GOVERNMENT COLLEGE (AUTONOMOUS)
RAJAMAHENDRAVARAM

DEPARTMENT OF MICROBIOLOGY

QUESTION BANK FOR

CELL BIOLOGY AND GENETICS (C-8)

(III SEM - B.Sc Honours-Microbiology)

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II B.Sc Microbiology Honours -III Semester

Course-8 (CELL BIOLOGY AND GENETICS)

Question Bank-2024-25

Essay type questions (Select any Two from each Unit for Internal Choice)

Unit -I

Q.No	Questions	Marks	BL	CO	PO
1.	State Cell theory. Explain detailed structure of Chloroplast	2+6	1&2	1	
2.	Discuss detailed structure and functions of Mitochondria	8	2	1	
3.	Illastrate cell cycle and it s regulation	8	3	1	

Unit -II

Q.No	Questions	Marks	BL	CO	PO
1.	Review structure and functions of Cell membrane	8	6	2	
2.	Discuss about structure and functions of nuclear membrane	8	3	2	
3.	Define and describe about Oncogenes and suppressor genes.	8	1	2	

Unit -III

Q.No	Questions	Marks	BL	CO	PO
1.	Generalise different Intracellular signal transduction pathways.	8	3	3	
2.	Explain about Programmed cell death	8	2	3	
3.	State structure and importance of Lampbrash and polytene chromosomes	8	2	3	

Unit -IV

Q.No	Questions	Marks	BL	CO	PO
1.	Compare and evaluate Mendal's Law of segregation and independent assortment	8	4&5	4	
2.	Define and distinguish between Incomplete dominance and co-dominance.	8	1&5	4	
3.	Define Multiple alleles, Lethal alleles, Epistasis and Pleiotropy.	4x2=8	1	4	

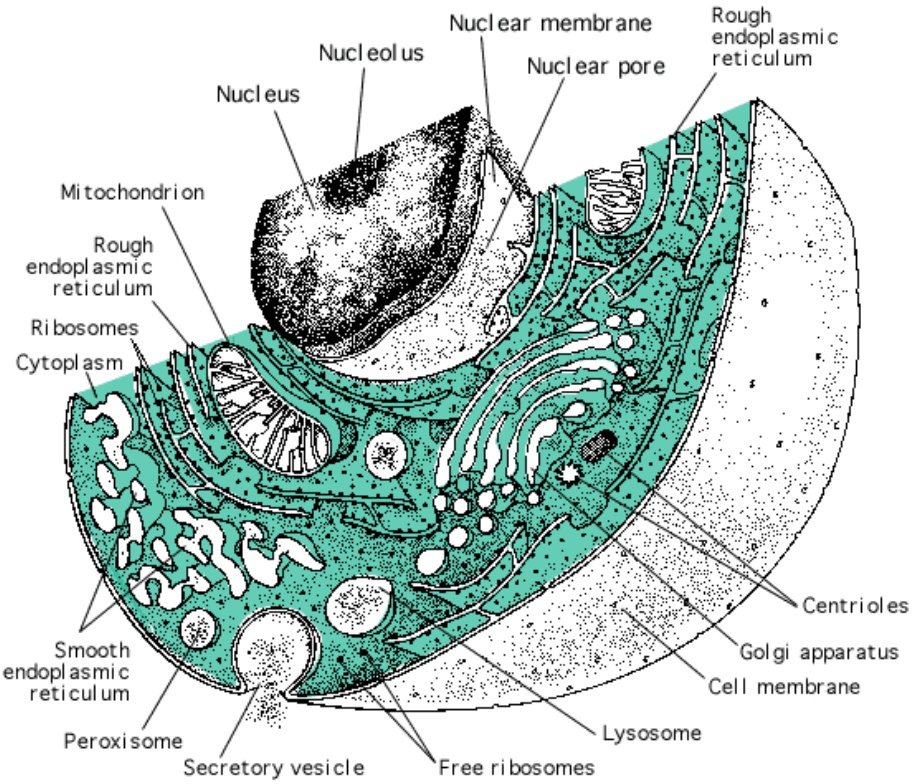
Unit -V

Q.No	Questions	Marks	BL	CO	PO
1.	Define crossing over. Interpret molecular mechanism of Crossing over	8	1&3	5	
2.	Evaluate the role of Natural selection and its role in Genetic drift and genetic shift	8	4		
3.	Write a short note on A. Sex linked inheritance B. extra chromosomal Inheritance	4+4	2	5	

Q.NO. 1. State Cell theory. Explain detailed structure of Chloroplast

Cell theory

In a 1665 publication called Micrographia, experimental scientist Robert Hooke coined the term "cell" for the box-like structures he observed when viewing cork tissue through a lens. In the 1670s, van Leeuwenhoek discovered bacteria and protozoa. Later advances in lenses, microscope construction, and staining techniques enabled other scientists to see some components inside cells.



Structure of an Animal Cell: The cell is the basic unit of life and the study of the cell led to the development of the cell theory.

By the late 1830s, botanist Matthias Schleiden and zoologist Theodor Schwann were studying tissues and proposed the unified cell theory. The unified cell theory states that: all living things are composed of one or more cells; the cell is the basic unit of life; and new cells arise from existing cells. Rudolf Virchow later made important contributions to this theory.
The generally accepted portions of the modern Cell Theory are as follows:

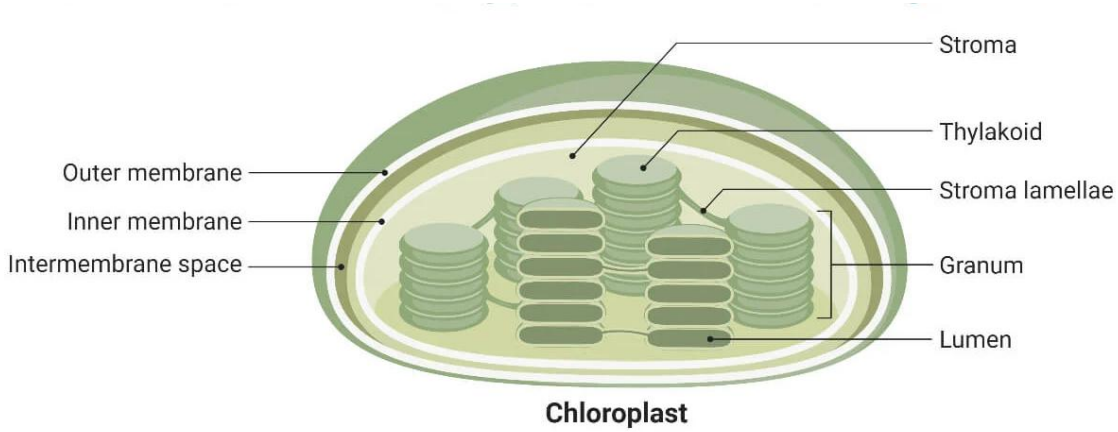
1. The cell is the fundamental unit of structure and function in living things.
2. All organisms are made up of one or more cells.
3. Cells arise from other cells through cellular division.
4. Cells carry genetic material passed to daughter cells during cellular division
5. All cells are essentially the same in chemical composition
6. Energy flow (metabolism and biochemistry) occurs within cells

Chloroplasts

The word *chloroplast is* derived from the Greek words *chloros*, which means green, and *plastes*, which means "the one who forms".

Chloroplasts are membrane-bound plastids that contain a network of membranes embedded into a liquid matrix and harbor the photosynthetic pigment called chlorophyll.

- It is this pigment that imparts a green color to plant parts and serves to capture light energy.
- Chloroplasts can be found in the cells of the mesophyll in plant leaves.
- There are usually 30-40 per mesophyll cells.



Chloroplast

TYPES OF PIGMENTS

Chlorophyll

Chlorophyll is a green pigment located within the chloroplast. More specifically, it is found in the thylakoid membranes. The chlorophyll consists of 75% of chlorophyll a and 25% of chlorophyll b. The chlorophyll absorbs energy from sunlight and the synthesis of food molecules in the chloroplast.

Carotenoids

Carotenoids are the pigments present in chlorophylls which are located in the thylakoid membrane. Pigments like yellow and orange are present in it. Carotenoids are related to vitamin A. They are important because they can absorb a certain wavelength of light that can not be absorbed by chlorophylls. Carotenoids are involved in a function known as photoprotection.

Xanthophylls

The carotenoids are carotenes and xanthophylls. Xanthophylls are present in the brown and green algae.

Phycobilin

Phycobilin is found only in red algae and *Cyanobacteria.* It has a relatively narrow distribution. Phycoerythrin and phycocyanin are other accessory pigments belonging to this family. Phycoerythrin makes red algae commonly red and phycocyanin causes the *Cyanobacteria* to appear blue-green.

STRUCTURE OF CHLOROPLAST

Chloroplasts found in higher plants are generally biconvex or planoconvex shaped.

In different plants, however, chloroplasts may have different shapes, varying from spheroid, filamentous saucer-shaped, discoid or ovoid-shaped.

They can be found in the cells of the mesophyll in plant leaves. They are vesicular and have a colorless center. The average size of the chloroplast is 4-6 Âµ in diameter and 1-3 Âµ in thickness.
The chloroplast has an inner and outer membrane with an empty intermediate space in between. Inside the chloroplast are stacks of thylakoids, called grana, as well as stroma, the dense fluid inside of the chloroplast. These thylakoids contain the chlorophyll that is necessary for the plant to go through photosynthesis. The space the chlorophyll fills is called the thylakoid space. A chloroplast thus has the following parts:

Envelope (Outer membrane):

It is a semi-porous membrane and is permeable to small molecules and ions, which diffuses easily. The outer membrane is not permeable to larger proteins.

Intermembrane Space:

It is usually a thin inter-membrane space about 10-20 nanometers and it is present between the outer and the inner membrane of the chloroplast.

Inner membrane: The inner membrane of the chloroplast forms a border to the stroma. It regulates the passage of materials in and out of the chloroplast. In addition to regulation activity, fatty acids, lipids, and carotenoids are synthesized in the inner chloroplast membrane.

Stroma: Stroma is an alkaline, aqueous fluid that is protein-rich and is present within the inner membrane of the chloroplast. The space outside the thylakoid space is called the stroma. The chloroplast DNA chloroplast ribosomes and the thylakoid system, starch granules and many proteins are found floating around the stroma.

Thylakoid System:

The thylakoid system is suspended in the stroma. The thylakoid system is a collection of membranous sacs called thylakoids. The chlorophyll is found in the thylakoids and is the sight for the process of light reactions of photosynthesis to happen. The thylakoids are arranged in stacks known as grana. Each granum contains around 10-20 thylakoids.

SEMI AUTONOMOUS NATURE

Like the mitochondria, they are also known as semi-autonomous cell organelles as they have their DNA and complete machinery to synthesize some of the required proteins.

While some other proteins depend upon nuclear DNA and cytoplasmic ribosomes.

Chloroplast and mitochondria are the only two organelles having their DNA.

