MUTATION

A sudden, inheritable change in an organism is known as a mutation. Darwin called them "sports," while Bateson referred to them as "saltation" or "discontinuous variations."

Hugo de Vries discovered mutations in Oenothera lamarckiana (Evening Primrose). He identified 834 mutations in 54,343 plants of Oenothera. However, later researchers realized that what De Vries called 'mutations' were actually chromosome aberrations and polyploids.

Types of mutations :

1. **On the basis of direction, mutation involves two types**
   - (i) **Forward mutation :** Wild type – Mutant type
   - (ii) **Backward mutation :** Mutant type – wild type.

2. **On the basis of dominance or recessiveness.**
   - (i) **Dominant mutation** (ii) **Recessive mutaiton**

3. **On the basis of tissue :**
   - (i) **Somatic :** It takes place in somatic cell or Vegetative cell . It does not inherit in the next generation but in plants, It can transmit next generation through vegetative propagation.
   - (ii) **Germinal :** It occurs in germinal cell or reproductive cell. It transmits or inherits from generation to generation.

4. **On the basis of cytology mutation involves following types.**
   - (A) **Chromosomal Mutation** (B) **Genomatic Mutation** (C) **Gene Mutation**

(A) Chromosomal mutation or chromosomal aberration

Structural changes & Numerical changes of chromosomes include in this category. Structural changes of chromosomes involve following types.

(i) Intrachromosomal type :

Deletion : A part of chromosome is lost from terminal part (Terminal deletion) or from intercalary part (Intercalary deletion)

Ex: (i) Deletion of a segment of short arm of V^{th} pair of chromosome in human causes Cri du chat syndrome(cat cry syndrome) in child

Ex: (ii) Notched wing in Drosophila.

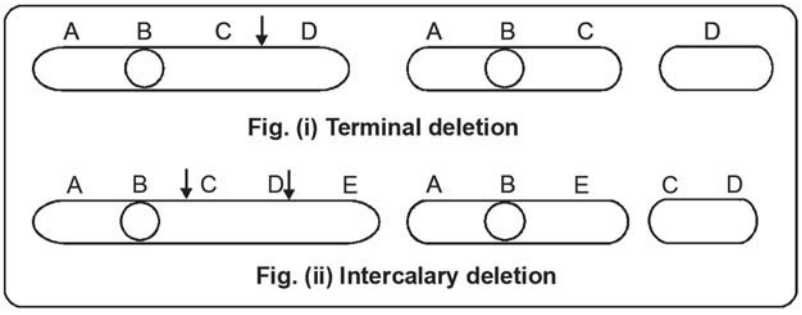

Fig. (i) Terminal deletion

Fig. (ii) Intercalary deletion

Inversion : In this type chromosome break at two intercalary places. The broken segment is inverted up to 180º and rejoined at chromosome. It is called inversion. It is of two types.

Paracentric inversion : Inversion without centromere.

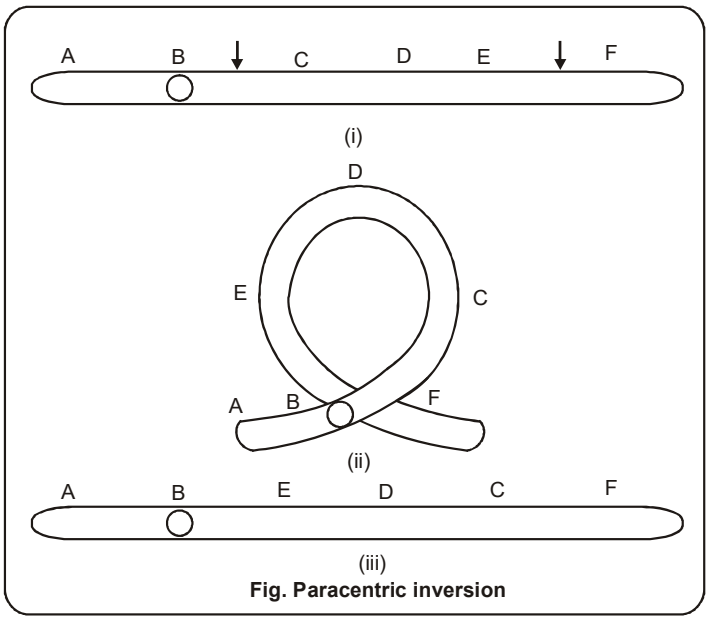

Fig. Paracentric inversion

Pericentric inversion : Inversion with centromere.

