

**Course- B.Sc. (Botany Honours), Part -3**

**Paper-VI (Group-A), Cytogenetics**

**Topic- Chromosomal Aberrations**

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# Chromosomal Aberrations

Alteration in the structure of individual chromosome or chromosomal aberration may occur spontaneously or by induction. Such changes may result in quantitative alteration of genes or rearrangement of genes. The breakage and reunion of chromatid segments result in a number of abnormalities in the chromosome structure. Thus origin of structural changes is caused by breaks in the chromosome.

Any broken end may unite with any other broken end, thus potentially resulting in new linkage arrangements. Depending upon the number of breaks, their locations, and the pattern in which broken ends join together, a wide variety of structural changes are possible (Fig. 12.1). The first cytological demonstration of chromosomal rearrangement in plants was made in maize by B. McClintock.

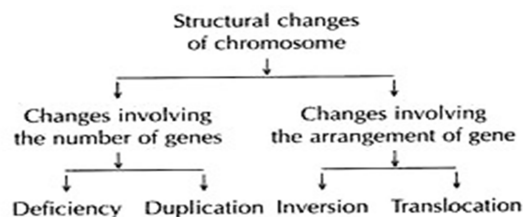


Fig. 12.1: Different types of structural changes of chromosome

## Types of Chromosomal Aberration:

Four different kinds of structural changes of chromosome have been demonstrated

- (i) Deficiency (parts of chromosome lost or deleted),
- (ii) Duplication (parts of chromosome added or duplicated),
- (iii) Inversion (sections of chromosome detached and reunited in reverse order), and
- (iv) Translocation (parts of chromosome detached and joined to non-homologous chromosome).

Of the various chromosomal aberrations, inversions and translocations only represent changes in position of chromosome segments of different sizes, the total chromosome mass remaining unchanged. All segments are present in the original dosage, but distributed in a new way, i.e. qualitative alterations.

In cases of deletions or deficiencies and duplications, quantitative alterations occur in the chromosome complement, with certain chromosome segments being lost or doubled.

Structural homozygotes are those in which alterations such as translocation or duplication occur in both the homologous chromosomes and as such termed as translocation homozygote or duplication homozygote. In cases, where only one chromosome of the pair is structurally altered, the term structural hybrid or hetero-zygote is used.

## Deficiency of Chromosomal Aberration:

Deficiency or deletion represents a loss in chromosomal material and was the first chromosomal aberration indicated by genetic evidence. This evidence, presented by Bridges in 1911 in *Drosophila melanogaster*, showed a deletion of the X-chromosome that included the Bar locus.

### Deficiency or deletion are of two types:

#### (i) Terminal deletion:

A single break near the end of a chromosome would be expected to result in a terminal deficiency;

#### (ii) Intercalary deletion:

If two breaks occur, a section may be deleted and an intercalary deficiency is created.

### Duplication of Chromosomal Aberration:

Duplication represents additions of chromosome parts. A chromosome segment is present in more than two copies.

### Types of Duplication of Chromosomal Aberration:

Duplications are of different types on the basis of position of duplicated segment (Fig. 12.11):

- (i) Tandem duplication – adjacent region;
- (ii) Displaced homo-brachial duplication – at a displaced position of the same arm;
- (iii) Displaced heterobrachial duplication – on the different arm of the same chromosome;

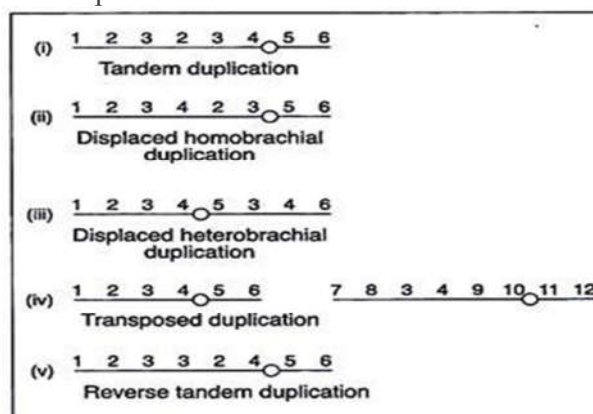


Fig. 12.11: Different kinds of duplications

- (iv) Transposed duplication – on a different chromosome;
- (v) Reverse tandem duplication – duplicated segment found as a reverse repeat at adjacent region.

