CHROMOSOMAL ABERRATIONS

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Chromosomal Aberrations

Chromosomes have definite structure and organization, which is normally constant from one cell division to next. Sometimes undergo certain structural modifications which are known as chromosomal aberrations.

Chromosomal aberrations may be

- **Deletion – Loss of a chromosome part**
- **Duplication – Segment of a chromosome is repeated**
- **Translocation – Part of a chromosome breaks off and attaches to another, non-homologous chromosome**
- **Inversion – Part of a chromosome is oriented in the reverse of its usual direction**

DEFICIENCY OR DELETION

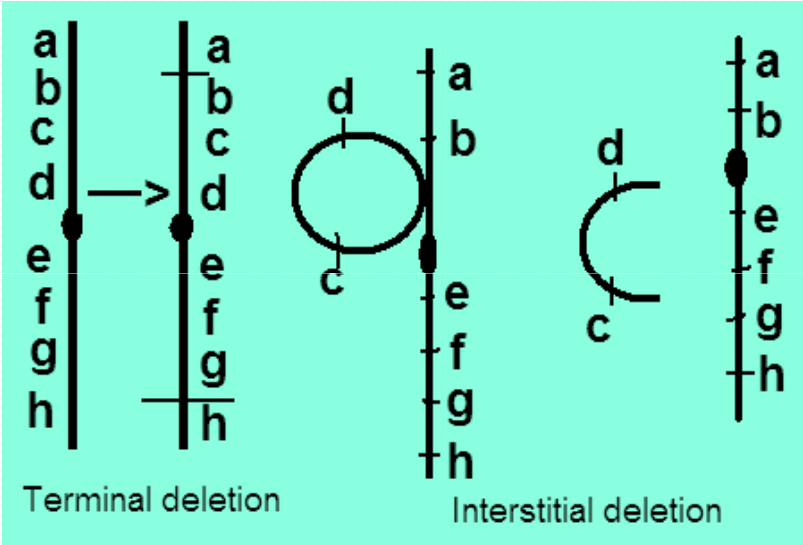
This is due to loss of a segment in the chromosome. Deletions are of two types, *i.e.*, terminal or interstitial

If a single break occurs near the end of a chromosome, it results in a terminal deletion.

The interstitial deletion results from the breakdown in the chromosome followed by the reunion on the broken ends.

Interstitial deletions are more common and terminal deletions are rare.

Deletion …



i. Genetic Significance:

If the missing genes are of physiological importance, the organism will not survive.

ii. Pseudodominance:

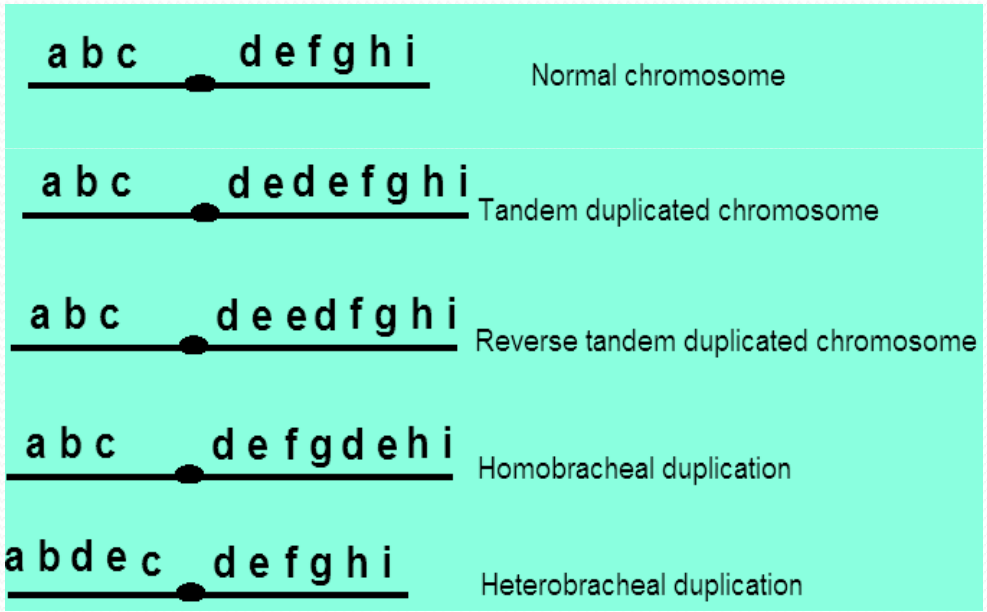
If dominant 'A' is lost in a deletion, then recessive 'a' will express itself. This expression of a recessive trait is called pseudodominance.

