Structural aberrations of chromosomes

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CHROMOSOMAL ABERRATIONS

- Chromosomal changes are abnormal structural and numerical changes
- They are also called ***chromosomal aberrations*** or ***chromosomal mutations***
- Gross structural changes of chromosomes are called ***structural*** *aberrations* or ***chromosome re arrangements***

- Numerical changes are called ***ploidy changes*** or ***numerical aberrations***

STRUCTURAL CHANGES

Structural changes of chromosomes involve the gain , loss or relocation of chromosome segments and genes

- **Intrachromosomal aberrations (homosomal aberration)**
- **Interchromosomal aberrations**

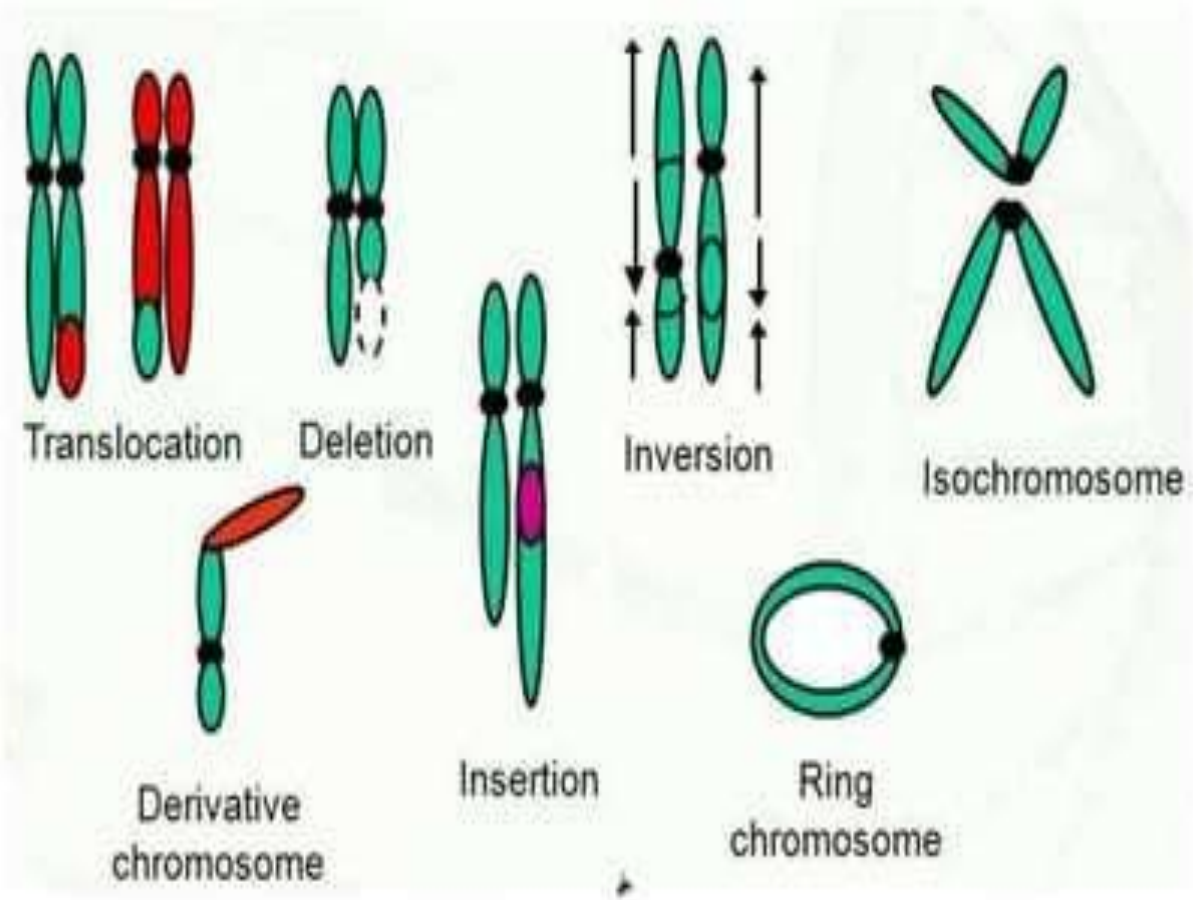
(i) Alleosomal aberrations

(ii) Heterosomal aberrations

Chromosomal aberration are of four basic types

1. **Deficiency or deletion**
2. **Duplication**
3. **Inversion**
4. **Translocation**

