Chromosomal Mutation-I

(Cytogenetics : Changes in Structure of Chromosomes)

Genetics makes extensive use of deviations from the norms, and the study of chromosomes is no exception. The chromosomes of each species has a characteristic morphology (structure) and number. But, sometimes due to certain accidents or irregularities at the time of cell division, crossing over or fertilization some alterations in the morphology and number of chromosomes take place. The changes in the genome involving chromosome parts, whole chromosomes, or whole chromosome sets are called **chromosome mutations**.

Chromosome mutations are inherited once they occur and are of the following types :

A. Structural changes in chromosomes :

1. Changes in number of genes

(a) Loss : deletion
(b) Addition : Duplication

2. Changes in gene arrangement :

(a) Rotation of a group of genes 180 within one chromosome : inversion

(b) Exchange of parts between chromosomes of different pairs : translocation.

B. Changes in number of chromosomes :

1. Loss, or gain, of a part of the chromosome set (aneuploidy)

2. Loss, or gain, of whole chromosome set (euploidy)

(a) Loss of an entire set of chromosomes (haploidy)

(b) Addition of one or more sets of chromosomes (polyploidy).

Both types of changes (structural and numerical) in chromosomes can be detected not only with a microscope (**cytologically**) but also by standard genetic analysis.

STRUCTURAL CHANGES IN CHROMOSOMES

For better understanding of the abnormalities of chromosome structure, let us consider two important features of chromosome behaviour :

1- During prophase I of meiosis, homologous regions of chromosomes show a great affinity for pairing and they often go through considerable contortions in order to pair.
2- structural changes usually involve chromosome breakage; the broken chromosome ends are highly "reactive" or "sticky", showing strong tendency to join with broken ends.

Types of Structural Changes in Chromosome

Structural changes in chromosome may be of the following types:

1. **deficiency** or **deletion** which involves loss of a broken part of a chromosome.

2. **duplication** involves addition of a part of chromosome (i.e., broken segment becomes attached to a homolog which, thus, bears one block of genes in duplicate).

3. **inversion** in which broken segment reattached to original chromosome in reverse order, and

4. **translocation** in which the broken segment becomes attached to a nonhomologous chromosome resulting in new linkage relations.

Variation in Chromosome Morphology

Various changes in chromosome structure often produce variation in chromosome morphology such as isochromosomes, ring chromosomes and Robertsonian translocation.

1. Isochromosomes.

An isochromosome is a chromosome in which both arms are identical. It is thought to arise when a centromere divides in the wrong plane, yielding two daughter chromosomes, each of which carries the information of one arm only but present twice. For example,

telocentric X chromosome ofDrosophila may be changed into an “attached-X” which is formed due to misdivision of the centromere.

2. **Ring chromosomes.**

Chromosomes are not always rod-shaped. Occasionally ring chromosomes are encountered in higher organisms. Sometimes breaks occur at each end of the chromosome and broken ends are joined to form a ring chromosome. Crossing over between ring chromosomes can lead to bizarre anaphase.

3. **Robertsonian translocation.**

Thus, Robertsonian translocation is an eucentric reciprocal translocation where the break in one chromosome is near the front of the centromere and the break in the other chromosomes is immediately behind its centromere. The resultant smaller chromosome consists of largely inert heterochromatic material near the centromere; it normally contains no essential genes and tends to become lost. Thus, Robertsonian translocation results in a reduction of the chromosome number.