Duplication:

When a segment of the chromosome is represented two or more times in the chromosome of a homologous pair *i.e.*, in duplicate

or

when a segment of a chromosome is repeated it is known as duplication.



Duplication continued:

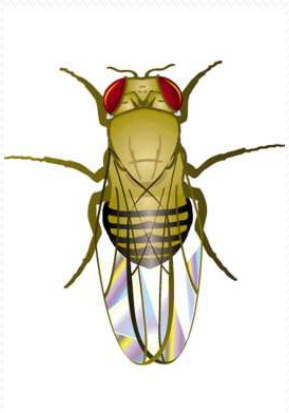
The duplications are not deleterious. Moreover the duplication is useful in evolution of new genetic material

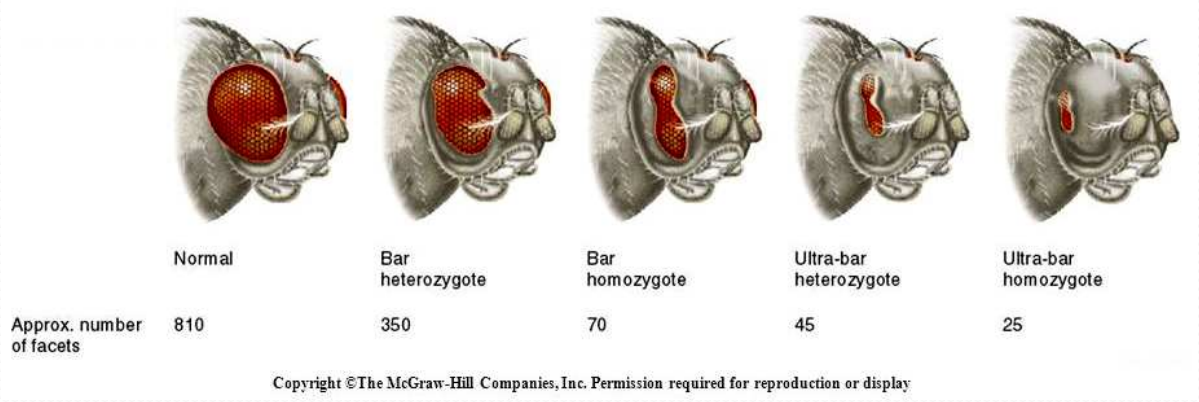
position effect *i.e*, reallocation of chromosomal material without altering its quantity may result in an altered phenotype

***eg*., Position Effect in Drosophila**

Duplication continued:

***eg.*, Position Effect in Drosophila, the wild type eye is large and has an 810 facets. It has two bar regions, B, one on each X chromosome. If bar region undergoes duplication, the size of eye is reduced.**





