Structure and Types of DNA

DNA (Deoxyribonucleic Acid) is the molecule that carries genetic information of most of the living organisms and many viruses. It is composed of a double helix structure and exists in several forms depending on its sequence and environment.

Structure of DNA

1. **Primary Structure**:
    - DNA is a polymer made of repeating units called **nucleotides**.
    - Each nucleotide consists of three components:
        - **Phosphate Group**
        - **Deoxyribose Sugar** (5-carbon sugar)
        - **Nitrogenous Base**: Adenine (A), Thymine (T), Cytosine (C), or Guanine (G).

These nucleotides are linked together in a long chain through **phosphodiester bonds** between the phosphate group of one nucleotide and the sugar of the next nucleotide. This forms the **sugar-phosphate backbone** of the DNA strand, with the nitrogenous bases extending inward to form complementary base pairs (A with T, and C with G) between the two strands of DNA

2. **Double Helix**:
    - DNA exists as two complementary and antiparallel strands twisted into a double helix.
    - **Base Pairing Rules**:
        - Adenine (A) pairs with Thymine (T) via **2 hydrogen bonds**.
        - Cytosine (C) pairs with Guanine (G) via **3 hydrogen bonds**.
    - The strands are held together by hydrogen bonds and stacked base interactions.
3. **Backbone**:
    - The **sugar and phosphate groups** form the **backbone of DNA**.
    - The **bases are attached to the sugar and** face inward, **forming pairs**.
4. **Orientation**:
    - One strand runs in the **5' to 3' direction**, and the complementary strand runs in the **3' to 5' direction**.

Types of DNA Based on Structure

1. **B-DNA** (Biological Form):
    - **Right-handed helix** and the **most common form under physiological conditions.**
    - Has **10.5 base pairs per turn** and a wide major groove and narrow minor groove.
2. **A-DNA**:
    - **Right-handed helix**, **more compact than B-DNA**.
    - **Forms under dehydrated conditions**.
    - Has **11 base pairs per turn** with a **deeper major groove** and **shallower minor groove.**
3. **Z-DNA**:
    - **Left-handed helix** with a **zig-zag backbone**.
    - Forms in regions of **DNA with high GC content** or under **high salt conditions**.
    - May play a role in **gene regulation**.

Types of DNA Based on Function

1. **Genomic DNA**:
    - Found in the chromosomes of eukaryotes and prokaryotes.
    - Carries the organism's genetic information.
2. **Mitochondrial DNA (mtDNA)**:
    - Found in mitochondria, inherited maternally.
    - Circular in structure and codes for mitochondrial proteins.
3. **Plasmid DNA**:
    - Extrachromosomal, circular DNA found in bacteria and some eukaryotes.
    - Often carries genes for antibiotic resistance or other traits.
4. **Viral DNA**:
    - DNA found in some viruses, which can be linear or circular, single- or double-stranded.

Significance of DNA

- **Genetic Information**: Encodes instructions for protein synthesis and hereditary traits.

- **Replication**: Ensures the transfer of genetic information during cell division.
- **Genetic Variation**: Mutations and recombination in DNA are sources of diversity.

In summary, DNA's structure—a double helix—and its types enable it to efficiently store, replicate, and transmit genetic information essential for life.

Structure and Types of RNA

RNA (Ribonucleic Acid) is a single-stranded nucleic acid that plays vital roles in gene expression, protein synthesis, and regulation of cellular processes. Unlike DNA, RNA contains **ribose sugar** and the nitrogenous base **uracil (U)** instead of thymine (T).

Structure of RNA

1. **Single-Stranded**:
    - RNA is generally single-stranded but can fold into complex three-dimensional structures due to base pairing within the same strand.
2. **Components of RNA**:
    - **Ribose Sugar**: A 5-carbon sugar with one more hydroxyl group (-OH) than deoxyribose in DNA.
    - **Nitrogenous Bases**:
        - Purines: **Adenine (A)** and **Guanine (G)**
        - Pyrimidines: **Cytosine (C)** and **Uracil (U)** (Uracil replaces thymine in RNA).
    - **Phosphate Group**: Links nucleotides through phosphodiester bonds.
3. **Base Pairing**:
    - When RNA forms secondary structures, base pairing occurs (A-U and C-G).
4. **Folding**:
    - RNA can fold into various shapes (e.g., stem-loops or hairpins) due to intra-molecular hydrogen bonding, enabling it to perform diverse functions.

Types of RNA

There are several types of RNA, each with specific roles in cells. The major types are:

1. Messenger RNA (mRNA):

- **Function**: Carries genetic information from DNA to ribosomes for protein synthesis.

- **Structure**:
    - Linear and single-stranded.
    - Contains codons (three-nucleotide sequences) that specify amino acids during translation.

2. Transfer RNA (tRNA):

- **Function**: Brings amino acids to ribosomes during protein synthesis.
- **Structure**:
    - Folded into a cloverleaf or L-shape.
    - Contains an **anticodon** region that pairs with mRNA codons and an amino acid attachment site.

3. Ribosomal RNA (rRNA):

- **Function**: Forms the core structural and catalytic components of ribosomes, where proteins are synthesized.
- **Structure**:
    - Highly folded with complex secondary structures.
    - Combines with proteins to form ribosomes.

Differences Between DNA and RNA

Feature	DNA	RNA
Sugar	Deoxyribose	Ribose
Bases	A, T, G, C	A, U, G, C
Strands	Double-stranded (helix)	Single-stranded
Stability	Stable	Less stable
Function	Genetic information storage	Protein synthesis, regulation, etc.

Functions of RNA

1. **Protein Synthesis**:
    - mRNA, tRNA, and rRNA coordinate/interact to translate genetic information into proteins.

Summary

RNA is a versatile molecule with a single-stranded structure and multiple types, each specialized for specific cellular roles. Its ability to act as a messenger, adapter, structural component, and regulator makes it essential for genetic and biochemical processes in all living cells.

Gene

A **gene** is a **basic unit of heredity that carries information from one generation to the next**. It is made up of a specific sequence of **DNA (Deoxyribonucleic Acid)** that provides instructions to make proteins, which are essential for the structure, function, and regulation of the body's cells and tissues. In short, **genes are like the instruction manuals for building and running an organism**. They are **inherited from parents and passed down to offspring**, making them the foundation of life and evolution.

Key Features of Genes:

1. **Location**: Genes are **located on chromosomes**, which are found in the **nucleus of a cell.**
2. **Structure**: A gene has two main regions:
    - **Coding Region**: Contains the **instructions to make a protein**.
    - **Regulatory Region**: **Controls when and how much of the protein is made**.
3. **Function**: Genes determine traits like **eye color**, **height,** and even the risk of certain diseases. They also guide processes like growth, repair, and metabolism.
4. **Variation**: Changes or mutations in genes can lead to differences in traits or cause genetic disorders.

Types of Genes:

1. **Structural Genes**: Code for proteins that make up the body.
2. **Regulatory Genes**: Control the **activity of other genes**.
3. **Non-coding Genes**: Help in processes like regulating gene expression and making functional RNA.

Interesting Facts:

- Humans have about **20,000–25,000 genes**.
- Not all DNA is part of a gene; only about **1-2% of human DNA** codes for proteins.

Genetic Code

The **genetic code** is the set of rules by which the information in DNA or RNA is translated into proteins, which perform important functions in the cell. It is like a biological "dictionary" that tells the cell how to convert a sequence of nucleotides (the building blocks of DNA and RNA) into a sequence of amino acids (the building blocks of proteins).

Key Features of the Genetic Code:

1. **Triplet Code**:
    - The genetic code is read in groups of three nucleotides, called **codons**.
    - Each codon corresponds to a specific amino acid or a stop signal.
2. **Universal**:
    - The genetic code is nearly the same in all living organisms, from bacteria to humans, showing its importance in evolution.
3. **Non-ambiguous**
    - Each codon corresponds /codes to a specific amino acid.
4. **Redundancy or Degeneracy**:
    - Multiple codons can code for the same amino acid. For example, both **UUU** and **UUC** code for phenylalanine.
5. **Start and Stop Signals**:
    - A specific codon, **AUG**, acts as the start signal and also codes for the amino acid methionine.
    - Stop codons (**UAA**, **UAG**, **UGA**) signal the end of protein synthesis and are called as termination codons.
6. **Non-overlapping and Continuous**:
    - Codons are read one after another, without overlapping or skipping bases.
7. **Polarity**

    Genetic codons have a ***fixed polarity.*** Each triplet codon is read from 5' → 3' direction and the beginning base is 5' followed by the base in the middle then the last base which is 3'.

