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MATS CENTRE FOR DISTANCE & ONLINE EDUCATION

Cell Biology & Genetics

Bachelor of Science (B.Sc.)
Semester - 2

SELF LEARNING MATERIAL

DSCC

ODL/MSS/BSCB/201

Cell Biology & Genetics

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MODULE INTRODUCTION

Course has five chapters. Under this theme we have covered the following topics:

S.No	Module No	
01	Module 01	CELL
02	Module 02	CHROMOSOME
03	Module 03	DNA, THE GENETIC MATERIAL
04	Module 04	GENE EXPRESSION
05	Module 05	GENETIC VARIATION

This book delves into the intricate world of cellular biology, exploring the fundamental structures and functions that underpin life. From the complexities of the cell envelope and the ultra-structure of organelles to the mechanisms of gene expression and genetic variation, each chapter is crafted to enhance your understanding of these essential biological concepts. We encourage you to engage with all the activities presented in each chapter, regardless of their perceived difficulty, as they are designed to reinforce your knowledge and stimulate critical thinking. By actively participating in these exercises, you will deepen your comprehension of cellular processes and their significance in the broader context of biology.

Module 1

UNIT 1.1Cell

Objectives

- Understand the structure and function of a cell, including its components such as the nucleus, cytoplasm, and cell membrane.
- Differentiate between prokaryotic and eukaryotic cells, highlighting key distinctions.
- Identify and describe the roles of organelles such as the mitochondria, endoplasmic reticulum, and Golgi apparatus in cellular processes.
- Explain the processes of cell division, including both mitosis and meiosis, and their significance in growth and reproduction.
- Describe the structure of the nucleolus and its role in ribosome synthesis, rRNA production, and overall cellular activity.

1.1.1Introduction

A cell is a structure with a large volume of cytoplasm surrounded by a semi-permeable membrane known as the plasma membrane. It encases cytoplasm, several cellular organelles, and the nucleus or nuclear material. Cells are categorised into two types—Prokaryotic and Eukaryotic—based on membrane organisation, the diversity and structure of cytoplasmic organelles, and the complexity of the nuclear area. These phrases were proposed by Hans Ris in the 1960s.

1.1.2History and Origin

Loewy and Siekevitz (1963) described a cell as a "unit of biological activity enclosed by a semi-permeable membrane and capable of self-reproduction in an environment devoid of other living systems." The examination of cells has been facilitated by the use of light microscopes. In 1665, Robert Hooke, utilising a light microscope, discovered that a slice

of cork comprises small spaces encased by rigid walls. He originally employed the term "cell" to characterise his examinations of the "texture of a cork specimen." Subsequently, A. Van Leeuwenhoek (1632-1723) examined different unicellular creatures and cells, including bacteria, protozoa, red blood cells, and sperm. He detected nuclei in certain erythrocytes, facilitated by the advancements in microscopy. In 1809, Mirble M. asserted that all plant tissues consist of cells. In that year, J.B. Lamarck elucidated the significance of cells in living beings. In 1831, Robert Brown observed the nucleus in specific plant cells. Dutrochet (1837) boiled Mimosa cells in nitric acid to isolate the cells, concluding that all organic tissues consist of globular cells bound by basic adhesive forces. Schwann, T. (1839) posited that all living entities are composed of cells following the examination of several animal and plant tissues.

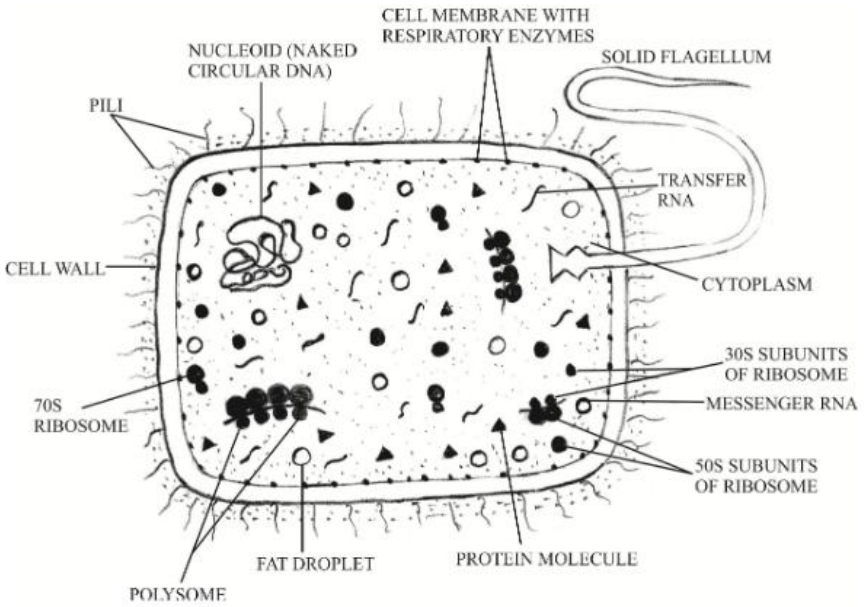


Fig. 1.1: Bacterial Cell

Contemporary microscopes are far more intricate than those utilised in the 1600s by Antony van Leeuwenhoek, a Dutch tradesman renowned for his lens-making expertise. Notwithstanding the constraints of his antiquated lenses, van Leeuwenhoek scrutinised the motility of protista (a category of

unicellular organisms) and sperm, which he collectively designated as “animalcules.”In the 1665 publication Micrographia, experimental scientist Robert Hooke introduced the term "cell" to describe the box-like formations he discovered in cork tissue under a lens. In the 1670s, van Leeuwenhoek identified bacteria and protozoa. Subsequent advancements in lenses, microscope design, and staining methodologies allowed additional scientists to observe some components within cells.

1.1.3Cell Theory

The cell is the basic building block of life, and studying it led to the development of cell theory. In the late 1830s, botanist Matthias Schleiden and zoologist Theodor Schwann studied tissues and put out the unified cell idea. The unified cell hypothesis asserts that all living entities are composed of one or more cells, which serve as the fundamental units of life, and that new cells arise from pre-existing cells. Rudolf Virchow later made important contributions to this theory. Schleiden and Schwann believed that cells originated from spontaneous generation; nevertheless, this notion was ultimately disproven. Rudolf Virchow is famous for saying, "Omnis cellula e cellula," which translates "All cells come from cells that already exist."But the parts of the theory that didn't have anything to do with where cells came from were able to stand up to scientific scrutiny and are still widely accepted by scientists today.

The following are the most well-known parts of modern Cell Theory:

1. The cell is the basic building block of living things and how they work.

2. Every living thing is made up of one or more cells.

3. Cells come from other cells as they divide.

The extended form of the cell theory may also encompass:

• Cells convey genetic material to daughter cells during cellular division.

• All cells have the same basic chemical makeup.

• Cells are where energy transmission (metabolism and biochemistry) happens.

1.1.4 Basic properties of cell

1. Cells are Highly Complex and Organized

2. Cells Possess a Genetic Program and the Means to Use it

3. Cells need Organic Compounds to Generate Essential Macromolecules

4. Cells are dependent to Water, making up more than 70% of the cell

5. Cells are Capable of Producing More of Themselves

6. Cells Acquire and Utilize Energy

7. Cells Carry Out a Variety of Chemical Reactions

8. Cells Engage in Mechanical Activities

9. Cells Die very Fast and Regenerate very Fast to Make New Cells

10. Cells are Able to Respond to Stimuli

11. Cells Are Capable of Self-Regulation

12. Cells Evolve Prokaryotic Cells

All cells, regardless of their type, possess four essential structural components:

1. Plasma Membrane – a protective outer layer that defines the boundary between the cell's internal environment and the external surroundings.

2. Cytoplasm – a gel-like substance (cytosol) that fills the cell and houses the organelles and other cellular structures.

3. DNA – the cell's genetic blueprint, containing instructions necessary for growth, function, and reproduction.

4. Ribosomes – molecular machines responsible for assembling proteins based on genetic instructions.

1.1.5 Difference between prokaryotic Cell & eukaryotic Cell

A prokaryote is a primitive, single-celled organism that lacks a structured nucleus or membrane-bound organelles. This will soon be identified as significantly distinct in eukaryotes. Prokaryotic DNA is found in the nucleoid, which is the cell's core portion. Most prokaryotes have a cell wall made of peptidoglycan, and many species have a polysaccharide capsule.The cell wall is another layer of protection, helps keep the cell's shape, and stops it from drying out. The capsule helps the cell stick to surfaces in the environment. Some prokaryotes have flagella, pili, or fimbriae. Flagella help things move. During a reproductive process called conjugation, pili help move genetic material from one cell to another. Fimbriae help bacteria stick to host cells.

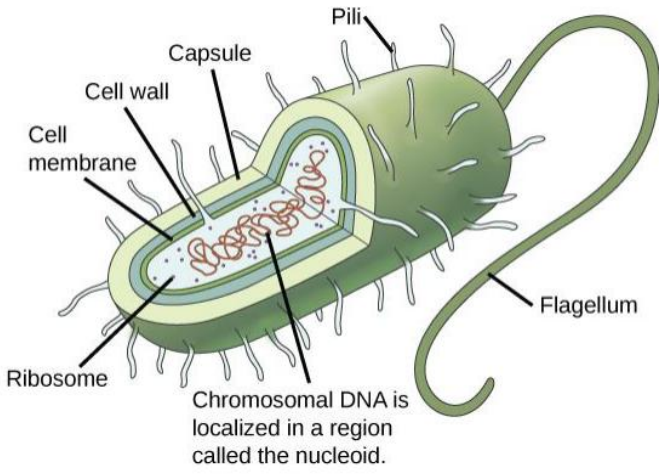


Fig.1.2 General Structure of a prokaryotic Cell

1.1.6 Summary

The cell is the fundamental unit of life, enclosed by a plasma membrane and containing cytoplasm, organelles, and genetic material. Cells are classified into prokaryotic (simple, no true nucleus or membrane-bound organelles) and eukaryotic (complex, with a nucleus and organelles). The study of cells began with Robert Hooke's discovery of "cells" in cork (1665) and van Leeuwenhoek's observations of microorganisms. Later contributions from Brown, Schleiden, Schwann, and Virchow led to the Cell Theory, which states that all living things are made of cells, cells are the basic units of structure and function, and new cells arise from existing ones. Modern extensions highlight genetic continuity, biochemical similarity, and cellular energy processes.
Cells share core features: plasma membrane, cytoplasm, DNA, and ribosomes. They are highly organized, self-regulating, responsive, capable of reproduction, and essential for life's biochemical activities. Prokaryotic cells differ from eukaryotic ones by lacking a nucleus and membrane-bound organelles, often possessing cell walls, capsules, pili, and flagella for survival and reproduction.

Self Assessment Questions

Multiple Choice Questions

1. Who first used the term "cell" to describe the box-like structures observed in cork tissue?

a) Robert Brown

b) Robert Hooke

c) Anton van Leeuwenhoek

d) Theodor Schwann

2. Which of the following is ***not*** one of the universal structural components found in all cells?

a) Plasma membrane

b) Ribosomes

c) DNA

d) Mitochondria

3. Which of the following was *not* part of the original Cell Theory proposed by Schleiden and Schwann?

a) All living organisms are made up of one or more cells
b) The cell is the basic unit of structure and function in living things

c) Cells arise from pre-existing cells

d) Cells pass on genetic material during division

4. What is the main difference between prokaryotic and eukaryotic cells?
a) Prokaryotes have DNA, while eukaryotes do not
b) Prokaryotes lack a true nucleus and membrane-bound organelles
c) Eukaryotes have no plasma membrane
d) Eukaryotes are always multicellular

5. Which scientist is credited with the statement *"Omnis cellula e cellula"* ("All cells come from cells that already exist")?
a) Matthias Schleiden

b) Theodor Schwann

c) Rudolf Virchow

d) J.B. Lamarck

Short Answer Questions

1. Define a cell and mention its two main types.

2. Mention any four basic properties of cells.

3. State the three main points of Cell Theory

Long Answer Questions

1. Trace the history of cell discovery and development of cell theory.

2. Compare prokaryotic and eukaryotic cells in detail.

3. Describe the basic properties and structural components of a cell.

Answers

1- b 2 – d 3 – d 4 – b 5 – c

UNIT 1. 2

Cell Development

1. 2.1 Cell Size

Prokaryotic cells, measuring 1 µm to 5 µm in diameter, are considerably smaller than eukaryotic cells, which range from 10 to 100 µm in diameter. The diminutive dimensions of prokaryotes facilitate the rapid diffusion of ions and organic molecules throughout the cell upon entry. Likewise, any waste generated within a prokaryotic cell can rapidly disperse out. In eukaryotic cells, distinct structural modifications have evolved to improve intracellular transport.

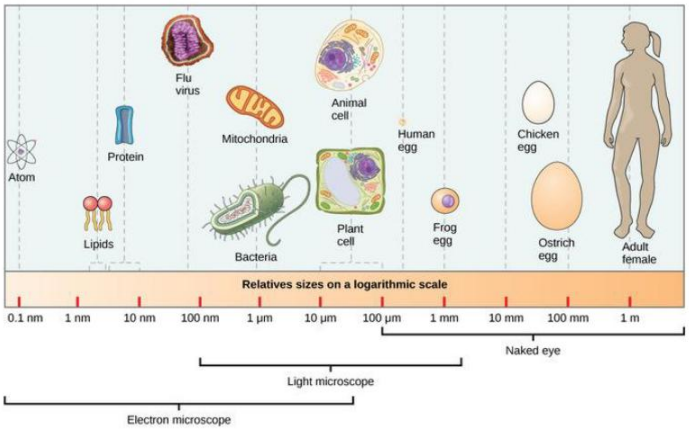


Fig.1.3 Microbial Size

Cellular diminutiveness is essential for all cells, whether of being prokaryotic or eukaryotic. Let us investigate the reasons for this phenomenon. Initially, we will examine the surface area and volume of a standard cell. While not all cells possess a spherical morphology, the majority prefer to approximate a spherical form. The surface area of a sphere is calculated using the formula $4\pi r^2$, and its volume is determined by the formula $4/3\pi r^3$. Consequently, as the radius of a cell expands, its surface area climbs with the square of the radius, whereas its volume escalates with the cube of the radius, resulting in a significantly more rapid increase. Consequently, as a cell enlarges, its surface area-to-volume ratio diminishes. The same logic would be applicable if the cell were cubic in

shape. If the cell enlarges excessively, the plasma membrane will lack adequate surface area to facilitate the diffusion rate necessary for the augmented volume. As a cell enlarges, its efficiency diminishes. One method to enhance efficiency is by division; another is by creating organelles that execute specific functions. These changes resulted in the emergence of increasingly complex cells known as eukaryotic cells.

1.2.2 Eukaryotic Cell Structure

A eukaryotic cell has a plasma membrane, cytoplasm, and ribosomes, just like a prokaryotic cell.

But eukaryotic cells are different from prokaryotic cells in that they have:

1. a nucleus that is surrounded by a membrane

2. a lot of organelles that are surrounded by membranes, like the endoplasmic reticulum, Golgi apparatus, chloroplasts, and mitochondria

3. a few chromosomes that look like rods

A eukaryotic cell is said to have a "true nucleus" since its nucleus is surrounded by a membrane. Just like the organs in your body have specific jobs, organelles (which means "little organ") have specific jobs in cells. They let different parts of the cell do different things.

1.2.3 The Nucleus & Its Structures

The nucleus is usually the most visible part of a cell. Eukaryotic cells have a real nucleus, which means that the cell's DNA is surrounded by a membrane. The nucleus holds the cell's DNA and controls the making of proteins and ribosomes, which are the organelles that make proteins. The nuclear envelope is a double-membrane structure that makes up the outside of the nucleus. The nuclear envelope's inner and outer membranes are made up of two layers of phospholipids. The pores in the nuclear envelope control how ions, chemicals, and RNA travel between the cytoplasm and nucleoplasm. The nucleoplasm is the semi-solid fluid that holds the chromatin and nucleolus inside the nucleus.DNA, the genetic material,

also makes up the structures called chromosomes, which are present in the nucleus. In prokaryotes, DNA is organised into a single circular chromosome. Eukaryotes have chromosomes, which are long, thin structures.

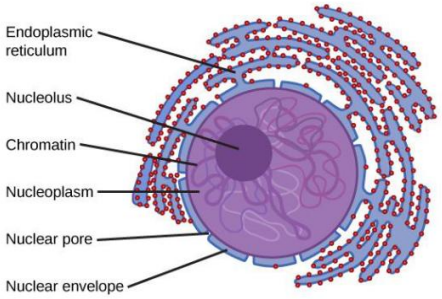


Fig.1.4 Nucleus

1.2.4 Other Membrane-Bound Organelles

Mitochondria are oval-shaped organelles with two membranes. They have their own DNA and ribosomes. People sometimes call these organelles "energy factories" because they do cellular respiration, which makes adenosine triphosphate (ATP), the main chemical that moves energy across the cell. The endoplasmic reticulum makes lipids and modifies proteins, whereas the golgi apparatus sorts, tags, packages, and sends out proteins and lipids. Peroxisomes are small, round organelles with only one membrane. They break down amino acids and fatty acids by oxidising them. Peroxisomes also get rid of a lot of poisons that can get into the body. Vesicles and vacuoles are membrane-bound sacs that are employed for storage and transport. The membranes of vesicles can join with the plasma membrane or other membrane systems inside the cell. This is a very small distinction between vesicles and vacuoles, which are a little bit bigger than vesicles. All of these organelles are present in every eukaryotic cell.

1.2.5 Animal Cells Versus Plant Cells

Even while all eukaryotic cells have the organelles and structures listed above, there are several important differences between plant and animal cells. Animal cells have lysosomes and a centrosome, while plant cells do not. Animal cells have a centrosome near the nuclei that

organisesmicrotubules and lysosomes that help the cell digest food.

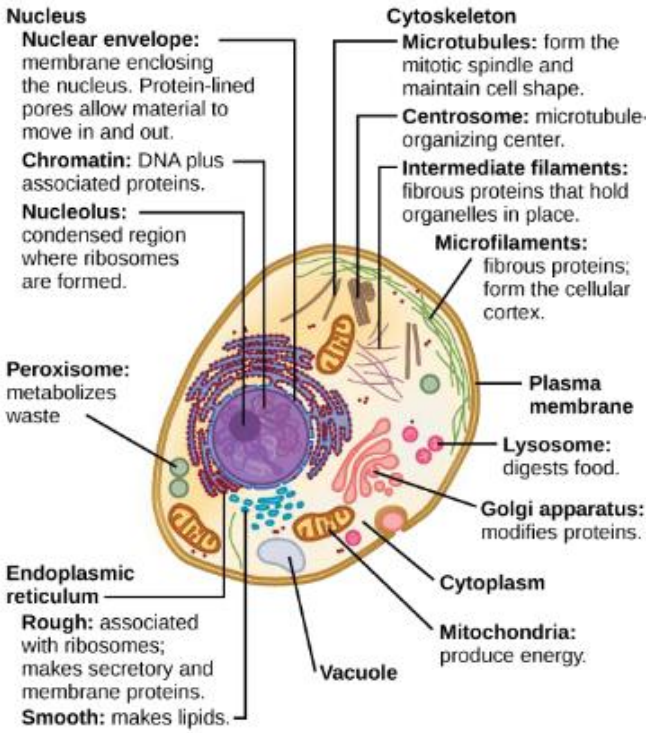


Fig..1.5 Animal Cell

Furthermore, unlike animal cells, plant cells feature a cell wall, a sizable central vacuole, chloroplasts, and various specialised plastids. The central vacuole is crucial in controlling the cell's water content under shifting external conditions, while the cell wall shields the cell, gives it structure, and offers structural support. The organelles that perform

photosynthesisare called chloroplasts.

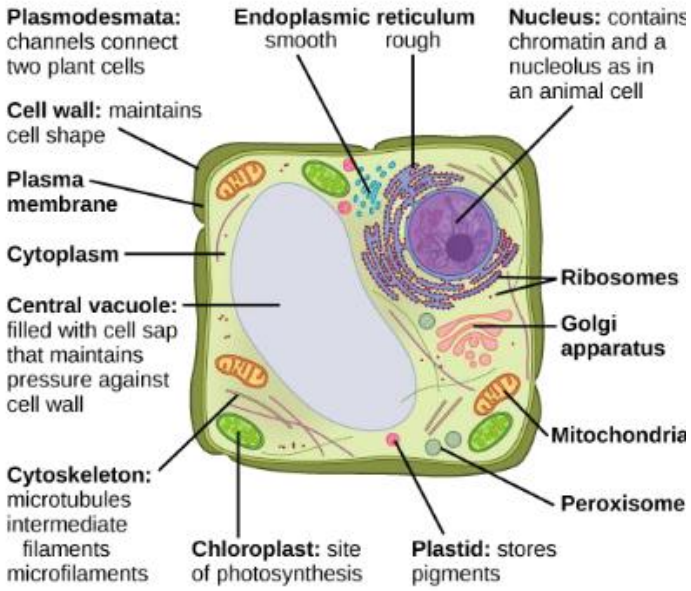


Fig.1.6 Plant Cell

1.2.6 Cell Envelope

Plasma membrane or plasma-lemma is a bio membrane that occurs on the outside of the cytoplasm in both prokaryotes and eukaryotic cells. It separates the cellular protoplasm from its external environment. Prokaryotic cells do not have internal membranous partitions. Several cell organelles, including the nucleus, mitochondria, plastids, lysosomes, Golgi bodies, peroxisomes, and others, are covered by the latter in eukaryotic cells. The reticulum's endoplasm is lined with biomembranes. They also occur on thylakoids inside plastids or cristae inside the mitochondria. A membrane known as the tonoplast divides vacuoles from the cytoplasm. Every biomembrane is dynamic by nature, exhibiting constant changes in size, shape, structure, and function. Schwann made the discovery of the plasma membrane in 1838. In 1855, Nageli and Cramer gave it the name "cell membrane." It was Plowe who named the membrane the plasma lemma (1931).