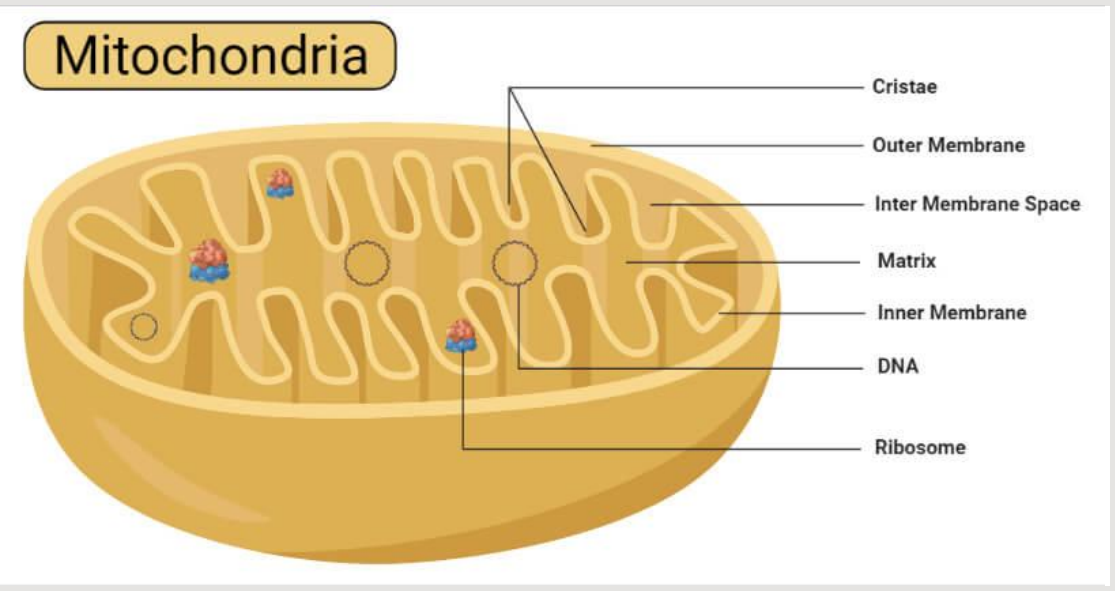
FUNCTIONS OF CHLOROPLAST

1. Chloroplasts are the sites for photosynthesis, which comprises a set of light-dependent and light-independent reactions to harness solar energy and convert it into chemical energy.
2. The components of chloroplast participate in several regulatory functions of the cell as well as in photorespiration.
3. Chloroplasts also provide diverse metabolic activities for plant cells, including the synthesis of fatty acids, membrane lipids, isoprenoids, tetrapyrroles, starch, and hormones.
4. Plants lack specialized immune cells—all plant cells participate in the plant response.
5. The chloroplasts with the **nucleus** and cell membrane and **ER** are the key organelles of pathogen defense.
6. Chloroplasts can serve as cellular sensors.

2.Q. Discuss detailed structure and functions of Mitochondria

Mitochondria are oxygen-consuming ribbon-shaped cellular organelles of immense importance floating free throughout the cell.
They are known as the "powerhouse of the cell" since these organelles supply all the necessary biological energy to the cell by oxidizing the substrates available.
The enzymatic oxidation of chemical compounds in the mitochondria releases energy.
Since mitochondria act as the power-houses, they are abundantly found on those sites where energy is earnestly required such as sperm tail, muscle cell, liver cell (up to 1600 mitochondria)microvilli, oocyte (more than 300,000 mitochondria), etc. Typically, there are about 2000 mitochondria per cell, representing around 25% of the cell volume.
In 1890, mitochondria were first described by Richard Altmann and he called them bioblasts. Benda (1835) named it Mitochondria.
It is the cell organelle which is a filamentous and granular structure.

- It is present in higher plants, animals, and some microorganisms.
- It is absent in bacteria but is found in algae, protozoa, and fungi.
- To carry out the energy metabolism, mitochondria have got the lipoprotein framework.
- It consists of the different enzymes and coenzymes.
- Mitochondria also consist of specific DNA and ribosomes.
- Ribosomes are involved in protein synthesis whereas specific DNA is involved in the cytoplasmic inheritance.
- Depending on the function of the cell, the mitochondria are distributed accordingly.
- The distribution of mitochondria varies according to species and cell type.
- But some cell may contain a large number of mitochondria as:
    - 50,000 in *Chaos chaos*
    - 140,000 to 150,000 in eggs of sea urchin
- The size of the mitochondria is 0.5 to 2.0 µm, so it cannot be seen clearly under the light microscope.
- It is also found sometimes the length reaches 7 µm.



Mitochondria Structure

STRUCTURE OF MITOCHONDRIA

Mitochondria are mobile, plastic organelles that have a double-membrane structure. It ranges from 0.5 to 1.0 micrometer in diameter. It has four distinct domains: the outer membrane, the inner membrane, the intermembrane space, and the matrix.

- The organelle is enclosed by two membranes—a smooth outer membrane and a markedly folded or tubular inner mitochondrial membrane, which has a large surface and encloses the matrix space.
- The intermembrane space is located between the inner and outer membranes.
- The number and shape of the mitochondria, as well as the numbers of cristae they have, can differ widely from cell type to cell type.
- Tissues with intensive oxidative metabolism— e. g., heart muscle—have mitochondria with particularly large numbers of cristae.
- Even within one type of tissue, the shape of the mitochondria can vary depending on their functional status.
- Both mitochondrial membranes are very rich in proteins.

Mitochondria consists of a mitochondrial membrane and a mitochondrial chamber.

1.MITOCHONDRIAL MEMBRANE

It consists of two membranes. They are:

a. Outer membrane

- It is a smooth membrane.
- It is made up of 40% **lipids** and 60% **proteins**.
- Due to the presence of the pores or porins, it is permeable.

b. Inner membrane

- It is made up of 20% lipids and 80% proteins.
- It is the selectively permeable membrane.
- Since the membrane is folded inwards and there is the presence of cristae, it is said as the rough membrane.
- Cristae are the numerous and finger-like projections.
- Tennis racket-like particles are present in each crista.
- It was called the electron transport particles (ETP)by Parsons in 1963.
- In each mitochondrion about 104-105 particles are present.
- ATPase or ATP synthetase is the enzyme present in it.
- The stalk also consists of the coupling factors which connect the respiratory chain with the elementary particle.

2. MITOCHONDRIAL CHAMBERS

Two chambers are present in the mitochondria i.e Outer and inner chamber.

a. Outer chamber (peri-mitochondrion space)

- Between the outer and the inner mitochondrial membrane, a space is present in between them which is known as the peri-mitochondrion space.
- Few enzymes are also present in the fluid present in it.

b. Inner chamber

- It is present in the inner part of the inner membrane.
- A semi-fluid matrix is present in it which consists of:
    - Water

- Minerals
- Protein particles
- 70s ribosomes
- RNA
- Circular DNA
- Enzymes

Mitochondrial Matrix

- The mitochondrial matrix which is the liquid (colloidal) area encircled by the inner membrane, contains the soluble enzymes of the **Krebs cycle** which completely oxidize the acetyl-CoA to produce CO2, H2O and hydrogen ions. Hydrogen ions reduce the molecules of NAD and FAD, both of which pass on hydrogen ions to respiratory or electron transport chain where oxidative phosphorylation takes place to generate energy-rich ATP molecules.

Mitochondria also contain in their matrix single or double circular and double-stranded DNA molecules called mt DNA and also the 55S ribosomes, called mitoribosomes.

FUNCTIONS OF MITOCHONDRIA

- Mitochondria stores and releases energy in the form of ATP (Adenosine triphosphate). It occurs by the oxidation of carbohydrates, proteins, and fats. It will be further utilized in the different metabolic activities. So, mitochondria are known as the **powerhouse of the cell** or storage batteries of the cell.
- Mitochondria help in the formation of the heme of hemoglobin.
- During cellular respiration, mitochondria form the different intermediate products. They are utilized for the synthesis of cytochromes, chlorophyll, ferredoxin, steroids, alkaloids, pyrimidines, etc.
- Calcium can be stored and released by the mitochondria.
- It helps in the formation of amino acids.

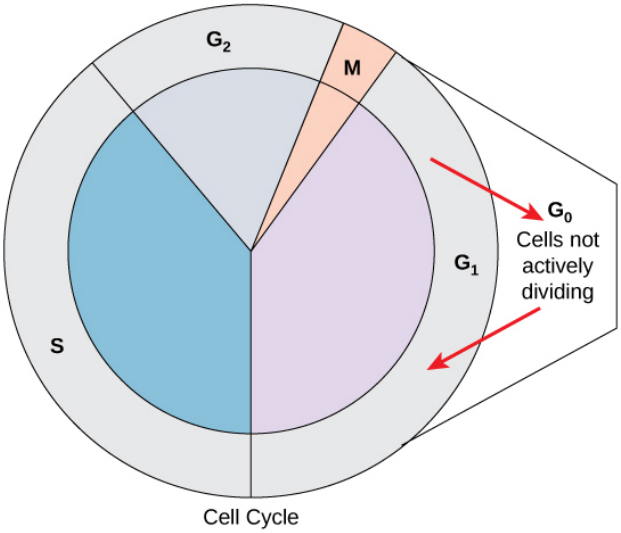
Q-3. Illustrate cell cycle and it s regulation

T**he cell cycle is the sequence of events occurring in an ordered fashion which results in cell growth and cell division.**

- The cycle begins at the end of each nuclear division and ends with the beginning of the next.
- A cell cycle acts as a unit of biological time that defines the life history of the cell.
- The cell cycle is a continuous process that includes all significant events of the cell, ranging from duplication of DNA and cell organelles to subsequent partitioning of the cytoplasm.
- In addition, the process of cell growth where the cell absorbs nutrients and prepares for its cell division is also a part of the cell cycle.
- The process of the cell cycle occurs in various phases, all of which are specialized for a particular stage of the cell.
- The overall process and steps of the cell cycle might differ in eukaryotic and prokaryotic organisms as a result of the differences in their cell complexity.
- Three main cycles are involved in the cell cycle; chromosome cycle, cytoplasmic cycle, and centrosome cycle.

- The chromosome cycle involves DNA synthesis that alternates with mitosis. During this cycle, the double-helical DNA of the cell replicates to form two identical daughter DNA molecules. This is followed by mitosis to separate the cell into two daughter cells.
- The cytoplasmic cycle involves cell growth that alternates with cytokinesis. During growth, the cell accumulates nutrients and growth factors and doubles the contents of the cytoplasm. Eventually, the cytoplasm divides via cytokinesis to equally divide the cytoplasmic contents into two cells.
- The final cycle is the centrosome cycle where the centrosome is divided so that it can be inherited reliably and duplicated accordingly to form two poles of the mitotic spindle fibers.

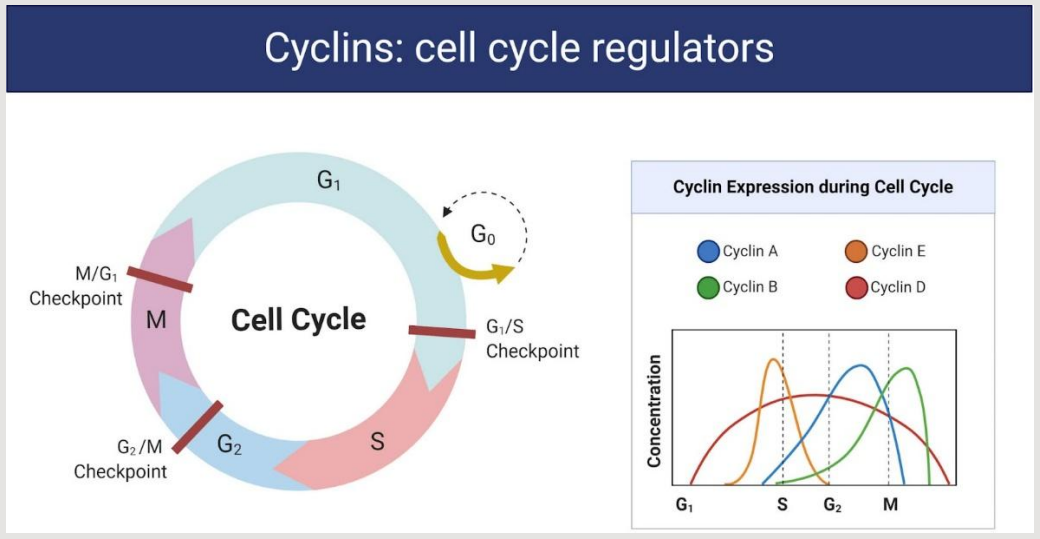
- The cell cycle is regulated by various stimulatory and inhibitory factors that decide whether the cell needs to divide or grow.
- The cell cycle is divided into different phases (according to Howard and Pelc), each of which is defined by various processes.



CELL CYCLE REGULATION:

1. Cyclins

- Cyclins are a group of proteins that together work to regulate different phases of the cell cycle as core regulators.



