VIVEKANANDA COLLEGE

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NAAC ACCREDITED ‘A’ GRADE

Topic: Chromosomal Aberration

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Name of the Department: Department of Botany

Introduction

Chromosomal aberrations are chromosomal mutations or variation in the structure of chromosomes involving changes either in the total number of gene loci on a chromosome or a rearrangement of their order.

- Changes involving the number of gene loci include deletion/deficiency
- Changes involving the rearrangement of gene loci include inversion and translocation.

The change may take place at the same locus in both homologues of a pair when it is called a structural homozygote or it may occur in any one of the chromosomes of a homologous pair when it is called a structural heterozygote. Sometimes in a structural heterozygote a dominant gene is lost from one homologue, allowing the expression of the recessive gene in the other homologue, this is called pseudodominance.

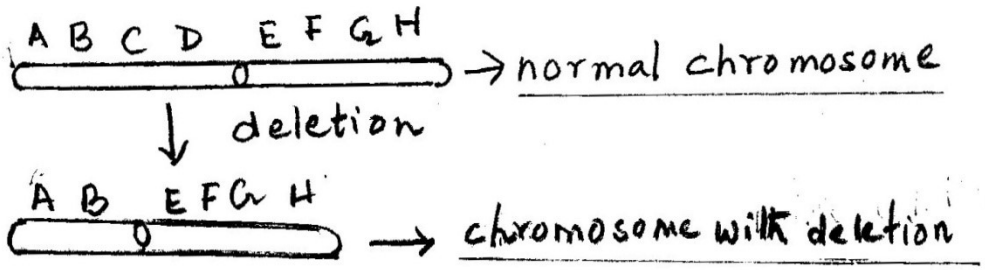
Deletion/Deficiency

This type of chromosomal aberration occurs when a chromosomal segment is lost due to breaks appearing in chromosomes on exposure to toxic chemicals, drugs or radiations.

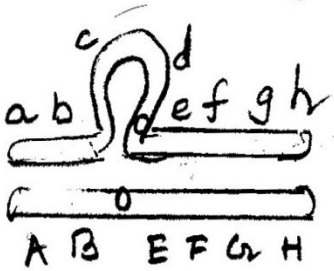
Loss of a segment from the end of a chromosome is caused by a single break and is termed deficiency. Whereas, loss of an intercalary segment caused by two breaks is termed deletion. The breaks occur at random and may involve one chromatid (chromatid break) or both chromatids of a chromosome (chromosomal break).

The length of the segment deleted may include one or more genes. Usually the broken segment does not survive if it lacks a centromere and the acentric fragment is lost. If the broken fragment carries a centromere it functions as a genetically deficient chromosome. When a large segment is lost from a chromosome it is deleterious.

Deletion Diagrams



Pairing of deleted chromosome with its homologue at pachytene by loop formation



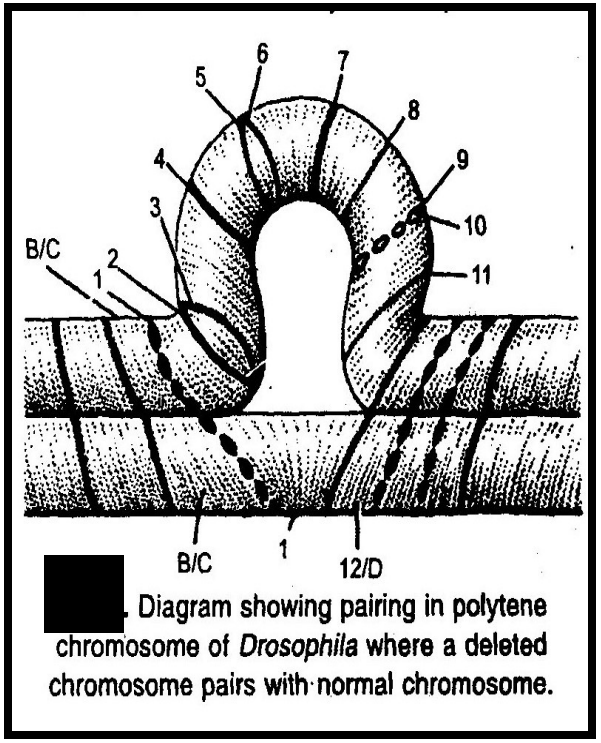


Diagram showing pairing in polytene chromosome of *Drosophila* where a deleted chromosome pairs with normal chromosome.

Taken from GENETICS by V.B.Rastogi

Phenotypic effect: A phenotypic effect of deletion is observed in human beings. Deletion of a part from the short arm of chromosome V in man results in '*cri-du-chat*' or cat cry syndrome where the child shows a mewing cat-like cry and improper development of mental faculty.