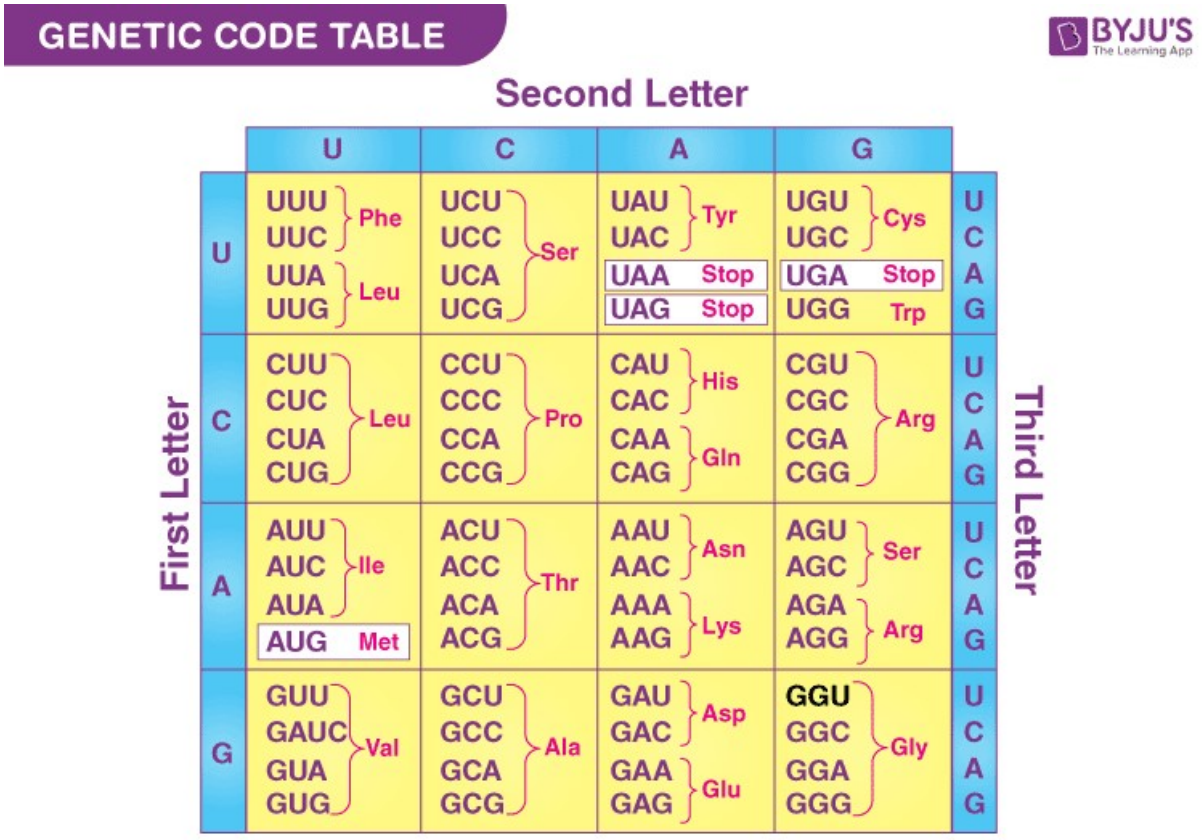
Example of Codons:

- **UUU**: Phenylalanine;
- **AUG**: Methionine (Start Codon);
- **UAA/UAG/UGA**: Stop Codon

Process of Translation:

1. **DNA to RNA**: DNA is transcribed into messenger RNA (mRNA) in the nucleus.
2. **Codons to Proteins**: In the ribosome, each codon in mRNA is matched with a complementary transfer RNA (tRNA) carrying an amino acid.
3. **Protein Formation**: The amino acids are linked together to form a protein chain.

In summary, the genetic code is the universal language of life, enabling cells to produce proteins based on genetic instructions. It ensures that genetic information is accurately translated into the molecules that sustain life.



The Central Dogma of Biology

The Central Dogma of Biology explains how genetic information flows within biological organisms to create proteins (building blocks of life). The central dogma states that information flow is unidirectional (in one direction) that is from DNA to RNA to protein. It was first proposed by Francis Crick in 1958 and is often summarized as:

DNA → RNA → Protein

Steps of the Central Dogma

1. **Replication (DNA → DNA):**

- DNA makes a copy of itself.

- This happens during cell division so that each new cell gets the same genetic information.

2. **Transcription (DNA → RNA):**

- DNA is used as a template to make RNA.

- Specifically, a type of RNA called mRNA (messenger RNA) is produced.

- This process happens in the nucleus of eukaryotic cells.

3. **Translation (RNA → Protein):**

- mRNA is used as a template to make proteins.

- Proteins are made up of amino acids, which are assembled based on the sequence of nucleotides in the mRNA.

- This occurs in the ribosome, which reads the mRNA sequence three nucleotides (a codon) at a time.

- Exceptions to central dogma of biology:

There are some exceptions to central dogma of biology:

1. **Reverse Transcription**: In retroviruses like HIV, RNA can be converted back into DNA using an enzyme called reverse transcriptase.

2. **Non-coding RNA**: Some RNA molecules (e.g., **rRNA, tRNA, and microRNA**) are not translated into proteins but play essential roles.

Why Is the Central Dogma Important?

TERMS FOR YOUR UNDERSTANDING

a) The Flow of Information

1. **DNA**: Stores the genetic blueprint.

2. **RNA**: Acts as a messenger and intermediary.

3. **Protein**: Performs most of the functions in the cell (enzymes, structural support, etc.).

b) **Template**: a thing that is used as a model for producing other similar examples

CHROMOSOMES

What is a Chromosome? – Definition

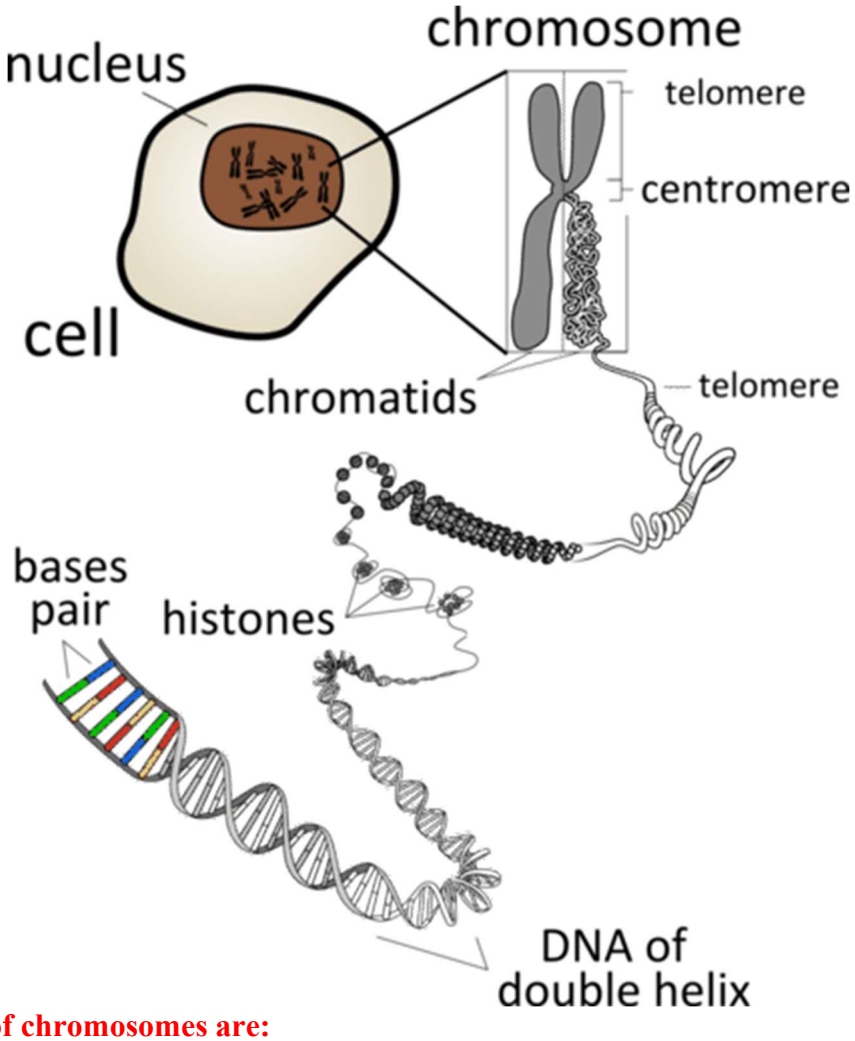
Chromosomes are thread-like structures present in the nucleus, which carries genetic information from one generation to another. In Eukaryotic cells, genetic material is present in the nucleus in chromosomes, which is made up of **highly organized DNA molecules** with **histone proteins** supporting its structure. They play important role in cell division, heredity, variation, and mutation.

- Chromosome means '**coloured body**', that which is in reference to its staining ability by certain dyes.
- **W. Waldeyer** in 1888 coined the term 'chromosome'.
- The number of chromosomes in **any species is constant** for all the cells.
- The number of **chromosomes in gametes (e.g. sperms, egg)** is **half of the somatic cell** and **known as a haploid set of chromosomes**, which is the result of meiosis during sexual reproduction.
- Chromosome number is **preserved in the mitotic division of somatic cells**, which is required for an organism to grow, repair and regenerate.
- Chromosome number **varies in different species**.
- Each cell has a pair of each kind of chromosome known as a **homologous chromosome**
- A human cell contains total 23 pair of chromosomes (2n, total 23×2=46), of which 22 are autosomes and 1 sex chromosome.

Chromosome Structure

- Chromosomes are **made up of chromatin**, which contains a **single molecule of DNA** and histone proteins.

- Each chromosome **contains hundreds and thousands of genes** that have **genetic code for several proteins in the cell**.



Main parts of chromosomes are:

- **Chromatid:** Each chromosome has **two symmetrical structures called chromatids** or **sister chromatids** which is **best visible in mitotic metaphase**.
  - Each **chromatid contains a single DNA molecule and positively charged histone proteins**
  - At the **anaphase of mitotic cell division**, **sister chromatids separate and migrate to opposite poles**
- **Centromere and kinetochore: Sister chromatids** are **joined by the centromere**.
  - **Spindle fibres during cell division** are attached at the **centromere**
  - The **number and position of the centromere differs in different chromosomes**
  - The **centromere** is called **primary constriction**
  - **Centromere divides the chromosome into two parts**, the **shorter arm is known** as **'p' arm** and the longer arm is known as **'q' arm.**
  - The **kinetochore** is a **protein complex on the centromere** that **helps in chromosome attachment to spindle fibers during cell division**.