Mechanism

- The concentration of these cyclins usually remains low for the most part but peaks dramatically if they are needed during the cycle.
- The activation of the cyclin proteins is stimulated by the binding of the growth factors to the receptors on the cell, which activate the transcription of the cyclin genes.
- Most of the cyclin proteins act by binding themselves to the cyclin-dependent kinases, which form a complex. The complex is then responsible for the regulation of the cell cycle.
- Some cyclin proteins like the cyclin D of the G1 phase (or G1 cyclin) act as rate-limiting proteins for cell cycle progression. G1 cyclins accelerate G1 transition by the overexpression of the cyclin genes.
- Even though cyclins do not have any enzymatic activity on their own, they induce different processes in the cell cycle by providing binding sites for other enzymes.

2. Cyclin-dependent kinases (CDKs)

- Cyclin-dependent kinases (CDKs) are a group of enzymes that work to regulate different processes in the cell cycle after activation by the binding of a cyclin molecule.

Mechanism

- The cyclin molecules that bind to these kinases provide additional sequences to the enzymes that are required for their enzymatic activity.
- The CDKs usually have specificity towards different cyclin molecules, and the binding of cyclin to the CDK molecule determines the specificity of the enzyme towards its substrate.
- The mechanism of action of these enzymes might differ among different kinases regulation different phases of the cell cycle.
- The activated CDKs in the interphase undergo phosphorylation and cause inactivation of the retinoblastoma protein (Rb).

3. Maturation-promoting factor (MPF)

- Maturation-promoting factor or M-phase promoting factor (MPF) is a large-sized diffusible protein that regulates the M-phase of a cell cycle.

Mechanism

- During the interphase, the inert subunit of MPF is inactive due to the presence of an enzyme, Wee1.
- The activation of the MPF unit is brought about by CDC25, which results in the binding of the cyclin molecule to the kinase subunit.
- After the binding of cyclin to cyclin-dependent kinase, and the activation of CDK, transition into the M phase begins.
- The MPF molecules then act by adding phosphate molecules to the nuclear envelope, which causes the breakdown of the membrane.
- Besides, it also triggers the formation of spindle fibers as a result of microtubule instability.
- The MPF kinase also phosphorylases several substances like histone H1, which then promotes chromosome condensation.
- The activity of MPF is further regulated by other components like p34. The phosphorylation of p34 regulates the activity of MPF.

4. Anaphase-promoting complex/cyclosome (APC/C)

- Anaphase-promoting complex (APC) is a protein that regulates the M phase of the cell cycle by inhibiting the action of MPF and causes the destruction of cyclin molecules.
- This molecule is important during the transition of a cell from metaphase to anaphase of the M phase.
- The APC is an enzyme that functions in the cell cycle by a different mechanism than CDKs.

5. p53

- p53, also called TP53 or tumor protein, is a gene that encodes for the protein that regulates cell proliferation and also acts as a tumor suppressor.
- The p53 gene is often termed the 'guardian of the genome' as it helps in conserving stability of the genome by preventing genome mutation.
- In eukaryotic organisms, it is important as it suppresses cancer.
- It also stimulates apoptosis if DNA damage is detected that is irreparable.

6. Retinoblastoma protein (Rb)

- Retinoblastoma protein is a nuclear phosphoprotein that helps in cell cycle regulation while also acting as a tumor suppression protein.
- The primary function of Rb is to prevent excessive cell growth during the cell cycle progression.
- It acts as a negative regulator of the cell cycle as inhibiting the process.
- The protein is expressed in both cycling and resting cells which functions by inhibiting a variety of nuclear proteins involved in the cell cycle.
- It regulates the transition of a cell from the G1 phase to the S phase by inhibiting DNA replication.

Q-4-Review structure and functions of Cell membrane

Plasma Membrane: Structure, Composition, Functions

Membranes are lipid structures that separate the contents of the compartment they surround from its environment.

Plasma membranes separate the cell from its environment while other membranes define the boundaries of organelles and provide a matrix upon which complex chemical reactions can occur.

The plasma membrane, also known as the cell surface membrane or plasmalemma, defines the boundary of the cell.

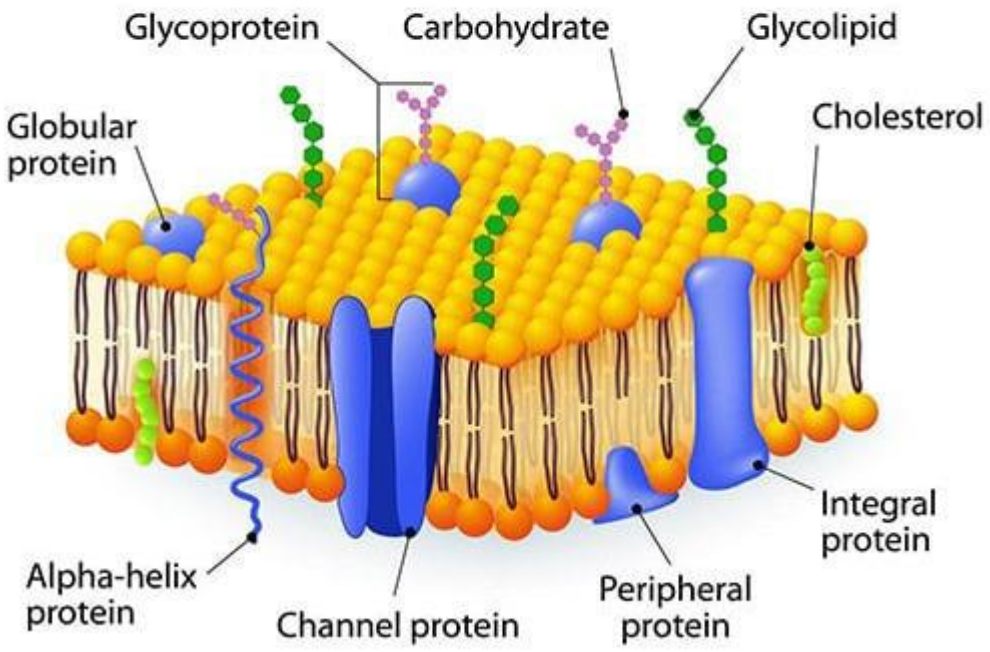
It is a phospholipid bilayer with embedded proteins that encloses every living cell.

It regulates the movement of materials into and out of the cell and facilitates electrical signaling between them.

It is said to be semi-permeable because it allows certain molecules but not others to enter into the cell.

It serves some specific functions such as controlling the flow of nutrients and ions into and out of the cells, mediating the response of a cell to external stimuli (a process called signal transduction), and interacting with bordering cells.

CELL MEMBRANE



Structure of Plasma membrane: All biological membranes are constructed according to a standard pattern. They consist of a continuous bilayer of amphipathic lipids approximately 5 nm thick, into which proteins are embedded. In addition, some membranes also carry carbohydrates (mono- and oligosaccharides) on their exterior, which are bound to lipids and proteins. The proportions of lipids, proteins, and carbohydrates differ markedly depending on the type of cell and membrane.

• The plasma membrane consists of a lipid bilayer containing embedded and peripheral proteins. The major component of membranes is lipids.

• The lipids in the plasma membrane are in the form of phospholipids, which contain a polar head group attached to two hydrophobic fatty acid tails; the head group faces the aqueous environment, the fatty acid tails the interior of the bilayer.

1. Glycerol-based lipids contain a glycerol backbone, and consist of phosphatidic acid (PA), phosphatidylethanolamine (PE), phosphatidylcholine (PC), phosphatidylserine (PS), phosphatidylglycerol (PG), phosphatidylinositol (PI), and cardiolipin (CL).

2. The one sphingosine-based lipid is sphingomyelin (SM).

3. Cholesterol is present in eukaryotic membranes and maintains membrane fluidity at a variety of temperatures. Fluidity is also determined by the content of unsaturated fatty acids in the membrane, which are liquids at room temperature, and the chain length of the fatty acids (shorter chains are more fluid than longer chains).

• The embedded proteins in the plasma membrane function as either channels or transporters for the movement of compounds across the membrane, as receptors for the binding of hormones and neurotransmitters, or as structural proteins.

• The peripheral membrane proteins provide mechanical support to the membrane through the inner membrane skeleton or the cortical skeleton. An example of this is spectrin in the red blood cell membrane. These can be removed from the membrane by ionic agents.

• The third type of membrane proteins is the glycophosphatidylinositol (GPI) glycan-anchored proteins. One example of a GPI-anchored protein is the prion protein, present in neuronal membranes.

• The plasma membrane glycocalyx consists of short chains of carbohydrates attached to proteins and lipids which extend in the aqueous media and both protects the cell from digestion and restricts the uptake of hydrophobic molecules. Note:

• Membrane lipids are strongly amphipathic molecules with a polar hydrophilic "head group" and a polar hydrophobic "tail." In membranes, they are primarily held together by the hydrophobic effect and weak Van der Waals forces and are therefore mobile relative to each other. This gives membranes a more or less fluid quality.

• Lipids and proteins are mobile within the membrane. If they are not fixed in place by special mechanisms, they float within the lipid layer as if in a two-dimensional liquid; biological membranes are therefore also described as being a "fluid mosaic".

Functions of Plasma membrane: The most important membranes in animal cells are the plasma membrane, the inner and outer nuclear membranes, the membranes of the endoplasmic reticulum (ER) and the Golgi apparatus, and the inner and outer mitochondrial membranes. Lysosomes, peroxisomes, and various vesicles are also separated from the cytoplasm by membranes. In plants, additional membranes are seen in the plastids and vacuoles. Membranes and their components have the following functions:

1. Enclosure and insulation of cells and organelles. The enclosure provided by the plasma membrane protects cells from their environment both mechanically and chemically.

The plasma membrane is essential for maintaining differences in the concentration of many substances between the intracellular and extracellular compartments.

2. Regulated transport of substances This determines the internal milieu and is a precondition for homeostasis—i. e., the maintenance of constant concentrations of substances and physiological parameters. Regulated and selective transport of substances through pores, channels, and transporters is necessary because the cells and organelles are enclosed by membrane systems.

3. Signal Transduction Reception of extracellular signals and transfer of these signals to the inside of the cell as well as the production of signals.

4. Enzymatic catalysis of reactions. Important enzymes are located in membranes at the interface between the lipid and aqueous phases. This is where reactions with apolar substrates occur. Examples include lipid biosynthesis and the metabolism of apolar xenobiotics. The most important reactions in energy conversion—i. e., oxidative phosphorylation and photosynthesis also occur in membranes.

5. Interactions with other cells For the purposes of cell fusion and tissue formation, as well as communication with the extracellular matrix.

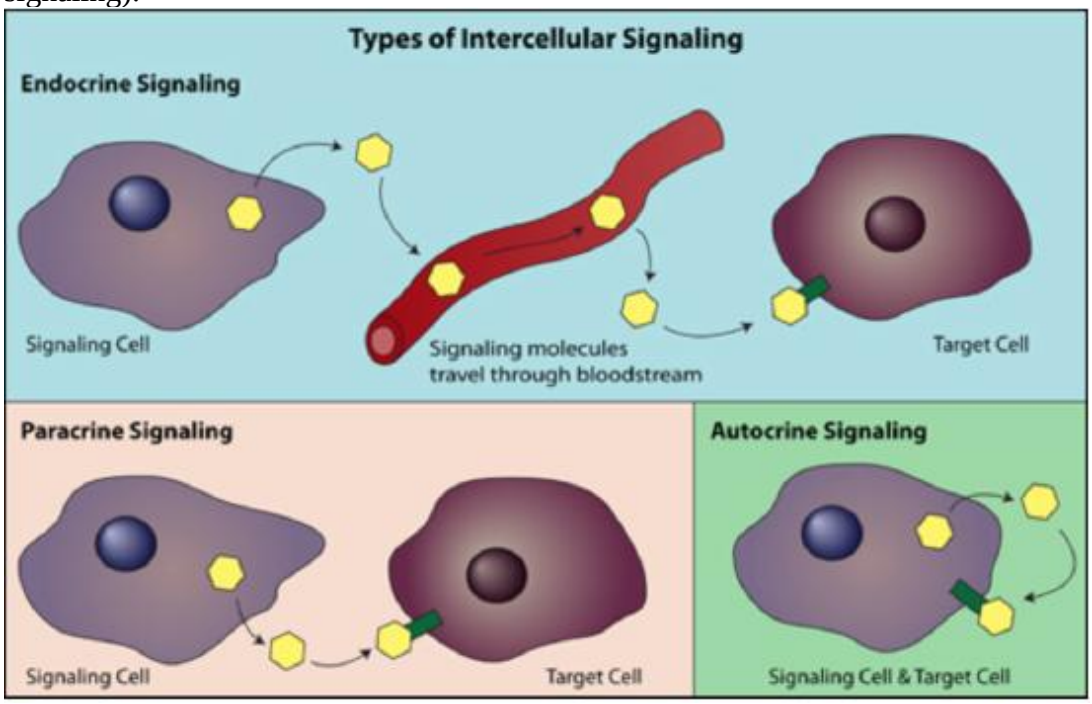
6. Anchoring of the cytoskeleton To maintain the shape of cells and organelles and to provide the basis for movement processes.