### Inversion of Chromosomal Aberration:

Inversion represents reverse gene order in the chromosome.

### Types of Inversions:

Inversions can be of two types:

- (i) Paracentric inversion and
- (ii) Pericentric inversion.

Paracentric inversions are those inversions where inverted segments do not include centromeres. On the other hand, in a pericentric inversion, inverted segment includes centromere.

### Paracentric Inversion:

In paracentric inversion, a single crossover or an odd number of crossovers in inverted region results in the formation of a dicentric chromosome (having two centromeres) and an acentric chromosome (with no centromere). Of the remaining two chromatids, one remains normal and the other carries the inversion.

The dicentric chromatid and the acentric chromatid are observed at anaphase I in the form of a bridge and a fragment (Fig. 12.14). Crossing over within and outside inversion lead to various kinds of deficiencies and duplications.

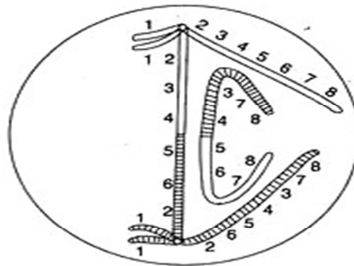


Fig. 12.14: Dicentric bridge and an acentric fragment at anaphase I

### Pericentric Inversion:

In pericentric inversion, the pachytene configuration observed is similar to that of paracentric inversion. But the products of crossing over and configurations at subsequent stages of meiosis differ. Two of the four chromatids will have deficiencies and duplications. No dicentric bridge or acentric fragment are formed (Fig. 12.15).

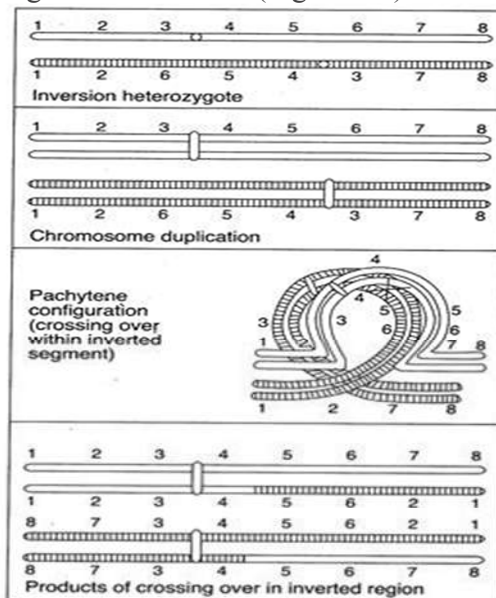


Fig. 12.15: Chromosome pairing and products of crossing over in a pericentric inversion heterozygote (after P. K. Gupta)

As the two chromatids resulting from crossing over have deficiencies and duplications, the gametes having these chromosomes do not function and lead to considerable gametic or zygotic lethality. The plants show pollen sterility. The only crossovers which can be recovered are double crossovers, and the observed frequency of recombination between any two genes is considerably reduced.

Thus inversions are called crossover suppressors. This property of inversion has been utilized in the production of CIB stock, used by Muller for detection of sex linked lethal mutations. Three different kinds of non-crossover progenies (1 : 2 : 1) are obtained by selfing of an inversion heterozygote (Fig. 12.16).

Gametes			
	♀	♂	
		Normal gamete	Gamete with inversion
Normal gamete		Normal	Inversion heterozygote
Gamete with inversion		Inversion heterozygote	Inversion homozygote

Fig. 12.16: Progenies obtained by selfing of an inversion heterozygote

### Translocation of Chromosomal Aberration:

Sometimes a part of a chromosome becomes detached and joins to a part of a non-homologous chromosome, thus producing translocation. Translocations have been described in a number of plants and are important factors in the evolution of certain plant groups such as *Datura* and *Oenothera*.

### Types of Translocation:

Three types of translocations are observed:

#### (i) Simple translocation:

The broken part gets attached to one end of non-homologous chromosome.

#### (ii) Shift translocation:

Broken part gets inserted interstitially in a non-homologous chromosome.