- **Secondary constriction and nucleolar organisers:**
    - Other than centromere, chromosomes possess secondary constrictions.
    - Secondary constrictions, are known as the **nucleolar organiser**

- **Telomere:** Terminal part of a chromosome is known as a telomere.
    - **Telomeres are polar, which prevents the fusion of chromosomal segments**
- **Satellite:** It is an **segment of chromosome that is separated by the rest of the chromosome by a secondary constriction.**
    - The **chromosomes with satellite** are known as **sat-chromosome**

CHROMOSOMES DURING DIFFERENT STAGES OF MITOSIS/MEIOSIS

✓ At interphase, chromosomes **are visible as thin chromatin fibres present in the nucleoplasm**.

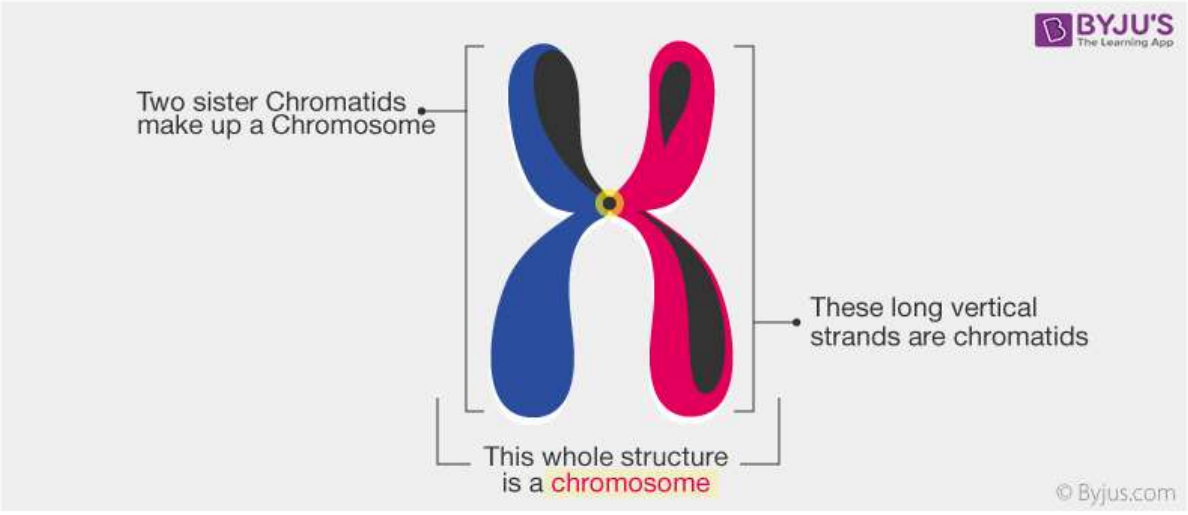
➢ During cell division, the **chromatin fibres condense** and **chromosomes are visible with distinct features**. Structure of a chromosome can be best seen during cell division.

✓ At prophase, the **chromosomal material is visible as thin filaments** known as **chromonemata**

✓ At interphase, bead-like structures are visible, which are **an accumulation of chromatin material called chromomere.** Chromatin with chromomere looks like a necklace with beads

- The **darkly stained, condensed region of chromatin** is known as **heterochromatin.** It contains tightly packed DNA, **which is genetically inactive**
- The **light stained, diffused region of chromatin** is known as **euchromatin.** It contains **genetically active and loosely packed DNA**

Labelled diagram of chromosome is given below.

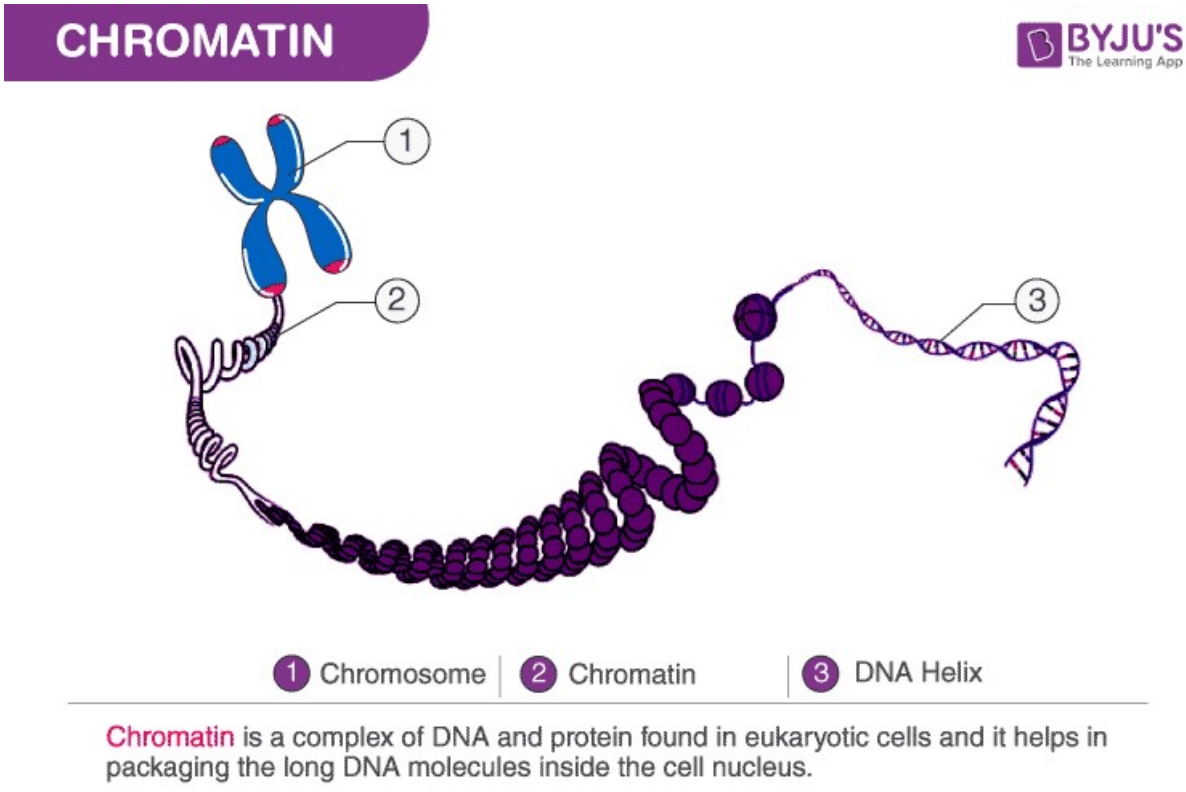


Chromosome Labelled Diagram

Structural Organisation of Chromatin

- Chromatin consists of **DNA** and **associated proteins**. DNA is packaged in a highly organised manner in chromosomes

1. **Nucleosomes** are the basic unit of chromatin.
    - DNA packing is facilitated by proteins called **histones.** DNA is wound around histone proteins to form a nucleosome
    - There are 5 types of histone proteins in the eukaryotic chromosomes, namely H1, H2A, H2B, H3 and H4
    - **Histones are positively charged due to the presence of amino acids** with **basic side chains** and it associates with **negatively charged DNA** due to the **presence of phosphate groups**
    - Histone proteins play an important role in **gene regulation**
    - A typical nucleosome **contains 200 bp of DNA helix**. The core particle of the nucleosome consists of approximately **146 base pairs (bp) of DNA coiled around a core of eight histone molecules (2 molecules of 4 histone proteins).** That is **linked by linker DNA of about 80 bp**.
    - **Nucleosomes prevent DNA from getting tangled**



Chromatin Structure

Read more: Organisation of Eukaryotic Genome

Types of Chromosome

Based on the number of centromeres present

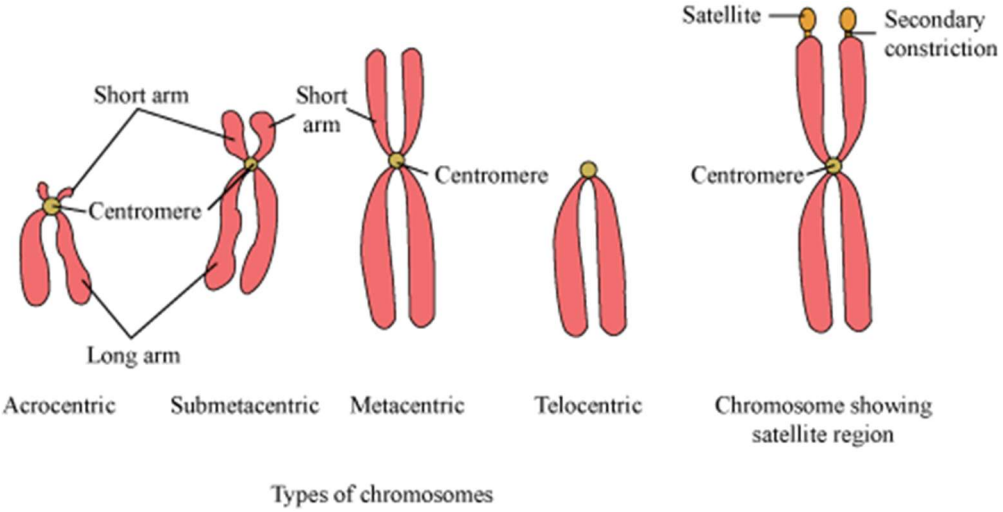
i. **Monocentric:** having only **one centromere**

ii. **Dicentric:** It has two centromeres.

iii. **Polycentric:** It has multiple centromeres

iv. Holocentric: having **diffused centromere spread along the entire length of a chromosome**

v. **Acentric:** chromosome **without a centromere**. It cannot attach to the mitotic spindle



