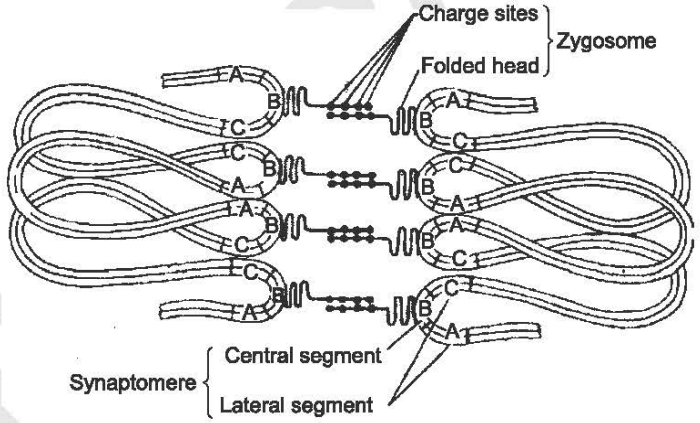


Fig. 2 : A diagrammatic representation of the synaptomeres and the zygosomes in a bivalent having two paired homologous chromosomes, to explain synaptomere-zygosome hypothesis.

The exact nature of events that lead to synapsis is still a subject of debate. A strong group of investigators believe that homologous chromosomes are prepared for synaptic pairing by the attachment of their telomeres to so-called **'attachment sites'** on the nuclear envelope. This is believed to be followed by attachment of zygosomes to central segments of synaptomeres and to adjacent zygosomes. When each synaptomere has a zygosome, pegs extend from chromosome folds and pairing extends in a zipper like action.

3. Function of Synaptonemal Complex

The appearance and disappearance of the synaptonemal complex coincide with the stages of meiosis in which pairing and recombination occur. This has led to the interpretation that they are functionally related. In leptonema, before pairing, single elements of snaptonemal complex are observed. Complete synaptonemal complexes are such at zygonema in the region of pairing.

Several pieces of evidence indicate that the synaptonemal complex is more directly related to the process of recombination. Some evidence, for example, is provided by the action of

inhibitors of DNA synthesis. At meiotic prophase, there is a small amount of DNA synthesis which, if inhibited can arrest the function of the synaptonemal complexes.

The synaptonemal complex has been interpreted as a protein framework that permits the proper alignment of the homologous chromosomes. However, since recombination by crossing over occurs at molecular level, it is necessary to assume that DNA fibres of the paired chromatids should reach the central component of SC within a distance of at least 1.0 μm for the recombination to take place. At diplonema the synaptonemal complex is shed from the bivalents with the exceptions of the regions in which the repelling homologues are held together each by a chiasma. Thus, a chiasma contains a piece of synaptonemal complex that will ultimately disappear and will be replaced by a chromatin bridge.

Q.4. Describe cell Signalling and its significance. Discuss various modes of cell signalling or cell communication.

Ans.

Cell Signalling

Cell Signalling is the phenomenon in which cells receive and respond to signals or chemical messages from their environment and from the neighbouring cells. Bacteria as well as prokaryotic and eukaryotic cells show cell to cell communication. This is achieved by a variety of signalling molecules secreted on the surface of one cell that bind to the receptors on the surface of plasm membrane of other cells. The stimulation arising from these contacts is converted to intracellular signals. These signals are then transmitted in the cell cytoplasm along diverse pathways and finally activate specific enzymes initiating a series of intracellular reactions that regulate virtually all aspects of cell activities.

Steps of Cell Signalling Transduction

The process of cell signalling typically includes :

1. **Stimulus or Signalling Molecules :** The stimulus is a molecule secreted into the extracellular space by some other cell that binds to the receptor at the outer surface of responding cell. The stimulus can also originate as a result of contact by another cell or by a non-cellular substrate. Such a stimulating agent is called **ligand.** The signalling molecules may be **hormones** secreted by endocrine cells and transported to the cells of target organ, neurotransmitters, nitric oxide, carbon monoxide, growth hormones and peptide hormones, plant hormones and eicosanoids.
2. **Surface Receptor :** Receptor is a transmembrane protein, which receives the stimulus on the exocytosolic or extracellular surface of plasma membrane. It remains embedded within the membrane. The main surface receptors are G-protein-coupled receptors, receptor protein-tyrosine kinases, cytokine receptors, non-receptor protein tyrosine kinases and receptors-linked to other enzymatic activities.
3. **Pathways of Intracellular Signal Transduction :** The signal received by the receptor on the outer surface of plasma membrane is transferred to its cytoplasmic surface. This is called **signal transduction.** The nature of stimulus received by the cell surface receptor is entirely different from the signal that is released to the cell cytoplasm.

   During signal transduction, the signal passes along a series of distinct proteins that form **signal transduction** pathway. In a signal transduction pathway, one protein changes the activity of the next protein in the series by adding of removing phosphate groups. These proteins are **kinases** and **phospatases.**

Following pathways are actively involved in signal transduction :

(i) Phospholipids and Ca^{2+}
(ii) NF-kB signalling
(iii) MAP kinase pathways
(iv) Cyclic GMP second messenger pathway
(v) Hedgehog, Wat, and Notch pathways
(vi) P13-kinase or Akt and mTOR pathways
(vii) cAMP pathway or second messengers and protein phosphorylation pathway.

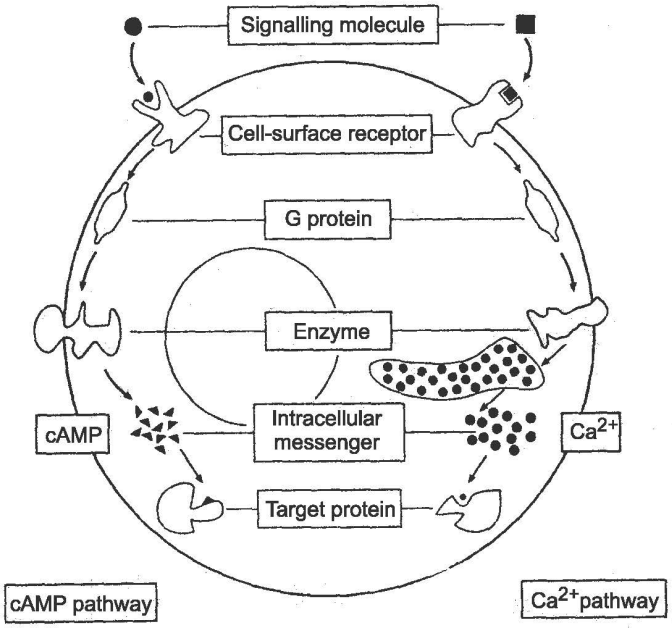


Fig. 1 : Summary of events occuring in a cell due to intracellular signals in response to an extracellular stimulus.

4. **Transmission of Signal to the Effector :** From the inner surface of plasma membrane, the signal is transmitted to the specific effector molecule present within the cell cytoplasm or to the nucleus. Here, the signal triggers cell's response. The cell response might involve a changed gene expression, an alteration of the activities of metabolic enzymes, a reconfiguration of the cytoskeleton, a change in the permeability or activation of DNA replication.

   The signals that are directly transduced to components of cytoskeleton cause their alternations and result in cell movement or change in cell shape. **Integrins** are the major receptors responsible for attachment of cells to the extracellular matrix, cell-cell junctions and also serve as receptors that activate intracellular Signalling pathways.

5. **Inactivation of Signalling Molecule :** At the end, the signal molecule is either inactivated or destroyed. This results in the cessation of the response.

Modes of Cell Signalling

Cell Signalling in multicellular animals can result from **cell-cell Signalling** or from the **action of secreted Signalling molecule :**

1. **Cell-Cell Signalling :** Cell to cell Signalling occurs from direct interaction of cell with its neighbouring cells. Therefore, it is also called **'communication through contact Signalling'.** It is found in cells whose plasma membranes are in direct contact. The Signalling molecules present on the plasma membrane of one cell or its matrix influence other cells in direct physical contact. The variety of receptors on the surface of plasma membrane interact with the Signalling molecules present on the surface of neighbouring cells.

   Cell-cell Signalling plays a critical role in regulating the behaviour of cells in animal tissues. For example, the integrins and cadherins that function as adhesion molecules also act as Signalling molecules. They regulate cell proliferation and survival in response to cell-cell and cell-matrix contacts. The cell-cell Signalling plays an important role during embryonic development.

2. **Signalling by Secreted Molecules :** The cell communication through multiple varieties of Signalling by secreted chemical molecules are divided into following categories :

   (i) Endocrine Signalling : In endocrine Signalling, the specialized endocrine cells secrete **hormones,** which travel through blood stream and influence target cells that are distributed throughout the body. The steroid hormones pass through the plasma membrane of target cells and travelling through the cytoplasm, reach the nucleus and activate genes. In animals, more than 50 different hormones are produced by endocrine glands, including the pituitary, thyroid, parathyroid, pancreas, adrenal and gonads.

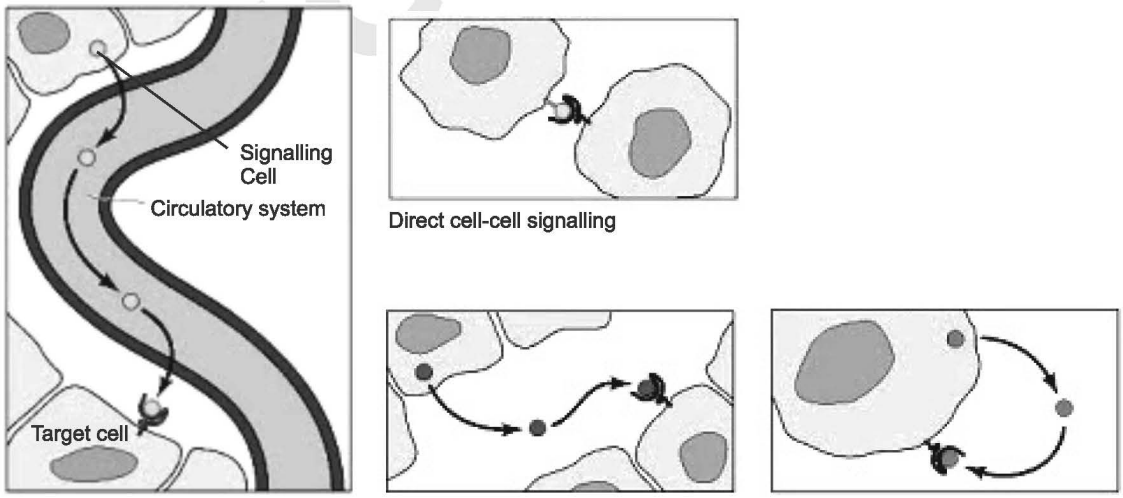


Fig. 2 : Different modes of cell Signalling : A. Endocrine Signalling, B. Paracrine Signalling and C. Autocrine Signalling.

(ii) **Paracrine Signalling :** The paracrine cells secrete **local chemical mediators,** which are taken up rapidly by neighbouring cells destroyed or immobilised. The mediators act on cells in the immediate environment.

(iii) **Synaptic Signalling :** It is paracrine Signalling, confined to nervous system. These cells secrete **neurotransmitter** at synapses. The neurotransmitter diffuses across the synaptic cleft and acts on the postsynaptic target cell.

(iv) **Autocrine Signalling :** In autocrine Signalling, cells respond to Signalling molecules that they produce themselves. For example, cells of vertebrate immune system respond to foreign antigens by producing growth factor which stimulates their replication.

During chemical Signalling, the target cells respond to chemical signal by means of specific proteins which act as receptors. These receptors bind the Signalling molecule and initiate the response.

Q.5. Summarise various pathways of intracellular signal transduction. Discuss significance of second messenger in signal transduction.

Ans.

Pathways of Intracellular Signal Transduction

The chain of reactions that transmits signals from the cell surface receptors to a variety of intracellular target is called **intracellular signal transduction.** The transcription factors are the targets of such Signalling pathways and thus, regulate gene expression. Thus, the intracellular Signalling pathways act to connect cell surface to the nucleus, leading to change in gene expression in response to extracellular stimuli. Some important pathways are :

1. cAMP Pathway

In the transduction of cell signals, **cyclic adenosine monophosphate** (cyclic AMP or cAMP) acts as a **second messenger.** It is released into cell interior as a result of binding of first messenger (a hormone or some other ligand) to a receptor at the outer surface of the cell. This was discovered by **Earl Sutherland,** his colleagues, **Edvin Krebs** and **Edmond Fischer.** The first messenger binds exclusively to only one type of receptors, whereas the second messenger is able to activate a variety of cellular activities. The use of second messenger enables cells to mount a large scale coordinated response. For example, cAMP stimulates glucose mobilisation by activating a protein-kinase.

Mode of Working of cAMP in Transduction of Cell Signal for Glucose Mobilisation : Cyclic AMP is formed from ATP by the action of an integral membrane protein called **adenylyl cyclase.** The catalytic domains of this protein reside at the inner surface of plasma membrane. Adenylyl cyclase is called effector because it brings about the cellular response by synthesising cAMP. The various steps in the induction of response by cAMP in glucose mobilisation are as follows :

(i) Binding of ligand, *i.e.,* the hormone glucagon or epinephrine to the outer surface of a liver cell causes a change in the conformation of hormone receptor.

(ii) The stimulus is transmitted across the plasma membrane and activates protein adenylyl cyclase at the inner surface of plasma membrane.

(iii) The activated adenylyl cyclase catalyses conversion of ATP into cAMP molecules. These readily diffuse into the cell cytoplasm. Each cAMP triggers a chain of reactions

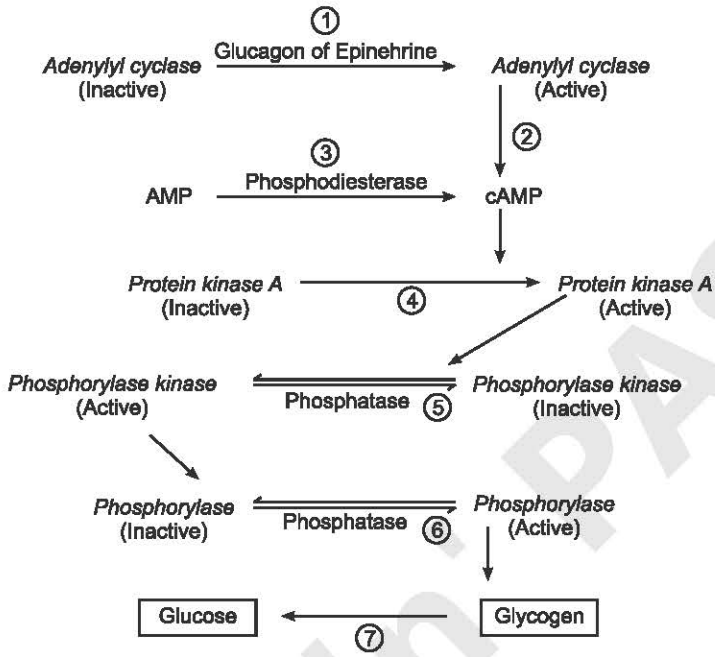


Fig. 1 : Reaction cascade triggered by glucagon or Epinephrine in the mobilisation of glucose.

which mainly include the modification of one enzyme by another. This is called **reaction cascade.**

The first step in the reaction cascade starts when hormone binds to receptor and stimulates formation of cAMP.

(iv) cAMP molecule binds to an allosteric site on a regulatory subunit of a specific cAMP-dependent protein-kinase, called **protein kinase A** (PKA). The regulatory subunits (R) normally inhibit the enzymatic activity of the catalytic subunits (C) of the enzyme. cAMP binding causes dissociation of inhibitory subunits, and the catalytic subunits become active. During glucose metabolism in liver cells, phosphorylation of *glycogen synthetase* inhibits its catalytic activity and thus prevents conversion of glucose to glycogen but induces synthesis of glycogen. On the contrary, phosphorylation of *phosphorylase-kinase* activates the enzyme to catalyse transfer of phosphate groups to phosphorylase molecules.

(v) Activation of phosphorylase stimulates breakdown of glycogen.

(vi) Glucose-1-phosphate formed in phosphorylase reaction is converted to glucose.

Significance of Second Messenger

Occurrence of second messenger like cAMP or Ca^{2+} ions in vertebrates and presence of reaction cascade is a very economical communication. Binding of a single hormone molecule at the cell surface is able to activate a number of adenylyl cyclase molecules, each of which is able to produce a large number of cAMP messengers. Thus formation of second messenger is an important amplification step in the process. Each cAMP molecule can activate only a single

PKA molecule which can phosphorylate a large number of phosphorylase kinase molecules. These in turn, phosphorylate an even large number of phosphorylase molecules, which can catalyse even much lager number of glucose phosphates.

Role of cAMP

cAMP exerts its effect by activating PKA (*protein kinase A*). Activation of PKA in liver cells in response to epinephrine leads to the breakdown of glycogen. Activation of this enzyme in kidney tubule cell in response to vasopressin causes reduction in the premeability of membrane to water and in a thyroid cell in response to TSH induces secretion of thyroid hormone.

2. Cyclic GMP (cGMP) Pathway

It is also an important second messenger found in animal cells. cGMP is formed from GTP by **guanylyl cyclases.** GTP is degraded to GMP by a *phosphodiesterase.* cGMP regulates ion channels and phosphodiesterases. It action is mediated by the activation of cGMP-dependent protein kinases. In vertebrate eye, cGMP serves as a second messenger during the conversion of visual signals received as light to nerve impulses. **Rhodopsin** in photoreceptor rod cells of retina is a **G-protein-coupled receptor.** Conformational change introduced by light in this receptor (rhodopsin) activates **G-protein transducin** which decreases the intracellular level of cGMP. This decrease is translated into a nerve impulse.

3. Calcium Ions (Ca^{2+}) Pathway

Ca^{2+} ions also act as cellular and intracellular messengers because they diffuse into the cytoplasm and bind to various target molecules, triggering specific response. Calcium ions play a key role in various important cellular activities such as cell division, secretion, endocytosis, fertilisation, synaptic transmission, metabolism and cell movement.

Under normal circumstances, the concentration of Ca^{2+} ions in the cytosol is maintained at a very low level. In contrast the concentration of these ions in the extracellular space or inside SER spaces or in mitochondria is even more than 1,000 times higher. SER spaces are specialised for calcium storage and release. These are, therefore, called **calcium sequestering compartments.**

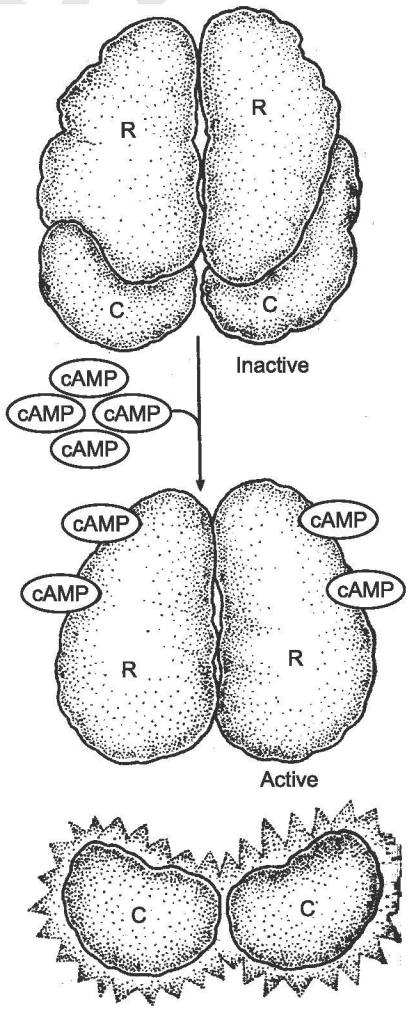


Fig. 2 : Regulation of protein kinase.

Change in Ca^{2+} ions concentration in the cytosol can occur in two different ways :

1. Ca^{2+} enter the cytosol from extracellular fluid through **voltage-gated Ca^{2+} channels.** This type of movement of Ca^{2+} ions occurs when terminal membrane of the neutron is depolarised by the action potential.
2. An extracellular Signalling molecule binds to a cell surface receptor which stimulates the release of calcium ions from the calcium-sequestering compartment inside the cell.

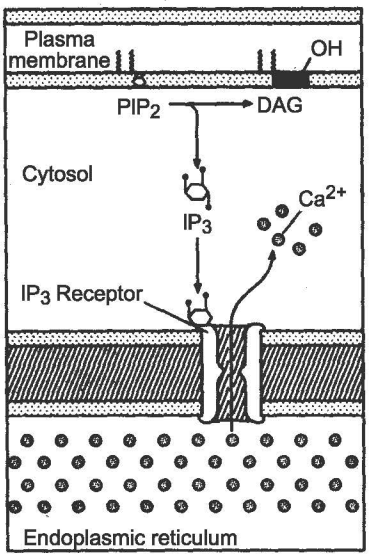


Fig. 3 : Ca^{2+} mobilization by IP_3 from cytosol into ER which serves as intracellular Ca^{2+} store.

4. Phospholipids Pathway

Membrane phospholipids like **phosphatidyl-inositol 4,5-biphosphate** (P_1P_2) act as second messenger in pathways of intracellular Signalling. PIP_2 is located on the inner or cytosolic surface of phospholipid bilayer. Its hydrolysis by *phospholipase C* is stimulated by a variety of hormones and growth factors and results in the formation of two second messengers-**diacylglycerol** and **inositol, 1, 4, 5-triphosphate** (IP_3). These activate protein kinase C and cause release of Ca^{2+} ions from intracellular stores. Increased Ca^{2+} level then activates a variety of target proteins, including protein kinases and phosphatases.

5. Inositol Triphosphate (IP_3)

Inositol 1, 4, 5-triphosphate (IP_3) is a sugar phosphate compound, soluble in water. It can diffuse through the membrane rapidly. It releases Ca^{2+} ions from calcium sequestering compartments.

IP_3 molecules diffuse through the plasma membrane into the cytosol and bind to a specific IP_3 receptor located on the cytoplasmic surface of intracellular compartments of smooth endoplasmic reticulum. The IP_3 reeptor also acts as a tetrameric Ca^{2+} channel. Binding of IP_3 opens the gated-channel and allows Ca^{2+} ions to diffuse out from ER into the cytoplasm.

The effect of IP_3 is usually because IP_3 is rapidly inactivated. IP_3 induces contraction of smooth muscles of stomach wall and wall of blood vessel and skeletal muscles.

The role of inositol triphosphate in signal transduction was suggested in 1953, when it was found that some extracellular signal molecules stimulate incorporation of radioactive phosphate into **phosphatidylinositol (PI).**

6. Calmodulin

Calmodulin is a small polypeptide chain of about 150 amino acid residues. It acts as a Ca^{2+} binding protein. It was discovered in late 1960s in the brain tissue. Calmodulin is found universally in plants, animals and eukaryotic microorganisms. It constitutes about 1% of the total protein mass of the cell.

Each molecule of calmodulin has four calcium binding sites. It has a dumb-bell shape with two globular ends connected by a long exposed α-helix. Each end has two Ca^{2+} binding domains each with a loop of 12 amino acid residues. On binding with Ca^{2+}, it undergoes conformational change. The allosteric activation of calmodulin by Ca^{2+} is analogous to allosteric activation of A-kinase by cyclic AMP.

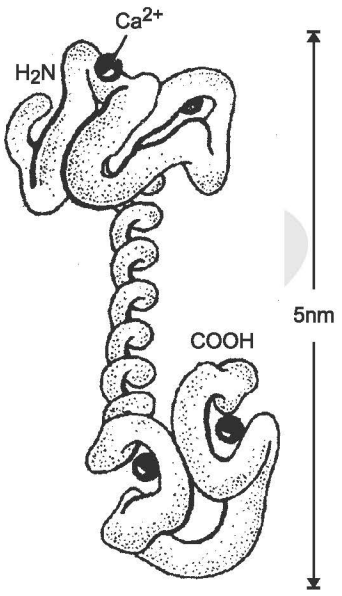


Fig. 4 : Structure of calmodulin

Calmodulin functions are multipurpose intracellular Ca^{2+} receptor. The binding of Ca^{2+} ions changes the conformation of protein calmodulin, causing its affinity for a number of proteins. The calcium calmodulin complex may bind to a protein kinase, a cyclic nucleotide phosphodiesterase or to the calcium transport proteins of plasma membrane. The effect of increase in free Ca^{2+} ions in the cytosol and activation of enzyme is shown as follows :

$$\text{Calmodulin} + Ca^{2+} \longrightarrow Ca^{2+}\ \text{Calmodulin Complex}$$
$$+\downarrow$$
$$\text{Active Enzyme} \longleftarrow \text{Inactive Enzyme Complex}$$

This complex helps in maintaining low concentration of Ca^{2+} ions in the cytosol by activating the system that is responsible for getting rid of excess of Ca^{2+} ions.

7. MAP Kinase Pathway

The MAP kinase pathway refers to a cascade of protein kinases that are highly conserved in evolution and play central role in signal transduction in all eukaryotes. The MAP kinases are regulators of cell growth and differentiation. MAP kinases stand for nitrogen-activated protein kinases. They are activated in response to a variety of growth factors. In animal cells, MAP kinase are coupled to growth factor receptors by the small GTP-binding protein, **Ras.** This initiates protein kinase cascade leading to MAP kinase (ERK) activation. ERK then phosphorylates a variety of cytosolic and nuclear proteins or transcription factors, which mediate gene induction. Some MAP kinase pathways mediate gene induction. Some MAP kinase pathways mediate responses of mammalian cells to inflammation and stress.

8. PI 3-Kinase/Akt or Serine-Threonine Kinase and mTOR Pathway

PIP_2 (phosphatidyl inositol 4,5-biphosphate) is usually cleaved into diacylglycerol and IP_3, but it can be phosphorylated to another second messenger PIP_3 (3, 4 5-triphosphate). This causes activation of protein-serine.

Threonine kinase (**Akt**) and is important for cell's survival. The target of Akt Signalling is the protein kinase mTOR which is a central regulator of cell growth and couples protein synthesis to the availability of growth factors, nutrients, and cellular energy.

9. JAK/STAT and TGF-β/SMAD Pathway

In these two pathways, the targeted transcription factors are phosphorylated directly by growth factor receptor associated protein kinases. **STAT proteins** are signal transducers and activators of transcription. They are present in inactive form in the cytoplasm and contain SH_2 domains. When cytokine receptors in the plasma membrane are stimulated by cytokine growth factors, the STAT proteins bind by their SH_2 domains to phosphotyrosine-containing sequences in the cytoplasmic domains of cytokine receptor polypeptides. The attached STAT proteins are then phosphorylated by JAK which are non-receptor protein-tyrosine kinases associated with cytokine receptors. The phosphorylated STAT proteins undergo dimerization. Their dimers separate and move to the nucleus to stimulate transcription of their target genes.

TGF-β represents a family of growth factors formed of protein-serine/threonine kinases. They phosphorylate transcription factors of the **Smads** family. The receptors of TGF-β are dimers of two polypeptides type I and II. The phosphorylated Smads enter the nucleus and activate the transcription of target genes.

10. NF-kB Signalling

Members of NF-kB family of transcription factors are activated in response to cytokines, growth factors and a variety of other stimuli. Their activation is mediated by phosphorylation and degradation of inhibitory 1kB subunits.

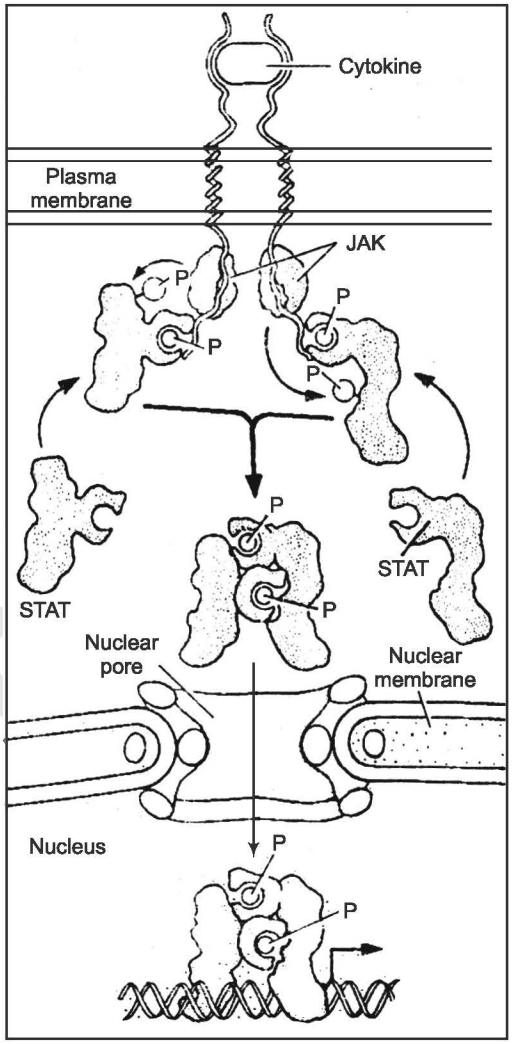


Fig. 5 : JAK/STAT protein pathway

UNIT-V

Mendelism and Sex Determination

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. What is developmental genetics?

Ans. The branch of genetics that deals with the role of genes in controlling the growth and development of an organism throughout its life cycle.

Development is the complex process through which a single-celled embryo transforms into a multicellular organism. Developmental processes are guided by information encoded in an organism's DNA and geneticists are trying to understand how this information leads to a fully formed organism.

Q.2. What is phenocopies?

Ans. When two genotypes produce the same phenotype due to different environments, one is called the phenocopy of the other, because they differ genotypically. For instance, in *Drosophila melanogaster,* normal body colour is brown and hereditary variant has yellow colour. It was observed that brown and yellow flies reproduce sincerely, irrespective of changes in the environment, because the genotypes differ in the two cases.

Cases are however, known where normal larvae, when raised on food containing silver salts, develop into yellow flies. This is a phenocopy of yellow mutant, but would give rise to brown flies in the normal environment.

Q.3. What is developmental noise?

Ans. In the above discussion, our emphasis has been on the effect of environment in determining the phenotype due to a specific genotype. However, even when environment and genotype are fixed (e.g. wild type at 16°C showing 1000 facets), different individuals or even two eyes of the same individual may differ in the number of facets (ranging from 980 to 1020 in wild type at 16°C). This uncontrolled variation in phenotype may be due to differences in local or internal environment during development, and is described as **developmental noise.**

Q.4. Define genotype-phenotype concept of Johannsen.

Ans. In order to make a definite distinction between hereditary and environmental variations. **Johannsen** in 1909 formulated the **genotype-phenotype concept.** According to him, the genotype of an individual represents sum total of heredity.

On the other hand, phenotype represents features which are produced by interaction between genotype and environment. A genotype can thus exhibit different phenotypes under different conditions. This is referred to as individual's **norm of reaction** to the environment. Therefore, similar genotypes may not have the same phenotype. Conversely, similar phenotypes do not necessarily mean same genotype.

Q.5. Write a very short note on sexuality in animals.

Ans. Before the development of light microscope in seventeenth century, the idea about sexuality in animals was based on speculation rather than facts. For instance, **W. Harvey** (1578-1657) speculated that all animals arise from eggs and that semen only plays a vitalizing role.

R. de Graff (1641-1673) observed that the progeny would have characteristics of father as well as mother and, therefore, suggested that both the parents should contribute to heredity. He also studied the development of embryo to some extent, although the egg was discovered by **Von Baer** in 1828. However, **A.V. Leeuwenhoek** observed sperms of several animals in 1677 and also suggested their association with eggs.

Q.6. Name three scientists who rediscovered Mendel's laws simultaneously.

Ans. When Mendel's laws were rediscovered simultaneously by **Hugo de Vries,** a Dutch biologist, **Carl Correns,** a German botanist and **Erich Von Tschermak,** an Austrian botanist.

Q.7. Why did Mendel chose garden pea for his experiments?

Ans. Mendel chose garden pea as plant material for his experiments, since it had the following advantages : (i) well defined characters, (ii) bisexual flowers, (iii) predominantly self-fertilization, (iv) easy hybridization. Besides these features, garden pea, being self-fertilized, had pure lines due to natural self-fertilization for a number of years. Therefore, any variety used was pure for the characters it carried.

Although hybridization experiments were conducted by earlier workers also as discussed in the previous section, but they considered the individual as a whole complex of characters. Mendel's success was mainly based on the fact that he considered a single character at one time. Seven pairs of contrasting characters were chosen for the study.

Q.8. Define monohybrid, dihybrid and trihybrid crosses.

Ans. Single characters each controlled by a single pair of genes or alleles were considered. Such crosses are known as **monohybrid crosses** and the F_2 ratio of 3 : 1 is known as the **monohybrid ratio.** Similarly crosses can be considered when two or three pairs of genes or alleles are involved. Such crosses will be called **dihybrid** and **trihybrid crosses** and the respective ratios as dihybrid and trihybrid ratios.

Q.9. Write a very short note on gene and allele.

Ans. Gene : Mendel presumed that a character is determined by a pair of **factors** or **determiners** present in each cell of the individual. These are known as genes in modern genetics. A gene is the unit of DNA responsible for the appearance and inheritance of a character.

Allele : Allele is a Greek word which means 'belonging to one another'. It refers to one of the two members of a gene pair. These represent alternatives of a character and are present on two separate chromosomes of a homologous pair, but at the corresponding loci. For example, in a gene pair **Tt, T** is present on one chromosome and **t** on the other homologue. **Alleles** or **allelomorphs** are a pair of genes representing the two alternatives of the same character and located at the same locus in the homologous chromosomes.

Q.10. Define the linkage.

Ans. According to the Law of Independent Assortment, any two or more than two pairs of characters assort independently of each other. Exceptions to this phenomenon were discovered due to linkage and the associated phenomenon of crossing over.

Q.11. Write different between dominant and Recessive.

Ans. A heterozygote possesses two contrasting genes or alleles but only one of the two is able to express itself, while the other remains hidden. The gene which gains expression in F_1 hybrid is known as **dominant gene,** which its allele is unable to express itself in presence of the dominant gene is the **recessive gene.**

Q.12. Define the haploid males in hymenoptera.

Ans. In the insect order Hymenoptera, which includes ants, bees, wasps and sawflies, etc. male individuals arise parthenogenetically (without fertilization) and therefore, have a haploid chromosome number. For instance, in honeybee, drones (males) have 16 chromosomes, while queen bee and workers have 32 chromosomes. In such cases, determination of sex is governed by haploid and diploid chromosome constitution.

Q.13. What are sex lethals? Explain.

Ans. Sex lethals (Sxl) is the master switch gene for somatic determination in *Drosophila* melanogaster. In XX animals Sxl becomes activated and imposes female development, in X (Y) animals, Sxl remains inactive and male development ensues.

Q.14. Give details in the Crew's hen.

Ans. A case of complex sex reversal was reported in 1923 by Crew, where a fertile female fowl (hen), which had already produced offspring, changed over to a fully fertile male (cock). This resulted due to a damaged ovary in the female. It is believed that ovary in female secreted a male suppressing hormone. Therefore, in the absence of ovary, testis could develop.

Q.15. Define the multiple allelism.

Ans. In each of the seven pairs of characters studied by Mendel, there were only two alternative forms for each character. This meant that only two alleles were present for each character. This also led to a belief that for each character there were two alternative forms, one dominant over the other. Later work showed that for a character there can be several phenotypes e.g. for rabbits the body colour can be of four or more types. Therefore, concept of alternative allelomorphs had to be modified by the concept of multiple allelism.

Q.16. What do you understand by reciprocal crosses?

Ans. A set of two reciprocal crosses means that the same two parents are used in two experiments in such a way that if in one experiment, 'A' is used as the female parent and 'B' is used as the male parent, in the other experiment 'A' will be used as the male parent and 'B' as the female parent.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Describe the germplasm theory of Weismann.

Ans.

Germplasm Theory

A. Weismann (1834-1914), for the first time, gave experimental evidence against **pangensis.** His popular experiments consisted of cutting the tails of mice and then studying the inheritance of the tail. Repeating such a treatment for 22 generations. Weismann found that complete tail structure was still inherited. These experiments of mutilation may appear now rather crude, but results can definitely be used as argument against **pangenesis** because once the tail was removed, the **pangenes** or **gemmules** for the tail will not be available and therefore this structure should not develop in the next generation if pangenesis holds good.

Weismann also proposed his own **germplasm theory** to account for heredity. According to this theory, the body of an individual can be divided into two types of tissues, **germplasm** and **somatoplasm.** The somatoplasm, was not able to enter the sex cells; consequently, the variations present in the somatoplasm will not be transmitted to next generation. The germplasm, on the other hand, was meant for the reproductive purpose only, so that any change occurring in germplasm will influence the progeny.

Germplasm theory of Weismann was a very significant advancement in our understanding of heredity, since this was for the first time that a distinction between hereditary and environmental variations could be made on a sound basis. However a distiction, between germplasm and somatoplasm in the sense of Weismann may be difficult to make. It is now known that the chromosomes are the main carriers of hereditary characters. This ideas was put forward in the form of **Chromosome Theory of Inheritance.** One may however, note that in recent years hereditary characters have been found in chloroplasts and mitochondria also.

Q.2. What do understand by Pangenes and acquired characters?

Ans.

Pangenes and Acquired Characters

It is pointed out earlier in this chapter that environmental variations have nothing to do with heredity. However, according to **J.B. Lamarck** (1744-1829), characters which are acquired during the lifetime of an individual are inherited. This concept is known as **Lamarckism** or **The Theory of Inheritance of Acquired Characters.** This theory was very popular in the eighteenth century to explain evolution and heredity. However, Lamarck did not point out the physical basis of this theory.

Charles Darwin (1809-1882) tried to examine the physical basis of heredity and inferred that every part of body produced very small invisible bodies called **gammules** or **pangenes,** which are transported through the blood stream to the sex organs and are assembled there into gametes. During fertilization, gemmules from both parents are brought together for redistribution to different organs during development, thus determining different characters. As is obvious, theory of **pangenesis** (*pan*– all; *genesis* – originating) proposed by Darwin is almost a copy of Lamarck's 'theory of inheritance of acquired characters' except that it suggested a physical basis. In the later part of the last century, through detailed study of cell structure and function, it was evident that Darwin's pangenesis was also based on imagination rather than on facts.

Q.3. Describe the transmission genetics.

Ans.

Transmission Genetics or Classical Genetics

The transmission genetics, sometimes also described as classical genetics makes the major part of the first half this book, although reference has been made to molecule aspects, wherever considered necessary.

These aspects include the following :

(i) **Mendelian genetics :** It involves study of both qualitative and quantitative (polygenic) traits and the influence of environment on their expression.

(ii) **Morganian genetics :** It includes **recombination** in all kinds of organisms, starting from higher plants and animals to fungi (*Neurospora*), bacteria and viruses. Since recombination is one of the sources for releasing hereditary variation, its study at all

levels particularly for preparation of linkage maps (also including molecular mechanism of recombination and preparation of molecular maps).

(iii) Non-Mendelian genetics : It involves a study of the role of cytoplasm and its organelles (particularly chloroplasts and mitochondria) in heredity. It has assumed special importance during the last three decades leading to study of characters located on these organellar genomes and preparation of chloroplast and mitochondria maps.

(iv) Mutations : These are another source of hereditary variation and have been studied at all levels—phenotypic level, biochemical level and molecular level. In a broad sense, these may include both chromosomal changes (structural and numerical) and also gene mutations.

Q.4. Write a short note on genomics.

Ans.

Genomics

With the availability of newer powerful tools, in mid 1990s later, efforts are being made to study the genome as a whole, rather than individual genes. In any organism, this will involve determining the nucleotide sequence of whole genome and assigning functions to all nucleotide sequences of this genome. Such a study is described as genome-wide study and the discipline is described as **'genomics'.** These **genome-wide studies,** described as genomics research, are being undertaken in several animal and plant systems and will lead to sequencing of the whole genome of humans and *Arabidopsis,* in the early years of the 21st Century.

