30

Chromosomal Mutations or Chromosomal Aberrations

The chromosomal mutations are the visible changes in the structure of chromosomes, involving changes either in the total number of genes or gene loci in a chromosome or their rearrangement. These are also known as **chromosomal rearrangements** or **chromosomal aberrations.** These arise from breaks in chromosomes and may be of the following four types –

A. Changes involving the Number of the Gene Loci

1. Deficiency or Deletion
2. Duplication

B. Changes involving the Arrangement of Gene Loci

3. Translocation
4. Inversion

1. Deficiency or Deletion

1.1 Definition – The deficiency is the deletion of a chromosomal segment resulting in the loss of genes. Depending upon the length of the lost segment, the genes lost may vary from a single gene to a block containing several genes.

1.2 Nature – The break in the chromosome may be caused by several agents such as chemicals, drugs and radiations. The break may occur at any time during cell cycle, either in the somatic or germ cells. The breakage usually occurs at random and either in both the chromatids

of a chromosome (**chromosome break**) or only in one chromatid (**chromatid break**).

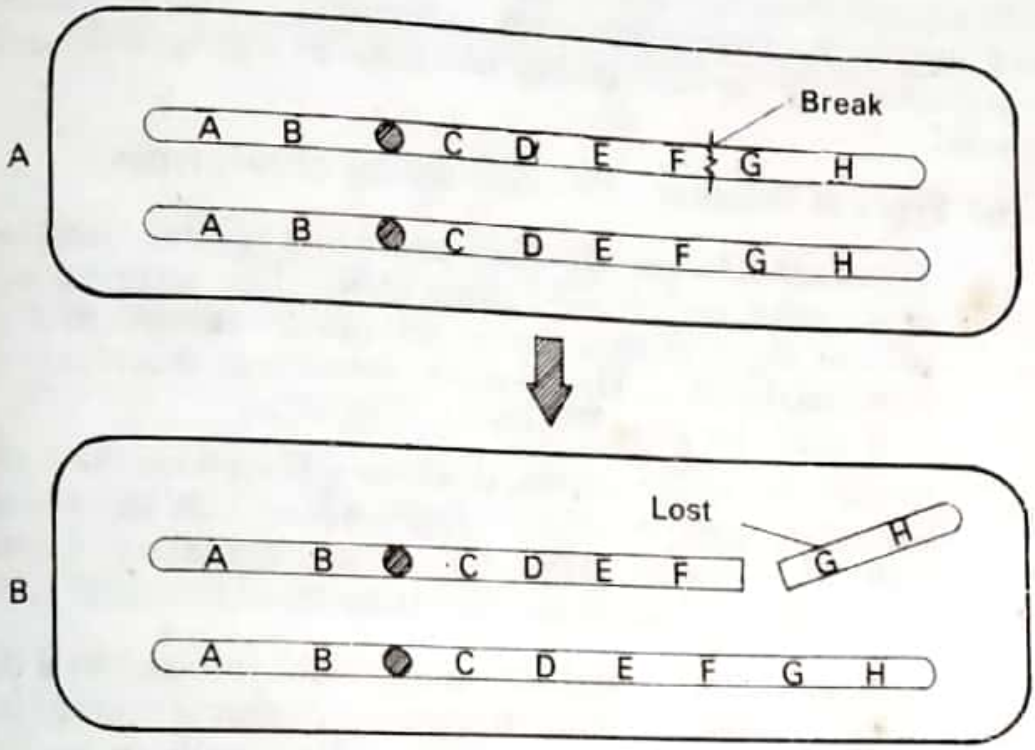
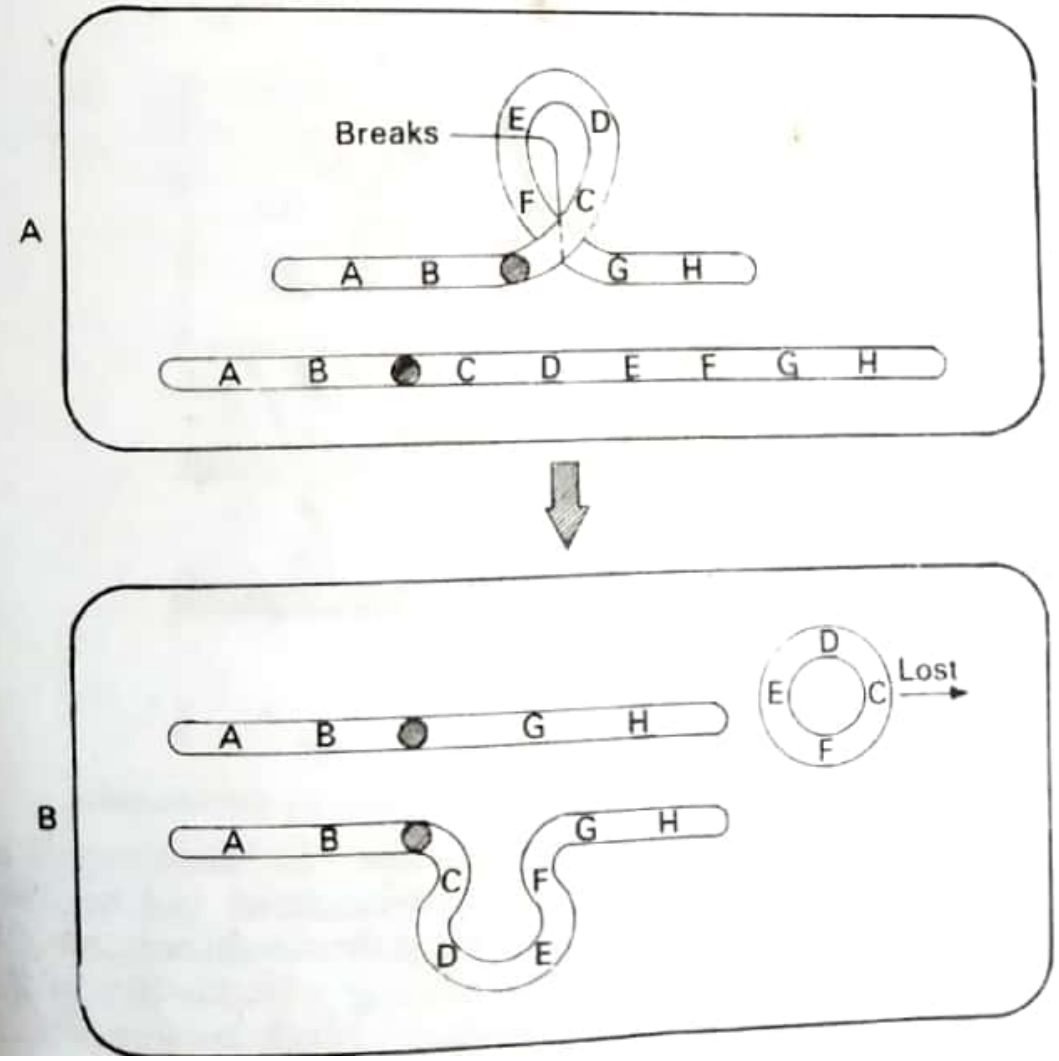