1.2.7 Chemical Nature of Membranes:

Chemically a bio membrane consists of lipids (20—40%), proteins (59—75%) and carbohydrates (1—5%). The important lipids of the membrane

are phospholipids, sterols (e.g., cholesterol), glycolipids, sphingolipids (e.g., sphingomyelin, cerebrosides). Carbohydrates present in the membrane are branched or un branched oligosaccharides,e.g.,hexose, fucose, hexoamine, sialic acid, etc. Proteins can be fibrous or globular, structural, carrier, receptor or enzymatic. About 30 kinds of enzymes have been recorded in different bio membranes, e.g. phosphatases, ATP-aseesterases, nucleases, etc. The lipid molecules are amphiatic or amphipathic, that is, they possess both polar hydro- philic (water loving) and nonpolar hydrophobic (water repelling) ends. The hydrophilic region is in the form of a head while the hydrophobic part contains two tails of fatty acids. Hydrophobic tails usually occur towards the centre of the membrane. Protein molecules alsopossess both polar and nonpolar side chains. Usually their polar hydrophilic linkages are towards the outer side. The nonpolar or hydrophobic linkages are either kept folded inside or used to establish connections with hydrophobic part of the lipids. Several types of models have been put forward to explain the structure of a bio-membrane. The more important are Lamellar and Mosaic.

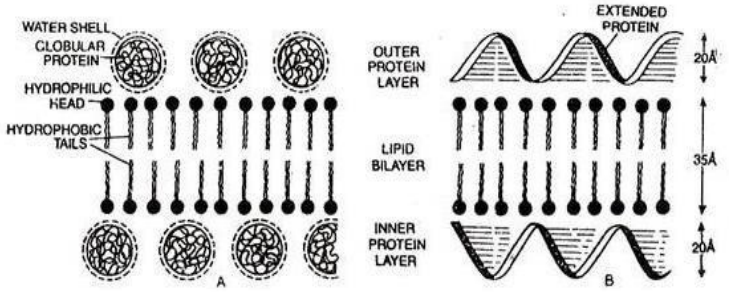
LamellarModels(=SandwichModels):

Theyaretheearlymolecularmodelsofbio membranes. Accordingto these models, bio membranes are believed to have a stable layered structure.

Danielliand Davson Model:

The first lamellar model was proposed by James Danielli and Hugh Davson in 1935 on the basis of their physiological studies. According to Danielli and Davson, a biomembrane contains four molecular layers, two of phospholipids and two of proteins. Phospholipids form a double layer.

The phospholipids bilayer is covered on either side by a layer of hydrated globular or a-protein molecules. The hydrophilic polar heads of the phospholipid molecules are directed towards the proteins. The two are held together by electrostatic forces. The hydrophobic nonpolar tails of the two lipid layers are directed towards the centre where they are held together by hydrophobic bonds and van der Waals forces.



Lamellar models of plasma membrane. (A) after Danielli and Davson (1935). (B) unit membrane, after Robertson (1959).

Fig. 1.7 DanielliandDavsonModel

Robertson Model:

J. David Robertson (1959) modified the model of Danielli and Davson by proposing that the lipid bilayer is covered on the two surfaces by extended or 3-protein molecules. A difference in the proteinsoftheouterandinnerlayerswasalsoproposed,e.g.,mucoprotinontheoutersideand non- mucoid protein on the inner side. Robertson worked on the plasma membrane of red blood cells under electron microscope. He gave the concept of unit membrane which means that:

(i) All cytoplasmic membranes have a similar structure of three layers with an electron transparent phospholipid bilayer being sand-witched between two electrons dense layers of proteins,

(ii) All bio membranes are either made of a unit membraneor a multiple of unit membrane. The unit membrane of Robertson is also called trailaminar membrane. It has a thickness of about 75 Å with a central lipid layer of 35 A thick and two peripheral protein layers of 20Aeach. According to Robertson, if a membrane contains more than three layers, or is thicker than 75A, it must be a multiple of unit membrane.

Mosaic Model:

Mosaic-Fluid Model. In 1972, Singer and Nicolson presented the most modern biomembrane model.

1. This concept states that the membrane is a mosaic of proteins and lipids rather than having a consistent distribution of both. Moreover, the membrane is quasi-fluid rather than solid.
2. It assumes that, similar to the lamellar model, lipid molecules are present in the aqueous bilayer. Protein molecules can be found both inside and outside of lipid bilayers.Extrinsic or peripheral proteins are those that are external, whereas intrinsic or integral proteins are those that are interior..
3. The intrinsic or integral proteins penetrate the lipid bilayer to varying depths and make up 70% of the total membrane proteins. The lipid

bilayer contains some of these. They are known as tunnel proteins, and they can construct channels for the transport of water and water-soluble compounds either alone or in combination.

4. 4. The membranes' structural and functional specialisation is provided by the proteins. Additionally, because the lipid bilayer is quasifluid, the membrane proteins may move laterally, giving the membrane flexibility and dynamism.

5. A few proteins work as carriers because they actively move various substances across the membrane, while many membrane proteins act as enzymes and some act as per-meases for assisted diffusion.Other proteins serve as hormone, antigen, and recognition centre receptors. Glycolipids or glycocalyx are created when a portion of the lipid on the outside complexes with carbohydrates.

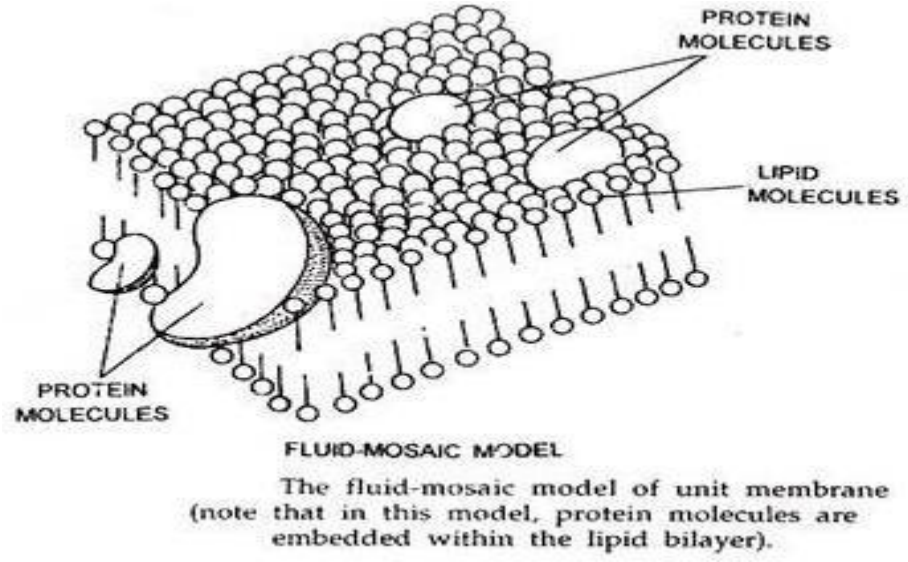


Fig. 1.8 Mosaic Model

ModificationsofCellMembrane:

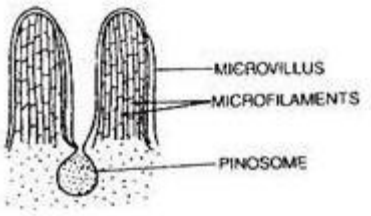
1.Microvilli:

They are finger like evaginations of 0.6—0.8 µm length and 0.1 µm diameter which are found on the free surface of cells engaged in absorption, e.g. intestinal cells, hepatic cells, mesothelial cells, uriniferous tubules. The surface having microvilli is called

striated border or brush border.Microvilli increase the surface area several times. They are supported by a web of microfilaments, actin along with myosin, tropomysoin, spectrin, etc. The narrow spaces in between microvilli take part in pinocytosis.

2.Mesosomes:

They are plasmalem main foldings found in bacteria. One type of mesosome is attached internally to the nucleoid. It is required for nucleoid replication and cell division.



Two microvilli and a pinosome developing in between.

Fig. 1.9 MesosomesJunctionalComplexes:

They are contacts between adjacent cells which in case of animal cells are separated by spaces of 150-200 Å filled with tissue fluid. The important ones are:

(i) Interdigitations:

There is interlocking of finger like membrane outgrowths between two adjacent cells.Interdigitations increase the area of the contact between two cells for exchange of materials.

(ii) IntercellularBridges:

Projections from adjacent cells make contact for rapid conduction of stimuli.

(iii) Tight junctions:

(Zonulae Occludentes, singular— Zonula Occludens). Here plasma membranes of two adjacentcellsarefusedataseriesofpointswithanetworkofridgesorsealingstrands.Tightjunctionsoccur in epithelia with high electrical resistance and where filtration is to occur through the cells, e.g., capillaries, brain cells, collecting tubules of kidneys.

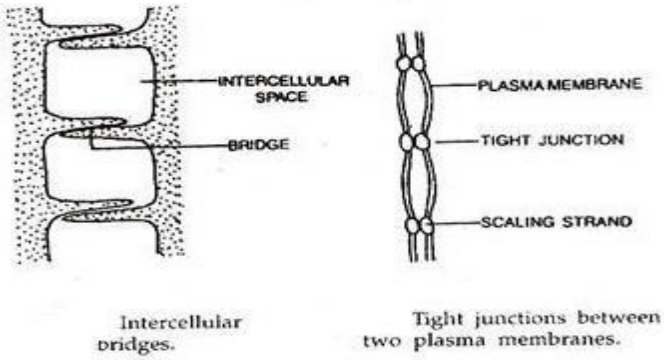


Fig. 1.10 Tight Junctions

(iv) Gap Junctions:

The adjacent cells have protoplasmic connections through special protein cylinders called connexons. Each connexon is made of six identical protein subunits around a hydrophilic channel.

(v) Plasmodesmata They are protoplasmic bridges amongst plant cells which occur in the areas of cell wall pits or pores.

(vi) Desmosomes:

(Maculae Adherentes, singular—Macula Adherens). Adjacent membranes possess disc-shaped thickenings of about 0.5 (am

diameter, a number of tonofibrils (= tonofilaments) and trans-membrane linkers embedded in dense intercellular material. Desmosomes function as spot welds and are hence called spot desmosomes. They occur in epithelia subjected to disruption.

(vii) TerminalBars:

(Belt Desmosomes, Zonulae Adherentes, singular—Zonula Adherens. Intermediary Junction). Terminal bars are desmosomes without tonofibrils. Bands of thickenings occur on the inner surface of membrane. The bands contain microfilaments and intermediate filaments.

1.2.8 Functions of Cell Membranes:

1.The major function of cellular membranes is compartmentalisation. As plasma membranes they separate the cells from their external environment. As organelle coverings, they allow the cell organelles to maintain their identity, specific internal environment and functional individuality.

2.The membranes allow the flow of materials and information between different organelles of the same cell as well as between one cell and another.

3.As plasmodesmata and gap junctions, the bio membranes provide organic connections between adjacent cells.

4.Plasma membranes as well as other membranes of the organelles have selective permeability, that is, they allow only selected substances to pass inwardly to selected degrees. The membranesare impermeable to others.

5.Bio membranes have the propertyof retentivity, that is, theydo not allow the outward passage of substances already permitted entry.

6.Plasma membrane possesses specific substances at its surface which

function as recognition centres and points of attachment.

7.Substances attached to cell membranedetermineantigen specificity. Glycophorins present on the surface of erythrocytes function as antigen determinants. Histocompatibility antigens signify whether a foreign cell or tissue should be incorporated or rejected.

8.Cell membrane has receptors for certain hormones. The hormone combines with its particular receptors and either changes membrane permeability or activates enzyme adenylate cyclase to produce cyclic AMP from ATP. cAMP then triggers a set of enzymes to perform a particular function.

9.Membranes have carrier proteins for active transport.

10. Cellmembranescontainenzymesforperformingcertainreactiononth eirsurface,e.g.,ATP-ase (for ATP synthesis and release of energy from ATP), phosphatases, esterases etc.

11. Certain cell membranes (e.g. plasma membrane in bacteria, thylakoid membranes of chloroplasts, inner mitochondrial membrane) possess electron transport systems.

12. Membrane infolds are used for bulk intake of materials by endocytosis.

1.2.9 Summary

Cells vary in size: prokaryotic cells (1–5 μm) are smaller than eukaryotic cells (10–100 μm). Small size ensures a high surface area-to-volume ratio, which aids diffusion and exchange. Larger eukaryotic cells evolved organelles to improve efficiency and intracellular transport.

Eukaryotic cells differ from prokaryotes by having a true nucleus and membrane-bound organelles such as mitochondria, endoplasmic reticulum, Golgi apparatus, chloroplasts, vacuoles, and lysosomes. The nucleus, enclosed by a double membrane with pores, stores DNA as chromosomes and contains the nucleolus for ribosome production.

Membrane-bound organelles perform specialized functions:

- Mitochondria – ATP production (energy).
- ER & Golgi – synthesis, modification, and transport of proteins and lipids.
- Peroxisomes – breakdown of fatty acids and detoxification.
- Vesicles/Vacuoles – storage and transport.
- Chloroplasts (in plants) – photosynthesis.

Plant vs Animal cells:

- Plant cells have a cell wall, large central vacuole, plastids (chloroplasts).
- Animal cells have centrosomes and lysosomes.

Cell membrane (plasma lemma): discovered by Schwann (1838), named in 1931 by Plowe. It is a dynamic biomembrane made of lipids (20–40%), proteins (59–75%), and carbohydrates (1–5%). Its structure has been explained by models:

- Danielli–Davson (1935) – protein-lipid-protein “sandwich”.

- Robertson (1959) – unit membrane (trilaminar structure).
- Fluid Mosaic Model (Singer & Nicolson, 1972) – flexible lipid bilayer with mobile proteins.

Specialized modifications:

- Microvilli – increase surface area for absorption.
- Mesosomes – bacterial infoldings linked to DNA replication and respiration.
- Cell junctions – tight junctions, gap junctions, desmosomes, plasmodesmata (plants) for communication and adhesion.

Functions of membranes include: compartmentalization, selective permeability, recognition, antigen specificity, hormone reception, active transport, enzymatic activity, electron transport, and endocytosis.

Self Assessment Questions

Objective Type Questions

1. Why are cells generally small in size?
a) To reduce energy requirements
b) To maintain a high surface area-to-volume ratio for efficient diffusion
c) To increase nuclear control
d) To store more organelles

2. What is the main structural difference between prokaryotic and eukaryotic cells?
a) Prokaryotic cells lack DNA
b) Prokaryotic cells lack a true nucleus and membrane-bound organelles
c) Eukaryotic cells have no plasma membrane
d) Eukaryotic cells cannot reproduce

3. Who proposed the Fluid Mosaic Model of the cell membrane?
a) Danielli and Davson

b) J.D. Robertson

c) Singer and Nicolson

d) Robert Brown

4. Which type of cell junction allows direct passage of ions and molecules between adjacent animal cells?

a) Tight junction

b) Gap junction

c) Desmosome

d) Plasmodesmata

5. What are microvilli mainly responsible for?

a) Cell division

b) Absorption by increasing surface area

c) Protein synthesis

d) DNA replication

Answers

1 – b 2 – b 3 – c 4 – b 5 - b

Short Answer Questions

1. Write two structural differences between plant and animal cells.

2. State the chemical composition of the cell membrane.

3. Describe the Fluid Mosaic Model of the cell membrane in brief.

Long Answer Questions

1. Describe the nucleus and its associated structures in detail.

2. Discuss the different models of cell membrane structure with their main features.

3. Compare the structure and functions of plant and animal cells.

1.3.1 Nucleus

The nucleus is usually the most apparent part of a eukaryotic cell. But prokaryotic cells don't have a clear nucleus. The nucleus holds the genome and gives the cytoplasm the information it needs to make things. It is inside a bilaminary nuclear envelope containing pore complexes that let things pass between the nucleus and the cytoplasm. It is normally in the middle of animal cells, with the cytoplasm surrounding it on all sides. But because plant cells have a huge central sap vacuole, it often gets pushed to one side of the cell.

The shape of the nucleus changes depending on the cell type. Although it is usually spheroid, certain cells can also have ellipsoid or flattened nuclei. The nucleus of some white blood cells (WBCs) has a dumbbell form. It has three lobes in human neutrophils.

Monooruninucleate cells, which make up the majority of cells, have only one nucleus. Binucleate cells, such as Paramecium, have two nuclei.A single cell can occasionally have more than two nuclei. These cells are referred to as multinucleated or polynucleate cells. In plants, these cells are called coenocytes (like siphonal algae), while in animals, they are called syncytial cells (like osteoblasts). Eukaryotic cells contain a separate nucleus, but prokaryotic cells, like bacteria, do not. The latter have nucleoid, which is DNA that is spread out throughout the cytoplasm. Also, adult mammalian red blood cells don't have a nucleus.The size of the nucleus varies and is typically associated with the amount of DNA present. Depending on the number of chromosomes (DNA content), the nuclear size varies.

In 1710, Dutch microscopist Antonie van Leeuwenhoek observed that the nucleus was a translucent area in the middle of blood cells in birds and amphibians. Fontana (1781) noted that each eel skin cell possessed an oval shape. Robert Brown first called a noticeable feature in the orchid cell the nucleus in 1831. He proposed the concept of nucleated cells and asserted that the nucleus is an integral component of cells.

There are various parts that make up the nucleus. The nuclear envelope, which is also called the karyotheca, is a thin yet clear layer that surrounds it. Inside the envelope, there is a transparent fluid called nucleoplasm, which is also known as nuclear sap or karyolymph. This is where the solutes of the nucleus dissolve.The nucleoplasm contains one or more

spherical structures termed nucleoli (plural: nucleolus), fine-intermingled nucleoprotein filaments commonly referred to as chromatin, and a network of protein-containing fibrils known as the nuclearmatrix. The nucleus lacks microtubules and membranes. Nevertheless, the nuclei of protozoans that create a mitotic spindle within the nuclear envelope include microtubules.

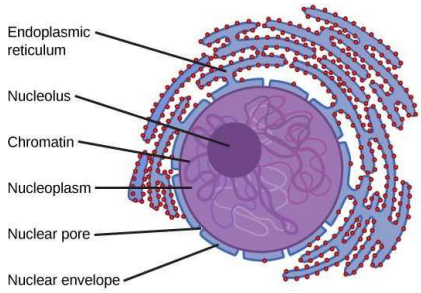


Fig. 1.11: Nucleus

1.3.2 ChemicalComposition

The nucleus is made up of about 9–12% DNA, 5% RNA, 3% lipids, 15% simple basic proteins like histones or protamines, and about 65% complex acid or neutral proteins. These proteins include enzymes like polymerases that help produce DNA and RNA, as well as organic phosphates and inorganic salts or ions like Mg++, Ca++, and Fe++.

1.3.3 Functions

The nucleus is the part of the cell that controls everything. It performs the following main purposes: • It protects the cell by directing the creation of structural proteins.

- • It controls cell metabolism by influencing the production of enzyme proteins.
- • It also carries genetic information about how the organism behaves, grows, and reproduces, in addition to its structure and metabolism.
- • It makes cells replicate when it needs to.
- • It is where the parts of ribosomes are made.
- • It helps cells differentiate by only keeping certain genes activated.
- • It makes genetic changes that cause evolution.

1.3.4 Summary

The cell is the fundamental unit of life, forming the structural and functional basis of all living organisms. Whether existing independently as unicellular organisms or combining into complex multicellular systems, cells carry out essential processes that sustain life. The journey of a cell from its origin to its specialized form is known as cell development, a highly regulated process involving cell division, growth, and differentiation. Through development, cells acquire specific functions, allowing the formation of tissues and organs necessary for the organism's survival. Within the nucleus of a cell lies the nucleolus, a dense, membrane-less structure that plays a crucial role in producing ribosomal RNA and assembling ribosomes—key components in protein synthesis. The nucleolus is active during interphase and temporarily disassembles during mitosis, reflecting its dynamic role in cellular activity.

Self Assessment

MultipleChoiceQuestions:-

1. Nucleusisseparatedfrom cytoplasm by nuclear membrane which is

   (a) Double,non-porous (b) Single,non-porous

   (c) Single,porous (d) Double,porous

2. Nucleolus is especially rich in:

   (a) DNAand proteins (b) DNAandlipids

   (c) RNAand proteins (d) RNAandlipids

3. Nuclearmembrane facilitates:

   (a) Synapsesofhomologouschromosomes

   (b) Nucleocytoplasmicexchangeofmaterials

   (c) Anaphasicseparationofdaughterchromosomes

   (d) Organizationof spindles

4. Nucleoplasmiscontinuouswithcytoplasmthrough:

   (a) Centriole(b) Nucleopore

(c) E.R(d) GolgiBody

5. Themajorcomponentofthenucleusis:

(a) DNA (b) RNA

(c) Lipids Proteins (d)

6. Chiefroleofnucleolusinanucleusconcerns:

(a) Organizationof chromosomes (b) DNAreplication

(c) Ribosomal synthesis (d) Chromatid separation

7. Nucleuswasdiscoveredby:

(a) RobertBrown (b) RobertHook

(c) Virchow (d) DeDuve

8. Nucleolarorganizerisassociatedwith:

(a) Synthesisofplasma membrane (b) Ribosome formation

(c) G6PD
(d)Disappearanceofnuclearmembrane

VeryShortQuestions:

1. What is the study of nucleus called??
2. Whodiscoveredthenucleus?
3. How many types of histones are found associated with DNA?

4. Whatisthecompositionofchromatin?
5. Whatare nucleosomes?
6. Whatisaninterphasenucleus?
7. GivetheroleofDNApresentinnucleolus?
8. Which has more DNA and less RNA, euchromatin or heterochromatin?
9. Where are nucleoli formed at the end of cell division?
10. Nametwotypesofchromatin.

Answer

1 d

2 c

3 b

4 b

5 d

6 c

7 a

8 b

Module 2 Chromosomes

Objectives

 Understand the structure and organization of chromosomes, including the role of histones, chromatin, and centromeres in maintaining chromosomal integrity and function.

 Explain the different types of chromosome variations, such as structural and numerical variations, and their effects on genetic stability and organismal traits.

 Describe the process of cell division, including both mitosis and meiosis, and understand how these processes ensure the accurate distribution of chromosomes to daughter cells.

