MUTATIONS

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•ILOS

1. Define mutations
2. Discuss the classification of mutations
3. Discuss causes of mutations
4. Explainchromosomes mutations
5. Explain gene mutations
6. Summary

•Introducion

A mutation is a change in the genetic material of living organisms, and it can be small or large, with effects that can be positive, negative, or even neutral. Mutations are responsible for the genetic diversity we see in nature and play a key role in the process of evolution over time.

• What is a Mutation ?

- A mutation is a change in the DNA sequence of an organism's genome.
- Mutations can be caused by errors during DNA replication, environmental factors, or even viruses.

Classification of mutations

• Based on the survival of an individual

- **Lethal mutation:** when mutation causes death of all individuals undergoing mutation are known as lethal
- **Sub lethal mutation:** causes death of 90% individuals
- **Sub vital mutation:** such mutation kills less than 90% individuals
- **Vital mutation:** when mutation don't affect the survival of an individual are known as vital
- **Supervital mutation:** This kind of mutation enhances the survival of individual

Based on the tissue of origin

1

Somatic Mutation

A mutation occurring in somatic cell is called somatic mutation.
In asexually reproducing species somatic mutations transmits from one progeny to the next progeny

2

Germinal Mutation

When mutation occur in gametic cells or reproductive cells are known as germinal mutation.
In sexually reproductive species only germinal mutation are transmitted to the next generation

Based on the direction of the mutation

1

Forward mutation

When mutation occurs from the normal/wild type allele to mutant allele are known as forward mutation

2

Reverse mutation

When mutation occurs in reverse direction that is from mutant allele to the normal/wild type allele are known as reverse mutation

- **Causes of mutations**

1-Spontaneous Mutations

2-Induced Mutations

- **Can occur naturally without any external influence:**

Errors in DNA replication

Mistakes can happen during DNA copying in cell division, leading to base substitutions, insertions, or deletions.

Errors in DNA repair

Faults in the mechanisms that correct DNA damage can lead to mutations.

- **Physical Mutagens**

1-Ionizing radiation (e.g., X-rays, gamma rays): Breaks DNA strands or causes large-scale damage.

2-Non-ionizing radiation (e.g., UV light): Induces thymine dimers, which interfere with DNA replication.

- **Chemical Mutagens**

1. Base analogs
2. Alkylating agents
3. Intercalating agents
4. Reactive oxygen species (ROS)

Biological Agents

01

Viruses:

Some viruses, like retroviruses, integrate their DNA into the host genome, causing mutations

02

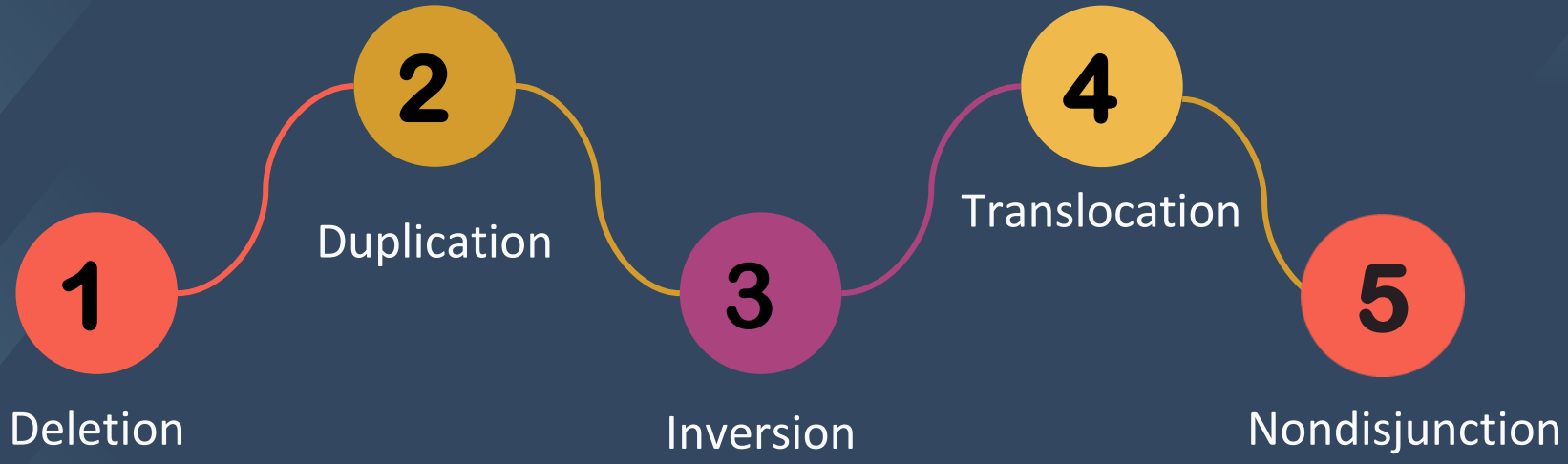
Transposons:

