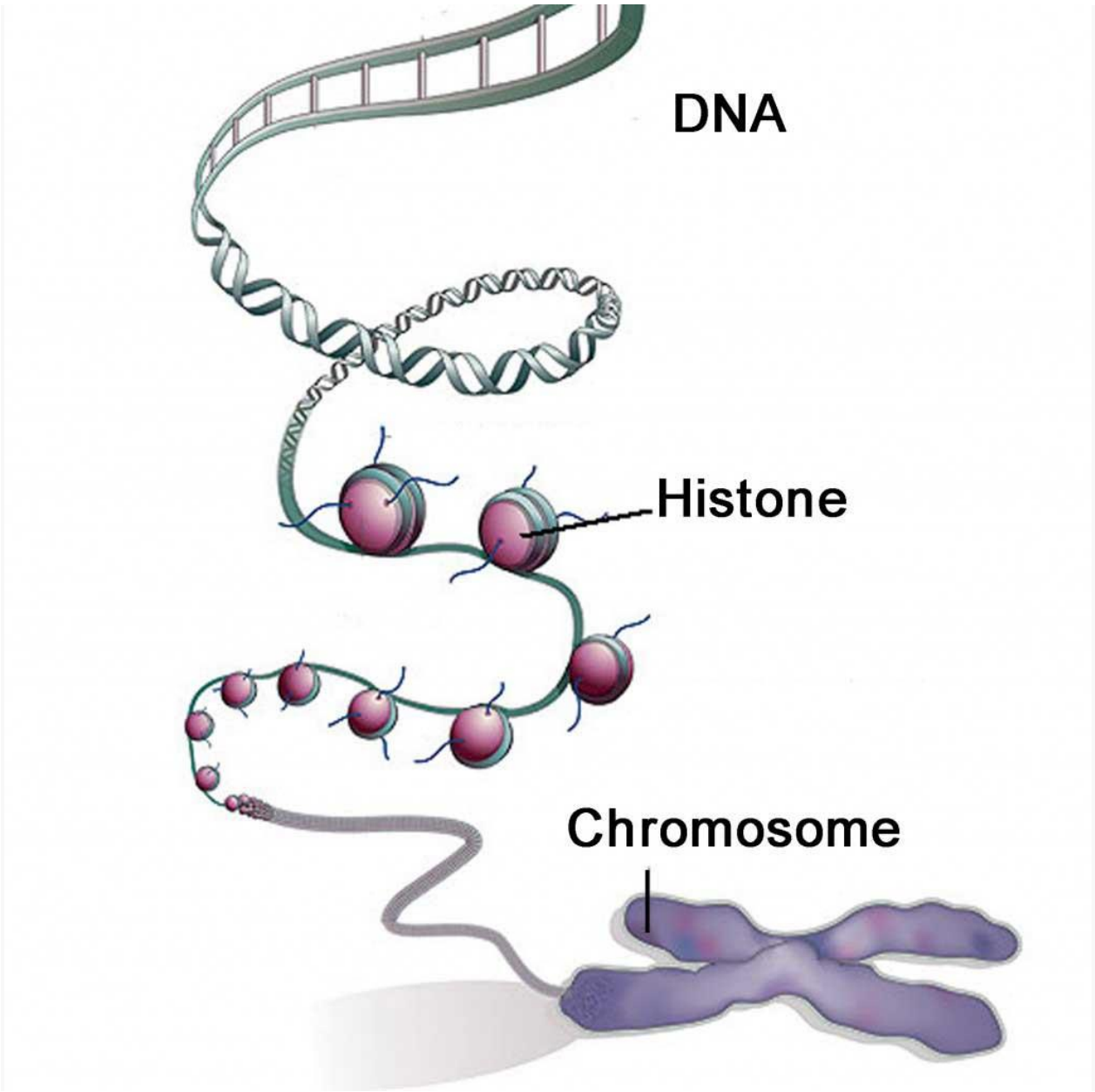
Major Types of Chromosomal Aberrations (Structure and Arrangement)

Some of the major types of Chromosomal aberrations are as follows:

The arrangement and presence of many genes on a single chromosome provides a change in genetic information not only through change in chromosome number but also by a change in chromosome structure.