Other plant (rice, maize, etc.) and animal (fruitfly, mouse, etc.) genomes are also being sequenced. It is hoped that the next decade (2000-2010) will be the decade of genomics research.

Function of Genomic

The traditional genetic functions of the genome include storage, propagation and transmission of the genetic material and rely on the use of the genetic information encoded in the DNA sequence.

Q.5. What do you known about preformation and epigenesis? Discuss.

Ans.

Preformation and Epigenesis

Although the basic idea of heredity that traits are transferred from one generation to the other was known since thousands of years in the past, but the actual physical link was not known. With the discovery of sexuality it could be suggested that hereditary traits must be transmitted either through egg or through sperm or both.

In 1679, **J. Swammerdam** studied development of insects and suggested that development of insects and suggested that development of an organism is a simple enlargement of a minute but preformed individual. This preformed individual was called **homunculus** and could be present in the sperm or in the ovum. During eighteenth and nineteenth centuries, there was a controversy among preformationists. Workers who attached more importance to the ovum were called **ovists** and they thought that homunculus was present in the ovum. Other workers like **Leeuwenhoek** who attached more importance to sperm were called **animalculists,** who insisted that a miniature but complete organism was present in the sperm. Preformation theory was soon rejected, because that homunculus was later found to be the creation of imagination of earlier workers and could never be observed in subsequent studies.

K.W. Wolff (1738-1794) proposed that neither egg nor sperm had a structure like homunculus but that the gametes contained undifferentiated living substance capable of forming the organized body after fertilization. Such an idea was called the theory of **epigenesis** This theory suggested that many new organs and tissues, which were originally absent, develop *de novo* due to mysterious vital forces. Such, however, was not the case and now we know that although the organism is not preformed, but its characteristics are predetermined by the sperm and the ovum taking part in fertilization.

Q.6. What do you mean by homozygous and heterozygous? Also write differences between homozygous and heterozygous.

Ans.

Homozygous and Heterozygous

Every organism possesses two gene for every character. If in an organism the two genes for a particular character are identical it is said to be **pure** or **homozygous** for that character. The prefix *homo* means 'the same' and **zygos** means 'a pair'. For example, tall plants with **TT** on dwarf plants with **tt** are homozygous. They produce only one type of gametes. If such plants are self-pollinated their offspring also have all characters similar to parent.

Heterozygous organism possesses contrasting genes of a pair. It receives two different alleles for the same character from its two parents. The prefix *hetero* means 'different' and *zygos* means 'a pair'. It means an organism with **T** and **t (Tt)** will be heterozygous. These produce two types of gametes.

Differences between Homozygous and Heterozygous

S.No.	Homozygous	Heterozygous
1.	Possess identical alleles of a gene (*e.g.* **TT**).	Possess dissimilar or contrasting alleles of a gene **(Tt).**
2.	Produce only one type of gametes (*e.g.* **T** only).	Produce two types of gametes (*e.g.* **T** and **t**).
3.	Breed true to the particular character, *i.e.,* all the offsprings possess the same genotype and phenotype when such parents are self-pollinated.	Produce three different genotypes when self pollinated which may have two or three different phenotypes.

Q.7. Write a short note on heterosis.

Ans.

Heterosis or Hybrid Vigour

Hybrids between two diferent races to varieties of animals and plants exhibit general increase in size, strength, stamina and vigour. This quality of hybrids is called as **hybrid vigour** or **heterosis.**

Early plant hybridisers had noticed that when cross-fertilisation is effected between two pure strains or races, differing in a number of characters, the resultant hybrids were superior to either of the parents with regard to a number of morphological and physiological features. The general increase was noticed in size, vigour, vitality, productivity, increased resistance of disease, more adaptability to the environment etc. The increase in the vigour of hybrid is not a permanent feature. If crosses are repeated the vigour is maintained, but if these are not crossed further the vigour decreases in successive generations.

The experiments on cross breeding were conducted by a number of scientists, like **Knight, Mendel, Darwin, Herber, Beal** and **Shull,** etc. **Shull** (1910) proposed the term heterosis for this superiority in hybrids.

Genetic Basis of Heterosis

The exact cause for the appearance of heterosis is still not properly understood.

(i) **Physiological stimulus or heterozygosity :** According to **Shull** (1910) heterosis is due to some kind of physiological stimulus produced by the union of dissimilar gametes from two separate races.

(ii) **Dominant factor hypothesis : Bruce** suggested that the superiority is governed by number of genes. Due to linkage it is not possible to get all the dominant genes at a time in any pure line breeding. Crossing of the two pure lines brings together these dominant genes in the hybrid. **Bruce** calculated that when dominant genes of one parent combine with those of the other one, there is an increase in the total number of dominant genes in the zygote. This is the most widely accepted hypothesis.

(iii) **Cytoplasmic nuclear reaction hypothesis :** According to **Michaelis, Shull** and **Lewis** heterosis is due to the interaction between nuclear and cytoplasmic systems.

Significance of Heterosis

Hybrid vigour has practical importance. It has been experimentally introduced in a large number of plants and animals. In plants, hybrid vigour is expressed in the form of height, viability, greater yield of fruits and seeds, large size of fruits and seeds, increased resistance to environmental factors, resistance of diseases and pests and better seed-germination, etc. It has been seen in tomatoes, onion, grasses, tobacco, cotton, sugarcane, brinjal, cucumber and several other ornamental plants.

Among animals, hybrid vigour is expressed in the form of milk production in cattle, increase in the number of eggs, better pork and beef output, and more strength and vigour. **Mule,** a hybrid of mare and donkey is definitely stronger and strudier than either of his parents. Similarly, hinney is a hybrid of horse and janette and zebronkeys is produced by a cross between zebra and donkey.

Q.8. What do you understand by pleiotropism? Explain with examples.

Ans.

Pleiotropism

According of Mendel, a specific gene produces only one specific phenotypic character. But cases have been studied in which one gene may produce several side effects, *i.e.,* a gene produces a major phenotypic trait but in addition to that influences some other phenotypic traits. This phenomenon of a single major gene influencing more than one character (multiple expression) is known as **pleiotropism** and such genes are known as **pleiotropic genes.** Pleiotropy may be due to true pleiotropic genes or the closely linked genes or groups of genes.

Examples : Following are the examples of pleiotropism :

(i) In *Drosophila,* the recessive gene for vestigial wings also affects structure of reproductive organs, reduction in the egg production, reduction in longevity, and the bristles in the wings.

(ii) In man, gene producing the disease **phenylketonuria** also produces a number of abnormal phenotypic traits, which are collectively known as **syndrome.**

The gene results in short stature, mental retardation, widely spaced incisors, pigmented patches on the skin and excessive sweating.

Q.9. What is Gynandromorphs in *Drosophila*?

Ans. Gynandromorphs in *Drosophila*

In *Drosophila,* occasionally flies are obtained in which a part of the body exhibits female characters and the other part exhibits male characters. Such flies are known as **gynandromorphs.** These are formed due to misdivision of chromosome and starts as female with 2A + 2X-chromosomes. One of the X-chromosomes is lost during the division of the cell with the result that one of the daughter cells possesses 2A + 2X chromosomes and the other 2A + X. If this event happens during first zygotic division, two blastomeres with unequal number of X-chromosomes are formed.

The blastomere with 2A + 2X-chromosomes develops into female half, while the second blastomere with 2A + X chromosomes produces male half and the resultant fly is a **bilateral gynandromorph.** The occurrence of gynandromorphs clearly indicates that the number of X-chromosomes determines the sex of the individual.

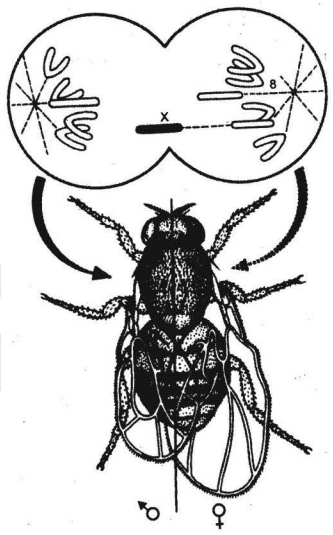


Fig. : Gynandromorph of *Drosophila* in which right half is male and left half is female.

Q.10. What do you understand by backcross and testcross? Explain them with the help of examples.

Ans. Backcross and Testcross

Crosses between F_1 offsprings with either of the two parents (hybrid) are known as **backcrosses.** When F_1 offsprings are crossed with the dominant parents all the F_2 offspring develop dominant character.

On the other hand when F_1 hybrids are crossed with recessive parent, individuals with both the phenotypes appear in equal proportion. While both the crosses are known as **backcross,** the second one is specified as **testcross.**

The **testcross** is a cross between heterozygous F_1 hybrid and the double recessive homozygous. The test cross is used to determine whether the individuals exhibiting dominant character the homozygous or heterozygous.

Examples :

(i) A cross between half heterozygous (F_1 hybrid) with homozygous tall pea plant produces all offsprings. But only 50% of them are all homozygous. The other 50% are heterozygous tall (Back cross).

(ii) A cross between tall heterozygous $\mathbf{F_1}$ hybrid with dwarf homozygous recessive $(\mathbf{P_1})$ produces tall and dwarf in equal proportion indicating that $\mathbf{F_1}$ hybrids are heterozygous (Testcross).

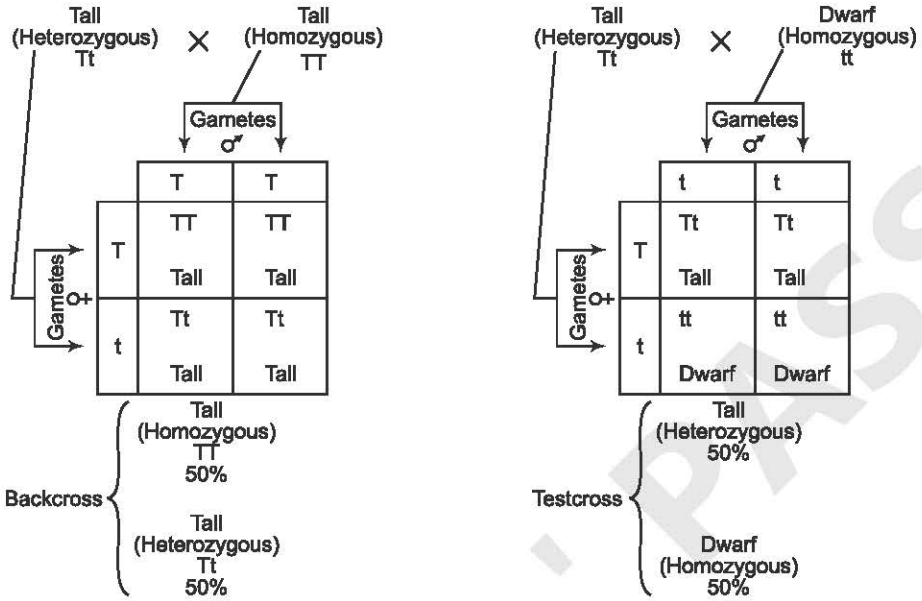


Fig. : Diagram of backcross and testcross, whereby a heterozygous tall pea plant is differentiated from homozygous tall pea plant.

Q.11. Write the differences between Klinefelter's and Turner's syndrome.

Ans. Differences between Klinefelter's and Turner's Syndrome

Klinefelter's Syndrome : Klinefelter discovered some **XXY** individuals which are phenotypic males. These have poorly developed male sex-organs. Their face and hair are somewhat faminine and these have somewhat enlarged breasts. They are, therefore, sexually sterile. Such abnormal males exhibits Klinefelter's syndrome. They possess 47 chromosomes. From these observations, it appears that **Y**-chromosome possesses some genes which promote the emergence of male characteristics and the **X**-chromosomes produces female characters or inhibits male characteristics.

Turner's Syndrome : About one out of every 300 females born is an abnormal female. These phenotypic females have **XO**-genotype (**AAXO**). They possess 45 chromosomes. Such **XO** females do not develop adult female characters and are functionally sterile. These have ovaries formed of bundles of connective tissue.

They are retarded physically as well as mentally. They average about 4 ft 10 inches and exhibit webbing of neck, low-set ears and under-developed breasts.

Q.12. Write a short note on free martinism.

Ans. Free Martinism

Another example of early influence of hormones on sex determination comes from free martins often found in cattles. **Lillie** and others found that where twins of opposite sex (one male and other female) are born, the male is normal but female is sterile with many male characteristics. Such sterile females are known as **free martins.** The scientific explanation for the formation of free martins is the effect of hormones of the male sex on the female. In cattle, the foetal membranes of the twins are fused in such a manner that they have a common circulation of blood. The female hormone is produced at a slightly later stage in the development and guides its development towards female side.

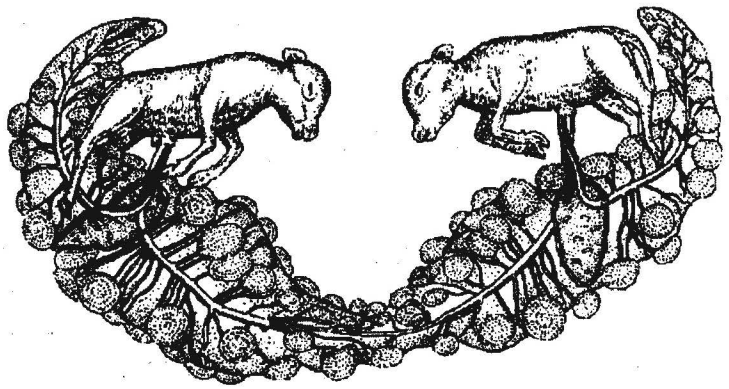


Fig. : Free martins in cattle

But since the twins have a common circulation and blood passes from one twin into the body of other twin, the male hormone, which is produced slightly in advance of female hormone, enters the body of female twin and before the female hormone onsets the development of female characteristics it is already differentiated in the guidance of male hormone. As a result the developing female is sterile.

Q.13. Describe the gene penetrance and their types.

Ans.

Gene Penetrance

Genes express themselves by producing visible phenotypic effect. Generally, the structural genes express themselves in all the individuals that carry them. **The ability of gene to express itself in an individual that is carrying it, is called penetrance.** All the factors in Mendelian experiment were able to express themselves completely, producing the expected phenotype. But case were noticed where structural genes failed to produce expected phenotypic ratio.

On this basis penetrance can be of two types :

1. Complete Penetrance

In complete penetrance a dominant gene expresses itself in all the organisms in which it is present producing the expected phenotype while the recessive gene produces the associated phenotype in all those organisms in which it is present in homozygous condition. Examples :

(i) In pea plants the red and white flower colour, tall and dwarf character, the seed colour and shape all exhibit complete penetrance.

(ii) In *Drosophila,* the recessive gene for wing character (vestigial wings) has complete penetrance.

(iii) In Guinea pigs the gene for coat colour exhibits complete penetrance.

2. Incomplete Penetrance

Some genes whether in homozygous or heterozygous condition fail to produce the phenotypic expression in all the organisms in which these are present, *i.e.,* these fail to produce cent-percent expression. These are said to exhibit **incomplete penetrance.** In these cases degree of penetrance is usually expressed as the percentage of individuals that actually express the phenotype determined by a particular genotype. Examples :

(i) In man, genes for **polydactyly, blue sclerotic** and **diabetes mellitus** all exhibit incomplete penetrance. The gene for blue sclerotic (produces sclerotic of blue colour) has about 90% penetrance, *i.e.*, it develops in about 90% persons, possessing it. Similarly, gene for polydactyly has 70% penetrance.

(ii) A gene in Lima beans causing partial chlorophyll deficiency in cotyledons leaves has 10 percent penetrance.

Q.14. Why do you understand by genetic imprinting. Explain.

Ans.

Genetic Imprinting

Genetic imprinting is a phenomenon, which involves differences in expression of genes inherited from mother and father. In other words, the chromosomes in the sperm and egg are differentially imprinted on marked as having come from the father or the mother, so that some maternally inherited genes and other paternally inherited genes are differentially expressed in the progeny.

An epigenetic changes in the germ cells, such as the inactivation of an X-chromosome, are reversed or altered during or just before meiosis, which leads to the production of sperms and eggs. After this reversal of epigenetic changes, the eggs and sperms are differentially imprinted. As a consequence of this, even if a chromosome in a sperm happens to have been inherited from the fathers' mother, it would still bear the male imprint. Such imprinting has been found to be crucial to normal development in mice and other mammals, because it silences and activates different sets of genes in the maternal and paternal chromosomes. These differences in paternal and maternal chromosomes complement eachother in directing normal development. This differentiation or reprogramming of the genome is believed to be achieved through changes in methylation patterns. In mice, differential methylation of paternal and maternal chromosomes has actually been demonstrated.

Q.15. Define sex linkage and their types.

Ans.

Sex-linkage

The mechanism of sex-linkage is slightly different from the ordinary linkage of genes because the sex-linked genes are present on the sex chromosomes which do not possess homologous parts.

Types

Depending upon whether the sex-linked genes are present on **X** or **Y** chromosome, the sex-linked inheritance can be of the following types :

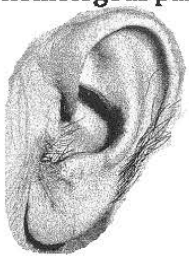


Fig. : Hair on pinna-a holandric character in man.

(i) Digenic : The sex-linked genes are present on the nonhomologous part of **X** chromosome and are passed on from male parent to male grandson through daughters of F_1 generation. Naturally male sex is heterogametic.

(ii) Diandric : In the case the **2X**-chromosomes of female behave as if these are completely homologous and female passes its one **X** received from female to male.

(iii) Hologenic : This character is directly passed from male to female.

(iv) Holandric : Genes which are in the nonhomologous part of **Y**-chromosome are passed on directly from father to son.

Q.16. Write a short note on sex-limited and sex-influenced genes.

Ans.

Sex-limited Genes

Sex-limited genes are present in the autosomes but their expression is determined by the presence or absence of one of the sex-hormones. Therefore, these express themselves only in one sex. These differ from the sex-linked genes, which are actually located in the sex chromosomes. The sex-limited genes control the expression of primary and secondary sexual characters. They express their effects in only one-sex and their action is clearly related to the sex-hormones. For example, beard-development in human beings is a sex-limited character. The genes for deep male voice and male musculature are expressed only in the presence of male sex-hormone. Similarly, genes for feminine voice, development of breast and feminine musculature are sex-limited characters. The abnormalities in hormone secretion may result in the development of breast in male or beard in female (hormone imbalance).

Sex-influenced Genes

The **sex-influenced genes** are present in the autosomes whose dominance is influenced by the sex of the bearer. These are expressed more frequently in one sex than in the other. For example, **pattern of baldness** is dominant in man and recessive in female. This is because the gene for baldness (B) in heterozygous state (Bb) expresses itself in male but not in female. It means gene B for baldness behaves as a dominant in male and as a recessive in female.

Genotype	Male	Female
BB	Bald	Bald
Bb	Bald	Not bald
bb	Not bald	Not bald

Horns in sheep and spotting in cattle are sex-influenced characters.

Q.17. Describe the master gene concept of sex determination.

Ans.

Master Gene Concept of Sex Determination

An international team of scientists (**Dr. David C. Page**) and others at Whitehead Institute for Biomedical Research and the Massachusettes Institute of Technology in Cambridge very recently (1987) have come to the following conclusion about sex determination in man.

The foetus having a master gene TDF (testes determining factor) develops testes and grows into a male. When this gene is absent the foetus develops ovaries and grows into a female. This gene is present on Y-chromosome and is believed to act as a biological switch, turning other genes on and off.

At the start, a human embryo has all the other genetic instructions it needed to become a male or female, it develops into a male only if the TDF gene is present, otherwise it develops into a female.

The above concept is based on the following observations :

(i) Maleness can be conferred by a minute piece of Y-chromosome, even when that piece is inherited with two complete X-chromosomes. Such males are sterile.

(ii) An individual develops as a female when the crucial part of Y-chromosome is missing. Such females remain sexually immature.

Q.18. Write the scope and significance of genetics.

Ans. **Scope and Significance of Genetics**

In the last few decades, the science of genetics has pervaded all aspects of biology so that it has assumed a central position of great significance in biology as a whole. While on the one hand, genetics is used for a study of the mechanism of heredity and variation, on the other hand it has provided tools for the study of the fundamental biological processes examined and taught in areas, like plant physiology, biochemistry, biosystematics, ecology, plant pathology, microbiology, etc. Consequently today every biologist should be bit of a geneticist. Genetics, in fact provided the modern paradigm (a prototype) for whole of biology. The science of genetics also had a tremendous impact in applied areas including medicine, agriculture, forestry, fisheries, law and religion. In view of this, all newspapers often address questions dealing with different aspect of genetics that may be of significance to common man, who is not a geneticist or a biologist. The recent upsurge of biotechnology has added further to the significance of the science of genetics, so that the products of genetics have also become a subject for discussion for Trade Related Aspects of Intellectual Properties (TRIPs) under the aegis of General Agreement on Tarrif and Trade (GATT) Patenting of life forms, which may or may not be the product of genetic manipulation is one such topic, which is receiving considerable attention of both developed and developing countries.

Genetics can be broadly classified in the following three areas for the convenience of a discussion on its scope and significance :

(i) **Transmission genetics** involving study of transmission of genetic material from one generation to the other.

(ii) **Molecular and biochemical genetics,** involving study of the structure and function of genes.

(iii) **Population and biometrical genetics,** involving study of the behaviour and effects of genes in population, often using mathematical models.

The above classification is arbitrary and the three areas are inter-related and even enter other areas of biology to answer some difficult questions. Significance of genetics also stems from the fact that the genetic material containing information for hereditary traits consists of nucleic acids only, across the entire spectrum of life on the earth.

More important of the two types of **nucleic acids, deoxyribose nucleic acid** (DNA) and **ribose nucleic acid** (RNA) is the former i.e., DNA, which has two unique properties :

(i) It can replicate and produce its exact copies.

(ii) It carries the genetic information, necessary to give form to an organism; this information is written into the sequence of four monomers called **nucleotides,** which make the polymer molecule, the DNA.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. Describe Mendels' laws of inheritance with the help of suitable example.

Ans. **Gregor Johann Mendel**

The Austrian monk **'Gregor Johann Mendel'** was the first person to explain the mechanism involved in the transmission of characters from parents to the offspring generation after generation. He is, therefore, considered as the pioneer of modern genetics and is called **"Father of Genetics".**

Mendel was born on July 22, 1822 in a gardner's family. In October 1843, Mendel entered Augustinian monastery at Brunn in Austria as a priest. There he earned the title Gregor in 1849.

He started his historic experiments with garden pea (*Pisum sativum*) in the monastery garden. The experimentation work continued for about nine years from 1856 to 1864. The results of these classical experiments and Mendel's conclusions were published in an obscure journal—*'The Annual Proceedings of the Natural History Society of Brunn'* in 1865.

Mendel's Experiments

Mendel studied the inheritance of seven different pairs of contrasting characters in garden pea (*Pisum sativum*) plant but considered only one pair at a time. He crossed two pea plants with alternate characters by **artificial pollination.** The resulting hybrids which resembled one of the two parents, were then crossed with each other. He pooled the data of many similar crosses, analysed the results and found that both traits reappeared in the F_2 offsprings in a definite ratio of 3 : 1.

Mendel's Laws of Inheritance

Mendel postulated three laws, which are now called after his name as Mendel's laws of heredity. These are :

1. Law of dominance and recessiveness
2. Law of segregation
3. Law of indpendent assortment

1. Law of Dominance

When two homozygous individuals with one or more sets of contrasting characters are crossed the characters that appear in the F_1 hybrids are **dominant characters** and those that do not appear in F_1 are **recessive characters.**

The dominance and recessiveness of genes can be explained on the basis of enzymatic functions of genes. The dominant genes are capable of synthesising active polypeptides or proteins that form functional enzymes, whereas the recessive genes (mutant genes) code for incomplete or non-functional polypeptides. Therefore, the dominant genes produce a specific phenotype while the recessive genes fail to do so. In the heterozygous condition also the dominant gene is able to express itself, so that the heterozygous and homozygous individuals have similar phenotype.

Critical Appreciation of Law of Dominance

Scientists conducted cross-breeding experiments to find out the applicability of law of dominance. The experiments were conducted by **Correns** on peas and maize, by **Tschermak** on peas, by **DeVries** on maize, etc., by **Bateson** and his collaborators on a variety of organisms, by **Davenport** on poultry, by **Hurst** on rabbits, by **Toyama** on silkmoth and by many others. These scientists observed that a large number of characters in various organisms are related as dominant and recessive.

List of characters in Plants and Animals related as Dominant and Recessive

S. No.	Name of Plant or Animal	Dominant characters	Recessive Characters
	In plants		
1.	**Maize**	(a) Full endosperm	Shrunken endosperm
		(b) Starchy seeds	Sugary seed
		(c) Coloured pericarp	Colourless pericap
2.	**Wheat and barley**	(a) Beardless	Bearded
		(b) Rivet wheat	Polished wheat
		(c) Late ripening	Early ripening
		(d) Susceptibility to rust	Resistance to rust
3.	**Tomato**	(a) Purple stem colour	Green stem colour
		(b) Red fruit colour	Yellow fruit colour
		(c) Tall plant	Dwarf plant
		(d) Spherical fruit shape	Oval fruit shape
	In Animals		
4.	***Drosophila***	(a) Red eyes	White eyes
		(b) Gray body colour	Black body colour
		(c) Long wings	Vestigial wings
		(d) Straight wings	Curved wings
5.	**Rabbit**	(a) Coloured coat	Albino
		(b) Angora fur	Short fur
6.	**Cattle**	(a) Horned character	Hornless character
7.	**Poultry**	(a) Rose comb	Single comb
8.	**Man**	(a) Brown eyes	Blue eyes
		(b) Black body	Albino colour
		(c) Long stature	Short stature

Importance of Law of Dominance

The phenomenon of dominance is of practical importance as the harmful recessive characters are masked by the normal dominant characters in the hybrids. In human beings a form of idiocy, diabetes, haemophilia, etc. are recessive characters. A person hybrid for any of these characteristics appears perfectly normal. Thus, harmful recessive genes can exist for several generations without expressing themselves.

Exceptions to Law of Dominance (Incomplete Dominance)

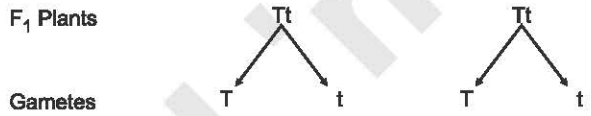
After Mendel, several cases were recorded by scientists, where F_1 hybrids exhibited as blending of characters of two parents. These hybrids were found to be midway between the two parents. This is known as **incomplete dominance** or **blending inheritance.** It means that two genes of the allelomorphic pair are not related as dominant and recessive, but each of them expresses itself partially. As for example, in **four-O'clock plant,** *Mirabilis jalapa,* when plants with red flowers **(RR)** are crossed with plants having white flowers **(rr),** the hybrid F_1 plants **(Rr)** bear pink flower. When these F_1 **plants** with pink flowers are self-pollinated they develop red **(RR),** pink **(Rr)** and white **(rr)** flowered plants in the ratio of 1 : 2 : 1 (F_2 generation).

2. Law of Segregation (Purity of Gametes)

The law of segregation states that when a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote (hybrid) the two members of the allelic pair remain together without being contaminated and when gametes are formed from the hybrid the two separate out from each other and only one enters each gamete.

Example : Pure tall plants are homozygous and, therefore, possess genes (factors) **TT;** similarly dwarf plants possess genes **tt.** The tallness and dwarfness are two independent but contrasting factors or determiners. Pure tall plants produce gametes all of which possess gene **T** and dwarf plants **t** type of gametes.

During cross fertilisation gametes with **T** and **t** unite to produce hybrids of F_1 generation. These hybrids possess genotype **Tt.** It means F_1 plants, though tall phenotypically, possess one gene for tallness and one gene for dwarfness. Apparently, the tall and dwarf characters appear to have become contaminated developing only tall character. But at the time of gamete formation, the genes **T** (for tallness) and **t** (for dwarfness) separate and are passed on to separate gametes. As a result, two types of gametes are produced from the heterozygote in equal numerosity. 50% of the gametes possess gene **T** and other 50% possess gene **t.** Therefore, these gametes are either pure for tallness or for dwarfness. (This is why the law of segregation is also described as **law of purity of gametes).**



Gametes unite at random and when gametes are numerous all possible combinations can occur, with the result that tall and dwarf appear in the ratio of 3 : 1. The results are often represented by Punnett square as follows :

	♂ \ ♀	Female Gametes T	Female Gametes t
Male Gametes	T	TT	Tt
	t	Tt	tt

F_2 Zygotes

Fig. : Punnett square : A very popular method of representing

Critical Appreciation of Law of Segregation

It has been confirmed by cytological studies that dominance or no dominance, the law of segregation hold good to all cases. Its far reaching applicability has made it is a rare **biological generalisation.**

3. Law of Independent Assortment

Definition : If the inheritance of more than one pair of characters (two pairs or more) is studied simultaneously, the factors or genes for each pair of characters assort out independently of the other pairs. Mendel formuated this law from the results of a dihybrid cross.

Explanation : The cross was made between plants having, **yellow and round cotyledons** and plants having **green** and **wrinkled cotyledons.** The F_1 hybrids all had yellow and round

seed. When these F_1 plants were self fertilized they produced four types of plants in the following proportion :

(a) Yellow and round	9
(b) Yellow and wrinkled	3
(c) Green and round	3
(d) Green and wrinkled	1

The above results indicate that yellow and green seeds appear in the ratio of 9 + 3 : 3 + 1 = 3 : 1. Similarly, the round and wrinkled seeds appear in the ratio of 9 + 3 : 3 + 1 = 12 : 4 or 3 : 1. This indicates that each of the two pairs of alternative characters *viz.* yellow-green cotyledon colour is inherited independent of the round-wrinkled character of the cotyledons. It means at the time of gamete formation the factor for yellow colour enters the gametes independent of **R** or **r**, *i.e.,* gene **Y** can be passed on to the gametes either with gene **R** or **r.**

Cytological Explanation of the Result

In the above experiment yellow and round characters are dominant over green and wrinkled characters which can be represented as follows :

(i) gene for yellow colour of cotyledons	**Y**
(ii) gene for green colour of cotyledons	**y**
(iii) gene for round character of cotyledons	**R**
(iv) gene for wrinkled character of cotyledons	**r**

Therefore, plants with yellow and round cotyledons will have their genotype **YYRR** and those with green and wrinkled cotyledons will have a genotype **yyrr.** These plants will produce gametes with gene **YR** and **yr** respectively. When these plants are cross pollinated, then union of these gametes will produce F_1 hybrids with **YyRr** genes.

When these produce gametes all the four genes have full freedom to assort independently and, therefore, there are possibilities of four combinations in both male and female gametes.

1. **RY** 2. **Ry** 3. **rY** 4. **ry**

This shows an excellent example of independent assortment. These gametes can unite at random producing in all 16 different combinations of genes, but presenting four phenotypes in the ratio 9 : 3 : 3 : 1.

		Pure round, yellow	
1. **RRYY-1** 2. **RRYy-2** 3. **RrYy-4** 4. **RrYY-2**	}	**9**	**Round, yellow**
5. **RRyy-1** 6. **Rryy-2**	}	**3**	**Round, green**
7. **rrYY-1** 8. **rrYy-2**	}	**3**	**Wrinkled, yellow**
9. **rryy-1**	}	**1**	**Wrinkled, green**

Critical appreciation of law of Independent Assortment

The law of independent assortment fails to have a universal applicability. Cytological studies have revealed that only those allelomorphs assort independently during meiosis, which are located in different homologous pairs of chromosomes. But, if the allelomorphs for different characters are present in the same homologous pair of chromosomes, these are passed on to the same gamete. Law of indpendent assortment does not apply to such cases.

Q.2. Write an essay on the chromosome theory of sex determination.

Ans. **Chromosome Theory of Sex Determination**

In majority, of diploid sexual animals are found a pair of sex chromosomes which are specialised for sex determination. These are represented by **X** and **Y.**

1. Sex Chromosomes and Autosomes

The X-chromosomes was first observed by German biologist, **Henking** in 1891 during the spermatogenesis in male bug and was described as **X-body.** The chromosome theory of sex determination was worked out by **E.B. Wilson** and **Stevens** (1902-1905). They named the X and Y chromosomes as **sex chromosomes** or **allosomes** and other chromosomes of the cell as **autosomes.**

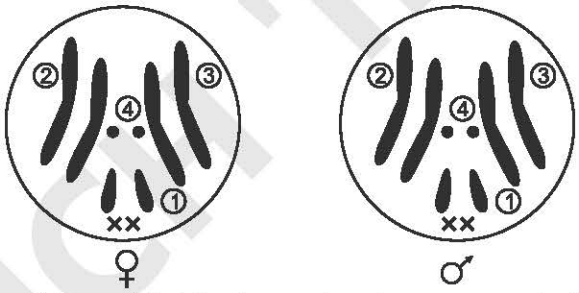


Fig. 1 : Chromosomes of *Drosophila*. The three pairs of autosomes, which are similar in both the sexes and XX or XY are sex chromosomes similar in female but dissimilar in male.

(i) **Sex chromosomes** carry genes for sex. **X-chromosome** carries female determining genes and **Y-chromosome** has male determining genes. The number of **X** and **Y** chromosomes determines the female or male sex of the individual.

(ii) **Autosomes** carry genes for the somatic characters. These do not have any relation with the sex.

2. Types of Chromosomal Mechanisms of Sex Determination

Following are the types of chromosomal mechanism of sex determinate :

I. Sex Chromosomes Undifferentiated

In primitive forms sex chromosomes or X and Y-chromosomes are not identified. The genes determining the sex seem to be located on certain autosomes. This is regarded as the most primitive type of sex determination.

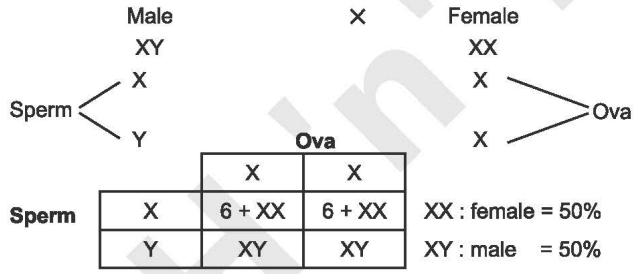
II. XX-XY Type or Lygaeus type

XX-XY type of sex determining mechanism was first studied in the milk weed bug, *Lygaeus turcicus* by **Wilson** and **Stevens.** Therefore, it is called **Lygaeus type.** There are two different patterns of sex determination in Lygaeus type :

(A) Female Homogametic XX and Male Heterogametic XY

The homogametic sex **(XX)** is female and produces ova all of one type, *i.e.,* having **X**-chromosome. The male is heterogametic—**XY** and produces sperm of two types, 50% of which possess **X**-chromosome and other 50% **Y**-chromosome. This is simple **XX-XY** type and is found in man, *Drosophila* and certain insects.

(a) Sex-Determination in *Drosophila* : In *Drosophila* total number of chromosomes is eight, of which six are autosomes, common to both male and female. The fourth pair of sex chromosomes. In male this is represented by **XY,** *i.e.,* karyotype of male *Drosophila* 6 + **XY** and in female **XX**, *i.e.,* 6 + **XX.** Ova produced by female are all similar possessing 3 + **X** chromosomes, whereas the sperm produced by male are 3 + **X** and 3 + **Y** in equal numbers.



(b) Sex Determination in Man : In case of man total number of chromosomes is 23 pairs of 46.

(i) In male (man) 44 + **XY**

(ii) In female (woman) 44 + **XX**

The **sperm** produced by male are of two types (a) 22 + **X,** (b) 22 + **Y,** whereas the **ova** all have 22 + **X** chromosomes.

Example Melandrium : In *Melandrium* (the garden pink) a variety of garden flower, sex is determined by a pair of **X** and **Y** chromosomes just as in animals. In *Melandrium* the **Y**-chromosome is longer than **X**-chromosome.

Modifications of XX-XY Mechanism : There are several modifications of this simple **XX-XY** type of mechanism :

(i) X and Y-chromosomes attached to a Pair of Autosomes : In this type **X** and **Y** are attached to a pair of autosomes and during maturation follow the autosomes.

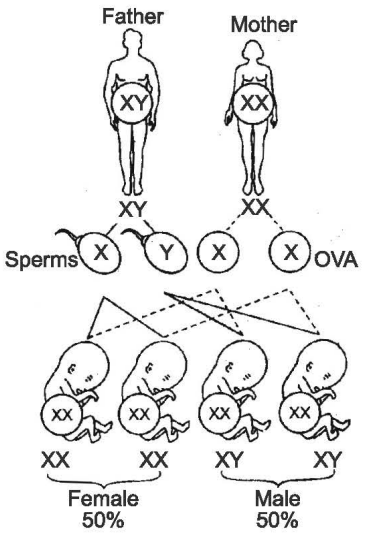


Fig. 2 : Sex determination in man, all the eggs carry X-chromosome but one-half of the sperm carry an X-chromosome and one-half carry a Y-chromosome.

(ii) X or Y Complex : In certain organisms either **X** or **Y** chromosome gets broken into two or more fragments forming **X**-complex or **Y**-complex. In *Tenodera, Mantis* and *Stegomantis,* **X**-chromosome is broken into two fragments. Therefore, the female possesses $X_1 X_1 X_2 X_2$ and male $X_1 X_2 Y$. In **Accola insect** male possesses 26 chromosomes and female possesses 30 chromosomes.

(iii) Male **20 + 5X + Y** and female **20 + 5X + 5X.** Sperm produced are (i) **10 + 5X** and (ii) **10 + Y,** whereas ova all possess **10 + 5X.**

(B) Female Heterogametic and Male Homogametic

In fowl, other birds and some fishes, certain moths and butterflies the female sex is heterogametic, with **X** and **Y**-chromosomes often represented by **Z** and **W** and laying two types of eggs, one half with **X** or **Z** chromosome and the other half with **Y** or **W** chromosome. The male sex is homogametic having **XX** or **ZZ** chromosomes. It produces sperm all of one type.

Q.3. Explain Bridge's Genic balance theory of sex determination in *Drosophila*. How does it differ from that of Goldschmidt's.

Ans.

Genic Balance Theory

Based upon the observations of ratio theory, Bridges put forward genic balance theory in which he suggested that every individual whether male or female possesses in its genotype genes for both male and female characteristics. Which sex will actually develop is decided by the preponderance of that type of genes. If there is excess of female determining genes a female develops or if the ratio of male determining genes exceeds that of female determining genes a male is formed. The sex chromosomes and autosomes are mere vehicles of genes. In *Drosophila,* the X-chromosome carries more genes that incline the development towards femaleness and the autosomes possess genes which incline the development towards maleness. Therefore, the deciding factor is the ratio between the number of X-chromosomes to autosomes. In *Drosophila,* given table represents the results of different ratios of **X**-chromosomes to the sets of autosomes.

Showing ratio of X and A chromosomes responsible for determination of sex in *Drosophila*

S. No.	Sex	Number of X-chromosomes	Sets of Autosomes	Sex-index ratio (X-A)
1.	Super female	XXX	AA	1.5 super female
2.	Normal female — Tetraploid	XXXX	AAAA	1.0
	Triploid	XXX	AAA	1.0
	Diploid	XX	AA	1.0
3.	Intersex	XX	AAA	.66
		XXX	AAAA	.75
4.	Normal male	X	AA	.50
5.	Intersex	X	AAA	.33

For a normal male the ratio between X-chromosomes and autosomes is .50 and for female 1.00. The ratio increases in superfemales and decrease in super males.