 Discuss the mechanisms of chromosomal recombination and how they contribute to genetic diversity during meiosis.

 Explore the significance of chromosome abnormalities, such as aneuploidy and polyploidy, and their consequences for human health and development.

UNIT 2.1

CHROMOSOME ORGANIZATION

2.1.1Chromosomes

The Greek words "chroma" (colour) and "soma" (body) are the roots of the English word "chromosome." The most significant and permanent component of the cell nucleus, chromatin, makes up these special cell organelles. They are able to reproduce themselves. They have a significant impact on differentiation, inheritance, mutation, and evolution in addition to controlling the structure and metabolism of cells.

2.1.2History

Nuclear filaments were found in the nucleus of Tradescantia pollen mother cells by W. Hofmeister in 1848. W. Flemming achieved the first precise count of chromosomes in a cell's nucleus in 1882. W. Flemming, Evan Benden, and E. Strasburger proved in 1884 that during mitosis, the number of chromosomes doubles through longitudinal division. In 1887, Beneden discovered that each

species had an equal number of chromosomes. W. Waldeyer first used the name "chromosomes" to refer to the nuclear filaments in 1888. In 1902, W.S. Sutton and T.Boveri proposed that chromosomes play a part in heredity, and Morgan verified this in 1933.

2.1.3 Different chromosome structures

1.1. Viral chromosome: A virus's chromosome contains a single nucleic acid molecule (DNA or RNA) that is encased in a protein coat known as the capsid. It could be round or linear. DNAviruses are viruses with DNA as their genetic material, while RNAviruses are viruses with RNA as their genetic material. The viral chromosome contains a small amount of genetic material that mostly controls the creation of other virus particles of the same kind in the host cell. In RNA viruses, the RNA frequently uses reverse transcription to guide the host's production of DNA complementary to itself. The DNA subsequently transcribes the RNA to create new virus particles. Retrovirus is the term for such a ribovirus. The retrovirus that causes AIDS.

2. Prokaryotic chromosomes: Prokaryotic chromosomes, like those seen in bacteria, contain a single circular two-stranded DNA molecule that is not membrane-enclosed. It has direct contact with the cytoplasm and is protein-free. Some RNA that seems to form a core packs the bacterial chromosome into the nucleoid. It has at least one persistent attachment to the plasma membrane. Most bacterial cells may also have some extra-chromosomal DNA molecules in addition to the main chromosome; these molecules are circular and double-stranded, but they are considerably smaller. We call them plasmids. The plasmid can be present in the cytoplasm of cells on its own or in conjunction with the primary chromosomal DNA, which is known as an episome.

3. Eukaryotic chromosomes: These are found in the nucleus and several other organelles, such as plastids and mitochondria. Nuclear and extra nuclear chromosomes are the names given to these chromosomes, respectively.

Double-stranded, long, linear DNA molecules make up nuclear chromosomes. They are linked to proteins.A nuclear envelope envelops them.Compared to prokaryotic chromosomes, DNA is involved in the coding of significantly more proteins.

Plasmids and mitochondria include extra nuclear chromosomes. They are short, circular, double-stranded DNA molecules. They don't associate with proteins. For the organelles that contain certain protein particles, less genetic information is available for their synthesis. Under the guidance of

nuclear chromosomes, other proteins are synthesised in the cytoplasm after being received.

2.1.4Chromosome Morphology

The eukaryotic chromosomes are stretched into long, thin chromatin fibres during the interphase stage, where they form the chromatinreticulum by lying crisscrossed. They double during the S-phase of replication. They are made up of two chromatids at this stage, which are joined at a centromere. The chromosomes compress and firmly coil up during cell division, becoming distinguishable during the metaphase stage. Although the number, size, shape, and location of eukaryotic chromosomes vary, their structure is remarkably consistent.

Number- Different species have anywhere from two to several hundred eukaryotic chromosomes. Every cell in a species has the same number of chromosomes, except for gametes. Because a species' chromosome count is consistent, it can be used to determine the species' taxonomic place and phylogeny.

Size: Not every chromosome in a species is the same size. They differ in size from species to species as well. However, the size of a species' specific chromosome is essentially constant. Larger chromosomes are found in creatures with fewer chromosomes than in those with more. The chromosomes of plants are generally larger than those of animals, and the monocots have larger chromosomes than the dicots.

Shape: At the metaphase stage, the chromosomes resemble thin rods that can be straight or curled to create a S or an arc. Depending on where the centromere is located, they may take on J or V forms during the anaphase stage.

Position: Every chromosome in a nucleus is separate from every other chromosome in its vicinity. They can therefore be found in any part of the nucleus.

Structure: The chromosome features two highly coiled sister

chromatids at the metaphase stage because it is a highly condensed nucleoprotein filament. The centromere, a section of the narrow region known as the major constriction of the metaphase chromosome, holds these chromatids together as they run parallel to one another along their length. During cell division, spindle microtubules attach to the kinetochore, a darkly stained, disc-like, fibrous structure found at the centromere of each chromatid. The places where force is applied to draw the chromatids towards the poles are known as kinetochores.Secondary constrictions are very thin regions that can be seen on one or more chromosomes. Satellite is the part of the chromosome that is cut off by secondary constrictions. A satchromosome is a type of chromosome that has a satellite. For a species, the size and shape of the satellite remains constant. The nucleolar organisers are secondary constrictions that are connected to the nucleoli.Nucleolar chromosomes are those that have nucleolar organising regions.

endpoints: Telomeres are the endpoints of chromosomes. The telomere serves a different purpose than the remainder of the chromosome. A chromosome may break and its fragments may reassemble after being exposed to X-rays, but no segment joins the telomere, indicating that the telomere has polarity and that it somehow "seals" the end.

Ultrastructure: Achromatids contain chromonema, a single, long, double-stranded DNA molecule, which is an extremely fine filament. To create nucleosomes, it is wound around histones. The chromatin fibre is made up of non-histone proteins and the nucleosome. Reactive groups found in the chromatin fiber—likely H1 histone molecules—act as "folders" and crosslink the fibre, transforming it into a massive, compact, coiled metaphase chromatid.

Chemical composition: The eukaryotic chromosome's chromatin is made up of roughly 35% DNA, 60% proteins, 5% RNA, a few metal ions, and certain enzymes.

Chromosome types: The chromosomes are categorised as follows based on the location and quantity of centromeres.

Metacentric: In metacentric chromosomes, the arms are equal and the centromere lies in the centre of the

chromosome.Thechromosome appears V-shaped during anaphase. Human chromosomal number three, for instance.

2.1.5Functions

(i) Genetic traits are passed down from parents to children via chromosomes.

(ii) They control the production of structural proteins, which aids in cell division, growth, and self-maintenance.

(iii) They regulate metabolism by guiding the synthesis of essential enzymes.

(iv) Throughout development, they direct cell differentiation.

(v) In daughter cells, they create nucleoli at nucleolar organiser sites.

(vi) By altering their genes, they create variants and aid in the species' evolution.

(vii) They contribute to determining sex.

(viii) Through reproduction, they preserve life's continuance.

2.1. 6 Summary

Definition & Importance

- Chromosomes (from Greek *chroma* = colour, *soma* = body) are permanent nuclear components made of chromatin.
- They self-replicate and control cell structure, metabolism, inheritance, mutation, differentiation, and evolution.

History

- 1848: W. Hofmeister observed nuclear filaments.
- 1882: W. Flemming counted chromosomes.
- 1884: Flemming, Benden & Strasburger described longitudinal division.
- 1887: Benden noted species-specific chromosome number.

- 1888: W. Waldeyer coined the term "chromosome."
- 1902: Sutton & Boveri proposed role in heredity; confirmed by Morgan (1933).

Types of Chromosomes

1. **Viral:** Single DNA/RNA molecule with protein coat (capsid). May be linear/circular. Retroviruses (e.g., HIV) use reverse transcription.
2. **Prokaryotic:** Single, circular, double-stranded DNA in nucleoid (protein-free). May have plasmids/episomes.
3. **Eukaryotic:** Nuclear chromosomes (linear DNA + proteins, inside nuclear envelope) and extranuclear chromosomes (mitochondria, plastids – circular DNA, protein-free).

Morphology (Eukaryotic Chromosomes)

- **Number:** Species-specific (used in taxonomy).
- **Size:** Constant within species; plants > animals; monocots > dicots.
- **Shape:** Straight/curved rods (S, J, V-shaped depending on centromere).
- **Structure:**
    - **Chromatids** joined at **centromere** with **kinetochores**.
    - **Secondary constrictions** → satellites (sat chromosomes).
    - **Telomeres** → seal chromosome ends, prevent fusion.
    - **Ultrastructure:** DNA wrapped around histones forming nucleosomes → chromatin fibres → chromatids.
- **Chemical composition:** ~35% DNA, 60% proteins, 5% RNA + metal ions/enzymes.
- **Types (based on centromere position):** Metacentric (arms equal, V-shaped), and others.

Functions of Chromosomes

1. Inheritance of traits.
2. Protein synthesis (structural proteins, enzymes).
3. Regulation of metabolism and growth.
4. Control of cell differentiation.
5. Formation of nucleoli (at nucleolar organiser regions).
6. Gene mutations → variation & evolution.
7. Sex determination.
8. Continuity of life through reproduction.

Self Assessment Questions

Objective Type Questions

1. Who first used the term "chromosome"?
a) W. Hofmeister
b) W. Waldeyer
c) W. Flemming
d) Benden

2. The genetic material of prokaryotic chromosomes is usually:
a) Linear, double-stranded DNA with proteins
b) Circular, double-stranded DNA without proteins
c) Linear RNA molecule with proteins
d) Circular, single-stranded RNA
Answer: b) Circular, double-stranded DNA without proteins

3. **Which of the following are extranuclear chromosomes?**
a) Plasmids and nuclear DNA
b) Plastids and mitochondria
c) Viral DNA only
d) Only nucleolus DNA

Short Answer Questions

1. Write any four functions of chromosomes.

2. Differentiate between prokaryotic and eukaryotic chromosomes.

3. What are telomeres? State their function.

Long Answer Questions

1. Describe the history of chromosome discovery and explain the contributions of different scientists.

2. Explain the morphology of eukaryotic chromosomes in detail with reference to number, size, shape, centromere, constrictions, satellites, telomeres, and ultrastructure.

3. Discuss the different types of chromosomes (viral, prokaryotic, and eukaryotic) with their structural features and functions.

Answers
1 – b 2 – b 3 - b

UNIT-2.2

2.2.1Chromosome Variation

Chromosomes are not static entities; they are subject to a range of variations in both number and structure. These variations are termed chromosomal aberrations or chromosome variations, and they have profound implications in genetics, development, evolution, and human disease.

Chromosome variations include changes in number (numerical) and structure (structural). They have biological, medical, and evolutionary implication

Chromosome variation is when a species has a different number or structure of chromosomes than usual. It is a primary source of genetic variation, but it can also cause birth defects, cancer, and evolutionary divergence. Chromosome variations occur either:

- Numerically: changes in chromosome number
- Structurally: alterations in chromosome architecture

These changes may arise spontaneously, due to mutagens, or inheritance. Cytogenetic tools like karyotyping, G-banding, and molecular cytogenetics help identify such variations.

2.2.2Numerical Chromosome Variation

Aneuploidy

Aneuploidy refers to the gain or loss of **one or more individual chromosomes** without affecting the entire set. It results from **nondisjunction** during meiosis I or II, or during mitotic divisions.

Mechanism:

- **Nondisjunction**: Failure of homologous chromosomes (or sister chromatids) to segregate properly.
- Can occur in **meiosis I, meiosis II**, or **mitosis** (mosaic aneuploidy).

Clinical Examples:

- **Down Syndrome** (Trisomy 21): Delayed development, intellectual disability.
- **Turner Syndrome** (Monosomy X): Short stature, gonadal dysgenesis in females.
- **Klinefelter Syndrome** (XXY): Infertility, feminized male phenotype.

- **Patau Syndrome** (Trisomy 13) and **Edward Syndrome** (Trisomy 18): Severe malformations, early mortality.

Effects of Aneuploidy:

- Gene dosage imbalance
- Developmental abnormalities
- Often lethal in early embryogenesis
- Some tolerate aneuploidy better (e.g., plants)

Euploidy

Euploidy involves the gain or loss of entire sets of chromosomes.

Polyploidy

Polyploidy is common in plants and rare in animals. It results in larger cell size, greater biomass, hybrid vigor, and sterility in some cases (e.g., seedless fruits).

Types of Polyploidy:

- **Autopolyploidy**: Multiple chromosome sets from the same species (e.g., autotetraploid potato)
- **Allopolyploidy**: Chromosome sets from different species (e.g., wheat, cotton)

Importance in Agriculture:

- Artificial polyploidy induced using colchicine
- Development of seedless fruits, high-yielding crops

2.2.3Structural Chromosome Variation

Structural variation refers to rearrangements of chromosomal segments, often due to breakage and faulty repair of chromosomes. These changes affect gene positioning, function, and expression.

Deletion (Deficiency)

Definition: Loss of a chromosome segment.

Terminal deletion: Loss from the end

Interstitial deletion: Loss from middle

Example:

- **Cri-du-chat syndrome**: Deletion of short arm of chromosome 5 (5p). Affected infants have a characteristic high-pitched cry, small head, and intellectual disability.

Duplication

Definition: Repetition of a segment of chromosome.

- **Tandem**: Repeated next to original sequence
- **Displaced**: Duplicated segment is elsewhere

Example:

- **Bar eye phenotype** in *Drosophila*: Caused by duplication on the X chromosome.

Effects:

- Can lead to **dosage imbalance**
- May allow evolution of **new gene functions** (gene redundancy)

Inversion

Definition: A chromosome segment is reversed end to end.

Paracentric inversion: Does not involve centromere

Pericentric inversion: Includes centromere

Effects:

- Inverted chromosomes may produce unbalanced gametes during meiosis
- Inversions are suppressors of recombination

Translocation

Definition: Transfer of a chromosome segment to a non-homologous chromosome.

Types:

Reciprocal translocation: Two-way exchange

Robertsonian translocation: Fusion of two acrocentric chromosomes

2.2.4 Summary

Chromosomal variations are changes in chromosome number or structure that impact genetics, development, disease, and evolution.

- **Numerical Variations:**
  - *Aneuploidy* → gain/loss of individual chromosomes (e.g., Down, Turner, Klinefelter syndromes).
  - *Euploidy/Polyploidy* → gain/loss of whole sets of chromosomes; common in plants, useful in agriculture (seedless fruits, hybrid vigor).
- **Structural Variations:**
  - *Deletion* → loss of a segment (e.g., Cri-du-chat).
  - *Duplication* → repetition of a segment (e.g., Bar eye in Drosophila).
  - *Inversion* → segment reversed (paracentric/pericentric).
  - *Translocation* → segment transferred to another non-homologous chromosome.

Self Assessment Questions

Objective Types Questions

1. The gain or loss of one or more individual chromosomes without affecting the entire set is called:
a) Polyploidy
b) Aneuploidy
c) Euploidy
d) Translocation

2. Down Syndrome is caused by:
a) Monosomy X
b) Trisomy 21
c) Trisomy 18
d) Reciprocal translocation

3. Which type of inversion involves the centromere?
a) Paracentric inversion
b) Pericentric inversion

c) Tandem duplication
d) Terminal deletion

Short Answer Questions

1. Differentiate between aneuploidy and polyploidy with suitable examples.

2. Explain how duplications can contribute both to genetic disorders and to evolution.

3. Write short notes on Cri-du-chat syndrome and its chromosomal basis.

Long Answer Questions

1. Describe the types of numerical chromosomal variations. Explain their causes, examples, and biological significance.

2. Discuss structural chromosomal variations in detail. Explain deletion, duplication, inversion, and translocation with suitable examples.

3. Polyploidy plays a major role in plant evolution and agriculture. Explain the types of polyploidy, their significance, and applications in crop improvement.

Answer

1 – b 2 – b 3 – b

UNIT 2.3
CELL DIVISION

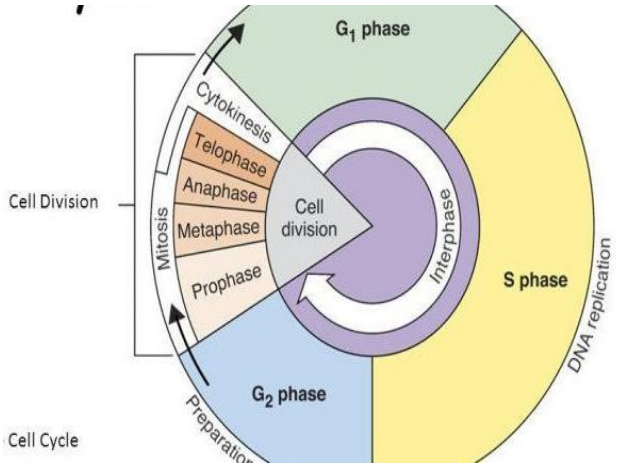


Fig. 2.1 Cell division

2.3.1 Introduction

In both unicellular and multicellular eukaryotes, the cell reproduction is a cyclic process of growth, nuclear division and usually cytoplasmic division called cell cycle.

- The cell cycle is a series of big molecular events that cause a cell to divide and make two daughter cells, each of which has chromosomes that are the same as those of the parent cell.Two main molecular processes take place during the cell cycle are duplication of parental chromosome during S phase and separation of chromosome equally to daughter cell duringM phase.
- Fourphases of Somatic Cell;
- G1(gap1)phase
- S(synthesis)phase
- G2(gap2) phase
- M (mitosis) phase

1.G1(gap1) phase:

- The G1 phase (first gap) is the initial stage of interphase. This is because, on a microscopic level, there isn't much change. But

the cell is quite active biochemically at the G1 stage.It is characterized by a change in chromosome from condensed state to more extended state and series of metabolic events that leads to initiation of DNA replication. During G1 phase, chromatin fibres become slender, less coiled and fully extended and more active for transcription. The transcription results in synthesis of RNAs (tRNA, mRNA and rRNA) ad series of proteins molecules required for initiation of DNA replication.

- The length of G1 phase varies from cell to cell and also the length of G1 phase is more than other three phase in cell cycle.
- G1phaserepresents25-40%ofgenerationtimeofacell.

The G1 phase is a particularly important part of the cell cycle since it is when the cell expands and gathers the building blocks of chromosomal DNA and the proteins that go with it, as well as enough energy to finish the job of copying each chromosome.

During the G1 phase, there is a clear point at which DNA synthesis begins. After the biochemical activities that happen at that point, the cell moves on to division.

2.S (synthesis)phase:

The synthesis phase of interphase is a time when DNA and histones are activelymade.

In the S phase, the number of chromosomes doubles due to DNA replication and related proteins. Some of the histone proteins are made during the G1 phase, while most of them are made during the S phase.

DNA replication is a semi-conservative and discontinuous process that leads to the production of identical pairs of DNA molecules.

After the chromosome has doubled, the sister chromatids are still securely connected to the centromeric region.

Centrosomes are found in the middle of each animal cell. They are next to a pair of rod-like items called centrioles, which are at right angles to each

other. Centrioles help cells divide in an orderly way. Plants and most fungi do not have centrioles.

The centrosome (centriole) also makes a copy of itself during the S phase. The two centrosomes will form the mitotic spindle, which is the structure that moves chromosomes during mitosis.

3.Gap2(gap2) phase:

G2phase comes after Sphase. This phase makes up 10–25% of the time it takes for a cell to grow.

In the G2 phase, chromosomes have two chromatids, which means that the cell has twice as much DNA.

In the G2 phase, the cell replenishes its energy reserves and synthesises proteins essential for chromosomal manipulation.

Some cell organelles are copied, and the cytoskeleton is broken down to make room for the mitotic phase.

There can be more cell proliferation during G2. Before the cell can enter the first stage of mitosis, it must finish getting ready for the mitotic phase.

4.M(mitotic) phase:

After G2 phase comes M phase. At this point, the cell splits into two daughter cells, each with the same number of chromosomes. After the M phase, the cell goes into the G1 phase, and the cell cycle starts over. Some cells, meanwhile, don't go into the G1 phase after mitosis. These cells are called G0 cells.

Mphase has the following sub-phases:

The nuclear membrane breaks down, spindle fibres develop, and DNA condenses into chromosomes (sister chromatids) during prophase.

During metaphase, the sister chromatids line up along the cell's equator by connecting their centromeres to the spindle fibres.

During anaphase, the mitotic spindle pulls sister chromatids apart at the centromere and towards opposite poles of the cell.

During telophase, chromosomes reach opposite poles and unwind into thin strands of DNA. The spindle fibres dissolve, and the nuclear membrane

reappears.

Cytokinesis is the actual splitting of the cell membrane. Animal cells pull apart, while plant cells make a cell plate that becomes the new cell wall. Cells go into the G0 (inactive) phase when they leave the cell cycle and are not getting ready to divide. Some cells stay in G0 phase forever.

2.3.2MITOSIS:MITOTICCELLDIVISION,STAGESAND SIGNIFICANCE

Mitosis is a kind of cell division in which a single haploid cell (n) or diploid cell (2n) splits into two haploid or diploid daughter cells that are the same as the parent cell.

Mitosis takes place in the somatic cells of both plants and animals. During this cell division, the two daughter cells have the same number of chromosomes as the parent cells.

The process of mitosis consists of the following stages or phases:

1. **Interphaseor Interkinesis**
2. **Karyokinesis**
3. **Cytokinesis**

1. Interphaseor interkinesis

Interphase is the time between two cell divisions, from the end of one to the start of the next. It is the longest phase in the cell cycle. Interphase looks dormant but is a metabolically active stage. It is divided into 3 sub-stages, viz. G1-phase, S-phase, and G2-phase.

i. G_1-PhaseorGap-1phase

- **The cell grows in size due to active biosynthesis.**
- **Formation of structural and functional proteins.**
- **Synthesis of mRNA, tRNA, and rRNA takes place.**

ii. S-PhaseorSyntheticphase

Replication of DNA takes place.
Synthesis of histone proteins takes place, which covers DNA.

iii. G_2-PhaseorGap-twophaseorSecondgrowthphase

RNA and protein are synthesized.
Centrioles get replicated (in case of animal cell).
Synthesis of spindle proteins takes place.