CHROMOSOME STRUCTURE ABNORMALITIES



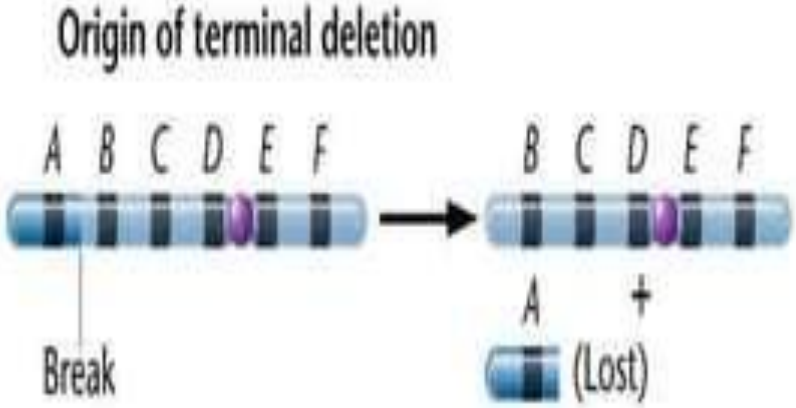
(1). DELETION OR DEFICIENCY

- Loss of a chromosome segment, together with the genes contained in it
- **Terminal deletion** – loss of a terminal segment, with a single break in the chromosomes
- **Intercalary deletion-** loss of an intercalary segment, with two breaks

*__Homozygous deletion__ – a segment is lost from both the chromosomes of a homologous pair

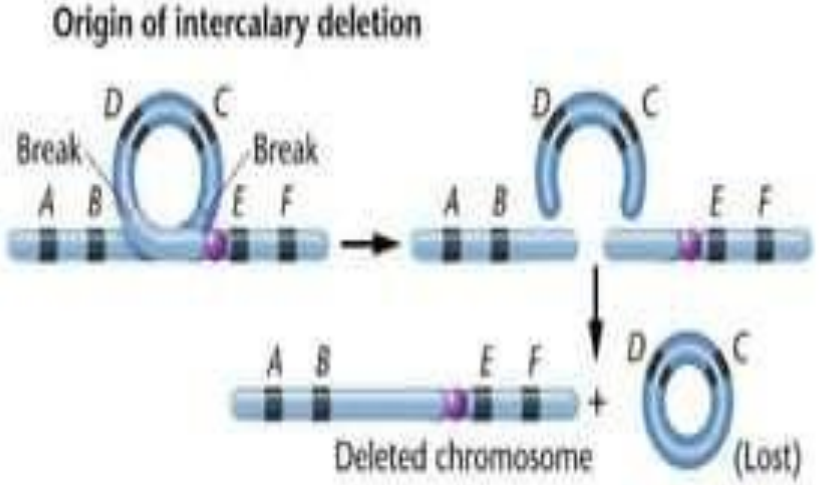
*__Heterozygous deletion__ - a segment is lost only from one of the two homologues

- **Terminal deletion**



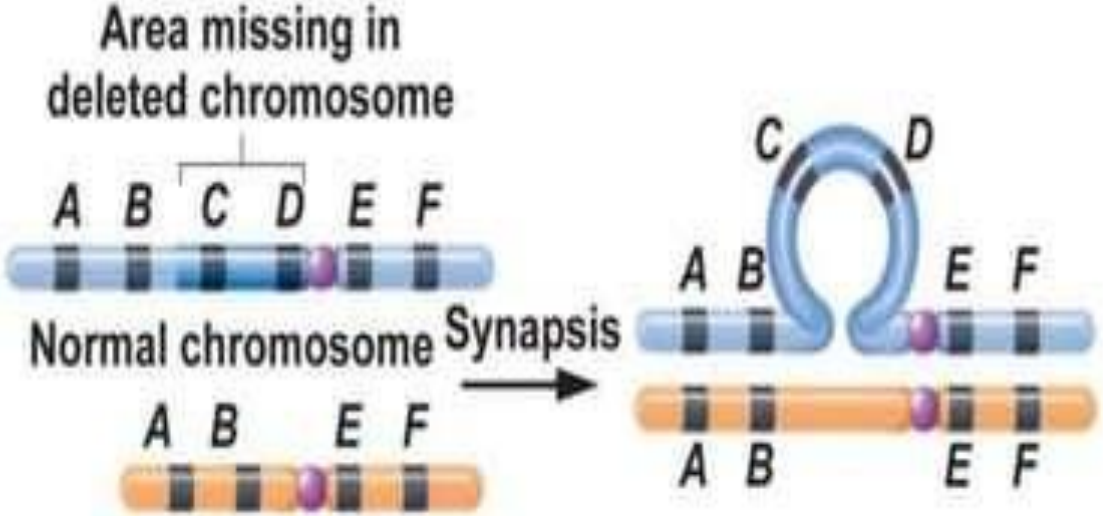
Deletion involving the terminal part of a chromosome and leading to an adhesive terminus.