Although, the ratio of X-chromosomes and autosomes is operative in determining sex in *Drosophila* but it is not a universal mechanism. **Coswing** has shown that in *Platypocilus* the sex determining factor is the ratio of autosomes to Y-chromosome.

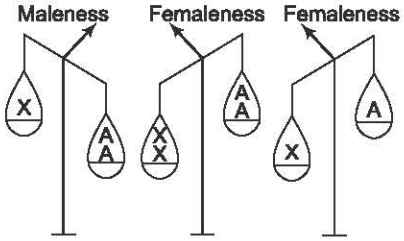


Fig. 1 : Diagrammatic representation for genic balance theory of sex determination. X-contains female determining genes; A-contains male determining genes.

Goldschmidt Theory

Classical work of **Goldschmidt** on diploid intersexes in gypsy moth, *Lymantria,* offers an important contribution towards understanding the mechanism of sex determination. *Lymantria* extends from England to Japan and exhibits striking sexual dimorphism.

While working on Lymantria **Goldschmidt** observed the following facts : When *Lymantria dispar* (European race) male are crossed with *L. japonica* females, the males and females of the progeny are all normal, but when the sexes were changed, *i.e.,* when females of *dispar* of European race were crossed with males of *japonica* race the females were all intersexes, whereas males were all normal.

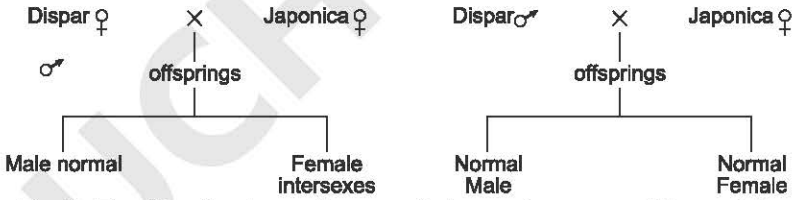


Fig. 2 : Results of reciprocal crosses between two races of *Lymantria*

Results of reciprocal crosses between two races of *Lymantria*

Cross	Parents	Males	Females
1.	European *L. dispar* (weak) female × *L. japonica* (strong) male	All normal	Intersexes
2.	*L. japonica* (strong) female × *L. dispar* (weak) male	All normal	All normal
3.	F_2 from cross 1	All normal	1/2 normal 1/2 intersexes
4.	F_2 from cross 2	1/2 normal 1/2 intersexes	All normal

To account for these observations **Goldschmidt** presumed that the factors for maleness are present on the **X**-chromosomes and for femaleness on Y-chromosome or in the cytoplasm and

that these factors vary in strength in different races. The sex factors are weak in European strain and strong in Japanese strain. Accordingly, if a European female (**XY**) is crossed to *japonica* male (**XX**), the females produced the intersexes because **X**-chromosome from *japonica* race with its maleness is not completely dominated by the relatively weak femaleness (**Y**) from European females. On the contrary males receiving one strong **X** from Japanese race and other weak **X** from European race result in the formation of normal males.

In the reciprocal cross between *japonica* female (strong) and *dispar* male (weak) 50% zygotes develop into **normal females** with weak **X** and strong **Y** and the other half into normal males with one strong and one weak **X**-chromosome.

The mechanism by which intersexes develop is briefed by **Goldschmidt.** The intersexes start development as females or males in the guidance of their chromosomal configuration and develop as such up to a certain critical point or turning point, after which their development is switched on towards the opposite sex due to the appearance of hormones which direct the development towards that direction. He has postulated certain hypothetical enzymes termed as ***andrase*** for initiation of maleness and ***gynase*** for femaleness. But experimental evidences in this connection are lacking.

Sexual Function of X and Y-Chromosomes

No doubt X and Y chromosomes are concerned with the determination of sex, both in animals as well as in plants, their exact role has been found to vary in different groups.

Function of X and Y Chromosomes in *Drosophila* : In *Drosophila,* **X** has been found to possess female determining genes and **Y** has no influence in determination of sex. The sex in *Drosophila* is, therefore, determined by the ratio of **X**-chromosomes to autosomes. **X/A = 0.50** gives rise to male. The fact has been established by the occurrence of **XO** males and **XXY** females, which are formed as a result of nondisjunction of sex chromosomes.

Q.4. Describe the sex-linked characters with suitable examples.

Ans.

Sex-Linked Characters

In man sex-linked characters belong to the following two classes :

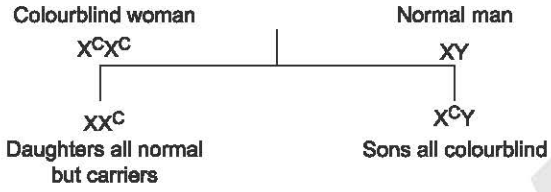
1. **X-linked Characters :** These are colour blindness and haemophilia. The genes of these characters are present on non-homologous part of X-chromosomes.
2. **Y-linked Characters :** The genes for these characters are located on Y-chromosome. These are also called holandric genes. These are genes for hypertrichosis, scaly skin, histocompatibility gene (H-Y antigen) and testis determining gene.

1. Inheritance of X-linked Characters

(a) Inheritance of Colourblindness in Man : Red green colourblindness or deutroranopia is a recessive X-linked character. Its gene is present on the nonhomologous part of X-chromosome. Y-chromosome is without its allele. Since male possesses only one X-chromosome, males with single gene for colourblindness develop this character. Females need two recessive genes for colourblindness, present one on each X-chromosomes.

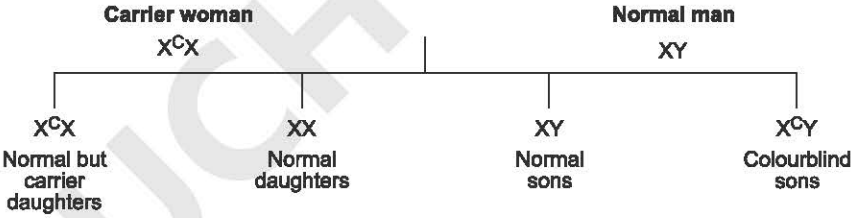
If X-chromosome with gene for colourblindness is repesented by X^c-and X-chromosome with gene for normal colour vision is represented by X, the results of different crosses can be represented as under :

(i) Colourblind Woman and Normal Man : If a colourblind woman is married to a man with normal colour vision, all her sons are colourblind and all her daughters have normal colour vision. But these daughters, are all carriers having one X-chromosome with gene for normal colour vision and other X-chromosome with gene for colourblindness (X^C).

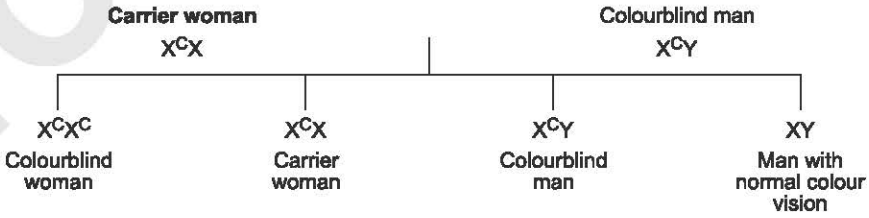


(ii) Colourblind Man and Normal Woman : If a colourblind man marries woman with normal colour vision, all her children (boys or girls) have normal colour vision.

(iii) Carrier Woman and Normal Man : If a carrier woman is married to a normal man, all her daughters will be normal, whereas 50% of the sons may be colourblind.



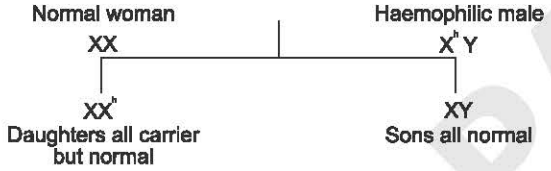
(iv) Carrier Woman and Colourblind Man : A woman could show colourblindness only if a carrier woman happens to marry a colourblind man.



(b) Inheritance of Haemophilia (Bleeder's Disease) in Man : Haemophilia is a recessive sex-linked character. Its gene is located on X-chromosome. In haemophilic persons, the blood fails to clot because of the deficiency in the formation of thromboplastin. The failure in the formation of thromboplastin is due to the absence of antihaemophilic globulin in the plasma of haemophilics. Therefore, the haemophilics bleed profusely from even a small cut. The disease haemophilia was very common in the royal families of Europe.

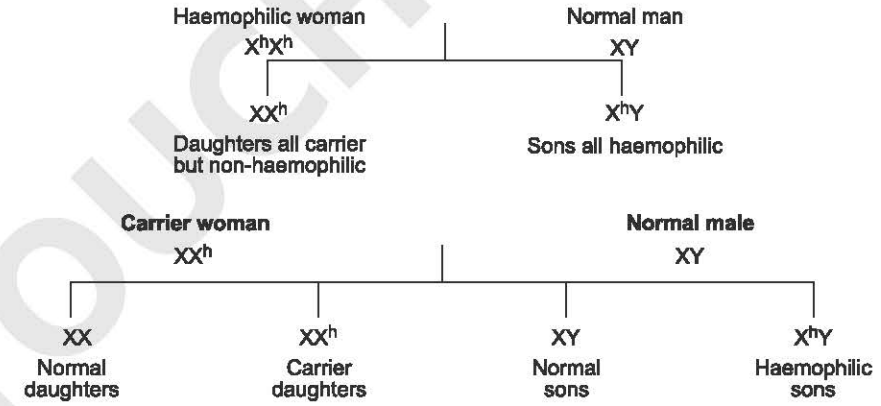
In antihaemophilic man globulin production is controlled by a gene present on the X-chromosome. Its recessive mutant gene, however, is unable to produce this substance in appropriate amount.

(i) If a Haemophilic Male Marries a Normal Female, all his daughters and sons are non-haemophilic. The daughters as a matter of fact are carrier, receiving one normal X-chromosome from mother and one X-chromosome with gene for haemophilia from father. But since this gene is recessive it fails to express itself. The sons on the other hand receive. X-chromosome from mother with normal gene and Y-chromosome with no gene for haemophilia, are normal.



(ii) If the Carrier Daughter of F_1 Generation is married to a normal man, approximately 50% of her sons are haemophilic. Of course daughters are all non-haemophilic, but half of them must be carriers. In case the carrier daughters are married to haemophilic man then half of females and half of males might express haemophila. It means that a female haemophilic could be obtained only if carrier female is married to a recessive haemophilic male.

(iii) If a Haemophilic Male Marries a Normal Male all her sons are haemophilic and daughters all non-haemophilic.



2. Inheritance of Y-Linked Characters Or Inheritance of Holandric Characters

A few genes are present on Y-chromosome of man and are called holandric genes or Y-linked genes. These genes do not have alleles on X-chromosome. Y-linked characters are transmitted from male parent to all his male offsprings. Whether recessive or dominant, Y-linked characters express them in every male offspring in every generation. It was reported by **Dronamraju** (1960).

Examples : Holandric genes in man are testis determining factor (TDF), hypertrichosis, (excessive hair growth on pinna), histocompatibility antigen gene (H-Y, antigen), gene for scaly skin **(ichthyosis hystrix gravior)** and genes for **height.**

Q.5. Discuss the phenomenon of sex reversal in mammals. How did the availability of XX males and XY females help in understanding and locating the genes responsible for sex determination.

Ans.

Sex Reversal in Mammals

Sex reversal in Mammals belongs to the following two classes :

1. Sex Reversal in Mice

In mice, XX male individuals and XY female individuals have been reported. The sex reversed XX males in mice resulted due to transfer of a sex reversing factor (*Sxr*) from a Y-chromosome, designated as Y^{Sxr}. This duplicated segment carrying *Sxr* was transferred to X-chromosomes due to crossing over during male meiosis and was then found to be on the distal arm of X-chromosome, which presumably pairs with a segment of Y-chromosome. The presence of *Sxr* on the distal arm of one of the two X's in *Sxr* XX males was actually confirmed.

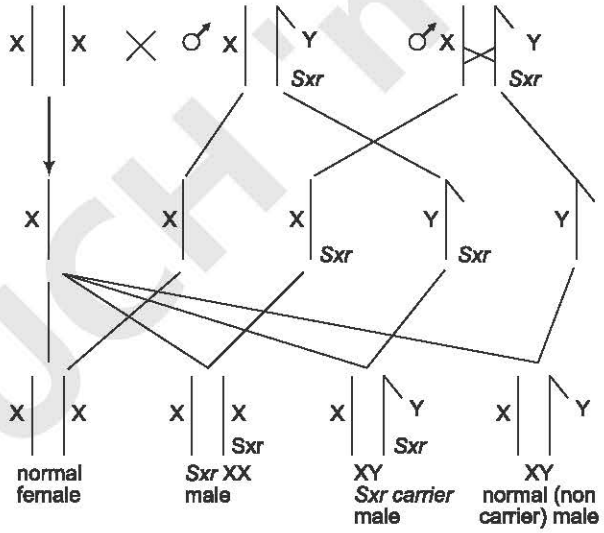


Fig. 1 : The inheritance of sex reversal gene *Sxr* carried by Y-chromosome (note its transfer to X-chromosome due to crossing over)

In the above situation, if a normal female is crossed with a XY *Sxr* carrier male, then normal female, *Sxr* XX males, XY *Sxr* carrier males and non-carrier XY males are obtained. Such a situation has actually been confirmed and can be confused with autosomally inherited genes, and therefore sometimes called a **pseudoautosomally inherited condition.**

Cases of sex reversed XY females or harmaphrodite mice have also been reported and atleast following two genes are said to be involved : (i) *Tdy* (or testis determining Y) gene lying in centromeric region of Y and (ii) *Tda*-1 (or testis determining autosomal) which is autosomal.

There may be other autosomal genes also and one (*Tas*) is located on chromosome 17 of mice causing sex reversal. Another gene is testis is ferminizing gene (*Tfm*) causing XY mice to become females.

2. Sex Reversal in Goats and Human Beings

Cases of XX sex reversal were also reported in goats and humans. In human beings, 1 in 20,000 males, have a female (XX) chromosome constitution and 1 in 100,000 females have a male (XY) chromosome constitution.

It has been shown that these XX sex reversed males may result from either of the following two reasons : (i) due to transfer of a segment (called *TDF* = testis determining factor) of Y-chromosome to X due to abnormal crossing over, in the same manner as the *Sxr* is transferred from Y to X in mice or (ii) due to mutation in a locus (which represses testis development), so that XX individuals now develop testes.

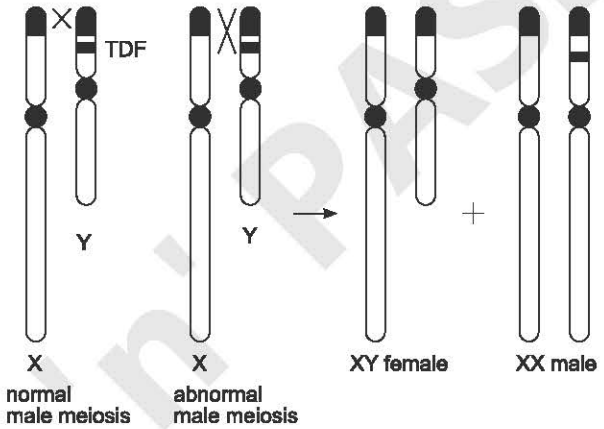


Fig. 2 : Chromosome constitution of XY male human and the transfer of a segment of Y to X-chromosome due to abnormal crossing over, leading to sex reversed XX males.

Most of the XX males are found to possess Y derived DNA sequences thus supporting the first model. It is possible that in different cases of XX sex reversed males, each of these two models holds good. In humans, XY-females have been shown to be due to a duplication on chromosome 21, carrying gene *DAX*1.

Q.6. Describe the Criss-cross inheritance with the help of diagrams.

Ans.

Criss-cross Inheritance

In a reciprocal cross in *Drosophila* when a red-eyed male was crossed to a white-eyed female, the F_1 offspring instead of being all red-eyed consisted of 50% red-eyed and 50% white-eyed and all the red-eyed offspring were females and all the male offspring were white-eyed. When these F_1 offspring were interbred, their F_2 offspring consisted of red and white-eyed individuals in equal proportion in both the sexes. In *Drosophila*, therefore, sex-linked traits such as white-eye colour follows a criss-cross inheritance. The male transmits his sex-linked traits to grandsons through his daughters.

From these observations **Morgan** concluded :

1. Criss-cross inheritance helps in establishing relationship between genes and sex chromosomes.
2. It provides evidence that sex-linked genes are located on X chromosome.
3. It helps in understanding mechanism of inheritance of sex-linked disorders in human beings.

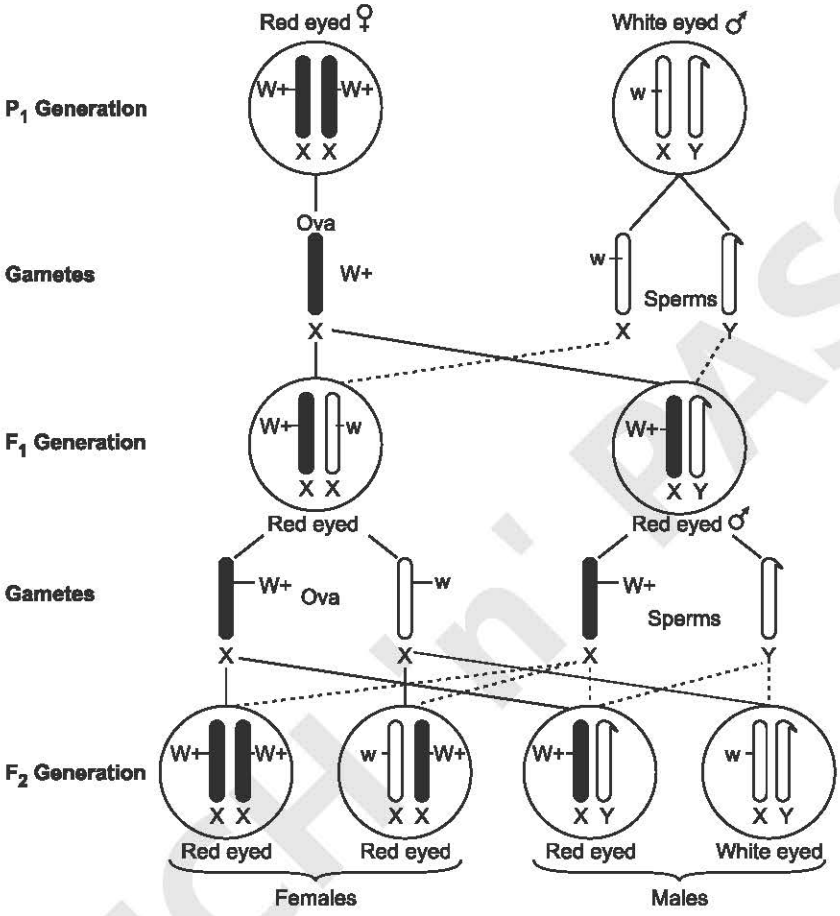


Fig. 1 : Sex-linked inheritance in *Drosophila melanogaster*. The transmission of sex chromosomes carrying eye colour gene W^+ and w in a cross between red-eyed female and white-eyed male.

The inheritance of white eye colour in *Drosophila* can be explained by assuming that :

(i) Gene for white eye colour is located in the X chromosome and Y chromosome is empty carrying no normal allele for white eyed colour.

(ii) The white eyed female possesses gene for white eyed colour (w) on both of its X chromosomes.

(iii) The white-eyed males receive X chromosome with w gene from the mother and Y chromosome with no such w-gene from father. Therefore, recessive w-gene in hemizygous state (*i.e.,* represented singly) produces white eye colour in all the male offspring.

(iv) The daughters receive one X chromosome with w gene from mother and one X chromosome with dominant W^+ gene for red eye colour from father. Therefore, all of them are red-eyed phenotypically, though heterozygous (W^+w).

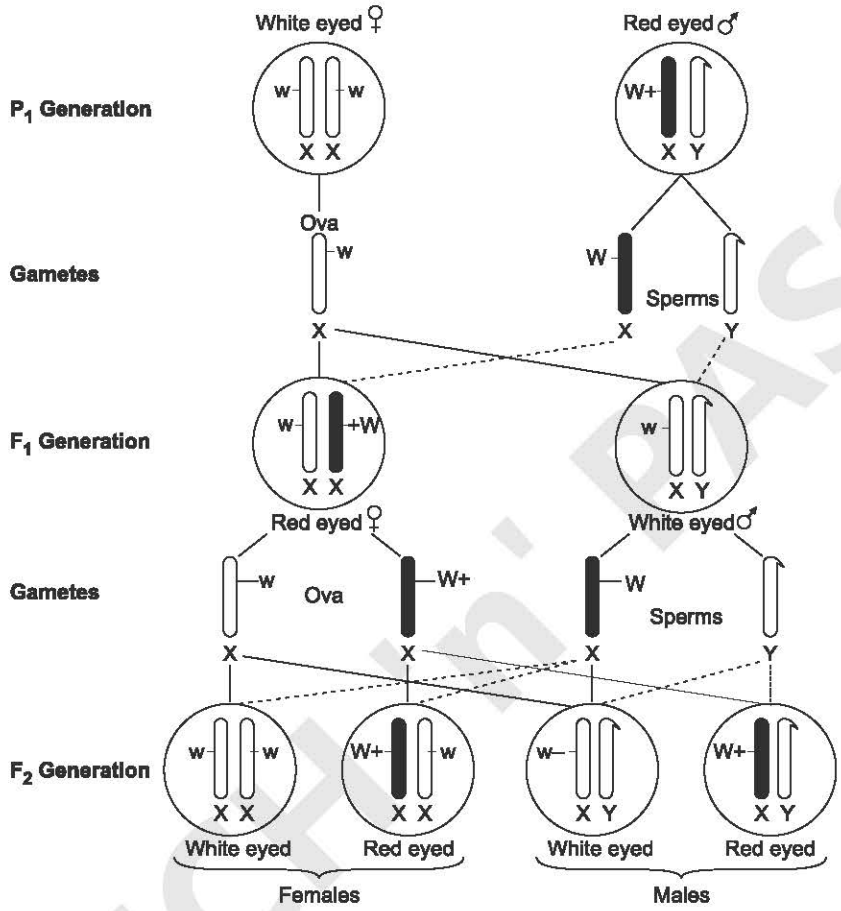


Fig. : Sex-linked inheritance in *Drosophila.* The transmission sex-chromosomes carrying eye colour gene W^+ and w in a cross between white-eyed female and red-eyed male.

Q.7. What is dosage compensation? How is this achieved? Discuss the difference in the mechanisms involved in humans and *Drosophila.*

Ans.

Dosage Compensation

In **XX-XY** mechanism of sex determination, there are two **X** chromosomes in normal females and only one **X** chromosome in males. It means females have two copies of every **X** linked gene and males have only one copy. This situation should create a genetic dosage problem for sex-linked genes between males and females. Animals overcome this problem through **dosage compensation.** The dosage compensation is the mechanism by which the effective dosage of **X** linked genes in both sexes is made equal or nearly equal, so that the X linked genes produce the same phenotypic effect in single or double dose.

Mechanism of Dosage Compensation

The dosage compensation can be achieved in two possible ways :

(i) Enhancement of gene expression almost two fold by stimulated rate of gene transcription of sex-linked genes from single **X** chromosome of males (in *Drosophila*).

(ii) Inactivation or facultative heterochromatisation of one of the two **X** chromosomes in females (**Lyonisation** in mammals).

(iii) By halving the activity of X linked genes on both X chromosomes in female (in worm *Caenorhabditis elegans*).

1. Dosage Compensation in *Drosophila*

In *Drosophila,* there is no inactivation of one of the **2X** chromosomes in females as in mammals. Instead, there is two fold increase in the transcription of **X** linked genes, present on single **X** chromosome in male *Drosophila.* It means more mRNA is synthesized to match the level of expression in males to the level of expression in females, that have **2X** chromosomes.

H.J. Muller in 1932, while studying eye pigment in *Drosophila,* noticed that the level of products of most sex-linked genes in two sexes remains the same inspite of different number of copies. He called this regulatory mechanism as **dosage compensation.** The first evidence that dosage compensation in *Drosophila* is achieved by regulating transcription of mRNA from **X** linked genes came from **Dr. A.S. Mukherjee** and **W. Beerman** (1965). In male *Drosophila,* the **X** linked genes are transcribed at twice the level of comparable genes in females. In mosaic individuals with **XX** and **XO** cells, the hyperactivity can be demonstrated in **XO** cells. The mutant and wild eye colour in both male and female flies develops with the same intensity. The level of several enzymes such as **6-PGD** (6-phosphogluconate dehydrogenase), **G-6PD** *(glucose-6 phosphate dehydrogenase), tryptophan pyrolase* and *fumarase* are found to be the same in both sexes.

In *Drosophila,* four autosomal male specific lethal genes are associated with dosage compensation. Their activity is regulated by **master switch gene (Sxl)** that controls sex determination by synthesizing sex-lethal protein. One autosomal gene, **mle (malelesss)** encodes a protein, that binds to numerous sites along **X** chromosome. It causes enhancement of genetic expression. The products of other three autosomal genes are required for **mle** binding.

The gene of **Sxl** in **XX** flies turns on the **tra** gene, which leads to female differentiation. The **Sxl** gene also inactivities one or more male specific autosomal genes, **mle.** In **XY** flies, **Sxl** is not expressed and autosomal genes are activated causing enhanced activity of **X** chromosome.

2. Dosage Compensation in Mammals

In mammals, dosage compensation of sex-linked genes is achieved by the inactivation of one of the two **X** chromosomes present in female. The inactivation of **X** chromosome is random, and may be either maternal or paternal, during early stage of development. This inactivation of either of the two **X** chromosomes results in the formation of a heterochromatic **Barr body** in the somatic cells.

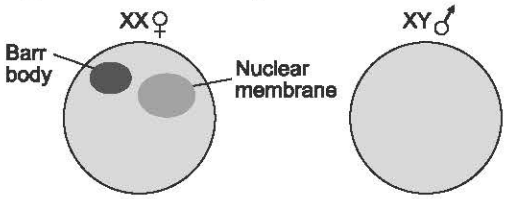


Fig. : A. Nucleus of a female mammalian cell with a Barr body (see arrow). B. Nucleus of a male mammalian cell without a Barr body.

This was discovered by **Murray Barr** and **Ewart G. Bertram** in cats. According to **Mary Lyon** (1961), out of the two **X** chromosomes, one remains functional and other becomes heterochromatic and forms a sex **chromatin body.** This is called **Barr body.** This mechanism is called **lyonization** or inactivation of **X** chromosome.

A **Barr body** is a darkly stained condensed, heterochromatic **X** chromosome observed in the nucleus of all cells of female mammals, that contain two or more X chromosomes. The number of Barr bodies in each mammalian cell is one less than the number of **X** chromosomes.

❑

UNIT-VI

Extensions of Mendelism, Genes and Environment

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Describe the genetics of seed colour in maize.

Ans. In maize, the purple colour of the seed is dependent on the dominant alleles of several genes. Plants which are homozygous for the recessive alleles of any one of these genes fail to develop anthocyanin and, therefore, their seeds are white in colour. About three genes **A, C** and **R** are found to be associated with the development of colour. The plants having genotypes **aaccRR** or **AAccRR** or **AACCrr** or **AAccrr** produce non-purple seeds.

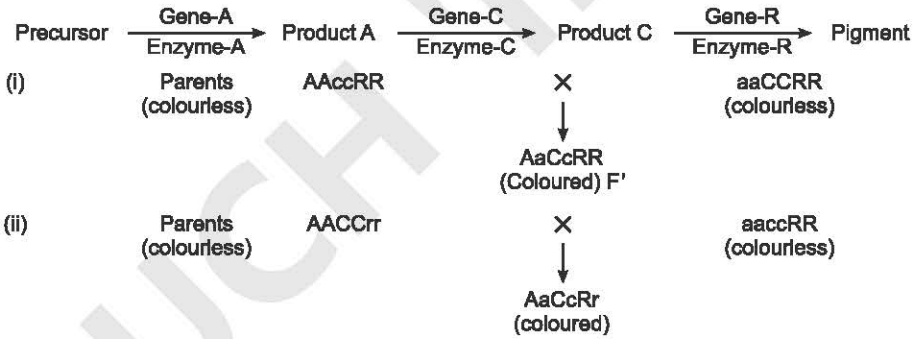


Fig. : Biochemical basis of development of seed colour in maize.

Q.2. How many types of skin colour are found in rabbits? Based on these colours, classify the rabbits.

Ans. In rabbits, four kinds of skin colour are known. Rabbits are accordingly classified as coloured (agouti), chinchilla, Himalayan albino and albino.

Q.3. Describe the importance of *R* locus on chromosome 10 in maize.

Ans. *R* locus on chromosome 10 is a complex locus, controlling anthocyanin colour of seed and the plant. Following four alleles were earlier designated as R^r, R^g, r^r and r^g. But later, due to their two independent functions, they were designated as *SP*, *Sp*, *sP* and *sp* respectively.

R^r = *SP* = coloured seed and coloured (red) plant

R^g = *Sp* = coloured seed and green plant

r^r = *sP* = colourless seed and coloured (red) plant

r^g = *sp* = colourless seed and green plant

The new designations were based on the assumption that *S* and *P* are two closely linked genes rather than forming a series of multiple alleles. But since alleles later called pseudoalleles, can undergo crossing over between them, the concept of multiple alleles may apply in this case also.

Q.4. Write very short note on universal donor and universal recipients.

Ans. Type O negative blood does not have any antigens. It is called the 'universal donor' type because it is compatible with many blood type. Type AB positive blood is called the universal recipient type because a person who has it can receive blood of any type.

Q.5. Write phenotypes and genotypes of 'Dorset' and 'Suffolk' sheep for horned trait in sheep.

Ans. Among *Dorset* sheep, both sexes are horned and homozygous (h^+h^+). In *Suffolk* sheep, both sexes are hornless and homozygous recessive (*hh*). When *Dorset* (horned) are crossed to *Suffolk* (hornless), the F_1 (h^+h) are found to be horned males and hornless females. When F_1's (h^+h) are intercrossed, in the resulting F_2 progeny, males segregated as 3 (horned) to 1 (hornless), while females segregated as 1 (horned) to 3 (hornless). It is thus obvious that expression of only heterozygote (h^+h) is influenced by sex, h^+h being horned in male and hornless in female. In other words, while horned character is dominant in male, it is recessive in female. This influence is believed to be mainly due to male and female hormones.

Phenotypic expression of different genotypes for horned character in male and female sheep are given as follows :

Genotype	Males	Females
h^+h^+	horned	horned
h^+h	horned	hornless
hh	hornless	hornless

Q.6. Describe sex influenced trait in human, for which genes are located on autosome.

Ans. Certain characters in human beings which are not located a sex chromosomes, are also believed to be sex influenced. For instance, white forelock (areas of skin completely without pigment), absence of upper lateral incisor teeth, a type of enlargement of terminal joint fingers, harelip (a fissure in upper lip), cleft palate (a fissure in roof of mouth) and stuttering (involuntary repeats of letters or words) are more common and more severely influenced in males.

Q.7. What is extrachromosomal inheritance?

Ans. Extrachromosomal inheritance is defined as a form of a non-mendelian pattern of inheritance that is governed by the DNA persent in the cytoplasm. It refers to the transmission of genes that occurs outside the nucleus, so also known as extra nuclear inheritance found in most eukaryotes.

Q.8. What is sigma factor?

Ans. Sigma factor are subunits of all bacterial RNA polymerases. They are responsible for determining the specificity of promotor DNA binding and control how efficiently RNA synthesis (transcription) is initiated.

Q.9. Differentiate between nuclear and cytoplasmic inheritance.

Ans. Cytoplasmic inheritance is the transferring of genes present in the organelles of the cytoplasm while nuclear inheritance is the transferring of genes present on the chromosome.

Q.10. Define male sterility.

Ans. **Male sterility** is defined as the failure of plants to produce functional anthers, pollen, or male gametes.

Q.11. What is the differences between suballeles and isoalleles?

Ans. In maize the **Rr** series for the purple aleurone and purple plant colour behaves as though it was a compound gene. **Stadler** has found that these genes from different strains are not exactly alike, because they result in different distributions of the pigment. These alleles are known as **suballeles.**

Alleles which produce the same phenotypic effect in homozygous state, but nevertheless differ, are referred as **isoalleles.** In *Drosophilia,* whites of different origin are not alike. When separate strains of whites are crossed to flies which carry one of the intermediate alleles (as eosin), the hybrids are not alike, some might show more dilution of eosin than others.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Describe briefly the genetics of MN blood groups.

Ans.

M-N Blood Groups

In the course of their investigations of human blood antigens, **Landsteiner** and **Levine** in 1927 discovered **M** and **N** antigens, which when injected into rabbits or guinea pigs, stimulated antibody production in the serum of the experimental animals.

Human population can be divided into blood group **M,** blood group **N** or blood group **MN** depending upon the presence of these antigens, but their **serum does not contain antibodies.** If human blood is injected into rabbit, the rabbit develops antibodies in its serum. These antibodies are found to be antagonistic to different antigens. **Landsteiner** and **Levine** in 1927 found that human population can be divided into three categories (groups) on the basis of their reaction which antibodies developed in rabbit. These categories are :

Blood groups M/N, their Genotype, Antibodies and Antigens in Man

Blood Group	Red Blood Cells reaction with Antibodies				Serum Antibodies
	Genotype	Anti-M	Anti-N	Cellular Antigens	
M	M/M	+	–	M	None
MN	M/N	+	+	M/N	None
N	N/N	–	+	N	None

In **MN** allelic series, no recessive alleles are known for the absence of either antigen. The pedigree analysis of **M** and **N** individuals presents the existence of 1 : 2 : 1 F_2 phenotypic ratio, showing phenomenon of **codominance.**

Ss Antigens

In 1947, another pair of antigens **S/s** were found to be associated with **M** and **N** series. The genes for **S** and **s** antigens (**MS** and **NS**) are not allelic to the **MN** genes. **Race** and **Sanger** (1968) have described these genes as codominant and closely linked and inter-related with **MN** genes. The different gene combinations of **MN** and **Ss** series can be : **MS, Ms, NS, Ns, MNSS, MNSs** or **MNss.**

Q.2. Write a short note on *Rh* blood group and erythroblastosis fetalis.

Ans. *Rh* Blood Groups and Erythroblastosis Fetalis

Rh factor was discovered by **Landsteiner** and **Wiener** in 1940 from rabbits immunized with the blood of **rhesus monkeys** (*Macacus rhesus*). The resulting antibodies in rabbit serum agglutinated blood of not only rhesus monkeys, but also of certain percentage of human beings and these human beings were called *Rh* + (*Rh* positive). The *Rh* antigen can produce antibodies in human serum also, which may be possible through wrong transfusion of blood. Therefore, to avoid agglutinization, cross-compatibility of *Rh* factor as well as ABO blood groups is necessary before transfusion of blood is made. *Rh* individuals should always be given *Rh* negative blood to avoid subsequent reaction due to antibody formation.

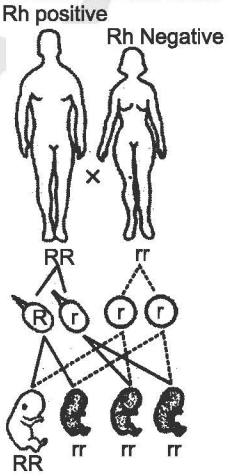


Fig. 1 : Rh Blood groups and its

A serious problem occurs, when father is *Rh* positive and mother is *Rh* negative. In such cases all children born will be *Rh* positive, if the father is homozygous (*RR*). Similarly, half the children will be *Rh* positive, if the father is heterozygous (*Rr*). If *Rh* negative mother carries a *Rh* positive fetus, in the first case of pregnancy, no serious problem due to *Rh*+ antigen in child's blood arises, since the concentration of antibodies produced in mother's blood due to immunization by child's *Rh*+ antigen, will be rather low.

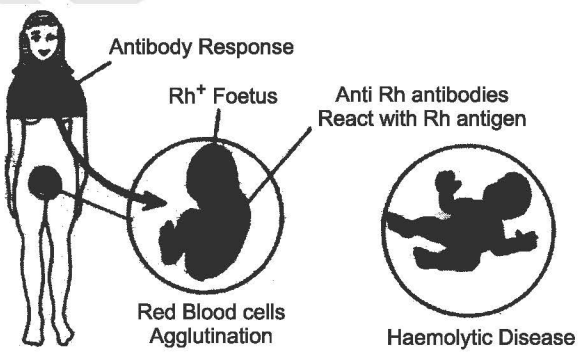


Fig. 2 : Erythroblastosis in man.

But subsequent *Rh*-positive children will increase the concentration of antibodies in mother's blood due to immunization and this blood while circulating through the fetus may cause death of the **fetus** due to **homolytic jaundice** and **anemia.** This disease is called erythroblastosis is fetalis.

Q.3. Describe the theories of multiple allelism.

Ans.

Theories of Multiple Allelism

(i) Theory of Point Mutation : According to this theory, the multiple alleles arise as a result of mutations of a gene at a given locus producing different effects.

(ii) Theory of Pseudoallelism or Close Linkage : According to this theory, multiple alleles do not represent gene mutations at a given locus but they occupy different loci which lie in close association in a chromosome. These closely linked genes have been called **pseudoalleles.** These affect the expression of normal genes showing the position effect.

In *Drosophila,* the normal eye colour is red, but there are other shades like white, pink, apricot, etc. When apricot flies are crossed with white, the F_1 hybrids are intermediate, proving allelic nature of the two genes. But recently **Lewis** found that these hybrid flies produced some red offsprings when crossing over is greatly increased in the vicinity of white locus. It means these red flies are cross-overs. **Lewis,** therefore, concluded that apricot and white are not actually alleles but they occupy adjacent loci and are pseudoalleles. These suppress the expression of normal gene.

(iii) Heterochromatin Theory of Allelism : Chromosomal breakage and rearrangement sometimes bring the heterochromatin next to genes, and suppresses their expression. In maize, there is evidence that position effects are sometimes due to the transposition of very minute fragments of heterochromatin, the fragments too small to be visible under the microscope.

If is often difficult to tell which of these three theories explains a particular case of allelism and, in fact, it is possible that all the three apply in different cases.

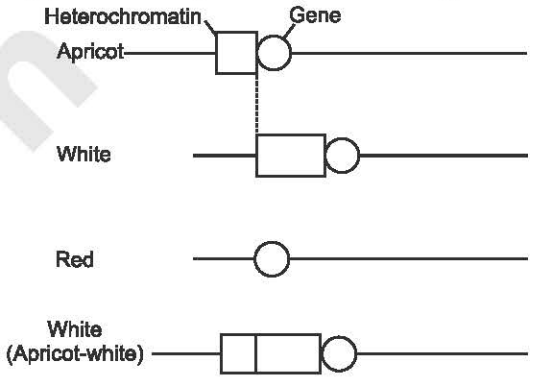


Fig. : Diagrammatic representation of heterochromatin theory of multiple allelomorphism.

Q.4. What do you understand by sex influenced traits.

Ans.

Sex Influenced Traits

In some organisms, traits are influenced by sex of the organism. In other words, male and female individuals, which are genotypically similar for a particular trait, give different expressions of the same trait, thus demonstrating that the trait is influenced by sex. Such traits are called sex-influenced traits. This can be illustrated with the help of example of horns in sheep, which are effectively controlled by a single gene, since other genes controlling this trait are always homozygous.