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The change in chromosome is due to alteration in genetic material through loss, gain or rearrangement of a particular segment. Such changes are called chromosomal aberrations. The modification brings about chromosomal mutations. Chromosomal mutations are very rare in nature but can be created artificially by 'X' rays, atomic radiation and chemicals, etc.

The structural changes in chromosomes are due to breaks in chromosome, or in its cell division subunit, i.e., chromatid. Each break produces 2 ends which may then follow three different paths. (Fig.43.1).

(a) They may reunite, leading to eventual loss of that chromosomal segment which does not contain the centromere.

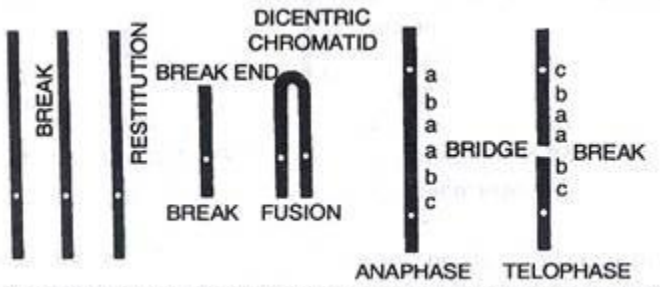

Fig. 43.1. Structural changes in the chromosome during various stages of cell division.

(b) Immediate reunion or reconstitution of the same broken ends may occur, leading to reconstitution of the original structure.

(c) One or both ends of one particular break may join those produced by a different break causing an exchange, or non reconstitutional union.

Mc Clintock (1941) studied in Zea Mays that chromosome breaks and duplication follows. A dicentric chromatid is found. During anaphase spindle fibres are attached to the two centromeres resulting in the formation of bridge from one pole to other. The bridge breaks causing deficiency or duplication.

Chromosomal aberrations are of 4 major types:

(a) Deletion (b) duplication (c) inversion and (d) translocation. (Fig. 43.2).

(A) Deletion or Deficiency:

Deletion or deficiency as the name suggests there is a loss of segment of chromosome. After break the part without centromere is lost. On the other hand the part attached to the centromere acts as deficient chromosome. Bridges (1917) for the first time observed deficiency in the Bar locus of Drosophila.

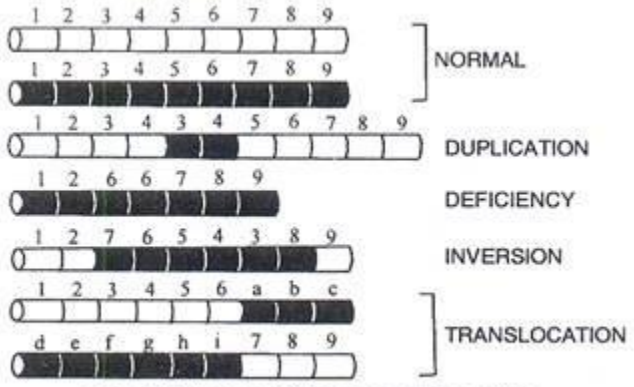

Fig. 43.2. Types of Chromosomal aberrations.

Two types of deletions are found:

Terminal deletion:

A single break near the end of the chromosome. Described in maize but otherwise not common.

Interstitial deletion:

Chromosome breaks and reunites but the part is lost from in between. (Fig. 43.3). Deletions are detected at the time of homologous pairing. If a part of chromosome is missing then the other chromosome also has to omit it in the form of bulging in order to make synapse. e.g., if a chromosome has 1, 2, 3, 4, genes. The part 2 is missing from one chromosome leaving, 1, 3, 4. The other homologous chromosome at the time of synapse bulge out or form loop at position 2.

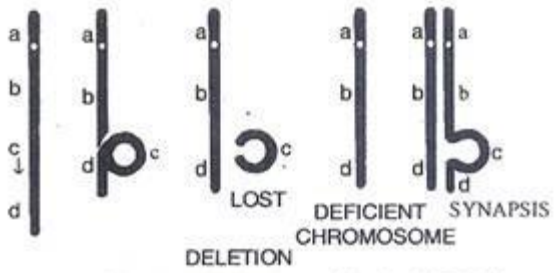

Fig. 43.3. Various steps involved in Deletion

If the missing segment is of physiological importance the individual will not survive. If dominant gene 'A' is missing the recessive allele 'a' may express itself. It is called pseudo dominance.