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2. Karyokinesis

- Karyokinesis is the division of the nucleus.
- It consists of the following four phases:

i. Prophase

- It is the first visible stage in karyokinesis.
- The chromosomes appear as long coiled threads called chromatids.
- The chromatin becomes shorter, thicker, and visible due to the condensation of DNA.
- The chromatins are now called chromosomes.
- Stainability of nucleus increases.
- Each chromosome starts to split longitudinally into two sister chromatids. These sister chromatids are attached with each other at the centromere.
- The nuclear membrane and nucleolus start to disappear, and by the end, they will completely disappear.

i. Metaphase

- Nuclear membrane and nucleolus completely disappear, and simultaneously, the appearance of spindle fibers occurs.
- Spindle fibers attach to the centromere of the chromosome.
- The chromosomes are arranged on the equatorial plane.
- The process of gathering chromosomes at the equator is called congression, and the plate formed is called the metaphase plate.

ii. Anaphase

- The centromere of each chromosome splits into two sister chromatids and forms two daughter chromosomes.
- The daughter chromosomes are pulled towards the poles due to the contraction of spindle fibers and stretching of interzonal fibers.
- During the polar movement, the chromosomes show different shapes, i.e., J, U, V, L, or I-shaped in appearance.
- At the end of anaphase, each pole will get one set of daughter

chromosomes.

- It is the shortest phase and is also known as the migratory phase.

iii. Telophase

- The daughter chromosomes reach their respective poles, uncoil, and become thin, long, and visible.
- The spindle fibers start disappearing and finally disappear.
- The nuclear membrane and the nucleolus reappear.
- Two nuclei are formed at the end of telophase. Both nuclei have the same number of chromosomes as the parent cell.

It is the last visible stage of karyokinesis and is also known as the reorganization phase.

2. Cytokinesis

- Cytokinesis is thedivisionofthe cytoplasm.
- Inplantcells,cytokinesisoccursbycellplateformation.
- During cytokinesis, many granular matrix formed by the golgibody and endoplasmic reticulum accumulates in the equatorial region. These granular matrix form cell plate. This plate divides the cell and by the end of telophase, cytokinesis is completed.
- Inanimalcells,cytokinesisoccursbycleavageorfurrow formation.

2.3.3 SIGNIFICANCEOFMITOSIS:

Mitosis produces 2 genetically identical cells, so mitosis maintains the genetic stability of organisms.

DNA remains constant, so mitosis keeps the chromosome number constant in a species.

Mitosis helps in the development of multicellular organisms.

Mitosis helps to replace old, dead, or damaged cells by new ones.

It helps in the recovery of wounds and injury of the body by the formation of new cells.

In unicellular organisms like Yeast and Paramecium, mitosis is a means of asexual reproduction.

Mitosis causes maturation and multiplication of germ cells and makes them ready for meiosis.

2.3.4MEIOSIS:MEIOTICCELLDIVISION,STAGES AND SIGNIFICANCE

- Meiosisisacelldivisioninwhichfourhaploidcellsarefo rmedfromasinglediploidcell.
- Itusuallyoccursinreproductiveorgans or gonadsof the organisms.
- Meiosisisalsoknownasreductionalcelldivisi

onbecauseofourdaughtercellsproducedcont
ain half the number of chromosomes than
that of their parent cell.

Summary

1. Prophase-I

- It occupies the longest duration in Meiosis-I.
- It is divided into five sub-stages or sub-phases.

i. Leptotene

- This phase starts immediately after interphase.
- The size of cell and nucleus increases
- The chromosomes appear long, uncoiled thread-like in structure bearing many bead-like

  structures called chromomeres.
- The nuclear membrane and nucleolus remain as it is.

ii. Zygotene

- Homologous chromosomes come closer and starts to pair up along their length.
- The pairing of homologous chromosomes is called Synapsis and the paired homologous

  chromosomes are referred as bivalents.
- The homologous chromosomes are held together by ribonuclear protein between them.

iii. Pachytene

- The chromosome become shorter and thicker.
- Each chromosome of the bivalents splits longitudinally to form two chromatids such that

bivalents is composed of four strands and is known as a tetrad.

- The process of crossing over starts (crossing over; a small fragment of chromosome

exchange between two non-sister chromatids of bivalent by breakage and rejoining).

- Crossing over is the most important genetic phenomenon of meiosis which causes variation

in genetic characters in offspring.

iv. Diplotene

- In this stage crossing over takes place.
- Bivalents (chromatids) repel each other.
- Homologous chromosome (two non-sister

chromatids) begins to separates but separation is not complete, they remains attached to a point with a knot like structure called chiasmata(singular – chiasma).

- The number of chiasmata varies. Depending upon the number of chiasmata, chromosome appears different shape.

1 chiasmata: cross like

2 chiasmata: ring like

Many chiasmata: series of loop

Nuclear membrane and nucleolus begins to disappear.

V. Diakinesis

- The chiasma moves towards the end of the chromosomes (tetrad) due to contraction of chromosomelastly slips over separating the homologous chromosome. This movement of the chiasmata towards the end of chromosome is called terminalization
- By the end of diakinesis the nuclear membrane and nuleolus get completely disappeared and the chromosomes are free in the cytoplasm.
- Spindle fibres begin to form

1.Metaphase-I

- The spindle fibres organized between two poles and get attached to the centromere of chromosomes
- Chromosome moves to equator
- The bivalent chromosomes are arranged in the equatorial plate in such a way that 2 metaphasic plates are formed.

3. Anaphase-I

- Spindle fibrescontracts and pulls the whole chromosomes to the polar region.
- The separated chromosome is known as dyad
- No splitting of chromosomes occurs so the centromere of each homologous chromosome does not divide. Thus, the chromosome number of the daughter nuclei is reduced to half.
- Now the separated chromosome moves toward opposite poles.

4. Telophase-I

- Two groups of chromosome formed at each pole and organized into nuclei.
- The nuclear membrane and nucleolus reappears.
- The chromosomes get uncoiled into chromatin thread
- The spindle fibres disappear totally.

Cytokinesis I

- Cytokinesis may or may not follow nuclear division (meiosis-I

Cytokinesis occurs by cell plate formation method in plant cell and furrowing method in animal cells.

Interphase II or Interkinesis

- The two cells or nuclei thus formed pass through a short stage called interphase-II
- Sometimes, interphase-II is absent.
- It is the resting phase between meiosis-I and meiosis-II.
- It is either very short or may be absen
- No DNA synthesis occurs.

2.3.4Meiosis-II(Homolyticorequationaldivision)

- Meiosis-IIisexactlysimilar to mitosis, soit is also known as meiotic mitosis.
    - In thisdivision,twohaploidchromosomesplitslongitudinallyanddistributedequallyto form 4 haploid cells.
    - Itcompletesin4 stages.

1. **Prophase-II**
2. **Metaphase-II**
3. **Anaphase-II**
4. **Telophase-I**I

1. Prophase-II:

- The dyad chromosomes become thicker and shorter.
- Nuclear membrane and nucleolus disappear.
- Spindle fibers start to form.

2. Metaphase-II:

- The dyad chromosomes come to the equatorial plane.
- Spindle fibers organize between poles and attach to the centromere of the chromosome.

3. Anaphase-II:

- Centromere of each chromosome divides and sister chromatids separate to form two daughter chromosomes.
- Spindle fibers contract and pull the daughter chromosomes apart towards opposite poles.

4. Telophase-II:

- Chromosomes become organized at respective poles into nuclei.
- Chromosomes elongate to form thin networks of chromatin.
- Nuclear membrane and nucleolus reappear.

Cytokinesis-II:

- The result of cytokinesis is four haploid daughter cells (gametes or spores).
- Cytokinesis takes place by cell plate formation in plant cells.
- Successive methods: Cytokinesis followed by each nuclear division resulting in 4 haploid cells. Eg. Monocot plants.
- Simultaneous methods: Cytokinesis occurs only after meiosis-II to form 4 haploid cells. Eg. Dicot plants.
- In animal cells, cytokinesis occurs by furrow formation or depression.

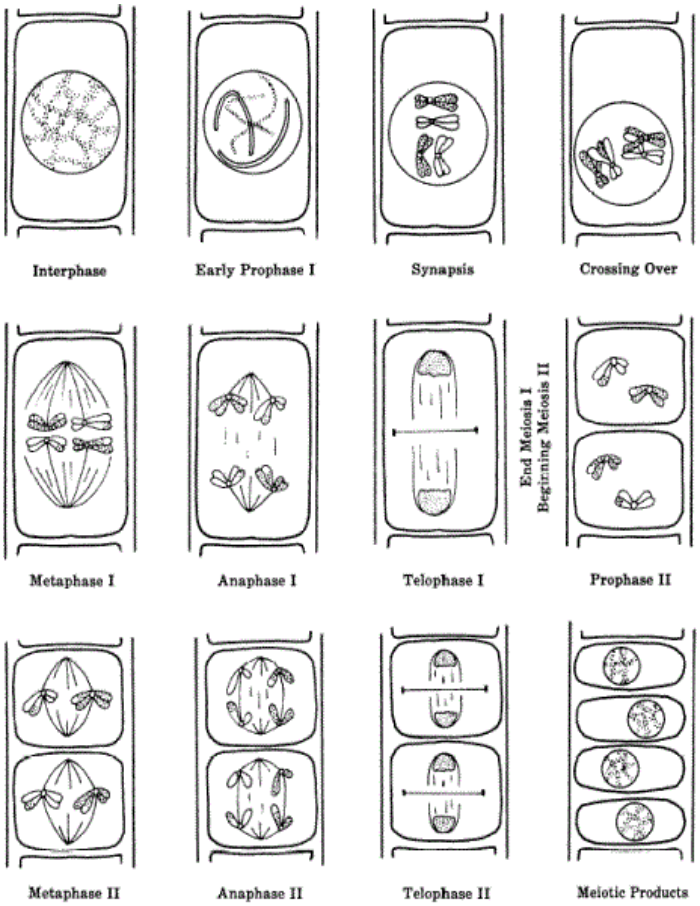

Fig. 2.3 Meiosis

THE IMPORTANCE OF MEIOSIS

1. By lowering the number of chromosomes in the gametes, meiosis aids in maintaining a consistent number of chromosomes.
2. Required for higher animals and plants to reproduce sexually
3. For sexual reproduction, meiosis aids in the creation of haploid gametes and spores.
4. A species' constant number of chromosomes from generation to generation
5. Genetic variations in offspring brought about by crossing over aid in the evolution of organisms.
6. Polypoid forms are formed through mutation caused by failure disjunction in meiosis.
7. During meiosis, maternal and paternal chromosomes are randomly distributed into daughter cells; this form of independent assortment results in variety.

Fig.7.1:Detailedschematicstructureofchromosomes

2.3.5 Summary

Within the nucleus of eukaryotic cells, **chromosomes** are intricately organized structures composed of DNA and proteins, primarily histones, that compact the genetic material into a manageable form. This **chromosome organization** ensures that vast lengths of DNA are efficiently packed while remaining accessible for processes like replication, transcription, and repair. Despite this highly conserved structure, **chromosome variation** occurs both naturally and as a result of mutations. These variations can include changes in chromosome number, such as in Down syndrome (trisomy 21), or structural alterations like deletions, duplications, and translocations, all of which can influence gene expression and phenotype. The transmission and maintenance of chromosomes are tightly regulated through **cell division**, which occurs in two major forms: mitosis and meiosis. Mitosis enables the equal distribution of genetic material into two identical daughter cells, supporting growth and tissue repair, while meiosis reduces the chromosome number by half to produce genetically diverse gametes, ensuring variation in sexually reproducing organisms.

Self Assessment Questions

1. The basic unit of chromatin structure is:

A) DNA helix
B) Histone
C) Nucleosome
D) Chromatid

Which histone protein is NOT part of the nucleosome core particle?

A) H2A
B) H2B
C) H3
D) H1

Euchromatin is:

A) Highly condensed and transcriptionally inactive
B) Loosely packed and transcriptionally active
C) Only found in prokaryotes
D) Present only in heterogametes

In metaphase chromosomes, DNA is most tightly packed in the form of:

A) Solenoid
B) Chromatin fiber
C) Supercoiled loops
D) Scaffolded loops

Telomeres are essential for:

A) Chromosome replication
B) Preventing chromosome end fusion
C) Coding for enzymes
D) Recombination

Module 3

DNA, THE GENETIC MATERIAL

Objectives

 Understand the structure of DNA, including its double-helix formation, nucleotide components, and the role of base pairing in maintaining genetic information.

 Explain the processes involved in DNA replication, repair, and recombination, and how these processes contribute to genetic stability and variability.

 Explore molecular diagnostic techniques used to detect genetic disorders, including PCR, DNA sequencing, and gene editing technologies, and their applications in healthcare.

 Describe the genetic code and how codons in mRNA translate into amino acids during protein synthesis, ensuring the accuracy of gene expression.

 Discuss the mechanisms of gene expression regulation, including transcription, translation, and post-translational modifications, and how they control cellular functions.

UNIT 3.1

STRUCTURE OF DNA

3.1.1 DNA

Deoxyribonucleic acid (DNA) is a nucleic acid that is made up of three components: a deoxyribose sugar, a phosphate, and a nitrogenous base. Deoxyribonucleic acid, DNA is the genetic material via which a cell is defined. It is a long molecule containing unique codes that give instructions for the synthesis of all body proteins.

Structure of DNA

- The structural model of DNA was initially proposed by James Watson and Francis Click.
- They found that DNA is a double-helical structure with two paired DNA strands with complementary nucleotide sequences.
- The double-stranded DNA molecule has two spiral nucleic acid chains that are twisted into a double helix shape. The twisting gives the DNA its compactness.
- DNA is made up of millions of **nucleotides.** Nucleotides are molecules that are composed of deoxyribose sugar, with a phosphate group and a nucleobase that is attached to it.
- Each nucleotide is tightly base paired with a complementary nucleotide on the opposite strand, i.e Adenine (A) paired with Thymine (T) or Guanine (G) paired with cytosine (C), and therefore one strand's sequence acts as a template for the new strand to be formed during replication.
- Nucleotides are bound to each other in strands via phosphodiester bonds forming a sugar-phosphate backbone.
- They form a bond that is between the third carbon atom on the deoxyribose sugar made up of one sugar thus it is designated as the 3′ (three prime) and the fifth carbon atom of another sugar on the next nucleotide as the 5′ (five prime).
- Any part of the sequence can be used to create or recognize its adjacent nucleotide sequence during replication.

- DNA fits within the nucleus by being closely packed into tight coils known as chromatins. The chromatins condense to form the chromosomes during cell division.
- Before DNA replication, the chromatins loosen up giving the replication machinery access to the DNA strands.

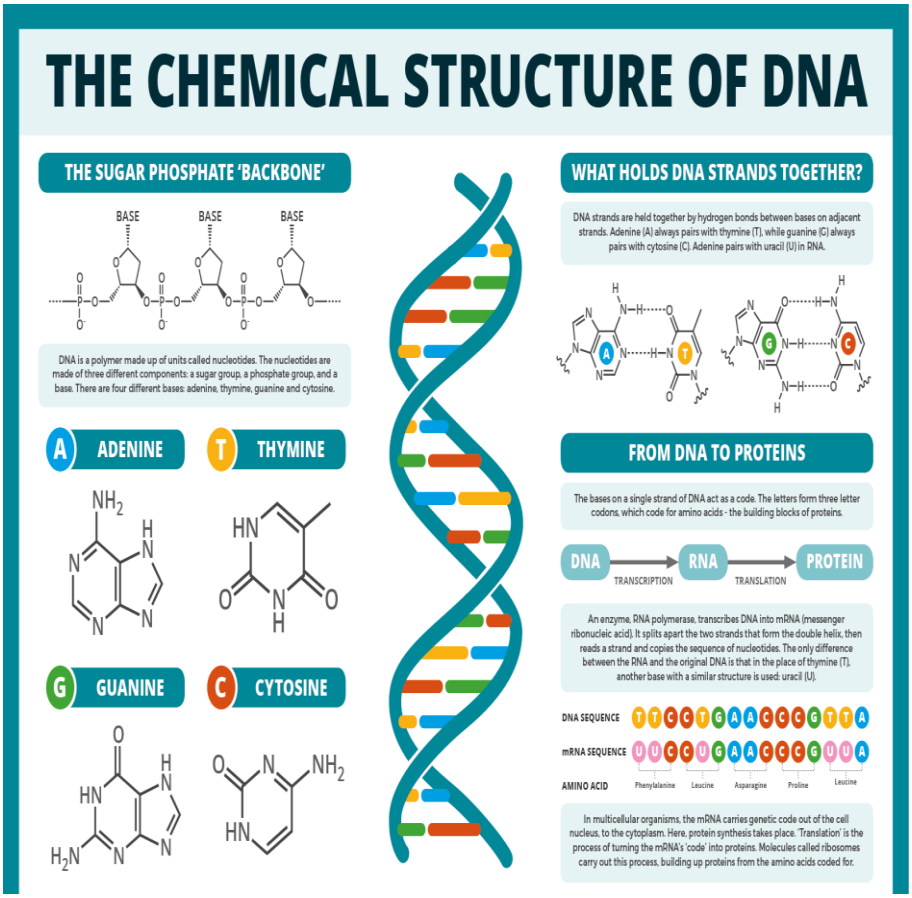

Fig. 3.1 DNA

James Watson and Francis Crick collaborated at the University of Cambridge in England in the 1950s to identify the structure of DNA. Maurice Wilkins and Linus Pauling were among the other scientists who were actively investigating this area. Pauling had used X-ray crystallography to find the secondary structure of proteins. By examining the patterns created by X-rays passing through a substance's crystal, a technique known as X-ray crystallography can be used to study molecular structure. The patterns provide crucial details about the target molecule's structure. Rosalind Franklin, a researcher at Wilkins' lab, was utilising X-ray crystallography to comprehend the structure of DNA. Franklin's

information helped Watson and Crick solve the DNA molecule's mystery. Important details from other researchers, including Chargaff's rules, were also accessible to Watson and Crick. Chargaff demonstrated that two of the four types of monomers (nucleotides) that make up a DNA molecule were consistently found in equal numbers, as were the other two types. This implied that they were inextricably linked. For their efforts in figuring out the structure of DNA, James Watson, Francis Crick, and Maurice Wilkins were granted the Nobel Prize in Medicine in 1962.

The structure of the two different forms of nucleic acids—ribonucleic acid (RNA) and deoxyribonucleic acid (DNA)—will now be discussed. Nucleotides, the building blocks of DNA, are composed of three components: a nitrogenous base, a phosphate group, and a deoxyribose (5-carbon sugar). DNA contains four different kinds of nitrogenous nucleotides. Cytosine (C) and thymine (T) are smaller, single-ringed pyrimidines, while adenine (A) and guanine (G) are double-ringed purines. The nitrogenous base that the nucleotide carries determines its name.

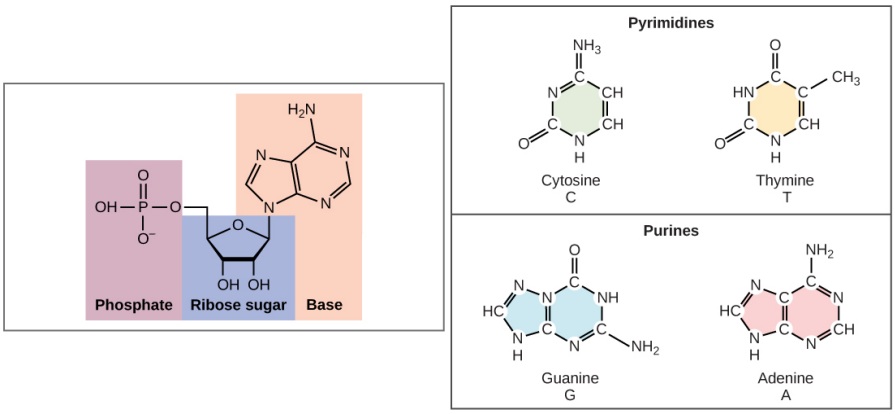

Fig. 3.2 Nitrogenous Bases

3.1.2 Summary

DNA (Deoxyribonucleic acid) is the genetic material that carries instructions for protein synthesis. It is composed of nucleotides, each consisting of a deoxyribose sugar, a phosphate group, and a nitrogenous base.

Watson and Crick (1953), using Rosalind Franklin's X-ray crystallography data and Chargaff's rules, proposed the **double helix model** of DNA. The molecule consists of two complementary strands held together by base pairing: Adenine (A) pairs with Thymine (T), and Guanine (G) pairs with Cytosine (C). The strands form a sugar-phosphate backbone with phosphodiester bonds and run in opposite directions (5′ to 3′).

DNA is tightly packed into **chromatin** and condenses into **chromosomes** during cell division. Its structure allows replication and accurate transmission of genetic information. Watson, Crick, and Wilkins received the Nobel Prize in 1962 for this discovery.

Self Assessment Questions

Objective Type Questions

1. Who proposed the double helix model of DNA in 1953?
a) Watson and Crick

b) Pauling and Franklin

c) Chargaff and Wilkins

d) Avery and Griffith

2. **In DNA, Adenine always pairs with Thymine through:**
a) Three hydrogen bonds
b) Two hydrogen bonds
c) Peptide bonds
d) Phosphodiester bonds

3. **Which component forms the backbone of the DNA molecule?**
a) Nitrogenous bases only
b) Deoxyribose sugar and phosphate group

c) Histone proteins

d) Purines and pyrimidines

Short Answer Questions

1. What are the three components of a DNA nucleotide?

2. Explain Chargaff's rule of base pairing in DNA.

3. Why is DNA described as a double helix?

Long Answer Questions

1. Describe the structure of DNA as explained by Watson and Crick.

2. Discuss the role of Rosalind Franklin and X-ray crystallography in the discovery of DNA structure.

3. Explain how the sugar-phosphate backbone and complementary base pairing contribute to DNA's stability and replication.