1. **Horned trait in sheep :** Among *Dorset* sheep, both sexes are horned and homozygous (h^+h^+). In *Suffolk* sheep, both sexes are hornless and homozygous recessive (hh). When *Dorset* (horned) are crossed to *Suffolk* (hornless), the F_1 (h^+h) are found to be horned males and hornless females. When F_1's (h^+h) are intercrossed, in the resulting

F_2 progeny, males segregated as 3 (horned) to 1 (hornless), while females segregated as 1 (horned) to 3 (hornless). It is thus obvious that expression of only heterozygote (h^+h) is influenced by sex, h^+h being horned in male and hornless in female. In other words, while horned character is dominant in male, it is recessive in female. This influence is believed to be mainly due to male and female hormones.

Phenotypic expression of different genotypes for horned character in male and female sheep are given as follows :

Genotype	Males	Females
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2. **Sex influenced autosomal traits in humans :** Certain characters in human beings which are not located on sex chromosomes, are also believed to be sex influenced. For instance, white forelock (areas of skin completely without pigment), absence of upper lateral incisor teeth, a type of enlargement of terminal joint fingers, harelip (a fissure in upper lip), cleft palate (a fissure in roof of mouth) and stuttering (involuntary repeats of letters or words) are more common and more severely influenced in males.

Q.5. Describe anticipation in genetics using suitable examples.

Ans.

Anticipation : Triplet Diseases

Anticipation is a phenomenon, where the signs and symptoms of some genetic conditions tend to become more severe and/or appear at an earlier age, when the disorder is passed on from one generation to the next generation. This phenomenon was often observed, when the disease is passed on from female and not when it is transferred from the male parent, so that the phenomenon is sex influenced. The term 'anticipation' was used for the first time by **Mott** in 1910, who observed that the trait 'insanity' appeared earlier in the offspring than in the parents. The phenomenon was later described by **Harper** and his coworkers in 1922 and by **Bell** in 1947. Although, initially several geneticists, including **Penrose, Haldane** and **Fisher** questioned the phenomenon of 'anticipation', the phenomenon is confirmed and a number of diseases are known to exhibit the phenomenon of anticipation. Most of this work involving anticipation was initially undertaken during 1980s and early 1990s.

Some Examples of human diseases showing anticipation are given as follows :

Disease	Repeat
I. Autosomal dominant	
(i) Several spinocerebellar ataxias	—
(ii) Huntington disease	CAG
(iii) Myotonic dystrophy	CTG
(iv) Dyskeratosiscongenita	TTAGGG (telomere repeat)
II. Autosomal recessive	
Friederich ataxia	GAA
III. X-linked	
Fragile X syndrome	CGG

The Sherman Paradox : In order to explain the abnormal inheritance pattern of fragile-X syndrome, an American geneticist named **Stephanie Sherman** observed that the effect of fragile X-syndrome occurred more frequently with passage of each generation. This is popularly described 'Sherman Paradox'. The phenomenon was explained by the expansion of permutations to full mutations, which occurs during female meiosis but not male meiosis. Thus NTMs may pass on the unexpressed pre-mutation to their offspring, but only their daughters may pass on the expressed full mutation.

Q.6. Write a note on genome imprinting.

Ans.

Genomic Imprinting

This process wherein a **gene** is differentially expressed depending on whether it has been inherited from the mother or from the father. Such "parent-of-origin" effects are known to occur only in sexually reproducing placental mammals. Imprinting is one of a number of patterns of inheritance that do not obey the traditional Mendelian rules of inheritance, which assume indifference about the parental origin of an **allele** (an allele is any one of two or more genes that may occur alternatively at a given site on a **chromosome**). Traits are, therefore, able to be passed down maternal or paternal lines.

Imprinting Mechanisms

Imprinting can occur when one of the gene's parental alleles is silenced throughout the embryonic development of the individual by an alternation in parental DNA made during parental gametogenesis (the formation of gametes, or sperm in males and eggs in females). The other parental allele is, therefore, allowed expression during embryonic development. A mechanism by which this occurs is DNA methylation (the addition of a CH_3, or methyl, group to specific regions of DNA) at impriting control regions (ICRs). Intracellular DNA-reading mechanisms exist after **fertilization** to check that the correct parental allele has been allowed differential expression.

Imprinting and Fetal Development

Imprinting has been able to explain certain predicaments of life in utero. A number of imprinted genes are related to embryonic and fetal growth and thus the extraction of resources from the uterine **environment** for growth. Mother and father, however, have different interests in how resources are extracted, because of asymmetrical parental investment in each given child. This arises from the fact that mothers can only have one child every nine months for approximately 20 years, whereas a father could conceivably impregnate many different women from puberty until death.

Systematic knockout (inactivation) studies of key imprinted genes, especially as performed on mice, have provided support for the hypothesis that imprinted genes that allow expression of paternally inherited alleles tend to drive more extraction of nutrients from the mother during gestation and after birth to produce a larger child. In contrast, imprinted genes that allow expression of maternally inherited alleles will tend to drive mechanisms to prevent the disproportionate utilization of resources by the fetus. A commonly cited example of this differential resource transfer in mice is the paternally expressed gene *lgf*2 (insulin-like growth factor 2), which enhances fetal growth and placental nutrient transport capacity, and the maternally expressed l*gf*2 receptor (*lgf2r*), which degrades excessive lgf2 protein.

Many of the effects of imprinted genes occur in the **placenta,** a crucial site for resource and nutrient transfer. For example, an overgrown placenta (hydatidiform mole) results when maternal imprints are missing. Additionally, in Silver-Russell syndrome (or Russell-Silver syndrome), a maternal uniparental disomy (both copies of a chromosome or partial chromosome are inherited from one parent), growth restriction is present. Similar effects are found in other cases of disordered imprinting. **Preeclampsia,** for example, in which disordered imprinting has been implicated, also demonstrates growth restriction in utero. Many of these diseases can be understood only within the context of imprinting as a common mechanism of parental conflict and manipulation of phenotypic outcome of children.

Q.7. What do you understand by sex-limited traits? Explain in brief.

Ans.

Sex-limited Traits

In cases of sex influenced traits, same allele had different expressions in male and female individuals, but both alternative forms of trait are known in each sex. For instance, both horned and hornless sheep are known as male and female. In still other cases, we find, that a particular trait may not express at all in one of the two sexes.

1. **Plumage in poultry :** In poultry, plumage is one such character, where **cock feathering** is never seen in female (i.e. it is sex limited), although **hen feathering** is common among males. In most breeds of domestic poultry, male is cock feathered and female is hen feathered, so that plumage of two sexes is strikingly different. However, in some cases (e.g. some Seberight bantams), both sexes are hen feathered while in still other cases (e.g. Hamburgh breed), males may be either hen feathered or cock feathered. It has been shown through appropriate crosses, that *hen feathering* (h^+) is a dominant character, so that both male as well as female with genotype h^+h^+ or h^+h will be *hen feathered.* However, homozygous recessive (*hh*) is *cock feathered* in male, but *hen feathered* in female, suggesting that in the presence of female hormone, *hh* cannot express its cock feathering character. This was confirmed through experiments of gonadectomies (removal of gonad). When ovary of *hh* female was removed, it resulted into cock feathering. Similarly in castrated males (testes removed), expression of h^+ is inhibited.

   Phenotypic expression of different genotypes for plumage in two sexes of poultry is given as under.

Genotype	Males	Females
h^+h^+	hen feathered	hen feathered
h^+h	hen feathered	cock feathered
hh	cock feathered	cock feathered

2. **Milk production in cattle :** Milk production in cattle is also sex limited, since genes for milk production are carried both by males and females, but they express only in females.
3. **Baldness in humans :** In man also, some traits have been considered to be sex limited e.g. premature baldness (baldness in twenties or early thirties). On the basis of statistical analysis, it was suggested that premature baldness is controlled by a dominant gene, which expresses only in the presence of a certain level of male (androgenic) hormone, which is never reached in females, but is always reached in normal male. This premature baldness is, however, very different than other cases of baldness which are explained by abnormalities in thyroid metabolism and infectious diseases.

Q.8. What is maternal effect? Explain with the example of pigment in flower moth.

Ans.

Maternal Effect

Maternal effects are defined as influences of the environment provided by mothers on offspring that act in addition to or in interaction with offspring genotype and other environment influences on offspring.

Pigment in Flower Moth

A distinct case of maternal effect was discovered in flour moth (*Ephestia kuhniella*) by **Caspari** (1936). Dark brown eyes and presence of pigment in other parts of the body in this moth are controlled by a dominant gene *A*, responsible for production of a pigment precursor **kynurenine**. Homozygous recessive *aa* lacks kynurenine, so that it exhibits absence of pigment and the eyes therefore have red colour. When heterozygote *Aa* (pigmented) is crossed to non-pigmented homozygous recessive *aa* (*aa* ♀ × *Aa* ♂), as expected, progeny segregates 1 *Aa* : 1 *aa*, which phenotypically gives the ratio 1 pigmented : 1 non-pigmented. In the reciprocal cross, pigmented *Aa* (♀) × non-pigmented *aa* (♂), the progeny (1*Aa* : 1 *aa*) had all the early larvae pigmented. In this case, however, when larvae matured, only half of them (*Aa*) were dark brown eyed, the other half (*aa*) were red eyed. These homozygous (*aa*) pigmented larvae received their egg cytoplasm from mother (*Aa*) and, therefore, had **kynurenine** in early stages of development.

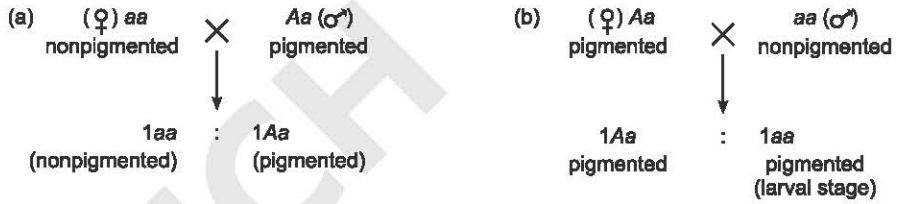


Fig. : Results of reciprocal crosses (♀ non-pigmented *aa* × ♂ pigmented *Aa*; ♀ Pigmented *Aa* × ♂ non-pigmented *aa*) in flour moth (*Ephestia kuhniella*) showing inheritance of pigment colour.

Q.9. Define the transgressive variations. Also give the differences between monogenic and polygenic inheritance.

Ans.

Transgressive Variations

In polygenic inheritance some offspring may exhibit more extremes than either parent or grandparent. For example, some children are shorter on taller than either parent or any of their more remote ancestors. The same is true with respect to intelligence. These are called **transgressive variations.**

Such variations occur because of polygenic inheritance brings together more dominant genes. **Punnett** and **Bailey** (1914 and 1923) crossed the large Golden Hamburg chicken with the smaller Sebright Banta. The F_1 was intermediate in weight but a few of the F_2 birds were heavier or lighter than either of the parental individuals. **Punnett** and **Bailey** presumed that four pairs of genes are associated with the weight in these chickens. The genotype of Golden Hamburg presumed to be **AABBCCdd** and Sebright Banta **aabbccDD.**

Differences between Monogenic Inheritance and Polygenic Inheritance

S.No.	Monogenic or Mendelian inheritance	Polygenic inheritance
1.	F_1 hybrid resembles one of the parents having dominant character.	F_2 hybrid exhibits character intermediate between the two parents.
2.	F_2 individuals distinctly fail into two categories and exhibit distinct phenotypes resembling either of the parents.	F_2 progeny exhibits a greater variability and a graded series of phenotype from one parent to other.
3.	Individuals with dominant phenotype are more numerous (2/3rd) and those with recessive phenotype are less in number.	Individuals with average or intermediate phenotype are more numerous than either of the parental characters.

Q.10. Define the plastid inheritance in 4'o clock.

Ans.

Plastid Inheritance

Although a number of cases are now known in plants, where inheritance of plastids is controlled by genes, there are also cases where this character is transmitted through the agency of cytoplasm alone. Since major part of cytoplasm in zygote is derived from egg, inheritance in such cases will be maternal.

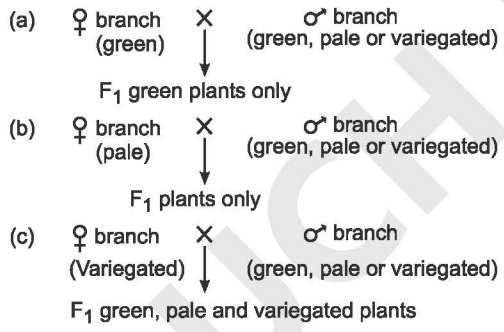


Fig. 1 : Plastid inheritance in four o'clock plant showing dependence on the nature of female branch.

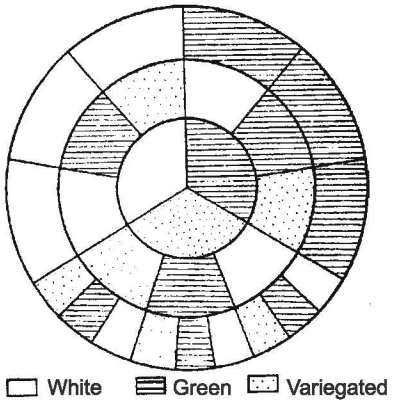


Fig. 2 : Plastid inheritance in four o'clock. The central circle represents the type of branch that produces flower which are pollinated. Intermediate circle represents branch from which pollen is used and outer circle shows progeny.

In four o'clock plant (*Mirabilis jalapa*), three kinds of branches with respect to occurrence of plastids may be found. These are : (i) completely green, (ii) completely pale green or (iii) variegated. In such cases, phenotype of progeny will depend upon phenotype of branch of which flowers are pollinated. Results from three kinds of branches used as female parents in figure.

As would be evident, when variegated branches are used as female source, both green and pale plastids are present in cells of female parent. Therefore, female gametes may carry either green or pale plastids or both. Consequently, three kinds of plants namely green, pale and variegated plants would be obtained.

Q.11. Write a short note on Twin's studies.

Ans.

Twin Studies

To evaluate the role of environment and heredity in human beings, **Spath** and **Galton** proposed the study of twins. **Identical twin** or **monozygotic twins (MD)** are two genetically homogeneous human beings, because these arise by the division of a single fertilised egg and inherit identical genotype. It means the genetic differences between monozygotic twins are absent and the observed phenotypic differences could be purely environment. Most identical twins are reared together and are exposed to very similar environments, some pairs are separated and are raised in different settings. For any particular trait, average similarities and differences are studied. Characteristics that remain similar in different environments are controlled by hereditary factors.

Similar studies can be made with **dizygotic** or **fraternal twins (DZ)** for the same sex. The fraternal twins develop from two different zygotes. Therefore, these differ both genetically and environmentally and are similar to any two siblings. By measuring both kinds of twins for the similarity and differences for a particular character, the role of environment and heredity can be evaluated.

Q.12. Describe briefly the monozygotic twins.

Ans.

Monozygotic Twins

Identical twins when reared in different environments show which character is more susceptible to environmental differences and reveal the extent of genetic influence of different characters. Although, this method has the advantage because it tells the effect of environment on identical genotypes, such cases are quite rare. **Newman, Freeman** and **Holzinger** collected data on 19 pairs of twins reared separately and 50 pairs each of identical and fraternal twins reared together and 50 nontwin siblings. The results are given below in table.

Average differences between Identical and Fraternal Twins and Siblings

S.No.	Trait	Identical Twins (MZ)		Fraternal Twins	Siblings
		Reared Together	Reared Apart		
1.	Height	1.7	1.8	4.4	4.5
2.	Weight	4.1	9.9	10.0	10.4
3.	Head length	2.9	2.2	6.2	—
4.	Head width	2.8	2.9	4.2	—
5.	Intelligence	5.9	8.2	9.9	9.8

From studying the above results, it can be concluded that height, head length and head width are genetically influenced characters and are little influenced by environment, because the value of concordance in MZ reared together and reared apart is very close. On the contrary body weight in influenced most by the environment.

Q.13. Explain the relationship between gene and environment.

Ans.

Genes and Environment

There exists an intimate relationship between the gene, the genetic makeup of the individual (genotype) and the environment to which the individual is exposed. The ultimate phenotype

is the sum total or net effect of all these factors. So, even if an individual possesses a certain genotype, there is no guarantee that the genotype will exhibit 100 per cent penetrance and full expressively. The degree to which a certain gene expresses itself into a phenotype, in a population or in an individual depends on the interacting influences of genotype and environment.

The genotype contains thousands of genes and action of many of them is inter-related. Thus, genes modify one another's effect. For example, level of expression of a trait is more similar among relatives than among unrelated individuals, provided relatives and unrelated individuals ar raised in fairly similar environments.

The genes that have secondary effect on a trait and change its expression are called **modifier genes.** For example, a **dilute gene** in cat reduces the intensity of pigmentation from back to gray. *Drosophila melanogaster,* when kept in laboratory culture for many years, the expression of a phenotype changes.

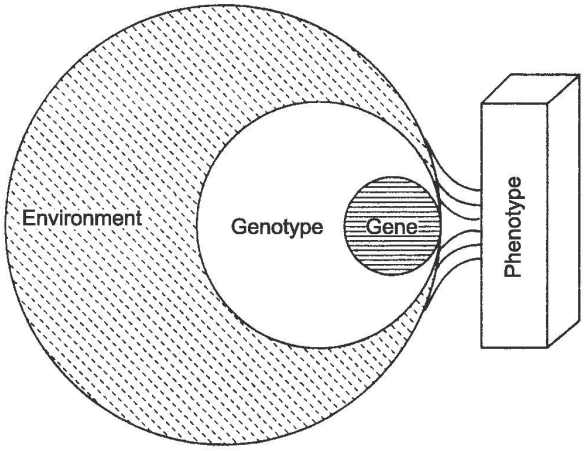


Fig. : Diagram showing relationship between gene, genotype, environment and phenotype

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. What is epistatis of gene? Explain their type and give suitable example.

Ans.

Epistatis (Inhibiting Genes)

Epistasis (Gr. act of stopping) is the interaction between non-allelic genes (present at separate loci) in which one gene masks, inhibits or suppresses the expression of other gene. The gene that suppresses the other gene is known as **inhibiting** or **epistatic factor** and the gene which is prevented from exhibiting itself is known as **hypostatic.** Although, it is similar to dominance and recessiveness but the two factors occupy two different loci.

Therefore, while dominance involves **intragenic** or **interallelic gene suppression,** the epistasis involves intergenic suppression.

Epistasis can be of the following types : 1. dominant epistasis, 2. recessive epistasis.

1. Dominant Epistasis (12 : 3 : 1 or 13 : 3 Ratio)

In dominant epistasis out of two pairs of genes the dominant allele (*i.e.,* gene A) of one gene masks the activity of other allelic pair **(Bb).** Since the dominant epistatic gene A exerts its epistatic influence by suppressing the expression of gene B or b, it is known as dominant epistasis.

Example (i) Dominant Epistasis in Dogs : In dogs white coat colour appears to be dominant. It develops due to the action of epistatic gene **I** which prevents the formation of pigment, controlled by hypostatic gene **B.** The hypostatic gene **B** produces black coat while its recessive allele **b** produces brown coat colour only when gene **I** is recessive. The progeny of dominant

gene **I** does not allow them to function and results in white colour. When two white coat dogs are crossed, they produce white, black and brown in the ratio of 12 : 3 : 1. The white dogs in this case possess gene for black or brown in colour but does not produce the pigment because of the presence of gene **I** in dominant state.

Example (ii) Fruit Colour in *Cucurbita* or Summer Squash : Fruits of *Cucurbita pepo* are white, yellow and green. White is dominant over both yellow and green and is produced by dominant inhibiting gene **W.** The yellow colour is produced by dominant hypostatic gene **G** and green by recessive hypostatic gene **g.** But these genes **G** and **g** express themselves only when **W** is recessive.

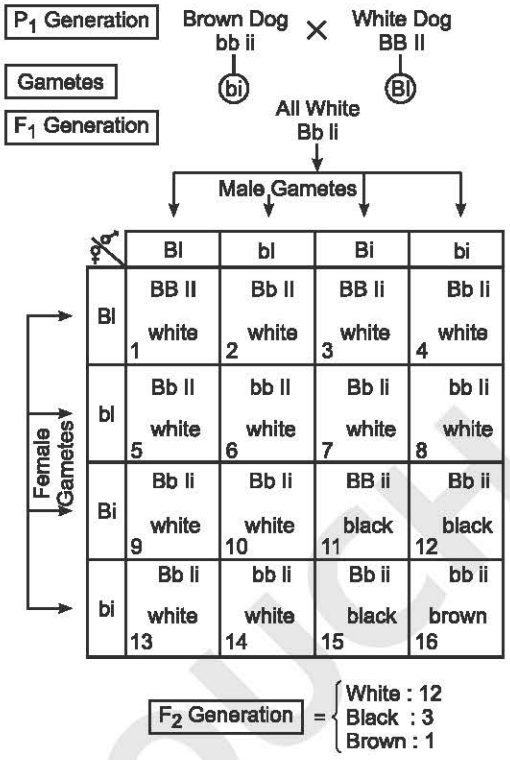


Fig. 1 : Interaction of inhibiting genes in dog for coat colour showing dominant epistasis.

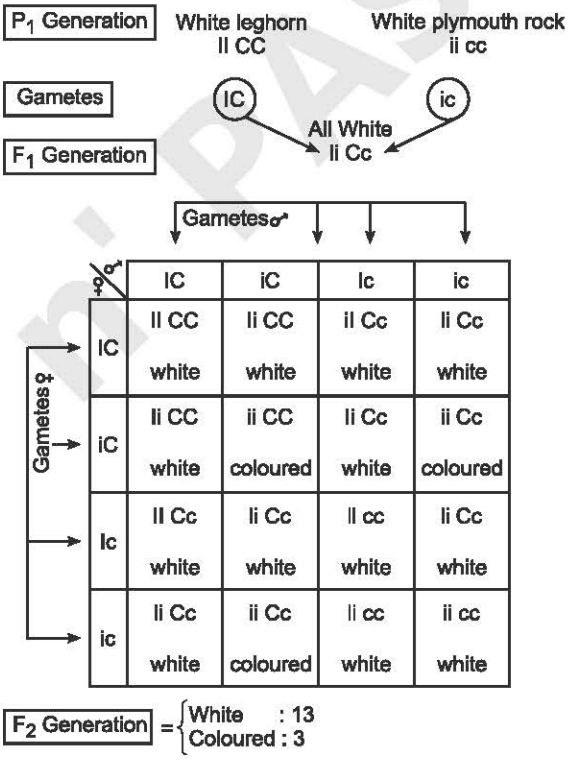


Fig. 2 : A cross between two white varieties of chickens producing white and coloured chickens in F_2 generation in the ratio of 13 : 3.

Example (iii) Coat Colour in Chickens : In poultry there are two kinds of white breeds—the white leghorn and the white plymouth rock. The white plumage of white leghorn is dominant over the coloured variety and the white plumage of white plymouth is recessive to coloured variety. When while leghorn is crossed with white plymouth in F_1 all are white, but in F_2 white and coloured forms appear in the ratio of 13 : 3, which are supposed to be the modified Mendelian dihybrid ratio. The dominance of white leghorn character is explained by that there may be present an inhibiting factor which prevents the development of pigment even when it is present. If :

I—inhibiting factor **i**—its recessive allele

C—colour factor for pigment **c**—its recessive allele

The white is produced :

(i) when both colour and inhibiting factors are present IC

(ii) when both colour and inhibiting factors are absent ic

(iii) the colour factor alone is absenet, *i.e.,* **ccII** or **ccIi.**

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

Epistasis due to recessive gene is known as **recessive epistasis,** *i.e.,* out of the two pairs of genes, the recessive epistatic gene masks the activity of the dominant gene of the other gene locus. The dominant **A** expresses itself only when the epistatic locus **C** also has the dominant gene, if the epistatic locus has recessive gene **c**, gene **A** fails to express.

Example : In mice, agouti colour, characterised by banding of hairs is controlled by gene **A,** which is hypostatic to recessive allele **c.** The dominant epistatic gene **C** in absence of **A** gives back coloured mice and in the presence of dominant gene **A** gives agouti, but dominant gene **A** is unable to produce agouti colour in the presence of gene **c.** Therefore, recessive **c** gene acts epistatic to gene **A.**

Epistasis in *Drosophila*

In *Drosophila,* two recessive wing mutants : (i) **apterous *(ap)*** produces small stubby wings (instead of normal transparent ones) and (ii) **cubitus interruptus (*ci*)** causes a small interruption in fourth longitudinal vein. The F_2 progeny of these two mutants in dihybrid cross produces 9 normal : 3 interrupted vein : 4 apterous wings.

Epistatic Genes in Man

In ABO blood group system of man, the formation of antigens **A** and **B** is controlled by two pairs of genes :

Gene **H** controls production of precursor substance **H.**

(i) Gene I^A converts the precursor—**H** into antigen **A.**

(ii) Gene I^B converts the precursor—**H** into antigen **B.**

(iii) Persons having blood group **O** and recessive allele **i** are unable to convert precursor—**H.** In them precursor—**H** remains as such in the blood and can be agglutinated by anti —**H.**

Majority of persons are either **HH** or **Hh** and produce precursor—**H.** Only a few persons are **hh.** They are unable to synthesise precursor—**H.** Such persons whether belonging to blood group **A, B** or **AB** do not form antigen—**A** or antigen—**B** and their **RBC** are not agglutinated by anti —**A,** anti —**B** or anti —**H.** Persons having this genotype are described to show **Bombay phenotype.** This phenotype is the result of recessive epistasis of recessive gene **h** over gene I^A **or** I^B.

Q.2. Write a detailed note on male sterility in plants.

Ans. **Male Sterility in Plants**

It is defined as the failure of plant to produce functional anthers, pollen or male gametes.

Types of male sterility

Male sterility in plants can be controlled by nuclear genes or cytoplasm or by both. Therefore, broadly there are atleast three different mechanisms for control of male sterility in plants. These three types would be briefly discussed in this section.

1. **Genetic male sterility :** In this type, male sterility is controlled by a single gene and is recessive to fertility, so that the F_1 individuals would be fertile. In the F_2 generation, the fertile and sterile individuals will segregate in 3 : 1 ratio (Fig. 1).

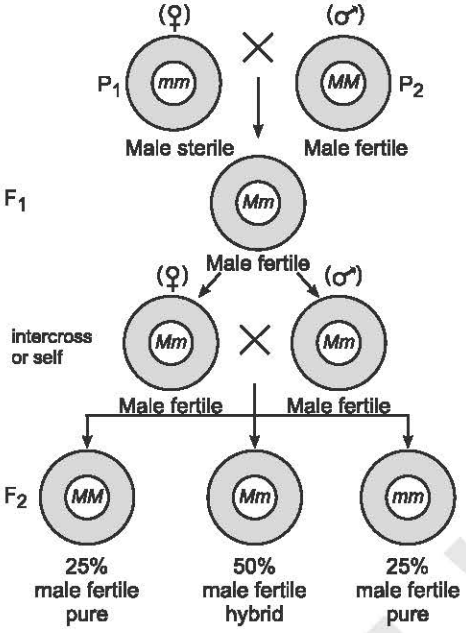


Fig. 1 : Inheritance of genetic male sterility in plants.

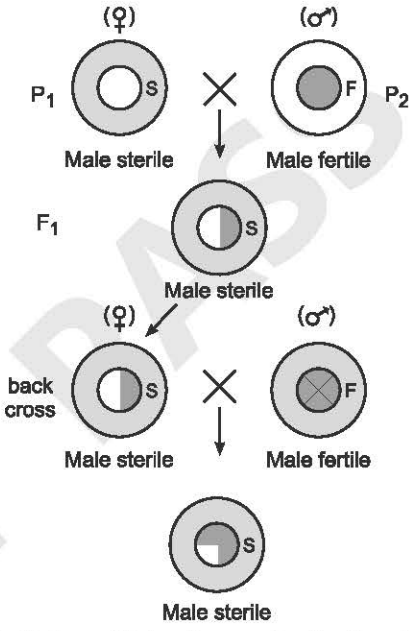


Fig. 2 : Maternal inheritance of cytoplasmic male sterility in plants.

2. **Cytoplasmic male sterility :** In several crops like maize, cytoplasmic control of male sterility is known. In such cases if female parent is male sterile, F_1 progeny would always be male sterile, because cytoplasm is mainly derived from egg obtained from male sterile female parent (Fig. 2).
3. **Cytoplasmic-genetic male sterility :** In certain other cases, although male sterility is wholly controlled by cytoplasm, but a restorer gene if present in the nucleus will restore fertility. For instance, if female parent is male sterile, then genotype (nucleus) of male parent will determine the phenotype of F_1 progeny. The male sterile female parent will have the recessive genotype (*rr*) with respect to restorer gene. If male parent is *RR*, F_1 progeny would be fertile (*Rr*). On

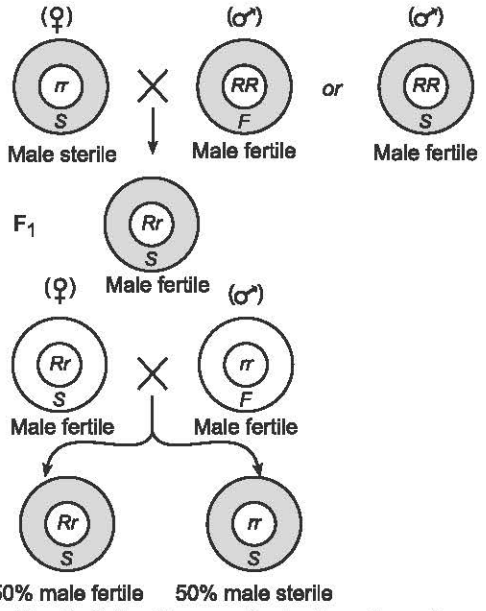


Fig. 3 : Inheritance of cytoplasmic male sterility and restoration of fertility due to restorer gene.

the other hand, if male parent is *rr*, the progeny would be male sterile. If F_1 individual (*Rr*) is testcrossed, 50% fertile and 50% male sterile progeny would be obtained.

During 1990-92, transgenic male sterile and fertility restorer plants have also been produced in *Brassica napus,* by transfer of *'barnase'* and *'barstar'* genes from *Bacillus amyloliquefaciens.*

Cytoplasmic Male Sterility in Maize

Rhoades in 1933, reported the analysis of first cytoplasmic male sterile plants in maize and demonstrated that male sterility was contributed by female parent and that nuclear genes had no influence. This was shown by corssing male sterile plants with wide range of fertile males and by observing that in subsequent generations all progenies were male sterile. Recent studies have proved that factors responsible for cytoplasmic male sterility are located in mitochondrial DNA (mtDNA).

Q.3. Define histogram. Discuss its role in understanding of polygenic inheritance.

Ans.

Histogram

A graphic representation of frequency distribution is called a **histogram.** To form a histogram from frequency distribution of heights, the various heights are marked off on the horizontal axis and rectangles are erected to represent the various classes.

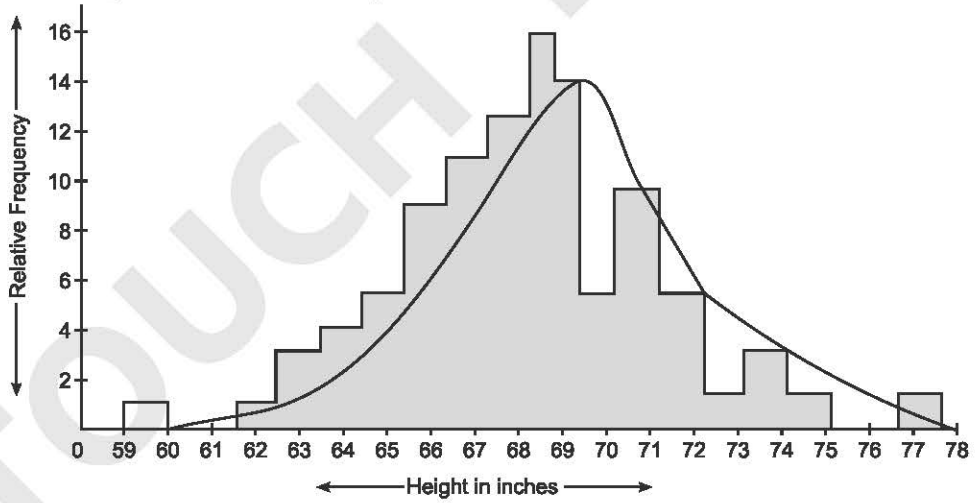


Fig. 1 : Histogram of height distribution of 100 men.

Looking at the histogram, it can be concluded that height of 68 inches is most common whereas the number of individuals declines towards its either end. Thus, in polygenic inheritance, the extreme phenotypes are rare and the intermediate ones are more frequent.

Histograms for Frequency Distribution of Characters in Monogenic and Polygenic Inheritance

In a monohybrid Mendelian segregation, the two parents belong to two distinct phenotypic classes : **homozygous dominant** and **homozygous recessive.** In F_1 due to the dominance of

one allele over the other the entire progeny shows only dominant phenotype. In F_2 generation the dominant and recessive characters (phenotypes) segregate out in the ratio 3 : 1.

In polygenic inheritance, though the parents fall into two distinct categories, F_1 progeny shows an intermediate character because dominant genes are lesser in number in F_1 generation. The F_2 progeny exhibits a wide range of variation so that a continuous curve is obtained in F_2 generation.

A comparison of the histograms showing the phenotypic distribution of F_2 of one, two or three gene segregation, indicates that the greater is the number of segregating gene pairs, the wider is the spread of the histogram curve. Therefore, from the nature of frequency distribution or of the spread of histogram, one can conclude the number of genes involved in polygenic inheritance.

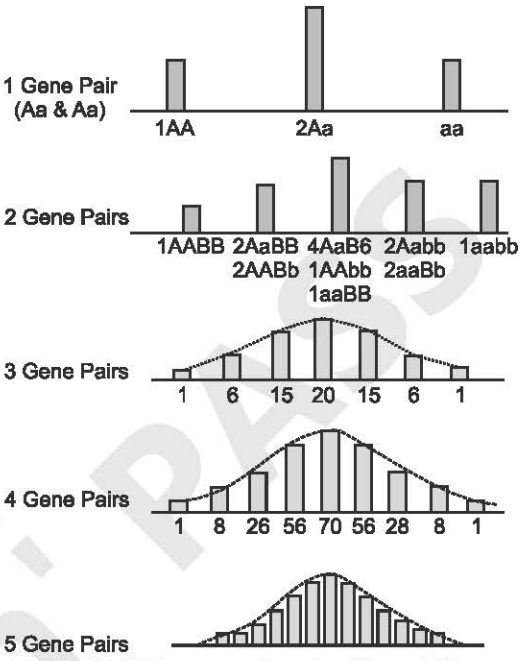


Fig. 2 : Histograms showing the relative frequency of phenotypes of F_2.

Some of the quantitative characters are controlled by single gene pairs as well as by more than one gene pairs in an additive or cumulative fashion. For example, tall character in sweet pea is controlled by polygenes as well as by a single gene pair. There is a range of variation between tall and dwarf plants due to different number of dominant genes in different phenotypes.

Q.4. What are multiple alleles? Give a brief account of multiple allelism.

Ans.

Multiple Alleles

The existence of genes is inferred from the processes of segregation and recombination of body characters. Only those genes are known which have an alternative expression. Mendel and his followers used the term **'allele'** or **allelomorph** to denote the alternative form of the normal gene. It means the genes for tall and dwarf characters of pea plant are alleles. The former is **normal allele** or **wild** and the later **mutant allele.** A gene can mutate several times producing several alternative expressions. Such genes are called **'multiple alleles'.** They occupy the same locus in homologous chromosomes and may or may not be a simple dominant and recessive relationship.

The **multiple alleles** can be defined "as a set of three, four or more allelomorphic genes or alleles, which have arisen as a result of mutation of the normal gene and which occupy the same locus in the homologous chromosomes."

Characteristics of Multiple Alleles

1. Multiple alleles occupy the same locus within the homologous chromosomes. It means only one member of the series is present in a given chromosome.
2. Multiple alleles control the same character, but each of them is characterised by different manifestations. **Sturtevent** has summarised it that **they carry the same function but with varying degree of efficiency.**

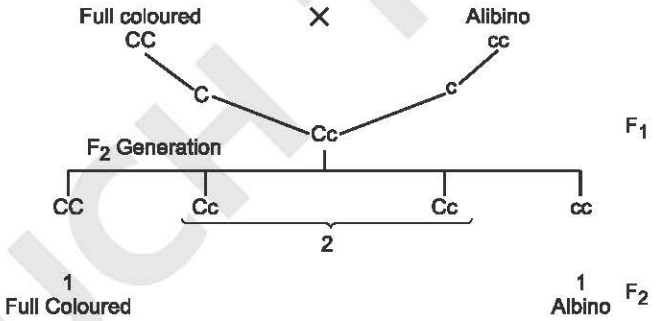
3. Crossing over does not occur in the multiple alleles.
4. Since only two chromosomes of each type are present in each diploid cell, only two genes of the multiple series are found in a cell and also in a given individual.
5. The gametes contain only one chromosome of each type, therefore, only one allele of the multiple series in each gamete.
6. The multiple alleles of a series are more often related as dominant and recessive. More commonly, the normal gene is dominant to all other mutant alleles. Even the intermediate members of the series may be related as dominant and recessive, or they may exhibit codominance.

   Therefore, multiple alleles act in some way to control the various steps in a chemical reaction.

Segregation and Recombination of Multiple Alleles

A normal gamete carries only one member out of the series and the union of two brings the two genes of an allelic series together. The two members may be in any possible combination.

Whenever the two members of an allelic series are crossed, it is found that their F_2 generation always contains homozygous, dominant, heterozygous and homozygous recessive in the ratio of 1 : 2 : 1. For example : A cross between fully coloured rabbit and albino results in **1 CC : 2 Cc : 1 cc in F_2** generation.



The principles underlying the segregation and combination are applicable to cross between any other two genes of the multiple series, *e.g.,* if chinchilla and albino are crossed, they will produce chinchilla (homo), chinchilla (hetero) and albino in 1 : 2 : 1 ratio. The appearance of F_1 and F_2 individuals will depend upon the degree of dominance of the genes involved. The alleles are not always related as dominant and recessive. Sometimes blending occurs between them.

Examples of Multiple Alleles

Many examples have been found in which more than two alternative alleles or multiple alleles for a character are present in a population.

1. Coat Colour in Rabbits

The best example of multiple allelomorphs is the albino series of coat colour in rabbits. The series consists of :

(i) Full colour or 'agouti' (wild type) — C dominant

(ii) Chinchilla (silver gray) —C^{ch}

(iii) Himalayan white (coat colour but extremities coloured) — C^{h}

(iv) Albino (pure white) —c

The crosses between homozygous agouti and albino rabbits produce uniform agouit F_1; interbreeding of the F_1 produces an F_2 ratio of 3 agouti : 1 albino. Two-thirds of these agouti individuals can be shown to be heterozygous.