Types of chromosomes

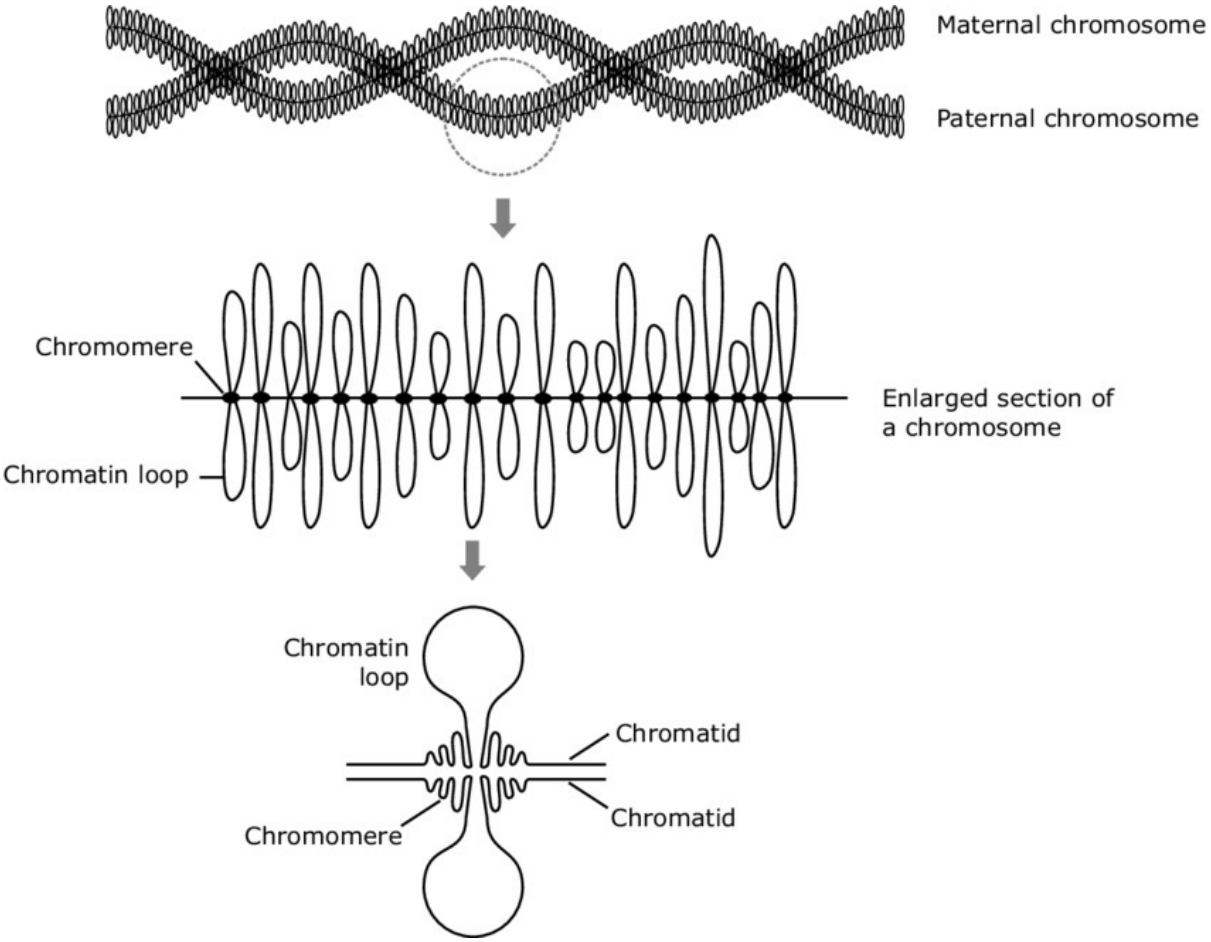
i. **Metacentric**: Centromere in the middle; **arms are equal in length**.
ii. **Submetacentric**: Centromere **slightly off-center**; one arm is longer.
iii. **Acrocentric**: **Centromere near one end**; one arm is very short.
iv. **Telocentric**: **Centromere at the end**; only one arm is visible.

Giant Chromosomes

I. **Polytene chromosomes** are giant chromosomes that arise from repeated rounds of DNA replication without cell division and were discovered by Balbiani. They are characterized by their large size and distinct banding pattern, which makes them ideal for studying gene activity and chromosomal organization. These chromosomes are commonly found in the salivary glands of certain insect larvae, such as *Drosophila*. The **dark and light bands** represent areas of **condensed** and **decondensed** chromatin, respectively, allowing scientists to map gene expression and study chromosomal rearrangements.

II. **Lampbrush chromosome** on the other hand, are **large meiotic chromosomes** found in the growing oocytes (female egg cell) of vertebrates(organisms with vertebrae/spinal cord), such as **amphibians(organisms which live on both land and water) and birds**. They are named for their **"brush-like" appearance**, with loops of **DNA extending outward from the main chromosomal axis**. These loops are regions of a**ctive RNA transcription**, supporting the intense metabolic activity required during oocyte development. Lampbrush chromosomes are particularly useful for studying tl activity and chromosomal structure during meiosis.

Both polytene and lampbrush chromosomes are specialized types of chromosomes that provide valuable information about **chromatin organization and gene regulation**.



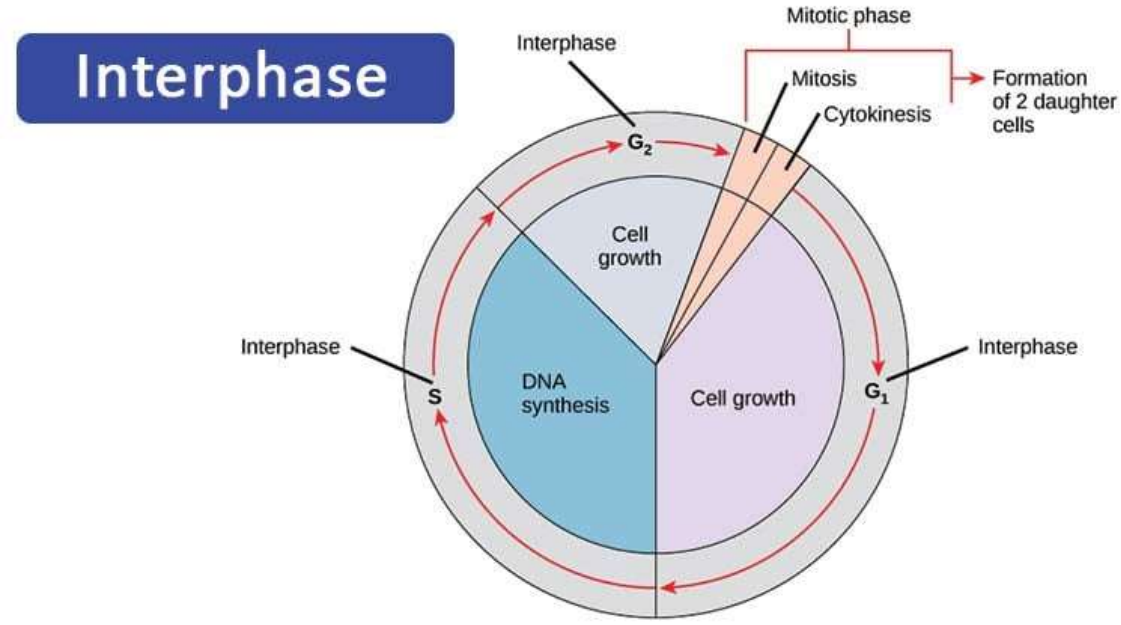
Functions of Chromosomes

- The main function of chromosomes is to **carry the genetic material from one generation to another**
- Chromosomes play an important role in the **growth, reproduction, repair** and regeneration process, which is important for their survival
- Chromosomes **protect the DNA from getting tangled and damaged**
- Histone and non-histone proteins help in the **regulation of gene expression**
- **Spindle fibres attached to the centromere** help in the movement of the chromosome during cell division
- **Each chromosome contains thousands of genes that precisely code for multiple proteins present in the body**

CELL CYCLE

Interphase

Interphase is the **phase of the cell cycle in which the cell prepares for division,** performing its normal functions and undergoing growth. It is the **longest phase of the cell cycle** and occurs before mitosis or meiosis. Interphase is divided into three distinct stages: G1, S, and G2.



Stages of Interphase

1. G1 Phase (Gap 1):
    - Growth and Preparation: The cell grows in size, and carries out its normal metabolic functions.
    - The cell also **prepares the necessary components** for **DNA replication**.

    - Checkpoint: At the end of G1, the cell checks whether it is ready to proceed to the next phase. If conditions aren't favorable, it may enter a resting state called the **G0 phase**, where it no longer divides (e.g., nerve or muscle cells).

2. S Phase (Synthesis):
    - DNA Replication: **The cell's DNA is replicated**, so that **each chromosome consists of two sister chromatids by the end of the phase.**
    - **This ensures that the daughter cells will receive an identical copy of the genome during cell division**.
    - Checkpoint: The cell **checks for any DNA damage and ensures proper replication.**
3. G2 Phase (Gap 2):
    - Final Preparations: **The cell continues to grow and produce proteins necessary for cell division.**
    - It also checks the replicated DNA for any errors and repairs them.
    - Checkpoint: The cell ensures that all **DNA has been properly replicated and that the cell is large enough to divide.**

Key Features of Interphase:

- Cell Growth: The cell increases in size and organelles are duplicated to ensure two functional daughter cells.
- DNA Replication: The genetic material is copied, so that each daughter cell will have the same genetic information.
- Metabolic Activity: The cell continues performing its specialized functions (e.g., protein synthesis, energy production) during interphase.
- No Cell Division: Although the cell is very active, it does not divide during interphase.

Importance of Interphase:

- **Preparation for Division**: Interphase ensures that the cell is properly prepared for mitosis or meiosis by growing, replicating DNA, and checking for errors.
- **Genetic Stability:** By accurately replicating DNA, interphase contributes to the maintenance of genetic stability in the organism.

