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Ajanta
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Date 10-6-2020

Qu. Write notes on Chromosomal Abberation?

1. Meaning of Chromosomal ~~Alteration~~ Abberation: Alteration in the structure of individual Chromosome or Chromosomal abberation 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 break in the Chromosome.

Structural Changes of Chromosome

- Changes involving the number of genes
  - Deficiency
  - Duplication
- Changes involving the arrangement of gene.
  - Inversion
  - Translocation

Different types of Structural Change of CHROMOSOME.

(2.) Types of Chromosomal Aberration :—

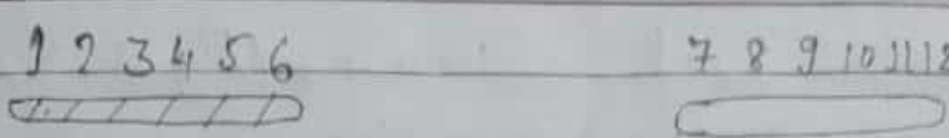
(I) Deficiency (parts of chromosomal lost or deleted),

(II) Duplication (parts of chromosome added or duplicated),

(III) Inversion (Section of chromosome detached and joined to non-homologous Chromose).

(IV) Translocation (parts of Chromosome detached and joined to non-homologous Chromosome).

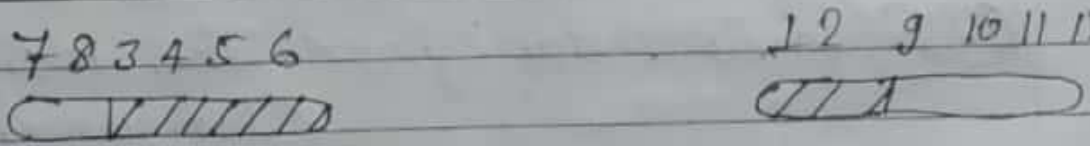
Of the various Chromosomal aberrations, inversions and translocations only represent and translocations only represent Changes in position of Chromosome segments of different sizes, the total Chromosome mass remaining unchanged. All segments are present in the original dosage, but distributed in a new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



Two nonhomologous Chromosome.

Deletion Deficiency

addition Duplication



Translocation

Inversion

Fig:- Different Kinds of Changes in Chromosome Structure.

Table 12.1 : Common Types of Chromosomal Abberations.

Type.	Change in Chromosome Structure.
Deficiency	Loss of a segment of Chromosome.
(1) Terminal deletion	Last Segment include telomere.
(2) Interstitial deletion	A Segment b/w telomere and Centromere is last.
Duplication.	
(1) Tandem duplication	The additional Chromosome segment located just after the normal Segment
(2) Reverse tandem duplication	Same as above but the gene sequence of the additional Segment is inverted.
(3) Displaced duplication	The additional Segment located in the same Chromosome but away from the Normal Segment.

Structural homozygotes are those in which alterations such as translocation or duplication occur in both the homologous Chromosome 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.

