Unit -06

Mutation :-

Sudden heritable change in genetic material or character of an organism is called mutation.

Mutation

- **Chromosomal mutation**
  - Change in chromosome No.
    - Aneuploidy
      - → Hypoploidy
        - a) monosomy
        - b) Nullisomy
      - → Hyperploidy
        - a) Trisomy
        - b) Tetrasomy
    - Polyploidy
      - Autopolyploidy
      - Allopolyploidy
  - Change in Chromosome Str.
    - Deletion
    - Duplication
    - Translocation
    - Inversion
- **Gene mutation / point mutation**
  - → On the basis of Nature of cell
    - a) Somatic mutation
    - b) Germinal mutation
  - → On the basis of direction
    - a) forward mutation
    - b) Backward mutation
  - → On the basis of Nature of change
    - a) Substitution
    - b) Deletion
    - c) Insertion
  - → On the basis of affects:
    - a) Same sense mutation
    - b) Non-sense mutation
    - c) Frameshift mutation
  - → On the basis of origin
    - a) Spontaneous mutation
    - b) Induced mutation.

01. Chromosomal Mutation :— Such mutation which occurs due to change in chromosome Number or change in chromosome structure.

a) on the basis of change in chromosome No.

i) Aneuploidy :— It refers to change in chromosome No. due to gain or loss of one or few chromosomes.
It is of two types :—

a) Hypoploidy :— It occurs due to loss of one or few chromosomes.

Monosomy :— Decrease in chromosome number due to loss of one chromosome.
Represented as 2n-1.

Nullisomy :— Decrease in chromosome Number due to loss of a pair of Homologous chromosome.
Represented as (2n-2).

b) Hyperploidy :— It refers to change in chromosome No. due to gain of one or few chromosomes.

Trisomy :— Increase in chromosome Number due to

gain of one chromosome.
Represented by – (2n+1)

Tetrasomy :– Increase in chromosome No. due to gain of a pair of homologous chromosome. Represented as (2n+2).

ii) Polyploidy :– Occurrence of more than two sets of chromosomes, called polyploidy.
It is of two types :–

a) Autopolyploidy :–
Polyploid having more than two sets of chromosomes of same species.
AAA – Autotriploid
AAAA – Autotetraploid

b) Allopolyploidy :–
Polyploid having more than two sets of chromosome of different species.
Eg :– AABB – Allopolyploid.

b) On the basis of change in chromosome structure.

i) Deletion :– It refers to loss of a segment of chromosome. The loss of segment may

be terminal or intercalary. C D E F (Terminal deletion)

A B C D E F Deletion → A D E F (Intercalary deletion)

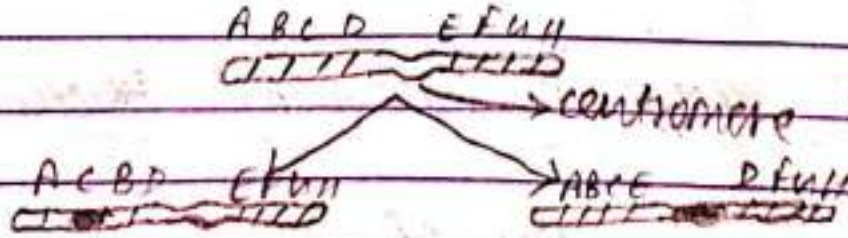
ii) Duplication :- It refers to occurence of a segment in excess of normal amount in a chromosome

A B C D E F G H Duplication → A B C B C D E F G H

iii) Translocation :- It refers to reciprocal or reciprocal exchange of segment b/w non-homologous chromosomes

A B C D E F G H Translocation → P Q R S T U V W

iv) Inversion :- It refers to rotation of a segment of a chromosome by 180° on its own axis. If it does not include centromere then it is called Paracentric inversion. If centromere included then it is called pericentric inversion.



Inverted segment (Paracentric inversion)

Inverted segment (Pericentric inversion)

c) Gene mutation or point mutation :- A gene mutation or point mutation is defined as an alternation in the sequence of nucleotides in DNA.

A) On the basis of nature of cell :-

i) Somatic mutation :- Mutation which occurs in somatic cells of an organism is called Somatic mutation. It is Non-heritable.

ii) Germinal mutation :- Mutation which occurs in reproductive / germinal cells of an organism is called Germinal mutation. It is heritable.

B) On the basis of Direction :-

i) Forward mutation :- Such mutation which changes wild genotype into mutant genotype is called forward mutation.

ii) Backward mutation :- The mutation which changes mutant genotype into wild genotype is called Backward mutation.