In human, deletion of chromosome 5 results in cri-du-chat syndrome, children cry like cat, they have small head and are mentally retarded.

Partial deletion of 18th chromosome results in a syndrome with large ears and long fingers.

In corn the deficiency is restricted to pollen sterility. The male haploid gemetophyte shows deficiency while female of it may receive metabolites from maternal tissue supplementing the deficiency. The omitted segment forms buckles. (Fig. 43.4)

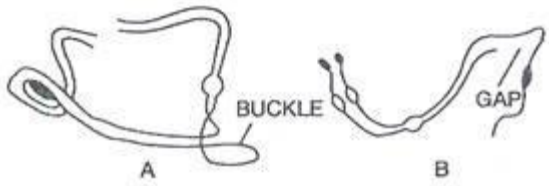

Fig. 43.4. Buckle formation during deletion.

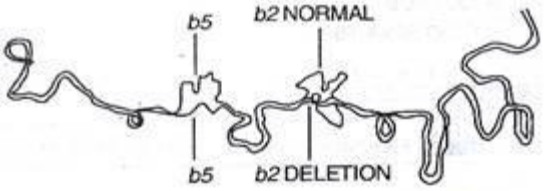

Fig. 43.5. Collapsed loop or brush in single strand DNA due deletion.

Deficiency in E. coli is also noted. The deletion points that the DNA is single stranded and looks like collapsed loop or brush. (Fig. 43.5).

(B) Duplication:

Here a segment of chromosome is repeated twice, i.e., duplicated. Duplication was discovered in Drosophila 'X' chromosome for the first time carrying wild type allele for vermilion (v^+) and has been transposed to an 'X' chromosome carrying the mutant vermilion allele (v), Bridges found that due to the fact that 'X' chromosome was carrying allele v and v^+ both it was wild type instead of vermilion. Equal properties of v and v^+ produced wild type effect. Such 'duplication females' when crossed with nonduplicated vermilion males all female progeny was vermilion and all male progeny, i.e., y was wild type. (Fig.43.6.)

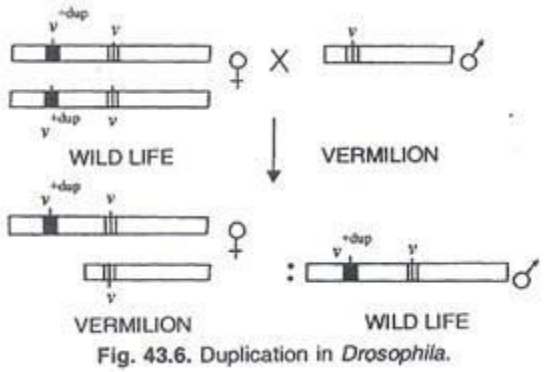

Fig. 43.6. Duplication in *Drosophila*.

Types of duplication:

Duplication is of various types. (Fig. 43.7)

Tandem duplication:

When the duplicating segment is near the centromeres e.g., the sequence on chromosome isabcdefghithe centromere is present between e and f the segment d e is repeated immediately after its normal position.

Reverse tandem:

When the segment is reversed in duplication, e.g., it is d e segment that is duplicated it will be duplicated as d e e d instead of d e d e.

Displaced tandem:

The segment is repeated somewhere away from its original location but on the same arm (homobrachial displacement) or on the other arm (heterobrachial displacement).

Transposition:

When the segment is duplicated on the non homologous chromosome it is called transposition.

Extra chromosomal:

Duplication involves centromere it is called extra chromosomal. In salivary gland chromosome duplications are common either as buckling in the duplication heterozygote or as cross pairing between sections of different chromosomes.

(C) Translocation:

Transfer of a section of one chromosome to non homologous chromosome is known as translocation. When there is exchange of segments on two non homologous chromosomes it is called reciprocal translocation. It also includes exchange of segments between non homologous parts of a pair of chromosomes, e.g., 'X' or 'Y' chromosomes. The segment is neither lost or added it is just exchanged.