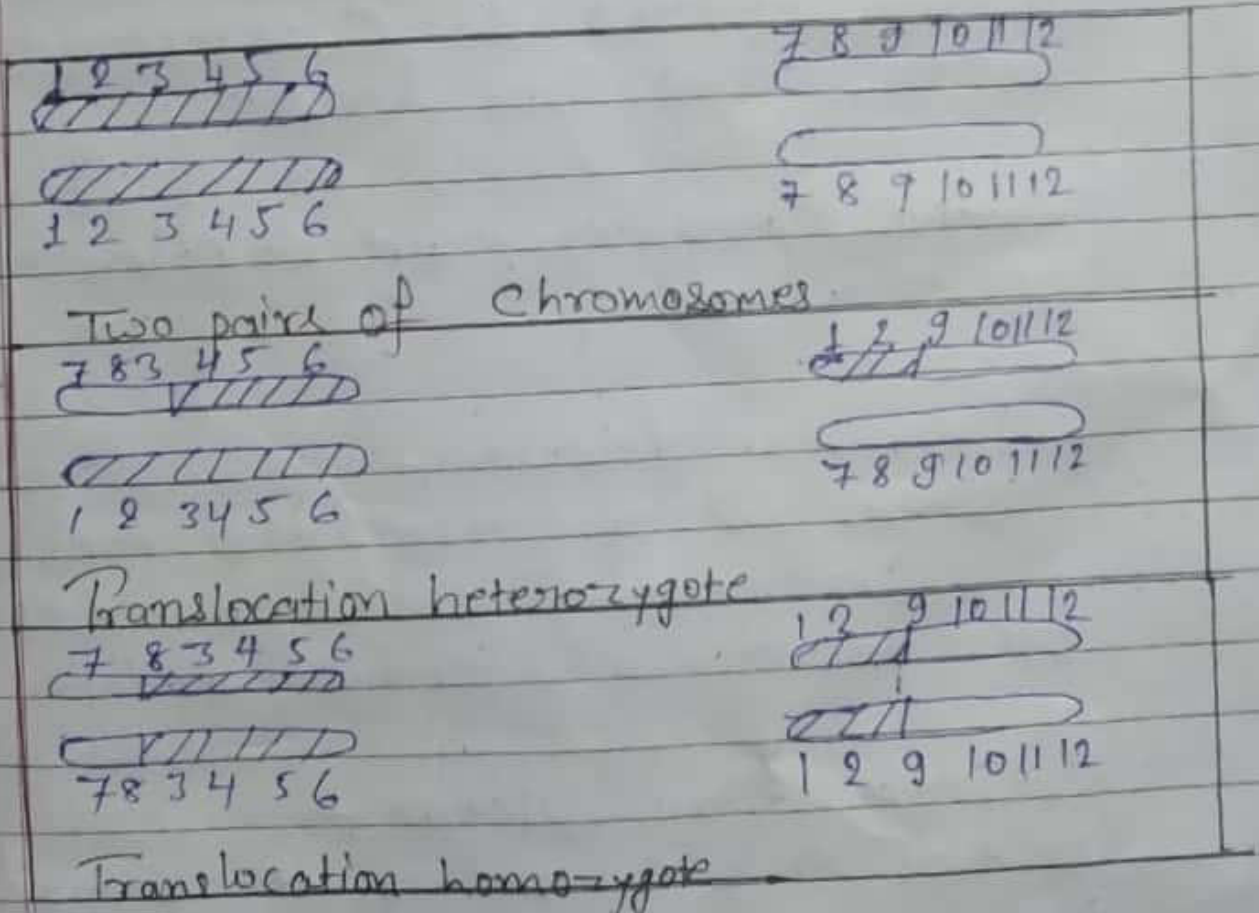


fig:- Chromosome Constitution of Structural heterozygote and a homozygote.

(3.) Deficiency of Chromosomal Aberration:-
Deficiency or deletion represents a loss in Chromosomal material and was the first Chromosomal materia and was the first Chromosomal aberration indicated by genetic evide This evidence, presented by Bridges in 1971 in Drosphila melangaster, showed a deletion of the X- Chromosome that included the Bar locus.

~~* Types~~ 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 terminal deficiency.

(II) Intercalary deletion:-
If two breaks occur, a section may be deleted and an intercalary deficiency is created.

* Origin :- Origin of intercalary deficiency is represented in fig. 12.4. Terminal deficiency might seem less complicated and more likely to occur than those involving two breaks.

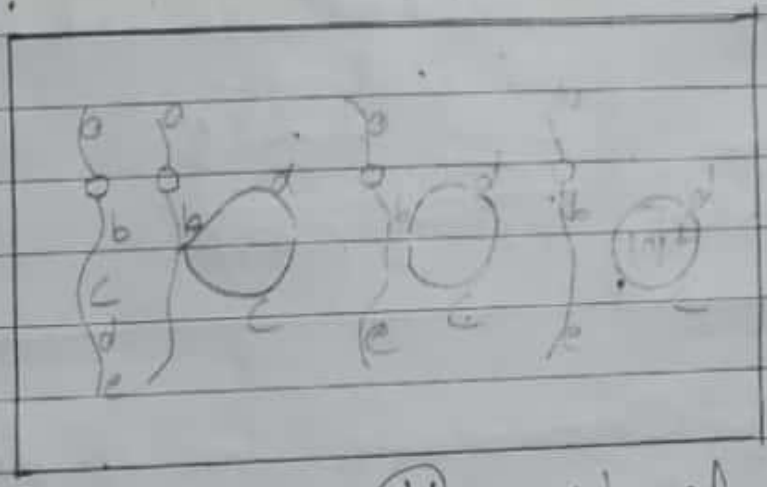


Fig :- 12.4: The origin of a deletion.

* Effect of Chromosomal Aberration :- Deficiencyes have an effect on inheritance also. In presence of a deficiency, a recessive allele will behave like a dominant allele and this phenomenon is called pseudo dominance. Thus Chromosome deficiency have greatly facilitated the cheaking of linkage maps.