Cytology: During meiotic pairing the part of the normal chromosome corresponding to the deleted part bulges out in the form of a loop over the deleted part. Such buckle-like loop formation can be clearly observed in the polytene chromosomes of *Drosophila*.

Significance: When the deletion is small no detectable morphological change in the chromosome is observed and no significant change in the organism occurs. But large deletion are detrimental to the organism. Deletions may be used to locate genes and construct genetic maps of the banded polytene chromosomes where loss of particular bands can easily be associated with loss of certain characters.

Duplication

This is a type of chromosomal aberration where a particular segment of the chomosome is present more than once (carrying the same block of genes) in the haploid complement. According to the positions of the repeated segment duplications may be of the following types:

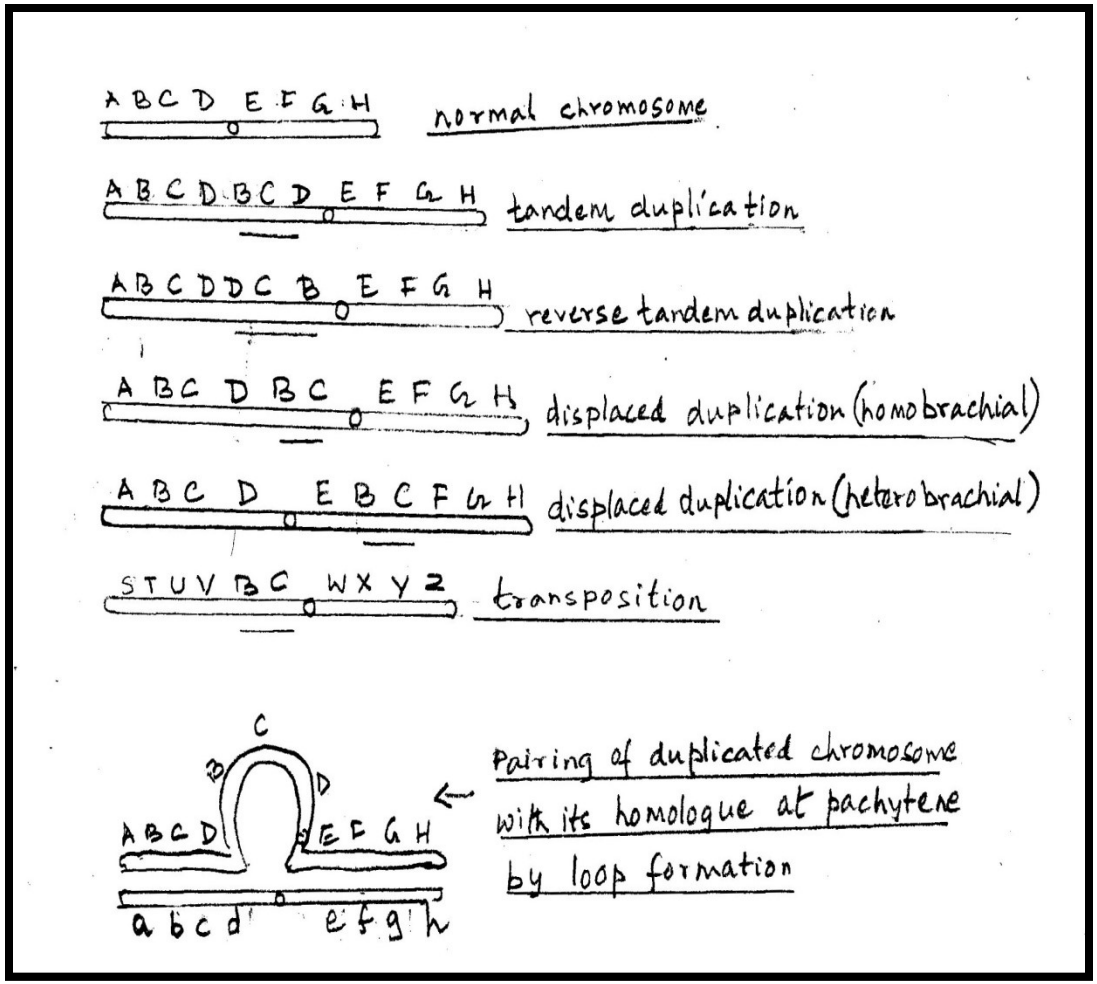
- Tandem duplication- here the repeated segment with the same gene sequence lies adjacent to the original segment.
- Reverse tandem duplication- here the repeated segment with the same genes is located adjacent to the original segment but the order of the genes is reversed.
- Displaced duplication- here the repeated segment with the sane gene sequence is located away from the original segment, either on the same arm of the chromosome (displaced homobrachial) or a different arm of the same chromosome (displaced heterobrachial) or to a non homologous chromosome (transposition).