C) On the basis of nature of change:—

i) Substitution mutation:— Such mutation which replaces a base by another base is called substitution mutation.
It is of following types:—
a) Transition:— Replacement of purine base by another purine or pyridine base by other pyridine base in called Transition.

b) Transversion:— Replacement of purine by pyridine or vice versa, called Transversion.

ii) Deletion:— mutation in which a base is lost.
iii) Insertion:— mutation in which a base is added.

D) On the basis of effects:—

i) same-sense mutation:— Mutation which fails to change in the nature of Amino acids specified a codon is called same-sense mutation.

ii) mis-sense mutation:— Mutation which causes change in nature of amino acid of a

polypeptide chain, called mid-zone mutation.

iii) Non-sense mutation:- mutation which changes a codon specifying an amino acid into either of termination codon (UAA, UGA, UAG), called Non-sense mutation.

iv) Frameshift mutation:- mutation which changes reading frame from point of insertion or deletion is called frameshift mutation.

B) On the basis of origin:-

i) Spontaneous mutation:- If origin of mutation is not known, called Spontaneous mutation.

ii) Induced mutation:- mutation which occurs in presence of mutagens, called induced mutation.

Ques:) Lac Operon model of E. coli :-

Lac operon model was proposed by Jacob and Monod.

The operon model

was based on the study of lac operon in E. coli. lac operon is an example of inducible system.

→ The lac operon has three structural genes and two contiguous units promoter and operator.

→ The structural gene of lac operons are Z, Y and a. The gene Z, Y and a have information for synthesis of β-galactosidase, permease and transacetylase respectively.

→ β-galactosidase is required for hydrolysis of lactose into glucose and galactose.
→ permease is responsible for pumping lactose from surrounding to E. coli.
→ Transacetylase function is not clear.

There are two cases:—
(i) in absence of lactose

b) In presence of lactose.

b) In absence of lactose.

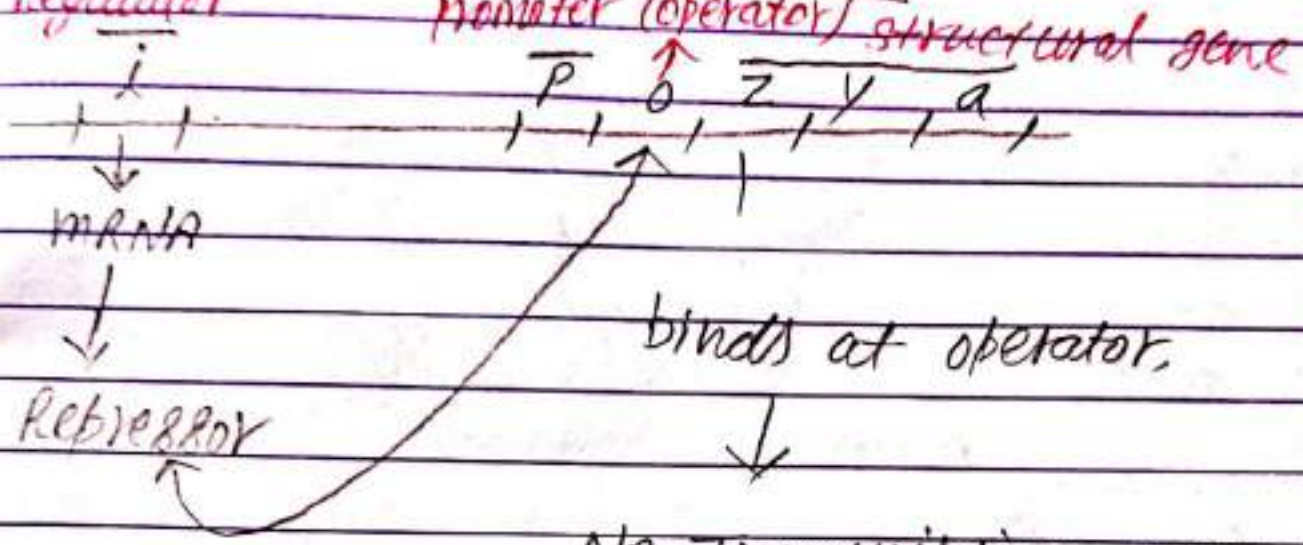
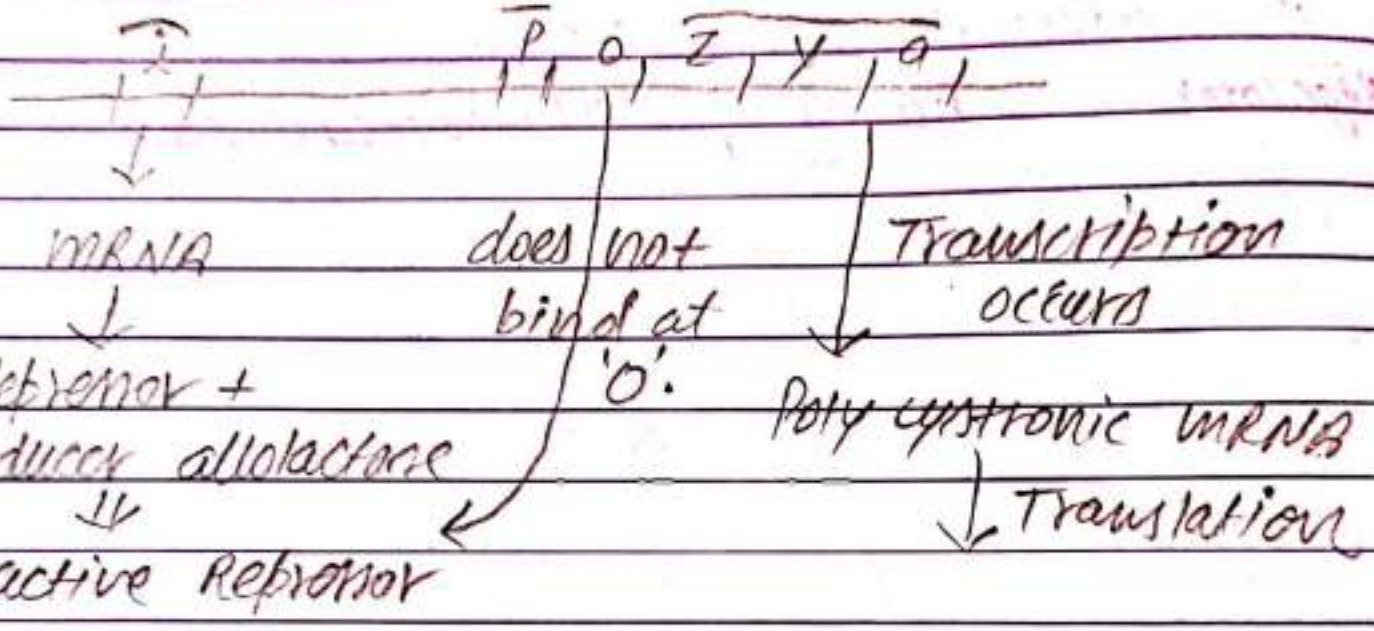