In summary, interphase is a **crucial part of the cell cycle where the cell grows, replicates its DNA, and prepares for cell division. It ensures that the cell is ready to divide into two daughter cells with identical genetic material.**

Mitosis

Mitosis is a type of cell division that produces two genetically identical daughter cells from a single parent cell. It occurs in somatic (non-reproductive) cells and is essential for growth, repair, and asexual reproduction in multicellular organisms.

Phases of Mitosis

Mitosis is divided into five stages:

1. **Prophase**:
    - Chromosomes condense and become visible as paired chromatids.
    - The nuclear membrane begins to break down.
    - The spindle fibers, made of microtubules, start forming.
2. **Prometaphase**:
    - The nuclear envelope disappears completely.
    - Spindle fibers attach to the **kinetochores** (protein structures on centromeres of chromosomes).
3. **Metaphase**:
    - Chromosomes align at the cell's equator, forming the **metaphase plate**.
    - Spindle fibers ensure each chromosome is properly attached to opposite poles.
4. **Anaphase**:
    - Sister chromatids separate and move to opposite poles as spindle fibers shorten.
    - Each chromatid becomes an independent chromosome.
5. **Telophase**:
    - Chromosomes decondense and return to their thread-like form.
    - The nuclear envelope re-forms around each set of chromosomes.
    - The spindle fibers disassemble.

Cytokinesis:

- Following mitosis, the cytoplasm divides, forming two daughter cells.
- In **animal cells**, this occurs through the formation of a **cleavage furrow**.
- In **plant cells**, a **cell plate** forms, which later becomes the cell wall.

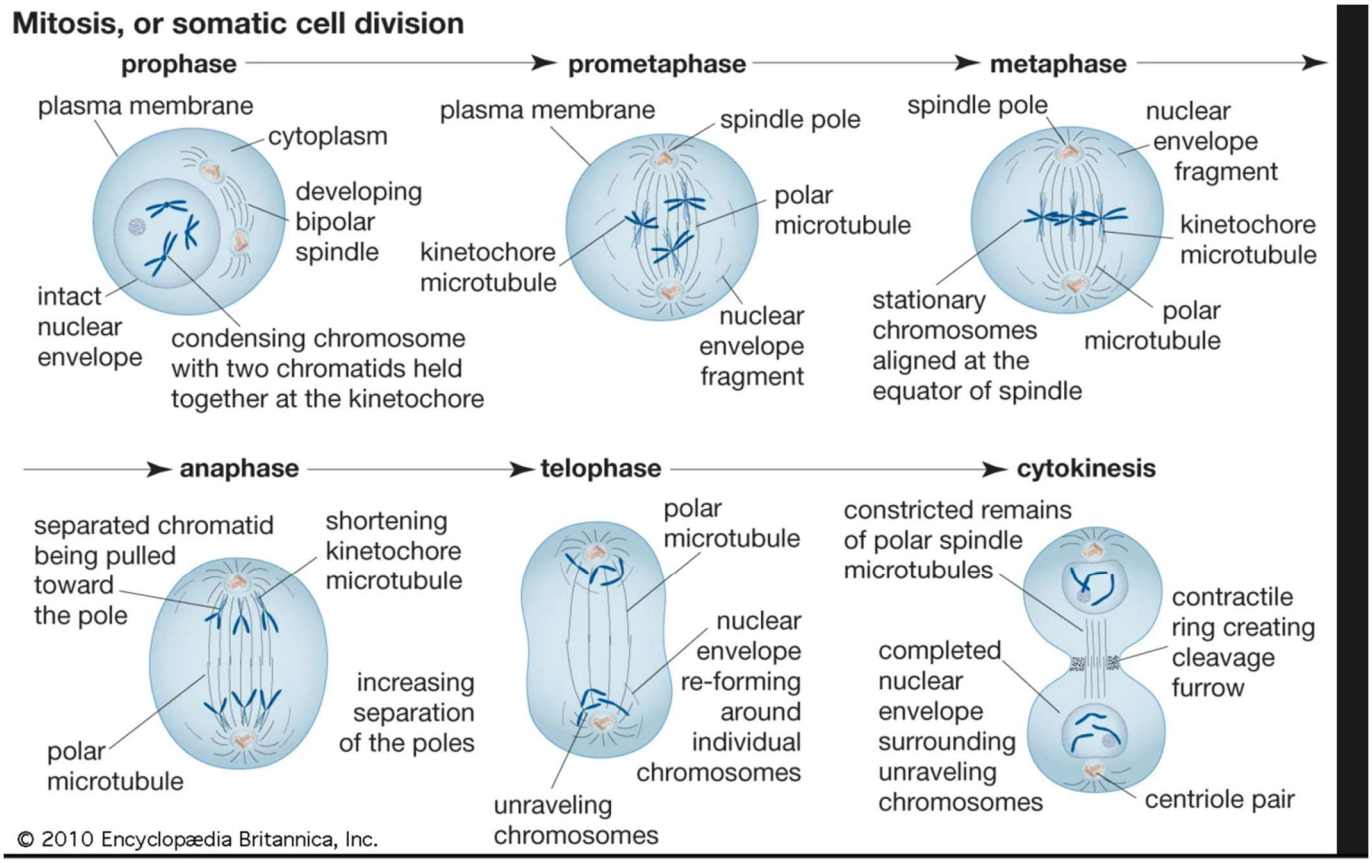
Importance of Mitosis:

1. **Growth**: Helps organisms increase in size by producing more cells.
2. **Repair**: Replaces damaged or dead cells.
3. **Asexual Reproduction**: In single-celled organisms like amoebas, mitosis is used to reproduce.
4. **Genetic Stability**: Ensures the daughter cells have the same genetic material as the parent cell.

Key Points:

- Mitosis maintains the chromosome number of the parent cell (e.g., a diploid cell produces diploid daughter cells).
- Errors in mitosis can lead to genetic disorders or cancer.

In simple terms, mitosis is the process by which cells make exact copies of themselves to support life and maintain the body's functions.



Meiosis

Meiosis is a type of cell division that reduces the chromosome number by half, producing four haploid cells from a single diploid cell. This process occurs in the reproductive cells (gametes) and is essential for sexual reproduction, ensuring genetic diversity.

Phases of Meiosis

Meiosis consists of two stages: **Meiosis I** (reductional division) and **Meiosis II** (equational division). Each stage has its own phases.

---

1. Meiosis I: Reduces the chromosome number from diploid (2n) to haploid (n).

a) Prophase I of Meiosis

Prophase I is the first and most complex stage of meiosis. It is crucial for genetic recombination and proper segregation of homologous chromosomes. This stage is divided into five sub-stages, each characterized by specific chromosomal events.

Sub-stages of Prophase I

1. **Leptotene**:
    - Chromosomes start to condense and become visible as thin threads.
    - Each chromosome consists of two sister chromatids, but they are not yet distinguishable.
    - The chromosomes attach to the nuclear envelope via protein complexes.
2. **Zygotene**:
    - Homologous chromosomes (one from each parent) pair up in a process called **synapsis**.
    - A structure called the **synaptonemal complex** forms between homologs, holding them together.
3. **Pachytene**:
    - Chromosomes condense further, and the homologs are fully synapsed.
    - **Crossing over** occurs, where non-sister chromatids of homologous chromosomes exchange genetic material at points called **chiasmata**.
    - This recombination is key for genetic diversity.
4. **Diplotene**:

- The synaptonemal complex dissolves, and homologous chromosomes begin to separate.
- However, they remain connected at the chiasmata, where crossing over occurred.

5. **Diakinesis**:
    - Chromosomes are highly condensed and visible under a microscope.
    - The nuclear envelope breaks down, and spindle fibers start to form.
    - Homologous chromosomes are still attached at chiasmata, but they are ready to move apart.

Key Features of Prophase I:

1. **Synapsis**: Pairing of homologous chromosomes.
2. **Crossing Over**: Exchange of genetic material increases genetic variation.
3. **Formation of Spindle Apparatus**: Prepares for the alignment and segregation of chromosomes.

Significance of Prophase I:

- Ensures genetic recombination through crossing over, which contributes to genetic diversity in offspring.
- Proper pairing and alignment of homologs ensure accurate segregation during Anaphase I.

Prophase I is a prolonged and intricate stage, setting the foundation for the unique outcomes of meiosis: the production of genetically diverse haploid cells.

b) Metaphase I:

- Homologous chromosome pairs (tetrads) align at the equatorial plate.
- Spindle fibers attach to the centromeres of each homolog.

c) Anaphase I:

- Homologous chromosomes are pulled apart to opposite poles, but sister chromatids remain together.

d) Telophase I and Cytokinesis:

- Chromosomes reach the poles, and the cell divides into two haploid cells.
- Each cell now has half the original chromosome number, but each chromosome still consists of two sister chromatids.

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2. Meiosis II: Similar to mitosis, it separates sister chromatids.

a) **Prophase II**:
   - Chromosomes condense, and a new spindle apparatus forms.
   - The nuclear envelope dissolves again (if it reformed in Telophase I).

b) **Metaphase II**:
   - Chromosomes align individually along the equatorial plate.
   - Spindle fibers attach to the centromeres.

c) **Anaphase II**:
   - Sister chromatids are pulled apart to opposite poles.

d) **Telophase II and Cytokinesis**:
   - Chromatids reach the poles, and nuclear envelopes reform.
   - Cytoplasm divides, resulting in four haploid daughter cells.