Q-5-Generalise different Intracellular signal transduction pathways.

Signal Transduction

As living organisms we are constantly receiving and interpreting signals from our environment. These signals can come in the form of light, heat, odors, touch or sound. The cells of our bodies are also constantly receiving signals from other cells. These signals are important to keep cells alive and functioning as well as to stimulate important events such as cell division and differentiation.

Signals are most often chemicals that can be found in the extracellular fluid around cells. These chemicals can come from distant locations in the body (endocrine signaling by hormones), from nearby cells (paracrine signaling) or can even be secreted by the same cell (autocrine signaling).



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Signaling molecules may trigger any number of cellular responses, including changing the metabolism of the cell receiving the signal or result in a change in gene expression (transcription) within the nucleus of the cell or both.

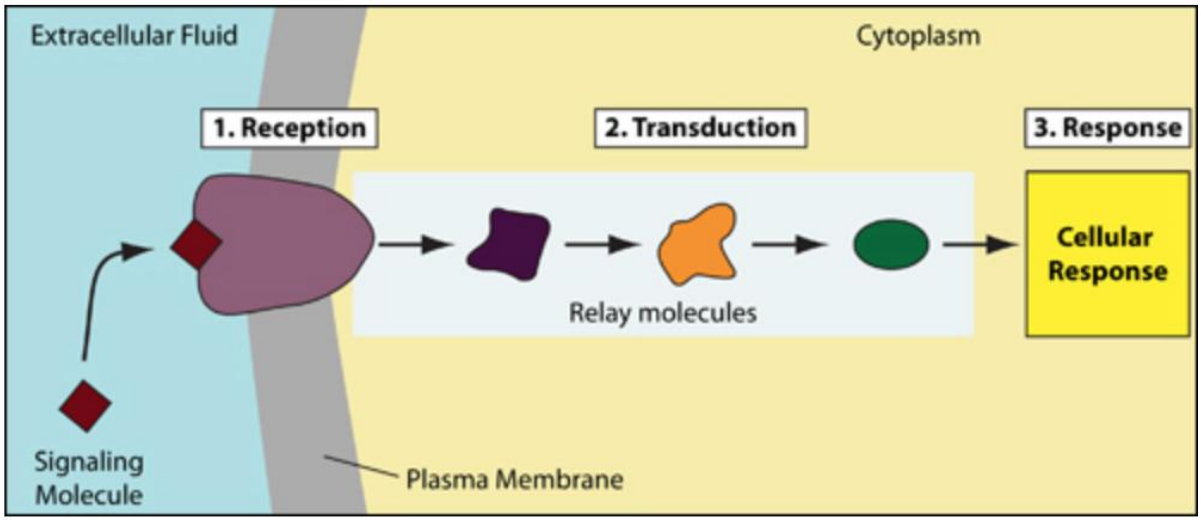
Overview of Cell Signaling

Cell signaling can be divided into 3 stages.

1. Reception: A cell detects a signaling molecule from the outside of the cell. A signal is detected when the chemical signal (also known as a ligand) binds to a receptor protein on the surface of the cell or inside the cell.

2. Transduction: When the signaling molecule binds the receptor it changes the receptor protein in some way. This change initiates the process of transduction. Signal transduction is usually a pathway of several steps. Each relay molecule in the signal transduction pathway changes the next molecule in the pathway.

3. Response: Finally, the signal triggers a specific cellular response.



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Reception

Membrane receptors function by binding the signal molecule (ligand) and causing the production of a second signal (also known as a second messenger) that then causes a cellular response. These type of receptors transmit information from the extracellular environment to the inside of the cell by changing shape or by joining with another protein once a specific ligand binds to it. Examples of membrane receptors include G Protein-Coupled Receptors and Receptor Tyrosine Kinases.

3. (CC BY-NC-SA)

Intracellular receptors are found inside the cell, either in the cytopolasm or in the nucleus of the target cell (the cell receiving the signal). Chemical messengers that are hydrophobic or very small (steroid hormones for example) can pass through the plasma membrane without assistance and bind these intracellular receptors. Once bound and activated by the signal molecule, the activated receptor can initiate a cellular response, such as a change in gene expression.

Transduction

Since signaling systems need to be responsive to small concentrations of chemical signals and act quickly, cells often use a multi-step pathway that transmits the signal quickly, while amplifying the signal to numerous molecules at each step.

Steps in the signal transduction pathway often involve the addition or removal of phosphate groups which results in the activation of proteins. Enzymes that transfer phosphate groups from ATP to a protein are called protein kinases. Many of the relay molecules in a signal transduction pathway are protein kinases and often act on other protein kinases in the pathway. Often this creates a phosphorylation cascade, where one enzyme phosphorylates another, which then phosphorylates another protein, causing a chain reaction.

Also important to the phosphorylation cascade are a group of proteins known as protein phosphatases. Protein phosphatases are enzymes that can rapidly remove phosphate groups from proteins (dephosphorylation) and thus inactivate protein kinases. Protein phosphatases are the "off switch" in the signal transduction pathway. Turning the signal transduction pathway off when the signal is no longer present is important to ensure that the cellular response is regulated appropriately. Dephosphorylation also makes protein kinases available for reuse and enables the cell to respond again when another signal is received.

Kinases are not the only tools used by cells in signal transduction. Small, nonprotein, water-soluble molecules or ions called second messengers (the ligand that binds the receptor is the first messenger) can also relay signals received by receptors on the cell surface to target molecules in the cytoplasm or the nucleus. Examples of second messengers include cyclic AMP (cAMP) and calcium ions.

Response
Cell signaling ultimately leads to the regulation of one or more cellular activities. Regulation of gene expression (turning transcription of specific genes on or off) is a common outcome of cell signaling. A signaling pathway may also regulate the activity of a protein, for example opening or closing an ion channel in the plasma membrane or promoting a change in cell metabolism such as catalyzing the breakdown of glycogen. Signaling pathways can also lead to important cellular events such as cell division or apoptosis (programmed cell death).

Q-6-Explain about Programmed cell death

Apoptosis is a normal genetically programmed cell death where an aging cell at the end of its life cycle shrinks and its remaining fragments are phagocytosed without any inflammatory reaction.

APOPTOSIS MECHANISM
The process of apoptosis is highly complex and sophisticated, involving an energy-dependent series of molecular events.
Three different pathways work on different mechanisms to achieve apoptosis. All three of these pathways converge at the same terminal pathway, which results in the sequential degradation of cellular organelles.

1. **Extrinsic or death receptor pathway**

- The extrinsic pathway that initiates apoptosis involves transmembrane receptor-mediated interactions.
- These interactions take place between ligands and their corresponding death receptors that are all part of the tumor necrosis factor (TNF) family.
- All members of the TNF receptor family share a common cysteine-rich extracellular domain with about 80 amino acids called the “death domain”.
- The death domain plays a vital role in transmitting the death signal from the cell surface to the intracellular signaling pathways.
- The events or interactions that take place in the extrinsic phase of apoptosis involve two

- models; FasL/FasR and TNF-α/TNFR1 models, both of which include the clustering and binding of receptors and their ligands.
- Upon ligand binding, cytoplasmic adapter proteins are activated, which causes the receptors to exhibit death domains.

2. The intrinsic or mitochondrial pathway

- The intrinsic pathway that initiates apoptosis involves a series of non-receptor-mediated processes that produce intracellular signals and act directly on targets within the cell.
- This pathway involves mitochondrial-initiated events.
- The factors that initiate the intrinsic pathway produce intracellular signals that might act in either a positive or negative fashion.

- Negative signals include the absence of certain growth factors, cytokines, and hormones that can lead to failure of inhibition of death programs, thereby triggering apoptosis. In simple words, the withdrawal of factors causes loss of apoptotic suppression and subsequent activation of apoptosis.
- The factors that act positively include, radiation, toxins, hypoxia, hyperthermia, viral infections, free radicals, among others.
- All of these factors cause changes in the inner mitochondrial membrane that causes the opening of the mitochondrial permeability transition (MPT) pore and release of two main groups of pro-apoptotic proteins from the intermembrane space into the cytosol.

3. Perforin/granzyme pathway

- Perforin/granzyme pathway is a novel pathway employed by cytotoxic T lymphocytes that exert their cytotoxic effects on tumor cells and virus-infected cells.
- This involves secretion of the transmembrane pore-forming molecule, *perforin*, with a subsequent release of cytoplasmic granules through the pore and towards the target cell.
- The granules consist of two crucial serine proteases; granzyme A and granzyme B that activate different proteins in the pathway leads to triggering of cell death.

4. Execution pathway

- Both the extrinsic and intrinsic pathways end at the point of the execution phase, considered the terminal pathway of apoptosis.
- This phase of apoptosis is initiated by the activation of various caspases (Factors) that activate cytoplasmic endonucleases and proteases.
- The cytoplasmic endonucleases degrade the nuclear material, whereas the proteases degrade the nuclear and cytoskeletal proteins.

Q.N-9: State structure and importance of Lamp brash and polytene chromosomes

Polytene chromosomes:

Introduction:

- This special type of chromosome is observed by Balbiani in salivary glands of the Chironomus larvae of Dipteran insects.
- Since they were discovered in the salivary glands, they were also called salivary gland chromosomes.
- The present name polytene chromosome was suggested by Kollar due to the occurrence of many chromonemata(DNA) in them.
- Thus, some cells of Drosophila, Chironomus and mosquitoes become very large having high DNA content.
- Polyteny of giant chromosomes is achieved by replication of the DNA several times without nuclear division and the resulting daughter chromatids do not separate but remain aligned side by side.

Detailed explanation:

- Chromonema is required to form Polytene chromosomes which are formed by chromosomal replication without nuclear division.
- The giant chromosomes are also formed by the process of somatic pairing between homologous chromosomes.
- The resultant daughter chromosomes remain aligned with each other and not separated from each other.
- This is very useful for the analysis of many facets of eukaryotic interphase chromosome organization and the genome as a whole.
- The salivary gland cells do not undergo mitosis and die during metamorphosis.
- This chromosome is symbolized by Balbiani rings or puffs which are swollen or puffy areas in polytene chromosomes.
- This chromosome is the site of mRNA synthesis and is multi- stranded in nature.
- The action of the basic dyes can result in the bearing of the number of dark bands in the polytene chromosome of varying size and intensity.
- The dark bands are presumed to be formed by the juxtaposition of chromomeres of the different chromonemata of a polytene chromosome.
- They are found in the permanent prophase stage of mitosis.
- In puffs, DNA is uncoiled for rapid transcription of RNA.
- Hence, the main function is the synthesis of RNA and proteins.

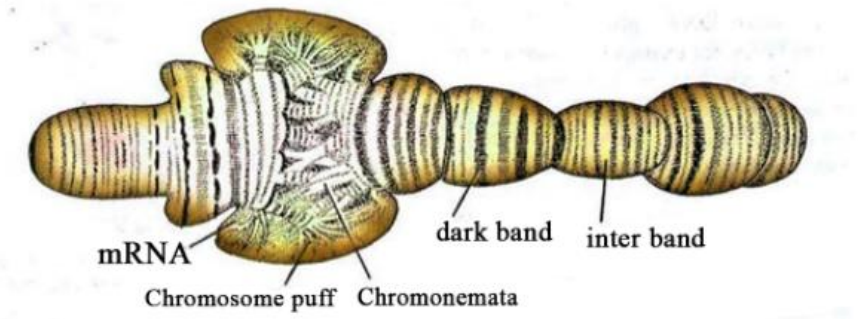


Fig.Polytene chromosome of an insect,showing bands and interbands and a puff or Balbiani ring.