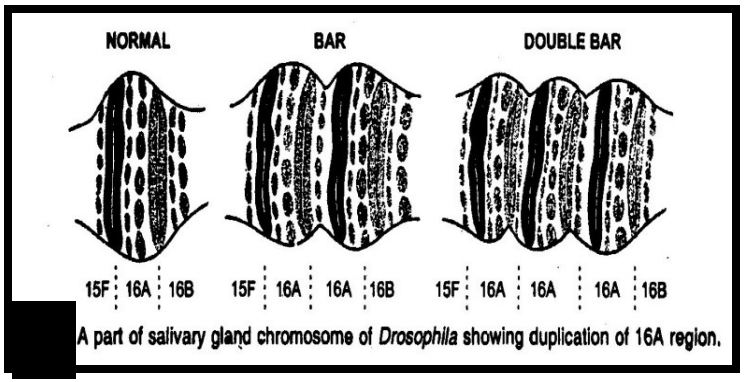
Phenotypic Effect: A very good example of duplication is the bar eye character (narrow, slit-like eye) of *Drosophila*. The bar eye character is a dominant X-linked character. It is controlled by 7 bands in the 16A region of the X-chromosome. In normal females the 16A region is represented once in the X-chromosome. In bar-eyed females the 16A region is represented twice. In double bar-eyed (eye narrower) females it is represented thrice.

Cytology: During meiotic pairing the duplicated part of the chromosome bulges out in the form of a buckle-like loop over the normal chromosome.

Significance: Duplications unlike deletions are more frequent and less deleterious. They do not lower the viability but produce abnormality of structure and function. Duplications play a significant role in evolution because they increase the number of genes on the chromosome. These additional genes might get modified by mutation. Under these conditions some lethal mutations might get converted to tolerable or beneficial ones. They may overcome the effect of deletion, thus reducing their effect.

Duplication Diagrams





A part of salivary gland chromosome of *Drosophila* showing duplication of 16A region.

Taken from GENETICS by V.B.Rastogi

Inversion

Sometimes the number of genes on a chromosome is not changed but the sequence of genes is altered by the rotation of a gene block through 180°

Inversions can be of two types:

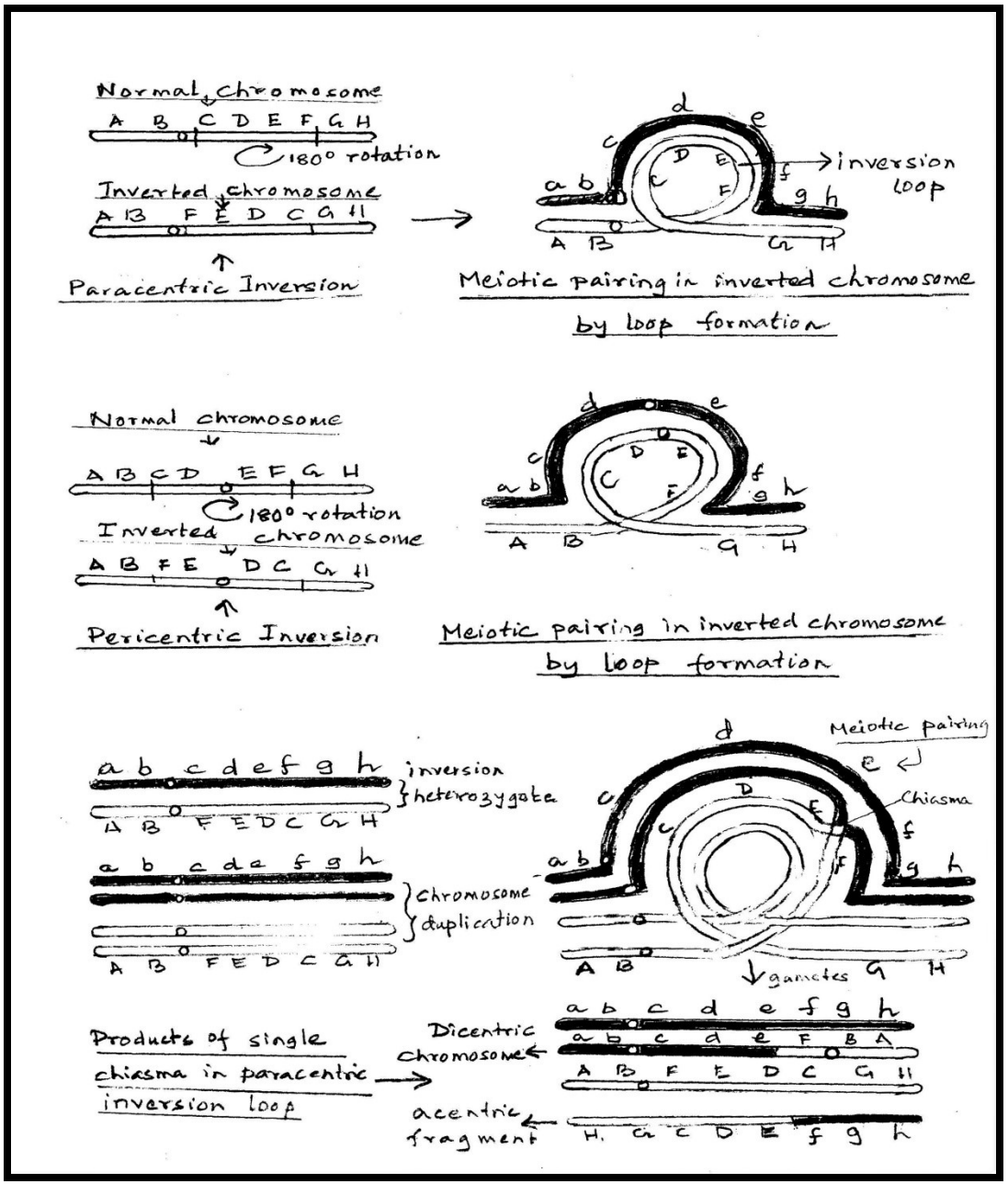
- Paracentric –where both breaks in a chromosome during inversion occur on the same arm (homobrachial) and thus the inverted segment does not include a centromere. Paracentric inversions do not change the morphology of the chromosome.
- Pericentric -where one break occurs on either side of the centromere, involving both the chromosome arms. Thus the inverted segment includes a centromere. If the breaks on both chromosome arms are not symmetrically placed then the chromosome morphology changes. For example a metacentric chromosome may become sub-metacentric.

