

MUTATION

Definition: Any sudden hereditary change in genetic material or character of an organism is called Mutation.

The term Mutation was introduced by Hugo de Vries a Dutch Scientist (1848-1935). The term was used for large spontaneous inheritable change which occur suddenly in naturally reproducing Population & Propounded 'Mutation Theory'. The term first coined by De Vries in 1900 by his observation in *Oenothera*. Systematic study of mutation was started in 1910 when Morgan genetically analyzed white eye mutant in *Drosophila*. H. J. Muller induced mutation in *Drosophila* by using X-Rays in 1927 & he was awarded with nobel Prize 1946.

Types: - 1) Somatic mutation → A mutation occurring in somatic cell is called somatic mutation. In asexually reproducing sps. Somatic mutations transmits from one progeny to next progeny.

2) Germinal mutation → When mutation occur in germ cells or reproductive cells are known as germinal mutation. In sexually reproducing species only germinal mutation is transmitted to the next generation.

③ Forward Mutation: - When mutation occurs from the normal/wild type allele to mutant allele are known as forward mutation.

④ Reverse Mutation: - When mutation occurs in reverse direction i.e. from mutant allele to the normal/wild type allele are known as reverse mutation.

Type of Trait affected: -

1) Visible mutation - Those mutation which affects on phenotypic characters & can be detected by normal observation are known as visible mutation.

2. Biochemical mutation :- Mutation which affect the production of biochemicals & which does not show any phenotypic character are called biochemical mutation.

Chromosome mutation

Any change in structure either loss or gain of segment is chromosome mutation. It may be differentiated into

5 types.

1. Deletion
2. Inversion
3. Translocation
4. ~~A~~ Nondisjunction
5. Duplication.

1) Deletion :- Due to breakage a piece of segment of a chromosome is lost.

2) Inversion :- The chromosome segment breaks off. The segment flips around backwards & then the segment reattaches.

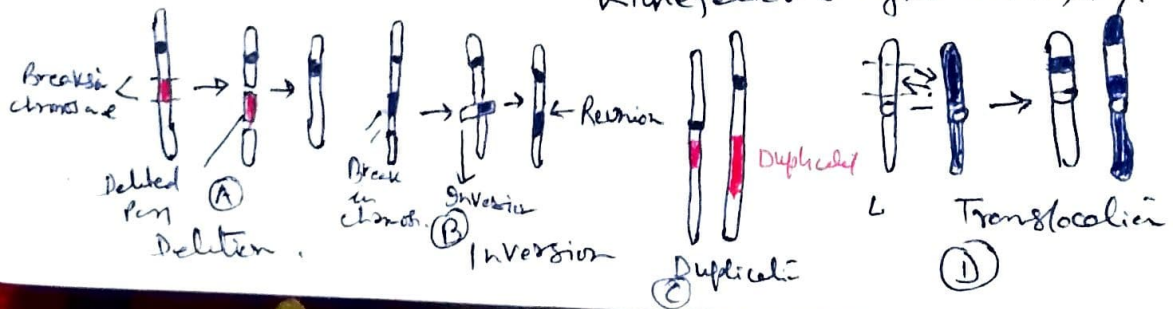
3) Duplication :- It occurs when a gene sequence is repeated.

4) Translocation :- In this two non-homologous chromosomes are involved. Part of one chromosome is transferred to another chromosome.

5) Nondisjunction :- Failure of chromosomes to separate during meiosis. It causes gametes to have too many or too few chromosomes. It cause so many disorders like : Down Syndrome - Three 21st.

Turner Syndrome - Single x-chromosome.

Klinefelter's Syndrome - xxy.



Molecular Basis of Mutation.

Due to gene mutations it can be classified as:

1. Point mutations.
2. Substitutions.
3. Insertions.
4. Deletions &
5. Frame shift mutation

1. Point Mutation: change of a single nucleotide. In this type deletion, insertion or substitution of one nucleotide in a gene. It occurs in haemoglobin gene. Sickle cell disease (anaemia) is result of one nucleotide substitution.

Frame Shift Mutation: Inserting or deleting one or more nucleotides in a DNA sequence.

It changes the 'reading frame' like changing a sentence. Due to this the protein built incorrectly.

EX: -

Original:

The fat cat ate the wee rat.

Frame shift. 'a' added.

The fat caa tet hew eer at (change the sequence of triplet code)

Substitution Mutations.

Normal DNA CGA - TGC - ^ATC
Alanine - Threonine - Stop.

Mutated DNA CGA - TGC - TTC
Alanine Threonine Lysine

It is a substitution mutation. A single nitrogenous base is substituted for another in a triplet code. It effect the amino acid A is substituted by Thymine & amino acid Lysine is formed.