fig:- Lac operon — in absence of lactose.

In absence of lactose, transcription of structural gene is not allowed, because repressor bind at operator. So, RNA polymerase is strictly prohibited / inhibited by repressor to move ahead and catalyse the process of transcription. In this way, transcription of structural gene Z, Y and a does not takes place in absence of lactose.

Operon in presence of lactose:-



When lactose is added to the medium on which E. coli is growing, transcription of structural gene is induced. Induction of transcription takes place in following manner:-

→ permeases present in bacterial cell allows entry of lactose in the cell. Where lactose converted into allolactose.

→ It is allolactose which acts as inducer. As inducer it binds with repressor.

→ In such condition, the repressor fails to binds at promoter and initiates transcription of structural genes Z, Y and A.

* Translation of Proteins in Prokaryotes:-

Synthesis of polypeptide chain by utilising information in form of sequence of codons in mRNA is called Translation.

Translation involves following four steps:—

01.) Activation of Amino acid
02.) Initiation
03.) Elongation
04.) Termination

01.) Activation of Amino acid:—

Amino acid is attached to 3' end of tRNA under catalytic effect of specific amino acyl tRNA synthetase. This is called

activation of Amino acid.

Amino acid (AA) + tRNA + ATP $\xrightarrow[Mg^{2+}]{\text{Aminoacyl tRNA synthetase}}$ AA-tRNA + AMP + PPi

Attachment of Amino acid to tRNA is significant because it activates amino-acid for polypeptide chain formation.

For initiation methionine is attached to $tRNA^{fmet}$, it is catalysed by methionyl $tRNA^{fmet}$ synthetase, ATP and Mg^{2+} are required.