Meiotic Behaviour: The chromosomes of a homologous pair having identical inversions (structural homozygote) undergo normal meiotic pairing . However, in a structural heterozygote with only one homologue carrying the inversion pairing occurs by loop formation. When crossing over occurs within the inversion loop, the products vary in case of paracentric and pericentric inversions.

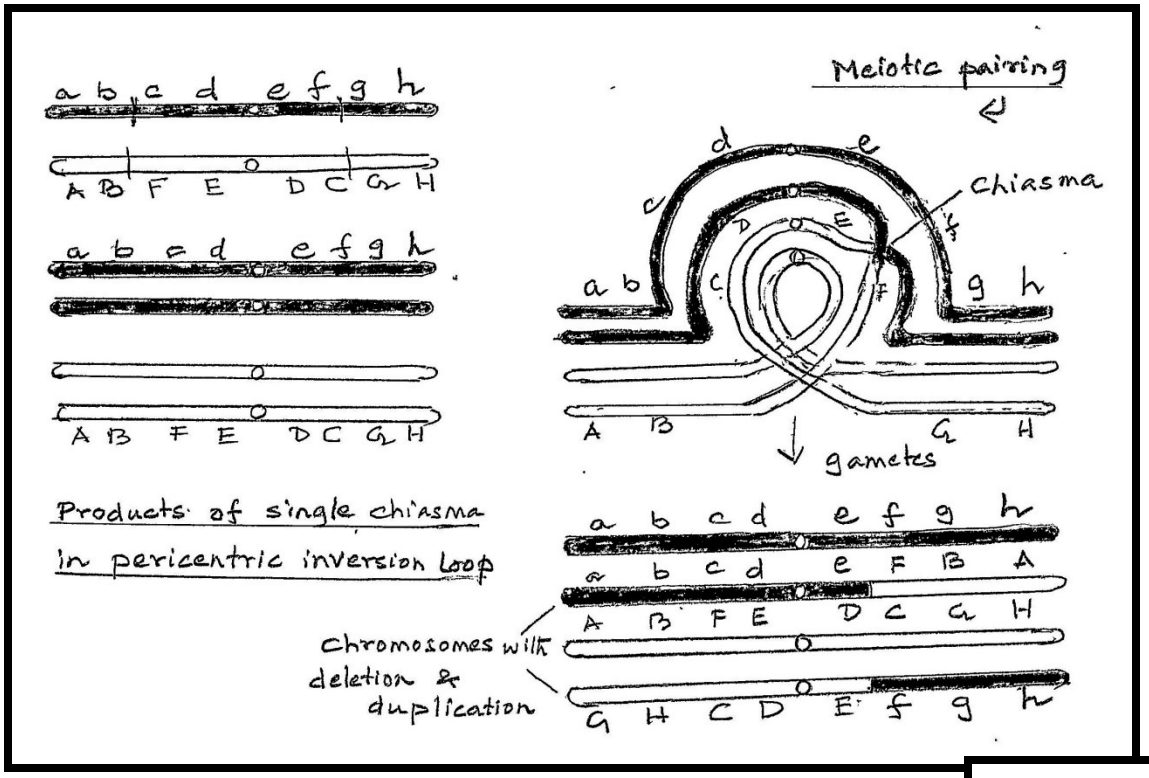
- In paracentric heterozygous inversion with one chiasma in the inverted region, the products include an acentric fragment and a dicentric chromosome (both of which are non-viable) in addition to a normal and an inverted chromosome (viable).
- In pericentric heterozygous inversion with one chiasma in the inverted region all the products are chromosomes with one centromere but one carries deletion and the other duplication which are rarely viable.