Similarly, crosses between chinchilla and agouti produce all agouti individuals in F_1 and 3 agouti : 1 chinchilla in F_2. Thus genes determining chinchilla and agouti colours appear to be alleles and agouti is dominant to chinchilla. Crosses between chinchilla and albino produce all chinchilla, and albino are also allelic and related as dominant and recessive. In the same way it has been determined that Himalayan coat colour is allelic to agouti or chinchilla or albino. It is recessive to agouti and chinchilla but dominant to albino. Therefore, the genes determining coat colour in rabbits form a series, and are related in the following manner :

The gene **C** for full colour is dominant over all other alternatives. **Chinchilla** (C^{ch}) is recessive to full colour but dominant to others (Himalayan and albino). Himalayan (C^{h}) is recessive to full colour and chinchilla but is dominant over albino. The different genotypes and their phenotypes, due to multiple allelic series for skin colour in rabbit are as follows :

Genotypes	Phenotypes
CC, Cc^{h}, Cc^{a}	Coloured (wild)
$c^{ch}c^{ch}$	Chinchilla
c^{ch},c^{h},$c^{ch}c^{a}$	Light grey
$c^{h}c^{h}$,$c^{h}c^{a}$	Himalayan albino
$c^{a}c^{a}$	Albino

2. Multiple Alleles in *Drosophila*

A large number of multiple alleles are known in *Drosophila.* One of them is the series of wing abnormality ranging in size from no wings to normal wings. The normal wings are dependent on the **Vg** allele. The extreme expression with no wings (just stumps) is the result of one allele **vg** in homozygous condition.

S.No.	Phenotype	Genotype
1.	Normal wings	Vg^{+} or
2.	Nicked wings	Vg^{ni}
3.	Notched wings	Vg^{no}
4.	Strap wings	vg^{st}
5.	Vestigial wings	Vg

A small stump is produced by gene vg, one of the members of the series. A narrow strap-like wing is produced by $\mathbf{vg^{st}}$, notched wings are associated with the allele $\mathbf{vg^{no}}$ and nicked with $\mathbf{vg^{ni}}$. These phenotypes are produced from the homozygous genotypes, their heterozygous combinations result in intermediate phenotypes, but wild type (normal gene) is dominant over all other members of the series.

3. Multiple Alleles in Mice

Coat colours of mice and cat also offer examples of multiple allelism. These are also found in various insects and plants. Blood groups in cattle are determined by as many as 80 alleles.

Two series of multiple alleles are found associated with the determination of coat (skin) colour in mice. Series number 1 is at the albino locus and comprises of four genes. The normal allele in this series is for **gray coat colour** and is represented by (+) in the figure. The other genes in the series are mutant genes—**albino** (a), **medium light grey** (a^m) and extreme light (a^e). All the three mutant genes are recessive to the normal allele.Various homozygous genotypes and phenotypes, due to two different multiple allelic series for skin colour in mice are as follows :

Albino series		Black series	
Genotype	**Phenotype**	**Genotype**	**Phenotype**
+/+	grey	+/+	normal grey
a/a	albino	b/b	black
a^m/a^m	medium light	Y/Y	yellow-dies (lethal)
a^e/a^e	extreme light	G^L/G^L	grey with light belly
a^e/a^e		b^L/b^L	black with light belly

The second series of the alleles is at the black locus and is on a different chromosome. Here the normal allele is one of the numerous genes necessary for **gray coat colour** (+). The mutant genes of the black series are : **black (b), yellow (y), gray body with light belly (G^L), and black body with light belly (b^L).** The light belly effect of both G^L and b^L is recessive to gray and so is to the black **(b)** but the yellow of the series is dominant to all.

4. Multiple Alleles in Man

ABO blood groups are controlled by a series of three multiple alleles. Different combinations of these alleles produce four blood types **A, B, AB** and **O.**

5. Multiple Alleles in Plants associated with Self Incompatibility

Self-sterility in tobacco, *Nicotiana,* is controlled by four genes designated as S_1, S_2, S_3 and S_4. Several other S-alleles were found, 37 members of multiple allelic series cause self-compatibility in evening promorse, *Oenothera,* and about 41 are found in red colour. Genotypes of progenies obtained due to crosses between various self-incompatible types are given as follows :.

Female	Male		
	S_1S_2	S_1S_3	S_2S_3
S_1S_2	—	S_1S_3, S_2S_3	S_1S_3, S_2S_3
S_1S_3	S_1S_2, S_2S_3	—	S_1S_2, S_2S_3
S_2S_3	S_1S_2, S_1S_3	S_1S_2, S_1S_3	—

Importance of Multiple Alleles

The occurrence of multiple alleles has been very wide and the knowledge of their place in general scheme of heredity has contributed vary largely to the analysis of genetical problems. **H.T. Morgan** has summarised the matter by saying, "Probably the most important bearing on the nature of gene is that derived from multiple alleles."

Q.5. Write an essay on blood groups in man.

Ans.

Blood Groups in Man

The description of various blood groups found in man are discussed as follows :

ABO Blood Groups

Blood from an individual cannot always be safely mixed with that of another individual without leading to some disastrous result. This fact became evident with the introduction of blood transfusions which sometimes cured and sometimes killed the patients. This is due to the fact that blood proteins of one individual differ from those of others. Extensive studies have shown that there are two types of substances in the blood known as **antigens** or **agglutinogens** and **antibodies** or **agglutinins.** These are designated by the English alphabets **A** and **B.** The antigens are located on R.B.Cs and antibodies occur in plasma. Normally, if a person has an antigen in his R.B.Cs his plasma has natural antibodies against the other antigen. A person having antigen-**A** in R.B.Cs has antibody **b** in his plasma. The agglutination occurs on account of the reaction between antigen and antibody. Recent chemical investigations have shown that antigens **A** and **B** are not proteins but mucopolysaccharides (sugars + amino acids) having a molecular weight of 300,000.

Depending upon the presence and absence of antigens and antibodies, four blood groups have been differentiated. These are designated as **A, B, AB** and **O** blood groups. The persons belonging to blood group **A** have antigen-**A** in their red blood cells and antibody **b** (anti-**b**) in the plasma. Persons of blood group **B** have the antigen-**B** in their red blood cells and antibody **a** (anti-**a**) in the plasma. Persons of group **AB** have antigen-**A** and **B** in the red blood corpuscles but no antibodies in the plasma. Persons of group **O** have no antigen in red blood cells but have both **a** and **b** or antibodies present in the plasma. The following table represents the antigens and antibodies present in persons of different blood groups :

Blood groups, their Antigens and Antibodies in Man

Blood groups	Antigen present in the RBC	Antibody present in plasma
A	A	anti-B or **b** or β
B	B	anti-A or **a** or α
AB	A and B	No antibodies
O	No Antigen	Both anti-A and anti-B or **a** and **b** or α and β

It will be noticed that blood corpuscles from individuals of blood group **O,** since they lack antigen, are not clumped by the serum of any blood group, so that person of group **O** can give blood to all but can take only from his own group. Hence, these are called **universal donors.** The serum from individuals of blood group **AB** does not cause clumping of corpuscles of any group. Hence, they can take blood from the persons of all the blood groups. So they are known as **universal recipients,** but can give blood only to the persons of their own group. The terms universal donor and universal recipient are no longer applicable after the discovery of **Rh** factor. Groups **A** and **B** can take and give their blood to their own groups, but in addition they can give blood to persons of group **AB** and receive blood from group **O.**

Possible Effects of Transfusion of Blood

Donor's blood groups	Reccipients Agglutinates the blood of				Can be given to	Can receive blood from	Remark
	O	A	B	AB			
O	—	—	—	—	O, A, B, AB	O	Universal donor
A	+	—	+	—	A & AB	O, A	—
B	+	+	—	—	B & AB	O, B	—
AB	+	+	+	—	AB	O, A, B, AB	Universal recipient

Inheritance of ABO Blood Groups

Berntstein (1925) discovered that the inheritance of different blood groups in man is determined by a number of multiple allele series.

For convenience group **O** is regarded as normal and allele **A** and **B** are said to represent two dominant mutations which have occured on the same locus. These represent codominant alleles. If normal gene is represented by +, the three genes will be +, **A** and **B.** Since + is recessive, individuals of group **O** will have a genotype +/+; gene **A** is dominant, individuals of

Blood group of parents	Genotypes and inheritance of genes	Blood group off springs
A and B	L^A L^A / L^B L^AL^B L^AL^B / L^B L^AL^B L^AL^B	AB
A and B	L^A I^O / L^B L^AL^B L^AI^O / I^O L^AI^O I^OI^O	AB, B, A and O
A and B	L^A I^O / L^B L^AL^B L^AL^B / L^B L^AL^B I^OL^B	AB and B
A and B	L^A L^A / L^B L^AL^B L^AL^O / I^O L^AL^O L^AL^O	AB and A
A and O	L^A L^A / I^O L^AI^O L^AI^O / I^O L^AI^O L^AI^O	A
A and O	L^A L^A / I^O L^AI^O I^OI^O / I^O L^AI^O I^OI^O	A and O

Fig. 1 : Diagram showing inheritance of ABO blood groups

blood groups **A** might have a genotype either **+/A** or **A/A** and similarly individuals of group B might have +B or B/B and individuals of group **AB** will have a genetic composition A/B.

Another method of representing these genes and the genotypes is :

L^A, L^B and I :

Blood group	**A**	**$-L^AL^A$**
Blood group	**B**	**$-L_BL_B$**
Blood group	**AB**	**$-L_AL_B$**
Blood **group**	**O**	**-II**

These blood groups are, therefore, inherited in the simple Mendelian fashion. Offsprings of all the four types of blood groups can be produced from a cross between heterozygous male and female for blood groups A and B.

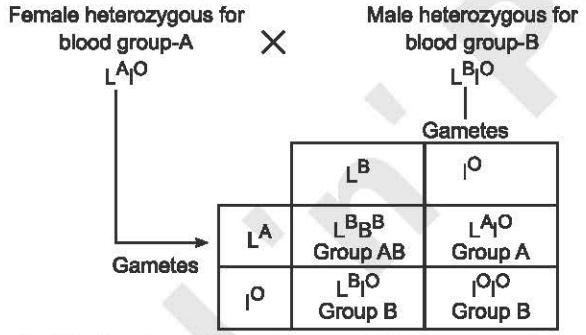


Fig. 2 : Mechanism of inheritance of blood groups in man.

Q.6. Write an essay on cytoplasmic inheritance.

Ans.

Introduction to Cytoplasmic Inheritance

The inheritance of most of the characters of an individual is governed by nuclear genes. But in some cases, the inheritance is governed by cytoplasmic factors or genes. When the transmission of characters from parents to offspring is governed by cytoplasmic genes; it is known as cytoplasmic inheritance or extra nuclear inheritance or extra chromosomal inheritance or non-mendelian inheritance or organellar inheritance.

The first case of cytoplasmic inheritance was reported by Conens in 1909 in four 'o' clock (*Mirabilis jalapa*) for leaf colour. Later on, cytoplasmic inheritance was reported by various workers in various organisms.

Features of Cytoplasmic Inheritance

Cytoplasmic inheritance differs from Mendelian inheritance in several aspects and exhibits some features.

(i) Reciprocal Differences : Characters which are governed by cytoplasmic inheritance invariably exhibit marked differences in reciprocal crosses in F_1, whereas in case of nuclear inheritance such differences are not observed except in case of sex linked genes.

(ii) Maternal Effects : In case of cytoplasmic inheritance, distinct maternal effects are observed. This is mainly due to more contribution of cytoplasm to the zygote by female parent than male parent. Generally ovum contributes more cytoplasm to the zygote than sperm.

(iii) Mappability : Nuclear genes can be easily mapped on chromosomes, but it is very difficult to map cytoplasmic genes or prepare linkage map for such genes. Now chloroplast genes in Chlamydomonas and maize, and mitochondrial genes in human and yeast have been mapped.

(iv) Non-Mendelian Segregation : The mendelian inheritance exhibits typical segregation pattern. Such typical segregation is not observed in case of cytoplasmic inheritance. The segregation when occurs, is different from mendelian segregation.

(v) Somatic Segregation : Characters which are governed by cytoplasmic genes usually exhibit segregation in somatic tissues such as leaf variegation. Such segregation is very rare for nuclear genes.

Criteria for Cytoplasmic Inheritance

The cases of cytoplasmic inheritance are found to exhibit maternal influence. The reason is very simple. Very little cytoplasm is contained in the sperm cell of an animal. Most of the cytoplasm is contributed to the zygote by the ovum or egg. Hence, if there are hereditary units in the cytoplasm, these will be transmitted to the offspring through the egg. The offspring, therefore, will exhibit maternal influence. This could be explained further by citing an example :

Suppose the cytoplasm carries units for black pigment and race A is dark and race B albino. The two races possess corresponding units for pigment in their cytoplasm. If a female from race A (black) is mated to a male from race B, all the offspring will get units for black pigment through the cytoplasm of mother and would be pigmented. On the other hand, if reciprocal cross is made offspring of race B female (albino) and race A male (black), get the albino unit from the egg cytoplasm and would be albino. Had the genes for black and albino present in the nucleus, there would have been no difference in the results of reciprocal crosses.

Some Examples of Cytoplasmic Inheritance in Animals

1. **Inheritance of Kappa Particles in *Paramecium* :** **Sonneborn** and his associates have described the transmission of some cytoplasmic particles known as **kappa particles** and their relation to nuclear genes in *Paramecium aurelia.* Individuals of a particular race of *Paramecium aurelia* called **'killer strain'** destroy other races of paramecia by secreting some toxic substance into the water in which they live. This substance is known as **paramecin,** and such individuals are called **killers** and the individuals destroyed by paramecin are known as **'sensitive'.** The paramecia of killer strain contain large number of particles in their cytoplasm. These particles are known as 'kappa particles' and are composed of deoxyribonucleic acid. These are self-duplicating and arise *de-novo.* These can mutate.

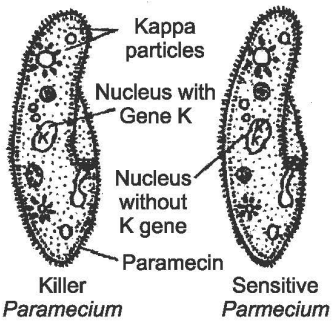


Fig. 1 : Diagram showing presence of gene K and kappa particles in Killer strain and their absence in sensitive strain of *Paramecium*.

Moreover these are present in the cytoplasm rather than in the nucleus, but they control a hereditary trait, the killer character. Like viruses, these can duplicate themselves independent of the nuclear division.

Although, the kappa particles are cytoplasmic particles and transmitted strictly through the cytoplasm, their maintenance and production of paramecin are controlled by a dominant gene **K** present in the nucleus of killer strain. The sensitive strain possesses its recessive allele *k*.

By following an appropriate procedure, it is possible to cross the individuals of killer and sensitive races. Observations on the descedants of such crosses have shown that individuals of killer shown that individuals of killer clones carry a dominant gene **K,** whereas those of sensitive clones posses **k.** When killer **KK** conjugates with sensitive **kk,** the exconjugants are **Kk.** These according to their genotype should be killer. In ordinary case of conjugation where only the nuclear material is exchanged and there is no exchange of cytoplasmic material, it has been found that each exconjugant gives rise to the organisms of its type, *i.e.,* sensitive produces sensitives and killer produces killers. This suggests that killer character is not governed by gene.

In the second case, when the conjugation is prolonged and cytoplasmic bridge between the conjugants is larger than usual permitting exchange of cytoplasm, all the offsprings are killers and their cytoplasm contains visible kappa particles. This means that the kappa particles from the killer strain are transferred to the sensitive strain through the exchange of cytoplasm so that sensitive individuals have also been converted into the killers. Therefore, **Kk** individuals may be either a killer or sensitive depending upon whether it has received kappa particles or not.

It is possible to convert killer clones into sensitive by making them undergo vary rapid fissions. If paramecia of killer strain are overfed they multiply very fast and kappa particles fail to keep pace with the division of the body of the animal, with the result that kappa particles are finally lost entirely. The gene **K** present in their genotype is unable to initiate the production of kappa particles, but can simply maintain them if these are already present or are introduced. Vice-versa gene **k** is unable to maintain the kappa particles. If kappa particles are introduced into a sensitive paramecia having gene **kk,** these are lost without producing any change.

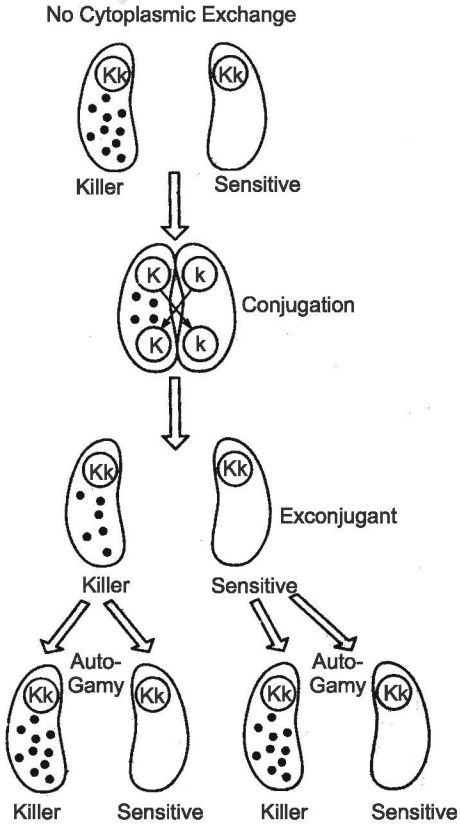


Fig. 2 : Inheritance of killer and sensitive characters in *Paramecium* during conjugation.

2. Inheritance of sigma particles in *Drosophila* : *Drosophila* flies are sensitive to CO_2 and they can be immobilized by exposing them to CO_2. **L. Heritier** and **Teisser** discovered that some strains of *Drosophila* are much more sensitive to CO_2 than the normal and are affected by much smaller concentrations. If these are exposed to higher concentration of CO_2 these soon become anesthetised and finally killed. Reciprocal crosses between sensitive and normal *Drosophila* produce different results. When females of the sensitive race are crossed to normal males, all the offsprings are sensitive. Of them, the females again transmit sensitivity during successive generations when crossed with nonsensitive males. On the contrary, when males of the sensitive stock are crossed with normal females only a few of the offsprings are sensitive. When each of the chromosomes of the sensitive females is replaced by homologous chromosomes from a normal resistant stock, the sensitive character remains unchanged. This indicates that the sensitive character in *Drosophila* is transmitted outside the chromosomes and can possibly be associated with the cytoplasm. These self-perpetuating units present in the cytoplasm and most probably associated with CO_2 sensitivity have been called **sigma,** the heat-labile particles.

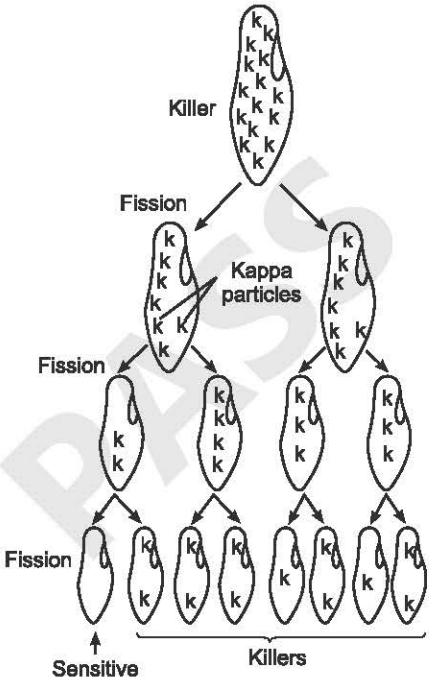


Fig. 3 : Diagram illustrating the formation of sensitive strain of *Paramecium* from a killer strin (K-represents kappa particles).

Q.7. What is the degree of gene expression. Discuss the differences between penetrance and expressivity of a trait with examples.

Ans.

Degree of Gene Expression

Genes express them by producing visible phenotype. Some genotypes express themselves faithfully in all the organisms in which these are present. Generally, the structural genes express themselves in all the individuals that carry them. All factors in Mendel's experiments expressed them completely producing expected phenotypes. Some genes do not produce the excepted phenotype in all cases. The expression of such genes is variable.

The measure of gene expression at population level and at individual level is denoted under following two heads :

1. Gene Penetrance

Gene pentrance is the degree of expression of gene at population level. It is the frequency with which a genotype is expressed in phenotype. It is the proportion of individuals of a given genotype that manifest a phenotype. Say for example, out of eight individuals with a particular genotype, only five produce the expected phenotype, the penetrance of this particular gene pair is = 5/8 = 0.625 or 62.5 per cent.

Penetrance may be of the following two types :

(i) Complete Penetrance : In complete penetrance a dominant gene expresses itself in all the organisms in which it is present producing the expected phenotype while the recessive gene produces the associated phenotype in all those organisms in which it is present in homozygous condition.

Examples of Complete Penetrance : In pea plants, the red and white flower colour, tall and dwarf characters, the seed colour and shape all exhibit complete penetrance.

(a) In *Drosophila*, the recessive gene for wing character (vestigial wings) has complete penetrance.

(b) In *Guinea pigs* the gene for coat colour exhibits complete penetrance.

(c) Human blood group genes also have cent per cent penetrance.

(ii) Incomplete Penetrance : Some genes whether in homozygous or heterozygous condition fail to produce the phenotypic expression in all the organisms in which these are present. It means some individuals fail to show a particular trait, even though the controlling genes for that trait are present. This is called **incomplete penetrance.** The incomplete penetrance of gene may cause a trait to skip a generation but reappear in the second generation. In these cases, the degree of penetrance is usually expressed as the percentage of individuals that actually express the **phenotype,** determined by a particular genotype.

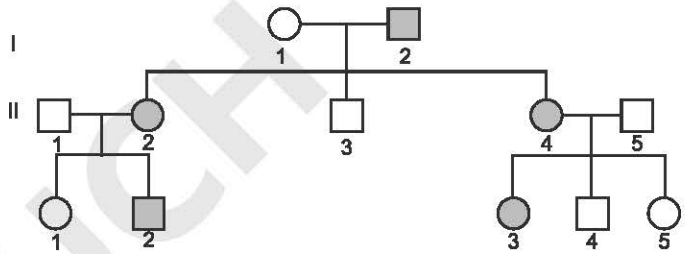


Fig. : A pedigree illustrating pattern of inheritance of an incompletely penetrant dominant gene.

Examples of Incomplete Penetrance :

(a) In man, genes for **polydactyly, blue sclerotic** and **diabetes mellitus** all exhibit incomplete penetrance. The gene for blue sclerotic (produces sclerotic of blue colour) has about 90% penetrance, *i.e.,* it develops in about 90% persons, possessing it. Similarly, gene for polydactyly has 70% penetrance. In this pedigree the unaffected individual II-4 is a daughter as well as mother of an affected individual.

(b) A gene in Lima beans causing partial chlorophyll deficiency in cotylendonous leaves has 10 per cent penetrance.

Effect of Environment on Penetrance : The present concept about inheritance explains that the phenotype of an organism is not exclusively dependent on the genotype, but it is the result of interaction between the genotype and environment and particular character is the outcome of interaction between gene in question, the genotype and an organism and the environment.

2. Gene Expressivity

Expressivity for gene is the degree or range of expression of a genotype in those individuals that are supposed to have it. The expressivity differs from penetrance because expressivity denotes the level of expression of phenotype, whereas penetrance is whether the phenotype has appeared or not. The genes may have **uniform** or **variable expresssivity.**

(i) Uniform Expressivity : A gene that expresses itself uniformly in all the individuals that carry it, has **uniform expressivity.** In Mendel's experiments, all the seven pairs of factors had uniform expressivity.

(ii) Variable Expressivity : The genes which do not have similar expression in all the individuals possessing them, have **variable expressivity.** Its effect may vary from mild to severe. The expressivity of a gene may be influenced by the environment or by the genetic makeup of the individual. The presence of complementary, supplementary, modifier or suppressor genes in the genotype of an individual change the expression of the gene in question.

Examples : The gene producing chlorophyll deficiency in Lima bean (*Phaseolus lunatus*) exhibits variable expressivity in addition to incomplete penetrance. In some seedlings, the complete cotyledonary leaves are without chlorophyll, in some only the leaf tip lacks the chlorophyll, while in others the margins of the leaves have no chlorophyll.

In man gene for polydactyly have variable expressivity and incomplete penetrance. Polydactyly is due to the dominant gene P, the normal condition with five digits in each limb is due to the genotype pp. It has been noticed that some heterozygous (Pp) persons are not polydactylous. Moreover, polydactylous persons mostly have an extra digit in only one of the hands or feet.

Q.8. In Wheat Kernel colour is controlled by three genes (R_1, R_2, R_3). If two parents differ for all the three genes (R_1 R_1R_2 R_2 R_3 R_3 and $r_1r_1r_2r_2r_3r_3$). What proportion of F_2 progeny will have the genotypes like that of one of the parents?

Ans.

Kernel Colour in Wheat

Kernel colour in wheat is a qualitative character and was studied by **H. Nilsson-Ehle** for the first time in 1908. It was argued that if one gene was considered or in other words, if the two parents differed due to one gene only a 3 : 1 ratio for red and white kernels was obtained in F_2 generation. However, out of three red, one was as red as one of the parents and two were lighter and were comparable to F_1 individuals. This indicated that the dominant alleles had a cumulative effect. If '*R*' stands for red colour and '*r*' for white, the two parents could be designated as *RR* and *rr*, the F_1 could be designated as *Rr* and F_2 would be obtained in 1*RR* : 2*Rr* :1*rr* ratio. In these three classes, *RR* should be red, *Rr* should be intermediate in colour and *rr* should be white.

In case there were two genes ($R_1 R_2$) involved a 15 : 1 ratio (15 coloured : 1 white) would be obtained. If different shades are taken into account 1 : 4 : 6 : 4 : 1 ratio will be obtained, provided R_1 contribute equally to the colour. However, it is known now that there are three genes involved in kernel colour in wheat. Obviously if the two parents differ for all the three genes in F_2 63 : 1 or 1 : 6 : 15.

Genotypic and Phenotypic Ratios in F_2 obtained due to a cross $R_1R_1R_2R_2 \times r_1r_1r_2r_2$.

Genotype	Genotypic ratio	Phenotype	Phenotypic ratio
$R_1R_1R_2R_2$	1 }	red (like) P_1	1
$R_1R_1R_2r_2$	2	darker than F_1	4
$R_1r_1R_2R_2$	2	and lighter than P_1	
$R_1r_1R_2r_2$	4	like F_1	coloured (15/16)
$R_1R_1r_2r_2$	1	(intermediate between	6
$r_1r_1R_2R_2$	1	P_1 and P_2)	
$R_1r_1r_2r_2$	2	lighter than F_1	4
$r_1r_1R_2r_2$	2		
$r_1r_1r_2r_2$	1}	colourless	1} colourless (1/16)

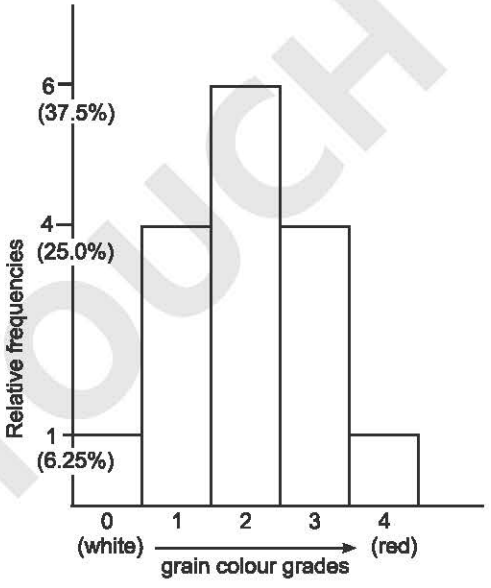


Fig. 1 : Relative frequencies (theoretical) of different grain colours in wheat in F_2 population derived from two strains differing in two gene pairs.

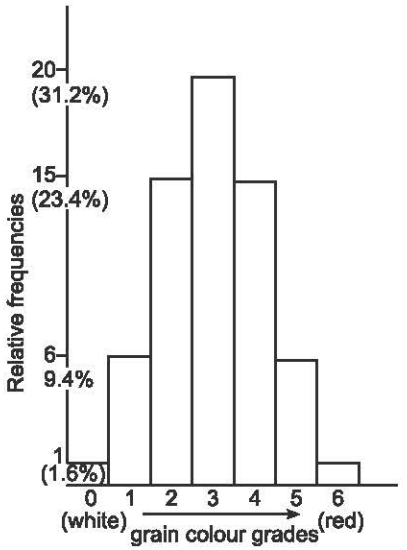


Fig. 2 : Relative frequencies (theoretical) of different grain colours in wheat in F_2 population derived from two strains differing in three gene pairs.

Expansion of binomial $(½ + ½)^n$ using different values of n with the help of Pascal's triangle.

n	Expansion of binomial $(½ + ½)^n$
1	1 : 1
2	1 : 2 : 1
3	1 : 3 : 3 : 1
4	1 : 4 : 6 : 4 : 1
5	1 : 5 : 10 : 10 : 5 : 1
6	1 : 6 : 15 : 20 : 15 : 6 : 1
7	1 : 7 : 21 : 35 : 35 : 21 : 7 : 1
8	1 : 8 : 28 : 56 : 70 : 56 : 28 : 8 : 1

20 : 15 : 6 : 1 ratio will be obtained (fig. 2). These ratios like 1 : 4 : 6 : 4 : 1 or 1 : 6 : 15 : 20 : 15 : 6 : 1 can be easily obtained by the expansion of binomial equation, $(1/2 + 1/2)^n$ where n is the number of alleles (no. of alleles will be double the number of genes, so that for 2 genes $n = 4$, and for 3 genes, $n = 6$). This expansion can be obtained by the use of **Pascal's triangle** given in table 2 (consult an elementary book on Statistics for Binomical Distribution).

By the study of kernel colour in wheat, **Nilsson-Ehle** reached the conclusion that the effect of each dominant allele was cumulative and therefore, he forwarded his multiple factor hypothesis. The hypothesis states that for a given quantitative trait there should be several genes, which were independent in their segregation, but had cumulative effect on phenotype. ❑

UNIT-VII

Human Chromosomes and Patterns of Inheritance

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Describe technique that are used in the study of chromosomes.

Ans. Techniques used include karyotyping, analysis of G-band chromosomes, other cytogenetic bendeting techniques, as well as molecular cytogenetics such as fluorescent in site hybridization (FISH) and comparative genomic hybridization (CGH).

Q.2. Write the characteristics of aneuploids involving sex chromosomes in humans.

Ans. Sex chromosome aneuploidies (individuals with abnormal numbers of sex chromosomes) are found not infrequently in general population and have characteristics deficits of cognitive ability : individuals with an extra-X-chromosome (XXY or Klinefelter's syndrome and XXX syndrome) have delays in the acquisition of language, as also do individuals with XYY syndrome.

Q.3. What do you understand by monozygotic and dizygotic twins?

Ans. To form identical or monozygotic twins, one fertilised egg (ovum) splits and develops into two babies with exactly the same genetic information. To form fraternal or dizygotic twins, two eggs (ova) are fertilised by two sperm and produce two genetically unique children.

Q.4. What do you understand by chromosome mapping in humans?

Ans. In humans, genetic mapping also called linkage mapping can offer firm evidence that a disease transmitted from parent to child is linked to one or more genes. Mapping also provides clues about which chromosome contains the gene and precisely where the gene lies on that chromosome.

Q.5. Who discovered DNA fingerprinting technique? Write its uses.

Ans. The techniques of DNA fingerprinting was developed for the first time in 1985 by **Alec Jeffreys** and his colleagues at Leicester university in UK.

Uses : This used as evidence in courts, to identify bodies, track down blood relatives and to look for cures for disease.

Q.6. Define pedigree analysis.

Ans. A chart which displays the affected members of a family by genetic diseases in the form of a family tree is called pedigree chart. The study of such a chart to detect genetic diseases in a family is called pedigree analysis. The diagram gives some common symbols in pedigree analysis.

Q.7. What is familial aggregator?

Ans. The first step in the identification of hereditary diseases is frequently a familial aggregation study. Such a study seeks to determine whether having relatives with disease increases one's risk of that disease. Familial aggregation refers to this clustering of diseases within families.

Q.8. Why a X-linked recessive trait common in males and nos in females.

Ans. Recessive X-linked traits appear more oftain in males than females because, if a male receiver a 'bad' allele from his mother, he has no chance of getting a 'good' allele from his father (who provides a Y) to hide the bad one.

Q.9. Define proband.

Ans. A proband is an individual being studied or reported on. A proband is usually the first affected individual in a family who brings a genetic disorder to the attention of the medical community.

SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Write short note on monozygotic twins.

Ans.

Identical or Monozygotic Twins (MZ Twins)

The **monozygotic twins** arise from a single zygote formed by fertilization of a single egg with a single sperm. Such twins are members of a clone and have the identical genotype. These arise by the separation of two or more blastomeres derived by mitotic divisions of original zygote.

Monozygotic twins (MZ) are **concordant type** *i.e.,* have identical characters. These are of the same sex. Identical twins are generally enclosed in one membrane and are attached to one placenta only.

Origin of Monozygotic Twins : It can be discussed as follows :

(i) Monozygotic twins may arise by the separation of two blastomeres of the first mitotic cleavage. The separated blastomeres develop further, implant as two DZ blastocysts in different types of the uterus and develop separate membranes and separate placenta. Their placentae are described as **separate dichorionic placentae.**

(ii) Monozygotic twins may develop within the blastocyst either in inner-cell mas or in the embryo derived from it. These usually have a single chorion but separte amniotic sacs and amnions. Their placenta is called **monochorionic diamniotic placenta.**

But these may have separate chorion and separate placenta also. Such placentae are called **dichorionic diamniotic.** These are more common.

(iii) **Conjoined** or **Siamese Twins** arise by incomplete separation of embryo at about 15 days or more after zygote formation.

Q.2. Describe aneuploidy in human.

Ans.

Aneuploidy in Human

Trisomics ($2n = 47$) and monosomics ($2n = 45$) are known in human beings. Trisomy or monosomy may involve a sex chromosome or an autosome.

Aneuploid chromosome numbers involving X-chromosome and the resulting phenotypes are listed in table.

Sex chromosomes	Sex	Phenotypes
XO (monosomic)	female	Turner's syndrome
XX (disomic)	female	normal
XXX (trisomic)	female	superfemale
XXXX (tetrasomic)	female	(mental abnormalities)
XXXXX (pentasomic)	female	(mental abnormalities)
XY (disomic)	male	normal
XYY (trisomic)	male	normal
XXY (trisomic)	male	
XXYY (tetrasomic)	male	Klinefelter's syndrome
XXXY (tetrasomic)	male	
XXXXY (pentasomic)	male	extreme Klinefelter's

(a) Turner's syndrome : Turner's syndromes are characterized by monosomy of XO type. These are immature females (sterile) with webbed neck.

(b) Klinefelter's syndrome : Klinefelter's syndromes were characterized by trisomy (XXY). These are male individuals, who are phenotypically fairly normal but have a very low sperm count and, are therefore sterile. Chromosome constitutions of other Klinefelter's syndromes are as follows :

Q.3. Describe the aminocentesis technique.

Ans.

Aminocentesis

In the past twenty years, a number of techniques have been evolved for the diagnosis of some of the genetic abnormalities during the foetal or intrauterine life of the foetus. This has increased the possibility of preventing or alleviating the effects of a genetic disorder if it can be detected as early as possible. **Amniocentesis** is one of the most widely used technique for this purpose.

In **amniocentesis,** a sample of amniotic fluid is obtained from the amniotic cavity around the foetus by inserting a needle through the lower abdomen of pregnant woman and through the wall of uterus. The amniotic fluid is now centrifuged. The cells form a sediment and removed.

(i) Some of these cells are transferred to the slides, fixed and stained. These are then studied for the study of karyotype, determination of sex, sex abnormalities and to identify trisomy, monosomy or other chromosome abnormalities.

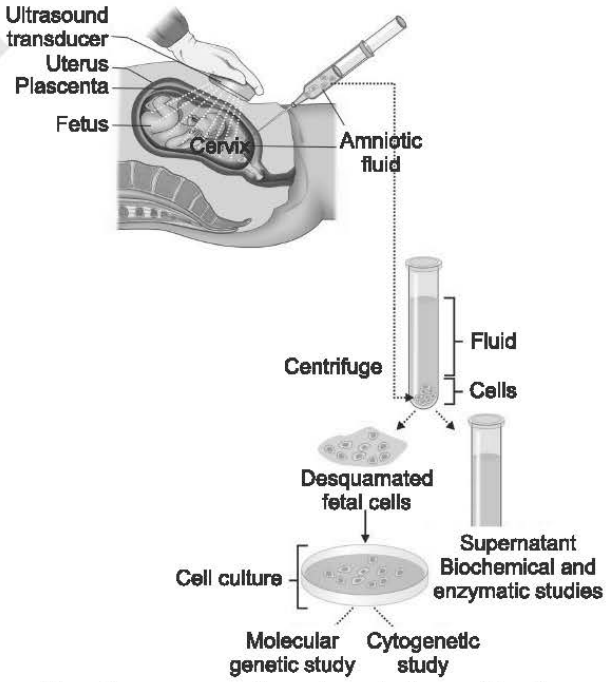


Fig. : The process of amniocentesis used for the prenatal diagnosis of genetic diseases.

(ii) Cells may be cultured in culture medium for about two weeks. These can be analysed biochemically for the presence of about 40 metabolic disorders caused due to absence of certain enzymes.

(iii) These may be used for detecting severe sex-linked hereditary diseases like haemophilia.

(iv) The amniotic fluid is used for determining the concentration of certain proteins which are also found associated with genetic defects in the organisms. For example, increase in α-fetoprotein concentration in amniotic fluid is associated with defects in the development of neural tube such as **spina bifida.**

Q.4. Write a short note on use of DNA fingerprinting in forensic science.

Ans.

DNA Fingerprinting

The technique of DNA fingerprinting was developed for the first time (1985, 86) by **Alec Jeffreys** and his colleagues at Leicester University in U.K. In this field, establishment of the identity of a person with the help of blood stains, semen (sperms) stains or hair roots will be possible with almost absolute certainty. In this technique, DNA will be isolated from blood stains, semen stains or hair roots and will be analysed for polymorphism, which has a very stable inheritance. The technique of DNA fingerprinting reveals such a great polymorphism that the possibility of two persons having same pattern of DNA fingerprints is very remote. The above technique has already allowed the identification of rapists in rape cases, and of mother and or father in case of doubtful parentage.

In India, DNA fingerprinting tests are carried out at the Centre for Cell and Molecular Biology (CCMB), Hyderabad. For this purpose, a test with the **Bkm** probe (banded krait minor satellite (DNA) earlier used for identification of sex chromosomes has been found to cost one-tenth of the cost of tests used in Europe and U.S.A. Paternity dispute cases are much more common in India and most of them are referred to **CCMB** for DNA evidence. The first such test on DNA fingerprinting was used in June, 1989 to settle a drawn-out paternity case in Madras. Ever since then, (Late) **Dr. Lalji Singh** (Former Director, CCMB, Hyderabad and Vice Chancellor, Banaras Hindu University), with the help of his coworkers, conducted DNA fingerprinting to settle more than 50 disputes.

Q.5. Write about the Lyon hypothesis (Barr body).

Ans.

Lyon's Hypothesis

In human beings, the sex can be identified by observing the nucleus of their resting cells (the **interphase nucleus**). In the interphase nucleus of cells in females a darkly stained chromatin mass is observed on one side. This is known as **sex-chromatin** or **Barr body** (after the name is its discoverer, **Murray Barr**, 1940).

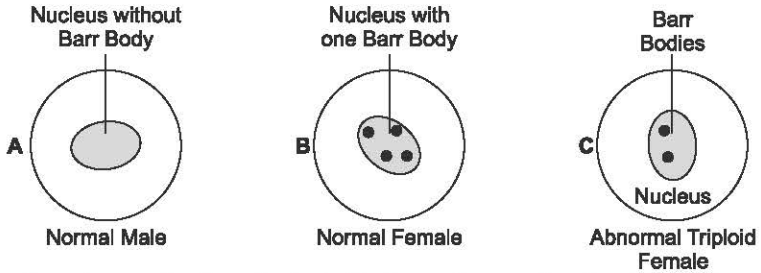


Fig. : Recognition of sex in man by Barr body A. Nucleus of normal male with no Barr Body; B. Nucleus of normal diploid female with one Barr body; C. Nucleus of triploid female with two Barr bodies.

