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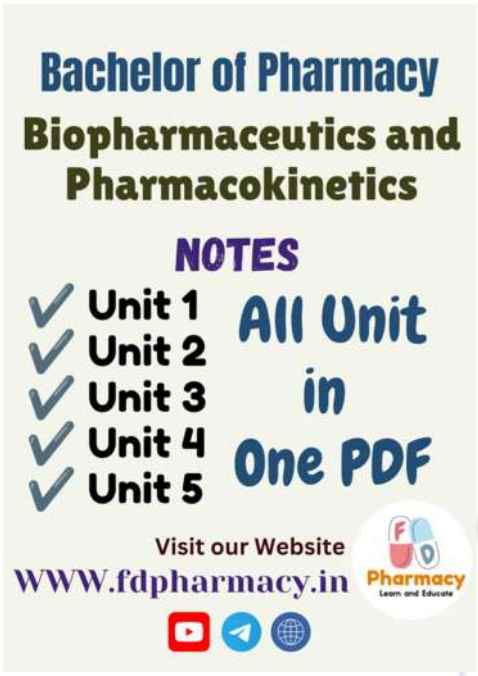
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Unit 3
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Unit 3
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PHARMACEUTICAL BIOTECHNOLOGY

UNIT 4

TOPIC :

- e) Mutation : Types of mutation/mutants.

Mutation

A mutation is a sudden, permanent, and heritable change in the genetic material (DNA or RNA) of an organism.

It alters the nucleotide sequence of genes and may affect the structure or function of proteins.

Mutations are the primary source of genetic variation and play an important role in:

- Evolution
- Adaptation
- Drug resistance
- Genetic disorders
- Cancer development

Types of Mutation

Mutations are broadly classified into:

1. Gene Mutations

- Point Mutation
- Frameshift Mutation

2. Chromosomal Mutations

- Deletion
- Duplication
- Inversion
- Translocation

Gene Mutation

- Gene mutations are changes occurring in the nucleotide sequence of a gene.
- These mutations affect a small portion of DNA and may alter protein synthesis.

Types of Gene Mutation

1. Point Mutation
2. Frameshift Mutation

1. Point Mutation

A point mutation is a mutation in which a single nucleotide base pair is changed, substituted, inserted, or deleted.
It may occur spontaneously or due to mutagens.
Point mutations are classified on two bases:

1. Based on Base Substitution
2. Based on Functional Change

A. Based On Base Substitution

1. Transition Mutation

A transition mutation occurs when one base is replaced by another base of the same type.

Examples

- Purine ↔ Purine
  - Adenine (A) ↔ Guanine (G)
- Pyrimidine ↔ Pyrimidine
  - Cytosine (C) ↔ Thymine (T)

Characteristics

- Most common type of base substitution.
- Causes minor structural changes in DNA.

2. Transversion Mutation

A transversion mutation occurs when a purine is replaced by a pyrimidine or vice versa.

Examples

- Adenine (A) ↔ Cytosine (C)
- Guanine (G) ↔ Thymine (T)

Characteristics

- Causes greater structural distortion.
- Less common than transition mutations.

B. Based On Functional Change

1. Silent Mutation

A mutation in which the altered codon still codes for the same amino acid.

Example

UUU → UUC
Both code for Phenylalanine.

Effect

- No change in protein structure.
- Usually no biological effect.

2. Missense Mutation

A mutation that changes one amino acid into another.

Example

Normal Codon: GAG → Glutamic Acid
Mutated Codon: GTG → Valine

Effect

- Produces altered protein.
- May change protein function.

Example Disease

- Sickle Cell Anemia

3. Nonsense Mutation

A mutation that converts a codon into a stop codon.

Example

UAU → UAA
(UAA is a stop codon)

Effect

- Premature termination of protein synthesis.
- Produces short, non-functional proteins.

2. Frameshift Mutation

→ A frameshift mutation occurs when one or more nucleotides are inserted or deleted from DNA, causing a shift in the reading frame of codons.

→ Since codons are read in groups of three nucleotides, any shift changes all subsequent codons.

Types Of Frameshift Mutation

1. Insertion Mutation

Addition of one or more nucleotides into DNA.

Example

Normal:
AUG – AAA – GGC
After Insertion:
AUG – UAA – AGG – C

Effect

- Reading frame shifts.
- Produces abnormal protein.

2. Deletion Mutation

Removal of one or more nucleotides from DNA.

Example

Normal:
AUG – AAA – GGC
After Deletion:
AUG – AAG – GC...

Effect

- Alters all downstream codons.
- Produces defective proteins.

Chromosomal Mutation

→ Chromosomal mutations are changes in chromosome structure involving large DNA segments.

→ They usually affect multiple genes simultaneously.

Types of Chromosomal Mutation

1. Deletion

Loss of a chromosome segment.

Effect

- Missing genetic information.
- Can cause severe abnormalities.

Example

Cri-du-chat Syndrome.

2. Duplication

A chromosome segment is repeated one or more times.

Effect

- Extra copies of genes are formed.
- Disturbs gene balance.

3. Inversion

A chromosome segment breaks off, rotates 180°, and reattaches in reverse orientation.

Effect

- Changes gene arrangement.
- Usually does not change gene quantity.

4. Translocation

A chromosome segment breaks and attaches to another non-homologous chromosome.

Effect

- Rearrangement of genetic material.
- May cause genetic disorders and cancers.

Mutants

Mutants are organisms or cells that have undergone mutation, resulting in permanent genetic changes.

Mutations may alter:

- Structure
- Metabolism
- Growth
- Drug resistance
- Virulence

Types of Mutants

1. Morphological Mutants

➢ Mutants showing changes in shape, size, color, or colony appearance.

Examples

- Change in colony color.
- Change in colony shape.
- Change in cell size.

2. Lethal Mutants

➢ Mutations that cause death of the organism or cell.

Example

Mutation affecting essential metabolic enzymes.

3. Conditional Lethal Mutants

➢ Mutants that survive under specific conditions but die under others.

Examples

Temperature-Sensitive Mutants

- Survive at one temperature.
- Die at another temperature.

Nutritional Mutants

- Require special nutrients for growth.

4. Biochemical Mutants

➢ Mutants with defects in metabolic pathways due to enzyme alterations.

Example

Auxotrophic mutants unable to synthesize certain amino acids or vitamins.

Mutagens

- A mutagen is any physical, chemical, or biological agent capable of inducing mutations in genetic material.
- Mutagens increase the frequency of mutations.

Types of Mutagens

1. Physical Mutagens

- Physical agents that damage DNA through radiation.

Mechanism

- DNA strand breaks.
- Abnormal base pairing.
- Chromosomal damage.

Examples

- X-rays
- Gamma rays
- Ultraviolet (UV) rays

2. Chemical Mutagens

- Chemical substances that alter DNA structure and nucleotide sequences.

Effects

- Base substitution
- Insertion mutations
- Deletion mutations

Examples

- Nitrous acid
- Mustard gas
- Ethidium bromide

3. Biological Mutagens

- Living organisms or biological elements capable of causing mutations.

Examples

- Viruses
- Transposons (Jumping genes)
- Certain bacteria

Importance of Mutation

1. Creates genetic variation.
2. Drives evolution and natural selection.
3. Helps microorganisms develop antibiotic resistance.
4. Useful in strain improvement programs.
5. Important in biotechnology and genetic engineering.
6. Helps in genetic research and molecular studies.
7. Contributes to adaptation in changing environments.