Fig. 30.1. Deletion.



The loss of chromosomal segment occurs when a portion of chromosome gets detached due to certain reasons and the lost segment does not survive, because it lacks the centromere. The portion of the chromosome carrying the centromere functions as a genetically deficient chromosome.

1.3 Types of Deletion – Deletion can be of two types –

(i) **Terminal deletion** – It refers to *the loss of a segment from one or the other end of the chromosome*. The terminal acentric part of the chromosome is unable to survive and causes terminal deletion. The terminal deletion is, therefore, caused by a single break in the chromosome.

(ii) **Intercalary or interstitial deletion** – It *involves the loss of an intercalary segment of the chromosome with the reunion of terminal segments*. Therefore, the intercalary deletion is caused by two breaks and the reunion of terminal parts.

Although, deficiency and deletion are synonymous, but a distinction is often made. The loss of terminal segment of chromosomes is often described as **deficiency**, whereas the loss in intermediary position of chromosome is designated by **deletion.**

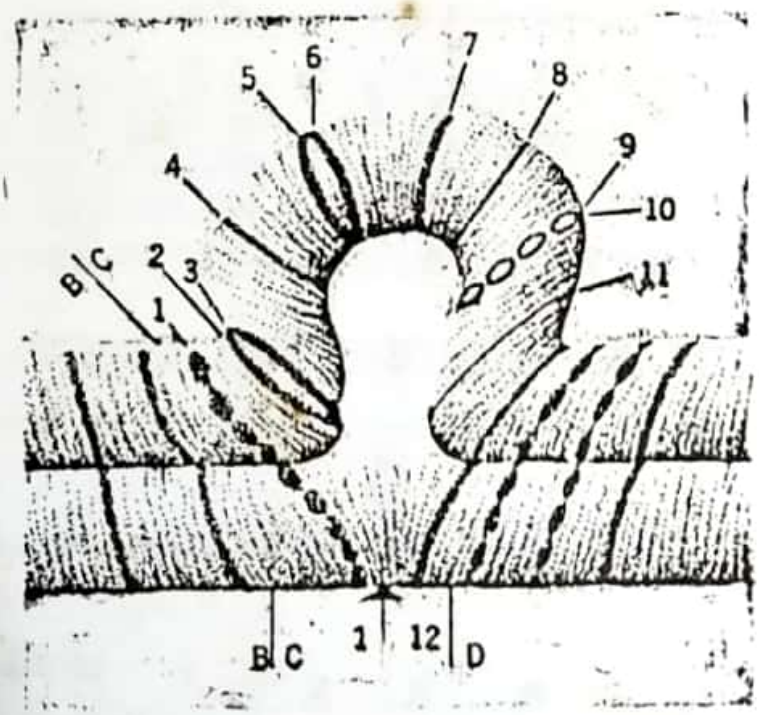


Fig. 30.3. Diagram showing pairing of a deleted chromosome.

1.4 Behaviour of deletion chromosome – In heterozygous condition both terminal as well as intercalary deficiencies can be observed during pachynema stage of meiosis or in the polytene chromosome. When an intercalary part of a chromosome is missing, a buckle-like projection is formed by the part of normal chromosome which corresponds to the missing part of the deletion chromosome.

1.5 Detection of deficiency – The deficiencies can be recognized –

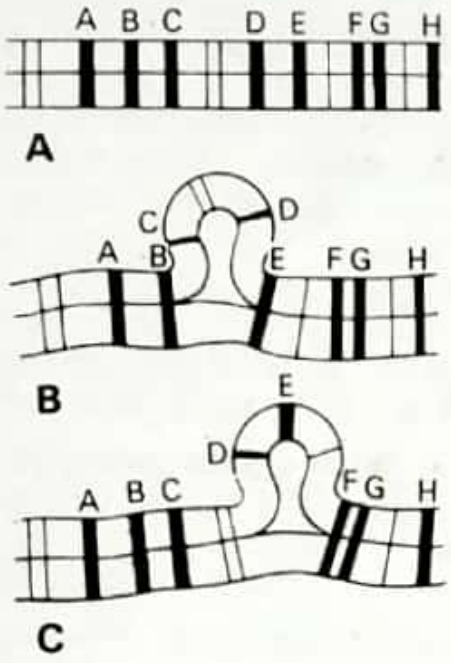


Fig. 30.4. Behaviour of deficiency chromosome during meiosis.
A – Pairing of two normal homologoues chromosomes.
B – Pairing of deletion chromosome showing deficiency of C-D genes with a normal chromosome.
C – Pairing of deletion chromosome with deficiency for D and E genes and a normal chromosome.

(i) **By the phenotypic effects shown in the organisms having deleted chromosomes** – An example of chromosomal deficiency was presented by BRIDGES (1917) and MOHR (1923) in *Drosophila*. A mutant produces notched margin of the wings and is, therefore known as **notch character**. It is in-