Intercalary deletion



An intercalary chromosome deletion occurs when a segment of DNA is lost from the interior of a chromosome.

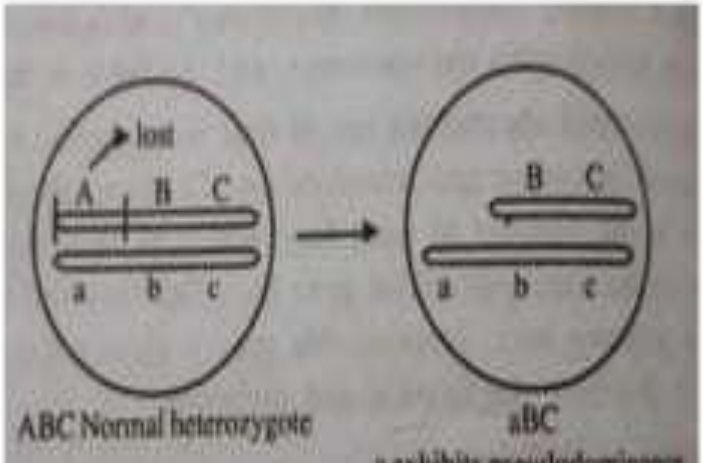
SYNAPSIS IN DELETION OF HETEROZYGOTE



a segment is lost only from one of the two homologues

PSEUDODOMINACE

The expression of the recessive alleles due to the deletion of the dominant alleles. Eg: waltizing mouse



EFFECTS OF DELETION

- Deletion involves the loss of genes so they have deleterious effect on organisms
- If the lost genes are crucial for viability, sterile gametes or non functional somatic cells may result
- In human genome deletion mutation