Answers

1 – a 2 – b 3 - b

UNIT 3.2

MOLECULAR DIAGNOSTICS

3.2.1 Introduction

Molecular diagnostics is a branch of laboratory medicine that utilizes molecular biology techniques to analyze biological markers in the genome and proteome. These diagnostics are pivotal in detecting and monitoring diseases, understanding genetic predispositions, and tailoring individualized treatment plans. The rapid advancement in molecular diagnostics has revolutionized modern medicine, particularly in oncology, infectious diseases, genetic disorders, and pharmacogenomics.

3.2.2 Historical Background

The foundation of molecular diagnostics lies in the discovery of DNA as the hereditary material and the development of techniques to analyze nucleic acids. Milestones include:

1953: Discovery of the double-helix structure of DNA by Watson and Crick.

1970s: Development of restriction enzymes and recombinant DNA technology.

1983: Invention of polymerase chain reaction (PCR) by Kary Mullis.

1990s: Human Genome Project initiation, leading to the mapping of the human genome.

These advances laid the groundwork for current molecular diagnostic methods.

3.2.3 Principles of Molecular Diagnostics

Molecular diagnostics typically involves the detection of specific sequences in DNA or RNA that may or may not be associated with disease. The process generally includes:

Sample collection and preparation

Nucleic acid extraction

Amplification (e.g., PCR)

Detection and analysis

The target can be a pathogen (in infectious disease testing), a gene mutation (in genetic testing), or gene expression levels (in cancer profiling).

Techniques in Molecular Diagnostics

Several molecular biology techniques are utilized, each with distinct principles and applications.

Polymerase Chain Reaction (PCR)

PCR is a technique used to amplify small segments of DNA. Variants include:

Real-time PCR (qPCR): Enables quantification of DNA.

Reverse transcription PCR (RT-PCR): Used for RNA analysis.

Digital PCR (dPCR): Provides precise quantification of nucleic acids.

Nucleic Acid Hybridization and Microarrays

This involves the hybridization of a labeled probe with complementary nucleic acid sequences. Microarrays allow the simultaneous analysis of thousands of genes.

Next-Generation Sequencing (NGS)

NGS provides high-throughput sequencing of DNA or RNA, allowing comprehensive analysis of genomes, transcriptomes, and epigenomes.

CRISPR-Based Diagnostics

CRISPR-Cas systems, known for gene editing, are being adapted for molecular diagnostics (e.g., SHERLOCK, DETECTR) offering highly sensitive and specific detection.

Isothermal Amplification Methods

Techniques like LAMP (Loop-mediated Isothermal Amplification) allow DNA amplification at a constant temperature, useful in point-of-care diagnostics.

Southern and Northern Blotting

Southern blot: Detects specific DNA sequences.

Northern blot: Detects specific RNA sequences.

Though now largely replaced by advanced techniques, they are foundational in molecular biology.

Applications of Molecular Diagnostics

Infectious Disease Diagnosis

Molecular diagnostics allows rapid and accurate detection of pathogens such as:

Viruses: HIV, HBV, HCV, SARS-CoV-2

Bacteria: Mycobacterium tuberculosis

Parasites and fungi

These methods are more sensitive and specific than traditional culture-based techniques.

Oncology

Used for:

- Identifying genetic mutations (e.g., BRCA1/2 in breast cancer
- Assessing gene expression profiles (e.g., Oncotype DX
- Minimal residual disease detection
- Liquid biopsies using circulating tumor DNA (ctDNA)

Genetic and Genomic Testing

Detects mutations causing inherited disorders such as:

- Cystic fibrosis
- Thalassemia
- Huntington's disease

Includes preimplantation, prenatal, and newborn screening.

Pharmacogenomics

Involves studying how genes affect drug response, aiding personalized medicine. Example:

CYP2C19 genotyping for clopidogrel therapy

Transplantation

Used for HLA typing and monitoring of transplant rejection through donor-derived cell-free DNA.

Forensic and Paternity Testing

DNA profiling used for:

- Crime scene investigations
- Paternity/maternity testing

Neurological and Rare Diseases

- Detection of mutations in genes associated with disorders like:
- Alzheimer's (e.g., APOE)

- Fragile X syndrome
- Muscular dystrophies

Advantages of Molecular Diagnostics

- High sensitivity and specificity
- Early and accurate diagnosis
- Rapid turnaround time
- Ability to detect non-culturable organisms
- Tailored treatment strategies

Challenges and Limitations

- High cost of equipment and reagents
- Need for skilled personnel
- Risk of contamination leading to false result
- Ethical concerns in genetic testing
- Limited accessibility in resource-poor settings

Quality Control and Regulatory Aspects

Regulations ensure test accuracy, reliability, and clinical relevance. Key organizations include:

- FDA (U.S.)
- CE-IVD (Europe
- CLIA (Clinical Laboratory Improvement Amendments)

3.2.4 Summary

Molecular diagnostics is a branch of laboratory medicine that uses molecular biology techniques to study DNA, RNA, and proteins for disease detection, monitoring, and personalized treatment. It plays a key role in oncology, infectious diseases, genetic disorders, pharmacogenomics, and transplantation.

Historical Background

- **1953:** Watson & Crick discovered DNA double helix.
- **1970s:** Restriction enzymes and recombinant DNA technology.
- **1983:** PCR invented by Kary Mullis.
- **1990s:** Human Genome Project mapped human genome.

Principles & Techniques

- Steps: sample collection → nucleic acid extraction → amplification → detection.
- **PCR & Variants:** qPCR (quantification), RT-PCR (RNA analysis), dPCR (precise quantification).
- **Hybridization & Microarrays:** detect multiple genes simultaneously.
- **NGS:** high-throughput genome/transcriptome sequencing.
- **CRISPR-based tests:** SHERLOCK, DETECTR for rapid detection.
- **Isothermal amplification (LAMP):** useful for point-of-care testing.
- **Southern/Northern blotting:** detect DNA/RNA (now mostly replaced).

Applications

- **Infectious diseases:** Detect viruses (HIV, SARS-CoV-2), bacteria (TB), fungi, parasites.
- **Oncology:** Detect mutations (BRCA1/2), gene expression profiling, liquid biopsy (ctDNA).

- **Genetic disorders:** Cystic fibrosis, thalassemia, Huntington's.
- **Pharmacogenomics:** Gene-based drug response (e.g., CYP2C19 for clopidogrel).
- **Transplantation:** HLA typing, rejection monitoring.
- **Forensics & paternity testing.**
- **Neurological/rare diseases:** Alzheimer's (APOE), Fragile X, muscular dystrophy.

Advantages

- High sensitivity & specificity
- Early and accurate diagnosis
- Rapid results
- Personalized treatments

Challenges

- High cost & need for skilled staff
- Risk of contamination → false results
- Ethical issues in genetic testing
- Limited access in low-resource settings

Regulation & Quality Control

- Ensured by **FDA (U.S.), CE-IVD (Europe), CLIA** standards for accuracy, reliability, and clinical safety.

Self Assessment Questions

Objective Type Questions

1. Which technique is considered the foundation of modern molecular diagnostics due to its ability to amplify DNA?

a) Next-Generation Sequencing (NGS)

b) Polymerase Chain Reaction (PCR)

c) Southern blotting

d) CRISPR-Cas

2. In pharmacogenomics, CYP2C19 genotyping is commonly used to guide therapy with which drug?

a) Clopidogrel

b) Insulin

c) Penicillin

d) Aspirin

3. **Which of the following is an advantage of molecular diagnostics over traditional methods?**

a) Longer turnaround time

b) Lower sensitivity

c) Ability to detect non-culturable organisms

d) Requires no specialized training

Short Answer Questions

1. What are the main steps involved in molecular diagnostics?

2. Define pharmacogenomics and give one example of its clinical application.

3. Mention two challenges or limitations of molecular diagnostics.

Long Answer Questions

1. Explain the principle and applications of PCR and its variants (qPCR, RT-PCR, dPCR) in molecular diagnostics.

2. Discuss the role of molecular diagnostics in oncology with suitable examples.

3. Describe the advantages and limitations of molecular diagnostics, hghlighting their impact on modern healthcare.

Answers

1 – b 2 – a 3 – c

UNIT 3.3

GENETIC CODE

The precise arrangement of DNA nucleotides that form three-letter words, or codons, and that dictates the amino acid sequence during protein synthesis is known as the genetic code. To put it another way, the genetic code is the collection of guidelines that living cells use to transfer information from genetic material (DNA or RNA sequences) into proteins (amino acid sequences).

1. Codons are triplets of nucleotides that are used to "read" the genetic code. Stated differently, a codon is a group of three nucleotide bases.

TABLE. 28.1. List of 20 amino acids which take part in protein synthesis

S.No.	*Amino Acid*	*Abbreviation*	*S.No.*	*Amino Acid*	*Abbreviation*
1.	Phenylalanine	Phe	11.	Histidine	His
2.	Leucine	Leu	12.	Glutamine	Gla
3.	Isoleucine	Ile	13.	Asparagine	Asn
4.	Methionine	Met	14.	Lysine	Lys
5.	Valine	Val	15.	Aspartic acid	Asp
6.	Serine	Ser	16.	Glutamic acid	Glu
7.	Proline	Pro	17.	Cysteine	Cys
8.	Threonine	Thr	18.	Tryptophan	Try
9.	Alanine	Ala	19.	Arginine	Arg
10.	Tyrosine	Tyr	20.	Glycine	Gly

Fig. 3.3 Amino Acids

2. One amino acid is encoded by three RNA bases in a triplet code.

3. Twenty amino acids and signals for the start and stop of transcription are represented by the 64 codons.

4. The code creates the amino acids that make up proteins using codons.

5. A single amino acid in a protein structure or a start or stop signal in protein synthesis is indicated by each triplet [codon].

6. The code determines the connection between the amino acid sequence in proteins and the base sequence in nucleic acids (DNA and complementary RNA).

7. The code describes the process by which living things store their genetic information.

3.3.2 Types

There are two types of genetic coding. DNA or RNA codons are two ways that the genetic code might show up. During translation, which makes polypeptides, messenger RNA (mRNA) has RNA codons, which are the codons that are actually "read."But transcription is how each mRNA molecule gets its nucleotide sequence from the gene that matches it [DNA]. A table of codons expressed as DNA is highly beneficial because DNA sequencing has sped up, and most genes are now found at the DNA level before they are found as mRNA or as a protein product.

3.3.3 DNA Codons

TABLE 28.2. The RNA Codons

	Second nucleotide				
	U	C	A	G	
U	UUU (Phe)	UCU (Ser)	UAU (Tyr)	UGU (Cys)	U
	UUC (Phe)	UCC (Ser)	UAC (Tyr)	UGC (Cys)	C
	UUA (Leu)	UCA (Ser)	UAA **STOP**	UGA **STOP**	A
	UUG (Ler)	UCG (Ser)	UAG **STOP**	UGG (Trp)	G
C	CUU (Leu)	CCU (Pro)	CAU (His)	CGU (Arg)	U
	CUC (Leu)	CCC (Pro)	CAC His	CGC (Arg)	C
	CUA (Leu)	CCA (Pro)	CAA (Gln)	CGA (Arg)	A
	CUG (Leu)	CCG (Pro)	CAG Gln	CGG (Arg)	G
A	AUU (Ile)	ACU (Thr)	AAU (Asn)	AGU (Ser)	U
	AUC (Ile)	ACC (Thr)	AAC Asn	AGC (Ser)	C
	AUA (Ile)	ACA (Thr)	AAA (Lys)	AGA (Arg)	A
	AUG (Met) or **START**	ACG (The)	AAG Lys	AGG (Arg)	G
G	GUU (Val)	GCU (Ala)	GAU (Asp)	GGU (Gly)	U
	GUC (Val)	GCC (Ala)	GAC Asp	GGC (Gly)	C
	GUA (Val)	GCA (Ala)	GAA (Glu)	GGA (Gly)	A
	GUG (Val)	GCG (Ala)	GAA (Glu)	GGG (Gly)	G

TABLE 28.3. The DNA Codons

	T		C		A		G		
T	TTT	Phe	TCT	Ser	TAT	Tyr	TGT	Cys	T
	TTC	Phe	TCC	Ser	TAC	Tyr	TGC	Cys	C
	TTA	Leu	TCA	Ser	TAA	STOP	TGA	STOP	A
	TTG	Leu	TCG	Ser	TAG	STOP	TGG	Trp	G
C	CTT	Leu	CCT	Pro	CAT	His	CGT	Arg	T
	CTC	Leu	CCC	Pro	CAC	His	CGC	Arg	C
	CTA	Leu	CCA	Pro	CAA	Gln	CGA	Arg	A
	CTG	Leu	CCG	Pro	CAG	Gln	CGG	Arg	G
A	ATT	Ile	ACT	Thr	AAT	Asn	AGT	Ser	T
	ATC	Ile	ACC	Thr	AAC	Asn	AGC	Ser	C
	ATA	Ile	ACA	Thr	AAA	lys	AGA	Arg	A
	ATG	Met*	ACG	Thr	AAG	Lys	AGG	Arg	G
G	GTT	Val	GCT	Ala	GAT	Asp	GGT	Gly	T
	GTC	Val	GCC	Ala	GAC	Asp	GGC	Gly	C
	GTA	Val	GCA	Ala	GAA	Glu	GGA	Gly	A
	GTG	Val	GCG	Ala	GAG	Glu	GGG	Gly	G

*When within gene; at beginning of gene, ATG signals start of translation.

Fig. 3.4 RNA & DNA Codons

This is how the codons look on the DNA strand that goes from 5′ to 3′.

They are similar to RNA codons, but thymine (T) is present instead of uracil (U). The antisense strand of DNA (3′ to 5′) is what actually makes mRNA.

•Codons are the 64 nucleotide triplets that make up the genetic code. With

three exceptions, every codon codes for one of the 20 amino acids that proteins need to be made. Because of this, the code becomes a little bit unnecessary. Most amino acids are encoded by more than one codon. AUG is a codon that does two things that are linked. It tells the ribosome to add the amino acid methionine (Met) to the growing polypeptide chain and starts the process of translation.

There are two kinds of codons: sense codons and signal codons.

Here is a definition of these:

1. Sense Codon: Sense codons are the codons that code for amino acids. There are 61 sense codons in the genetic code, and each one codes for 20 amino acids.

2. Signal Codons: Signal codons are codons that code for signals when proteins are made. There are four codons that make up the signal. These are AUG, UAA, UAG, and UGA.

There are two types of signal codons: (i) start codons and (ii) stop codons.

(i) Start Codons: A start codon is the codon that starts the process of translation. It is sometimes called an initiation codon since it starts the synthesis of the polypeptide chain.

This codon is AUG. This codon also codes for the amino acid methionine. In some cases, valine (GUG) codes for the start signal. In eukaryotes, methionine is the first amino acid, while in prokaryotes, N-formyl methionine is the first amino acid.

(ii) Stop Codons: These codons tell the polypeptide chain to stop growing. These codons are often called "termination codons" because they tell the polypeptide chain to stop and let go.

Some examples are the stop codons UAA, UAG, and UGA. Stop signal codons, which were once called non-sense codons, do not code for any amino acids.

Release factors are proteins that read the signals that stop or termination codons send. tRNA molecules are unable to interpret stop signals. The releasing factors in prokaryotes include RF1, RF2, and RF3. Factor RFI recognises the stop codons UAA and UAG, while factor RF2 recognises UAA and UGA. RF3's job is to get RF2 and RFI going. In eukaryotes, a single release factor (RF) recognises all three stop codons.

3.3.3 Genetic Code Properties

1. Triplet is the Code: Triplet is the genetic code. The 64 codons in the triplet code are enough to code for 20 amino acids as well as start and stop signals during polypeptide chain production. One amino acid is encoded by three RNA bases in a triplet coding.

2. Universality of the Code: The genetic code is nearly universal. In most genes in plants, animals, and microbes, the same codons are linked to the same amino acids and to the same START and STOP signals.

In the majority of them, an amino acid is assigned one or two of the three STOP codons. There have been some documented exceptions to the rule for the synthesis of nonstandard proteins such pyrolysine and selenocysteine in unicellular eukaryotes and the mitochondrial genome.

3. Commaless is the code:

The genetic code is thought to be commaless. Put otherwise, there are no boundaries between the codons and they are all continuous. When a single base in a commaless code is deleted, the entire amino acid sequence after the deletion point is changed.

4. The Genetic Code is Non-Overlapping:

In a non-overlapping genetic code, each nucleotide is part of only one codon. If a mutation changes a single nucleotide and results in the alteration of just one amino acid, it suggests that codons are read independently, without overlapping. Experiments involving Tobacco Mosaic Virus (TMV) confirmed this, as single base changes affected only one amino acid, reinforcing the concept of a non-overlapping code.

5. The Genetic Code is Unambiguous:

The genetic code consists of 64 codons, with 61 coding for 20 standard amino acids. Importantly, each codon specifies only one amino acid — never more than one. This one-to-one relationship ensures that the code is **non-ambiguous**. If the code were ambiguous, a single codon could produce multiple amino acids, which is not the case in any known organism.

6. The Genetic Code is Redundant (Degenerate):

Although the code is unambiguous, it is also redundant — most amino acids are encoded by more than one codon. For example:

- Tryptophan and methionine are each encoded by a single codon.
- Nine amino acids are encoded by two codons each.
- Isoleucine is encoded by three codons.
- Five amino acids are specified by four codons each.
- Three amino acids are encoded by six different codons.

This degeneracy acts as a buffer against mutations. A change in one base of a codon might still result in the same amino acid being produced, thereby preventing disruption in protein synthesis. This redundancy is systematic and often localized within codon "boxes," except in the cases of serine, leucine, and arginine, where the codons are distributed across multiple boxes. For example:

- All four codons for alanine start with GC.
- Codons for valine typically begin with GU.

3.3.4 Summary

The genetic code is read in a specific direction — from the 5′ end to the 3′ end of mRNA. This directionality is referred to as **polarity**. Reversing

the reading direction changes the codon and, consequently, the amino acid it encodes. For example: Reading **UUG** from left to right specifies one amino acid. Reading the same sequence in reverse as **GUU** results in a different amino acid.

Self Assessment Questions

Objectives Type Questions

1. How many total codons are present in the genetic code?

a) 20

b) 61

c) 64

d) 3

2. Which codon acts as both the start codon and codes for methionine?

a) UAA

b) AUG

c) UAG

d) UGA

3. **What does it mean when the genetic code is said to be "degenerate"?**

a) Each codon codes for multiple amino acids

b) More than one codon can code for the same amino acid

c) Codons overlap in reading

d) Codons have no fixed direction

Short Answer Questions

1. Define a codon. How many codons are there in the genetic code, and how many code for amino acids?

2. Differentiate between sense codons and signal codons with examples.

3. What is meant by the statement "the genetic code is universal"? Give one exception.

Long Answer Questions

1. Explain the role of start and stop codons in protein synthesis. How are they recognized in prokaryotes and eukaryotes?

2. Describe the major properties of the genetic code (triplet, non-overlapping, unambiguous, redundant, polarity, etc.) with examples.

3. Discuss the importance of degeneracy in the genetic code and explain how it helps protect organisms from harmful mutations.

Answers

1 – c 2 – b 3 - b

MODULE 4

UNIT 4.1

GENE

Objectives

- Understand the concept of a gene as the basic unit of heredity and its role in encoding information for protein synthesis.
- Describe the processes of DNA replication, transcription, and translation involved in the transfer of genetic information from one generation to the next.
- Explain the molecular mechanisms underlying gene expression and how the flow of genetic information is regulated within the cell.
- Explore the regulatory elements and factors involved in controlling gene expression, including promoters, enhancers, silencers, and transcription factors.
- Analyze the various modes of gene regulation in prokaryotes and eukaryotes, and discuss the importance of these processes in cellular differentiation and adaptation.

4.1.1 Introduction

Genes are the fundamental units of heredity and biological information that define the characteristics of living organisms. From determining eye color in humans to controlling metabolic pathways in microorganisms, genes orchestrate every biological function. In the field of biotechnology, understanding genes has revolutionized medicine, agriculture, forensics, and synthetic biology. For a B.Sc. Biotechnology student, mastering the concept of genes is foundational for all higher studies and applications.

This essay delves deeply into the structure, function, evolution, and manipulation of genes, offering an integrative view for biotechnology students.

4.1.2 Historical Perspective of Gene Concept

4.1.2.1 The Mendelian Beginning

Gregor Mendel, an Austrian monk, was the first to uncover the basic principles of inheritance using pea plants in the mid-19th century. He coined terms like "factors" (now known as genes) that control traits and showed how they segregate and assort independently.

4.1.2.2 Chromosome Theory and Beyond

In the early 20th century, Sutton and Boveri proposed that genes reside on chromosomes. Later, Thomas Hunt Morgan’s work with fruit flies provided concrete evidence linking genes to chromosomes.

4.1.2.3 Discovery of DNA as Genetic Material

Frederick Griffith's transformation experiment, followed by the work of Avery, MacLeod, and McCarty, confirmed that DNA carries genetic information. Watson and Crick’s elucidation of the double-helix structure in 1953 marked the birth of molecular genetics.

4.1.3 Definition and Structure of Gene

Definition

A **gene** is a specific sequence of nucleotides in DNA (or RNA in some viruses) that encodes the synthesis of a product, typically a protein or RNA molecule. It includes regulatory regions, exons, and introns (in eukaryotes).

Structural Components

- **Promoter:** Region where RNA polymerase binds to initiate transcription.
- **Exons:** Coding segments that are expressed.
- **Introns:** Non-coding segments spliced out during RNA processing.
- **Terminator:** Signals the end of transcription.

Gene Locus and Alleles

- **Locus:** The physical location of a gene on a chromosome.
- **Alleles:** Variants of a gene that produce different traits.

4.1.4 Types of Genes

Structural Genes - Encode proteins or RNAs (e.g., enzymes, structural proteins).

Regulatory Genes - Control the expression of other genes (e.g., repressors, activators).

Housekeeping Genes -Continuously expressed in all cells (e.g., actin, GAPDH).

Oncogenes and Tumor Suppressor Genes- Mutations can lead to cancer (e.g., p53, BRCA1).

Pseudogenes- Resemble functional genes but are non-functional due to mutations.