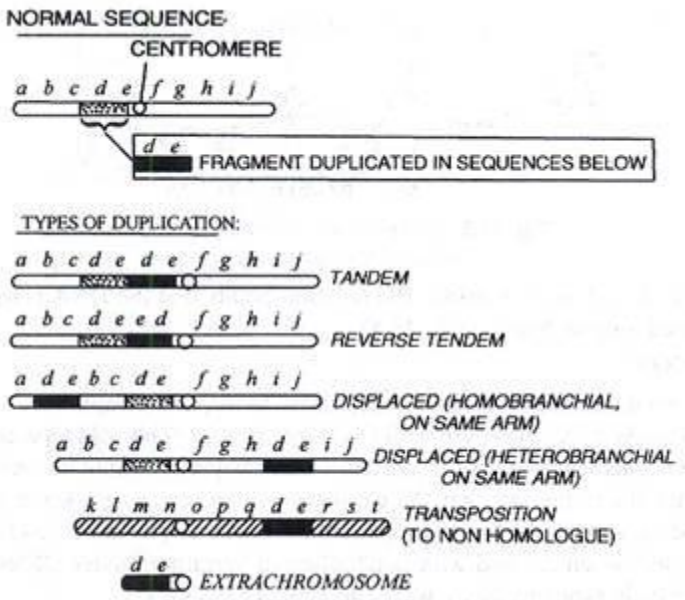

Fig. 43.7. Various types of duplication.

It was first observed in Drosophila by unusual behaviour of a particular 2nd chromosome gene called Pale. It is lethal in homozygous condition. Bridges observed that its lethality can be suppressed by the presence of another gene on the 3rd chromosome

which was also lethal in homozygous condition. Pale effect was caused due to deficiency for a small tip of 2nd chromosome including and plexus or balloon which links to 3rd chromosome gene between ebony or rough.

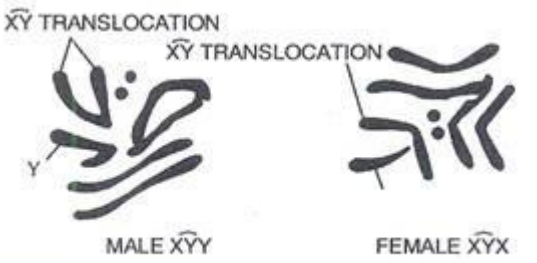

Fig. 43.8. Translocation in *Drosophilla* sex chromosome.

Stern in 1926 observed translocation of some allele (bobbed) on the 'Y' chromosome to the 'X' chromosome. (Fig.43.8)

Types of translocation:

(a) Simple translocation:

A single break in the chromosome and it is transferred onto the end of the other. (Fig. 43.9)

(b) Shift or intercalary translocation:

Common type of translocation involving 3 breaks so that a two break section of one chromosome (e.g., Pale) is inserted within the break produced in a non homologous chromosome. (Fig. 43.9B)

(c) Reciprocal translocation or Interchange:

Frequently observed translocation where single break in two homologous chromosomes produces an exchange of chromosome segment between them. (Fig. 43.9c)

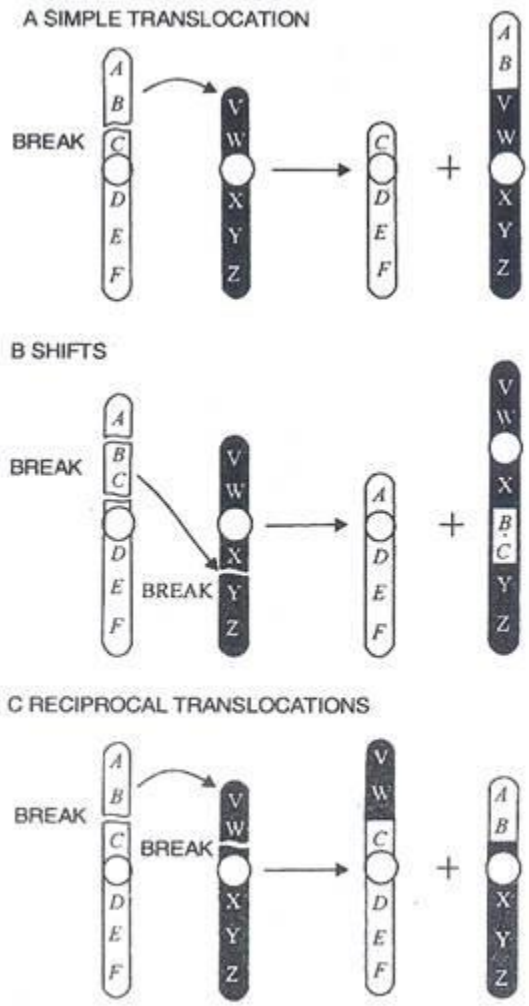

Fig. 43.9. Various types of translocations.