Granulocytic leukemia – deletion of long arm of chromosome 21

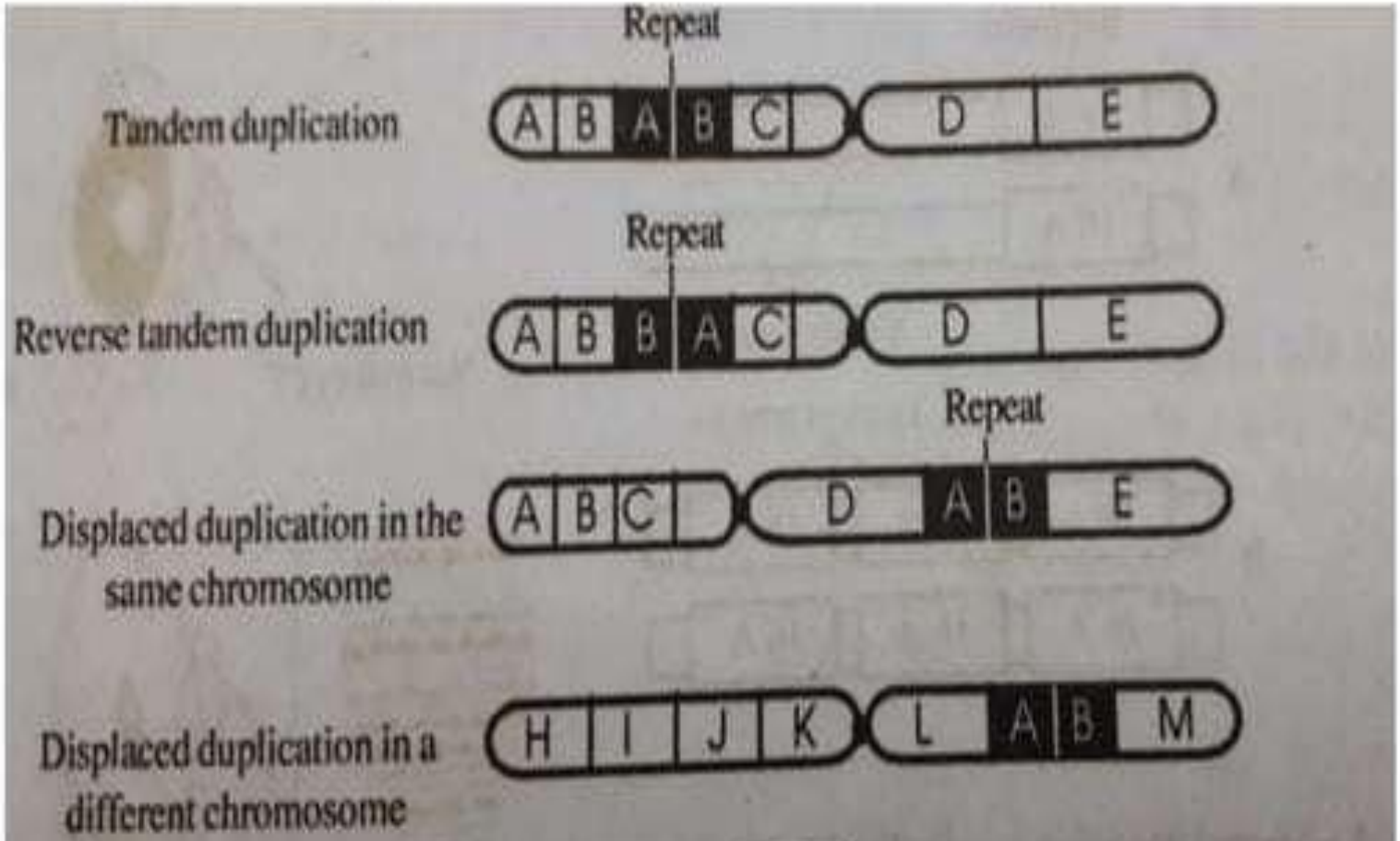
Cri-du-chat syndrome – heterozygous deletion in the short arm of chromosome 5

(2.) DUPLICATION

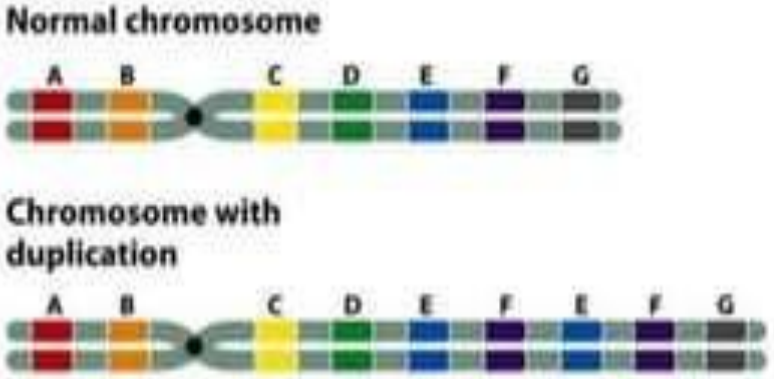
- Duplication is the doubling or repetition of chromosome segment during chromosome duplication
- As a result of it, a set of genes gets doubled or repeated
- The extra set of genes is generally called "***repeat***"
- Three types

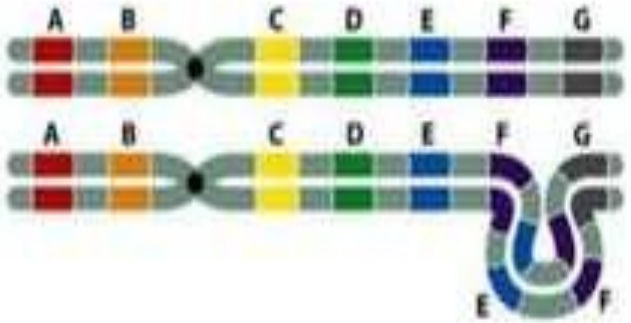
1. **Tandem Duplication** – The extra segment and the parent segment are next to each other and have the same order of genes
2. **Reverse tandem duplication** - The extra segment lies next to parent segment with reversed gene sequence
3. **Displaced duplication-** The duplicate segment lies some distance away from the parent segment