It has been explained by **Mary Lyon** that in early embryogenesis one of the two X-chromosomes becomes genetically inert and heterocycnotic and forms the **Barr body.** This is known as **Lyon's hypothesis.** It means there is one Barr body in female and none in male or the number of Barr bodies is always one less the number of X-chromosomes. In cases of sex anomalies an abnormal male has one Barr body and an abnormal female **(XO)** does not show Barr body.

In triploid males and females **(XXX)** or tetraploid males **(XXXY)** there are two or three Barr bodies :

S. No.	Particulars	Sex phenotype	Sex-chromosome	Number of Barr bodies
1.	Normal Male	Male	XY	0
2.	Normal Female	Female	XX	1
3.	Turner Syndrome	Female	XO	0
4.	Klinefelter Syndrome	Male	XXY	1
5.	Triple X-Syndrome	Female	XXX	2
6.	Triple X-Y Syndrome	Male	XXXY	2
7.	Tetra X-Syndrome	Female	XXXX	3
8.	Tetra X-Y syndrome	Male	XXXXY	3

Barr Body Technique : The cells from epithelial lining of buccal cavity, vagina or urethra are scrapped and smeared on a slide. These are stained with any of the basic dyes (methylene blue, Feulgen stain or haematoxylene) and are observed under high magnification of microscope. The Barr body is seen attached to the nuclear membrane.

Q.6. Describe detail of structural chromosomal aberrations in man.

Ans. Structural Chromosomal Aberrations in Man

Structural aberrations of all kinds like translocation, deletion, duplication, ring chromosomes, inversion and isochromosomes have been observed in man and are found to be associated with abortion and various congential diseases. Some of them are as follows :

1. **Translocation :** Translocation between chromosome 21 and either chromosome 14 or 15 has been observed in man. This also results in Down's syndrome or mongoloids. Such patients have 46 chromosomes with two normal 21 chromosomes and one unpaired large chromosome which is formed by the fusion of the long arms of

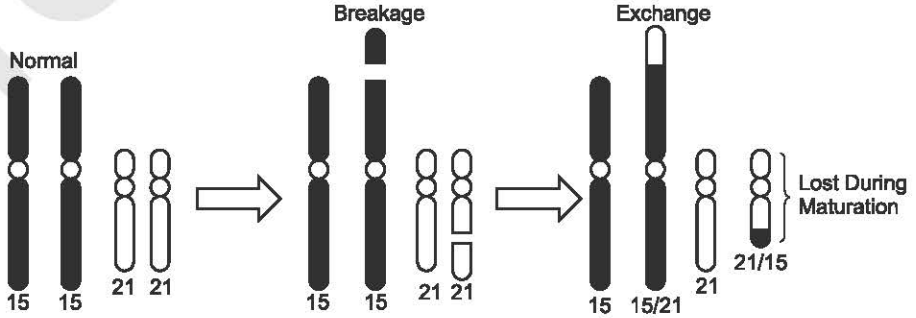


Fig. 1 : Traslocation betwen 21/15 chromosomes produces single large chromosome to produce Down's syndrome.

chromosomes 21 and 15. Therefore, in translocation or Down's syndrome the genetic material of chromosome 21 is present in triple dose and such cases are phenotypically indistinguishable from those with 21-trisomy.

The translocation mongolism runs in families and in such families the risk of a mongoloid child is one in three. Usually, the mother is a carrier of a balanced 21/15 translocation and phenotypically she is normal. But this translocation event is not confined to the mother only. Cases are known in which father has been the carrier.

2. **Deletion of Genes :** The effect of loss of a portion of a particular chromosome depends on the particular genes lost. A deletion involving loss of large number of genes is incompatible with life.
   (a) **Lejeune** (1963) has described the effect of loss of a portion of no. 5 chromosome. The affected infant has a rounded, moon-like face and utter feeble, plaintive cries similar to the mewing of cat (cat-cary syndrome or *cridu-chat* syndrome). These children remain mentally and physically retarded.
   (b) Deletion of a part of 21 chromosome produces **leukemia,** a cancerous malignancy arising in blood forming tissues. This was observed by **Peter Nowell** and D.A. **Hongrford** (1960) in Philadelphia and is commonly known as **Philadelphia chromosome syndrome.**
   (c) Deletion of some part of one **X**-chromosome has been found to produce some sex anomalies. The deletion of short arm produces clinical symptoms similar to **XO** Turner's syndrome.
   (d) Isochromosome of the long arm of **X**-chromosome also produces Turner's syndrome because the short arms are lost during isochromosome formation in 2nd meiotic division.

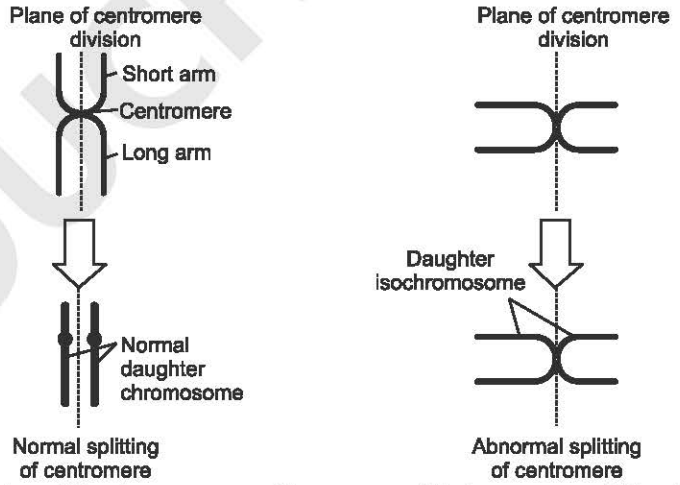


Fig. 2 : Formation of isochromosome of long arms of X-chromosome (After D.G. Harnden)

Q.7. Define dominant disorders.

Ans.

Dominant Disorders

The family pedigree of dominant inheritance is given in figure. It shows that the affected person has atleast one affected parent. However, the normal children of affected parents,

when marry normal persons have only normal offspring. This is because the harmful gene is dominant and can expresses itself even in heterozygous condition. The affected heterozygous parent transmits defective dominant gene to 50 per-cent of the children. Some of the dominant defective characters in man are as follows :

1. **Achondroplasia :** It is a form of dwarfs. The affected individuals are small and disproportionate with abnormally short arms and legs. Only 20% of these achondroplastic dwarfs are found to reach adulthood because most of them die in the first year.
2. **Tylosis :** The persons with tylosis have excessively thick skin on their palms and soles.

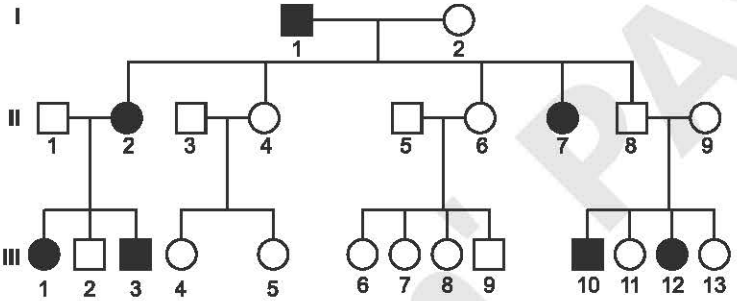


Fig. : Pedigree of dominant inheritance showing that dominant disorder is transmittted only by those individuals who display the disorder.

3. **Anonychia :** This disease is characterised by the absence of some or all nails of the fingers and toes.
4. **Dentinogenesis Imperfecta :** It is a disorder in which crowns of teeth wear down.
5. **Polydactyly :** Presence of extra fingers or toes.
6. **Brachydactyly :** Hands of such persons are short, stocky and thick (both palm and fingers are short).
7. **Marfan's Syndrome :** Have unusually long or tapering fingers with poor musculature and long and thin extermities.
8. **Huntington's Chorea :** The character appears late in the life between the age of 25-55 and is characterised by disorganized muscular movements and progressive mental deterioration.

However, lethal dominant defects fail to be transmitted because the possessor either dies before birth or before reaching the sexual maturity, except for Huntington's chorea which express between the age of 25-55 years.

Q.8. Write short note on Inheritance of autosomal recessive traits.

Ans. Inheritance of Autosomal Recessive Traits

The traits which are inherited as autosomal recessive are equally numerous in both males and females. But the trait expresses itself only when it is in homozygous condition. The heterozygotes for the recessive trait remain unnoticed.

If an individual homozygous for recessive trait marries a homozygous normal person, all the children are unaffected but are heterozygous carriers. The gene is passed unnoticed, unable to express itself in the presence of dominant normal gene.

Among the offspring of two heterozygous parents, one-fourth of the male and female children are expected to express the recessive trait. The traits like albinism, PTC tasting, deaf and mutism, phenylketonuria, alkaptonuria, xeroderma pigmentosum, microcephali, sickle cell anaemia and schizophrenia are all autosomal recessive traits.

Q.9. Describe the muscular dystrophy disease.

Ans.

Muscular Dystrophy

It refers to a group of more than 30 inherited (genetic) disease that cause muscle weakness. These conditions are a type of myopathy, a disease of the skeletal muscles. Over time, muscles shrink and become weaker, affecting your ability to walk and perform daily activities like brushing your teeth. The disease also can affect your heart and lungs.

Some forms of muscular dystrophy are apparent at birth develop during childhood. Some forms develop later during adulthood. Currently, then isn't a cure.

Muscle weakness is the primary symptom of muscular dystrophy. Depending on the type, the disease affects different muscles and parts of the body. Other signs of muscular dystrophy include :

1. Enlarged cell muscles.
2. Difficulty walking or running.
3. Unusual walking gait (like waddling).
4. Trouble swallowing.
5. Heart problems, such as arrhythmia and heart failure (cardiomyopathy).
6. Learning disabilities.
7. Stiff or loose joints.
8. Muscle pain.
9. Curved spine (scoliosis).
10. Breathing problems.

If your healthcare provider suspects muscular dystrophy, you or your child may undergo one or more of these diagnostic tests :

1. An **enzyme and protein blood test** checks for elevated levels of an enzyme called creatine kinase. High levels can indicate muscle damage caused by muscular dystrophy.
2. **Electromyography (EMG)** measures the electrical activity of muscles and nerves.
3. A muscle **biopsy** looks for cell changes in the muscle tissue.
4. Genetic tests identify gene mutations linked to muscular dystrophy.

Q.10. Write a short note inheritance of traits controlled by multiple alleles and multiple factors.

Ans.

Inheritance of Traits Controlled by Multiple Alleles and Multiple Factors

1. **Inheritance of Blood Groups :** Inheritance of blood groups A, B, AB and O is governed by three contrasting genes L^A, L^B and *l*. The *l* gene in homozygous condition produces blood group O and is said to represent the wild type. Two mutations at this locus have

resulted in the appearance of dominant gene L^A and L^B. These represent multiple alleles. An individual can possess only two out of these three genes. An offspring receiving gene L^A from one parent and L^B from other will have blood group AB. An individual belonging to blood group A may have a genotype L^AL^A or L^{AL°. Similarly, individual with blood group B can have a genotype L^BL^B or L^{BL°. The parents who are heterozygous for blood group A and for blood group B are expected to produce all the four types of offspring with blood group A, B, AB and O.

2. **Skin Colour :** The skin colour of a person is the result of an interaction between two pairs of genes.

Q.11. Write short note on Fragile-X-syndrome.

Ans.

Fragile-X-syndrome (FXS)

FXS is a genetic disorder. FXS is caused by changes in a gene that scientists called FMR1 gene when it was first discovered. The FMR1 gene usually makes a protein called FMRP. FMRP is needed for brain development. People who have FXS do not make this protein. People who have other fragile-X-associated disorders have changes in their FMR1 gene but usually make some of the protein.

FXS affects both males and females. However, females often have milder symptoms than males. The exact number of people who have FXS is unknown, but a review of research studies estimated that about 1 in 7,000 males about 1 in 11,000 females have been diagnosed with FXS.

Signs and Symptoms

Signs that a child might have FXS include :

(i) **Developmental delays** (not sitting, walking, or talking at the same time as other children the same age.

(ii) **Learning disabilities** (trouble learning new skills).

(iii) **Social and behaviour problems** (such as not making eye contact, anxiety, trouble paying attention, hand flapping, acting and speaking without thinking, and beign very active).

Males who have FXS usually have some degree of intellectual disability that can range from mild to severe. Females with FXS can have normal intelligence or some degree of intellectual disability. Austim spectrum disorder (ASD) also occur more frequently in people with FXS.

Testing/Diagnosis

FXS can be diagnosed by testing a person's DNA from a blood test. A doctor or genetic counselor can order the test. Testing also can be done to find changes in the FMR1 gene that can lead to fragile-X-associated disorders.

A diagnosis of FXS can be helpful to the family because it can provide a reason for a child's intellectual disabilities and behaviour problems. This allows the family and other caregivers to learn more about the disorder and manage care so that the child can reach his or her full potential. However, the results of DNA tests can affect other family members and raise many issues. So, anyone who is thinking about FXS testing should consider having genetic counselling prior to getting tested.

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. What is human karyotype? How does banding help in identification of individual chromosomes?

Ans.

Human Chromosomes

The normal diploid number of chromosomes in man is 46 (*i.e.,* 23 pairs). Till 1956, it was believed that each human cell contains 48 chromosomes. The new technique developed by two plant cytologists, **Tizo** and **Levan** in 1956 enabled the human geneticists to ascertain the correct chromosome number.

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

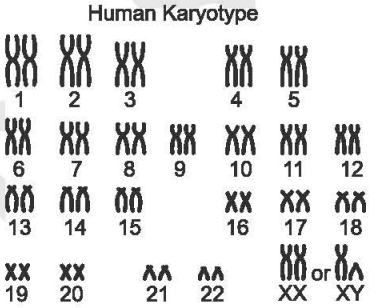


Fig. : A karyotype of human chromosomes based on drawing, using lengths of chromosomes, arm ratios and presence and absence of secondary constrictions.

Human Karyotype

Depending upon the position of centromere and relative length of two arms, human chromosomes are of three types—**metacentric, submetacentric** and **acrocentric.**

The photograph of chromosomes are artificially arranged in the order of descending length in seven groups as shown in the table below :

S. No.	Group	Size	Position of centromere	Idiogram number	Total number of chromosomes in dipoloid cells
1.	A	Large	Metacentric or submetacentric	1, 2, 3	3 pairs
2.	B	Large	Submetacentric	4, 5	2 pairs
3.	C	Medium	Submetacentric	6, 7, 8, 9, 10, 11, 12 & X	7 pairs + 1 in male (7 pairs + 1 pair in female)
4.	D	Medium	Acrocentric	13, 14, 15	3 pairs
5.	E	Small	Metacentric or submetacentric	16, 17, 18	3 pairs
6.	F	Smallest	Metacentric	19, 20	2 pairs
7.	G	Small	Acrocentric	21, 22, Y	2 pairs + 1 in male (2 pairs in female)

1. Karyotype and Idiogram

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

The term idiogram is the diagrammatic representation of a karyotype, which may based on mesurements of chromosomes in several or many cells.

This sort of arrangement of chromosomes represents relative morphology of chromosomes, the **karyotype.** This helps in proper identification and numbering of chromosomes. Any gross morphological change of abnormality in the shape or size of any of the chromosomes in easily identified.

2. Staining with Fluorescent Dyes

For identifying various regions of individual chromosomes a new technique is being practised since 1969. This involves staining the chromosomes with fluorescent dyes after certain treatments. The staining gives different patterns of bands and interbands (stained and unstained regions) along the length of chromosomes. The banding pattern of a particular chromosome remains constant for a particular treatment. At present four such banding patterns are known which are represented as Q, G, C and R banding patterns. According to a normal banding pattern (standarised), abnormalities in different chromosomes can easily be identified.

(i) Q-Banding : The Q-bands are fluorescent bands observed on human chromosomes by staining with **quinacrine (Q) mustard** and observing with UV light. By this staining Y-chromosome becomes brightly fluorescent while the distal end of each chromatid remains unstained.

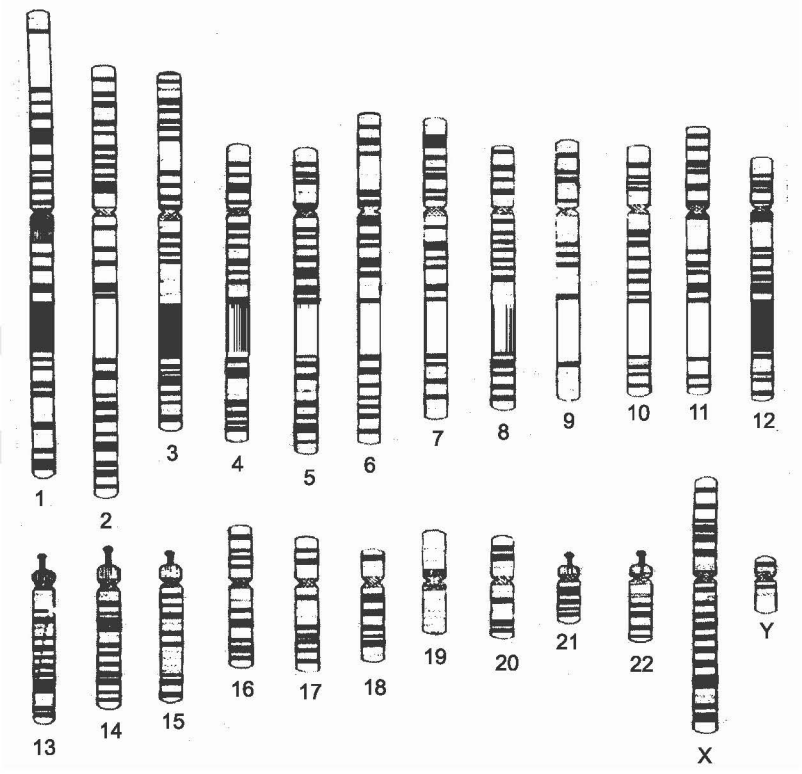


Fig. : Giemsa bands on human chromosomes.

(ii) **G-Banding :** The G-bands are produced by staining human chromosomes with **Giemsa stain.** These bands occur on the same locations as Q-bands. Their staining does not require fluroesent microscopy. There are many techniques of Giemsa staining. In **acid-saline-Giemsa** (ASG) **technique** cells are first incubated in citric and NaCl for 1 hr at 60°C and then treated with Giemsa stain.

(iii) **C-Banding :** The C-bands are regions of heterochromatin (C-represents **constitutive heterochromatin**). These are localized to particular sites on the chromosome. For example, in mouse these are restricted to the centrometric regions of every chromosome.

(iv) **R-Banding :** The R-bands are located in those zones of chromosomes that lie between the fluorescent Q-bands. These appear as green, brightly fluorescent bands with acridine orange staining.

Significance of Study of Human Chromosomes

Study of human chromosomes has helped a lot in correlating various human diseases, malformation and deformities with the abnormalities in the number and structure of chromosomes. These abnormalities may be in the autosomes or sex-chromosomes. The chromosomal abnormalities are present in 4-5 out of every 1000 live births and in one out of every five spontaneous abortions.

Q.2. What is 'Human Genome Project'? Discuss the technique used for chromosomes mapping in human beings. Discuss the progress already made in mapping and sequencing of human genome.

Ans.

Human Genome Project (HGP)

HGP was the international collaborative research program whose goal was the complete mapping and understanding of all the genes of human beings. All our genes together are known as our 'genome'.

The HGP benefited biology and medicine by creating a sequencing model organisms; developing high throughout sequencing technologies, and examining the ethical and social issues implicit in such technologies.

The techniques used for chromosome mapping may be :

(i) **Sequencing of Whole Genome :** Human genome, consisting of 23 chromosomes, contains about 3000 million base pairs of DNA and may contain ~100,000 genes. In order to understand the genome and the genes in humans, it was proposed during mid 1980s, that a detailed study of human genome be conducted, which should include determination of the nucleotide sequence of the whole 3000 Mb of DNA. After considerable debate, the need of this genome-wide study (study of whole genome) was accepted, and this project popularly described as **'Human Genome Project' (HGP)** was launched in USA on 1st October, 1990.

Besides several minor goals, the HGP had the following three major objectives : (i) preparation of dense genetic maps of the genome (ii) development of physical maps of the genome and (iii) determination of the complete sequence of entire DNA representing the human genome.

To facilitate the analysis of human genome, the HGP also proposed that preparation of maps and sequencing of genomes be also undertaken in small set of model organisms including *E. coli,* budding yeast *(S. cerevisiae),* nematode worm *(C. elegans), Drosophila* and the labortory mouse. The genetic maps and physical maps of human genome were thus prepared. The complete and accurate sequence of human genome became available by the year 2005. Each chromosome with its sequence was described as a pseudomolecule.

(ii) Chromosome Mapping in Humans : During the last two decades, human species has become a very favourable material for genetic studies due to the development of methods for parasexual analysis based on fusion of cells in culture. Indirect methods have, therefore, been used for chromosome mapping in humans.

Criteria used for this purpose included the following : (i) family linkage studies, (ii) segregation from cell hybrids, (iii) correlation of loss of marker from cell hybrids with radiation included loss of chromosome segment, (iv) linkage disequilibrium in population with another mapped marker and (v) *in situ* hybridization.

For preparation of genetic and physical maps, a variety of molecular markers were used. These molecular markers included RFLPs (restriction fragment length polymorphism), RAPDs (random amplified polymorphic DNAs), STS (sequence tagged sites), microsatellites or SSRs (simple sequence repeats) and ESTs (expressed sequence tags). The details about these molecular markers are beyond the scope of this book and are available in author's another book *"Elements of Biotechnology"*. Since crosses could not be planned for genetic analysis in humans, segregation patterns of the markers were studied within a panel of reference families (cell lines from 59 such families as a source of DNA have been maintained in Paris) and recombination frequencies calculated from this finite and limited sample size. As mentioned above, complete and saturated genetic and physical maps of human genome have already been made. As a result of these mapping efforts, positions of 30,000 genes are now known and roughly 200 disease associated genes have been identified in human genome.

Q.3. Name a few of the common chromosomal abnormalilties in man. Describe their chromosomal basis and their phenotypic characters.

Ans.

Chromosomal Abnormalities in Man

Two different types of sex-anomalies have been commonly observed in man, which are produced due to nondisjunction of **X-chromosomes.** These are :

1. Klinefelter's Syndrome (44 + XXY)

The persons with Klinefelter's syndrome are those sterile males who possess male appearance but have underdeveloped genitalia, sparse body their and exhibit some degree of breast development. They possess 47 chromosomes instead of normal 46. The interphase nuclei of their cells possess one Barr body. It means they possess XXY-sex chromosmoes instead of normal XY.

Klinefelter's syndrome arises by the nondisjunction of XX chromosmes. When an abnormal egg with XX chromosomes is fertilized with a sperm with Y-chromosomes, the zygote possesses XXX sex-chromosomes. This develops into an abnormal male. One out of every 500 male births possesses Klinefelter's syndrome.

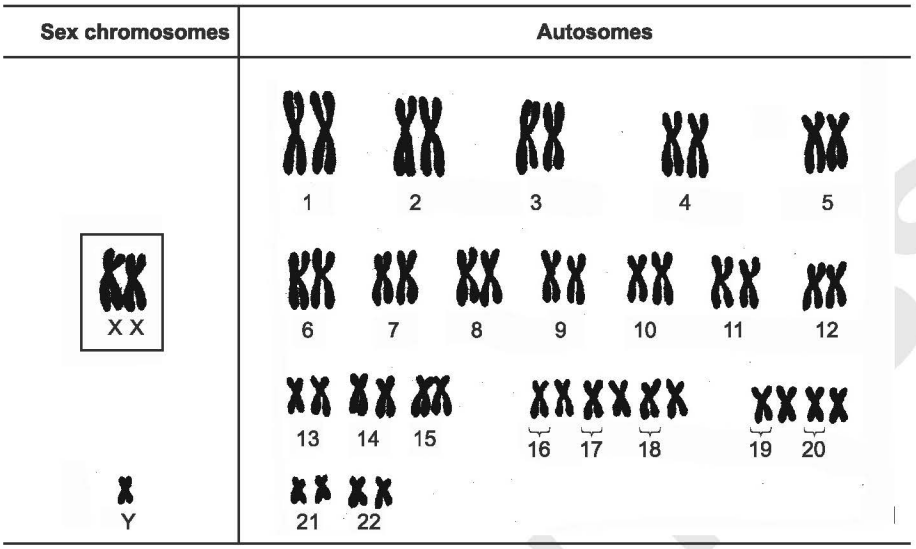


Fig. 1 : Karyotype of a man showing Klinefelter's syndrome.

2. Turner's Syndrome (XO)

The persons with Turner's syndrome are phenotypic females but their interphase nucleus is without a Barr body. These are sterile females with poorly developed ovaries and underdeveloped breasts. These exhibit webbed neck, low set ears and broad chest. Their intelligence is also below average. In such women, instead of normal ovaries only ridges of whitish tissue are present which are known as **'streak gonads'.**

Many scientists use the term **'gonadial dysgenesis'** to replace Turner's syndrome. One in every 25,000 births exhibits Turner's syndrome. They possess 45 chromosomes instead of 46. Since they possess no Barr body, it means they possess only one X-chromosome (XO).

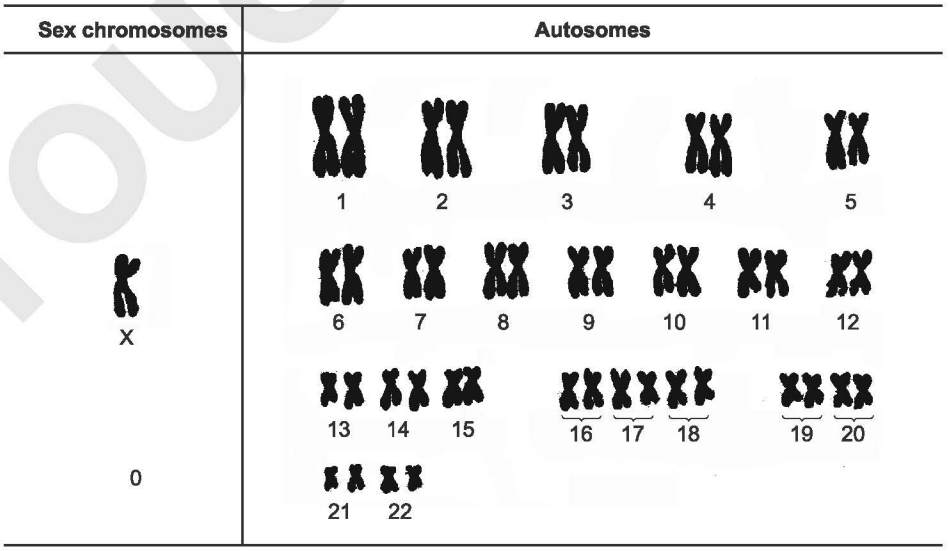


Fig. 2 : A woman showing Turner's syndrome.

Males with XXY (diplo-X), XXXY (triplo-X), XXXXY (tetra-X) or XXXXXY (penta-X) constitution were observed. All these extra X arise as a result of nondisjunction of sex-chromosomes.

The occurrence of XYY chromosomes abnormality was first observed in 1962 by T.H. **Hauschka,** et. al. The extra Y chromosome is strongly male determining. The extra Y chromosome leads to over production of male hormone, which causes over aggressiveness. So XXY men are prone to violence, criminality and antisocial behaviour. Such persons are usually taller than average, have barely normal **I.Q.** and suffer persistence. **XYY** genotype is doubted to be present in one out of 300 males.

3. Triplo-X Female

Such females are known as **superfemales.** These are usually mentally retarded and infertile. However, some triplo-**X** females are apparently normal and fertile. The children born to such females are found to be normal, presumably only **X**-bearing eggs are functional.

Sex Abnormalities Associated with Single Gene Pair

(i) **Pure Gonadial Dysgenesis :** Such females have **XY** chromosomes composition but have streak ovaries and lack secondary sexual characteristics. This develops because of a defective gene or genes, which suppress in some way the male determining function of Y-chromosome.

(ii) **Testicular Feminization :** In some exceptional cases, the individual is reared as female, has female features and attitute but genetically has normal **XY**-chromosomes. Such an individual has well developed breasts, but lacks ovaries and never menstruate. The vagina ends blindly inside. The gonads are a pair of undescended testes in the place of missing ovaries. The undescended testes produce estrogens producing secondary sexual characteristics. Such a condition appears because of some defective genes that alter the male-determining role of **Y**-chromosome and leads to sex reversal.

Q.4. What is pedigree analysis? How can this be used for identification of genes controlling specific traits controlled by (a) dominant allele, (b) recessive allele.

Ans.

Pedigree Analysis

Pedigree represent family members and relationships using standardized symbols. By analzing a pedigree, we can determine genotypes, identify phenotypes, and predict how a trait will be passed on in the future.

Genetic Analysis through Pedigree Charts

Although conventional genetic analysis in human beings is restricted due to small family size and the difficulty of experimental matings, pedigrees provided useful means of genetic analysis, particularly for single gene defects. Some examples of these single gene disorders **alcaptonuria, phenylketonuria, albinism** and **sickle cell anaemia.** Two other important triats in humans include (i) dominant gene for **ability to roll one's tongue** (found in 85% people) and (ii) dominant gene for ability to taste the bitterness of **phenylthiocarbamide (PTC).**

Identification of genes in human beigns is also difficult partly due to the difficulty is producing induced mutations, although such induced mutations in cell cultures can be produced. Single gene mutations can be autosomal or sex linked, and largely recessive. In all such cases, the data from pedigree can be pooled and 3 : 1 ratio in the progeny of two heterozygotes and 1 : 1

ratio in the progeny of a heterozygote and a homozygous recessive, can be demonsrated, although the ratio may be distorted in favour of dominant phenotype due to miscarriages caused by deleterious effect in homozygous recessive condition. Some of the single gene disorders identified from pedigrees are listed in table 2. Many others are known which are rare.

The application of Mendel's laws for a study of problems of heredity in humans will be most interesting, yet it is most difficult for the following reasons (besides others) : (i) controlled crosses can not be made, (ii) duration required for a study of three generations will be long (several decades). In view of the above, human geneticists often scrutinize family histories (pedigrees) in the hope that informative matings might have occurred by chance. This is called **pedigree analysis.** A member of a family having an exceptional phenotype (e.g. colour blind or deaf or dwarf), which first attracts the attention of a geneticist, is called **propositus.** The history of the exceptional character in the propositus is traced back in the family and a family tree is prepared using standard symbols.

Symbol	Explanation	Symbol	Explanation
□	Male	■ ● ♂ ♀	Affected individuals
○	Female	◧ ◐ ♂ ♀	Heterozygotes for autosomal recessives
□—○	Mating	◉ ♀	Carrier of sex linked recessive
□—○ with children	Parents and childern (1 boy 1 girl, in order of birth)	⊘	Death
△	Dizygotic twins	●	Abortion or stillbirth (Sex unspecified)
△	Monozygotic twins	■	
◇	Sex unspecified	I 1 2 ; II 1 2 3	Propositus method of identifying persons in a pedigree : here the propositus is child 2 in generation 2 or II 2
[2] ③	Number of children of sex indicated	□═○	Consanguineous marriage

Fig. 1 : Symbols used in human pedigree analysis.

Many human diseases or defects are governed by simple genes. Common examples of these traits include albino, Tay-Sachs disease, cystic fibrosis, phenylketonuria (PKU), etc. Following generalizations can be made : (i) a recessive character may appear in the progeny of both unaffected parents; (ii) two affected parents can not have an unaffected child; (iii) often these characters appear among children born from consanguineous marriages (e.g. first cousin marraige), which enhance the chances of mating among two heterozygotes for the same recessive trait. This also explains why often consanguineous marriages (or sagotra marriages among boy and girl from same gotra—ancestry) are forbidden in several castes in our country.

A typical pedigree for a rare recessive condition is shown in fig. Rarely the exceptional character may be governed by dominant alleles (e.g. Huntington's chorea; brachydactyly—very short fingers). This situation will be characterized by the following : (i) the condition occurs in every generation; (ii) unaffected parents can never transmit the condition to the offspring; (iii) two affected parents may have unaffected children; (iv) condition is passed on, on an average, to one half of the children of an affected individual. A typical pedigree of a rare dominant condition is shown in fig. 3.

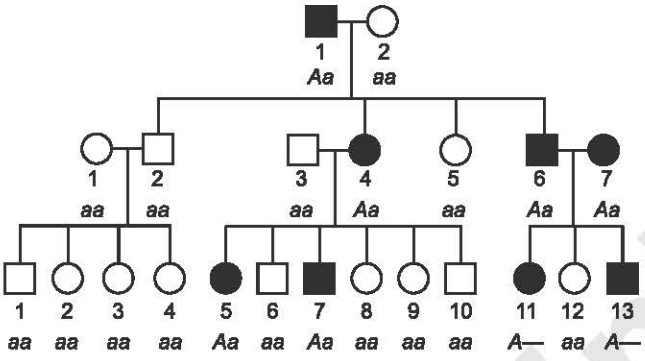


Fig. 2 : A pedigree involving an exceptional phenotype controlled by recessive allele 'a' (gene symbols are generally not given in pedigrees; they are given here as an aid to the readers; redrawn from Suzuki *et al.* 1986).

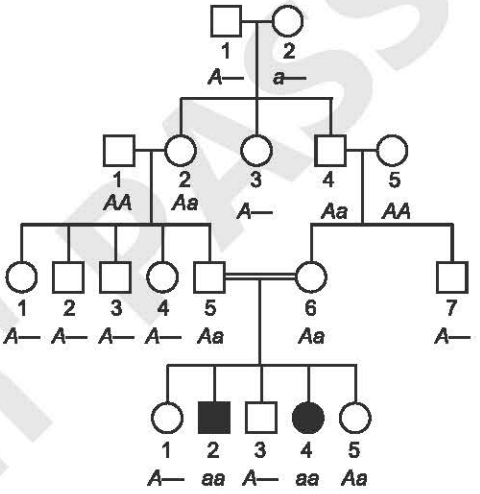


Fig. 3 : A pedigre involving a rare phenotype controlled by a dominant allele 'A' (redrawn from Suzuki *et al.* 1986.)

The pedigree analysis as above, helps in the identification of genes and gives information about the mode of its inheritance. The medical applications of such a genetic analysis, obviously, are far reaching. This information is particularly useful in day-to-day counselling of prospective parents, who fear genetic disease in their children.

Approximate Frequencies of some of the most common Recessive Deleterious Mutant Alleles in Man

Condition	Frequency per million birth	Frequency of carriers %	Frequency of allele %
Autosomal recessives			
1. Fibrocystic diseases	400	4	2
2. Phenylketonuria	100	2	1
3. Albinism	100	2	1
4. Tay-Sachs disease	10	0.6	0.3
5. Galactosaemia	5	0.4	0.2
Autosomal dominant			
6. Huntington's chorea	100	0.02	0.01
X-linked recessives			
7. Duchenne's muscular dystrophy	200	0.01 (female)	0.01 (feamles)
8. Haemophilia	10	0.005 (females)	0.005 (females)

UNIT-VIII

Infectious Diseases

SECTION-A VERY SHORT ANSWER TYPE QUESTIONS

Q.1. Give names of three pathogens causing infectious diseases.

Ans. A variety of microorganisms can cause disease. Pathogenic organisms are of five main types : viruses, bacteria, fungi, protozoans and worms. Pathogens of all classes must have mechanisms for entering their host and for evading immediate destruction by the host immune system. Most bacteria are not pathogenic.

Q.2. Name the pathogen, which causes diarrhoea.

Ans. *Rota virus* and *E. coli* are the two most common etiological agents of moderate-to-severe diarrhoea in low income countries. Other pathogens such as *Cryptosporidium* and *Shigella* sp. may also be important.

Q.3. Give the scientific name of a species of *Trypanosoma*.

Ans. African Trypanosomiasis also known as 'sleeping sickness' is caused by microscopic parasites of the species *Trypanosoma brucei gambiense*.

Q.4. Name any one respiratory disease, which is not caused by a protozoan.

Ans. Tuberculosis is not caused by a protozoan.

Q.5. What is pathogenesis?

Ans. Pathogenesis is the process by which a disease or disorder develops. It can include factors which contribute not only to the onset of the disease or disorder, but also to its progression and maintenance.

Q.6. What causes tuberculosis?

Ans. Taberculosis (TB) is caused by a type of bacterium called mycobacterium tuberculosis. It's spread when a person with active TB disease in their lungs caughs or sheezes and someone else inhales the expelled droplets, which contain TB bacteria.

Q.7. Define the process of somatogamy in brief.

Ans. Somatogamy takes place in fungi where formation of gametes is absent, *e.g.,* yeasts, mushrooms (*Agaricus*) and bracket fungi. The fusion involves two vegetative or somatic cells belonging to the same thallus or two physiologically different thalli.

Q.8. Write about the structure of bacterial flagellum in brief.

Ans. Bacterial flagella are the most primitive of all the motile organs. Each is composed of a single thin fibril (unistranded) as against the 9 + 2 fibrillar (11 stranded) structure of eukaryotic cells. It consists a few fine fibrils twisted tightly together into a rope-like structure. The flagellum is composed of protein **flagellin.** According to **Lowy** and **Hanson** (1965), bacterial flagellum is composed of globular subunits arranged in helices of various kinds. The diameter of each subunit is about 40-50 Å.

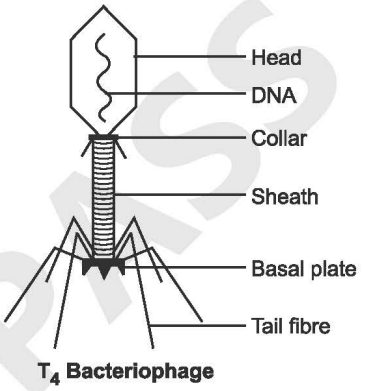
SECTION-B SHORT ANSWER TYPE QUESTIONS

Q.1. Define the structure of bacteriophage with the help of diagram.

Ans.

Structure of Bacteriophage

The phages may be spherical or comma-shaped but majority of them have tadpole-like appearance. The structure of T_4 **bacteriophage,** a parasite of bacterium *Escherichia coli,* is tadpole-shaped and is differentiated into head, collar and tail.



T_4 Bacteriophage

Fig. : Bacteriophage

(i) The **head** is polyhedral or hexagonal. Its **head capsid** is made up of about 2,000 **capsomeres,** each has a molecular weight of about 80,000. The head capsid encloses a circular double-stranded DNA about 53 μm long with more than 75 genes.

(ii) The tail is long and in the form of a hollow cyclinder. It consists of a central hollow **core,** surrounded by a spring-like **contractile sheath.** The sheath is formed of 144 subunits of protein, arranged in a helical pattern around the core.

The core with contractile sheath rests on a hexagonal **end plate.** Six tail fibres and 6 short **spikes** protrude out from the end plate. The tail fibres and spikes are formed of bundles of polypeptide chains.

Q.2. By giving a structural diagram, write about Tobacco Mosaic Virus (TMV).

Ans.

Tobacco Mosaic Virus (TMV)

TMV is the extensively studied plant virus. It was discovered by **Iwanowski** in 1893. This has been obtained in the form of fine crystals and needles by **Stanely** in 1935. TMV has a helical symmetry, cylindrical in structure, about 160-300 Å long. It has a molecular weight of 40 millions.

Its protein coat has 2310 identical protein subunits, each with a molecular weight of 18,000 daltons. Each subunit consists of a single chain of 158 amino acids of known sequence. RNA of the virus is single stranded. Its molecular weight is 2.4 million daltons having 6500 nucleotides.