Mobile genetic elements that move within the genome and disrupt normal gene function

Chromosomes Mutation

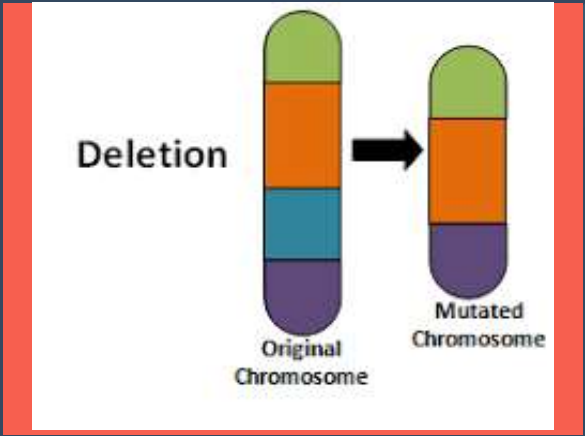
A chromosomal mutation is an abnormal change in the genetic makeup of chromosomes, whether at the level of a part of a chromosome or at the level of the entire chromosome, May lead to genetic diseases

Types of chromosome Mutation



Deletion

The loss of part of a chromosome, resulting in the loss of important genetic material



Diseases

Williams syndrome: Caused by a deletion of part of chromosome 7, it is characterized by cardiovascular problems and delayed development.

Duplication

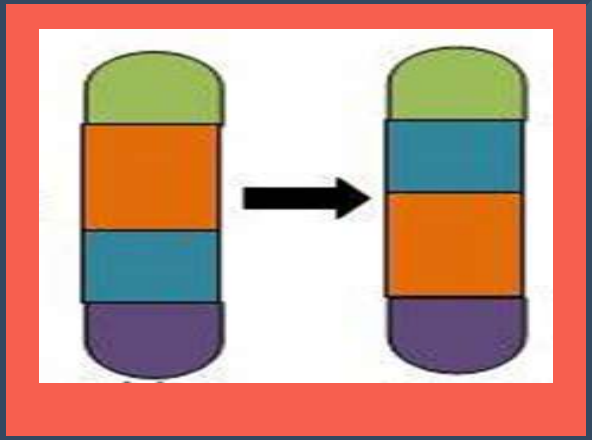
The repetition of a portion of a chromosome, resulting in an excess amount of genetic material.

Diseases

Down Syndrome : A person has three copies of chromosome 21 instead of two.

Inversion

A reversal of the order of a part of a chromosome (it is reversed

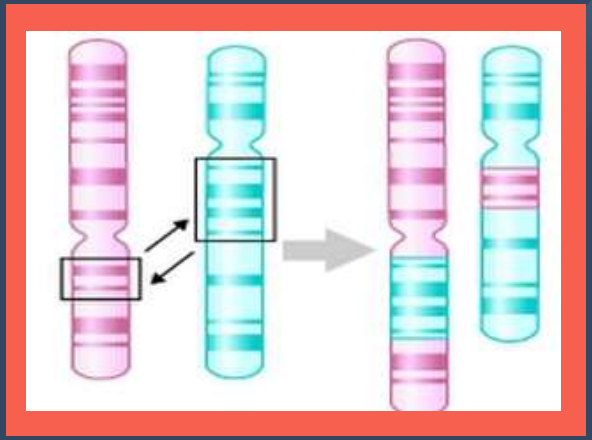


Diseases

Cri-du-chat Syndrome: The inversion occurs in chromosome 5, resulting from the deletion of a part of chromosome 5, specifically in the region called 5p

Translocation

The movement of part of a chromosome to another chromosome.