#### (iii) Reciprocal translocation:

When parts of chromosomes belonging to members of two different pairs become exchanged (Fig. 12.17).

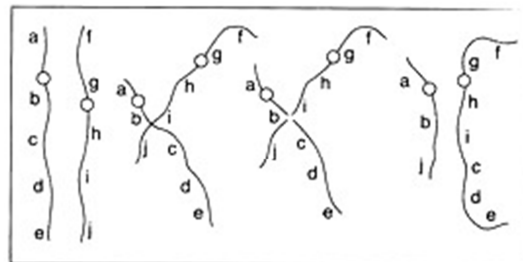


Fig. 12.17: The origin of a translocation

### Other Forms of Chromosomal Aberrations:

#### Centric Fusion and Fission:

Centric fusion is a process that leads to a decrease in chromosome number. Two acrocentric chromosomes join together to produce a metacentric chromosome. This phenomenon is also called Robertsonian translocation.

Dissociation or fission is process that leads to an increase in chromosome number. In dissociation, a metacentric (commonly large) and a small supernumerary metacentric fragment become trans-located, so that two acrocentric or sub-metacentric chromosomes are produced.

Direct fission of centromere of metacentric chromosome leads to two telocentric chromosomes (misdivision). Fusion and fission are the main mechanisms by which the chromosome number can be decreased and increased during evolution of the majority of animals and in some groups of plants (Fig. 12.23).

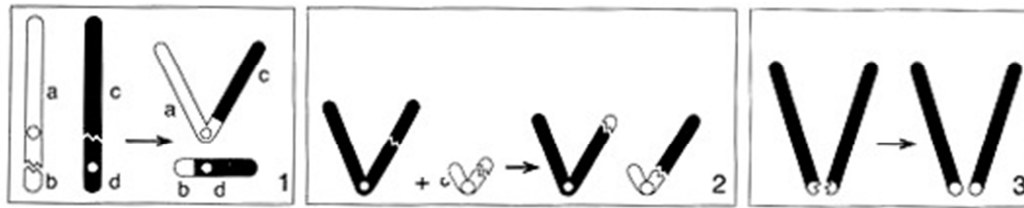


Fig. 12.23: (1) Fusion: The origin of a new V-shaped (metacentric) chromosome by centric fusion of two nonhomologous acrocentric chromosomes. Segment bd is lost. (2) Dissociation: A metacentric and a small supernumerary chromosomal fragment undergo a translocation, which results in two chromosomes (acrocentrics or metacentrics). (3) Fission: Direct division of centromere of a metacentric chromosome to two telocentric chromosomes

### Iso-chromosomes:

A new type of chromosome may arise from a break (i.e., a misdivision) at the centromere. As shown in Fig. 12.24, the two resultant telocentric chromosomes may open up to produce chromosomes with two identical arms (i.e., iso-chromosomes). This type of chromosome is produced in irradiated material. At meiosis they may pair with themselves or with a normal homologue.

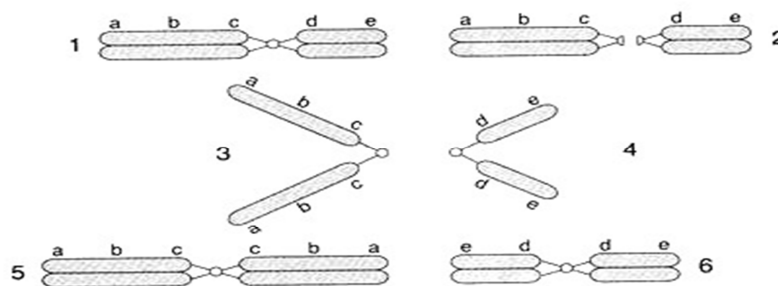


Fig. 12.24: Formation of isochromosomes: (1) Original chromosome; (2) Misdivision of the centromere at the beginning of mitotic anaphase; (3) and (4) The chromatids unfold into two isochromosomes; (5) and (6) In the next division, two complete isochromosomes are present. Note that in each isochromosome, the arms exhibit same genetic constitution

### Sister Chromatid Exchange:

A sister chromatid exchange is an interchange of DNA between sister chromatids in a chromosome, presumably involving DNA breakage followed by fusion. Sister chromatid exchanges are difficult to find using common cytological methods because the chromatids are morphologically identical.