Translocation homozygote forms the same number of homologous pairs as the normal homozygote as long as centromere is not lost.

The result of pairing and meiosis are different in translocation heterozygote bearing two translocated segments and their normal counterpart. (Fig. 43.10). A reciprocal translocation forms a 4 chromosome complex at the pachytene stage. The chiasmata between such chromosomes may form a quadrivalent which can then disjoin in

3 different segregation patterns in the first meiotic division. (Fig. 43.11).

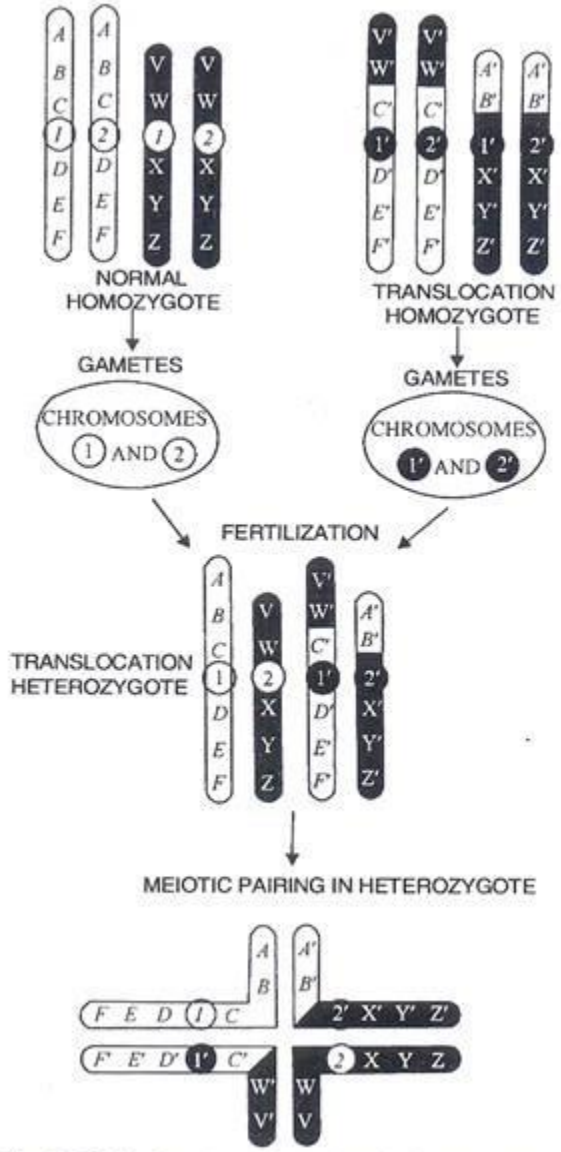

Fig. 43.10. Various types of translocation heterozygote.

(i) Alternate segregation:

Opposite or alternate nonhomologous centromeres go to the same pole in a zigzag fashion, so that the nontranslocated (1, 2) and translocated (1′, 2′) chromosomes are in separate gametes. Gametes

have complete balanced complement of genes without duplication or deficiency (Fig. 43.11).

(ii) Adjacent-1 segregation:
Non homologous adjacent chromosomes go to the same pole but each gamete contains both translocated and non translocated chromosome (1 2′, 1’2) both duplication deficiencies in each gametes are present (Fig. 43.11).

(iii) Adjacent-2 segregation:
Adjacent centromeres again go to the same pole but these are now homologous as well as containing both translocated and non translocated chromosomes (1, 1′; 2, 2′). Duplication and deficiencies produce unbalanced components of gene (Fig. 43.11C).

Adjacent-1 and adjacent-2 segregation produce unbalanced gametes. Fertile gametes of translocation heterozygotes will be mostly restricted to alternate segregation.