Methionine (met) + $tRNA^{fmet}$ + ATP $\xrightarrow[Mg^{2+}]{\text{Methionyl tRNA synthetase}}$ met-$tRNA^{fmet}$ + AMP + PPi

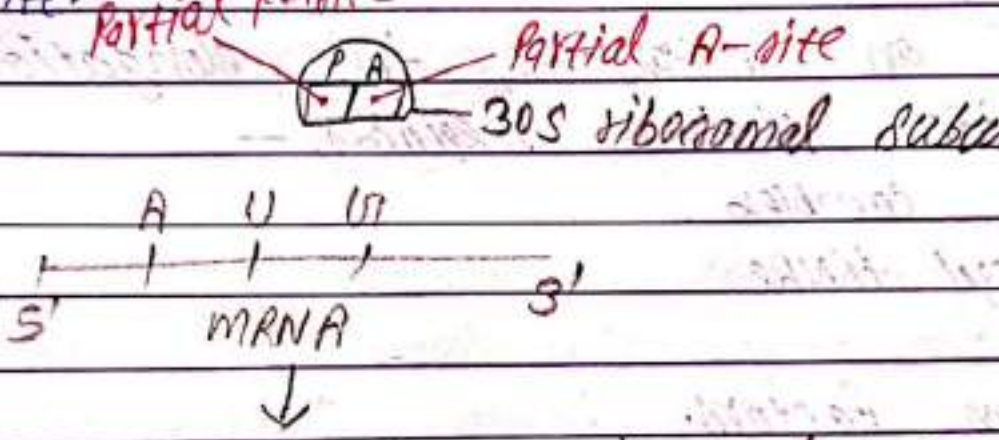
In prokaryotes, formylation occurs.

met-$tRNA^{fmet}$ + N-Formyl tetrahydro folate $\xrightarrow{\text{formylases}}$ fmet-$tRNA^{fmet}$ + tetrahydro-folate.

Initiation:-

Ribosome has two types of sites:-
P (Peptidyl site)
A (amino acyl) site

30 S ribosomal subunit binds with mRNA toward its 5' end. f-met tRNA^{fmet} has unique ability to bind at P-site.



initiation complex

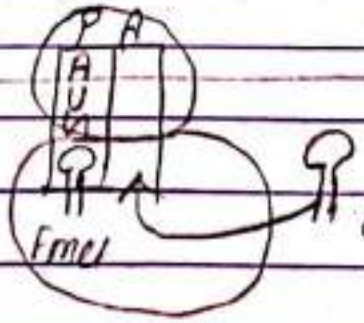
The correct position of initiation codon is Partial P-site.

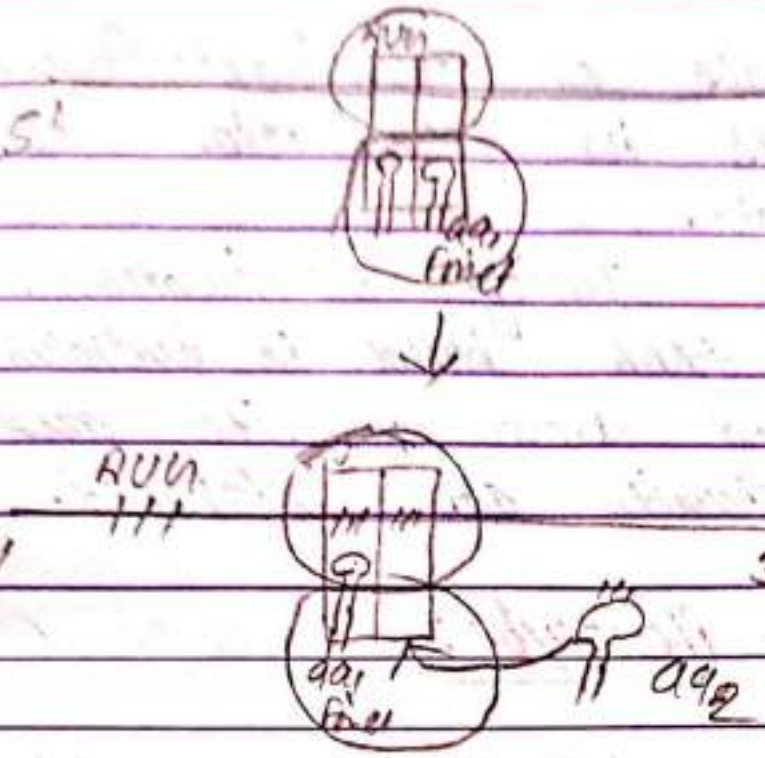
After Removal of initial factor (IF), 50 S ribosomal subunits bind with 30 S ribosomal subunits and forms in formation of 70 S ribosome.