Lamp brush chromosome:

Introduction:

- Lampbrush chromosomes were first observed in Salamander(amphibian) oocytes in 1882.
- He coined the name because the chromosomes look like the brushes which were used for cleaning the glass chimneys of old fashioned paraffin or kerosene lamps.
- This type of chromosome was observed by Flemming in 1882.
- Lampbrush chromosomes occur in the diplotene chromosomes bivalents of most in animal oocytes.
- It is also found in spermatocytes of several species, a giant cell of Acetabularia, and even in plants.
- These chromosomes are even larger than the polytene chromosomes.

Detailed explanation:

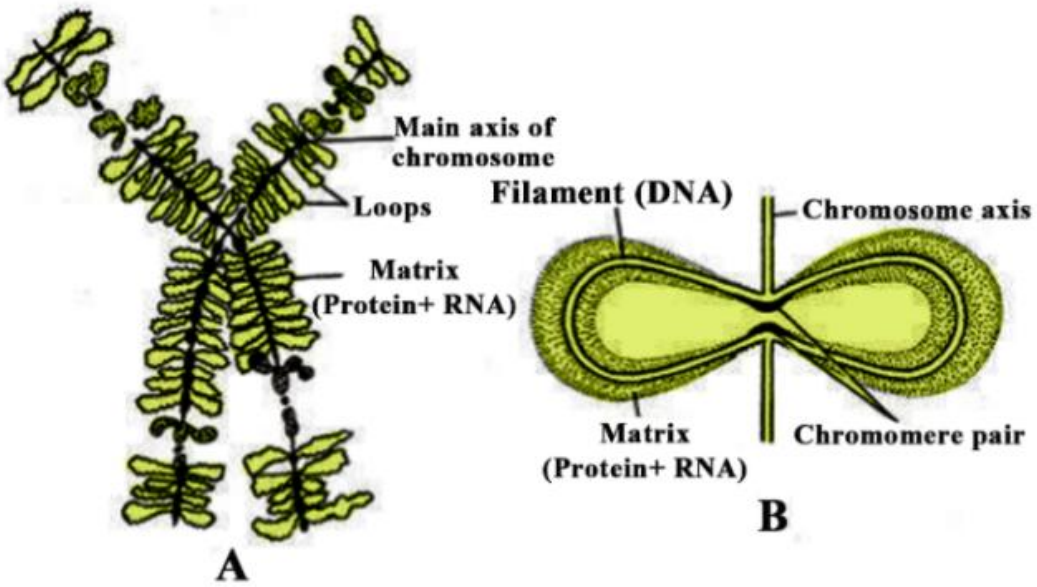


Fig. Lampbrush chromosome. A, Enlarge view of a part of lampbrush chromosome. B, One loop of a lampbrush chromosome

- Lampbrush Chromosomes are concerned with vitellogenesis (Yolk formation).
- This is the special kind of synapsed mid prophase or the Diplotene bivalent stage
- This chromosome occurs in pairs containing homologous chromosomes containing the point of contact known as chiasmata.
- The chromatids forming the chromosomes bear a large number of chromomeres which are separated by interchromomeric stretches.
- Many of the chromomeres give out lateral projections or loops which provide a lampbrush-like appearance to the chromosome pair.
- These loops contain one to several transcriptional units which show transcription of mRNA required for the synthesis of the substances for growth and development of meiocytes.
- Some mRNAs produced by lampbrush chromosomes may be stored as informosomes (mRNA + protein) for producing biochemicals during the early development of the embryo.
- The lateral loops are withdrawn followed by the shortening of chromosomes, after the full development of meiocytes.
- The main function is synthesis of RNA and proteins.
- Lampbrush chromosome is a model useful for studying chromosome organization, genome function and gene expression during meiotic prophase.
- This also allows the visualization of the individual transcription units.

Q.N.10: Compare and evaluate Mendal's Law of segregation and independent assortment

MENDEL'S LAW OF INDEPENDENT ASSORTMENT

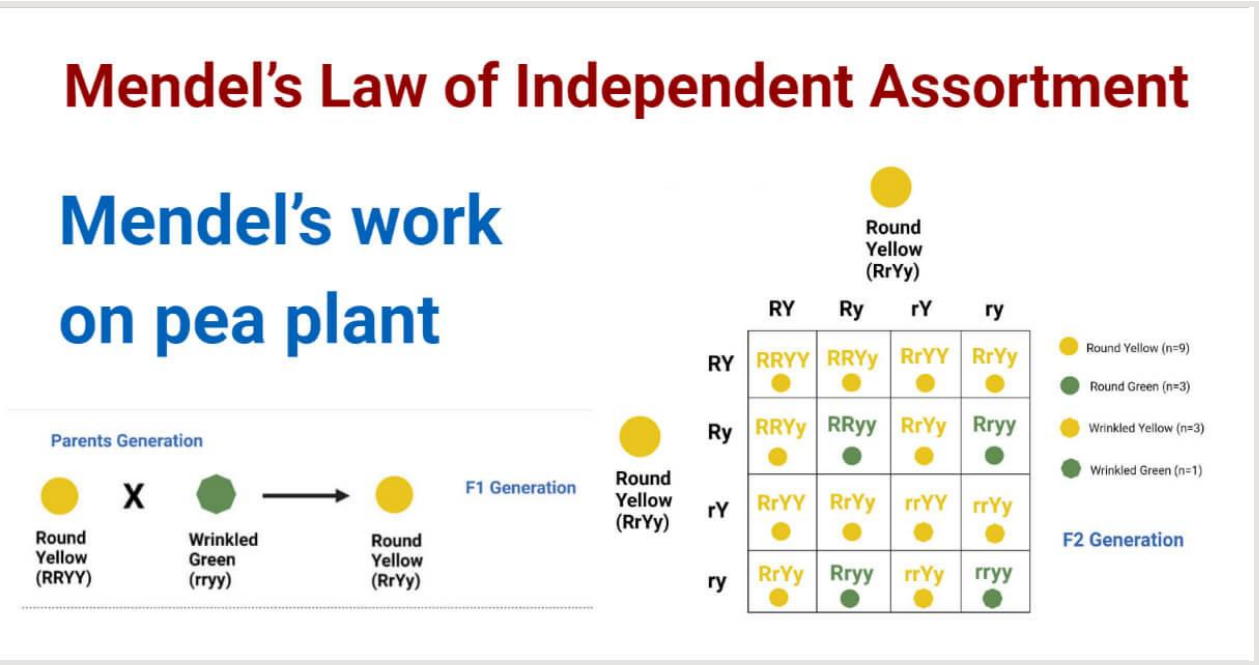
- The law of independent assortment, like the law of segregation, is based on meiosis cell division that occurs during sexual reproduction.
- During meiosis, the diploid chromosomes in the parents are separated to form the haploid gametes.
- The assortment of the chromosomes to the haploid gametes occurs independently of each other in a random manner.
- Thus, it is possible that the chromosomes from the same source can end up in different gametes, resulting in different characteristics.
- Independent assortment of genes is also based on recombination of chromosomes during meiosis which results in a further assortment of genes or characters.
- The pieces of chromosomes from the parents are assorted randomly during recombination resulting in the independent rearrangement.
- The mechanism of independent assortment can be studied by the example of a cross between a homozygous pea plant with yellow round seeds and a homozygous pea plant with green wrinkled seeds.
- The cross between these individuals produces distinct offspring where each pair of contrasting characters is expressed independently of the other characters.
- If the allele for the pea plant with yellow round seeds is YYRR and that with green wrinkled seeds is yyrr, the resulting offspring can have genotypes that include YyRR, YyRr, YYRr, YYrr, Yyrr, yyRR, and yyRr.
- This proves that even though the allele R is associated with the allele Y in the parent, it can express itself in other combinations.

EXAMPLE:

1. Mendel's work on pea plant

- Mendel used the dihybrid cross between a homozygous pea plant with yellow round seeds and a homozygous pea plant with green wrinkled seeds. The parents have the alleles YYRR and yyrr, respectively.
- During meiosis, the chromosomes are separated so that only half the genes are transported to the gametes. The possible genotypes of the gametes then become YR and yr.
- As the parents are crossed, the fusion of the gametes results in the F1 hybrid with the YyRr genotype.
- The F1 hybrids have the phenotype of yellow round seeds as the dominant allele Y for yellow color, and the dominant allele R for the roundness is expressed.
- The alleles in F1 hybrids can now be segregated into four parts; Y for yellow color, y for green color, R for roundness, and r for wrinkledness.
- The four alleles can combine in a number of combinations to produce different genotypes and phenotypes.
- On further crossing, four types of gametes can be formed; YR, Yr. yR, and yr. These gametes fuse to form sixteen individuals in the F2 generation.
- The ratio of the F2 hybrids is 9 yellow round: 3 yellow wrinkled: 3 green round: 1 green wrinkled.

- This proves that both the characters are inherited independently of one another and are expressed according to their dominance.

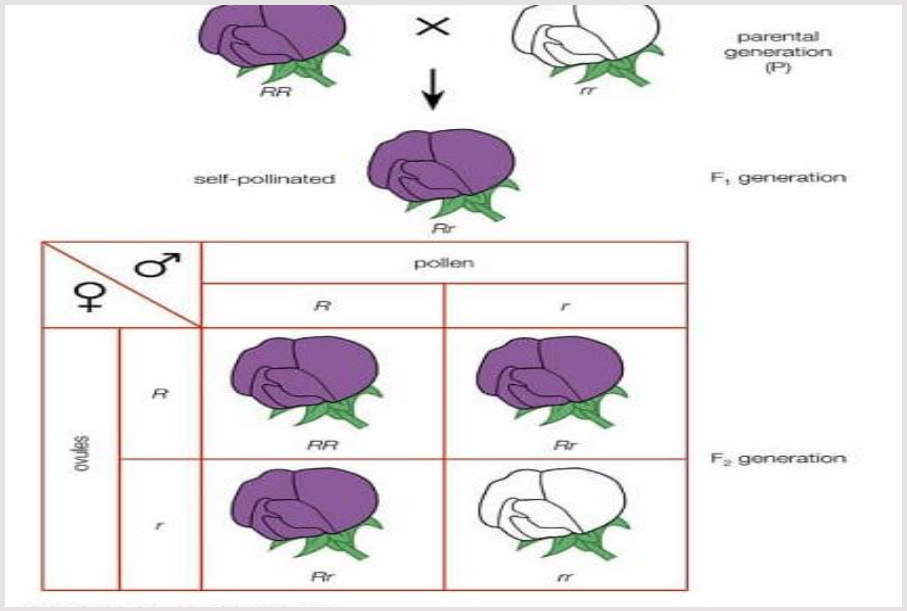


MENDEL'S LAW OF SEGREGATION

Mendel's Law of Segregation states that 'The hybrids or heterozygotes of F1 generation have two contrasting characters of dominant and recessive nature where the alleles though remain together for a long time do not contaminate or mix with each other and separate or segregate at the time of gametogenesis so that each gamete receives only one allele of a character either dominant or recessive.'

In simple words, the law states that only a single gene copy from a parent is distributed in a gamete, and the allocation of the gene copies is entirely random.
Mendel's law of segregation is based on a number of concepts;

- A gene exists in more than one form of an allele.
- During the formation of gametes, the allelic pair of a gene separate so that each gamete has a single allele.
- All organisms inherit two alleles for a genetic trait.
- the two alleles obtained for a trait are different as one is dominant and the other is recessive.
- The law of segregation enables the use of Punnett square for the estimation of resulting genotypes from a cross as it is based on the equal segregation of alleles.
- The law of segregation is significant as it introduced the concept of hereditary factors that remain as separate entities even when present together with other similar entities.
- The law was used to disprove a blending theory by the generation of traits encoded by recessive alleles in the F1 generation.