(ii) Interchromosomal type :

a) Duplication :

It occurs due to addition of a part of chromosome. Ex: Development of Bar eye in Drosophila. It involves following types.

Tendem duplication : When a chromosomal segement appears two time in a chromosome it is called Tendem duplication.

Reverse tendem duplication : If the sequence of duplicate part is reverse it is called Reverse tendem duplication.

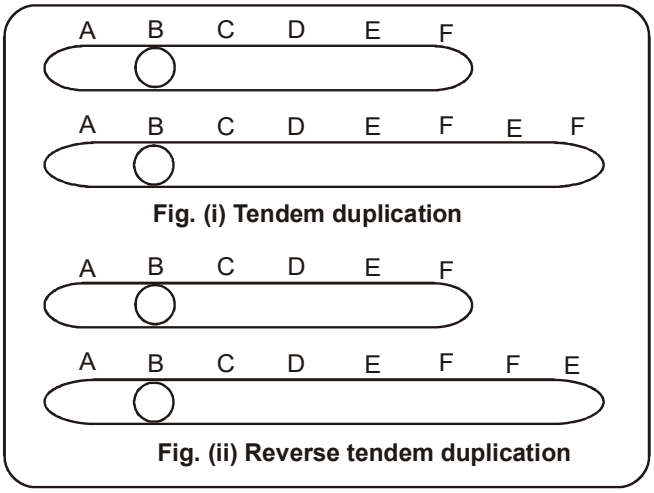

Fig. (i) Tendem duplication

Fig. (ii) Reverse tendem duplication

Displaced Duplication : Duplicating segment is inserted away from the same chromosome.

Transposed Duplilcation : Duplicating segment is inserted in a nonhomologous segment.

b) **Translocation :**

When the exchange of segments occur between two non homologous chromosomes. It is called translocation.

It is of two types.

Simple translocation : In which a segment of a chromosome transfers to the another nonhomologous chromosome.

Reciprocal translocation : In which a mutual exchange of chromosomal segments between two non-homologous chromosomes. Ex: Chronic myeloid leukemia is due to reciprocal translocation in between 9th and 22nd chromosomes.

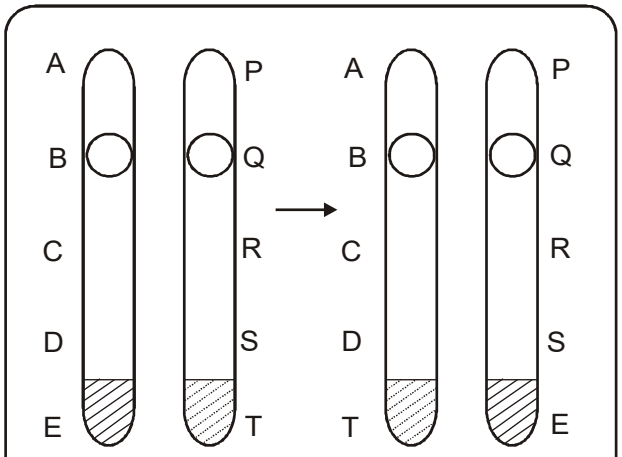

Fig. Reciprocal translocation

(B) Genomatic Mutation

(Changes in chromosomal Number) It involves aneuploidy and Euploidy (polyploidy).

1. **Aneuploidy :** In which one or few chromosomes are either deficient or in excess in a species. It is of two types.

(a) **Hypoploidy :** In which either one or few chromosomes are deficient. It involves two types.