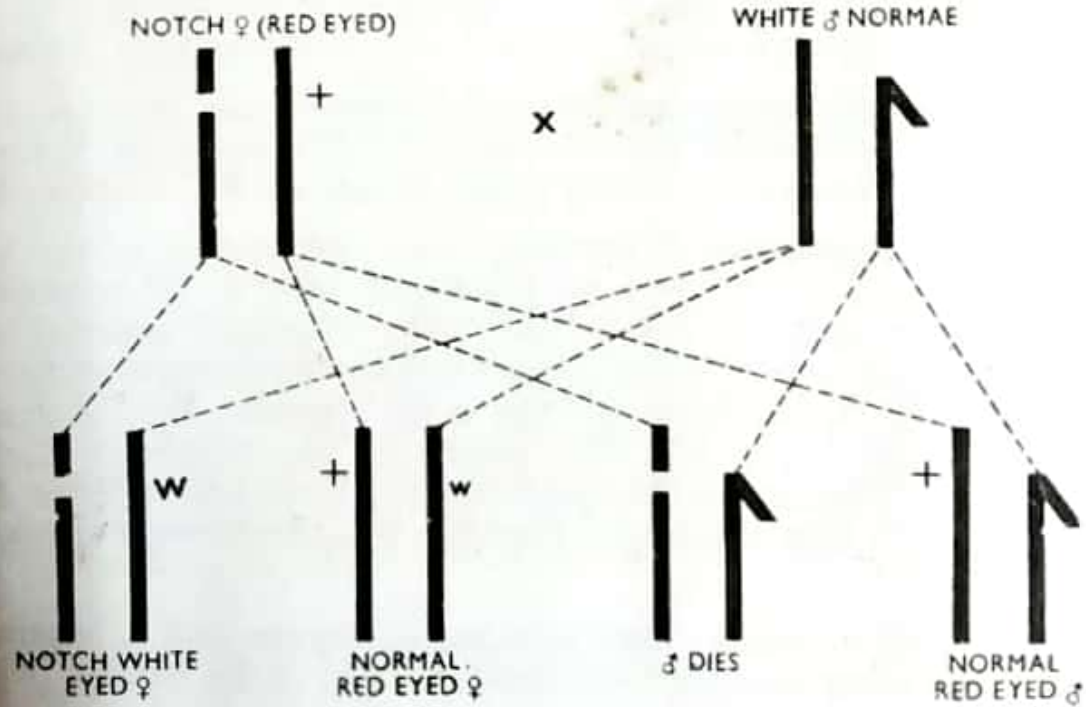


Fig. 30.5. Inheritance of notch deficiency in *Drosophila*.

herited as a sex-linked dominant in female but is lethal in male. Since the males with notch character are not viable, the females help in the inheritance of this character. When such notched females with red eye colour were crossed to white-eyed males having normal wings, the F_1 females with notched wings all exhibited white eye colour indicating as if the white colour is dominant. This pseudodominance is similar to the hemizygous condition found in males where Y-is without alleles somatic genes. It is shown by certain other genes present in the vicinity of white eye locus and is explained to occur by the loss of a piece of the X-chromosome containing these genes. These are **blond, beaded, snipped** and **carved.**

(ii) **By cytological studies of homologously paired chromosomes at meiotic prophase** – The normal chromosome when pairs with the deletion chromosome during prophase of meiosis, its part homologous to deleted segment buckles out.

(iii) **By cytological study of polytene chromosomes** – Here also the chromosome segment corresponding to the deleted part forms a buckle.

1.6 Occurrence of deficiency – Deficiencies have been observed in both animals and plants. In addition to *Drosophila*, a number of deficiency characters are found in man, such as :

(a) **Cri-du-Chat** or cat-cry syndrome caused by deletion in the short arm of chromosome-V.

(b) Deletion caused in the long arm of 22nd chromosome (Philadelphia chromosome).

In plants deficiencies are not easily transmitted to the offspring because pollens with deficiency chromosomes are found to be sterile. However, CREIGHTON and MECLINTOCK have shown that in maize small deficiencies are retained and inherited even in homozygous condition.

1.7 Spontaneous and induced deletions – The deletions usually occur spontaneously but these can be induced by radiation. X rays and ultraviolet radiations etc. readily induce deletion in the chromosomes.

1.8 Significance of deletion – Since deficiencies involve loss of genetic material, these have some deleterious effect on the organism and is dependent upon the amount and quality of genetic material lost. A deletion might be small enough not to cause any detectable morphological change either in the chromosome or in the organism. But the deletions of large size are usually deterimental to the organisms. Deletion heterozygotes having small deletions are most probably viable but abnormal. However, homozygosity for a deletion irrespective of the size is likely to be lethal.

Small deletions behave as recessive and are used in locating the genes and constructing the chromosome maps of the banded polytene chromosomes.