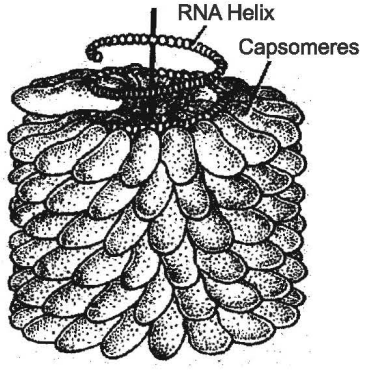


Fig. : Structure of Tobacco mosaic virus

TMV infects the leaves of tobacco plants. It produces chlorosis in plants. It is transmitted mechanically by rubbing or by insect vector, grasshopper. According to **Fraenkal Conrat** (1956), RNA is infectious. The protein part remains in host cytoplasm while RNA moves from cell to cell through plasmodesmata. Viral RNA, after entering the host cell takes control of the host cell metabolic activities and initiates its own replication.

Q.3. Explain the chemoautotrophic bacteria.

Ans. Chemoautotrophic Bacteria : These bacteria prepare their organic food from inorganic raw materials with the help of energy derived from exergonic chemical reactions. These reactions involve oxidation of some inorganic substance present in the external medium. The chemical energy obtained from oxidation reactions is trapped in ATP molecules. This energy is utilised in the assimilation of CO_2 with the help of hydrogen obtained from sources other than water.

Therefore, no oxygen is evolved in this process. A few chemoautotrophic bacteria are :

(i) **Nitrifying bacteria** obtain energy by oxidising ammonium ions to nitrite, e.g., *Nitrosococcus and Nitrosomonas.*

$$NH_4^+ + 2O_2 \longrightarrow NO_2 + 2H_2O + \text{Energy}$$

(ii) Another category of bacteria obtain energy by oxidising nitrites to nitrates, *e.g., Nitrobacter* and *Nitrocystis.*

$$2NO_2^- + O_2 \longrightarrow 2NO_3^- + \text{Energy}$$

(iii) **Sulphur bacteria** oxidise hydrogen sulphide to sulphur and get energy for chemosynthesis, *e.g., Beggiotoa.*

$$2H_2S + O_2 \longrightarrow 2S + 2H_2O + \text{Energy}$$

(iv) Some bacteria oxidise sulphur to get energy, *e.g., Thiobacillus thiooxidans.*

$$2S + 2H_2O + 3O_2 \longrightarrow 2H_2SO_4 + \text{Energy}$$

(v) **Iron bacteria** obtain energy by oxidising ferrous compounds to ferric compounds :

$$4FeCO_3 + 6H_2O + O_2 \longrightarrow 4Fe(OH)_3 + 4CO_2 + \text{Energy}$$

Nitrifying and sulphur oxidising bacteria participate in nitrogen and sulphur cycles in nature.

Q.4. Name the bacteriologist who discovered gram-positive and gram-negative bacteria. Tabulate the major differences between Gram-positive and Gram-negative bacteria.

Ans.

Gram-Positive and Gram-Negative Bacteria

Bacteriologist, named **Gram,** stained bacteria with crystal violet, stabilised with 0.5% iodine solution and washed with acetone or alcohol. He found that Gram positive bacteria turned to **deep violet** or **purple** but Gram negative bacteria remained turned **red.** Bacteria which turned to purple are called **Gram-positive** and those which become red are called **Gram-negative.**

Differences between Gram-Positive and Gram-Negative Bacteria

S.No.	Gram Positive Bacteria	Gram Negative Bacteria
1.	Retain Gram stain.	Do not retain Gram stain on washing with organic solvents like acetone or absolute alcohol.
2.	Wall is single layered.	Walls is two-layered.
3.	Cell wall is 20-80 nm.	Cell wall is comparatively thin, *i.e.,* 7.5-12 nm.
4.	Lack of pili or fimbriae	Pili or fimbriae present.

5.	Cell membrane invaginates and gets folded inside the cytoplasm of the cell forming the mesosome.	Mesosomes are absent.
6.	Cell wall contains only traces of lipids.	Contains up to 20% lipids.
7.	Pathogenic forms are fewer.	Pathogenic forms are more abundant.
	Examples : *Streptococcus, Bacillus, Clostridium, Streptomyces.*	**Examples** : *Salmonella, Pseudomonas, Vibrio, Rhizobium, Escherichia.*

Q.5. Describe the life cycle of bacteriophage.

Ans.

Life Cycle of Bacteriophage

Bacteriophage multiply inside bacterial cells only. They exhibit two types of life cycles :

1. Lytic Life Cycle (Replication Phage)

The **virulent phage** causes lysis (destruction) of host cell and exhibits lytic cycle. Each phage particle gets attached to the host cell wall with the help of its tail. The bacterial cell wall dissolves at the point of contact and DNA of bacteriophage migrates inside the bacterial cell. The protein coat of head is left outside on the host surface.

Once inside the host cells, the T_4 phage-DNA takes control of host cell machinery. It starts synthesising copies of phage DNA and coat proteins. Host cell DNA disintegrates and disappears. The new phages, are released by the rupture of bacterial cell wall. Within 30 minutes, about 100 new phage particles are formed from a single virus.

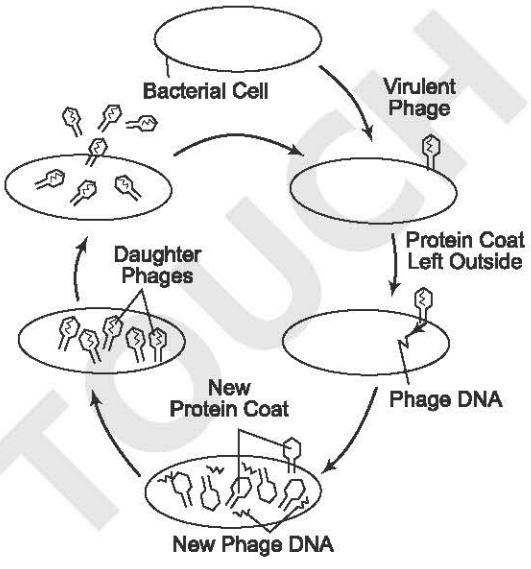


Fig. 1 : Lytic life cycle in T_2/T_4 bacteriophage.

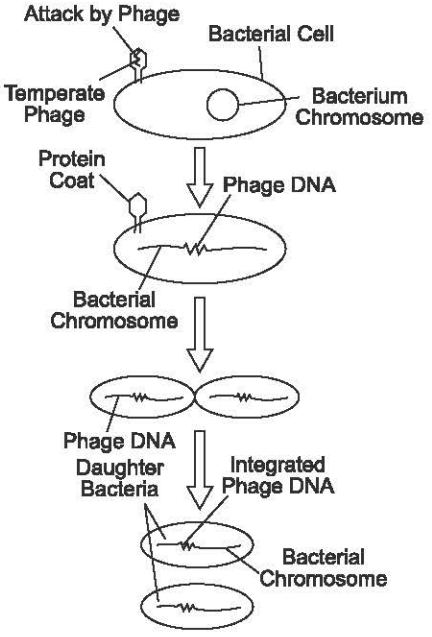


Fig. 2 : Lysogenic life cycle in temperate phage.

2. Lysogenic Life Cycle

Some phages live inside host cell without causing lysis of bacterial cell. Such novirulent viruses are called **temperate phages** or **symbiotic phages.** Phage λ may cause cell lysis or behaves as a symbiont, showing **lysogenic life cycle.**

In lysogenic cycle DNA of phage λ becomes integrated into *E. coli* chromosome. The integrated viral DNA is called **prophage.** It replicates along with bacterial chromosomes and is transmitted to bacterial progeny. Prophage can remain in the bacterial cells for many generations or may get isolated from host DNA. The isolated phage DNA multiples independently and causes bacterial lysis.

Q.6. Write differences between chemosynthesis and photosynthesis.

Ans. Differences between Chemosynthesis and Photosynthesis

S.No.	Chemosynthesis	Photosynthesis
1.	Does not require light as a source of energy.	Sunlight is essential for the synthesis of food.
2.	It can occur day and night.	It occurs during day time only.
3.	Energy required for the process is obtained by the oxidation of inorganic substances.	Sun is the source of energy.
4.	It is a slower process.	It is a rapid process.
5.	It occurs only in certain bacteria.	It occurs in cyanobacteria, some bacteria and in green plants.
6.	It is anoxygenic, *i.e.,* no oxygen is produced.	Except bacterial photosythesis, it is always oxygenic.

Q.7. Write a short note on heterothallism.

Ans. Heterothallism

The phenomenon of the existence of two genetically different and sexually compatible strains in separate thalli in a species is called heterothallism. This phenomenon was observed by an American botanist, **A.L. Blakeslee,** in 1904.

Blakeslee, while studying sexual process in Mucorales (*Rhizopus* and *Mucor*), observed that some species develop zygospores independently, whereas in others, they are formed only when two mycelia of different strains come in contact with each other. He coined the terms **homothallism** and **heterothallism** to explain this phenomenon. The homothallic species produce zygospores independently, whereas heterothallic species require the presence of opposite mating types. *Mucor mucedo* and *Rhizopus stolonifer* are examples of heterothallic species. In these species, although the two fusing hyphae are morphologically similar but differ physiologically. Blakeslee called them **plus** (+) and **minus** (–) strains. These strains do not produce zygospores singly but form zygospores when grown together.

Heterothallism as a Device to Prevent Inbreeding : In sexually reproducing haploid fungi, several genetically identical individuals occur together. They are formed from the same parent fungus by mitosis. But sexual reproduction between such haploid individuals of the same mating type does not take place.

Thus, heterothallism is a device to prevent **inbreeding,** *i.e.,* raising progeny by mating two genetically similar mating types of organisms and promote **outbreeding,** *i.e,* raising progeny by mating two genetically different mating types of organisms. This ensures genetic variability and produces offspring with new combinations of characters and ability to overcome unfavourable conditions.

Q.8. Define the karyogamy.

Ans. Karyogamy

It involves fusion of two haploid nuclei which come together in plasmogamy, thus, forming a **diploid** nucleus called **synkaryon.**

There is great variation in fungi regarding the duration between plasmogamy and karyogamy. In lower fungi, karyogamy follows plasmogamy almost immediately. In higher fungi like *Puccinia* and *Neurospora* and the mushrooms, karyogamy is much delayed and occurs just before meiosis. In the stage intervening between plasmogamy and karyogamy, the cells often contain two nuclei or **dikaryons.** Such cells are called dikaryotic cells and the phase as **dikaryotic phase.**

Q.9. Give a brief account of the zoospores.

Ans. **Zoospores** are thin-walled uninulceate structures formed in zoosporangium. Since these possess flagella and swim in water, these are called **zoospores.** These may be with one flagellum as in *Synchitrium* or with two flagella as in *Pythium.* Zoospores may be of two types :

(i) **Pear-shaped** or **pyriform** with two flagella placed at anterior end, and

(ii) **Kidney-shaped** or **bean-shaped** with two oppositely directed flagella inserted laterally in furrow.

Zoospores germinate to give rise to a new mycelium. Reproduction by zoospores occurs in Phycomycetes (*Albugo, Phytophthora* and *Saprolegnia*).

Dispersal of Spores : Zoospores produced by aquatic fungi are motile and possess flagella for swimming. Most terrestrial fungi produce nonmotile and minute spores in large numbers; These are easily dispersed by wind. On falling over a suitable medium, they germinate and the developing hyphae grow to produce new mycelia.

Q.10. Write a short note on role of fungi.

Ans.

Role of Fungi

Fungi play a key role is producing medicines, in bakery for making bread and as environmental decomposers.

1. **In Medicines :**
   (i) **Citrium** from *P. citrinum.*
   (ii) **Ergotine** from *Claviceps purpurea.*
   (iii) **Chaetomin** from *Chaetomium cochlides.*
   (iv) Life saving wonder drug, **Penicillin,** is obtained from *Penicillium notatum* and *P. chrysogenum.*
   (v) **e-Ephedrine** is synthesised by yeast from benzaldehyde.
   (vi) **Cortisone,** a steroid is prepared by the fermentation of glycosides by moulds like *Rhizopus nigricans* and *Aspergillus niger.*
2. **Vitamins :** Many fungi are rich in vitamins. **Thiamine** (B_1), **riboflavin** (B_2) and ergosterol, a precursor or vitamin D are obtained from yeast. **Vitamin A** is obtained from *Rhodotorulas gracilis.*
3. **In Bakery :** *Saccharomyces cerevisiae* (yeast) is used in bread making. It is a fermentation process in which CO_2 is produced which makes the Flour spongy. Yeast enzymes act on proteins which make them more digestible.
4. **As Environmental Decomposers :** Because of saprophytic nature, Fungi carry out the decomposition of dead remains of plants and animals and the organic matter. That are deposited in soil in the form of litter. Decomposition of litter and organic matter causes

release of various elements and compounds trapped in complex organic compounds. These inorganic compounds are again absorbed and used by the plants. By their decomposing activity, Fungi help keep the environment clean.

Q.11. Give a brief account of *Leishmania* sp. pathogenic in human beings.

Ans. The pathogenic species of Leishmania in human beings are as follows :

(i) ***Leishmania donovani*** **:** It causes **kala-azar.** This disease is common in East Asian countries, India and parts of Africa and America. The fever is accompanied with anaemia, enlargement of liver and spleen. Dogs and cats are reservoir host. The parasite is transmitted by a sand fly, *Phlebotomus argentipes* and other species. The parasite lives in the cells of liver, spleen, lymph glands and bone marrow.

(ii) ***Leishmania tropica*** **:** It causes **oriental sore** in humans. The disease is characterised by cutaneous sore on hands, feet and face. The disease spreads by sand flies. The parasite lives in the endothelial cells of skin capillaries.

(iii) ***Leishmania brasiliensis*** **:** It causes **espunda** disease in man. The disease is characterised by lesions on skin and mucous membrane of nose, mouth, pharynx and sometimes vagina. The disease is transmitted by sand flies.

Q.12. Write a short note on malarial parasite.

Ans. ***Plasmodium* (Malarial parasite)**

Various species of *Plasmodium* cause different types of malaria in man. It casual organism was discovered by **Laveran** in 1880, **Sir Ronald Ross** discovered that the disease spreads by mosquito bite.

Life cycle of *Plasmodium* is **digenetic,** *i.e.,* it completed on two hosts: Female Anopheles mosquito (the primary host) and man (the intermediate or secondary host). The female *Anopheles* mosquito acts as **vector** or carrier of malarial parasite.

The female *Anopheles* mosquito carries the **sporozoite** stage of the protozoan. When it bites man, sporozoites enter the blood. From blood sporozoites enter the liver and change to **schizonts.** The schizonts produce cryptomerozoites which enter RBCs and form **trophozoites.** These become amoeboid and form pseudopodia. They grow in size and become **schizonts.** They by multiple fission produce merozoites. Some merozoites beahve as gametes.

When female Anopheles sucks blood, gametes pass into the gut. In the gut of mosquito sexual reproduction takes place by the fusion of gametes : The zygotes, so formed, produce needle-shaped **sporozoites** which enter the blood of man by mosquito bite.

Q.13. Describe the brief account of *Entamoeba* histolytica.

Ans. ***Entamoeba Histolytica***

It was discovered by Lamble in 1859 and Losch gaves its pathogenic nature in 1875. Life cycle of *Entamoeba* is monogenetic, *i.e.,* it completes its life cycle in a single host (*i.e.,* man).

Entamoeba resides in the upper part of the human large intestine and causes **amoebiasis** (amoebic dysentery). Symptoms of this disease are abdominal pain, repeated motions with blood and stool. Parasite has only one pseudopodium. Contractile vacuole is absent. It feeds on red blood corpuscles by damaging the wall of large intestine and produces ulcers.

E. histolytica occurs in two forms—(i) **magna** (trophozoite), the pathogenic form found in the mucosa and submucosa of intestine forming ulcers and (ii) **minuta** (nonpathogenic form.) found

in the lumen of the intestine. A mature cyst is called **quadrinucleate cyst.** It has **four nuclei** and two **chromatoid bodies.** The quadrinucleate cyst is the infective stage of *Entamoeba.* On entering the intestine of new host, thy single cyst of *E. histolytica* produces eight amoebae.

Q.14. Enumerate characteristic features of Platyhelminthes.

Ans. Platyhelminthes are soft-bodied, elongated, dorsoventrally flattened and legless, worm-like animals. They are bilaterally symmetrical, triploblastic, acoelomate animals with organ-system level of organisation but without respiratory, circulatory and skeletal systems. They are represented by about 13,000 species. The term **Platyhelminthes** was coined by **Gegenbaur** (1859) for flatworms.

General Characteristics

1. **Habit and Habitat :** Flatworms are free-living, commensal or parasitic organisms. The free-living worms may be terrestrial or freshwater. Majority of flatworms are endoparasites.
2. **Symmetry and Body Plan :** The body is bilaterally symmetrical and dorsoventrally flattened. They exhibit **blind sac body plan.**
3. **Germ Layers :** Flatworms are **triploblastic.**
   (i) The ectoderm gives rise to **epidermis** (the outer layer)
   (ii) The endoderm gives rise to **endodermis** (the inner layer) and
   (iii) The embryonic mesoderm forms the middle layer which is formed of **mesenchyme** or **parenchyma.**
4. **Level of Organisation :** Flatworms exhibit definite **organ-system level of organisation.** Flatworms are the first and the simplest animals that have organs and organ systems. The systems are simple.
5. **Body cavity :** Body cavity or coelom is absent, *i.e.,* flatworms are **acoelomate.** Space between the body organs and body wall is filled **with mesenchyme** or **parenchyma** derived from embryonic mesoderm.
6. **Cephalisation :** Flatworms have definite anteior and posterior ends. Concentration of sense organs at the anterior end marks the beginning of **cephalisation.**
7. **Skeleton :** Flatworms are without any skeleton except that they have thick **cuticle.** It protects their body from host's digestive juices. The cuticle may bear chitinous hooks or spines for attachment.
8. **Alimentary Canal :** It is a highly branched tube with mouth but no anus. A muscular suctorial pharynx is present for taking in food. Highly branched intestine distributes digested food to various cells of the body and compensate for circulatory system.
9. **Respiration :** Free-living forms respire through body surface. Parasitic forms are mostly anaerobes.
10. **Excretory organs :** These are **protonephridia** which end in specialised **flame cells.** The flatworms are **ammonotelic.**
11. **Circulatory System** is absent.
12. **Nervous System :** It is formed of a simple **brain** having two masses of nervous tissue, called **ganglia.** Ganglia lie at the anterior end and are connected to nerve cords that

extend lengthwise and are connected by a series of transverse nerve connections. Thus, it gives the appearance of a ladder.

13. **Sense Organs :** These are absent in parasitic forms. In free-living planarians, there is a pair of eye spots at the anterior end or cephalic region. These are sensitive to light.
14. **Organs of Adhesion :** These are hooks, suckers and spines.
15. **Reproduction :** Reproductive organs are highly developed. Free-living planarians have great power of **regeneration.** Planarians are **unisexual** whereas other helminths are **hermaphrodite.**
16. Life cycle in parasitic forms complicated with a number of larval stages.

Q.15. Write short note on sheep liver fluke.

Ans. ***Fasciola hepatica* (Sheep Liver Fluke)**

Liver fluke is endoparasite in the liver of Sheep and Goat. It remains attached to the bile ducts and feeds on bile, blood cells and cellular debris. It respires **anaerobically.**

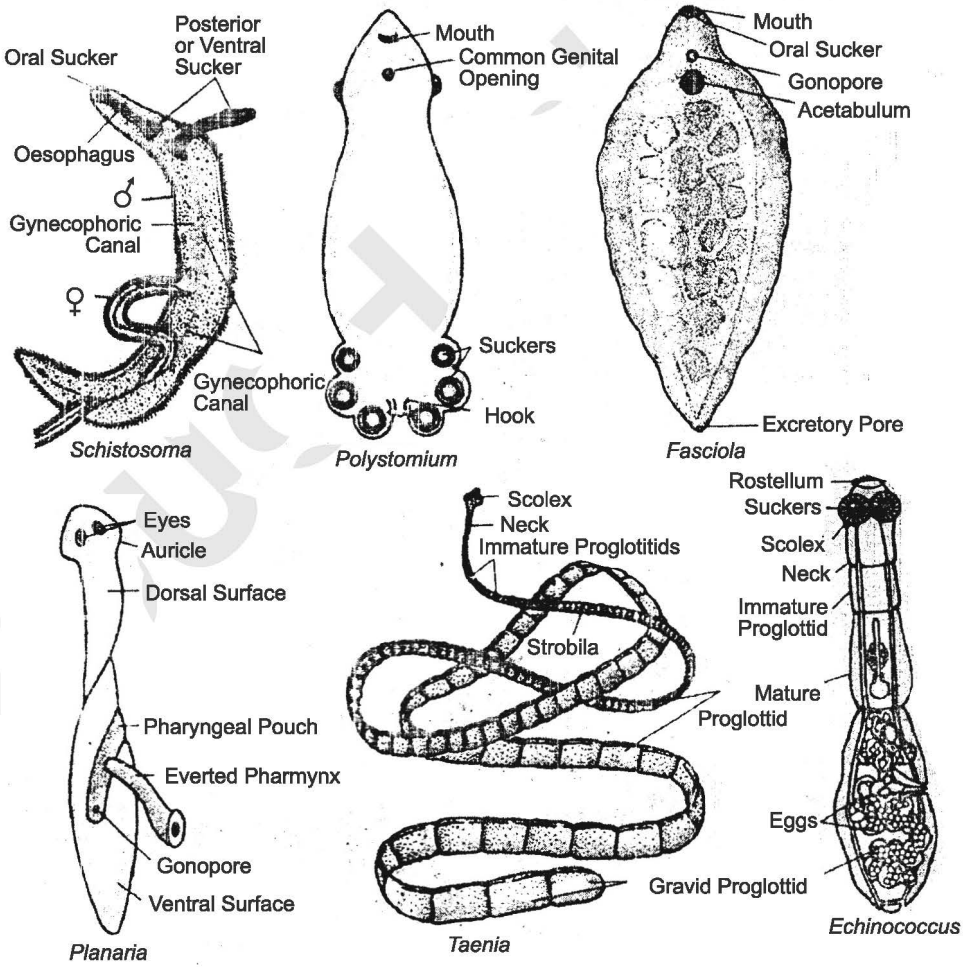


Fig. : Some common Platyhelminthes

Fasciola has flat, leaf-like body and dark brown in colour. Body is triangular with a small conical projection from the anterior end. It is called head lobe. It bears terminal **mouth**, surrounded by oral sucker. A large cup-shaped **ventral sucker** lies behind the head lobe. **Gential aperture** is a minute median aperture anterior to ventral sucker. **Excretory pore** lies midventrally at the posterior end. Phanyona is suctorial intestine is highly branched to supply digested food to all body parts. Anus is absent. Liver flukes are hermaphordite.

Liver fluke is a **digenetic trematode.** Life cycle is completed on two hosts. The primary host is sheep and intermediate host is snail, *Limnaea.* The life cycle includes several larval stages : **miracidium, sporocyst, redia, cercaria** and **metacercaria.** Development shows polyembryony.

Q.16. Describe in short external features of *Schistosoma.*

Ans. *Schistosoma* is a blood fluke. Because of living inside blood vessels, it has become thin, elongated vermiform. Its body is pinkish or greyish pink in colour. Sexes are separate and sexual dimorphism is much pronounced. Female is thin and lies in the gynaecophoric canal of male which is formed by the folds of ventral bodywall. Following structures are visible under the microscobe :

1. **Suckers :** Blood fluke has two suckers :
    1. **Anterior sucker** or **oral sucker** around mouth
    2. **Posterior sucker** or **ventral sucker** lies slightly behind the oral sucker on the ventral surface. It has a small stalk and is also called **acetabulum.**
2. **Gynaecophoric Canal :** It is present on the ventral surface of male fluke and is formed by the folds of ventral bodywall. It encloses mature female :
3. **Apertures :** These are :
    1. **Mouth**, at the anterior end of the body, surrounded by oral sucker.
    2. **Gonopore,** a median aperture just behind the acetabulum.
    3. **Excretory pore,** a medium aperture at the posterior end of body.

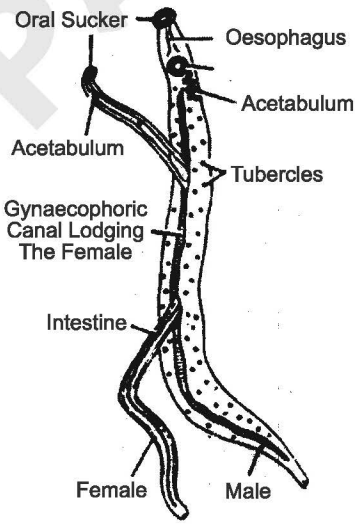


Fig. : ***Schistosoma***

SECTION-C LONG ANSWER TYPE QUESTIONS

Q.1. Describe structure of a virus with the help of digrams. Why viruses considered both living as well as non-living?

Ans.

Viruses

Viruses are infectious agents that typically consist of a molecule of nucleic acid in a protein coat. These are too small to bee seen by light microscope and multiply only within the living cells of the host. Viruses may have either RNA or DNA as their genetic material. It may be single or double-stranded. Thus viruses are **obligate intracellular parasites.** Their host cells may be bacteria, plant cells, animals cells and also human cells.

Viruses were first observed by **Iwanowski** in 1892. He described the detailed structure of **tobacco-mosaic virus (TMV).**

Size and Shape

Viruses are the smallest living entity, as far as known. The size of viruses is expressed in terms of millimicrons (mμ), [1 mμ = 1/1000 of a micron (μ) = 1/100 of a millimeter]. Mostly viruses are less than 0.1 μ or 300 mμ in diameter.

In form (shape), viruses differ widely. They may be rod-like, elongated like a piece of electric cable (TMV), rounded (mumps virus, herpes virus, influenza virus) or tadpole-like (T-even bacteriophage). The form of some viruses, especially the large pox viruses are complex. Rabies virus is bullet-shaped.

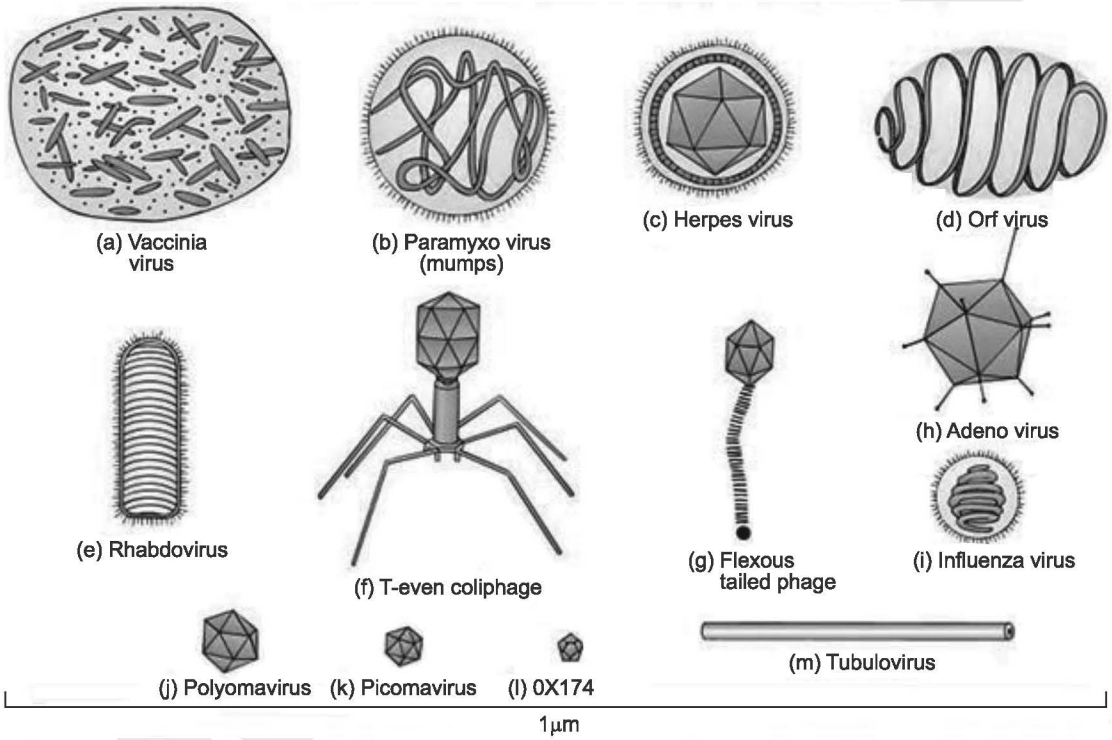


Fig. 1 : Morphology of various virus particles.

The Envelope

It is the outermost membranous envelope found in a few icosahedral and helical animal viruses. It is usually 100-150 Å (0.1-0.15 μm) thick. Some plant viruses and bacteriophages also bear an external envelope. The envelope consists of a phospholipid bilayer in which proteins are embedded.

The virus envelope posseses the protein of a host cell as well as proteins specified by the virus. Sufficient amount of carbohydrate is also present in viral envelopes. Glycoproteins and glycolipids are the major carbohydrate constituents of enveloped viruses. The source of lipid components of virus envelope is form host cells. Phospholipids, cholesterol, fatty acids and glycolipids are the major type of lipids found in viruses envelopes.

Structure

The complete single particle of virus is called **virion.** The protecting protein coat of virus is known as capsid, and the smaller subunits of protein are termed as capsomeres. The central core consists of nucleic acid (DNA or RNA).

Inside the host cells, RNA or DNA exists in the form of replicating nucleic acid molecules devoid of protein coat.

1. **Capsid :** The capsid is composed of a large number of protein subunits all of the same shape. These units are known as capsomeres. The arrangement of capsomeres determines the shape of the virus particle. Among viruses there are three different types of symmetry : (i) cubic, (ii) helical and (iii) complex.

   The capsid protects the nucleic acid against the action of nuclease enzyme. Some capsid-proteins help in binding the virion to the surface of the host cell, while a few act as enzymes to dissolve the surface layer of host cell for the penetration of virion nucleic acid.

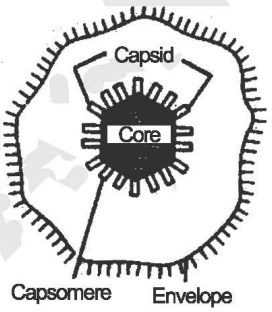


Fig. 2 : A generalized diagram of a virion.

2. **Nucleic Acid or Genetic Material :** A virus may contain DNA or RNA which may be single stranded or double stranded, linear or circular.

   Following types of nucleic acids are found in viruses :

   (i) Single-stranded DNA (ssDNA) viruses : Most of the animal viruses like *Herpes* virus, Hepatitis B virus chicken pox viruses and also cancer causing viruses.

   (ii) Double-stranded DNA (dsDNA) viruses : Bacteriophages.

   (iii) Single-stranded RNA (ssRNA) viruses : All plant viruses and some animal viruses causing polio, mumps, measles, cold and influenza, corona virus.

   (iv) Double stranded RNA (dsRNA) viruses : TMV, influenza virus or double stranded reoviruses and some plant viruses.

   Some animal viruses (tumour viruses and reoviruses) have RNA together with DNA as hereditary material. The transfer of message is :

   $$RNA \rightarrow DNA \rightarrow RNA \rightarrow Protein$$

   The nucleic acid has genetic information for the synthesis of variety of proteins that either become components of capsids of new virions or redirect the host cell metabolism to synthesise viral proteins.

Non-living Characters of Virsues

(i) Viruses can be crystallised like other inorganic and organic compounds and can be stored like other non-living substances.

(ii) Virsues do not show any metabolic activities because they do not have their own enzyme system.

(iii) Virsues do not show any growth.

(iv) They do not show any response to external environment.

(v) They do not multiply outside the host cells.

Living Characters of Viruses

(i) Viruses are living because they can multiply though within a living system.

(ii) Their genetic material possesses the property of recombination and inheritance.

(iii) Their genetic material undergoes mutations.

Because of their ability to utilise host's enzyme system for the manufacture of their own proteins and enzymes, and their ability to transfer genetic material to offsprings indicates that viruses share some living characters.

Q.2. Define bacteria. Also discuss various modes of reproduction found in bacteria.

Ans.

Bacteria

Bacteria are unicellular microorganisms. They are very diverse, have a variety of shapes, and have the ability to live in just about any environment, including in and on human body. Not all bacteria cause infections. Those that can cause disease are called pathogenic bacteria. Your body can be more prone to bacterial infections when your immune system is compromised by a virus. The disease state caused by a virus enables normally harmless bacteria to become pathogenic. Antibiotics are used to treat bacterial infections. Some strains of bacteria have become resistant to antibiotics, making them difficult to treat. This can happen naturally, but also happens because of the overuse of antibiotics.

The common diseases caused by bacteria include the following : (i) strep throat; (ii) urinary tract infection (UTI); (iii) bacterial gastroenteritis including *Salmonella* food poisoning and *E. coli* infection; (iv) bacterial meningitis; (v) lyme disease; (vi) tuberculosis; (vii) gonorrhea; (viii) cellulitis.

Reproduction in Bacteria

Bacteria reproduce by three methods : Binary fission, sporulation and sexual reproduction.

Binary Fission

Under favourable conditions, binary fission is the common method of multiplication in bacteria. Mature bacterium divides into two identical daughter cells by amitosis. During this process bacterial chromosome (nucleoid) replicates to produce two nucleoids while attached to the mesosome. A new mesosome develops which attaches to the daughter chromosome. Membrane synthesis occurs in between the two mesosomes, divides the cytoplasm and produces two daughter protoplasts each with its own nucleoid.

Under favourable conditons, many bacteria divide after every twenty minutes. In just 24

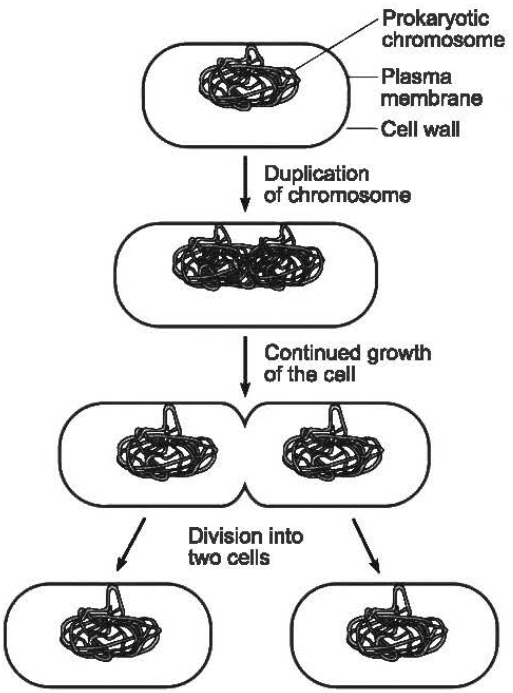


Fig. 1 : Mechanism of binary fission in a bacterium

hours, a single bacterium dividing after every 20 minutes will produce 4.7×10^{21} descendents with a weight of 2,000 tonnes. But, practically, it does not happen, so as either the process slows down or comes to an end due to the exhaustion of water and food and accumulation of poisonous substances.

Sporulation

Bacteria produce several types of spores for reproduction. **Endospores** are thick-walled, high esistant spores produced within the bacterial cells in adverse environment or in presence of harmful waste products. These are formed by Gram positive, rod-shaped bacteria belonging to two genera, *Bacillus* and *Clostridium.* At the time of endospore formation, the protoplast containing nuclear body stores food and undergoes dehydration to form a thick wall around it.

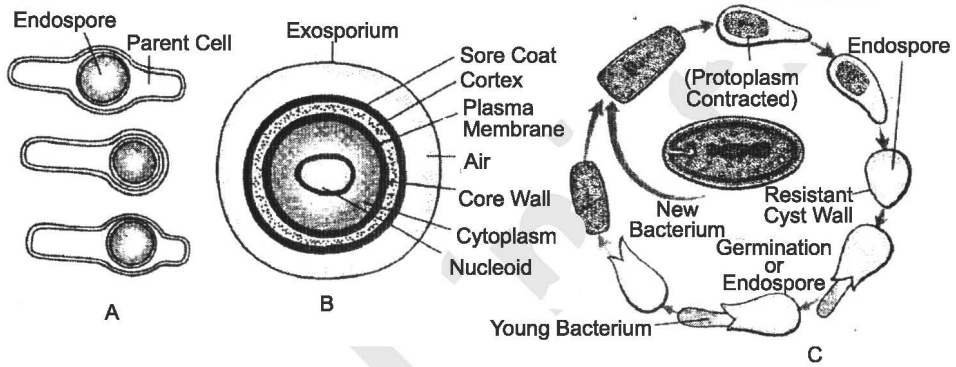


Fig. 2 : Endospores : A. Types of endospores according to their position in parent cell; B. Structure of endospore in section; C. Formation and germination of endospore.

Endospore is metabolically inert and can survive unsuitable conditions of temperature and drought. When dry, the endospores can withstand temperature at 100°C for several hours. Under favourable conditions, spores imbibe water and become metabolically active again.

Only two of the pathogenic bacteria, viz. tetanus-causing and anthrax-causing produce endospores. Fortunately, none other pathogenic bacteria produce spores.

Other Types of Spores : Formation of sporangiospores, arthospores (oidia), conidia, gonidia (swarm cells) and cysts are some other types of spores produced in some bacteria.

Sexual Reproduction (Parasexuality)

Sexualilty in bacteria was demonstrated by **Laderberg** and **Tatum** (1946) for which they were awarded the Nobel Prize. Typical sexual reproduction is absent in bacteria because these do not form gametes. However, exchange of genetic material does take place. Therefore, it is called **genetic recombination** or parasexuality. It occurs by three methods :

1. **Conjugation :** It is primitive form of sexual reproduction discovered in *Escherichia coli* by **Lederberg** and **Coli**. Bacteria that exhibit conjugation are **dimorphic,** *i.e.,* they have two types of cells : male (F^+) or donor and female (F^-) or recipient. The male (donor) cells possess **pili** and **F** or **fertility factor** on its plasmid. The female (recipient) cells does not have either of these structures.

(i) Sexduction : The donor cell gets attached to the recipient cell with the help of pili. In the region of contact, a pilus grows in size and forms cytoplasmic bridge or conjugation tube. The plasmid of donor cell with fertility factor undergoes replication. At this stage, a conjugation tube is formed through which a copy of fertility factor is transferred to the recipient cell, which is also converted into F^+ cell. This phenomenon is called **sexduction.**

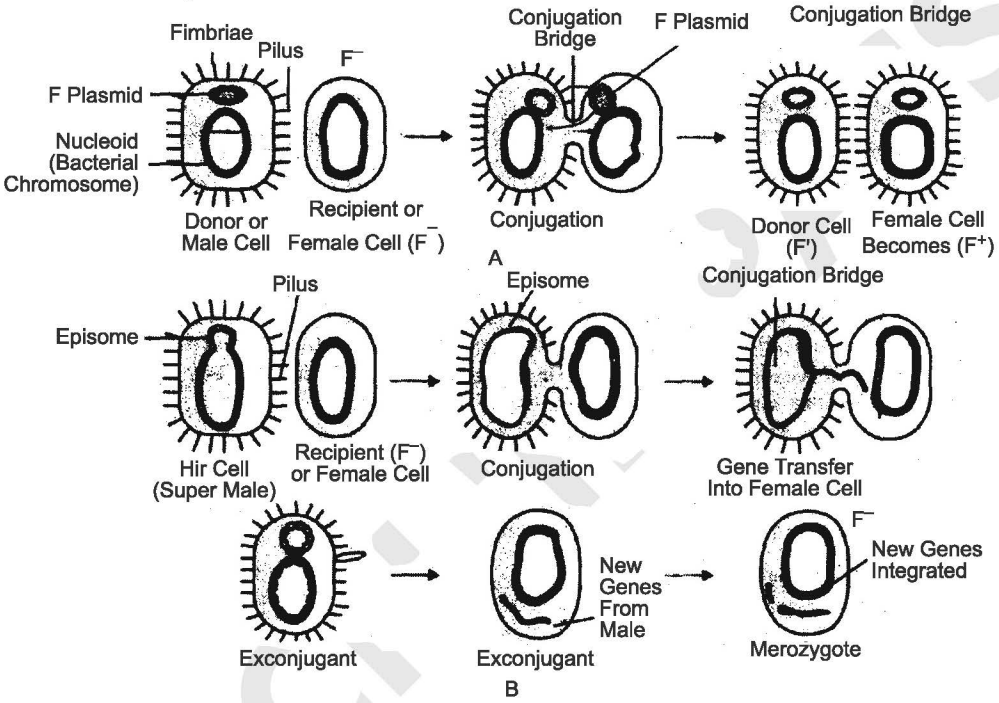


Fig. 3 : Conjugation in bacteria : A. Sexduction (Sterile male method); B. Transfer of chrmosomal genes (Fertile male method.