Monosomic (2 n – 1) : In which one chromosome is deficient. Ex: Turner's syndrome (44 + X 0), It is also found in Gossypium.

Nullisomic (2n – 2) : A pair of chromosome is deficient.

(b) **Hyperploidy :** In which one or few chromosomes are in excess. It is of two types.

Trisomy (2n + 1) : One chromosome is additional in a pair of chromosome. it represents as 2n + 1.

Ex: Datura, In human Trisomy in 21^{st} pair,Trisomy in 18^{th} pair and Trisomy in 13^{th} pair are respectively called down's syndrome. Edward syndrome and patau syndrome. Double trisomic has two different chromosomes in triplicate (2n + 1 + 1).

Tetrasomic (2n + 2) : A pair of chromosome is additional or a chromosome is found fourtimes

Ex: Super female 44 + XXXX, Double tetrasomic (2n + 2 + 2).

Pentasomic (2n + 3) : A chromosome occurs five-time Ex: superfemale (44 + XXXXX) rare.

Point of remember

(1) **Double monosomic :** In double monosomic one chromosome is deficent in each chromosome of a pair. It represents as 2n – 1 – 1.

(2) **Mixed Aneuploids :** In which both hypoploidy and hyperploidy occur in two different pair of chromosome. Ex: 2n + 1 A – 1B.

(3) **Euploidy :** In which chromosome number is exact multiple of genome Ex: monoploidy, diploidy, polyploidy.

Polyploidy :

When an organism has more than two sets of chromosomes, we call it polyploid, and this condition is known as polyploidy. Polyploidy is categorized based on the number of genome occurrences, such as triploid (3n), tetraploid (4n), pentaploid (5n), hexaploid (6n), and so on.

In nature, polyploidy happens when chromosomes don't separate properly during anaphase, either due to nondisjunction or the absence of spindle formation.

Polyploids with an odd number of genomes, like triploids and pentaploids, are unable to reproduce sexually and rely on vegetative propagation. Examples include plants like Banana and Pineapple.

Polyploidy involves three types :

(i) **Autopolyploidy** (ii) **Allopolyploidy** (iii) **Autoallopolyploidy**

(i) Autopolyploidy :

It is a numerical increase of the same genome such as Autotriploidy (AAA) Ex: Rice, Gram, Maize.

(ii) Allopolyploidy :

It is formed by hybridisation between two species followed by doubling of chromosomes such as (AABB) Ex: Wheat, Tobacco, artificially produced two allopolyploids are Raphanobrassica and Triticale.

(iii) Autoallopolyploidy :

One genome is in more than diploid state such as (AAAABB) Ex: Helianthus tuberosus.

Point of remember

Polyploidy can be artificially induced by application of colchicine and granosan.

(C) Gene Mutations

Quick and lasting alterations in the gene or cistron structure, caused by changes in the sequence or type of nucleotides, are known as gene mutations. The majority of gene mutations involve a change in a single nucleotide, and these are referred to as point mutations. When mutations occur in more than one base pair, they are called gross mutations. Gene mutations typically happen during the DNA replication process, earning them the name "copy error mutations."

The first recorded instance of a point mutation was documented by Seth Wright in 1791. He observed a lamb with short legs, known as an ancon sheep.

Mutagens

Mutations can be caused by certain things called mutagens or mutagenic agents. There are two main types of mutagens:

Physical Mutagens:

Radiations are a significant type of physical mutagen. H.J. Muller, who first used X-rays to increase the mutation rate in Drosophila, started a new field in inducing mutations. Muller is often referred to as the father of Actinobiology.

Chemical Mutagens:

There are many chemical mutagens, and they can be more harmful than radiations. Mustard gas, used by C. Auerbach et al. during World War II, was the first chemical mutagen. Chemical mutagens fall into two groups:

(a) Those that can cause mutations in both replicating and non-replicating DNA, like nitrous acid.

(b) Those that only cause mutations in replicating DNA, such as acridine dyes and base analogues.