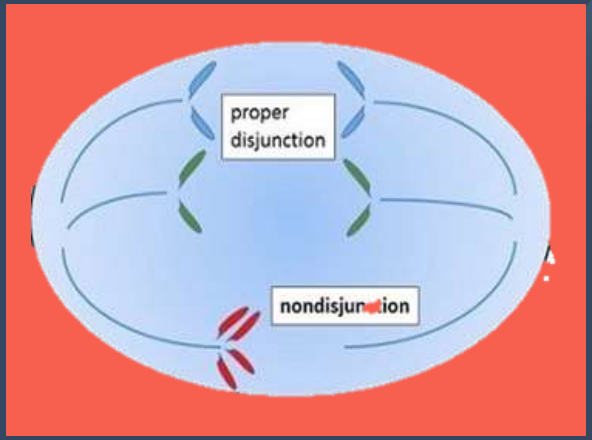


Diseases

In leukemia : translocations occur between chromosome 9 and chromosome 22, forming the Philadelphia chromosome, which increases the proliferation of cancer cells.

Nondisjunction

It is when chromosomes do not separate properly in cells.



Diseases

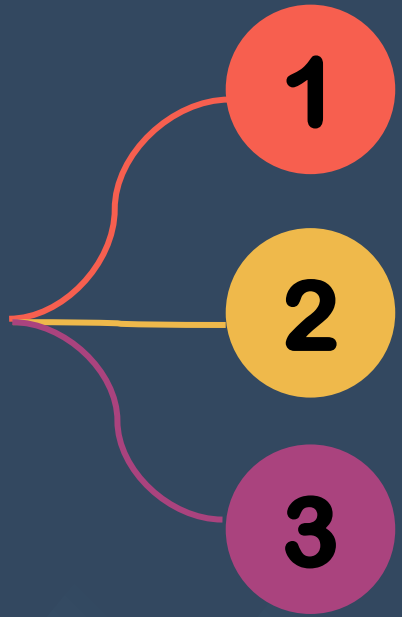
Patau Syndrome : occurs when a person has three copies of chromosome 13.

Gene Mutation

Refers to a permanent alteration in the DNA sequence that makes up a gene. These changes can occur in various ways and can have different effects on an organism.

- Types of gene mutation

Point Mutations



Silent mutations

Missense mutations

Nonsense mutations

- Types of gene mutation

Frameshift mutation

1. Insertions
2. Deletions

Point Mutations

These involve a change in a single nucleotide base in the DNA sequence.

Silent Mutations

No change in protein function

Example of Diseases

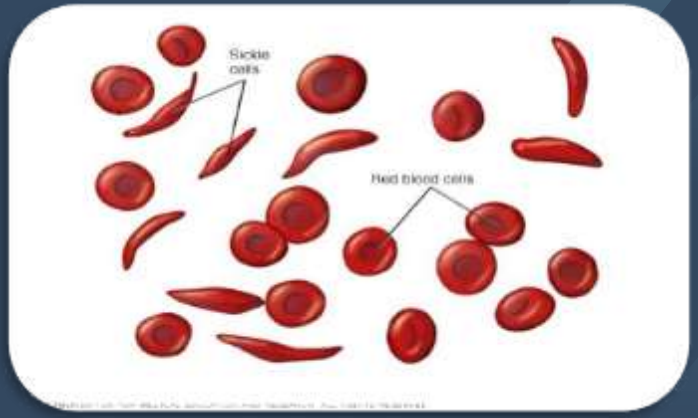
rarely cause diseases directly but may affect gene regulation or expression, contributing to conditions like cancer.

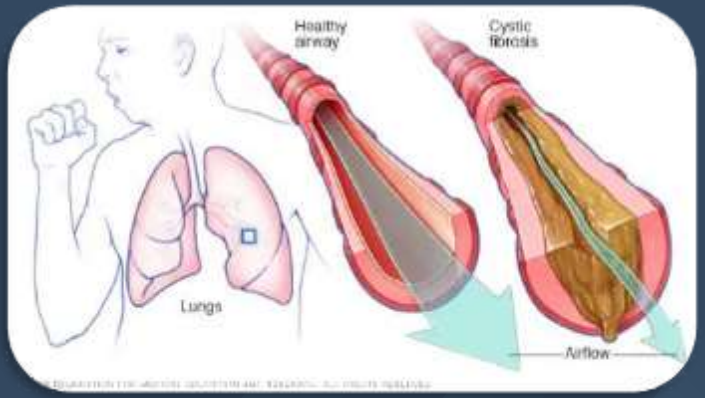
2. Missense mutations

Result in a different amino acid, potentially altering protein function.