4.1.4 Summary

Genes are the fundamental units of heredity, encoding proteins or RNAs that determine traits and biological functions. Historically, Mendel discovered inheritance principles, Sutton and Boveri linked genes to chromosomes, and Watson and Crick revealed DNA's double-helix structure. A **gene** consists of promoters, exons, introns, and terminators, located at specific **loci**, with different

alleles producing trait variations. **Gene types** include structural, regulatory, housekeeping, oncogenes/tumor suppressors, and pseudogenes. Understanding genes is essential for biotechnology applications in medicine, agriculture, and research.

Self Assessment Questions

Objective Type Questions

1. Who first discovered the basic principles of inheritance using pea plants?

A) Thomas Hunt Morgan

B) Gregor Mendel

C) Frederick Griffith

D) Watson and Crick

2. Which experiment confirmed that DNA is the genetic material?

A) Mendel's pea plant experiment

B) Griffith's transformation experiment

C) Morgan's fruit fly experiment

D) Hershey-Chase experiment

3. Which part of a gene signals the start of transcription?

A) Exon

B) Terminator

C) Promoter

D) Intron

4. **What are alleles?**

A) Non-coding regions of DNA

B) Variants of a gene producing different traits

C) Genes that encode proteins only

D) Genes that control other genes

5. **Which type of gene is continuously expressed in all cells?**

A) Oncogene

B) Regulatory gene

C) Housekeeping gene

D) Pseudogene

Answers

1 – b 2 – b 3 –c 4 – b 5 -c

Short Answer Questions

1. What is an allele?

2. Name the two main types of genes involved in cancer.

3. What is the function of a promoter in a gene?

Long Answer Questions

1. Describe the historical development of the gene concept.

2. Explain the structural components of a gene.

3. Discuss the types of genes and their importance.

UNIT - 4.2

TRANSFER OF GENETIC INFORMATION

4.2.1 DNA Replication

DNA replication is the process in which new copy of DNA is produced from parent DNA. When two strand of DNA are separated, each strand act as template for the formation of new strand. This process is called DNA replication.

Modes of DNA Replication

- Semiconservative Replication - each strand of DNA duplex used when forming new DNA
- Conservative Replication - original DNA duplex remains intact, new DNA has only new molecules.
- Dispersive Replication - original DNA gets scattered in new DNA, which contains new/old molecules on each strand.

- Replication Stages
- I. The process begins at the origin (OriC), where the initiator protein identifies particular locations within the OriC and opens the helix at the A-T rich area (which has relatively few triple bonds) where the primosome is formed onto strands.
- • When replication proceeds in both directions, two replication forks are created.
- II. The majority of replication time is spent on elongation; pol III adds additional nucleotides to the template strand;

the lagging strand undergoes a more complex process than the leading strand.

- III. Termination: the circular chromosome's termination point is situated across from the origin; DNA gyrase prevents new DNA molecules from entwining; eukaryotic DNA replication: the primary variation in the quantity of DNA replicated

4.2.2 Mechanism of DNA Replication

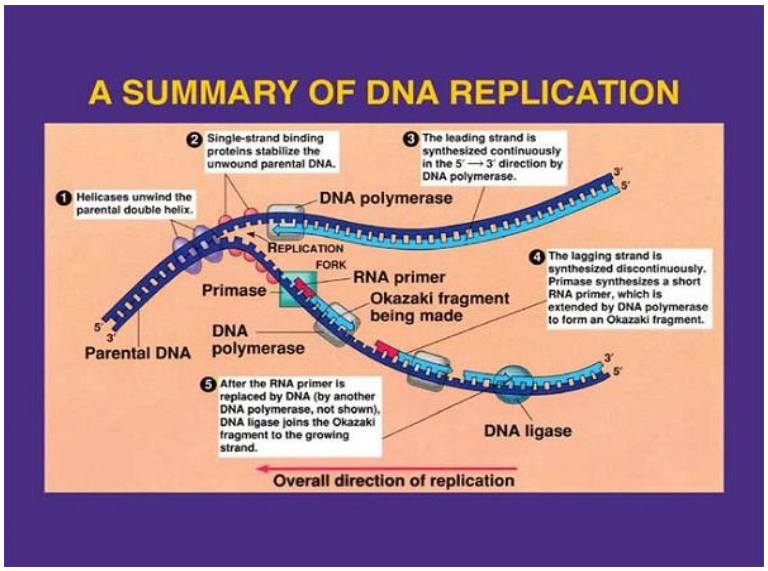

Fig. 4.1 : DNA Replication

4.2.3 Steps of DNA replication

1. Initiation
2. Chain Elongation
3. Termination

1. Initiation

- Replication starts at a specific point of DNA called origin of chromosomal replication (Ori C). In prokaryotic cell, there is one Ori whereas in Eukaryotic cell number of Ori is more than one.

- Ori C is a 245 base pair region of the chromosome and bears DNA sequence elements i.e. 2 types of repeated sequence.
  i. Four 9 mer motif which is the binding site for DnaA.
  ii. Three 13 mermorif which is the initial site of single stranded DNA formation.
- There is a formation of replication bubble due to the presence of origin of replication.
- At first, double stranded DNA starts to separate and uncoil due to breaking of hydrogen bond present between nitrogen bases by helicase. *The unwinding DNA molecule takes place once in every ten nucleotide pair in eukaryotic DNA.*
- Each uncoiled parental DNA strand acts as template DNA strand for the synthesis of new complementary strands.
- When two strands unwind and separate incompletely, they form Y-shape where active synthesis occurs. This region is called replication fork.
- Each separated strands are stabilized by single stranded binding protein.
- As the two strands are separated, supercoiling occurs which is removed by DNA topoisomerase.
- Initiation of replication requires RNA primer, which is a small strand of RNA, synthesized by primase.

2. Chain Elongation

- Chain elongation proceeds from the initiation site by the addition of deoxyribonucleotides at 3'-OH end of the primer by DNA polymerase III.
- DNA Polymerase III forms continuous strand of DNA on 3'→5' template. The continuous strand of DNA is called leading strand.

Since the direction of movement of replication fork and direction of leading strand synthesis are same, leading strand is synthesized continuously after its initiation.

- However, in other template strand 5'→3', there is discontinuous formation of DNA and thus more RNA primer are required for the formation of whole strand. Due to discontinuous formation, smaller fragments are formed, which are called Okazaki fragments. DNA ligase joins these Okazaki fragments to form complete lagging strands. Since the direction of movement of replication fork is opposite to direction of lagging strand synthesis, it cannot be synthesized continuously.
- After completion of chain elongation RNA primer is removed by exonuclease activity of DNA polymerase I and the gap is filled with complementary bases.

3. Termination

- Replication must be terminated to produce two daughter DNA molecule and to regulate and co-ordinate replication with cell division.
- When two replication fork meets Ter-Tus complex, DNA synthesis stops. And the daughter DNA are produced.
- In bacteria DNA is circular. Therefore two interlinked daughter DNA are obtained at completion of replication. Such interlinked DNA are called catenanes. Finally DNA topoisomerase IV cuts one DNA, removes it out of other and finally reseals it. So that two daughter DNA are separated. This process is known as decatenation.

Step 1: Formation of Replication Forks

The double-stranded molecule needs to be "unzipped" into two single strands before DNA can be reproduced. Adenine (A), thymine (T),

cytosine (C), and guanine (G) are the four nucleotides that make up DNA and create pairings between the two strands. Cytosine only bonds with guanine, whereas adenine only pairs with thymine. These base-pair connections must be broken in order to unwind DNA. The enzyme DNA helicase is responsible for this. DNA helicase separates the strands into a Y shape called the replication fork by breaking the hydrogen bonds between base pairs. This region will serve as the template for the start of replication.

Both strands of DNA have directed ends, denoted by 5' and 3'. The side group to which the DNA backbone is connected is indicated by this notation. A phosphate (P) group is joined to the 5' end, and a hydroxyl (OH) group is joined to the 3' end. Since it only moves in the 5' to 3' direction, this directionality is crucial for reproduction. Nevertheless, the replication fork is bidirectional, with the leading strand orientated 3' to 5' and the trailing strand orientated 5' to 3'. To account for the orientation difference, the two sides are thus duplicated using two distinct procedures.

Step 2: Binding primer

Replicating the leading strand is the easiest. A brief segment of RNA known as a primer attaches itself to the 3' end of the DNA strand after the strands have been split. Replication always begins with the primer binding. DNA primase is an enzyme that produces primers.

Step 3: Lengthening

By a process known as elongation, enzymes called DNA polymerases are in charge of producing the new strand. Bacteria and human cells include DNA polymerases of five distinct types. Polymerase III is the primary replication enzyme in bacteria like E. coli, whereas polymerase I, II, IV, and V are in charge of error checking and repair. During replication, DNA polymerase III attaches itself to the strand at the primer location and starts introducing complementary base pairs. The main polymerases involved in DNA replication in eukaryotic cells are alpha, delta, and epsilon. The

newly created strand is continuous since replication on the leading strand proceeds in the 5' to 3' direction.

The lagging strand binds to several primers to start replication. The distance between each primer is just a few bases. After then, DNA polymerase adds Okazaki fragments—fragments of DNA—to the strand in between primers. Because the freshly formed fragments are fragmented, this replication process is discontinuous.

Step 4: Discontinuation

All of the RNA primers from the original strands are eliminated by an enzyme known as exonuclease once the continuous and discontinuous strands have been produced. The proper bases are then used in place of these primers. The freshly created DNA is "proofread" by another exonuclease to identify, eliminate, and replace any mistakes. Okazaki fragments are joined to form a single, cohesive strand by another enzyme known as DNA ligase. Since DNA polymerase can only add nucleotides in the 5′ to 3′ direction, the ends of linear DNA provide a challenge. Telomeres are repeating DNA sequences found at the ends of the parent strands. To stop adjacent chromosomes from merging, telomeres serve as protective caps at the end of chromosomes. The synthesis of telomere sequences at the ends of DNA is catalysed by a unique kind of DNA polymerase enzyme known as telomerase. When finished, the parent strand coils into the well-known double helix shape with its complimentary DNA strand. Replication ultimately results in two DNA molecules, each containing one new strand and one strand from the parent molecule.

4.2.4 Summary

DNA Replication is the process of producing a new DNA copy from a parent molecule, with each original strand serving as a template.

Modes of Replication

1. Semiconservative: Each new DNA contains one original and one new strand.
2. Conservative: Original DNA remains intact; new DNA is entirely new.
3. Dispersive: Original DNA is scattered among new strands.

Stages of DNA Replication

1. Initiation:
    - Begins at origin of replication (OriC).
    - Helicase unwinds DNA; single-stranded binding proteins stabilize strands.
    - Primase synthesizes RNA primers.
    - Supercoiling removed by topoisomerase.
    - Formation of replication fork.
2. Elongation:
    - DNA polymerase adds nucleotides 5'→3'.
    - Leading strand: synthesized continuously.
    - Lagging strand: synthesized discontinuously as Okazaki fragments, later joined by DNA ligase.
    - RNA primers removed and replaced by DNA.
3. Termination:
    - Replication ends at Ter-Tus complex in prokaryotes.
    - Circular DNA interlinks (catenanes) are resolved by topoisomerase IV.
    - Results in two daughter DNA molecules, each with one new and one parental strand.

Key Enzymes: Helicase, primase, DNA polymerases, ligase, topoisomerase, telomerase.

Additional Notes:

- DNA replication is bidirectional.

- Leading strand synthesis is continuous; lagging strand is discontinuous.
- Telomeres protect chromosome ends in eukaryotes.

Self Assessment Questions

Objective Type Questions

1. What is the primary template for DNA replication?

A) RNA

B) Single-stranded DNA

C) Double-stranded DNA

D) Protein

2. Which mode of DNA replication produces two DNA molecules, each with one original and one new strand?

A) Semiconservative

B) Conservative

C) Dispersive

D) Fragmented

3. Which enzyme unwinds the DNA double helix during replication?

A) DNA polymerase

B) Helicase

C) Ligase

D) Primase

4. What is the function of primase in DNA replication?

A) Joins Okazaki fragments

B) Unwinds DNA strands

C) Synthesizes RNA primer

D) Removes supercoiling

5. In which direction does DNA polymerase synthesize the new strand?

A) 3’ → 5’

B) 5′ → 3′

C) Both directions simultaneously

D) Depends on enzyme type

Answers

1 – b 2 – a 3 – b 4 – c 5 -b

Short Answer Questions

1. What is the function of DNA helicase in replication?

2. Define the replication fork.

3. What are Okazaki fragments?

Long Answer Questions

1. Explain the three modes of DNA replication.

2. Describe the steps of DNA replication in prokaryotes.

3. Differentiate between leading and lagging strands during DNA replication.

UNIT- 4.3

GENE EXPRESSION

4.3.1 Introduction

For a cell to function properly, necessary proteins must be synthesized at theproper time. All cells control or regulate the synthesis of proteins frominformation encoded in their DNA. The process of turning on a gene to produceRNA and protein is called **gene expression**. The DNA of a microbial cell consists of genes, a few to thousands, which do notexpress at the same time. At a particular time only a few genes express andsynthesize the desired protein. The other genes remain silent at this moment andexpress when required. Requirement of gene expression is governed by theenvironment in which they grow. This shows that the genes have a property toswitch on and switch off. The Genetic Code that 20 different amino acids constitute different protein. Allaresynthesised by codons. Therefore, synthesis of all the amino acids requiresenergy which is useless because all the amino acids constituting proteins are notneeded at a time. Hence, there is need to control the synthesis of those amino acids (proteins)which are not required. By doing this the energy of a living cell is conserved andcells become more competent. Therefore, a control system is operative which isknown as gene regulation.There are certain substrates called inducers that induce the enzyme synthesis. For example, if yeast cells are grown in medium containing lactose, anenzymelactase is formed. Lactase hydrolyses the lactose into glucose and galactose. Inthe absence of lactase, lactose synthesis does not occur. This shows that lactose induces the enzyme lactase. Therefore, lactase is knownas inducible enzyme. In addition, sometimes the end product of metabolism hasinhibitory effect on the synthesis of enzyme. This phenomenon is called feedback or end product inhibition.From the outgoing discussion it appears that a cell has auto-control mediated by the gene itself. For the first time Francois Jacob and Jacques Monod (1961) at

the Pasteur Institute (Paris) put forward a hypothesis to explain the induction and repression of enzyme synthesis. They investigated the regulation of activities of genes which controls lactose fermentation in E. coli through synthesis of an enzyme, β-galactosidase. For this significant contribution in the field of biochemistry they were awarded Nobel Prize in Medicine in 1965.

4.3.2 Regulation of Gene Expression in Prokaryotes

Gene expression of prokaryotes is controlled basically at two levels i.e. transcription and translation stages. In addition, mRNA degradation and proteinmodification also play a role in regulation. Most of the prokaryotic genes that areregulated are controlled at transcriptional stage

4.3.3 Transcriptional Control in Prokaryotes

In this general strategy in a living organism that chemical changes occur by ametabolic pathway through a chain of reactions. Each step is determined by theenzymes. Again synthesis of an enzyme comes under the control of geneticmaterial i.e. DNA in living organisms. Enzymes (proteins) are synthesisedviatwo steps: transcription and translation.

Transcription refers to synthesis of mRNA. Transcription is regulated at oraround promoter of gene. However, if RNA polymerase has bound, againitcan modulate transcription. By doing so the amount of gene product synthesized is also modulated. The coding region is also called structural gene. Adjacent to it are regulatory regions that control the structural genes. The regulatory regions are composed ofpromoter (for the initiation of transcription) and an operator (where a diffusible regulatory protein binds) regions.The molecular mechanisms for each of regulatory patterns vary widely but usually fall in one of two major groups: negative regulation and positive regulation. In negative regulation an inhibitor is present in the cell and preventstranscription. This inhibitor is called as repressor. An inducer i.e. antagonist repressor is required to permit the initiation of transcription. In a positive regulated system an effector molecule (i.e. a

protein,molecule or molecular complex) activates a promoter. The repressor proteins produce negative control, whereas the activator proteins produce positive control.Since the transcription process is accomplished in three steps (RNA polymerasebinding, isomerization of a few nucleotides and release of RNA polymerase frompromoter region), the negative regulators usually block the binding, whereas the activators interact with RNA polymerase making one or more steps. Fig. 10.19 shows the negative and positive regulation mechanism of the genes. Innegative regulation (A) an inhibitor is bound to the DNA molecule. It must beremoved for efficient transcription. In positive regulation (B) an effector moleculemust bind to DNA for transcription.

i. The Lac Operon Model (Jacob-Monod Model): Jacob and Monod (1961) introduced the operon model to elucidate the regulation of gene expression. An operon consists of multiple different genes arranged in tandem, regulated by a shared regulatory region.

A typical operon comprises repressor, promoter, operator, and structural genes. An operon makes a polycistronic message because all the information from the structural genes is on one molecule of mRNA.

The lac operon is the regulatory mechanism that allows operons to use lactose as a carbon source. Jacob and Monod (1961) were the first to study it in depth. Lactose is a disaccharide made up of glucose and galactose (Fig. 10.20).

The lactose-utilizing system has two categories of components: structural genes (lacZ, lacY, and lacA), whose products are essential for the transport and metabolism of lactose, and regulatory genes (lacI, lacO, and lacP).

The lac operon is made up of these two parts.

One of the most important things about operon is that it lets structural genes that are regulated by regulatory genes work together.

Second, operon demonstrates polarity, which means that the genes Z, Y, and A make the same amount of three enzymes: β-galactosidase (by lacZ), permease (by lacY), and acetylase (by lacA). They are made in a certain order, with β-galactosidase coming first and acetylase coming last.

(i) The Structural Genes: The structural genes make one long polycistronic mRNA molecule. The number of structural genes is the same as the number of proteins. Each structural gene is regulated autonomously and transcribes mRNA molecules individually.

This relies on the substrates that will be used. Three structural genes (Z, Y, and A) are linked to the use of lactose in the lac operon (Fig. 10.21A).LacZ breaks the β-1 → 4 bond in lactose and releases the free monosaccharides. The result is β-galactose.

(ii) The Operator Gene: The operator gene is roughly 28 base pairs long and lies next to the lacZ gene. The base pairs in the operator area are palindromic, which means they have two-fold symmetry from a point (Fig. 10.22). The operator and the promoter region are the same.

In vitro, the lac repressor proteins (a tetramer of four subunits) attach to the lac operator and protect part of the promoter region from DNase digestion.

The repressor proteins attach to the operator and make an operator-repressor complex that stops the transcription of the Z, Y, and A genes by stopping RNA polymerase from being released to start transcription (Fig. 10.21b).

The bacteriophage λ has two operators, OL and OR, that have different base sequences. The lambda repressor (gpcl) is made quickly, attaches to OL and OR, and stops the making of mRNA and the proteins gpcll and gpcII.

(iii) The Promoter Gene: The promoter gene is around 100 nucleotides long and is connected to the operator gene. Gilbert (1974) and Dickson (1975) determined the whole nucleotide sequence of the lac operon

regulatory region. The promoter gene is in the middle of the operator gene and the regulator gene.

The promoter region, like operators, has a palindromic sequence of nucleotides (Figs. 10.22 and 10.23). Proteins with subunits organised in a symmetrical way can recognise these palindromic sequences. This part of the CRP site that binds to a protein termed CRP (cyclic AMP receptor protein) has two folds of symmetry. The CRP gene codes for the CRP.

These components are:

(i) The recognition sequence,

(ii) The binding sequence, and

(iii) An mRNA initiation site.

(iv) The Repressor (Regulator) Gene:

Repressor gene determines the transcription of structural gene.

It is of two types:

i. active

ii. inactive repressors.

It codes for the amino acid of a specific repressor protein.

Once they are made, the repressor molecules spread out from the ribosome and attach to the operator without an inducer. In the end, RNA polymerase can't move and mRNA isn't made. As a result, protein synthesis does not take place. This kind of mechanism happens in the active repressor's inducible system.

Also, when an inducer (like lactose) is present, it attaches to repressor proteins and makes an inducer-repressor complex. This complex is unable to bind to the operator. The repressor changes structure and becomes inactive when a complex forms. As a result, the structural genes can make polycistronic mRNAs, which then make enzymes (proteins).

In the reversible system, however, the regulator gene makes a repressor protein that is inactive and doesn't bind to the operator. As a result, structural genes make proteins.

But a corepressor can turn on the repressor proteins. The repressorco-repressor complex is made up of the co-repressor and repressor proteins. This complex attaches to the operator gene and stops the making of proteins.

Jacob and Monod (1961) could not identify the repressor protein. Gilbert andMuller – Hill (1966) succeeded in isolating the lac repressor from the Lac mutantcells of E. coli inside which the lac repressor was about ten times greater than thenormal cells. The lac repressor proteins have been crystallized. It has a molecular weight of about 1,50,000.It consists of four subunits-each has 347 amino acid residues and molecularweight of about 40,000 Daltons. The repressor proteins have strong affinity for asegment of 12-15 base pairs of operator gene. This binding of repressor blocks the synthesis of mRNA transcript by RNA polymerase. The lac operon is induced when E. coli cells are kept in medium containinglactose. The lactose is taken up inside the cell where it undergoes glycosylationi.e. molecular rearrangement from lactose to allolactose. The galactosyl residue is present on 6 rather than 4 position of glucose (Fig. 10.20). Glycosylation is doneby β-galactosidase that is constitutively present in the cell before induction.Allolactose is the real inducer molecule. The lac repressor protein is an allostericmolecule with specific binding sites for DNA and inducer. Allolacctose binds tolac repressor to form an inducer- repressor complex. Binding of inducer torepressor allosterically changes the repressor lowering its affinity for lacODNA.Consequently repressor is released from lacO due to changes in threedimensional conformations. This is called allosteric effect. After being free lacO allows the RNA polymerase to form mRNA transcript. Here, allolactose acts asthe effector molecule and checks the regulatory protein from binding to lacO (operator) gene.

ii. Positive Regulation of the lac Operon-Catabolic Control: Cyclic AMP (cAMP) is the small molecule which is distributed in animal tissues,and controls the action of many hormones. It is also present in E.

coli and theother bacteria. The cAMP is synthesized by the enzyme adenyl cyclase. (Fig.10.24). Its concentration is directly regulated by glucose metabolism.The Lac operon has an additional positive regulatory control mechanism toavoid the wastage of energy during the synthesis of lactose-utilizing proteinswhile there is adequate supply of glucose.When E. coli grows in a medium containing glucose the cAMP concentration inthe cells falls down. This mechanism is poorly understood. However, the noteworthy point is that cAMP regulates the activity of lac operon (and other operons also).In contrast when E. coli cells are fed with alternate carbon source e.g. succinate,cAMP level increases. The crp locus expresses the enzyme adenylate cyclase thatconverts the ATP to cAMP.
How does cAMP increase the process of transcription, is not known clearly. Ithas been shown experimentally that cAMP binds to the proteins expressed bycrp locus which is known as cAMP receptor protein (CRP) or catabolic activatorprotein (CAP) (Fig. 10.25). Therefore, CRP-cAMP complex binds to the CAP-binding site present on lacpromoter. The CRP -cAMP bound complex promotes the helix destabilizationdownstream, and facilitates RNA polymerase binding. This results in efficientopen promoter formation and in turn transcription.