Q.N.11. Define and distinguish between Incomplete dominance and co-dominance.

Incomplete Dominance

- Incomplete dominance is a mechanism of dominance in heterozygotes, where the dominant allele does not entirely overcome the phenotypic expression of the recessive allele, and there occurs an intermediate phenotype in the heterozygote.
- Incomplete dominance is also called partial dominance or semi-dominance as the phenotype resulting from the genotype is a blend of dominant and recessive alleles.
- An example of this is observed in flowers where the dominant allele is red, and the recessive is white. However, the heterozygous flowers from these alleles might appear pink due to incomplete dominance.
- In incomplete dominance, the dominant allele cannot completely dominate the recessive allele, as a result of which, the resulting phenotype becomes a mix of both.
- Incomplete dominance is important as it explains the existence of a mix of two alleles that are not described by Mendel in his experiment.
- Mendel explained the Law of dominance to indicate that one of the two alleles is dominant as it always dominates the recessive character.
- Mendel couldn't study incomplete dominance as the pea plant he selected for his experiment didn't show incomplete dominance.
- However, his model can still be used to determine the results of crosses of alleles by incomplete dominance. According to his model, the resulting F1 generation will be in the genotypic ratio of 1:2:1 and the phenotypic ratio will be red: pink: white.
- This result indicates that the alleles are still inherited according to Mendel's rule even with incomplete dominance.
- In quantitative genetics, if the phenotype of heterozygous alleles is exactly between (numerically) that of the two homozygotes, this is considered as no dominance. That is, for dominance to occur, the phenotype of heterozygote must lie closer to one of the homozygotes.

EXAMPLE

Wavy hair in humans

- Curly hair is the dominant trait in humans, whereas straight hair is the recessive trait.
- In heterozygous species, the resulting phenotype is wavy hair which is an intermediate between straight and curly.
- Thus, wavy hair results from incomplete dominance where the phenotype results due to the mixing of the two traits.
- Wavy hair, thus, represents a novel phenotype different from straight or curly hair.
- Offsprings formed from two parents with homozygous genotypes will have a genotypic ratio of 1:2:1 with the phenotypic ratio or curly: wavy: straight.

CO-DOMINANCE

Co-dominance is the mechanism of dominance seen in some alleles where both alleles of a gene in a heterozygote lack the dominant and recessive relationship, and each allele is capable of some degree of phenotypic expression.

- Co-dominance is sometimes considered as no dominance at all as the heterozygote shows the phenotypes of both homozygotes.
- Thus, heterozygote genotype gives rise to a phenotype distinctly different from either of the homozygous genotypes.
- For codominant alleles, all upper case base symbols with different superscripts are used. The upper case letters indicate that each allele can express itself to some degree even when in the
- presence of its alternative allele.
- An example of co-dominance can be observed in plants where the dominant phenotype is red, and the recessive phenotype is white, and the heterozygote will have flowers with pink and white spots.
- Like incomplete dominance, co-dominance was also not explained by Mendel as the model he chose didn't express co-dominance.
- However, his model can still be used to determine the results of crosses of alleles by co-complete dominance. According to his model, the resulting F1 generation will be in the genotypic ratio of 1:2:1 and the phenotypic ratio will be red: spotted: white.
- Co-dominance can usually be easily detected in plants and animals with two different colors, but it might also occur in some less visible traits like the blood type.
- Thus, co-dominance is different from incomplete dominance as in co-dominance both the alleles co-exist but separately but in incomplete dominance, the phenotype will be a blend of the two alleles.
  EXAMPLE:

Blood type in humans

- Blood type in humans is determined on the basis of the gene for the proteins that appear on the outside of the blood cells.
- The alleles present are A, B, and O, where A and B represent two different proteins, but O represents the absence of any proteins.

- The existence of A and B proteins, like two colors in flowers, can occur together as a result of co-dominance.
- Thus, if both the proteins A and B are inherited to the offsprings, and both are expressed, AB blood type might occur in the offsprings.
- However, the blood type O represents a dominant/recessive relationship where if A and B genes are expressed, then O doesn't get expressed.

Q.N.12: Define Multiple alleles, Lethal alleles, Epistasis and Pleiotropy

Multiple allele:
Different variants of a gene located at the same location on homologous chromosomes are known as alleles or allelomorphs. Multiple alleles refer to genes that have more than two allelic forms.

- A haploid cell only has one allele, while a diploid cell might have either of the two alleles present on each homologous chromosome.
- Multiple allelism is the term used when more than one allele controls a characteristic. A gene may have many alleles that reside at the same chromosomal location.
- As different expressions of the same gene, multiple alleles affect the same trait.
- The wild-type allele predominates over the mutant alleles in most cases. All other alleles are viewed as variations of the wild type, which is the accepted norm.
- The mutant or variant allele may have an intermediate influence on phenotype or be either dominant or recessive.
- At the population level, there are several alleles, but an individual can only have two alleles for a specific gene.
- The ABO blood type system used by people is one of the most well-known examples of multiple alleles.
- Three alleles, A, B, and O, make up the ABO system. Specific glycoproteins found on the surface of red blood cells are encoded by these alleles.
- The O allele does not code for any antigen, while the A allele does for the A antigen, the B allele does for the B antigen, and so on. Because the A and B alleles are codominant, both antigens will be expressed on a person's red blood cells if they inherit one of each.

PLEOTROPHY

Pleiotropy refers to the phenomenon in which one gene influences several attributes, wherein one allele may cause a variety of phenotypic features in several alleles. This is possible because a gene may produce a protein or RNA molecule that functions in several cellular or metabolic pathways. As a result, modifications to the gene sequence may affect a variety of biological functions, producing pleiotropic effects.

Ex; The gene that causes sickle cell anaemia is one instance of pleiotropy. This gene produces the haemoglobin protein, which is in charge of transporting oxygen in red blood cells.
LETHAL GENES

For the alleles that Mendel studied, it was equally possible to get homozygous dominant, homozygous recessive, and heterozygous genotypes. That is, none of these genotypes affected the survival of the pea plants. However, this is not the case for all genes and all alleles.

Many genes in an organism's genome are needed for survival. If an allele makes one of these genes nonfunctional, or causes it to take on an abnormal, harmful activity, it may be impossible to get a living organism with a homozygous (or, in some cases, even a heterozygous) genotype.

Example: The yellow mouse

A classic example of an allele that affects survival is the lethal yellow allele, a spontaneous mutation in mice that makes their coats yellow. The dominant allele progeny all will die in the F1 generation.

EPISTASIS:

Epistasis is the interaction between two non-allelic genes where the phenotypic expression of one gene is masked or suppressed by the expression of one or more other genes.

This states that certain characters are governed by the expression of two or more genes. Gene interaction occurs as a result of interaction between two or more allelic or non-allelic genes of the same genotype. As a consequence of this, variation in particular phenotypic characters occur.

The expression of one gene relies on the expression (i.e., presence or absence) of another gene, i.e., the expression of genes is not independent of each other.

- Example: Fruit colour in Summer Squash (*Cucurbita pepo*)
- It modifies the classical ratio of 9:3:3:1 into 12:3:1 for the F2 hybrid.
- White, yellow, and green are the three different colors of squash fruits.
- The allele for white color is dominant over both yellow and green. Similarly, yellow is dominant over green color.
- If the dominant allele W is present, it masks the phenotypic effect of the yellow and green allele, and thus, the fruit becomes white.

Q.N.13: Define crossing over. Interpret molecular mechanism of Crossing over

CROSSING OVER

Crossing over refers to the interchange of parts between non-sister chromatids of homologus chromosomes during meiotic prophase (pachytene). In other words, crossing over results from exchange of genetic material between non-sister chromatids involving breakage and reunion at precise point. The term crossing over was first used by Morgan and Cattell in 1912.

2. Feature of Crossing Over:

The main features of crossing over are given below:

1. Crossing over takes place during meiotic prophase, i.e., during pachytene. Each pair of chromosome has four 2. Crossing over occurs between non-sister chromatids. Thus one chromatid from each of the two homologus chromosomes is involved in crossing over.

3. It is universally accepted that crossing over takes place at four strand stage.

4. Each crossing over involves only two of the four chromatids of two homologus chromosomes. However, double or multiple crossing over may involve all four, three or two of the four chromatids, which is very rare.

5. Crossing over leads to re-combinations or new combinations between linked genes. Crossing over generally yields two recombinant types or crossover types and two parental types or non-crossover types.

6. Crossing over generally leads to exchange of equal segments or genes and recombination is always reciprocal. However, unequal crossing over has also been reported.
7. The value of crossover or recombinants may vary from 0-50%.
8. The frequency of recombinants can be worked out from the test cross progeny. It is expressed as the percentage ratio of recombinants to the total population (recombinants + parental types). Thus,

$$\text{Crossing over frequency (\%)} = \frac{\text{No. of recombinants}}{\text{Total progeny}} \times 100$$

Cases of two strand crossing over, somatic crossing over, sister strand crossing over and unequal crossing over are also known.

Molecular Mechanism of Crossing Over:
There are two important theories viz:
1. Copy choice theory and
2. Breakage and reunion theory to explain the mechanism of crossing over.
These are briefly presented below:
i. Copy Choice Theory:
This theory was proposed by Belling. This theory states that the entire recombinant section or part arises from the newly synthesised section. The non-sister chromatids when come in close contact they copy some section of each other resulting in recombination. According to this theory, physical exchange of preformed chromatids does not take place.
The non-sister chromatids when come together during pairing, copy part of each other. Thus, recombinant chromosome or chromatids have some alleles of one chromatids and some of other. The information may be copied by one strand or both the strands. When only one strand copies, non-reciprocal recombinant is produced

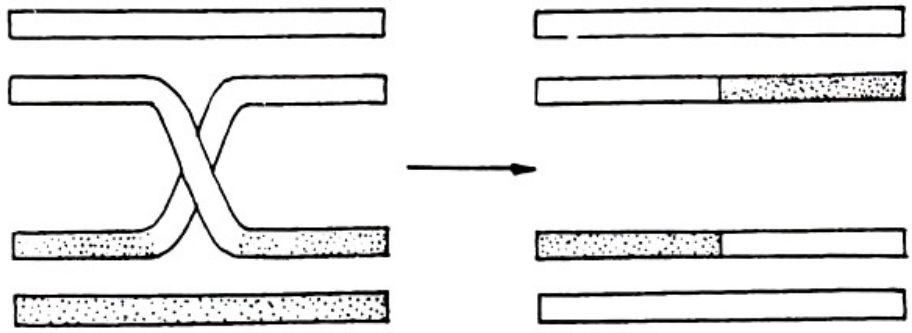


Fig. 9.1. Crossing over according to copy choice theory.

If copy process involves both strands of chromosomes, reciprocal recombinants are produced. Assume, there are two chromosomes, viz., AB and ab. When their chromatids come in close contact they copy each other and result in Ab and aB re-combinations besides parental combinations (Fig. 9.1).
This theory has two objections:
1. According to this theory breakage and reunion does not occur, while it has been observed cytological.
2. Generally crossing over takes place after DNA replication but here it takes place at the same time.
ii. Breakage and Reunion Theory:
This theory states that crossing over takes place due to breakage and reunion of non-sister chromatids. The two segments of parental chromosomes which are present in recombinants

arise from physical breaks in the parental chromosomes with subsequent exchange of broken segments (Fig. 9.2).
The breakage results due to mechanical strains that result from the separation of paired homologous chromosomes and chromatids in each chromosome during pachytene stage. The broken ends of non-sister chromatids unite to produce chiasmata resulting in crossing

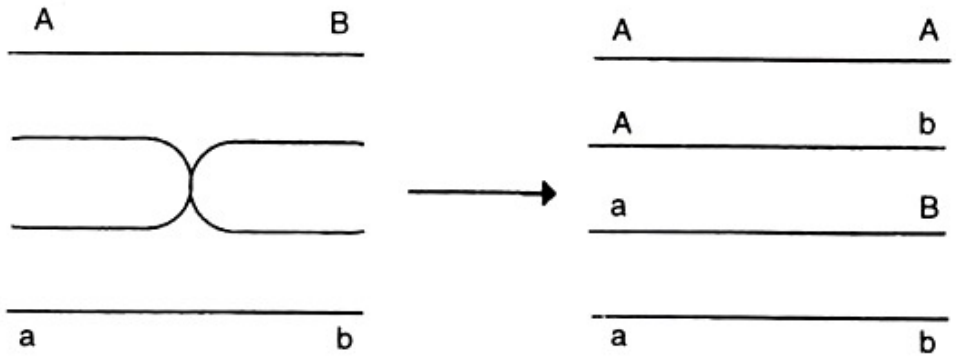


Fig. 9.2. Crossing over according to breakage and reunion theory.

