PRINCIPLES OF INHERITANCE AND VARIATION

MUTATION

MUTATION :

Sudden inheritable change in an organism is called mutation.

Darwin coined the term **sports** and **Bateson** coined the term **saltation or discontinuous variations** for them.

Mutation was discovered by **Hugo de vries** in **Oenothera lamarckiana (Evening primerose).** He observed 834 mutations in 54343 plants of **Oenothera**.

Later workers found that 'mutations' observed by De Vries were actually **chromosome aberration 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 :

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

(a) Intrachromosomal type :

(I) 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.

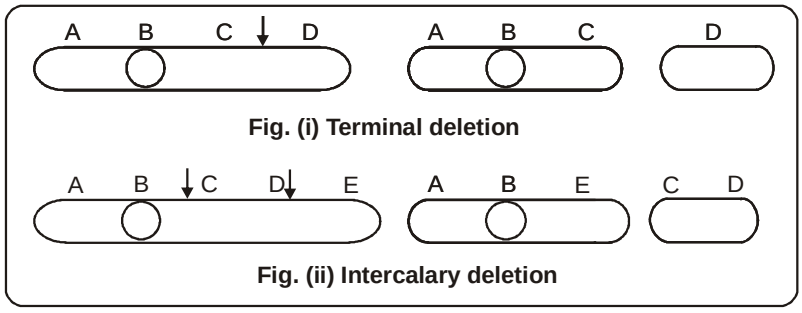

Fig. (i) Terminal deletion

Fig. (ii) Intercalary deletion

(II) 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.

(i) Paracentric inversion : Inversion without centromere.

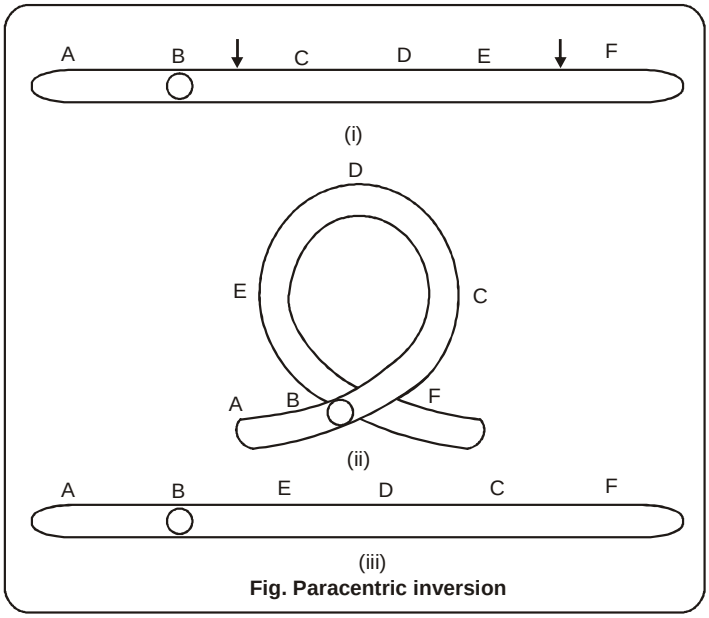

Fig. Paracentric inversion

(ii) **Pericentric inversion :** Inversion with centromere.

(b) Interchromosomal type :

(I) Duplication :

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

It involves following types.

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

(ii) **Reverse tendem duplication :** If the sequence of duplicate part is reverse it is called Reverse tendem duplication.

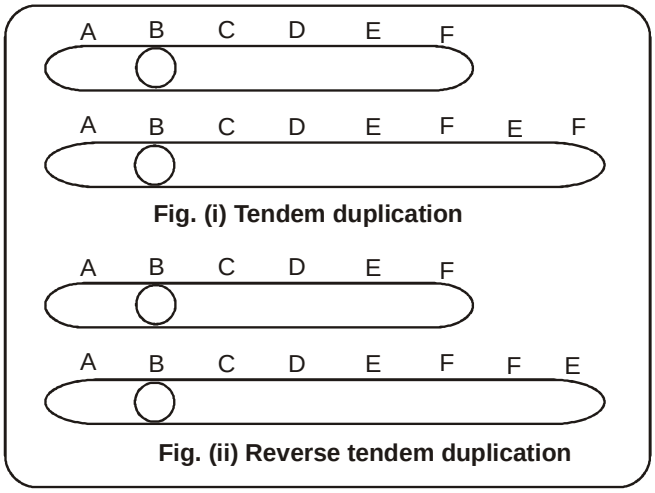

Fig. (i) Tendem duplication

Fig. (ii) Reverse tendem duplication

(iii)**Displaced Duplication :** Duplicating segment is inserted away from the same chromosome.

(iv) **Transposed Duplilcation :** Duplicating segment is inserted in a nonhomologous segment.

(II) Translocation :

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

It is of two types.

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

(b) **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.

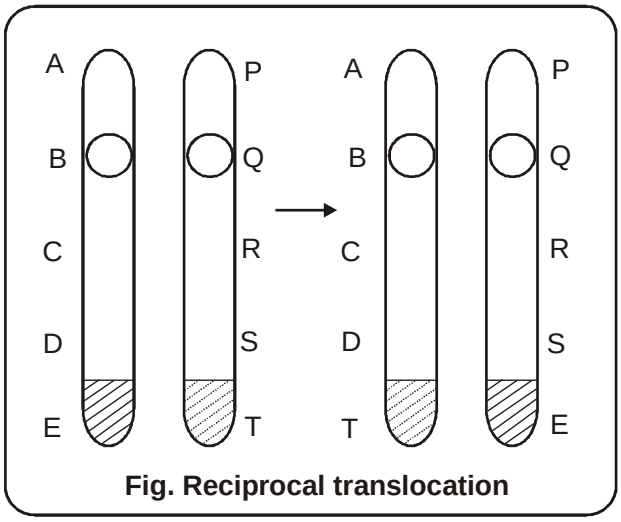

Fig. Reciprocal translocation

(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.

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

(ii) **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.

(i) 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)**.

(ii) 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).**

(iii)**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 :

If an organism has more than two sets of chromosomes, It is called Polyploid this phenomenon is called polyploidy.

On the basis of occurence of number of genome, polyploid is called **triploid (3n), tetraploid (4n), Pentaplaoid (5n), Hexaploid (6n)** etc.

In nature, polyploidy appears due to the failure of chromosomes to separate at the time of **anaphase** either due to **nondisuction** or due to **no formation of spindle**.

Polyploids with odd number of genomes **(Ex: triploids, pentaploids)** are sexually sterile. so that they perform reproduction by vegetative propagation **Ex: Banana, 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 :

Sudden stable changes in the structure of gene or cistron due to change in nucleotide sequence and nucleotide type are called gene Mutations.

Most of the gene mutations include change in single nucleotide. These are called **point mutations.** If mutations takes place in more than one base pair is called **gross mutation**. Usually Gene mutations appear during replication of DNA therefore It is called **copy error mutation.**

Seth Wright (1791) firstly recorded point mutation. He observed **short legged lamb (ancon sheep).**