Possible way in which a duplicate segment is incorporated



Homozygous &Heterozygous duplication





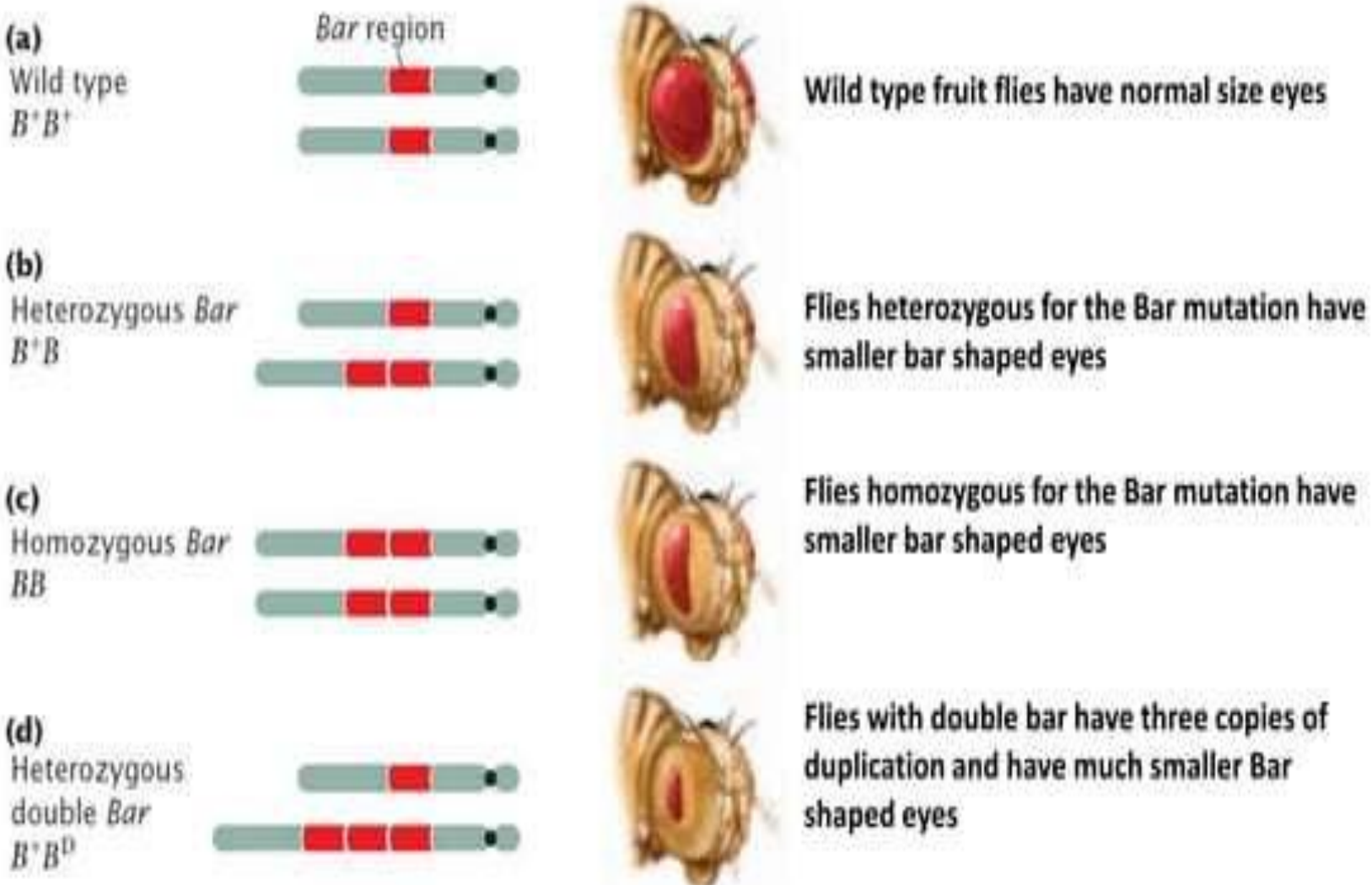
Homozygous duplication: both of the homologues of a pair of chromosomes will have homologous duplicated sequences