Chromosome Mapping:
Chromosome map refers to a line diagram which depicts various genes present on a chromosome and recombination frequency between them. Such maps are also known as genetic maps or linkage maps. The process of assigning genes on the chromosomes is known as chromosomal mapping.

The mapping of chromosomes is done with the help of three point test cross. A three point test cross is a cross of a trihybrid (F1 differing in three genes) with its homozygous recessive parent. The three point test cross provides useful information on two important aspects, viz:
(1) About the sequence of genes, and
(2) About the recombination frequencies between genes. This information is essential for mapping of chromosomes.

Types of Crossing Over:
Depending upon the number of chiasmata involved, crossing over may be of three types, viz., single, double and multiple as described below:
i. Single Crossing Over:
It refers to formation of a single chiasma between non-sister chromatids of homologous chromosomes. Such cross over involves only two chromatids out of four.
ii. Double Crossing Over:
It refers to formation of two chiasmata between non-sister chromatids of homologous chromosomes. Double crossovers may involve either two strands or three or all the four strands. The ratio of recombinants and parental types under these three situations are observed as 2:2:3:1 and 4 : 0, respectively.
iii. Multiple Crossing Over:
Presence of more than two crossovers between non-sister chromatids of homologous chromosomes is referred to as multiple crossing over. Frequency of such type of crossing over is extremely low.

Significance of Crossing Over:
Crossing over is useful in three principal ways, viz:
(1) Creation of variability,

(2) Locating genes on the chromosomes, and
(3) Preparing linkage maps as described below:
i. Creation of Variability:
Crossing over leads to recombination or new combination and thus is a potential genetic mechanism for creating variability which is essential for improvement of genotypes through selection.
ii. Locating Genes:
Crossing over is a useful tool for locating genes in the chromosomes.
iii. Linkage Maps:
Crossing over plays an important role in the preparation of chromosome maps or linkage maps. It provides information about frequency of recombination's and sequence of genes which are required for preparation of linkage maps.

Q.N.14. Evaluate the role of Natural selection and its role in Genetic drift and genetic shift

Genetic Drift- Definition, Types, Examples
Genetic drift can be defined as the loss of alleles from a population by chance. It is one of the four elements that influence the evolution of a gene pool, along with mutation, gene flow, and natural selection.
Genetic Drift
It is also referred to as the "Sewall Wright effect" based on the theory given by the author in 1969. All populations experience genetic drift. However, small populations are more frequently affected by it. Random variations in allele frequencies in small populations limit genetic diversity, increasing homozygosity and decreasing the capacity of an organism to adapt through evolution.

EFFECT OF POPULATION:

- Small populations are more vulnerable to the effects of genetic drift. Contrarily, large populations are buffered against the effects of chance.
- It is significantly less likely that the **b** allele would be lost and that **B** allele would attain 100% frequency or fixation in such a short amount of time if there were a population of 1000 rabbits as opposed to just 10.
- Alleles are quickly driven to fixation in the smallest population, whereas allele frequencies are more stable in larger populations.

TYPES OF GENETIC DRIFT

There are two major types of genetic drift. They are:

1. Population bottleneck
2. The founder effect

1. Population bottleneck

- It can cause a radical change in allele frequencies in a very short time.
- A population bottleneck occurs when population size abruptly decreases due to unpredicted circumstances, such as the unexpected demise of individuals due to an environmental disaster, habitat degradation, predation, or hunting.
- A new populace is formed when a small number of individuals survive, and this new population's gene frequency undergoes a drastic change.

- Independent of selection, some genes (including rare alleles) of the original population may see a significant increase in proportion, while others may see a significant decline or completely disappear.
- Additionally, the resulting population contains a small fraction of the genetic diversity of the original population.

Example of the Population bottleneck

Due to a population bottleneck caused by humans in the 1890s, northern elephant seals' genetic variation decreased.

At the end of the 19th century, hunting had reduced their population to as low as 20 individuals. Their number has now increased to around 30,000, but their genes still bear the scars of this bottleneck.

They have far less genetic diversity than a group of southern elephant seals that were not subject to as severe a hunting impact.

2. Founder effect

- When a small number of individuals leave a population to start a new subpopulation, it is known as the founder effect.
- The founder effect significantly reduces the genetic diversity in the original population.
- An allele whose frequency was extremely low in the original population may see a significant increase in frequency in the new population due to the founder effect.

Example of Founder effect

It can be observed in the prevalence of the disease in the new population because of disease-related alleles.

Genetic drift:

Genetic drift is a mechanism of evolution that refers to random changes in allele frequencies within a population. Here are some key points:

1. **Random Sampling**: Genetic drift occurs due to random sampling of organisms, particularly in small populations, where chance events can significantly alter allele frequencies.
2. **Bottleneck Effect**: This occurs when a population undergoes a drastic reduction in size due to events like natural disasters. The surviving population may have a different genetic makeup than the original.
3. **Founder Effect**: When a small group breaks away from a larger population to establish a new one, the new population may carry only a small fraction of the genetic variation present in the original group.
4. **Neutral Evolution**: Genetic drift is often considered a neutral process, as it does not necessarily favor advantageous traits but can lead to the fixation or loss of alleles regardless of their effect on fitness.
5. **Implications for Evolution**: Over time, genetic drift can lead to significant evolutionary changes, especially in isolated populations, potentially contributing to speciation.
6. **Population Size**: The effects of genetic drift are more pronounced in smaller populations, where random events can have a larger impact on allele frequencies.

Understanding genetic drift is crucial for studying population genetics and the evolutionary processes that shape biodiversity.

GENETIC SHIFT

Genetic shift, often referred to as "antigenic shift," is a significant change in the genetic makeup of a virus, particularly in influenza viruses. This phenomenon occurs when two different strains

of a virus infect the same host cell and exchange genetic material. As a result, a new virus strain is formed, which can have different surface proteins (antigens). This shift can lead to the emergence of new viral strains that may evade the host's immune system, potentially resulting in widespread outbreaks or pandemics. Antigenic shift is distinct from antigenic drift, which involves smaller, gradual mutations over time.

Natural selection plays a crucial role in influencing genetic shift within populations. Here's how it works:

1. **Survival of the Fittest**: Natural selection favors individuals with traits that enhance their survival and reproductive success in a specific environment. These advantageous traits become more common in the population over generations, leading to a shift in allele frequencies.
2. **Adaptation**: As environmental conditions change, certain traits may become more beneficial, driving a shift in genetic makeup as those traits are selected for. This process leads to the adaptation of populations to their environments.
3. **Genetic Variation**: Natural selection acts on existing genetic variation within a population. If a beneficial mutation arises, it may spread rapidly through the population due to the selective advantage it provides.
4. **Directional Selection**: This form of natural selection favors one extreme of a trait distribution, leading to a shift in the average trait value over time. For example, larger size might be favored in a predator-rich environment, resulting in a genetic shift toward larger body sizes.
5. **Balancing Selection**: In some cases, natural selection maintains genetic diversity by favoring multiple alleles, preventing a complete shift toward a single trait. This can lead to a more stable genetic structure over time.

Overall, natural selection is a driving force that shapes genetic diversity and can lead to significant shifts in the genetic composition of populations.

Q.N.15. WRITE A SHORT NOTE ON- A. SEX LINKED INHERITANCE B. EXTRA CHROMOSOMAL INHERITANCE

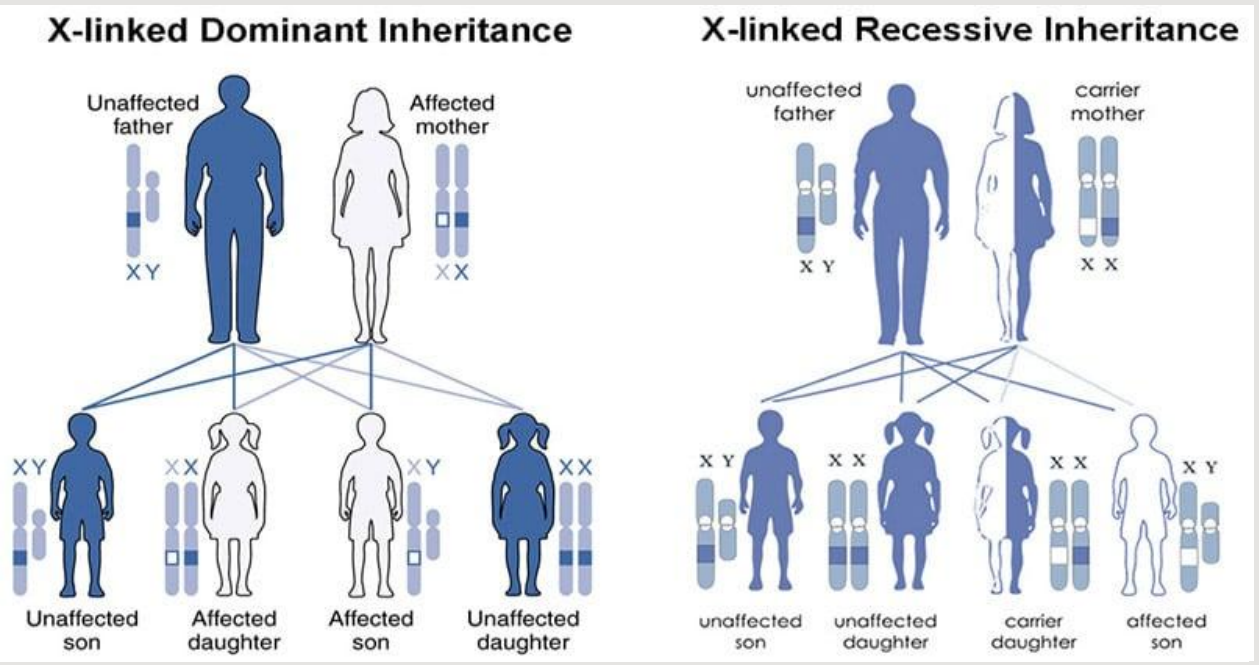
SEX-LINKED INHERITANCE:

- Sex-Linked Inheritance is the inheritance of a trait (phenotype) that is determined by a gene located on one of the sex chromosomes.
- The genes which occur exclusively on the X chromosome or on the analogous Z chromosome (in birds and other species) are called X- or Z -linked genes while the genes which exclusively occur in Y chromosome are called holandric genes.
- The inheritance of such X- or Z-linked and holandric genes is called sex-linked inheritance.

CHARACTERISTICS OF SEX-LINKED INHERITANCE

It has been observed that the genes occurring only in the X chromosomes are represented twice in female (because female contains 2X chromosomes) and once in male (because male has only one X chromosome).

The differential region of each chromosome (i.e., X) contain genes that have no counterparts on the other kind of sex chromosome. These genes, whether dominant or recessive, show their effects in the male phenotype. Genes in the differential regions are called hemizygous ("half zygous") in the males.



INHERITANCE OF X-LINKED RECESSIVE GENES

The X-linked recessive genes show criss-cross pattern of inheritance.

In criss-cross inheritance, an X-linked recessive gene is transmitted from P1 male parent (father) to F2 male progeny (grandsons) through its F1 heterozygous females (daughters), which are called carriers) and different F1 and F2 results (ratios) in the reciprocal crosses.

