

**Cytogenetics** is branch of biology devoted to the study of chromosomes and their implications in genetics.

**CHROMOSOMAL ABERRATIONS:** Occasionally, spontaneous (without any known causal factor, Natural occurrence) variations in the structure and number of chromosomes have been observed in nature. These variations are called chromosomal aberrations and can be due to either (a) structural changes or (b) numerical changes.

### **Origin of structural aberration**

Chromosomes are structures with definite organization. However, through various means they may be broken and their normal structure disrupted. X-rays, atomic radiations and various chemicals are among the agents that can cause breaks in chromosomes. Breaks also sometimes occur under natural conditions, where in most instances the reason for breakage is not known.

**Breakage-fusion-bridge Cycle:** In the gametophyte and endosperm of corn, ends of chromosomes that have recently been broken behave as though they were “sticky,” as is shown by their tendency to adhere to one another. Extensive studies of broken chromosomes in corn have been made by Barbara Mc Clintock. She found that following reduplication of a broken chromosome the two sister chromatids may adhere at the point of previous breakage. The fused sister chromatids would be unable to separate readily. In effect, they constitute a single chromatid with two centromeres, a dicentric chromatid. As the centromeres move to opposite poles at anaphase, the dicentric chromatid stretches out, forming a chromatin bridge from one pole toward the other. This bridge eventually breaks, but the break does not always occur at the point of previous fusion. Therefore, chromosomes may be formed that show structural aberrations such as duplications or deficiencies if compared with an original type.

Thus, if the original chromosome is     **C B A**    

the type     **C B A A**    

is a duplication type, since region A is represented twice.

The type     **C B**    

is a deficiency, since region A is absent.

When a chromosome bridge breaks, perhaps as a result of tension caused by the movement of two centromeres of the dicentric chromatid two new broken ends are formed. Each of these has the same qualities of adhesiveness that gave rise to the original fusion. This situation permits repetition of more similar events as described above, in cyclic series. Spontaneous production of

chromosome aberrations through breakage -fusion -bridge cycles may occur in this manner for some time. But when a broken chromosome is introduced into the sporophytic generation such cycles cease, as the broken ends heal in the zygote.

**Structural chromosomal aberrations:** Structural chromosomal changes include those chromosomal aberrations which alter the chromosome structure i.e. the number of genes, the sequence or kind of genes present in the chromosome(s) and do not involve a change in chromosome number.

**Types of structural chromosomal aberrations:** These aberrations may be confined to a single chromosome or more than one chromosome. Hence they are of two types.

1. Intra-chromosomal aberrations
2. Inter-chromosomal aberrations

**I. Intra -chromosomal aberrations:** When aberrations remain confined to a single chromosome of a homologous pair, they are called intra -chromosomal aberrations. Includes deletions, deficiency and inversions.

**II. Inter-chromosomal aberrations:** When aberrations occur in non-homologous chromosomes and the resulting fragments are inter-changed by both the nonhomologous chromosomes, they are known as inter-chromosomal aberrations. Ex: Translocations.

**I. Intra-chromosomal aberrations:** They may be of the following types :

**1. Deletions or deficiencies:** Deletion is due to the loss of a part of a chromosome. In a deletion, a chromosome lacks either a terminal or an interstitial segment which may include only a single gene or a part of a gene. Hence it is of two types.

i) Terminal deletion: If a break occurs near the end of chromosome and a small piece of terminal chromosome is lost, it is called terminal deletion.

ii) Interstitial or intercalary deletion: Sometimes two breaks may occur at any two points and the broken ends of the original chromosome get fused or reunited and as a result, an interstitial deletion is formed. The deleted fragment is acentric. Consequently the acentric fragment is immobile and will be lost during cell division.

Both types of deletions can be observed during pachytene stage of meiosis or in the polytene chromosomes. Deficiencies can be artificially induced using radiations such as X-rays or gamma ( $\gamma$ ) rays. However, in majority of the cases, deficiencies are intercalary because, when a terminal part of the chromosome is lost, it can not be repaired unless it unites with another broken end.

### **Genetic significance / effects of deletions:**

1. Organisms with homozygous deletion do not survive to an adult stage because a complete set of genes is lacking (lethal effect).
2. Small deficiencies, if present in heterozygous condition (deficiency heterozygote) can be tolerated by the organism. In such individuals during pachytene stage of meiosis the unpaired segment of the normal chromosome of an intercalary deletion heterozygote produces a characteristic loop in a bivalent. In case of terminal deletion heterozygotes, the segment towards one end of the normal chromosome remains unpaired. Loops can be observed in salivary gland chromosomes of *Drosophila* or giant chromosomes, which are found in a permanent state of pairing. Therefore even small deficiencies could be detected in these chromosomes.
3. Deficiencies have an effect on the inheritance also. In presence of deficiency a recessive allele will behave like a dominant allele. When an organism heterozygous for a pair of alleles Aa loses a portion of the chromosome bearing the dominant allele 'A', the recessive allele being in the hemizygous condition will be expressed phenotypically. This phenomenon is known as pseudo-dominance.
4. This principle of pseudo -dominance has been utilized for location of genes on specific chromosomes in *Drosophila*, maize and other organisms. Thus, deletions are important cytological tools for mapping genes.
5. Deletions play an important role in species formation and creating variability through chromosomal mutations.

In *Drosophila*, deletions were recorded on X-chromosome in the regions containing genes-w (for white eye), fa (for facet eye) and v (for vermilion eye colour).

**2. Duplications or Repeats:** Duplication occurs when a segment of chromosome is represented two or more times in a chromosome of a homologous pair. The extra segment may be a free fragment with a centromere or a chromosomal segment of the normal complement. As a result, in one chromosome of the homologous pair, there will be deletion, while in other there will be a duplication. Duplication was first reported in *Drosophila* by C.B. Bridges in 1919.

Duplications are of four types:

1. **Tandem:** The extra chromosome segment may be located immediately after the normal segment in precisely the same gene sequence.
2. **Reverse tandem:** The gene sequence in the extra segment of tandem duplication is in the reverse order i.e. is inverted. (eg . cb in place of bc)
3. **Displaced:** The extra segment may be located in the same chromosome but away from the normal segment and with same sequence.
4. **Reverse displaced:** The gene sequence in the extra segment of a displaced duplication is in the reverse order i.e. is inverted (eg. ed in place of de)

If duplication is present in only one of the two homologous chromosomes, at pachytene stage of meiosis, cytological observations characteristic of deficiency will be obtained in duplication also.

**Genetic significance / effects of duplications:**

1. Duplications are not as harmful as deletions.
2. Some duplications are useful in the evolution of new genetic material such as new alleles due to unequal crossing over and thereby genetic diversity.
3. Large duplications can reduce fertility.
4. The phenotype may be altered.

One of the classical examples of duplication in *Drosophila* is that of bar eye. Bar eye is a character where the eyes are narrower as compared to normal eye shape. This phenotypic character is due to the duplication for a part of chromosome. By the study of giant salivary gland chromosomes of *Drosophila melanogaster*, it could be demonstrated that 'Bar' character was due to duplication in the region 16A of x chromosome. Barred individuals (16A 16A) gave rise to ultra bar (16A 16A 16A) and normal wild type (16A) due to unequal crossing over.