Heterozygous duplication: one of the homologues of a pair of chromosomes will have homologous duplicated sequences

EFFECTS OF DUPLICATION

- Duplications are more frequent and less deleterious than deletions
- Duplications are believed to be the important raw materials of organic evolution
- Large duplications affect the regulation of gene activity and there by causes phenotypic alterations which in turn promotes speciation and evolution

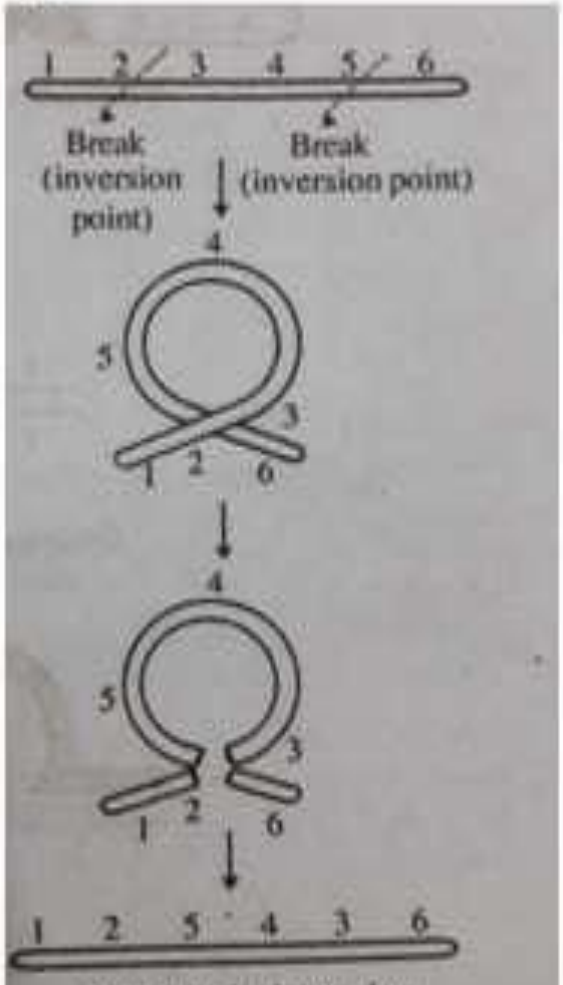
The Bar-eye phenotype in Drosophila



(3.) INVERSIONS

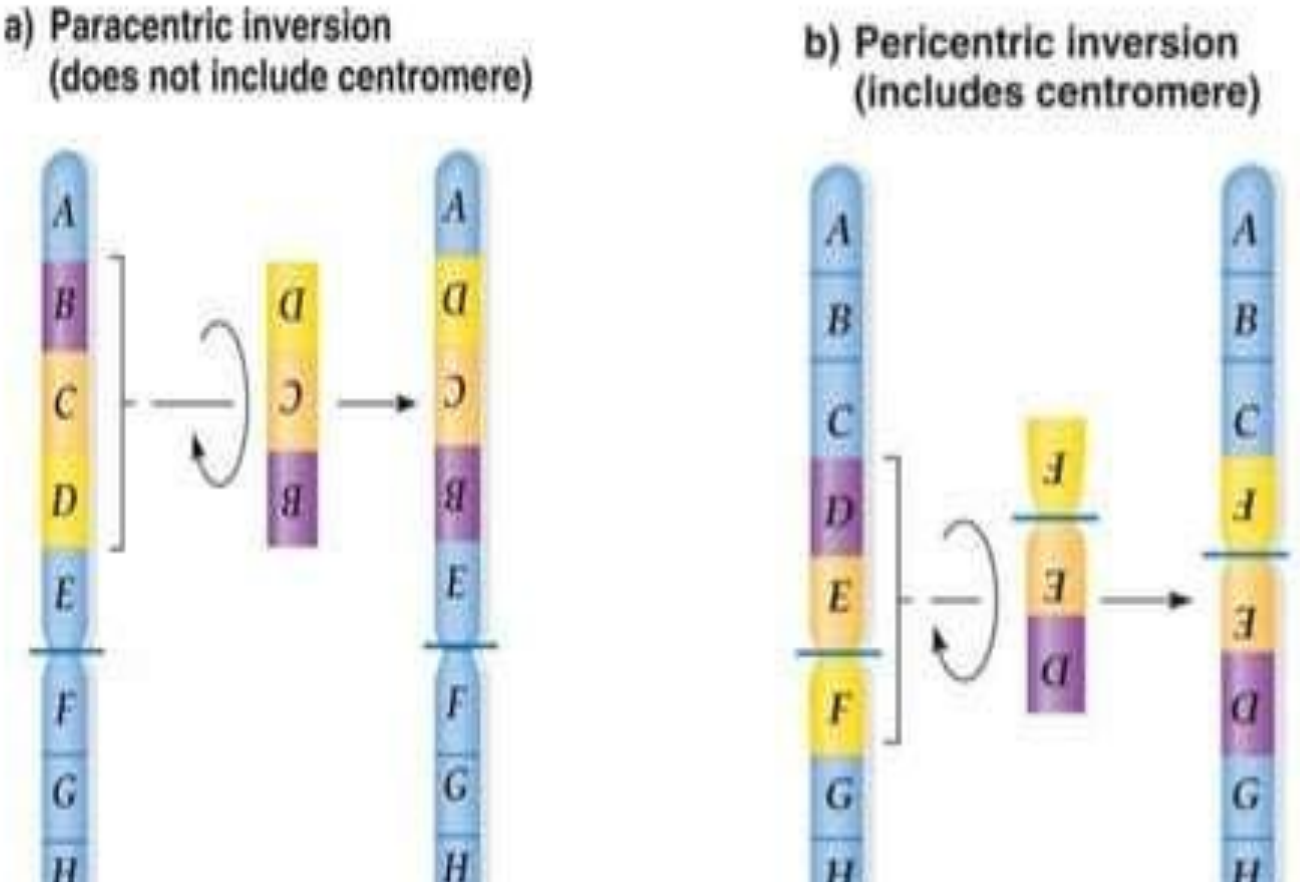
- Inversion the reversal of the linear order of a chromosome segment and its gene sequence
- Belived to be occur in intercalary segments – **intercalary inversion**
- ***Breakage re union hypothesis:*** **Intercalary inversion** involves the breakage of a chromosome segment and its subsequent re-insertion into the same location in a reversed orientation
- In this case two breaks occur in a chromosome – inversion points
- ***Paracentric Inversion** –The rotation(180°) and reinsertion of an **acentric** segment
- ***Pericentric Inversion-** The rotation(180°) and reinsertion of a **centric** segment
- Paracentric inversion is of two types
- (i) **Intraradial or homobranchial inversion** – single inversion in one arm

Origin of an inversion