(ii) Conjugation : Sometimes, the plasmid of male cell gets attached with the bacterial chromosome to from **episome.** Such a male cell with episome is called *Hfr* (high frequency of recombination). The episome causes a break in the bacterial chromosome so that some of the genes of the male cell are transfer to the female cell along with the plasmid. The transferred genes recombine with the corresponding genes in the female cell.

2. **Transduction :** Transduction is the process in which the genetic material (a portion of a host DNA) of one bacterium is transferred to another bacterium through the agency of temperate bacteriophage (i.e., bacterial virus). This process was discovered by **Zinder** and **Lederberg** (1952) in bacteria, *Salmonella typhimurium.*
3. **Transformation :** This phenomenon was discovered by **Griffith** (1928) in *Diplococcus pneumoniae* (pneumonia causing bacteria). He obserevd that bacteria of nonvirulent strain developed characteristics of the virulent strain, when injected in mice along with heat-killed lysogenic bacteria of virulent strain. **Avery et al** (1944) found that virulent strains are developed due to transfer of DNA segments from the dead cells to the living cells.

Q.3. What do you know about Fungi? Describe various modes of asexual reproduction in fungi.

Ans.

Fungi

There are millions of different fungal species on Earth. Fungi can be found just about everywhere in the environment, including indoors, outdoors, and on human skin. They cause infection when they overgrow. Fungi cells contain a nucleus and other components protected by a membrane and a thick cell wall. Their structure can make them harder to kill. Some new strains of fungal infections are proving to be especially common such as *Candida aurus*, and have prompted more research into fungal infections.

Following are some of the fungal human diseases : (i) vaginal yeast infection, (ii) thrush; (iii) ringworm; (iv) athlete's foot; (v) jock itch; (vi) fungal nail infection (onychomycosis).

Reproduction in Fungi : Fungi have three types of reproduction : Vegetative, asexual and sexual.

Asexual Reproduction

It occurs through the formation of **spores** or **propagules**. These may be formed asexually or sexually. These are single-celled propagules which are separated from the body and disseminated.

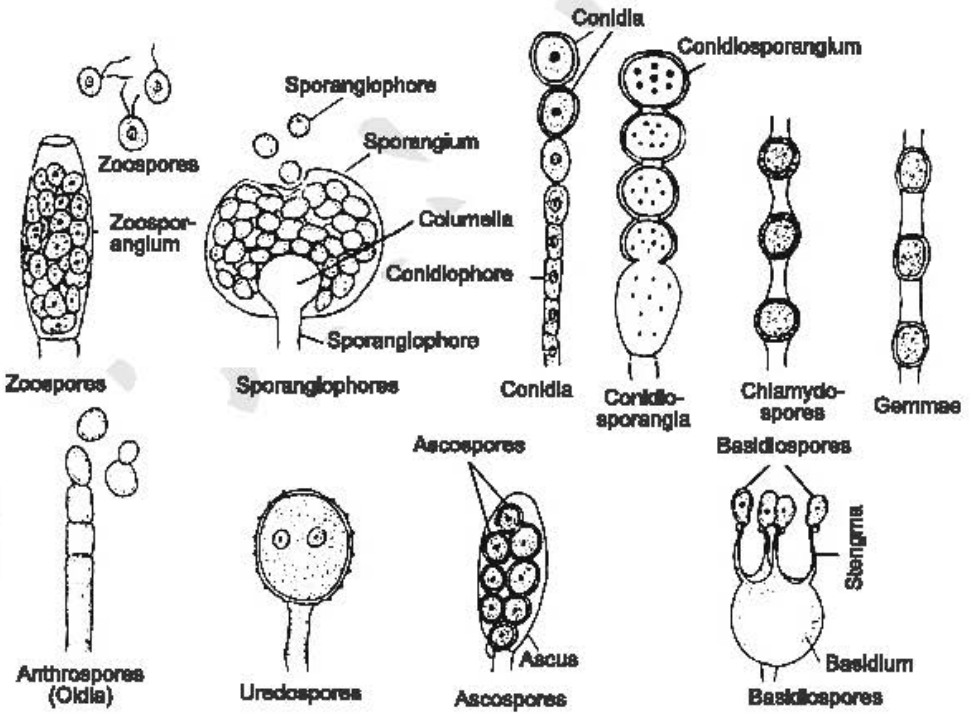


Fig. : Types of fungi spores.

The spores may be motile or non-motile, thin-walled or thick-walled. Spores produced sexually, *i.e.*, after meiosis are called meiospores, *e.g.*, ascospores and basidiospores. The spores produced asexually are called mitospores. These are genetically similar to the parent. They are mostly formed in large number.

Asexual reproduction in fungi usually takes place by following types of spores :

1. **By Zoospores** : These are thin-walled uninculeate structures formed in zoosporangium. Since these possess flagella and swim in water, these are called **zoospores.** These may be with one flagellum as in *Synchitrium* or with two flagella as in *Pythium.* Zoospores may be of two types :
   (i) **Pear-shaped** or **pyriform** with two flagella placed at anterior end, and
   (ii) **Kidney-shaped** or **bean-shaped** with two oppositely directed flagella inserted laterally in furrow.

   Zoospores germinate to give rise to a new mycelium. Reproduction by zoospores occurs in Phycomycetes (*Albugo, Phytophthora and Saprolegnia*).
2. **Aplanospores or Sporangiospores :** These are thin-walled and nonmotile spores produced in a sporangium and dispersed by wind. On liberation, they germinate into new mycelia. The hypha that bears terminal sporangium is called sporangiophore, *e.g., Mucor* and *Rhizopus.*
3. **Oidia :** In some fungi, like ***Coprinus*** and ***Mucor***, the hyphae break up into numerous fragments known as **oidia** or **arthrospores.** The oidia germinate on suitable substratum and give rise to new mycelia.
4. **Chlamydospores :** In some fungi like smuts, the vegetative cells change into small thick-walled rounded spores known as **chlamydospores.** These may be terminal or intercalary in position. They remain viable for several years. On the approach of favourable conditions, they germinate into same type of mycelia.
5. **Conidia :** These are nonmotile, thin-walled exogenous spores produced singly as in *Pythium* or in chains on special hyphal branches, called **conidiosphores.** They are arranged in **acropetal succession** (the topmost youngest) as in *Penicillium* or in **basipetal succession** (the upper one oldest) as in *Aspergillus.*
6. **Ascospores :** These are nonmotile meiospores produced in special sacs, called **asci** (singular: ascus). An ascus contains 8 ascospores because meiosis is accompanied by mitosis. Ascospore formation is a characteristic of Ascomycetes (*Saccharomyces* or yeast).
7. **Basidispores :** These are nonmotile meisopores which are formed exogenously or short outgrowths, the **sterigmata,** on a club-shaped structure called **basidium.** These are formed in Basidiomycetes.
8. **Binucleate Spores :** These spores are meant for the multiplication of dikaryotic mycelium. Thus, they are dikaryotic, *e.g.,* **aecidisopores, uredospores** and **teleutospores.** These spore are found in Puccinia or wheat rust.

Dispersal of Spores

Zoospores produced by aquatic fungi are motile and possess flagella for swimming. Most terrestrial fungi produce nonmotile and minute spores in large numbers. These are easily dispersed by wind. On falling over a suitable medium, they germinate and the developing hyphae grow to produce new mycelia.

Q.4. What are endoparasite? Enumerate characteristic features of *Taenia solium.*

Ans. Parasites may be grouped into ectoparasites and endoparasites. Parasites who live outside the host are called **ectoparasites** whereas who live inside the host are called **endoparasites.**

Endoparasites are of two forms : intercellular and intracellular parasites. Intercellular parasites are those that inhabit the spaces of the body of the host. Intercellular parasites are endoparasites that live within the cell of the host.

Characteristics Features of *Taenia solium*

Taenia is a digenetic cestode. It is an endoparasite in the small intestine of man. It is attached to the intestinal wall by hooks and suckers present on scolex. It is commonly found in pork-eating persons.

Taenia has a long, white, ribbon-shaped body as long as four metres. It is dorsoventrally flattened and is differentiated into three regions :

1. **Scolex :** It is knob-like anterior end of the body, of the size of **pin's head.** It bears four cup-shaped **suckers** by which scolex is attached to host's intestinal wall. Scolex bears a conical **rostellum,** surrounded by two rows of curved chitinous **hooks.** The rostellar hooks get inserted into the intestinal wall of host.
2. **Neck :** It is the region of proliferation and adds new segments to the body throughout the life of tapeworms.
3. **Strobila :** The rest of the body is called **strobila.** It consists of about 850-1100 proglottids. These are of three types :
   (i) **Immature proglottids** are undifferentiated segments just behind the neck. The reproductive organs are either absent or in different stages of development.
   (ii) **Mature proglottids** or **reproductive proglottids** have fully mature male and female reproductive organs.

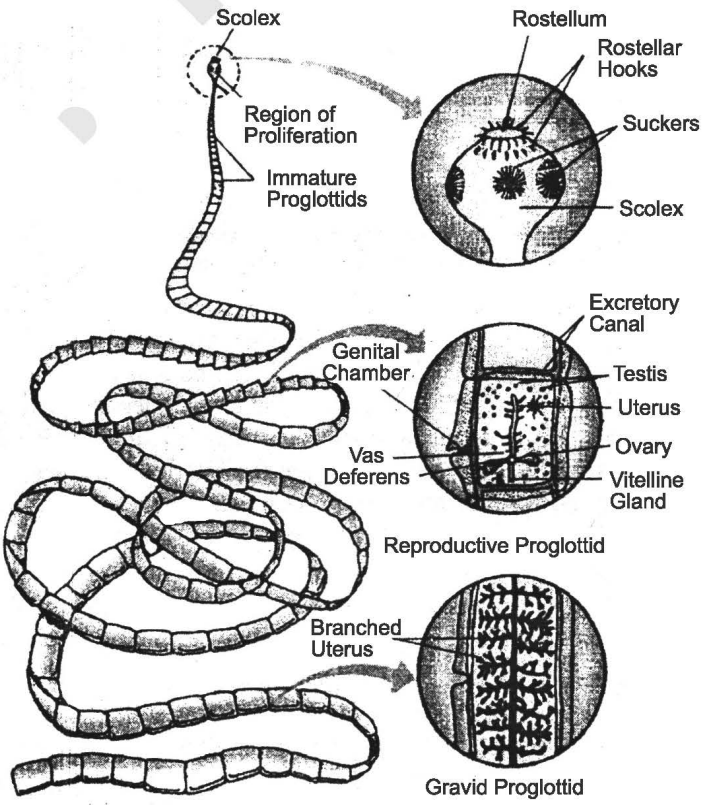


Fig. : *Taenia solium* with different types of proglottids

(iii) **Gravid proglottids** have large and highly branched uterus filled with embryonated eggs. These segments are present in the posterior part of body and are longer than broad. All other reproductive organs (except uterus) get degenerated.

The posterior-most gravid segments deatch from the strobila and pass out in the host faeces. This is called **apolysis.**

Taenia is without mouth, alimentary canal and anus. It absorbs digested food from host intestine through body surface.

The circulatory and respiratory systems are also absent. Respiration is anaerobic.

Fertilisation is internal. Eggs are capsulated. The embryonated eggs containing **hexacanth lavae** come out of primary host (Man) in host's faeces enclosed in gravid proglottids. The secondary host (Pig) is infected when it feeds on infected human faeces. Embryo develops into **bladder worm** or **cysticerus larva** in the pig muscles. Man gets infected by eating undercooked pork, containing cystricerci.

Economic Importance : Tapeworm causes **taeniasis** characterised by abdominal pain, indigestion, nausea, vomiting, etc. Its cysticercus larva causes **cysticercosis.**

Q.5. Describe the structure, polymorphism and life cycle of *Trypanosoma*. What are the diseases caused by its different species?

Ans.

Trypanosoma

The trypanosomes are digenetic which spend a part of their life-cycle in the blood of vertebrates and a part within the alimentary canal of blood-sucking invertebrates like leeches, ticks and insects. Common trypanosome parasites of man are *T. rhodesiense* and *T. gambiense* causative of sleeping sickness, widely spread in Africa.

Structure

Trypanosoma is unicellular and microscopic having elongated and fusiform body. The anterior end is pointed and posterior one blunt. It measures 10 to 40μ in length and 2.5 to 10μ in width.

Its body is enclosed in a strong, tough but elastic **pellicle,** which is followed by a layer of **ectoplasm** and the inner mass of **endoplasm.** In the centre of the body is a large vesicular **nucleus** which contains a large **karyosome.** Behind the nucleus and near the posterior end of the body is present a spherical, rod-shaped or disc-shaped **parabasal body** or **kinetoplast** and a small **blepheroplast** immediately in front of it. The two are connected by a delicate **rhizoplast.** The **flagellum** arises from the blepheroplast at the posterior end and extends forward along the outer margin of the body, and is connected to it by the **undulating membrane.** A small portion of the flagellum extends freely beyond the anterior end. Elongated contractile fibres, are **myonemes,** are visible in the periplast in large forms. The **contractile vacuoles** are absent but **volutin granules** in the form of green refractile bodies are found scattered in the endoplasm of medulla.

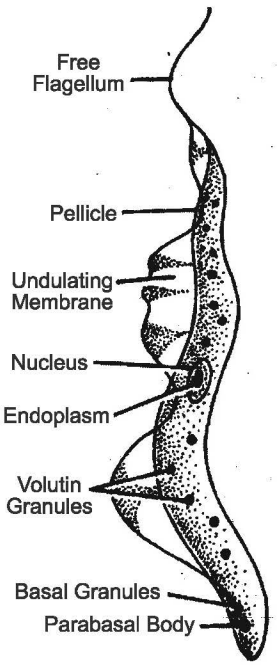


Fig. 1 : *Trypanosoma*

Polymorphism in *Trypanosoma*

Trypanosoma is **polymorphic** occuring in four morphologically different forms :

1. **Leptomonad stage** with elongated body having a free flagellum and an anteriorly packed kinetoplast.
2. **Crithidial stage** with flagellum and the kinetoplast situated in the middle of body but in front of nucleus.
3. **Trypanosoma stage** with kinetoplast situated far behind the nucleus near the posterior end.
4. **Lieshmanial state** without flagellum and with anteriorly placed kinetoplast.

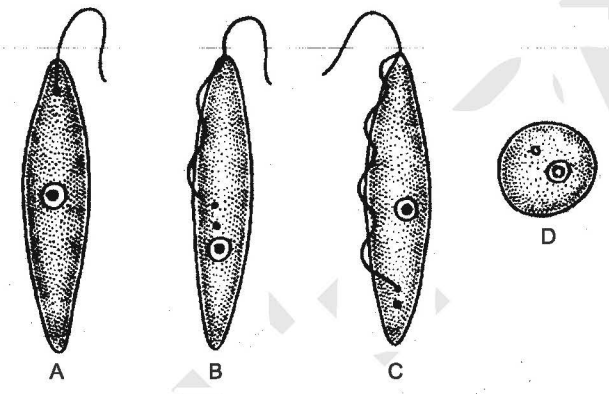


Fig. 2 : Polymorphism in *Trypanosoma* : A. Leptomonad stage, B. Crithidial Stage, C. Trypanosoma stage, D. Leishmanial Stage

Life Cycle of *Trypanosoma*

Sexual reproduction is unknown in *T. gambiense.* The asexual reproduction takes place by longitudinal **binary fission.**

The life-history of *T. gambiense* and *T. rhodesiense* is **digenetic** and is completed in two hosts.

The secondary part of the life-cycle is spent in the blood or cerebrospinal fluid of man or some game animal. This represents the **definitive** or **vertebrate host.** The rest of the life-cycle is completed in the alimentary of Tse-tse-fly, *Glossina palpalis* or *G. tachinoides.* This is the **secondary host** or **intermediate host.**

1. Life-Cycle in Man

(i) Inoculation : The parasite is introduced into the blood stream of may by the bite of Tse-tse fly. To take its bloody meal, the fly makes a local cutaneous puncture through which is meta-cyclic form of *Trypanosoma* enters the blood-stream.

(ii) Multiplication : In human blood, the metacyclic forms transform into **long slender forms.** These swim by the beating of flagellum and the vibratile movement of the undulating membrane. These multiply by longitudinal binary fission.

(iii) Metamorphosis : When the absorption of glucose ceases due to antibodies produced in blood, trypanosomes stop multiplying. They new form short **stumpy** forms. These are devoid of flagellum During transformation to short stumpy forms, the

intermediate forms with somewhat shortened body and a free flagellum also appear. The stumpy forms do not feed and die if not sucked by the tse-tse fly along with the blood meal.

(iv) Relapse of Infection : Some of the long and slender forms which do not undergo transformation, survive and continue to multiply in blood leading to future relapses of the infection.

2. Life Cycle in Tse-tse Fly

(i) Transfer to the Tse-tse fly : The tse-tse fly sucks the trypanosomes along with its bloody meal. These remain unchanged in midgut for few hours.

(ii) Development in Midgut : The short-stump forms develop in the insect's mid-gut into long, slender forms and multiply by longitudinal binary fission.

(iii) Development in Salivary Glands : The long, slender forms make their way into the salivary glands via oesophagus and mouthparts of the insect. Here the parasites get attached and change into the **crithidial forms.** These multiply in the lumen of the salivery glands and transform into the slender trypanosome forms, known as **metacyclic forms.**

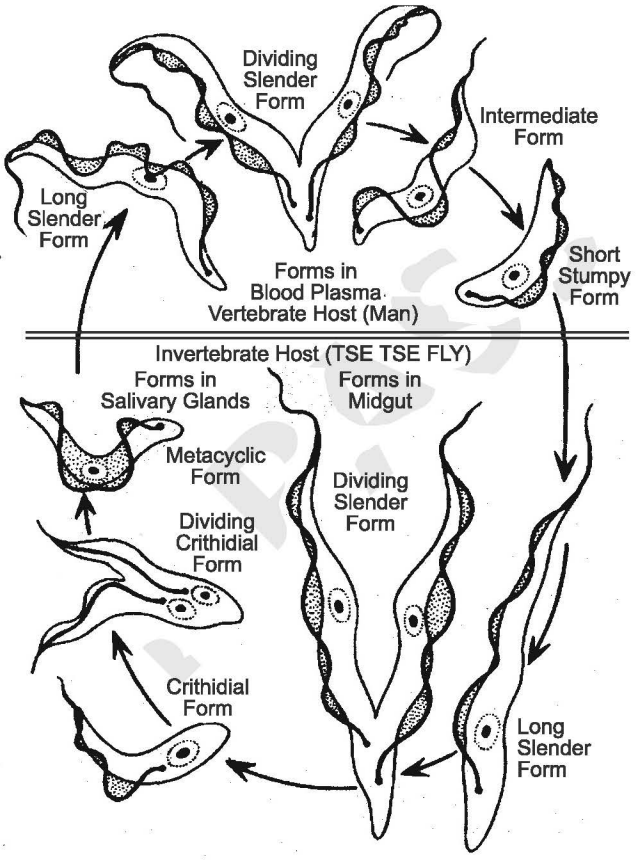


Fig. 3 : Life cycle of *Trypanosoma gambiense.*

Pathogenicity

Presence of trypanosomes in blood causes **gambien fever,** which persists only for few days. Later on, enlargement of lymph glands, anaemia and weakness occur. These pathological conditions are followed by the enlargement of spleen and liver. On entering the cerebrospinal fluid, these cause **sleeping sickness,** in which patient losses conciousness first at regular intervals and finally enters into a state of coma ending in death.

Diseases caused by *Trypanosoma* sp. parasite

Tryponosoma sp.	Vertebrate host	Vector	Diseases	Epidemiology
T. brucei brucei	Horses, pigs, cattle, rodents	*Glossina* sp.	Magana	Tropical Africa
T. brucei gambiense	Human, monkeys, dogs, pigs, etc.	*Glossina* sp.	Sleeping sikness	West Africa

T. brucei rhodesiens	Human, pigs	*Glossina* sp.	Sleeping sikness	East Africa
T. crusi	Human, domestic and wild animals	Reduvivid bugs *(Triatoma rhodnieus)*	Charge disease	South America
T. evansi	Horse, dogs	*Tabanus* sp.	Surve	India, Africa, Australia, South and Central America

Prevention of Disease

Direct attacks on tse-tse fly help in checking the spread of sleeping sickness. These include :

1. elimination of breeding grounds.
2. general cleanliness, and
3. protection against bite.

Q.6. Briefly describe the structure and life cycle of *Giardia*. And also discuss the role of this structure in the formation of cysts and their role in causing the disease.

Ans.

Giardia

Giardia is a genus of anaerobic flagellated protozoan parasites of the phylum metamonada that colonise and reproduce in small intestines of several vertebrates, causing giardiasis. Their life cycle alternates between a swimming trophozoite and an infective, resistant cyst. *Giardia* were first described by the Dutch microscpist **Antonie van Leeuwenehoek** in 1681. The genus is named after French zoologist **Alfred Mathieu Giard.**

Structure of *Giardia*

Giardia lamblia (also known as *G. intestinalis*) lives as a flagellate parasite in the small intestine of man. It causes a disease called giardiasis which causes digestive disturbances. Heavy infection interfere with normal absorption as the flagellates adhere to the mucosa. *Giardia* present both trophic and cystic forms (pear-shaped). The trophozoites measure 9-20 microns by 6-20 microns. The protoplasm of the oval body is clear. The body is flattened dorsoventrally. The dorsal surface is convex. In the body, there are two nuclei and four pairs of flagella arranged symetrically. 'The trophozoites transform to cysts and reproduction within the cyst occurs by binary fission. Cysts come out with stool. Infection occurs through contaminated food or drink.

Life cycle, *Giardia* cysts are the infective stage of *G. intestinalis.* As few as 10 cysts can cause infection. These cysts are ingested by consuming contaminated food or water, or fecalorally. They can survive outside the body for several months, and are also relatively resistant to chlorination, UV exposure and freezing. When cysts are ingested, the low pH of the stomach acid produces excystation, in which the activated flagella breaks throught the cyst wall. This occurs in the small intestine, specifically the duodenum. Excystation releases trophozoites, with each cyst producing two trophozoites. Within the small intestine, the trophozoites reproduce asexually (longitudinal binary fission) and either float free or are attached to the mucosa of the lumen. Some trophozoites the encyst in the small intgestine. Encystation occurs

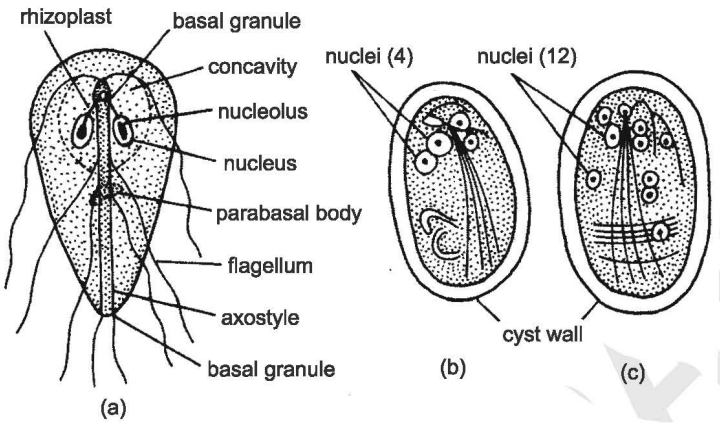


Fig. 1 : *Giardia lamblia* : (a) Dorsal view of Trophozoite, (b) Cyst with 4 nuclei, (c) with 12 nuclei.

most likely as as result of exposure to bile salts and fatty acids and a more alkaline environment. Both cysts and trophozoites are then passed in the feces, and are infectious immediately or shortly afterward. Person-to-person transmission is possible. Animals can also be infected with Giardia.

Pathogenesis

Giardia lives in the **intestines** of infected humans or other animals, individuals of which become **infected** by ingesting or coming into contact with contaminated foods, soil, or water tainted by the feces of an infected carrier.

The symptoms of giardiasis, which may begin to appear 2 days after infection, include violent **diarrhoea** excess gas, stomach or abdominal cramps, upset stomach and nausea. Resulting dehydration and nutritional loss may need immediate treatment. A typical infection can be slight, resolve without treatment, and last between 2-6 weeks, although it can sometimes last longer and/or be more severe. Coexistence with the parasite is possible (symptoms fade), but an infected individual can remain a carrier and transmit it to others. Medication containing **tinidazole** or **metronidazole** decreases symptoms and time to resolution. **Albendazole** is also used and has an **anthelmintic** (anti-worm) property as well, ideal for certain compounded issues when a general vermicidal agent is perferred *Giardia* causes a disease called **giardiasis** which cause the villi of the small intestine to atrophy and flatten, resulting in malabsorption in the intestine. Lactose intolerance can persist after the eradication of *Giardia* from the digestive tract.

Symptoms of Giardiasis

Some people (~ 10%) can carry *Giardia* parasites without experiencing any symptoms. Symptoms of giardiasis generally show up one or two weeks after exposure. Common symptoms include the following : fatigue, nauseas, diarrhea or greasy stols, loss of appetite, vomiting, bloating and abdominal cramps, weight loss, excessive gas, headaches, abdominal pain.

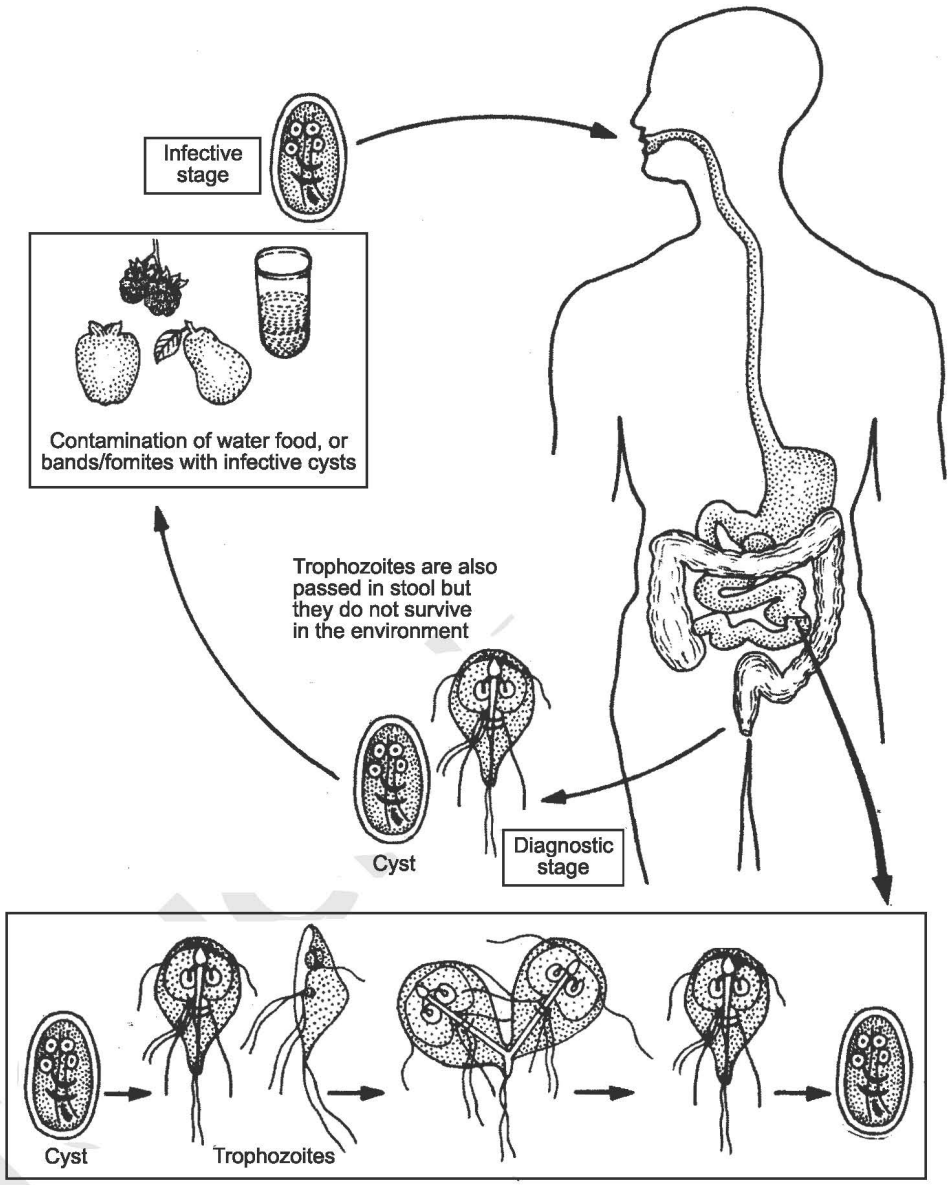


Fig. 2 : Life cycle of *Giardia intestinalis*.

Disease Control

Giardia germs are generally present in stool, soil or other contaminated places, since anything that gets contaminated by stool or due to work in the field, can spread the germs. Good hygiene is the best measure for disease control.

This may involve following precautions/practices—(i) wash your hands frequently with soap and water, especially when you are likely to spread the germs (*e.g.* after working in the field); (ii) keep away from people and places, infected with diarrhea until the diarrhoea has stopped;

(iii) clean, sanitize, or disinfect all places/objects, which may spread the disease; (iv) keep away from water that may be contaminated; when at the pool or lake, do not swallow the water, and do not allow children to swim if sick with diarrhoea; (v) minimize contact with stool of all animals, especially young animals and do not touch your face or mouth after being near the animals; (vi) drink either bottled water or water that was boiled for a minute and then stored, or else use water filter.

Q.7. Describe distribution morphology and life cycle of *Wuchereria bancrofti.*

Ans. ***Wuchereria bancrofti* (*Filaria bancrofti*)**

Wuchereria is a digenetic nematode, which lives in the lymphatic vessels and lymph nodes of man.

Distribution

It is found in the tropical and subtropical countries like India, West Indies, Southern China, Japan, Pacific Island, West and Central Africa and South America. In India it is largely confined to the coastal areas and the banks of big rivers.

Morphology

The adult worm is a long hair-like transparent nematode often creamy-while in colour. The female is usually larger than the male, measuring 8-13 cms in length and 0.2 to 0.3 mm in thickness. The male is only 2.5 to 4 cm long and 1 cm broad. Both male and female are filiform with tapering ends. The tail end of male is curved and carries two spicules. The male and female remain coiled together and are separated with difficulty. The female is ovoviviparous or lays eggs, which contain well-developed embroys. The embroys are known as **microfilariae.**

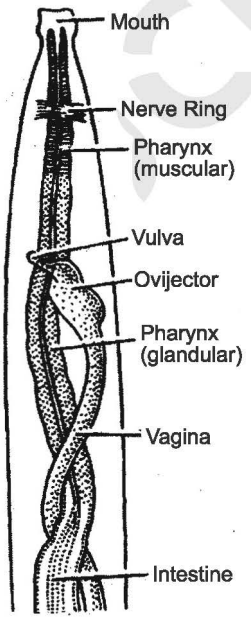


Fig. 1 : *Wuchereria bancrofti,*

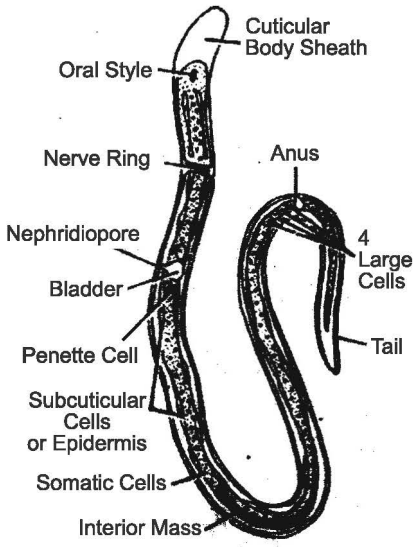


Fig. 2 : Larval stage of *Wuchereria*

Life Cycle

The life-cycle of *Filaria* is completed in two hosts, the primary host is man and secondary host is mosquito.

1. **Life Cycle in Man :** The adult worm produces microfilariae which are active larval forms and can move both with and against the blood stream. Each of them is a colourless, transparent and cylindrical animal with blunt head and rather pointed tail. It is about 290 μ in length and 6-7 μ in breadth. The microfilaria is enclosed in a hyaline sheath which projects beyond the ends of the embryo so that it can move inside the sheath. The elongated body contains granules arranged along the central axis from head to tail. At places, the granular layer is interrupted by nerve ring, anterior V-shaped spot, posterior V-shaped spot, a few genital cells and central body or inner cell mass.

   The microfilariae are unable to develop further in the human body unless they are taken up by the intermediate host. Microfilariae, as soon as they are discharged, enter the blood streams and at night move into the peripheral vessels. If not sucked up by the mosquito they die with in 70 days.

2. **Life Cycle in Mosquito :** The microfilariae are sucked up by the mosquito (*Culex fatigans*) along with bloody meal. These are collected in the anterior part of the stomach where these cast off the hyaline sheath, penetrate the gut wall and within an hour or two migrate to the thoracic muscles. Inside, these increase in size and within next two days the slender snake-like organisms, change to thick short sausage-shaped forms with short spiny tail. These measure 121-150 μ in length and 10-17 μ in breadth and represent **first stage larvae.** These possess rudimentary alimentary canal.

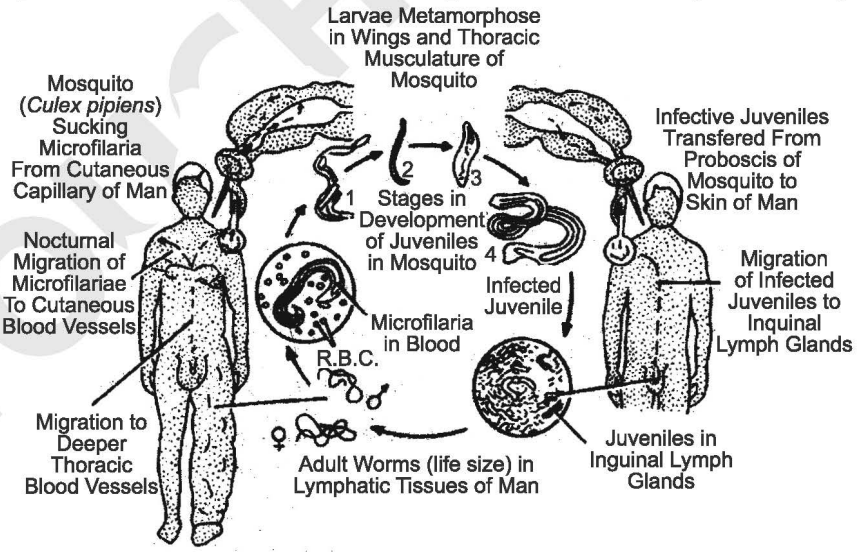


Fig. 3 : Life cycle to *Wuchereria bancrofti*

Within 3-7 days the larvae grow rapidly, moult once or twice and attain a size of about 225-330 μ. These are second stage larvae. On 10 or 11th day the metamorphosis is complete. The tail atrophies and the digestive system, body cavity and genital organs

are developed fully. These constitute third stage larvae measuring 1500-2000 μ in length and 18 to 23 μ in breadth. These now enter the proboscis sheath of the mosquito and represent the **infective stage.**

3. **Infection in Man :** When the infected mosquito bites a man the third stage larvae are deposited usually in pairs on the skin near the wound. The larvae are attached by the warmth of the skin and they enter the body either through the puncture or penetrate through the skin. From here they reach the lymphatic channels and settle down at some spot to metamorphose into the adults. Within five to eighteen months the adults become sexually mature and start new generation of microfilariae.

Pathogenicity and Clinical Symptoms

The pathogenic effect is produced by the adult worm living or dead. The living adult causes mechanical irritation and deposits metabolites. As a result the lymph vessels get obstructed. The dead worms also block the lymph vessels. All these initiate proliferation of endothelial cells of lymph vessels leading to inflammatory thickening of the wall of lymhatic vessels.

As a result periodic attacks of fever occur and the tissues surrounding the lymph nodes and other organs of reticuloendothelial system such as liver, spleen, vulva, leggs and groins become greatly enlarged producing tumour-like solildity. This condition is known as **elephantiasis.**

Treatment

The drugs used in elephantiasis are divided into three categories depending upon their effect :

(i) On microfilariae : Diethyl carbamazine (Hetrazan).

(ii) On adult worms : Mel. W, arsenical preparation.

(iii) On infective larvae and immature adult : Para-melamminyl phenylstibonate (Msb).

❑

MODEL PAPER

Cytology, Genetics and Infectious Diseases

B.Sc.-I (SEM-I) [M.M. : 75

Note : Attempt all the sections as per instructions.

Section-A : Very Short Answer Type Questions

Instruction : Attempt all **FIVE** questions. Each question carries **3 Marks**. Very Short Answer is required, not exceeding 75 words.

1. Define plasmodesmata.
2. Write about 'sliding filament model' of muscle contraction.
3. Describe the small nucleolar RNAs.
4. Write a short note on bim genes.
5. Who discovered DNA fingerprinting technique? Write its uses.

Section-B : Short Answer Type Questions

Instruction : Attempt all **TWO** questions out of the following 3 questions. Each question carries **7.5 Marks**. Short Answer is required not exceeding 200 words.

6. What are the exceptions of cell theory?

Or How are intermediate filaments linked with genetic diseases?

7. Describe the structure of the Nuclear Pore Complex (NPC) with the help of a suitable diagram.

Or Write a note on checkpoint of cell cycle.

8. What do understand by Pangenes and acquired characters?

Or Describe the theories of multiple allelism.

Section-C : Long Answer Type Questions

Instruction : Attempt all **THREE** questions out of the following 5 questions. Each question carries **15 Marks**. Answer is required in detail, between 500-800 words.

9. Give a details account of fluid mosaic model of plasma membrane.

Or Write shorte note on EMP pathway.

10. Describe the structure, chemical composition and functions of chromosome.

Or Describe cell Signalling and its significance. Discuss various modes of cell signalling or cell communication.

11. Describe Mendels' laws of inheritance with the help of suitable example.

Or Write a detailed note on male sterility in plants.

12. Name a few of the common chromosomal abnormalilties in man. Describe their chromosomal basis and their phenotypic characters.

Or Describe distribution morphology and life cycle of *Wuchereria bancrofti.*

13. What are endoparasite? Enumerate characteristic features of *Taenia solium.*

Or Describe the Criss-cross inheritance with the help of diagrams.

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