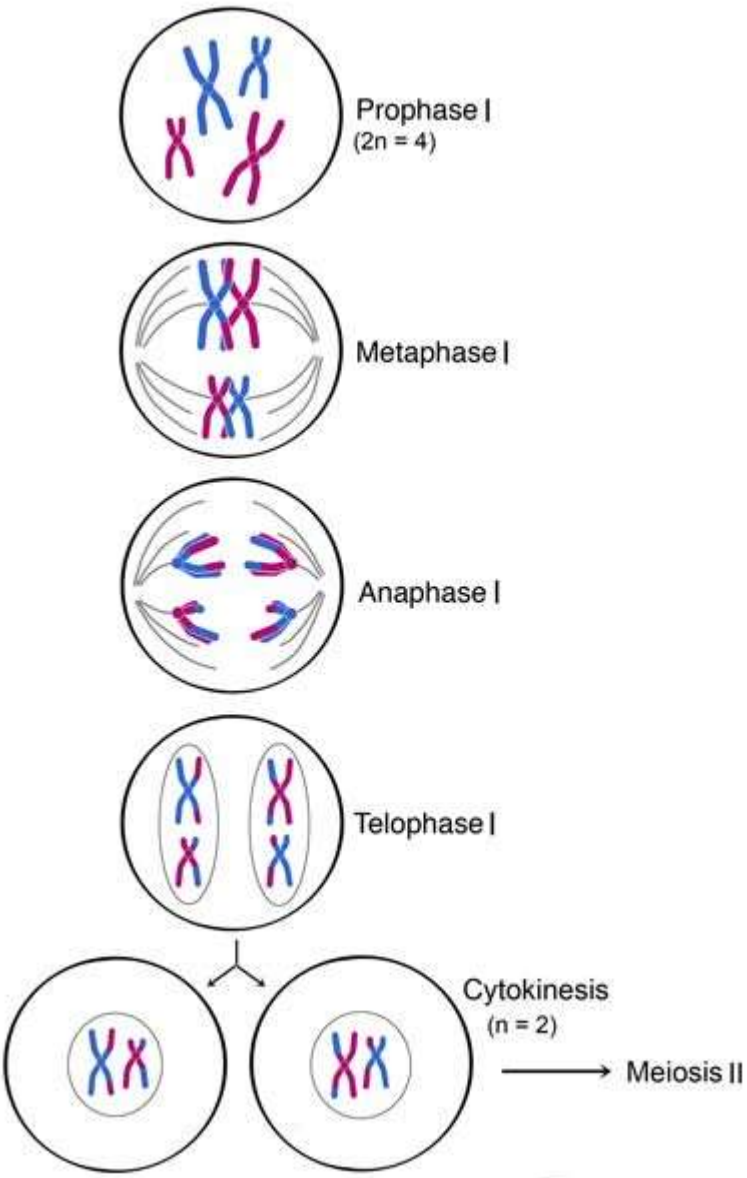
Key Features of Meiosis:

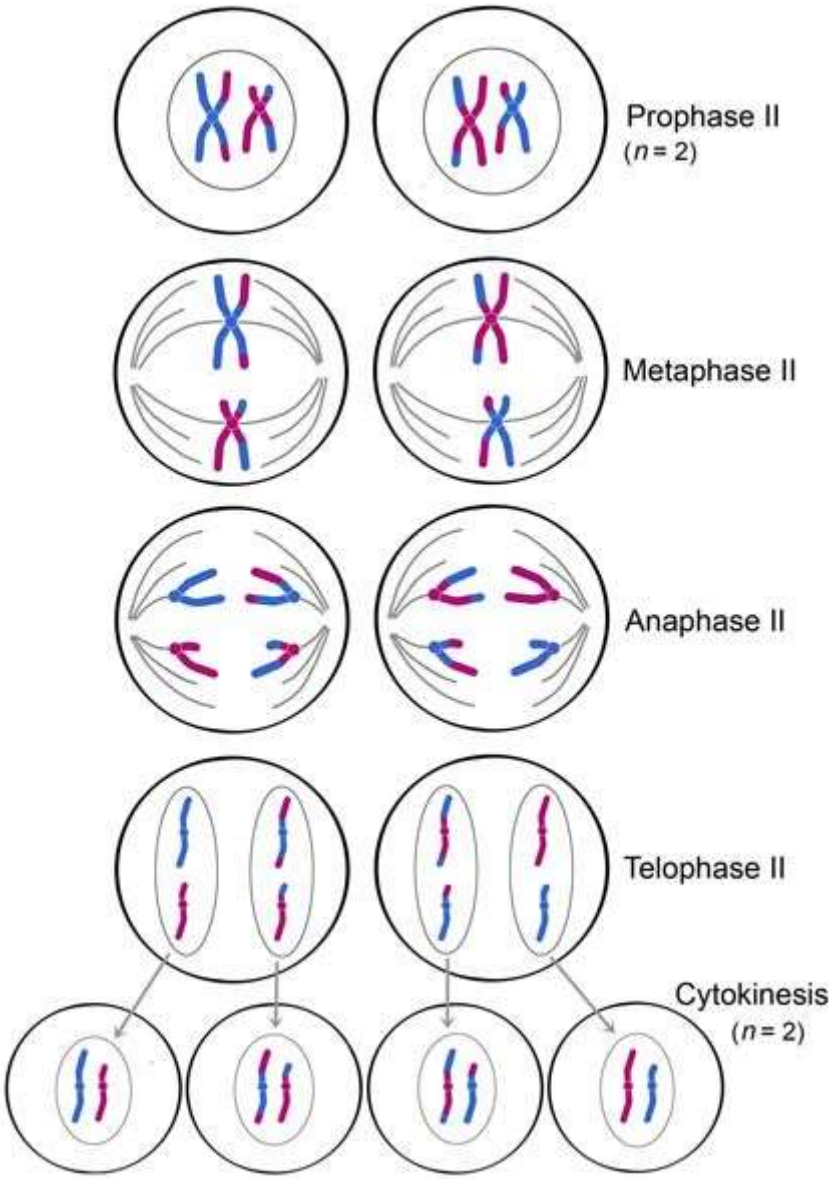
1. **Reduces Chromosome Number**: Essential for maintaining chromosome count in sexually reproducing organisms.
2. **Genetic Variation**:
   - **Crossing Over**: Exchange of genetic material during Prophase I.
   - **Independent Assortment**: Random alignment of homologous chromosomes during Metaphase I.

Significance of Meiosis:

1. **Formation of Gametes**: Produces eggs and sperm in animals, and spores in plants.
2. **Genetic Diversity**: Essential for evolution and adaptation through recombination and independent assortment.
3. **Maintaining Chromosome Number**: Prevents doubling of chromosomes in each generation.

In summary, meiosis is the process by which sexually reproducing organisms create haploid cells, enabling genetic diversity and continuation of species.


Prophase I
(2n = 4)
Metaphase I
Anaphase I
Telophase I
Cytokinesis
(n = 2)
Meiosis II



Mutation - A Genetic Change

Mutation Definition

"Mutation is the change in our DNA base pair sequence due to various environmental factors such as UV light, or mistakes during DNA replication."

What Are Mutations?

The DNA sequence is specific to each organism. It can sometimes undergo changes in its base-pairs sequence. It is termed as a mutation. A mutation may lead to changes in proteins translated by the DNA. Usually, the cells can recognize any damage caused by mutation and repair it before it becomes permanent.

A mutation is a sudden, heritable modification in an organism's traits. The term "mutant" refers to a person who exhibits these heritable alterations.

Classification & Types of Mutations

Mutation Classifications	Types	Description	Examples of Human Disease(s)
Point mutation	Substitution	During replication, **one base is inserted** incorrectly, replacing the pair at the appropriate location on the complementary strand.	Sickle-cell anemia
	Insertion	In replicating DNA, **one or more additional nucleotides are added**, frequently causing a frameshift.	One form of beta-thalassemia
	Deletion	During replication, **one or more nucleotides may be "skipped" or removed**, which usually causes a frameshift.	Cystic fibrosis
Chromosomal mutation	Inversion	The **flipping and reinserting of a single chromosomal region**.	Opitz-Kaveggia syndrome
	Deletion	When a **chromosome segment is lost**, all the genes in that segment are also gone.	Cri du chat syndrome
	Duplication	**A chromosomal segment is repeated**, increasing the concentration of the genes in that area.	Some cancers

	Translocation	**A section of one chromosome is inappropriately joined to another chromosome**.	One form of leukemia
Copy number variation	Gene amplification	An increase is made in the **tandem copies of a locus.**	Some breast cancers
	Expanding trinucleotide repeat	There **are more repeating trinucleotide sequences than usual.**	Fragile X syndrome, Huntington's disease

Causes of Mutations

The mutation leads to genetic variations among species. **Positive mutations are transferred to successive generations.**

E.g. Mutation in the gene coding for haemoglobin causes sickle cell anaemia. The R.B.Cs become sickle in shape. However, in the African population, this mutation provides protection against malaria.

A mutation in the gene controlling the cell division leads to cancer.

Let us have an overview of the causes and impacts of mutation.