- **Paracentric Inversion**

Pericentric Inversion



Inversions cont.

- Based on the no. of the inverted segments and the no. of breaks in chromosome two major kind of inversions can be recognized

*__Single Inversions__ – only one segment is inverted

*__Complex inversions__ - more than one segment would be inverted

- __Homozygous inversion__ – both the homologues are inverted
- __Heterozygous inversion-__ one homologue is un inverted and its companion is inverted

Complex inversions

- **(i) Independent inversions:**

Inversions occur in different regions of the chromosome and they are separated from one another by un-inverted (normal) segment

- **(ii) Direct tandem inversions:**

There are two or more inverted segments which are directly adjacent to each other, i.e., the inverted regions are not separated by normal regions

- **(iii) Reversed tandem inversions:**

The two inverted segments are adjacent to each other but their positions are mutually interchanged i.e., g. the first segment lies in place of the second and vice-versa

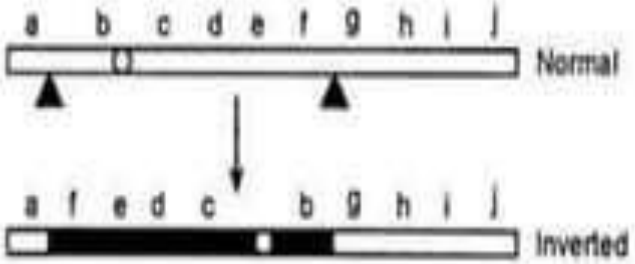
- **(iv) Included inversions:**

One inversion is confined within another inversion, i.e., a segment within an inverted segment is inverted again; as a result, the second inverted segment possesses the normal gene sequence for the concerned segment