Example Disease:

- Sickle Cell Anemia





3. Nonsense mutations

Create a premature stop codon, leading to a truncated protein.

Example Disease:

- Cystic Fibrosis

- **Frameshift Mutation**

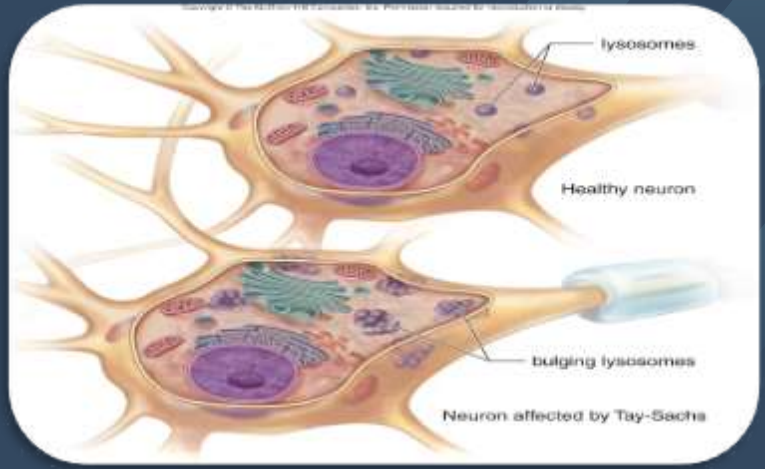
Altering the reading frame of the gene and usually resulting in a nonfunctional protein.

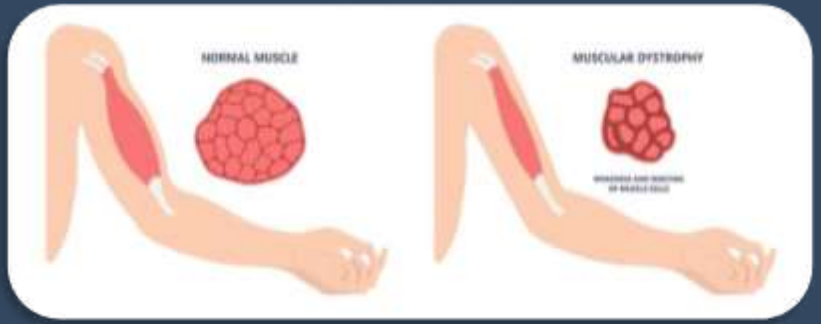
1.Insertions

Addition of one or more nucleotide bases.

Example of Diseases:

- Tay-Sachs Disease





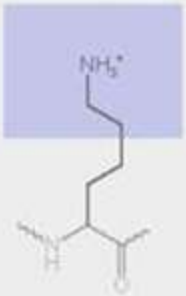
2.Deletions

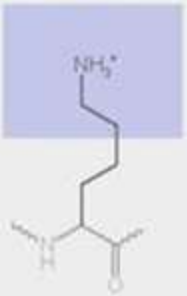
Removal of one or more nucleotide bases.

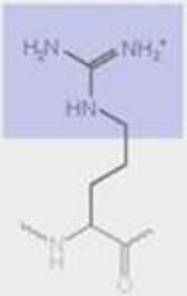
Example of Diseases:

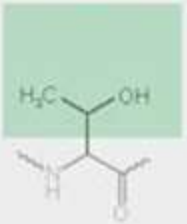
- Duchenne Muscular Dystrophy

	No mutation	Point mutations			
		Silent	Nonsense	Missense	
				conservative	non-conservative
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
protein level	Lys	Lys	STOP	Arg	Thr









basic
polar

Summary

Definition: Mutations are changes in genetic material, driving genetic diversity and evolution.

Causes: They can occur spontaneously or be induced by physical, chemical, or biological factors.

Types: Mutations are categorized by their effects, origin, direction, and scale (gene or chromosomal).

Chromosomal Mutations: Involve large-scale changes like deletions, duplications, inversions, translocations, and nondisjunction, causing conditions like **Down Syndrome** or **leukemia.**

Gene Mutations: Include point mutations and frameshift mutations (insertions/deletions), ranging from silent changes to severe diseases **(e.g., cystic fibrosis).**

Reference

- *https://www.britannica.com/science/mutation-genetics*
- *https://www.genome.gov/genetics-glossary/Mutation*
- *https://www.bioexplorer.net/chromosomal-mutations.html/*
- *https://www.Pubmed .gov.com*

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