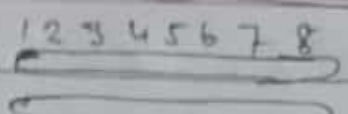
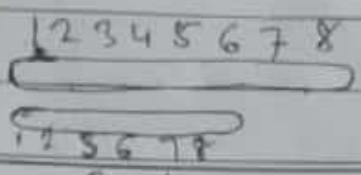
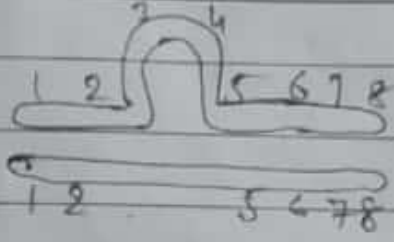
A pair of homologous Chromosomes	
Deficency heterozygote	
Pachytene Configuration	

Fig:- Chromosome in a deficiency heterozygote.

(4) Origin of Duplication of chromosomal Aberration:-
Duplication originates out of unequal Crossing over (Fig. 12.6)

Fig. 12.6 The origin of a duplication.

Effect of Duplication of Chromosomal Aberration:-

The duplication was critically examined in the B (bar) locus of the X-chromosome of Drosophila. The Bar Charactor is due to duplication in region 16A of X-chromo-some (Fig. 12.8).

Bared eye individuals - some (16A 16A) give rise to ultra-bar (16A 16A 16A) and normal wild type (16A) due to unequal Crossing over (Fig. 12.9) Barred eyes have different phenotypes in homozygous bar and heterozygote.

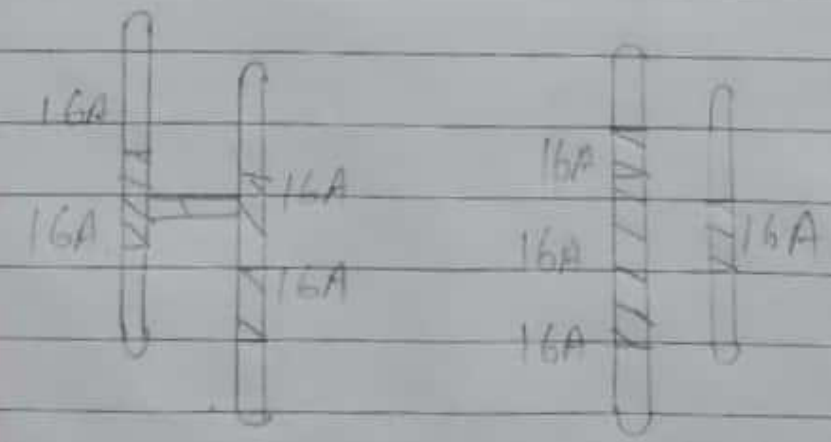


Fig:- 12.9: An ultrabar Condition resulting from 'Bar' due to unequal Crossing over.

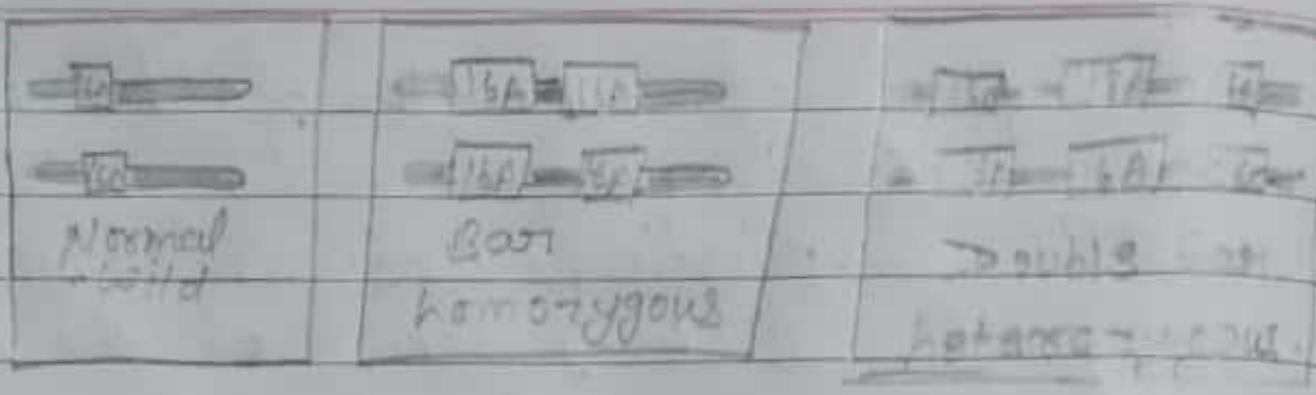


Fig:- Digram Showing position effect.