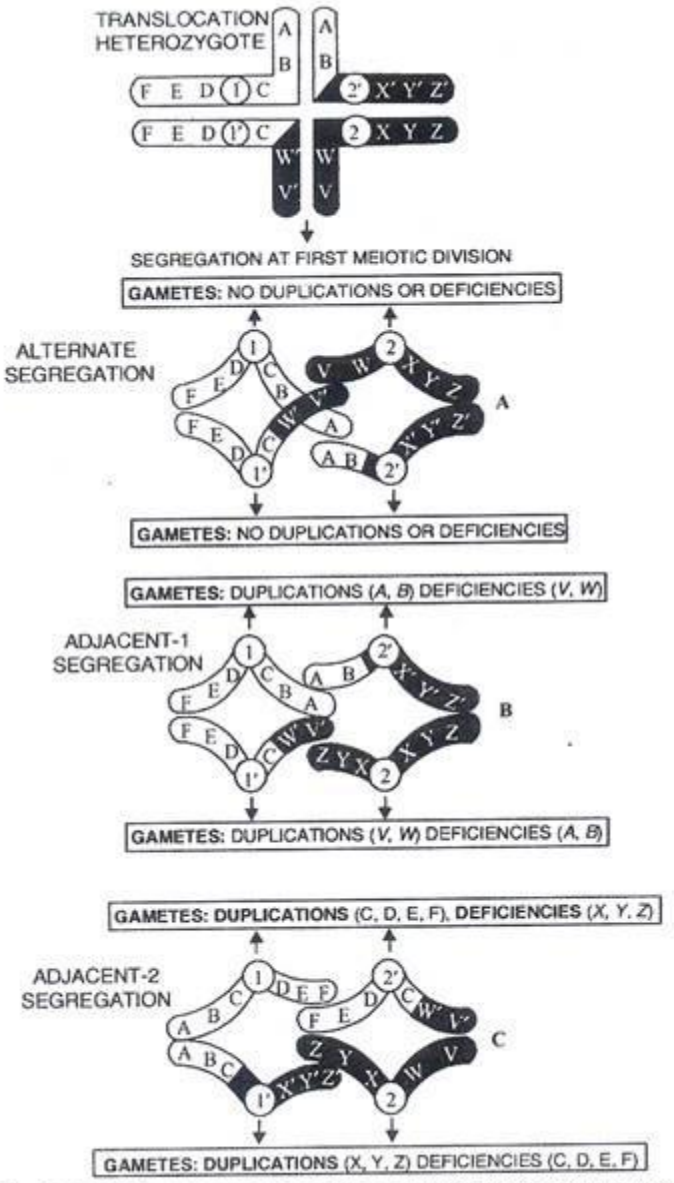

Fig. 43.11. Different segregation pattern in translocation heterozygote.

Synapsis of heterozygous translocation chromosomes showing cross like configuration later on opens out a ring or a figure of eight (Fig. 43.12).

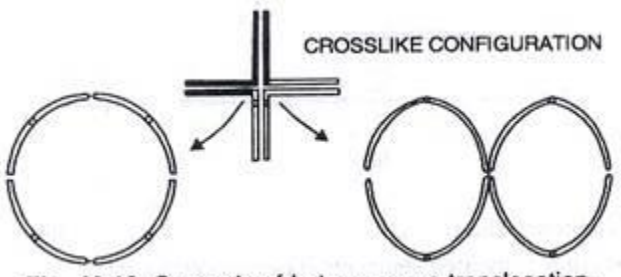

Fig. 43.12. Synapsis of heterozygous translocation.

Consequences of such segregation are that independent assortment between genes and nonhomologous chromosomes will be inhibited. Because of duplications and deficiencies neither of the single mutant phenotypes will appear in the offspring. Translocation heterozygotes have low fertility. If extent of duplication and deficiency is small, unbalanced gametes or zygotes may not necessarily be lethal.

(D) Inversions:

A section of the chromosome becomes changed by rotation at 180° is called inversion. The order of the genes in it are reversed.

Inversion arises by the formation of loops on a chromosome. Breaks may occur at the point of intersection of the loops (Fig. 43.13). Reunion of the broken ends takes place in a new combination, and inverts. Inversion heterozygotes are formed by loops and bulges in pairs.