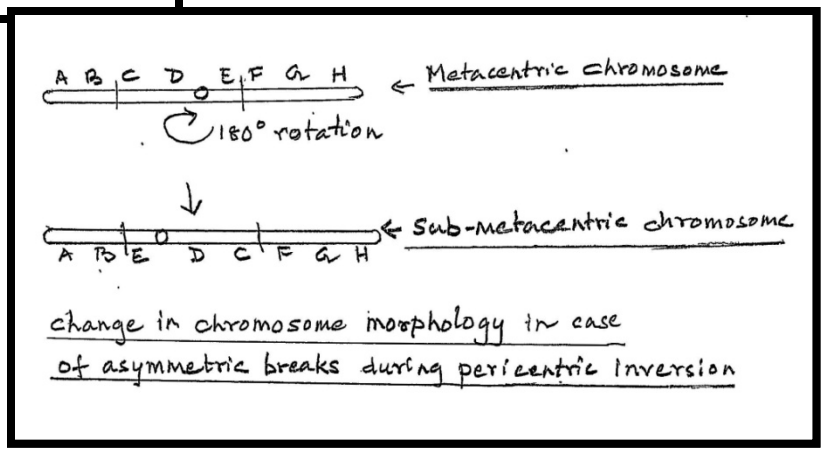
Significance: There may be phenotypic changes due to position effect. Chromosome morphology may be changed in pericentrics with asymmetric breaks. Inversions suppress crossing over and help preserve beneficial arrangement of genes. Sterility of gametes result from crossing over in paracentric and pericentric inversions.

Inversion Diagrams



Inversion Diagrams cont..





Translocation

This is a type of chromosomal aberration in which a segment of a chromosome is transferred to another non-homologous chromosome. Translocation may be non-reciprocal where a segment of a chromosome is shifted to another non-homologous chromosome or it may be reciprocal where there is a mutual exchange of chromosome segments between two non-homologous chromosomes. These may again be structural homozygotes or heterozygotes.

Meiotic behaviour of translocation heterozygotes:

Homozygous translocations cannot be detected cytologically as they form regular bivalents during pachytene of meiosis. Genetically they may influence the phenotype due to position effect. They are non-lethal and do not influence the viability of gametes. However,in heterozygous reciprocal translocations the two normal and two translocated chromosomes from a cross-shaped structure in order to undergo pairing during pachytene of meiosis.

The occurrence of cross over and chiasma formation in each arm of the quadrivalent results in the formation of a ring of 4 chromosomes at diakineses and metaphase-I (either open or twisted). The anaphasic movement towards the poles takes place in one of the 3 ways:

- **Alternate segregation**: The ring is twisted and alternate chromosomes move to the same pole. The normal chromosomes move to one pole and the translocated ones to the other. Therefore gametes receive the full complement and are all viable.
- **Adjacent-1 segregation**: The ring is open and one normal and one translocated chromosome move to each pole (non-homologous centromeres move to same pole). Half the gametes are non-viable as they carry deletions and duplications.
- **Adjacent-2 segregation**: The ring is open and one normal and one translocated chromosome move to each pole (homologous centromeres move to same pole). Half the gametes are non-viable as they carry deletions and duplications.

Sometimes multiple trans locations may occur involving more than 2 pairs of chromosomes resulting in the formation of multivalent ring.

Significance: Heterozygous reciprocal translocation results in sterility of the gmetes. Translocations lead to formation of new species.

Translocation Diagrams