The mutation is caused due to the following reasons:

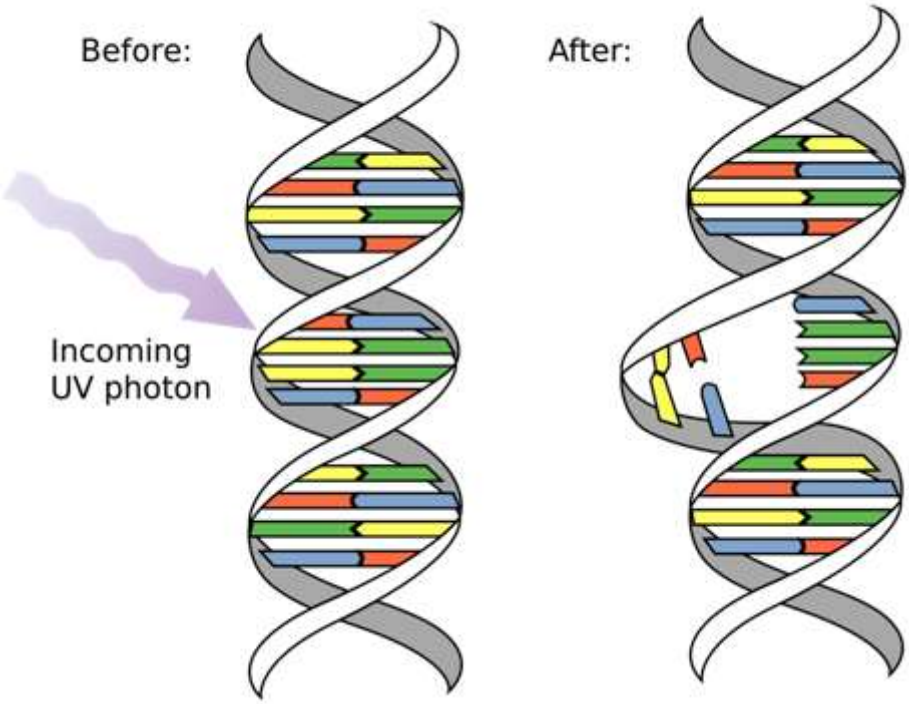
Internal Causes

Most of the mutations occur when the **DNA fails to copy accurately**. All these mutations lead to evolution. During cell division, the DNA makes a copy of its own. Sometimes, the copy of the DNA is not perfect and this slight difference from the original DNA is called a mutation.

External Causes

When the DNA is exposed to certain **chemicals (Ethyl methyl sulfate, nitrogen mustards, mitomycin)** or **radiations (X-rays, UV rays, gamma radiations), it causes the DNA to**

break down. The ultraviolet radiations cause the thymine dimers to break resulting in a mutated DNA.



DNA Mutation

Somatic mutations and Germline mutations

There are several mutations that cannot be passed on to the offsprings. Such mutations occur in the somatic cells and are known as **somatic mutations. The germline mutations** can be passed on to successive generations and occur in the reproductive cells.

Spontaneous and Induced Mutations

Mutations are also classified as **spontaneous** or **induced**.

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1. Spontaneous Mutations:

These mutations happen naturally without any external influence. They occur during normal cellular processes, such as DNA replication, repair, or recombination.

Examples:

- Point mutations caused by DNA polymerase errors.
- Small insertions or deletions due to slippage during replication.

2. Induced Mutations:

These mutations result from exposure to external agents called **mutagens**, which increase the mutation rate.

Causes:

a) **Physical Mutagens**: **Radiation** (Ultraviolet (UV) radiation, X rays and gamma rays)
b) **Chemical Mutagens**:
    - **Alkylating Agents**. Add alkyl groups to DNA bases, altering base pairing.
    - **Intercalating Agents**: Insert between DNA bases, causing insertions or deletions.
c) **Biological Agents**:
    a. Viruses or transposable elements that integrate into the genome.

Examples:

- DNA damage caused by UV light.
- Mutations induced by chemicals like ethidium bromide or mustard gas.

Key Differences:

Aspect	Spontaneous Mutation	Induced Mutation
Cause	Natural cellular processes	External agents (mutagens)
Occurrence	Random and rare	Depends on exposure to mutagens
Examples	DNA replication errors, deamination	UV-induced thymine dimers, chemical damage

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In summary, **spontaneous mutations** are natural errors in DNA, while **induced mutations** arise from external environmental factors. Both types play crucial roles in evolution, genetic variation, and disease.

Effects of Mutation

Beneficial Effects of Mutation

1. **Few mutations result in new versions of proteins** and help the organisms to adapt to changes in the environment. Such mutations lead to evolution.
2. **Mutations in many bacteria result in antibiotic-resistant strains of bacteria that can survive in the presence of antibiotics.**

3. A **unique mutation found in the population of Italy protects them from atherosclerosis, where fatty materials build up in the blood vessels.**

Negative effects of Mutations

1. Genetic disorders can be caused by the mutation of one or more genes. Example cystic fibrosis is one such genetic disorder caused by the mutation in one or more genes.
2. Cancer is another disease caused by the mutation in genes that control/regulate the cell cycle.

Transposons

Transposons, also known as "jumping genes," are segments of DNA that can move from one location to another within a genome. Transposons were discovered by **Barbara McClintock**, an American scientist, in the 1940s and 1950s while studying the genetics of maize (corn). Her work showed that genes are not always fixed in place—they can move around. They are found in almost all organisms and are an important part of genetic evolution and diversity. Here's an overview:

Types of Transposons

1. **Class I Transposons (Retrotransposons):**

- Move through a "copy and paste" mechanism.

- These transposons are transcribed into RNA and then reverse-transcribed back into DNA by reverse transcriptase before inserting into a new genomic location.

- **Examples:**

  - retrotransposons like LINEs (Long Interspersed Nuclear Elements) and SINEs (Short Interspersed Nuclear Elements).

2. **Class II Transposons (DNA Transposons):**

- Move through a "cut and paste" mechanism.

- These transposons **do not involve an RNA intermediate**.

- Often encode the enzyme transposase, which facilitates their excision and reinsertion.

- Example: The ***Ac/Ds* system in maize** discovered by Barbara McClintock.

Importance of Transposons

1. **Genetic Diversity**: They contribute to mutations and recombination (a combining of genes or characters different from what they were in the parents) events, which results in evolution.

2. **Gene Regulation:** Transposons can affect gene expression by disrupting or modifying regulatory elements.

3. **Applications in Science**:

- Tools in genetic engineering.

- Model systems for studying genome dynamics.

Risks of Transposons

1. **Genomic Instability**: Their movement can cause harmful mutations or chromosomal rearrangements.

2. **Disease Association**: Linked to various diseases, including cancer, due to their disruptive potential.

MEANING OF TERMS USED FOR YOUR UNDERSTANDING

1. **Evolution:** the development of plants, animals, etc. over many thousands of years from simple early forms to more advanced ones
2. **Genetic Diversity: (विविधता)**
3. **Mutation**: A mutation is a change in a DNA sequence.
4. **Recombination**: the rearrangement of genetic material, especially by crossing over in chromosomes during meiosis or by the artificial joining of segments of DNA from different organisms.
5. **Genetic engineering:** the modification and manipulation of an organism's genes using technology.
6. **Genome dynamics** refers to the changes and movements in an organism's DNA over time, shaping its structure, function, and evolution
7. **Gene expression** is the process by which the information encoded in a gene is turned into a protein/function.

Experimental Evidence for DNA as Genetic Material

The discovery that **DNA** is the genetic material that carries and transmits genetic information was a result of several key experiments over time. Below are some of the major experimental studies that provided evidence for DNA being the genetic material:

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1. Griffith's Experiment (1928) – Transformation in Bacteria

Objective: To understand how bacterial strains cause disease.

Experiment:

- Griffith used **Streptococcus pneumoniae** bacteria, which existed in two forms:
    - **Smooth (S)**: Virulent (disease-causing) due to a protective capsule.

    - **Rough (R)**: Non-virulent (harmless).
- He injected mice with:

1. Live S strain (mice died).
2. Live R strain (mice lived).
3. Heat-killed S strain (mice lived).
4. Mixture of heat-killed S strain and live R strain (mice died).

Result:

- The mice injected with a mixture of heat-killed S strain and live R strain died, and **live S bacteria were found in their blood**, despite the heat-killed S strain being non-viable.
- This suggested that something from the dead S strain was transforming the R strain into a virulent form.

Conclusion:

- Griffith concluded that the transforming factor was likely a **genetic material** that could transfer genetic information between bacterial cells.

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2. Avery, MacLeod, and McCarty's Experiment (1944) – Identifying the Transforming Principle

Objective: To identify the chemical nature of the "transforming principle" from Griffith's experiment.

Experiment:

- They treated the extract from heat-killed S strain bacteria with different enzymes that destroyed proteins, RNA, and DNA.
- After each treatment, they tested whether the R strain was still transformed into the S strain.

Result:

- Only when **DNA** was destroyed, the transformation did not occur, meaning the transforming factor was **DNA**.

Conclusion:

- They provided evidence that **DNA** was the substance responsible for the transfer of genetic information, proving DNA as the genetic material.

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3. Hershey-Chase Experiment (1952) – Bacteriophage Infection

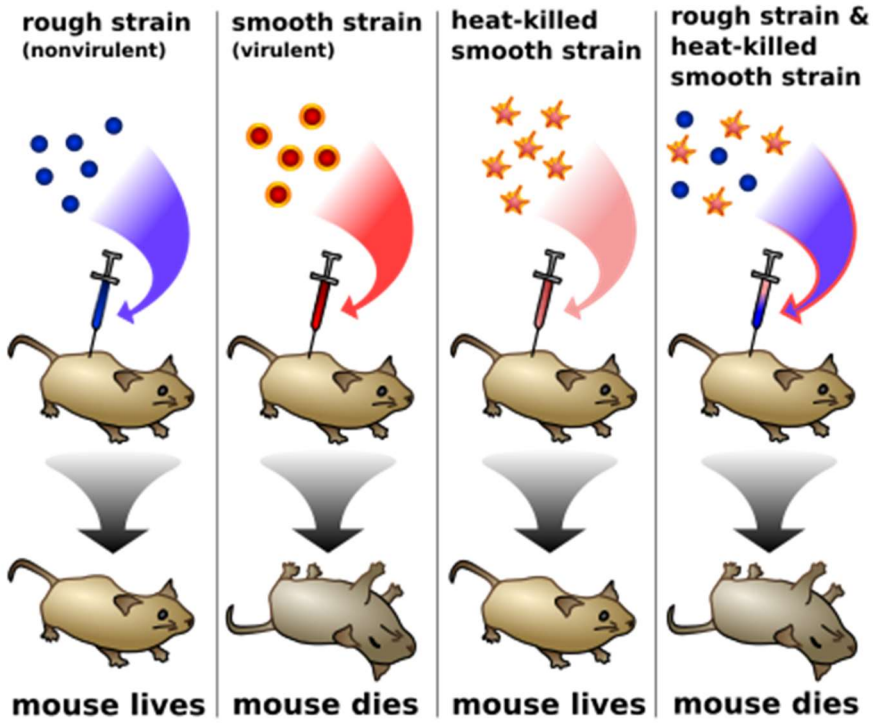
Experiment: The experiment began with the culturing of Bacteriophages (viruses that infect bacteria) in two types of medium. One set of bacteriophage (A) was cultured in a medium of radioactive phosphorus whereas another set (B) was cultured in a medium of radioactive sulfur. They observed that the first set of viruses (A) consisted of radioactive DNA but not radioactive proteins. This is because DNA is a phosphorus-based compound while protein is not. The latter set of viruses (B) consisted of radioactive protein but not radioactive DNA.