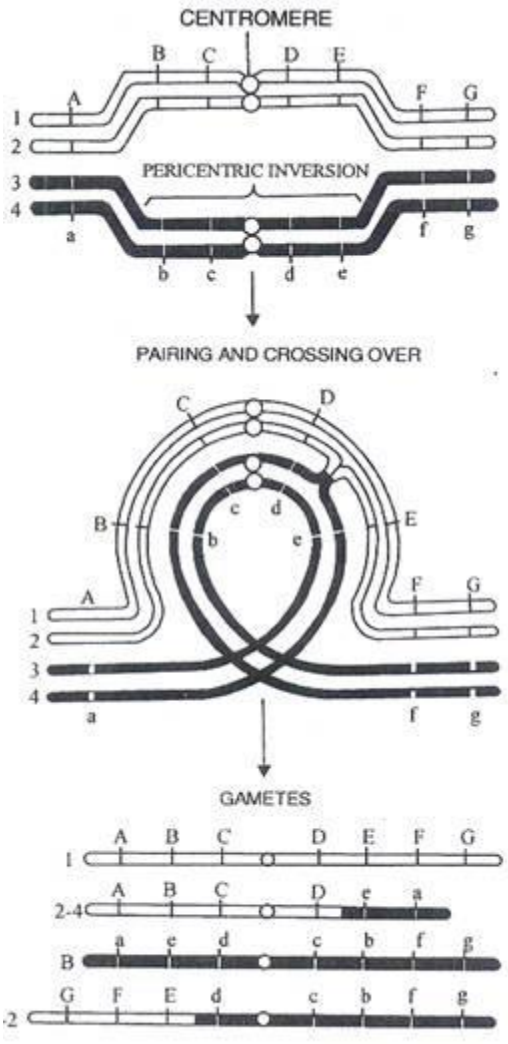

Fig. 43.13. Inversions.

Types of Inversion:

Paracentric inversion: The inverted segment doer no; include centromere. Pericentric inversion: Inverted segments include centromere.

Paracentric inversion:

A single crossing over inverted region will result into formation of a dicentric chromosome (with 2 centromeres) and an acentric chromosome (with no centromere). Of the remaining 2 chromatids one will be normal and the other will carry inversion. The dicentric

chromatid and acentric chromatid will be observed at anaphase I in the form of a bridge and a fragment (Fig. 43.14). Double crossover shows deficiencies and duplication (Fig. 43.15) giving rise to variations in anaphase I configurations.

Pericentric inversion:

In pericentric inversion centromere is in the inverted segments. In pachytene stage 2 of the 4 chromatids resulting after meiosis will have deficiencies and duplications. No dicentric bridge or acentric fragment will be observed (Fig.43.16). In pericentric inversion, if two breaks are not situated equidistant from the centromere, a change in shape of chromosome results. A metacentric chromosome may become submetacentric and vice versa (Fig. 43.17).

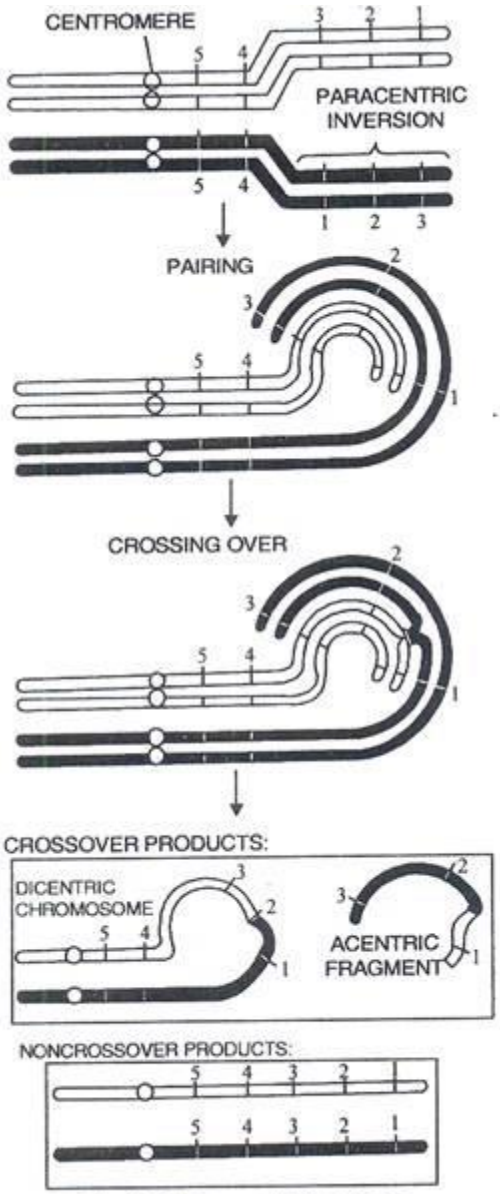

Fig. 43.14. Paracentric inversions.

