

## 10. CHROMOSOMAL ABERRATIONS

Occasionally, spontaneous (without any known causal factor) 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. An initially single deviation from the normal can give rise to a whole series of unusual cytological events.

### 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 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

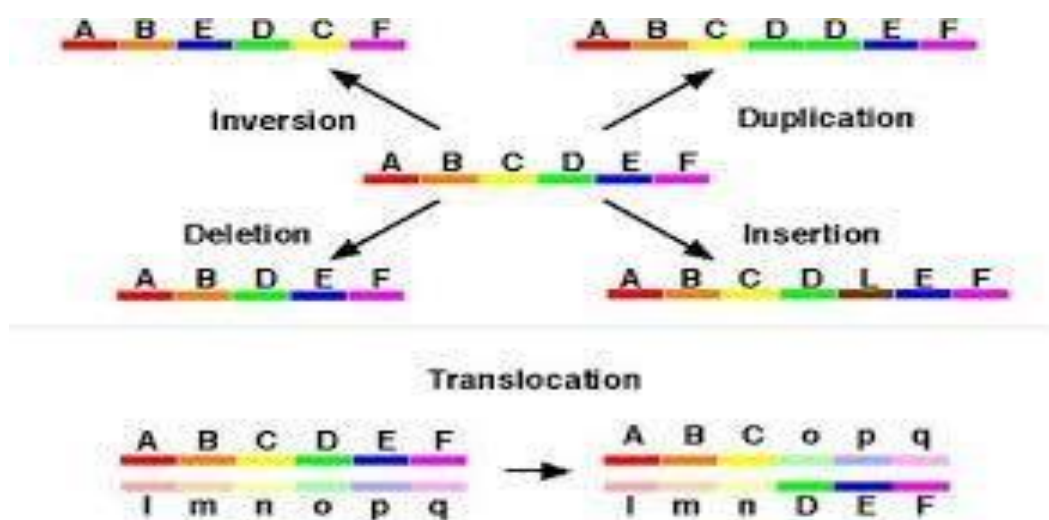
is a deficiency, since region A is absent.

When a chromosome bridge breaks, perhaps as the 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.

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



**Fig.:** Different types of structural chromosomal aberrations

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

**A. 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.

- a) **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.
- b) **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. If the chromosome has a centromere, it will persist. Otherwise it 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. However, in majority of the cases, deficiencies are intercalary because, when a terminal part of the chromosome is lost, it cannot 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).

**B. 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.

- a) **Tandem:** The extra chromosome segment may be located immediately after the normal segment in precisely the same orientation (i.e. having the same gene sequence)
- b) **Reverse tandem:** The gene sequence in the extra segment of a tandem duplication is in the reverse order i.e. is inverted. (eg .cb in place of bc)
- c) **Displaced:** The extra segment may be located in the same chromosome but away from the normal segment.
- d) **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.
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 ultrabar (16A 16A 16A) and normal wild type (16A) due to unequal crossing over.

**C. Inversions:** When a segment of a chromosome is oriented in reverse direction, such a segment is said to be inverted and the phenomenon is termed as inversion. Gene sequence in an inverted segment is exactly the opposite of that in its normal homologous pair. Inversions would involve two breaks followed by reunion of interstitial segment in a reverse order i.e. the segment rotates by  $180^\circ$ . (Let us imagine that a chromosome 1-2-3-4-5-6-7-8 gives rise to another chromosome having the order 1-2-6-5-4-3-7-8, the segment 3-4-5-6 has rotated here at  $180^\circ$  giving an inverted order of genes 6-5-4-3)

Inversions can be of two types depending upon whether centromere is involved or not in inversions.

(a) **Paracentric inversions:** Paracentric inversions are those inversions, where the inverted segment does not include centromere.

(b) **Pericentric inversions:** The inverted segment includes the centromere in pericentric inversions. (pericentric means surrounding the centromere or on the periphery of centromere).

**Cytology of inversions:** When both the members of a homologous pair have similar type of inversion, it is called inversion homozygote. Meiosis is normal in inversion homozygotes. When only one chromosome of a homologous pair has inversion it is called inversion heterozygote. Due to an inverted segment in one of the two homologous chromosomes, the normal kind of pairing is not possible in an inversion heterozygote. In order to enable pairing of homologous segments, a loop is formed by each of the two chromosomes. This kind of configuration will be observed in paracentric as well as pericentric inversions. However, the products of crossing over and the subsequent stages of meiosis will differ in these two kinds of inversions.

1. **Product of paracentric inversion:** A single crossing over or an odd number of crossovers in an inverted region will result into the formation of a dicentric chromosome (having two centromeres) and an acentric chromatid (without centromere) when two chromatids are involved in the crossing over. These

dicentric chromatid and acentric chromatid will be observed at anaphase I in the form of a bridge and a fragment.

2. **Product of pericentric inversion:** In a pericentric inversion, although at pachytene, the configuration observed is similar to that described above for paracentric inversion, the products of crossing over and the configurations of subsequent stages of meiosis differ. In this case, two of the four chromatids resulting after meiosis will have deficiencies and duplications. Unlike paracentric inversion, no dicentric bridge or acentric fragment will be observed at anaphase I. However, in pericentric inversion, if the two breaks are not situated equidistant from the centromere, this will result in the change in shape of the chromosome. For instance, a metacentric chromosome may become sub-metacentric and vice-versa.