03) Elongation :—

In Prokaryotes, the elongation of Polypeptide chain requires:—

Initiation complex
amino acyl tRNAs
GTP
Elongation factors.





P site has fmet $tRNA^{fmet}$ and A-site is occupied by amino acyl tRNA. f-methionine is separated from $tRNA^{fmet}$ and peptide bond is formed b/w -COOH of f-methionine and $-NH_2$ of amino acid of A site. As a result $tRNA^{fmet}$ is removed from p-site. peptide bond is catalysed by peptidyl transferase.

(4) Termination :-

Elongation of polypeptide chain continues till the arrival of one of the termination codon (UAG, UGA & UAA).

at A-site. Presence of termination codon at A-site results in termination of Polypeptide chain.

During termination, terminal peptidyl tRNA bond is hydrolysed. tRNA is removed from P-site and both ribosomal subunits are separated.

* Genotype and Phenotype :-

Genotype :- The term Genotype was coined by Wilhelm Johansen.

Genetic constitution of an organism is called as Genotype.

Eg :- In pea plant, the gene representing the colour of the flower has two alleles.

Violet flower — 'V'

White flower — 'v'

Phenotype :-

Observable characteristics of an organism is called Phenotype.

Phenotype = Genotype + Environment.

Eg:- Colour, Shape etc.

Character	Genotype	Phenotype
01. meaning	The hereditary information of organisms, in the form of the gene in the DNA and remain same throughout life	The visible characteristics are phenotype, which is the expression of the genes. but these character change with the period like the stage from infant to adult.
02. Consist of	The hereditary characters of the organisms, which may or may not get expressed in the next generation. The same genotype produces the same phenotype in a particular environment.	These characters are not inherited. Thus we can say that same phenotypes may or may not belong to the same genotype.

03. It appears	Inside the body, as genetic material.	Outside the body, as physical appearance.
04. Inherited	These are partly inherited from an individual to the offspring as one of the two alleles, during the reproduction process.	The phenotype is not inherited.
05. Determined by	Using scientific methods like polymerase chain Reaction (PCR) to find out the kind of genes on the allele.	By observing the organisms.
06. Affected By	It is affected by genes.	It is affected by the genotype and other environmental conditions.

of. Examples.	RH disease, blood groups, eye colour, Height etc.	weight, body physique, eye colour, hair colour.

* **Adaptation :—** An inherited characteristics that helps an organism to survive long enough to reproduce more successfully in its changing environment.

Types of Adaptation :—

01. Structural adaptation
02. Behavioral adaptation

Adaptation for animals

01.) **Structural Adaptation :—** Actual body parts or colouration that helps an organism survive in their environment.

Eg :—

1) camouflage :— Blending in with the environment

for protection from predators.
Use:- obtaining food and protection.

b) Copying / mimicry:- copying a behaviour or appearance, for protection and obtaining food.

c) Bent hind legs:- prey run fast to escape and predators run fast to catch prey.

d) Behavioural Adaptation:- ways an organism act to help them survive in their environment
Eg:-

a) Migration:- seasonal or periodic movement of animals in response to change in climate or food availability or to ensure reproduction.

b) Hibernation:- Adaptive winter survival technique where animal becomes inactive and all body processes slow down.

c) Living in a group:- more eyes in a group to watch out for prey or predator, protection.

Adaptation for Plants:—

a) Structural Adaptation:—

a) Adaptation on the body:— Holdfast, empty space for water storage, tallness, thorns.

b) Seeds:— all seeds have adaptation to better enable it to survive long enough to plant itself

c) Protection:— Thorn, bad taste, colouration

b) Behavioural Adaptation:—

a) Tropism:— movement of a plant toward or away from a stimulus. Toward is called positive, away is called Negative.

phototropism:— When the plant senses light and the shoots grow toward the light source.

b) Dormancy:— A state of rest or inactivity.

* Genetic method of classification bacteria.
(Genotypic classification)

01) Ribosomal RNA Sequence analysis:— This has emerged as a major method for classification. It

has been used to establish a phylogenetic tree. In addition it is now also used to rapidly diagnose the pathogen responsible for an infection, to help select appropriate therapy and to identify non cultivatable microorganisms.

02.) Universal Phylogenetic tree:—

Woese has developed a "universal Phylogenetic tree" for all living organisms that establishes a tripartite division of all living organisms — Bacteria, archaea, eucarya. His work is based on a comparison of 16S rRNA sequences. These sequences are highly conserved and undergo change at a slow, gradual and consistent rate. They are therefore useful for making comparison among the different living organisms.

03) DNA — DNA hybridization.

04.) G + C content —