Such chromatid exchanges were first described in studies in which  $^3\text{H}$ -thymidine was added during a replicating cycle which was followed by another cycle in a non-radioactive medium. Analysis of this phenomenon has been greatly facilitated by the use of bromodeoxyuridine (BrdU), a thymidine analogue that can be incorporated into the DNA of replicating cells instead of the original base.

If BrdU is followed by a fluorescent dye (Hoechst 33528), the fluorescence of the segments that contain BrdU is greatly diminished in comparison with those of the original base. Furthermore, there is also a similarly decreased staining with the Giemsa stain.

The use of this technique, however, has been unable to discover whether the chromatid exchange could occur spontaneously or whether it is induced by the BrdU.

It has, though, been of great help in differentiating the various inherited diseases characterized by chromosome fragility, which have an increased frequency of sister chromatid exchanges and a tendency to have associated neoplasia. Some of the diseases (e.g. Bloom's

syndrome, Fanconi's anemia, and ataxia-telangiectasia) are presumably related to defects in DNA repair.

Sister chromatid exchange has also been important in studying the effect of mutagens on the chromosomes. Various mutagenic drugs that are alkylating agents, such as mitomycin C and nitrogen mustard, produce a great number of breaks and chromatid exchanges (Fig. 12.25). The intimate association of sister chromatid exchange with mutagenesis and carcinogenesis may have important medical implications.



Fig. 12.25: Cultured cell exposed to the mutagenic chemical agent 8-methoxypsoralen and to bromodeoxyuridine. The regions stained only lightly with Giemsa and those in which the DNA was synthesized in the presence of BrdU. Observe the numerous sister chromatid exchanges in each chromosome (after S. Latt)

### **Effects of Chromosomal Aberration:**

In most cases, homozygosity for deficiencies or deletions has a deleterious effect and leads to death. Duplications may have more desirable effects than the loss of chromosome substances. Even in this category, there is a disturbance of chromosome balance and in instances of large duplications, a reduction in fertility as well as in vigour may occur.

Translocation in *Oenothera lamarckiana* produces 50% non-viable seeds. The viable seeds are all translocation heterozygotes (balanced lethal system). In *Rhoeo discolor*, the only translocation heterozygotes are survivors. In *Clarkia*, *Paeonia*, translocation and normal homozygotes are also common.

Sometimes in *Oenothera*, *Rhoeo*, chromosomes disjoin in an irregular manner, new translocations are produced and crossing over between different complexes may take place. All these changes produce recognisable phenotypic effects.

The occurrence of inversions is less recorded than translocations. In flowering plants with vegetative reproduction, for instance, in *Tulipa*, heterozygosity for inversions has, however, turned out to be frequent and in *Paris quadrifolia*, every plant seems to be heterozygous for one or several inversions (Muntzing).

That, inversions are common in plants with vegetative reproduction is due to the fact that structural alterations arise and accumulate in them without particular disadvantages. Reproduction is not affected on account of structural aberrations. Since these plants in question reproduce exclusively or predominantly in vegetative way, the aberrations affecting sexuality and seed setting are of no prime importance.

### **Detection of Chromosomal Aberrations:**

The alterations of chromosome structure can however be detected through comparative analysis of karyotypes. The gross chromosomal changes and their location can conveniently be studied through clarification of chromosomal details and their comparison with unaltered genotypes.

The study of meiosis too provides with a powerful method of detection, provided the changes are adequate to bring out the detectable changes in meiotic behaviour.



The study of detection includes formation of loops for the deficiency, inversion bridges for inverted segments as well as ring formation for structural heterozygotes. The formation of multivalent also clearly indicates the duplication of chromosomes. As such, meiotic analysis can provide clear indication of the changes the chromosomes have undergone affecting their structure.

However, gradually a number of modified methods have come up through which finer segments of chromosome can be microscopically differentiated. These methods permit identification of minute chromosome segments which otherwise become difficult to resolve through karyotype or pachytene analysis or study of meiotic details.

The two methods which are now widely applied for detection of chromosomal and genomic alterations are (1) chromosome banding and (2) In situ hybridization (ISH).