- The X-linked recessive phenotype is usually found more frequently in the male than in the female. This is because an affected female can result only when both mother and father bear the X-linked recessive allele (e.g., XA Xa × Xa Y), whereas an affected male can result when only the mother carries the gene.
- Usually none of the offspring of an affected male will be affected, but all his daughters will carry the gene in masked heterozygous condition, so one half of their sons (i.e., grandsons of F1 father) will be affected.
- None of the sons of an affected male will inherit the X-linked recessive gene, so not only will they be free of the defective phenotype; but they will not pass the gene along to their offspring.

Example

In Drosophila, the gene for white eye colour is X linked and recessive to another X-linked, dominant gene for red-eye colour.

When white-eyed male was mated with a red-eyed female the F1 flies were all red-eyed. F2 generation of it included 3: 1 ratio of red and white-eyed flies. But all white eyed flies of F2 generation were males only. When normal female of F1 is crossed with normal male 50% of males were white-eyed and 50% were red-eyed It shows that the recessive allele is expressed in male only.

- The common sex-linked disorders that are mostly found in humans are mostly recessive. They include disorders like Color-blindness and Haemophilia.

Extrachromosomal Inheritance

Extrachromosomal inheritance, also known as cytoplasmic or extranuclear inheritance, refers to the inheritance of traits that are not controlled by chromosome genes. Instead, they are determined by genetic materials located outside the chromosomes. This form of inheritance occurs in the cytoplasm of cells and involves genes present in cytoplasmic organelles like mitochondria and plastids. The extrachromosomal hereditary factors have the ability to self-replicate and can be transmitted sexually or asexually. It is important to study these non-chromosomal factors to gain a comprehensive understanding of heredity.

CHARACTERISTICS OF EXTRACHROMOSOMAL INHERITANCE

There are several characteristics associated with extrachromosomal inheritance:

1. Extrachromosomal inheritance does not follow the typical Mendelian inheritance patterns.
2. The inheritance of extrachromosomal factors is independent of genes located within the cell nucleus.
3. In some cases, extrachromosomal traits are inherited exclusively from the mother. This is because the egg contributes more cytoplasm to the zygote compared to the male parent.
4. Extrachromosomal inheritance can lead to characteristic phenotypic changes that are not inherited in a Mendelian pattern.
5. Extrachromosomal genes can exhibit vegetative (somatic) segregation which is rare in nuclear genes.

SIGNIFICANCE

- Extrachromosomal inheritance is important for understanding evolutionary processes. It helps to study inheritance patterns and explore relationships between different species or groups.
- Maternal inheritance in extrachromosomal elements like mitochondrial DNA helps trace maternal lineages and study human population history. It is valuable for understanding ancestral relationships.
- Mutations or changes in extrachromosomal elements can cause genetic disorders or diseases. Studying the mechanisms of extrachromosomal inheritance is important for understanding these inherited disorders.
- Mitochondrial DNA has unique characteristics that make it useful for forensic identification. Its circular shape and multiple copies make it more resilient than nuclear DNA. The presence of specific genes and hypervariable regions enables mtDNA to act as a fingerprint for identification purposes.
- Extrachromosomal inheritance has also been useful in mapping the chloroplast and Mitochondrial genome in many species.

COURSE 8: - CELL BIOLOGY AND GENETICS

UNIT-1

Multiple Choice Questions (MCQ)

1. Which statement is part of the cell theory?
   a) All organisms are made up of multiple cells.
   b) Cells arise only from pre-existing cells.
   c) Cells do not contain hereditary material.

d) Energy flows outside of cells.
Answer: b) Cells arise only from pre-existing cells.

2. Which organelle is responsible for packaging and modifying proteins in eukaryotic cells?
   a) Mitochondria
   b) Lysosomes
   c) Golgi apparatus
   d) Endoplasmic reticulum
   Answer: c) Golgi apparatus
3. What is the main function of peroxisomes?
   a) Protein synthesis
   b) Lipid synthesis
   c) Detoxification of harmful substances
   d) ATP production
   Answer: c) Detoxification of harmful substances
4. Which phase of the cell cycle involves the duplication of DNA?
   a) G1 phase
   b) S phase
   c) G2 phase
   d) M phase
   Answer: b) S phase
5. Which cytoskeletal structure is primarily involved in muscle contraction?
   a) Microtubules
   b) Intermediate filaments
   c) Actin filaments
   d) Golgi network
   Answer: c) Actin filaments

Fill in the Blanks

6. Mitochondria are often referred to as the _____ of the cell because they produce ATP through cellular respiration.
Answer: powerhouse
7. The cytoskeleton is composed of actin filaments, intermediate filaments, and _____, each contributing to the cell's shape and internal organization.
Answer: microtubules
8. The G1 checkpoint in the cell cycle ensures that the cell is ready to enter the S phase and begin _____ replication.
Answer: DNA

True or False

9. True or False: Lysosomes are involved in the breakdown of cellular waste and damaged organelles.
Answer: True
10. True or False: Intermediate filaments are primarily involved in the transport of vesicles throughout the cell.
Answer: False

UNIT-2

Multiple Choice Questions (MCQ)

1. The primary function of the Na+/K+ pump in the cell membrane is to:
   a) Transport glucose into the cell
   b) Maintain the electrochemical gradient by moving 3 Na+ ions out and 2 K+ ions in
   c) Facilitate passive diffusion of ions
   d) Pump calcium ions into the endoplasmic reticulum
   Answer: b) Maintain the electrochemical gradient by moving 3 Na+ ions out and 2 K+ ions in
2. Which process involves the cell engulfing large particles or debris from the extracellular environment?
   a) Pinocytosis
   b) Phagocytosis
   c) Exocytosis
   d) Diffusion
   Answer: b) Phagocytosis
3. The nuclear pore complex primarily functions to:
   a) Regulate the movement of molecules between the nucleus and the cytoplasm
   b) Produce ribosomal RNA
   c) Anchor the nuclear envelope to the cytoskeleton
   d) Synthesize DNA
   Answer: a) Regulate the movement of molecules between the nucleus and the cytoplasm
4. Which gene, when mutated, is most commonly associated with promoting the development of cancer?
   a) p53
   b) BRCA1
   c) Ras
   d) Calmodulin
   Answer: c) Ras
5. Which of the following is a function of tumor suppressor genes?
   a) Promote cell growth and division
   b) Initiate DNA repair mechanisms
   c) Inhibit apoptosis
   d) Increase the rate of cellular metabolism
   Answer: b) Initiate DNA repair mechanisms

Fill in the Blanks

6. The calmodulin protein binds to _____ ions to regulate various cellular processes, including signal transduction pathways.
Answer: calcium (Ca^{2+})

7. Exocytosis is a process where vesicles fuse with the plasma membrane to release their contents into the _____ space.
   Answer: extracellular

8. The nucleolus is the site within the nucleus responsible for synthesizing _____.
Answer: ribosomal RNA (rRNA)

True or False

9. True or False: The nuclear lamina is a mesh-like structure that provides mechanical support to the nuclear envelope and regulates nuclear events such as DNA replication.
Answer: True
10. True or False: Oncogenes normally function to suppress tumor growth, but when mutated, they can lead to uncontrolled cell division.
Answer: False

UNIT-3

Multiple Choice Questions (MCQ)

1. Which organelle is primarily involved in the sorting and modification of proteins before they are transported to their destination?
   a) Mitochondria
   b) Golgi apparatus
   c) Lysosomes
   d) Nucleus
   Answer: b) Golgi apparatus
2. G protein-coupled receptors (GPCRs) activate which molecule after binding to a ligand?
   a) mTOR
   b) G proteins
   c) ERK
   d) DNA
   Answer: b) G proteins
3. Which signal transduction pathway is activated by growth factors and leads to cell proliferation?
   a) GPCR Pathway
   b) ERK/MAPK Pathway
   c) mTOR Pathway
   d) Apoptotic Pathway
   Answer: b) ERK/MAPK Pathway
4. Programmed cell death, or apoptosis, is characterized by all of the following EXCEPT:
   a) Cell shrinkage
   b) DNA fragmentation
   c) Inflammation
   d) Caspase activation
   Answer: c) Inflammation
5. Polytene chromosomes are primarily found in:
   a) Human liver cells
   b) Drosophila salivary glands
   c) Frog oocytes
   d) Mammalian neurons
   Answer: b) Drosophila salivary glands

Fill in the Blanks

6. The mTOR signaling pathway is crucial for regulating cell growth and _____, responding to nutrients, growth factors, and energy status.
Answer: metabolism

7. Lampbrush chromosomes are large, extended chromosomes found in the oocytes of _____ and amphibians, with loops of chromatin active in transcription.
Answer: birds
8. Stem cells have the ability to differentiate into specialized cell types and are also capable of _____ division, giving rise to identical stem cells.
Answer: self-renewing

True or False

9. True or False: In the GPCR signaling pathway, GTP-bound G proteins activate downstream effectors such as adenylyl cyclase or phospholipase C.
Answer: True
10. True or False: Apoptosis is an uncontrolled cell death process that results in damage to neighboring cells and tissue.
Answer: False

UNIT-4

Multiple Choice Questions (MCQ)

1. In a monohybrid cross, which of the following is the phenotypic ratio observed in the F2 generation according to Mendel's law of dominance?
   a) 1:2:1
   b) 3:1
   c) 9:3:3:1
   d) 1:1
   Answer: b) 3:1
2. Which of Mendel's laws states that alleles for different traits are inherited independently of one another?
   a) Law of Segregation
   b) Law of Dominance
   c) Law of Independent Assortment
   d) Law of Linkage
   Answer: c) Law of Independent Assortment
3. What type of inheritance is shown when both alleles in a heterozygote are fully expressed, as in AB blood type?
   a) Incomplete dominance
   b) Co-dominance
   c) Pleiotropy
   d) Epistasis
   Answer: b) Co-dominance
4. In pedigree analysis, a filled-in square represents a(n):
   a) Male without the trait
   b) Female without the trait
   c) Male with the trait
   d) Female with the trait
   Answer: c) Male with the trait
5. In a dihybrid cross, the phenotypic ratio of the F2 generation is typically:
   a) 1:2:1
   b) 9:3:3:1

c) 3:1
d) 1:1
Answer: b) 9:3:3:1

Fill in the Blanks

6. Multiple alleles refer to a gene having more than two _____, such as the ABO blood group system.
Answer: alleles
7. Lethal alleles can cause death when present in the _____ state, often resulting in altered Mendelian ratios in offspring.
Answer: homozygous
8. Pleiotropy occurs when a single gene affects _____ traits, as seen in conditions like Marfan syndrome.
Answer: multiple

True or False

9. True or False: In incomplete dominance, the heterozygote exhibits a phenotype that is an intermediate between the two homozygous phenotypes.
Answer: True
10. True or False: Epistasis occurs when one gene masks or alters the expression of another gene at a different locus.
Answer: True

UNIT-5

Multiple Choice Questions (MCQ)

1. Linkage refers to:
   a) Genes located on different chromosomes
   b) Genes that are inherited together because they are located on the same chromosome
   c) Genes that do not assort independently
   d) Both b and c
   Answer: d) Both b and c
2. During crossing over, genetic material is exchanged between:
   a) Non-homologous chromosomes
   b) Homologous chromosomes
   c) Sister chromatids
   d) None of the above
   Answer: b) Homologous chromosomes
3. Recombination frequency is used to measure:
   a) The mutation rate in a population
   b) The intensity of linkage between genes
   c) The rate of natural selection
   d) The rate of genetic drift
   Answer: b) The intensity of linkage between genes
4. Genetic drift is most pronounced in:
   a) Large populations
   b) Small populations

c) Stable environments
d) Diverse ecosystems
Answer: b) Small populations

Fill in the Blanks

5. Speciation occurs when populations of the same species become _____ and evolve into different species over time.
Answer: reproductively isolated
6. Sex-linked inheritance typically involves genes located on the _____ chromosomes.
Answer: sex
7. Extra-chromosomal inheritance refers to the transmission of genetic material found outside the _____.
Answer: nucleus
8. Crossing over results in new combinations of alleles, which contributes to genetic _____ in a population.
Answer: variation

True or False

9. True or False: The process of natural selection acts only on phenotypes, not genotypes.
Answer: True
10. True or False: In linkage, genes that are located far apart on the same chromosome assort independently.
Answer: False

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