4.3.4 **Summary**

These units explore the concept of a gene as the fundamental hereditary unit responsible for encoding proteins, which determine an organism's traits. They explain how genetic information is transferred through key processes—DNA replication, transcription into RNA, and translation into proteins—following the central dogma of molecular biology. Furthermore, the regulation of gene expression is examined in depth, highlighting how cells control when and how genes are activated or silenced via mechanisms such as promoters, transcription factors, operons, and epigenetic modifications. This regulation ensures proper development, cellular function, and adaptation across different organisms.

Self Assessment Questions

Objective Type Questions

1. What is the fundamental unit of heredity?

A) Chromosome

B) Gene

C) Protein

D) RNA

2. Which process transfers genetic information from DNA to RNA?

A) Replication

B) Translation

C) Transcription

D) Mutation

3. What is the sequence of information flow in the central dogma of molecular biology?

A) RNA → DNA → Protein

B) DNA → Protein → RNA

C) DNA → RNA → Protein

D) Protein → RNA → DNA

4. Which of the following is NOT involved in gene regulation?

A) Promoters

B) Transcription factors

C) Operons

D) Ribosomes

5. What ensures that genes are activated or silenced at the right time?

A) DNA replication

B) Gene expression regulation

C) Protein folding

D) RNA splicing

Short Answer Questions

1. Define a gene.

2. What is the central dogma of molecular biology?

3.Name two mechanisms by which gene expression is regulated.

Long Answer Questions

1. Explain the processes of DNA replication, transcription, and translation in the flow of genetic information.

2. Describe the role of promoters, transcription factors, and operons in regulating gene expression.

3. Discuss the importance of gene regulation in cellular function, development, and adaptation.

Answers

1 – b 2 – c 3 – c 4 –d 5 - b

MODULE 5

GENETIC VARIATION

Objectives

Understand the concept of genetic variation and the various sources, such as mutation, genetic recombination, and gene flow, that contribute to diversity in populations.

Describe the types of chromosome mutations, including structural changes (e.g., deletion, duplication, inversion) and numerical changes (e.g., aneuploidy), and their effects on organisms.

Explain how chromosome mutations can lead to genetic disorders and provide examples of diseases caused by chromosomal abnormalities.

Discuss the different types of gene mutations (e.g., point mutations, frameshift mutations) and their potential impact on protein function and overall cellular processes.

Analyze the role of gene mutations in evolution and how they contribute to genetic diversity, with a focus on their potential advantages, disadvantages, and relationship to natural selection.

UNIT 5.1

GENETIC VARIATIONS

5.1.1 Introduction

Genetic variation refers to differences in the DNA sequences among individuals within a population. It is the basis for biological diversity and evolution and plays a crucial role in how organisms adapt to their environment, respond to diseases, and evolve over generations. In biotechnology, understanding genetic variation is vital for applications like crop improvement, medical genomics, conservation biology, and personalized medicine.

5.1.2 Definition of Genetic Variation

Genetic variation is defined as the heritable differences in the genetic makeup (DNA sequences) of individuals within a species or population. It can occur at the level of single nucleotides (point mutations), larger segments of DNA (insertions, deletions, duplications), or entire chromosomes.

These variations can influence physical traits (phenotype), susceptibility to disease, behavior, and adaptability to environmental changes.

5.1.3 Types of Genetic Variation

Single Nucleotide Polymorphisms (SNPs)

- The most common type of genetic variation.
- Occurs approximately once every 300 nucleotides in the human genome.
- Can affect protein structure, function, or gene regulation.

Insertions and Deletions (Indels)

- Addition or loss of small segments of DNA.
- Can lead to frameshift mutations in coding regions, altering protein synthesis.
- Can also be neutral if they occur in non-coding regions.

Copy Number Variations (CNVs)

- Large segments of DNA (≥1 kb) are duplicated or deleted.
- Can influence gene dosage and contribute to diseases like autism or cancer.

Structural Variants

- Includes inversions, translocations, and chromosomal rearrangements.
- May disrupt gene function or regulatory regions.

Microsatellites and Minisatellites (STRs and VNTRs)

- Short tandem repeats (STRs) are repeating sequences of 2–6 base pairs.

- Highly polymorphic and used in DNA fingerprinting.

5.1.4 Sources of Genetic Variation

Mutation

- Spontaneous mutations occur naturally due to errors in DNA replication.
- Induced mutations are caused by mutagens such as UV radiation, chemicals, or viruses.
- Mutations can be beneficial, neutral, or harmful.

Genetic Recombination

- Occurs during meiosis when homologous chromosomes exchange segments (crossing over).
- Leads to new combinations of alleles in gametes.

Independent Assortment

- During meiosis, chromosomes assort independently, creating diverse genetic combinations in offspring.

Gene Flow (Migration)

- Movement of alleles between populations through migration.
- Increases genetic variation within populations and reduces differences between populations.

Genetic Drift

- Random changes in allele frequency due to chance events, especially in small populations.
- Can reduce genetic variation over time.

5.1.5 Summary

Genetic variation refers to the heritable differences in DNA sequences among individuals within a population, forming the basis of biological diversity, evolution, and adaptability. It can occur at multiple levels, including single nucleotides (SNPs), small insertions or deletions (Indels), large DNA segments (copy number variations), structural rearrangements (inversions, translocations), and repetitive sequences (microsatellites and minisatellites). These variations arise from sources such as spontaneous or induced mutations, genetic recombination during meiosis, independent assortment of chromosomes, gene flow between populations, and genetic drift in small populations. Genetic variation is essential for evolution, adaptation to environmental changes, and has important applications in biotechnology, including crop improvement, medical genomics, conservation, and personalized medicine.

Self Assessment Questions

Objective Type Questions

1. What is genetic variation?

A) Differences in phenotype only

B) Heritable differences in DNA sequences among individuals

C) Differences in environmental conditions

D) Random mutations that cannot be inherited

2. Which of the following is the most common type of genetic variation?

A) Copy Number Variation (CNV)

B) Single Nucleotide Polymorphism (SNP)

C) Structural Variant

D) Microsatellite

3. Which process during meiosis leads to new combinations of alleles in gametes?

A) Independent assortment

B) Gene flow

C) Genetic recombination (crossing over)

D) Genetic drift

4. Gene flow increases genetic variation by:

A) Random mutations within a population

B) Migration of alleles between populations

C) Independent assortment of chromosomes

D) Chromosomal duplication

5. Which source of genetic variation is more influential in small populations and can reduce diversity over time?

A) Mutation

B) Genetic drift

C) Gene flow

D) Recombination

Short Answer Questions

1. Define genetic variation.

2. What is a single nucleotide polymorphism (SNP)?

3. Name two sources of genetic variation.

Long Answer Questions

1. Explain the different types of genetic variation with examples.

2. Describe the role of mutation, recombination, and independent assortment in generating genetic variation.

3. Discuss the significance of genetic variation in evolution and biotechnology.

Answers

1 – b 2 –b 3 – c 4 – b 5 - b

UNIT 5.2

CHROMOSOME MUTATION

5.2.1 Introduction

- Inheritance is based on genes that are faithfully transmitted from generation to generation in microorganisms (in all organisms). Despite the biochemical mechanisms that facilitate such transmissions faithfully, **sudden change in the sequence of nucleotide bases in genes can and do occur.** These changes, called mutations introduce variability into the gene pool and are heritable.
- The word mutation is derived from Latin **word mutate, meaning to change**. Thus, mutations are the permanent changes in the genes. Mutation in the broad sense include all the changes in the hereditary material which can alter the character of any individual.
- Mutation is an **important phenomenon because it is the ultimate source of genetic variation** and provides the raw material for evolution. All the genes would exist in only one form and no mutants (alleles) would be produced without mutation. Thus the mutations are essential to provide new genetic variability to allow microorganisms to evolve and adapt to environmental changes.
- At the same time, **if mutations occurred too frequently they would totally disrupt the transmission of genetic information from generation to generation.**

5.2.2 Mutation - Definition

✓ Mutation refers to sudden heritable change in the phenotype of an individual. In the molecular term, mutation is defined as the permanent and relatively rare change in the number or sequence of nucleotides.

✓ A mutation is a change in gene potentially capable of being transmitted —Synder

- ✓ **A mutation is a sudden and discontinuous change in a gene occurring rarely for any particular gene and capable of producing a change great or small in some part of body.** —Collins

5.2.3 Discovery

- ✓ Mutation was first discovered by Wright in 1791 in male lamb which had short legs.
- ✓ Later on mutation was reported by Hugo de Vries in 1900 in Oenothera, Morgan (1910) in Drosophila (white eye mutant) and several others in various organisms.
- ✓ The term mutation was coined by de Vries.

The chromosomal mutation is the process of change in the chromosomes as a result of rearranged chromosome parts and changes in the number of individual chromosomes or chromosome set present in the genome.

- Chromosome mutations can be detected either by microscopic examinations or genetic analysis, or both. This distinguishes chromosomal mutations from gene mutations that cannot be detected microscopically.
- Chromosomal mutations are the result of certain accidents or irregularities in the chromosomes at the time of cell division, crossing over, or fertilization. These incidents cause alterations in the morphology and number of chromosomes.
- Chromosomal mutations are also called chromosomal aberrations, chromosomal abnormality, or chromosomal disorders, all indicating a possible alteration in the morphology and structure of the chromosome.
- Chromosomal mutations lead to abnormalities in the function of the cell and organism, as chromosomal mutations can result in abnormal gene numbers or positions.
- These are known to cause different genetic diseases that can be hereditary and are transferred from one generation to another.

- These mutations, however, do not always affect the functioning of the cell as some mutations might affect regions of chromosomes that do not make up the genetic makeup of the organism.
- Even though gene mutations are usually more severe than chromosomal mutations, some chromosomal mutations might result in gene mutations.

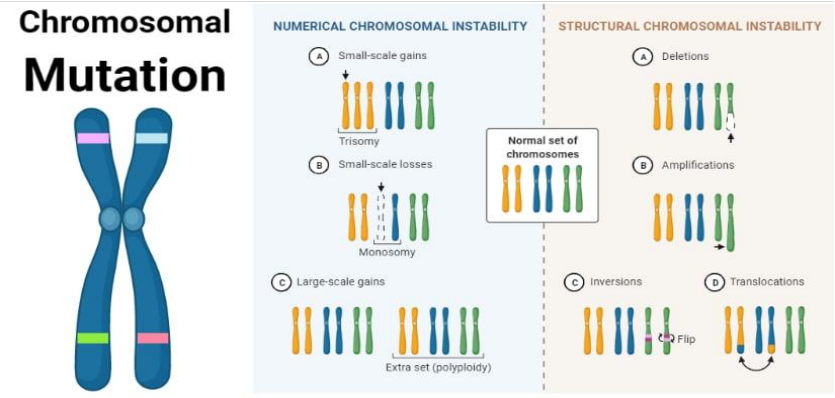

Fig. 5.1 Chromosomal Mutation

Causes

- Chromosomal mutations take place either due to the changes in the structure of the chromosomes or due to the abnormality in the chromosome number.
- In both of the cases, the most important cause includes the mistakes during chromosome division or crossing over.
- Some portions of chromosomes might lag during anaphase, which causes them to be lost during nuclei reorganization. Some might even be digested by nucleases.
- In the case of duplication, a portion or the entire chromosome might be duplicated as a result of some mistake during chromosome division.
- Most of the structural mutations occur at crossing over, where the abnormal breaking or separation of the chromosome can result in chromosomal mutations.
- In the case of numerical mutations, irregularities in nuclear division or accidents or mutagens can change the genomes of the cells.

- The formation of aberrant genomes in living organisms is caused by irregularities during meiosis. These can occur either naturally or are generated artificially.
- Mutagens like radiation and chemical molecules might also be involved in chromosomal mutations.
- Chromosomal mutations are hereditary if present in the heritable part of the chromosomes and thus can be passed down from one generation to the other.
- Depending on the cause, extent, and position of the mutation, chromosomal mutations can produce a wide range of effects in the organisms.

5.2.4 Types of Chromosomal Mutation

Chromosomal mutations can be broadly categorized into two groups; chromosomal mutations I and chromosomal mutations II.

1. Chromosomal Mutations I

Chromosomal mutations I include structural mutations that arise as a result of alterations in the structure of the chromosomes. Structural changes in chromosomes usually occur due to the property of the chromosomes to form pairing and undergo contortions, as well as due to the tendency to break and form sticky ends. Structural mutations are further divided into different types depending on the mechanism of the process;

a. Inversion

- Inversion is a type of structural mutation where a part of chromosomes or a set of genes rotates by 180° on its own axis.
- There is no net loss or gain of genes but simply a rearrangement of the sequence. A part of the chromosome is broken and then rejoined in a different direction.
- Inversion mutation can be detected cytologically in the meiotic nuclei by the detection of an inversion loop in the paired homologs.
- The genetic behavior of the changed chromosome depends on the location of the centromere from the site of inversion.

- If the inversion doesn't include the centromere, it is called paracentric inversion, while if the centromere is included, it is called pericentric inversion.
- If identical inversion occurs in each member of the homologous chromosome, normal distribution of the chromosome occurs. However, inversion heterozygotes might produce deletions and duplication during crossing over.

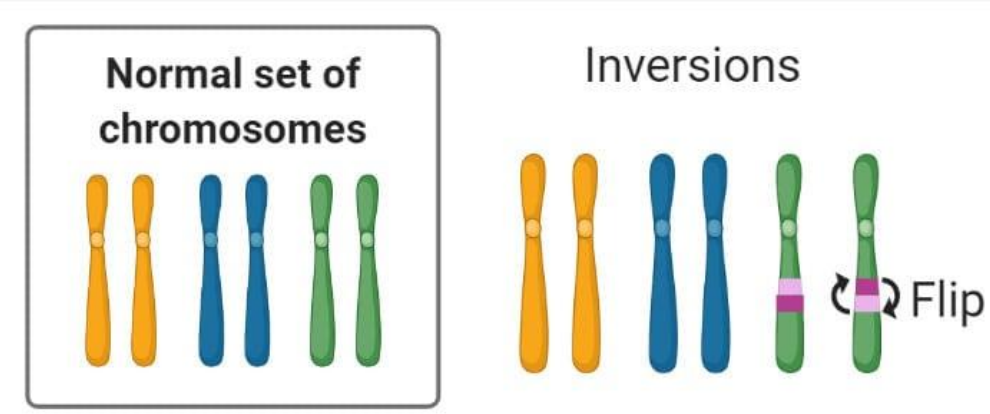

Fig. 5.2 Invesion

Examples of Inversion

- An example of chromosomal inversion can be observed in the insect, *Coelopa frigid*, where the chromosomal inversion results in the production of differences in phenotype.
- Chromosomal inversion occurs only in the larger species as the smaller species cannot survive the mutation.
- The changes observed as a result of the mutation are a three-fold difference in size in males. The change heterokaryotype has higher viability than the original structure.
- Thus, inversion mutation in the species acts as an evolutionary asset in the species, which increases its fitness in the ecosystem.

b. Deletion

- Deletion is a type of structural mutation that occurs due to the loss of a part of a chromosome as a result of the breakage of the chromosome.

- The deletion occurs due to the loss of a portion of a chromosome, usually due to lag during anaphase and digestion by nucleases.
- Chromosomes that have undergone deletion cannot revert back to normal and, if transmitted to the next generation, can be hereditary.
- Deletion can either be terminal or intercalary. The terminal deletion occurs due to the loss of the terminal section of a chromosome. This involves a single break n the chromosome.
- The intercalary deletion occurs due to the loss of an intermediate section of the chromosome. This involves two breaks on either end of the deleted section.
- Deletion is lethal in the case of homozygous chromosomes, and even in heterozygous chromosomes, there are slim chances of survival. Heterozygous chromosomes can, however, survive smaller deletion.

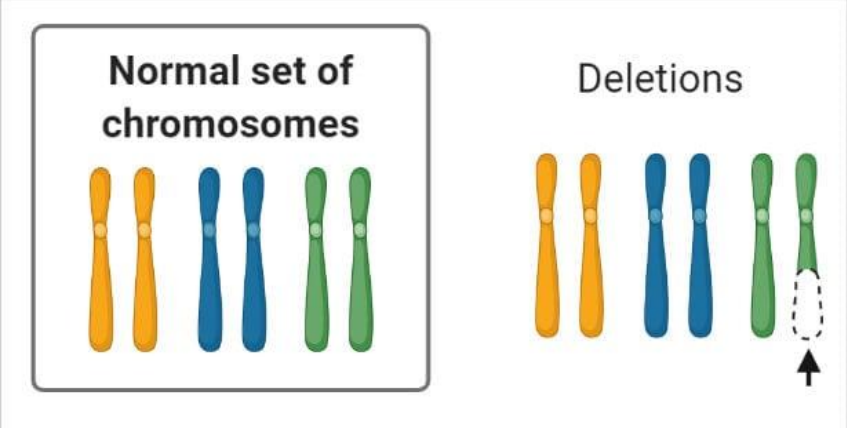

Fig. 5.3 Deletion

Examples of Deletion

- Deletion of the short arm of chromosome 5 in humans results in a distinctive cat-like cry in babies.
- It is also known as the French name ‘cri du chat’ syndrome, indicating the cat-like cry.
- These individuals tend to be mentally slow with an IQ below 20 and have different forms of malformation in the larynx, moon faces, saddle noses, and small mandibles.
- The syndrome can be inherited from one generation to another can might even affect the ears and the size of the head.

c. Duplication/Amplifications

- Duplication is a type of structural mutation where a part of a chromosome is present in excess of the normal composition.
- The genes present in a cell might exist in more than two doses as a result of duplication.
- When the duplication is present in only one of the two homologous chromosomes, the chromosome with the supplicated segment forms a loop to match the position of the homologous regions.
- Duplication in the chromosome can occur in multiple ways; tandem duplication, reverse tandem duplication, displaced duplication, transposed duplication, and extra-chromosomal duplication.
- Tandem duplication occurs when the duplicated region is present just beside the normal corresponding section of the chromosome.
- Reverse tandem duplication occurs when the sequence of genes in the duplicated region is just the reverse of the normal sequence.
- In displaced duplication, the duplicated region is not situated next to the normal section.
- When the duplicated regions become attached to a different nonhomologous chromosome, it is called transposed duplication.
- An extra-chromosomal duplication is formed when the duplicated part has a centromere, resulting in an independent chromosome.

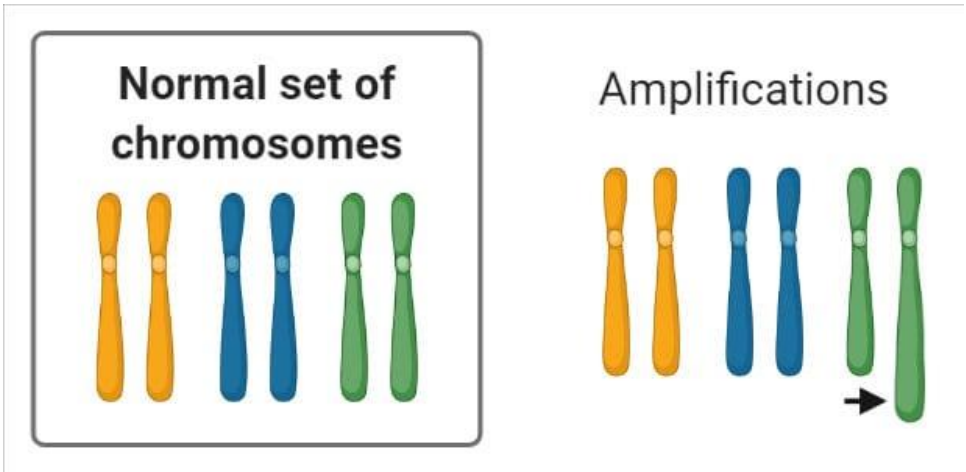

Fig. 5.4 Amplification

Examples of Duplication/Amplifications

- The duplication of a segment of the X-chromosome, called section 16A, in *Drosophila* is a classic example of duplication.
- Section 16A codes for the bar trait in *Drosophila,* which is characterized by a narrower, oblong, bar-shaped eye with a few facets.
- Each duplicated section 16A intensifies the bar phenotype, which increases the narrowing effect.
- The duplication of the section acts as a genetically dominant factor, and the phenotype intensifies if the duplicated genes occur on the same chromosome.

d. Translocation

- Translocation is a type of structural mutation resulting from the shift or transfer of a part of a chromosome or a set of genes to a nonhomologous chromosome.
- There is no net gain or loss of chromosomes or genes during translocation but a rearrangement.
- There are three different types of translocations depending on the pattern of rearrangement; simple translocation, shift translocation, and reciprocal translocation.
- Simple translocation involves a single break in the chromosome where the broken piece then attaches to one of the ends of a nonhomologous chromosome.
- Shift translocation involves the insertion of a broken segment of a chromosome interstitially in a nonhomologous chromosome.
- Reciprocal translocation is the exchange of a translocated segment with another in order to create two translocated chromosomes.
- The exchange of chromosome sections between nonhomologous chromosomes generates new linkages with possible new phenotypes.