The host for infection was *E.coli* bacteria. The bacteriophages(viruses) were allowed to infect bacteria by removing the viral coats through a number of blending and centrifugation.

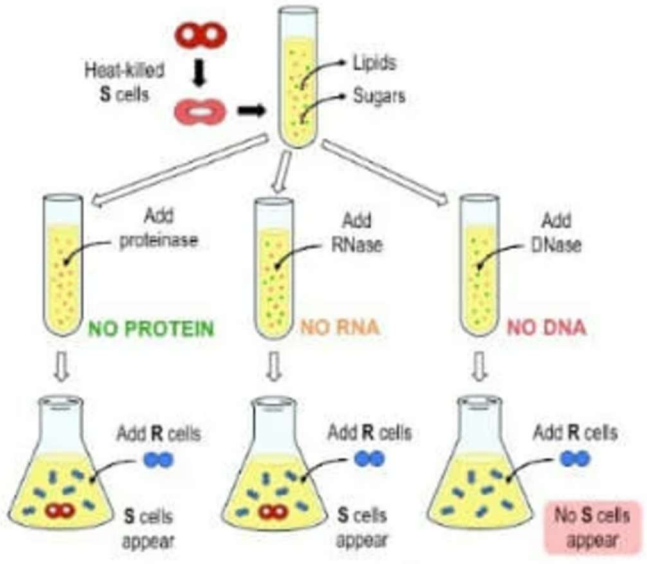
Observation: *E.coli* bacteria which were infected by radioactive DNA bacteriophages (viruses) (A) were radioactive but the ones that were infected by radioactive protein bacteriophages (viruses) (B) were non-radioactive.

Conclusion: Resultant radioactive and non-radioactive bacteria infer that the viruses that had radioactive DNA transferred their DNA to the bacteria but viruses that had radioactive protein didn't get transferred to the bacteria. Hence, DNA is the genetic material and not the protein.

Together, these experiments established **DNA** as the molecule responsible for storing and transmitting genetic information in living organisms.



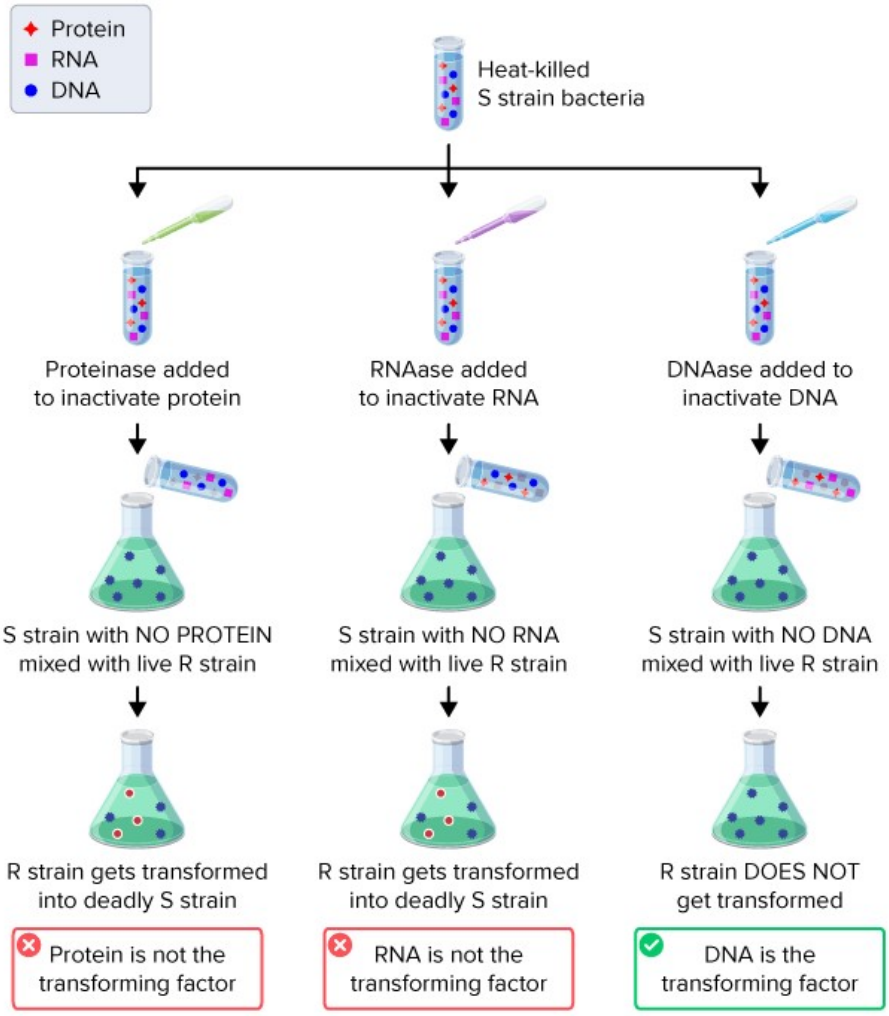
Griffith's experiment



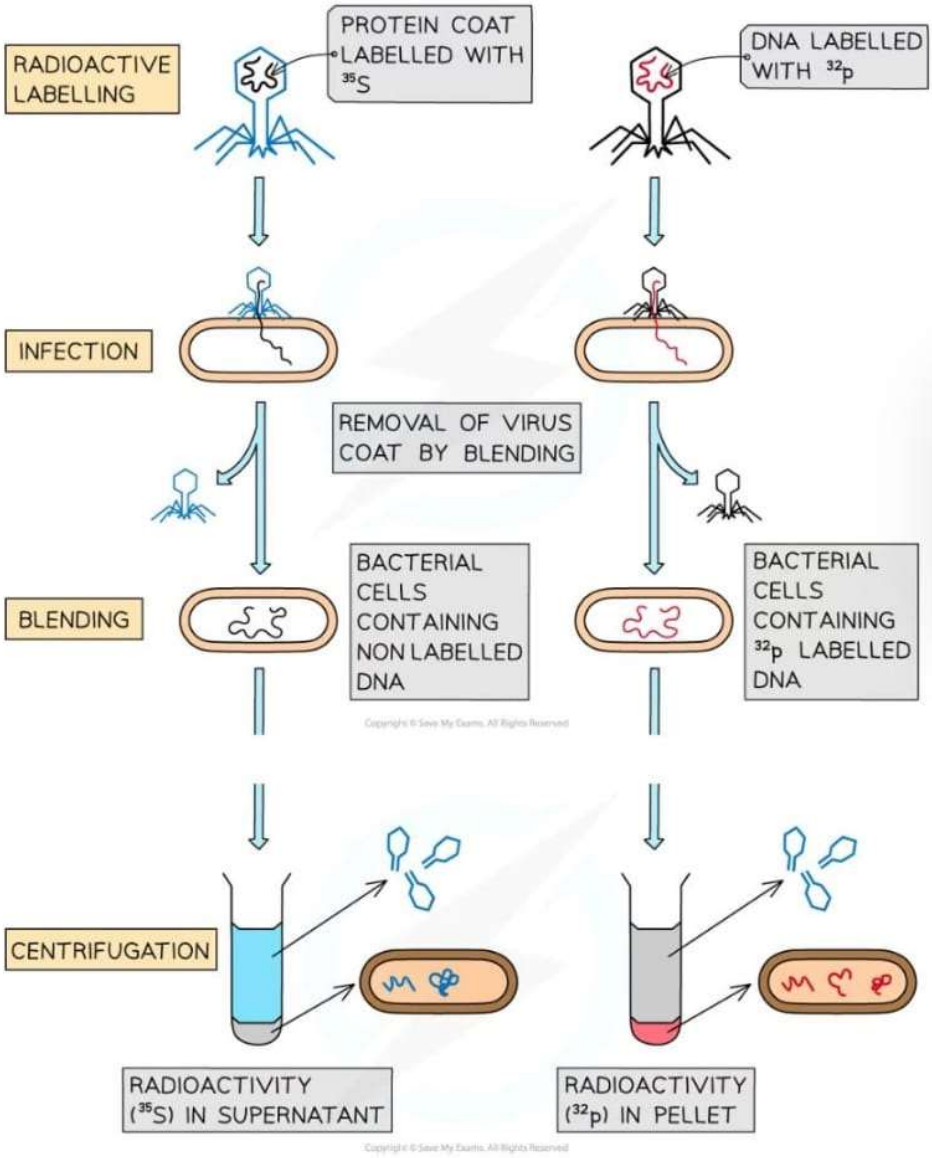
Avery, McCarty, and MacLeod Experiment

DNA

Avery, MacLeod, and McCarty's Experiment (1944) – Identifying the Transforming Principle



Avery, MacLeod, and McCarty's Experiment (1944) – Identifying the Transforming Principle



Hershey and Chase's experiment provided unequivocal proof that DNA is the heritable material

Feedback