### **Genetic consequences of inversions**

1. Simple inversions do not have primary effects other than change in chromosome shape.
2. A peculiar kind of position effect occurs due to suppression of the transcription of gene.
3. Normal linear pairing of homologous chromosomes is not possible.
4. Heterozygosity will be maintained from generation to generation.
5. Among the four chromatids resulting after crossing over, two chromatids would have deficiencies and duplications. The gametes having these chromosomes will not function normally and lead to high sterility. Therefore, there should be considerable gametic or zygotic lethality. In plants there will be sufficient pollen sterility. However, since the products of single crossing over will not function and the only crossover products recovered will be double cross overs, the observed frequency of recombination between any two genes of interest will be considerably reduced. Due to this reason, inversions (especially paracentric inversions) are often called crossover suppressors. This reduction in crossing over is not the actual reduction in cytological crossing over, but is the result of lack of recovery of the products of single cross overs. This property of inversion has been utilized in the production of CIB stock used by Muller for the detection of sex-linked lethal mutations in *Drosophila melanogaster*.

**D. Shifts:** Shifts are altered forms of inversions. In this type of aberrations, the genes are in the right order but a segment is shifted either to the right or to the left.

**E. Isochromosome:** Isochromosome is the one in which both the arms are identical in both gene content and morphology. It arises when the centromere divides in wrong plane yielding two daughter chromosomes each of which carries the information of one arm only but present twice.

**II. Interchromosomal aberrations:** These are of the following types.

**A. Translocation:** Integration of a chromosome segment into a non-homologous chromosome is called translocation. It involves shifting of one part of chromosome to another non-homologous chromosome. The phenomenon of translocation was discovered by C.B. Bridges in 1923 in *Drosophila* and by Hugo de Vries in *Oenothera lamarckiana*.

Translocation is of two types

- 1. Simple translocation:** In simple translocation, the terminal segment of chromosomes is integrated at one end of a non-homologous chromosome. However, they are rare.
- 2. Reciprocal translocation:** If two non-homologous chromosome exchange segments which need not be of same size, it results in reciprocal translocation. Production of reciprocal translocation requires a single break in each of the two non-homologous chromosomes followed by reunion of the chromosome segments thus produced. An individual having reciprocal translocation may be either a translocation homozygote or a translocation heterozygote. When both the chromosomes from each pair are involved, it produces translocation homozygote and when only one chromosome from each pair of two homologues is involved, it gives rise to translocation heterozygote.

In a translocation homozygote, the two homologues of each of the two translocated chromosomes are identical in their gene content. As a result, they form normal bivalent and there is no detectable cytogenetic aberration (peculiarity). In a translocation heterozygote, one member from each of two homologous pairs is involved in reciprocal translocation, while the remaining chromosomes of the two concerned pairs are normal. Due to the pairing between homologous segments of chromosomes, a cross-shaped (+) figure involving four chromosomes will be observed at pachytene. These four chromosomes at metaphase I will form a

quadrivalent, which may exhibit any one of the following three orientations. (1) Alternate (2). Adjacent I (3) Adjacent II

1. **Alternate:** In this case, the centromeres lying alternate to each other in the cross shaped figure move to the same pole. In other words, the adjacent chromosomes will orient towards opposite poles. As a result, the two normal chromosomes move to one pole, while the two translocated chromosomes move to the opposite pole. Such a segregation can take place only when the cross shaped figure of four chromosomes is twisted to form a figure of '8'.
2. **Adjacent-I:** In adjacent I orientation, adjacent chromosomes having nonhomologous centromeres will orient towards the same pole. In other words, the chromosomes having homologous centromeres will orient towards the opposite poles. Thus, a ring of four chromosomes will be observed.
3. **Adjacent – II:** In adjacent II orientation, the adjacent chromosomes having homologous centromeres will orient towards the same pole. In this case also a ring of four chromosomes will be observed. In both Adjacent-I and adjacent-II disjunctions, one normal and one translocated chromosome move to the opposite poles.

Adjacent-I and adjacent-II disjunctions will form gametes which would carry duplications or deficiencies and as a result would be non - functional or sterile. Therefore, in a plant having translocation in heterozygous condition, there will be considerable pollen sterility.

#### **Genetic significance of translocation heterozygotes:**

1. They produce semi sterile plants with low seed set.
2. Some genes which earlier assorted independently tend to exhibit linkage relationship.
3. The phenotypic expression of a gene may be modified when it is translocated to a new position in the genome.
4. The presence of translocation heterozygosity can be detected by the occurrence of semi-sterility and low seed set. This can then be confirmed at meiosis by quadrivalent formation. Functional gametes will be formed only from alternate disjunction, which will give rise to three kinds of progeny *viz.*, normal, translocation heterozygotes and translocation homozygotes in 1:2:1 ratio.



### **Role of structural chromosomal aberrations in plant breeding**

1. They are useful in the identification of chromosomes
2. Utilization of vigour as in case of duplication.
3. Useful in genome analysis.
4. Useful for the transfer of desirable characters through translocation.
5. They have evolutionary significance.