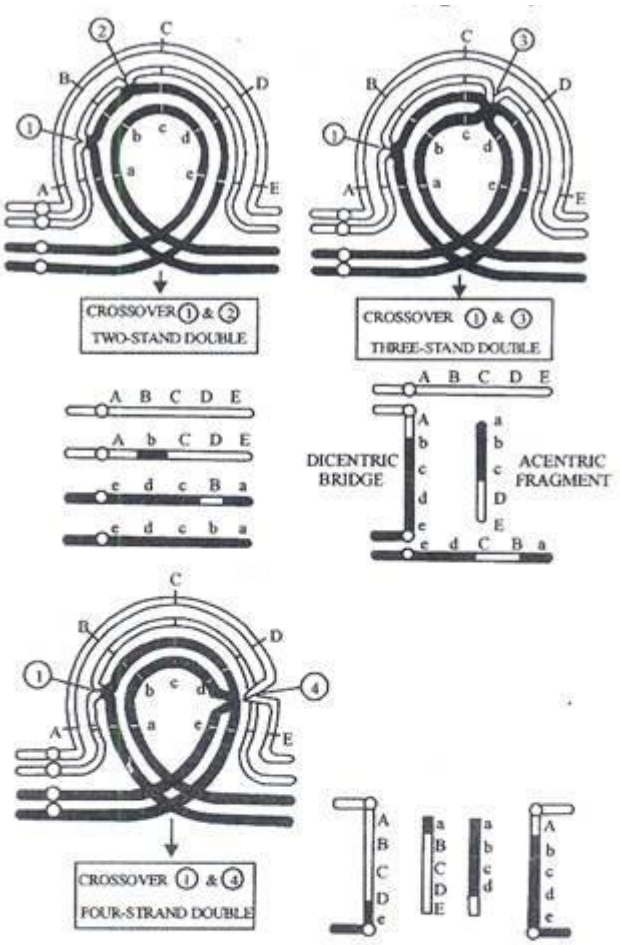

Fig. 43.15. Paracentric inversions result of double cross.

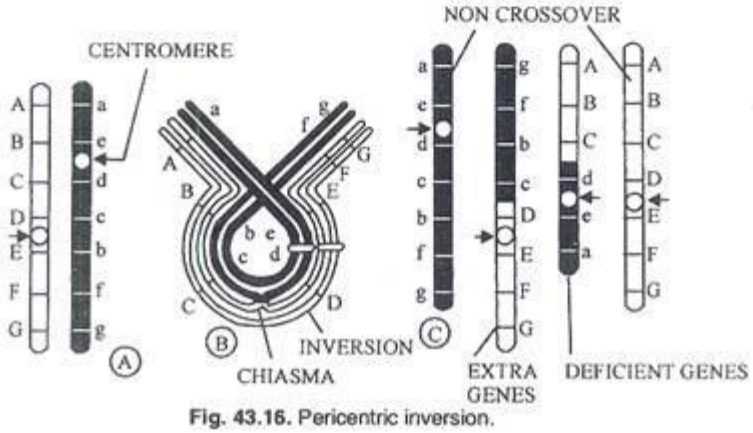

Fig. 43.16. Pericentric inversion.

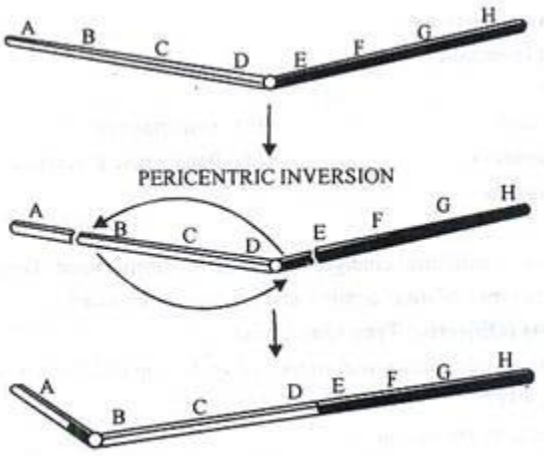

Fig. 43.17. Pericentric inversion and metaphase.