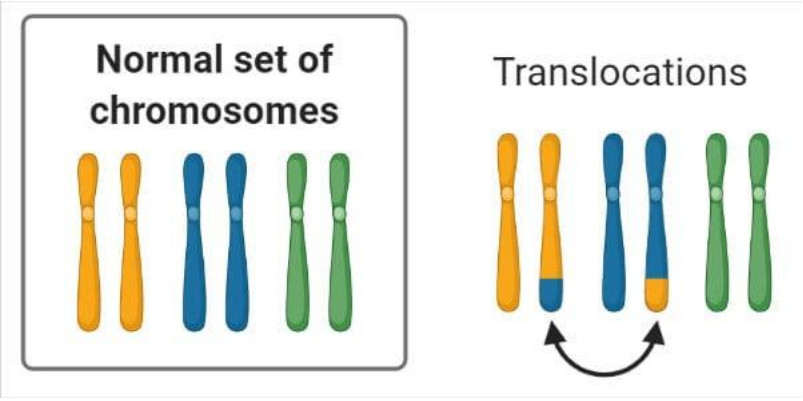

Fig. 5.5 Transloaction

Examples of Translocation

- A rare series of reciprocal translocation can be observed in *Oenothera* involving all 7 of the chromosome pairs.

- Multiple translocations exist within the set, which produces different lethal combinations.
- The translocation results in a ring-like structure of the chromosome where the gametes usually do not survive the mutation.
- Viable gametes are formed only when the linkages are alternate disjunction from the ring structure.

2. Chromosomal Mutations II

Chromosomal mutations II include mutations that are caused by the alterations in the number of chromosomes in a cell. The change in the number of whole chromosomes is called heteroploidy. It produces phenotypic changes, modifications of phenotypic ratios, and alteration of linkage groups. Heteroploidy can be further divided into two different categories depending on the changes in the entire set of chromosomes or in the single whole chromosome.

a. Aneuploidy

- Aneuploidy is a type of mutation that changes parts of a chromosome set, resulting in either the loss of one or more chromosomes or the addition of chromosomes.
- Aneuploidy resulting from the loss of chromosomes is called hypoploidy, whereas that due to the addition of chromosomes is called hyperploidy.
- Hypoploidy usually occurs due to the loss of a single chromosome (monosomy) or due to the loss of a pair of chromosomes (nullisomy).
- Hyperploidy, in turn, might involve the addition of a single chromosome (trisomy) or the addition of a pair of chromosomes (tetrasomy).
- Aneuploids are caused as a result of nondisjunction during mitosis or meiosis.
- If aneuploidy occurs in the gametes of plants, those do not survive, but in animals, some genetic imbalance leading to higher mortality or reduced fertility might occur.

Examples of Aneuploidy

- Down's syndrome is an example of aneuploidy which is associated with a trisomic condition for one of the smallest human autosomes (chromosome no. 21).
- Down's syndrome is the most common chromosomal abnormality in live births and exhibits 50 different physical characteristics.

- The characteristics can range from mild and moderate mental retardation to internal epicanthal folds and swollen tongue.
- The main cause of the trisomy is the nondisjunction of chromosome 21 during oogenesis. Down syndrome in children can also occur due to nondisjunction chromosome pairs in spermatogenesis.

b. Polyploidy

- Polyploidy is a type of euploidy (changes in the entire set of chromosomes) where an organism has more than two sets of genomes (2x).
- Polyploidy includes different combinations like triploid, tetraploid, pentaploid, hexaploid, and octoploid.
- Polyploidy higher than tetraploid is not common in natural environments, but it can be observed in some crops and ornamental flowers.
- Polyploidy can be further divided into three groups; autopolyploids, allopolyploids, and autoallopolyploids.
- Autopolyploids are polyploids that consist of the same basic set of chromosomes but multiplied to form multiple sets.
- Allopolyploids are the polyploids that result from the doubling of chromosome number in a hybrid from two different species.
- The most common morphological effect of polyploidy is gigantism that is commonly seen in large-sized pollen and cells. Besides, it also reduces the fertility of plants to varying degrees.

Examples of Polyploidy

- An example of polyploidy can be observed in ‘doob’ grass (*Cynodondactylon*) which is mostly cultivated in South Asia.
- It is triploid and sterile but can be propagated vegetatively. Polyploidy results in large-sized plants that have decreased osmotic pressure with increased water content.
- The rate of cell division is low, and thus, the plant growth rate also decreases. These also have reduced auxin content which decreases their rate of respiration.

5.2.5 Summary

Mutations and Chromosomal Mutations

Mutation:

- Mutations are permanent, heritable changes in genes or nucleotide sequences.
- They introduce genetic variability, essential for evolution, but excessive mutations can disrupt heredity.
- First observed by Wright (1791) in lambs; term coined by Hugo de Vries (1900).

Chromosomal Mutations:

- Involve changes in chromosome structure or number.
- Can be detected cytologically or genetically.
- Causes: errors during chromosome division, crossing over, fertilization, or exposure to mutagens (radiation/chemicals).
- Can be hereditary if occurring in germline cells.

Types of Chromosomal Mutations I (Structural):

1. Inversion: Chromosome segment rotates 180°, can be paracentric (not including centromere) or pericentric (including centromere).
2. Deletion: Loss of a chromosome segment; terminal or intercalary; can be lethal in homozygotes.
3. Duplication/Amplification: Extra copies of chromosome segments; types include tandem, reverse tandem, displaced, transposed, and extra-chromosomal.
4. Translocation: Segment shifts to a nonhomologous chromosome; types include simple, shift, and reciprocal translocations.

Types of Chromosomal Mutations II (Numerical):

1. Aneuploidy: Changes in part of chromosome set; includes hypoploidy (loss) and hyperploidy (gain). Examples:
    - Down's syndrome: Trisomy of chromosome 21 due to nondisjunction.

2. Polyploidy: Changes in the whole set of chromosomes (more than 2x); types include autopolyploidy, allopolyploidy, and autoallopolyploidy.
    - Effects: gigantism, reduced fertility, slower growth.
    - Example: triploid ‘doob’ grass (Cynodon dactylon).

Significance:

- Mutations provide raw material for evolution and adaptation.
- Structural mutations can alter gene function or produce new phenotypes.
- Numerical mutations affect organism viability, development, and morphology.

Self Assessment Questions

Objective Type Questions

1. Who first observed mutations and in which organism?
A) Hugo de Vries in Oenothera
B) Wright in male lamb
C) Morgan in Drosophila
D) Mendel in pea plants

2. Which type of chromosomal mutation involves a 180° rotation of a chromosome segment?
A) Deletion
B) Inversion
C) Duplication
D) Translocation

3. What is the cause of Down’s syndrome?
A) Deletion of chromosome 5

B) Nondisjunction of chromosome 21
C) Reciprocal translocation
D) Inversion of X chromosome

4. Which chromosomal mutation results in extra copies of a chromosome segment?
A) Deletion
B) Translocation
C) Duplication/Amplification
D) Inversion

5. Polyploidy refers to:
A) Loss of a single chromosome
B) Changes in the whole set of chromosomes
C) Exchange of segments between nonhomologous chromosomes
D) Rotation of a chromosome segment

Short Answer Questions

1. Define chromosomal mutation.

2. What is aneuploidy and give an example.

3. Name two causes of structural chromosomal mutations.

Long Answer Questions

1. Explain the different types of structural chromosomal mutations with examples.

2. Describe numerical chromosomal mutations and their effects on organisms.

3. Discuss the significance of chromosomal mutations in evolution and heredity.

Answers

1 – b 2 – b 3 – b 4 – c 5 - b

UNIT 5.3

ROLE OF GENE MUTATION

5.3.1 Mutations

Mutations have certain general characteristics which are summarised as below:

(i) Mutations are random i.e., they may come in a gene. However, some gene show higher mutation rates than others.

(ii) Mutations are generally lethal or harmful to the organism, a small proportion (0.1%) of all the induced mutations are useful.

(iii) Mutations are recurrent, i.e., the same mutation may occur repeatedly or again and again.

(iv) Induced mutations generally show pleiotropy (single gene affecting two or more different characters) often due to mutations in closely linked genes.

(v) Mutations provide the raw material for evolution.

(vi) Origin of mutation is unpredictable and haphazard.

(vii) Mutations are reversible i.e., an allele that arose through mutations of a gene can in turn mutate back to the original form of the gene. This is known as back mutation.

This can be represented as follows:

A and a = alleles

$A \underset{v}{\overset{u}{\rightleftharpoons}} a$ u and v = represents the mutation rates in the two directions.

(viii) A number of different mutational possibilities exists for any particular gene. Different mutations at the same locus give rise to multiple allelic series. For example, in Drosophila, the sex-linked white-eye locus (w) is represented by a large number of different alleles. These include eosin and apricot as well as white and the wild type allele.

(ix) Some genes increase the spontaneous mutations rates of some other genes of the genome, such genes are called mutator genes. Some genes are termed as anti-mutator genes which suppress or prevent the mutation of other genes.

(x) Many agents, both physical and chemical increase the frequency of mutations, they are said as mutagenic agents.

(xi) Some mutant alleles do not mutate back. They do not exhibit reverse mutation. Such mutant alleles are believed to be formed by deletions.

Some of the important characteristics of mutations are briefly presented below:

i. Nature of Change:

Mutations are more or less permanent and heritable changes in the phenotype of an individual. Such changes occur due to alteration in number, kind or sequence of nucleotides of genetic material, i.e., DNA in most of the cases.

ii. Frequency:

Spontaneous mutations occur at a very low frequency. However, the mutation rate can be enhanced many fold by the use of physical and chemical mutagens.

The frequency of mutation for a gene is calculated as follows:

Frequency of gene mutation = M / M + N

where, M = number of individuals expressing mutation for a gene, and

N = number of normal individuals in a population.

iii. Mutation Rate:

Mutation rate varies from gene to gene. Some genes exhibit high mutation rate than others. Such genes are known as mutable genes, e.g., white eye in Drosophila. In some genomes, some genes enhance the natural mutation rate of other genes. Such genes are termed as mutator genes.

The example of mutator gene is dotted gene in maize. In some cases, some genes decrease the frequency of spontaneous mutations of other genes in the same genome, which are referred to as anti-mutator genes. Such gene has been reported in bacteria and bacteriophages.

iv. Direction of Change:

Mutations usually occur from dominant to recessive allele or wild type to mutant allele. However, reverse mutations are also known, e.g., notch wing and bar eye in Drosophila.

v. Effects:
Mutations are generally harmful to the organism. In other words, most of the mutations have deleterious effects. Only about 0.1% of the induced mutations are useful in crop improvement. In majority of cases, mutant alleles have pleiotropic effects. Mutations give rise to multiple alleles of a gene.

vi. Site of Mutation:
Muton which is a sub-division of gene is the site of mutation. An average gene contains 500 to 1000 mutational sites. Within a gene some sites are highly mutable than others. These are generally referred to as hot spots. Mutations may occur in any tissue of an organism, i.e., somatic or gametic.

vii. Type of Event:
Mutations are random events. They may occur in any gene (nuclear or cytoplasmic), in any cell (somatic or reproductive) and at any stage of development of an individual.

viii. Recurrence:
The same type of mutation may occur repeatedly or again and again in different individuals of the same population. Thus, mutations are of recurrent nature.

5.3.2 Classification of Mutation

The mutation can be classified as follows:

1. Changes in genes

2. Changes in chromosomal number (polyploidy, haploidy, heteroploidy)

3. Changes in the arrangement of the chromosomal segments due to

(a) Intra-chromosomal segmental rearrangements (Inversions)

(b) Inter-chromosomal segmental rearrangements (Translocations)

(c) Losses and duplication of chromosomal segments (Deletions and Deficiency)

Mutations can be classified in various ways. A brief classification of mutations on the basis of:

(1) Source,

(2) Direction,

(3) Tissue,

(4) Effects,

(5) Site,

(6) Character, and

(7) Visibility

Mutations have been broadly categorized as somatic and germinal mutations. When a mutation occurs in a somatic cell, it does not change the whole organism, but produces a phenotypic change in the organ to which the mutant cell belongs. The resulting individual is a mosaic for mutant and normal tissues. golden delicious apples, emperor seedless grapes, some horticultural varieties of flowering plants, and white sectors in the red eyes of Drosophila males are examples of somatic mutations. Germinal mutations take place in cells of the germ line. A classic example of germinal mutations and perhaps also the one first recorded is that of short-legged sheep.

5.3.3 Kinds of Mutations:

According to the phenotypic expression the mutation may be classified in the following types:

(i) Somatic Mutation:

When changes in genes occur in the somatic or vegetative cells of the individuals, these are referred to as somatic mutation. Hugo de-Vries termed it as sports are saltation's. It has been found by Emerson in endosperm of maize and in many tissues of plants. Chimeras have been developed of such nature.

(ii) Spontaneous Mutation:

These spontaneous or gene mutation generally are developed by natural agencies like light, temperature etc. There are various characters which are

gene mutation. In mice spontaneous gene mutations determine coat colour which may be variously coloured like black, brown, spotted etc. In Drosophila there are many wild or normal type genes and their mutants like white eyes, pink eyes, yellow or black body colour and vestigial wing etc. Likewise, there are other gene affecting characters.

(iii) Germinal Mutation:

If the mutation occurs in the reproductive cells of gonads, then these are said as germinal mutation. Such type of mutation may be genetic occurring in the gametes of individuals or zygotic originating in the fused diploid gamete. Different sex linked mutations are of these types and pass from generation to generation.

(iv) Biochemical Mutation:

Such mutants influence the production of chemicals within an organism or causes the prevention of some enzymatic formation thus constituting biochemical mutation. Beadle and Tatum have studied in detail in Neurospora. Alcaptonuria and phenyl ketonuria described under gene nature are also biochemical mutations.

(v) Spurious Mutations:

These are hidden mutations appearing in the generation as a result of crossing over or other means. For example, in Drosophila, the gene for pink eyes remain usually hidden but it comes to light after crossing over. The appearance of recessive genes produced by crossing-over constitute spurious mutation.

(vi) Anomozygous Mutation:

These changes have developed due to structural (chromosomal aberrations) or numerical (polyploids) variations in the chromosomes.

(vii) Reverse Mutation:

It has been found in certain bacteria that are not capable to produce vitamins and other nutrients for their growth which normal type of bacteria can do efficiently. Sometimes such deficient mutants revert or change to normal condition is termed as reverse mutation. The chief cause of reverse mutation is radiation. Even then these reverse mutations as a rule occur rare.

(viii) Induced Mutation:

When gene changes are artificially produced or induced by means of experiments, such change constitutes induced mutation. The agent which

cause these induced mutations are called mutagenic agents which may be x- ray's radiation, various chemicals etc. Various chromosomal breaks make these changes.

Generally mutation have harmful effect on organisms. The individuals which carry them, reduce their viability.

Depending upon their effect on their viability of the individuals, it may be classified in to four groups:

(i) Lethal

(ii) Sub-lethal

(iii) Sub-vital

(iv) Vital

Muller (1927) firstly produced mutation successfully in Drosophila by x-ray treatment.

(i) Lethal:

Lethal mutations kill each and every organisms which carry them. Dominant lethals, therefore, cannot be studied because they can not survive even in the heterozygous state. Thus, we have to consider only recessive lethals. Recessive lethals would kill the individuals that carry them in homozygous state, e.g., albina chlorophyll mutation.

(ii) and (iii) Sub-lethal and Sub-vital.

It reduces the viability but do not kill the individuals carrying them. Sub-lethals will kill more than 50% individuals, where as sub-vitals less than 50%. A large majority of mutations are sub-lethals and sub-vitals, thus are of no value in crop improvement.

(iv) Vital:

Vital mutations do not reduce the viability of the individuals carrying them. Practically, crop improvement needs only such mutations. It occurs in very low frequency as compared to the other types.

Stages at which Mutations Occur

Mutations may occur at any stage in the development of the organism. If mutation comes in the primordial germ cells, all the gametes derived from these primordial germ cells will be carrying the mutant character.

If it happens in one of the gamete, this leads a mutant individual in the progeny. If mutation takes place in one of the daughter chromosomes of the dividing zygote, one part of the body of the individual will be carrying the mutation.

The later it appears, smaller will be the part of the body carrying the mutation. This type of individual is called Mosaic, for example, in Drosophila normal red eye with a speck of white or with one white and one red eye. Mutations also take place in the somatic tissue of any part of the body.

Mutation appears suddenly and never occurs gradually in a single individual and transmits to its progeny. Mutations have been observed in Oenothera, maize, man and other plant and animal species like Drosophila. In recent years, micro-organisms have been found to be the most favourable material to study this phenomenon.

Frequency of Mutations:

Different genes have different rates of mutability. Mutation rate may be defined as the number of changes at one special locus from one allele to another, measured in a biological unit of time, i.e., generation in a given population.

The rate of mutation varies from one organism to another and even from one variety to another in the same organism. The mutation rate varies considerably from one locus to another in the same variety.

5.3.4 Summary

Genetic variations are the differences in DNA sequences among individuals within a population, forming the raw material for evolution and contributing to biological diversity. These variations arise through multiple mechanisms, including **chromosome mutations** discussed in **Unit 14**, which involve large-scale structural changes such as deletions, duplications, inversions, and translocations of chromosome segments. Such alterations can disrupt gene function or regulation, potentially leading to developmental disorders or diseases. Additionally, changes in chromosome number, such as aneuploidy (e.g., trisomy 21 in Down syndrome), can result in severe phenotypic consequences. **Unit 15: Role of Gene Mutation** explores how mutations at the level of individual genes—such as point mutations, insertions, or deletions—can alter the nucleotide sequence and affect protein synthesis. Some gene mutations are silent or benign,

while others can impair or enhance protein function, leading to genetic disorders, increased disease susceptibility, or even beneficial adaptations. Together, chromosome and gene mutations play a crucial role in shaping genetic variation, driving evolution, and influencing health and disease in populations.

SELF ASSESSMENT

Q1. Genetic variation refers to:
a) Differences in the genetic makeup of individuals within a population
b) Similarity in the genetic makeup of individuals within a population
c) The process of DNA replication
d) The regulation of gene expression

Q2. Which of the following is a major source of genetic variation in a population?
a) Asexual reproduction
b) Mutation
c) Genetic drift
d) Gene flow

Q3. The exchange of genetic material between homologous chromosomes during meiosis is called:
a) Crossing over
b) Independent assortment
c) Genetic drift
d) Chromosomal inversion

Q4. Which of the following is true about genetic recombination?
a) It occurs only in prokaryotes
b) It results in genetically identical offspring
c) It contributes to genetic variation by creating new combinations of alleles
d) It only happens during mitosis

Q5. A change in the structure or number of chromosomes that leads to genetic disorders is known as:
a) Gene mutation
b) Chromosome mutation
c) Epigenetic modification
d) Allelic variation

Q6. Which of the following is an example of a chromosomal mutation?
a) Substitution of a nucleotide
b) Insertion of a base pair
c) Deletion of part of a chromosome
d) Change in protein structure

Q7. Down syndrome is caused by:
a) A gene mutation in chromosome 21
b) A trisomy of chromosome 21
c) A deletion on chromosome 13
d) A translocation of chromosome 5

Q8. A gene mutation that results in a change in a single nucleotide is known as:
a) Point mutation
b) Frameshift mutation
c) Inversion mutation
d) Translocation

Q9. Which of the following is a potential consequence of a gene mutation?
a) No change in the protein function
b) A change in protein function, leading to a genetic disorder
c) Replication of the mutated gene without any effects
d) Elimination of the gene from the genome

Q10. Gene mutations that occur in gametes can:
a) Affect only the individual in which they occur
b) Be inherited by the offspring
c) Be corrected by the immune system
d) Lead to mutations in somatic cells

Short Answer Questions

Q1. Explain the role of genetic recombination in increasing genetic variation in a population. Include the mechanisms that contribute to this variation during meiosis.

Q2. Describe chromosome mutations and provide examples of the different types of chromosomal mutations, such as deletion, duplication, inversion, and translocation. How can these mutations impact an organism?

Q3. Discuss the types of gene mutations, including point mutations and frameshift mutations, and explain how each type can affect the function of proteins. Provide examples of diseases caused by each type of mutation.

Q4. Explain how chromosomal abnormalities like trisomy and monosomy can result in genetic disorders, with specific reference to Down syndrome (trisomy 21) and its impact on individuals.

Q5. Discuss the role of mutations in the evolution of a species. How do gene mutations contribute to the genetic diversity within a population, and what is the role of natural selection in shaping the frequency of these mutations?

Long Answer Questions

Q1. Discuss the importance of genetic variation in evolution. Explain the sources of genetic variation in populations, with a focus on mutation, genetic recombination, and gene flow. How do these mechanisms contribute to the diversity of traits in a population?

Q2. Explain the concept of chromosome mutations and describe the different types, such as deletion, duplication, inversion, and translocation. How do chromosomal mutations affect the structure and function of genes, and what are the potential consequences for the organism? Provide examples of diseases or disorders caused by chromosomal mutations.

Q3. Gene mutations play a critical role in the evolution of organisms. Explain the various types of gene mutations (such as point mutations, insertion, deletion, and frameshift mutations), their effects on the genetic code, and how they influence protein synthesis. Provide examples of genetic disorders caused by specific gene mutations and discuss their impact on human health.

Q4. Chromosomal abnormalities can lead to significant genetic disorders. Describe trisomy and monosomy in detail, and explain how these chromosomal anomalies contribute to conditions such as Down syndrome, Turner syndrome, and Klinefelter syndrome. Discuss the genetic basis of these conditions and the impact on affected individuals.

Q5. Mutations are essential to the process of natural selection and the evolution of new traits. Explain the role of mutations in generating genetic diversity within a population. How do gene mutations influence evolutionary processes, and how does natural selection act on these mutations to shape the genetic makeup of future generations? Discuss the

potential advantages and disadvantages of mutations in different environments.

Answers

1 –a

2-b

3- a

4 –c

5 – b

6 – c

7 – b

8 – a

9 – b

10 - b

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