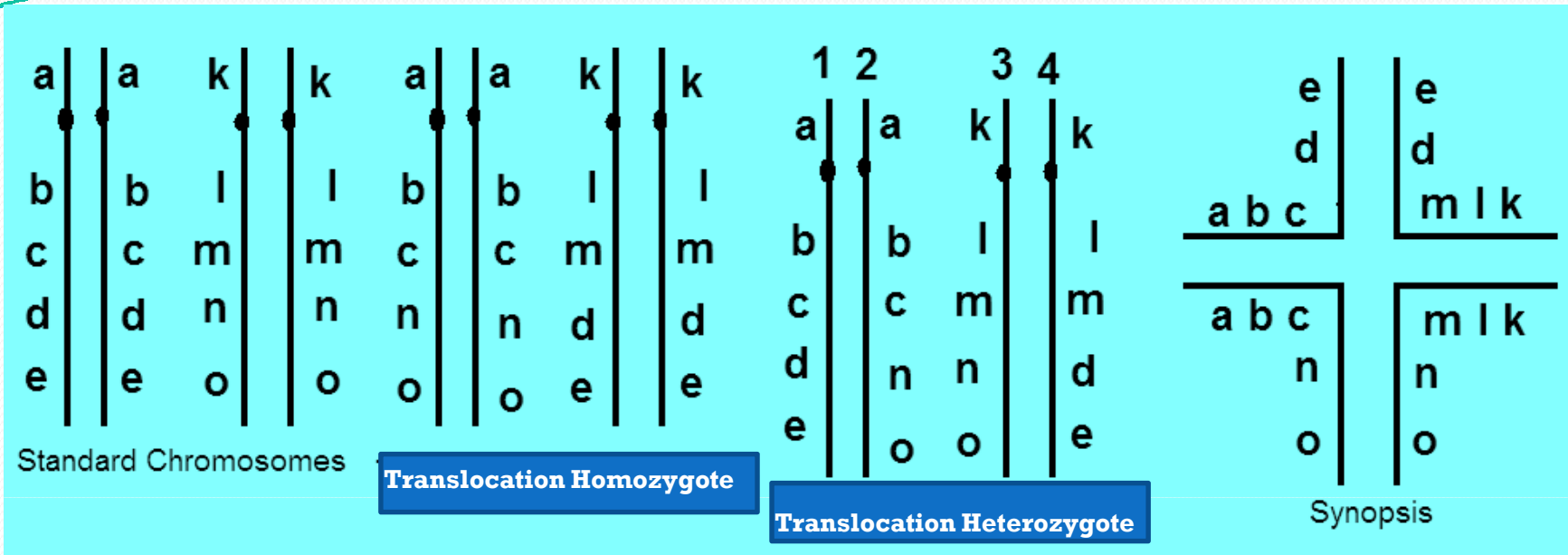
TRANSLOCATION

The shifting or transfer of a part of a chromosome or a set of genes to a non homologous one is called translocation. Translocation may be three types.

- **Simple translocation:** The broken piece gets attached to one end of the chromosomes.
- **Shift translocation:** The broken segment of one chromosome gets inserted interstitially in a chromosome.
- **Reciprocal translocation:** A segment from one chromosome is exchanged with a segment from another non homologous one, so that in reality two translocation chromosomes are simultaneously achieved.

They are of two types *i.e.*, Homozygotic and heterozygotic translocation.

TRANSLOCATION continued



Homozygotes do not have any cytological peculiarities and undergo regular pairing during meiosis and cannot be detected cytologically.

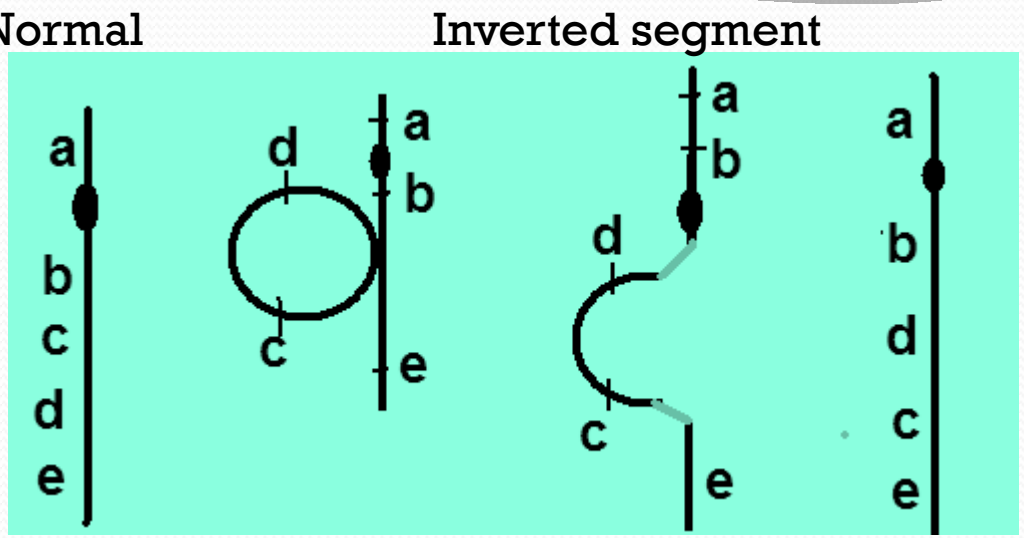
In translocation heterogzygotes, breaks occur on only one of the chromosomes of a homologous pair. In heterozygotic translocation a considerable degree of meiotic irregularities occur.

INVERSION:

It is an intrachromosomal aberration, in which a section of the chromosome becomes changed by rotation through 180 degrees. The order of the genes in the section is thus reversed.

In a diploid organism, when out of two homologous chromosomes one chromosome undergoes the inversion then it is called inversion heterozygote.

Inversions are of two types: *i.e.*, pericentric and paracentric inversions.



Pericentric inversions includes the centromere while in paracentric inversions the centromere is outside the inverted segment.

When crossing over occurs within the inverted segment of a paracentric inversion, then acentric and dicentric chromatids formed. The acentric chromoatids fail to move to either pole due to lack of centromere. The dicentric chromatids have two centromeres and are connected by a bridge, breaks and contains duplications and deficiencies.

Acknowledgements

to

1. Internet

2. Cell Biology by C B Powar