- **(v) Overlapping inversions:**

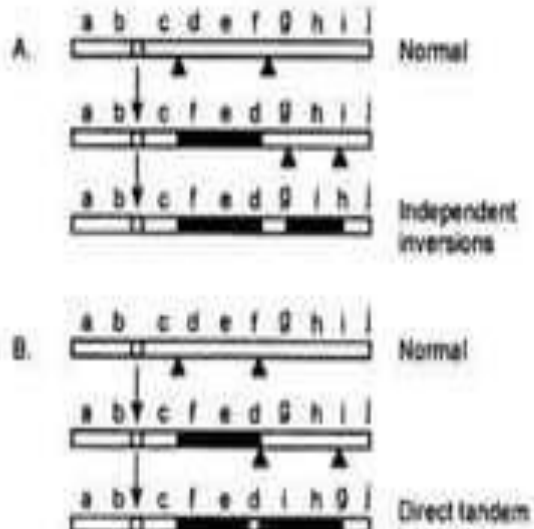
Such inversions have a common segment, i.e., a part of an inverted chromosome segment is inverted again together with an adjacent segment which was not included in the first inverted segment

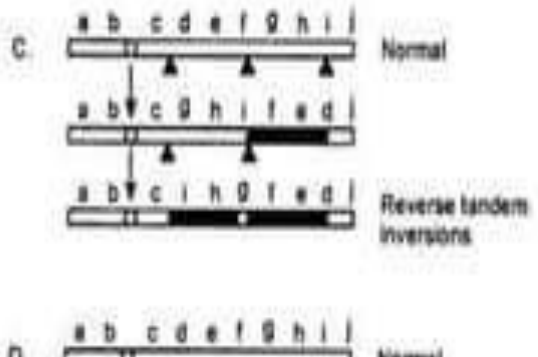
- **Single Inversions**

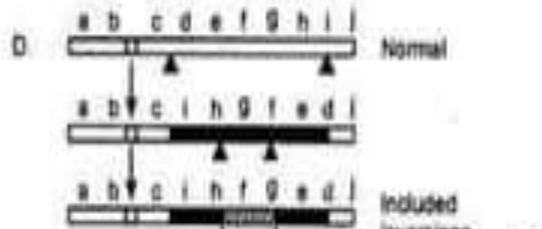


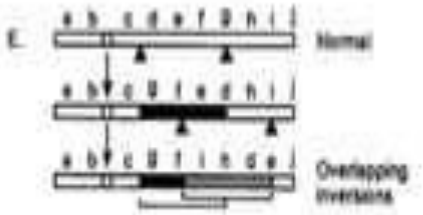
only one segment is inverted

Complex inversions









EFFECTS OF INVERSION

- Helps to retain the original gene combination by reducing the crossing over frequency
- Inversion enhances the position effect & there by produces phenotypic changes
- Inversion causes chromosome polymorphism with in a population leading to karyotype evolution
- Inversion produces balanced gene complexes(super genes) through reduction of crossing over, which in turn confers greater biological fitness and adaptability & increases the evolutionary potentiality

(4.)TRANSLOCATION

- Translocation is the re-organization of chromosomes through the transfer or change in position of chromosome segments with out gain or lose of genes

(i)Intrachromosomal translocation {chromosomal shift} -Change in position of a segment within a chromosome, either from one arm to the other or from one location to another in the same arm

(a)Intraradial- Translocation from one location to another in the same arm

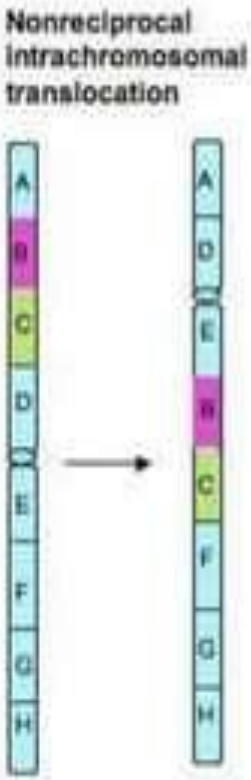
(b)Extraradial- Translocation from one arm to the other

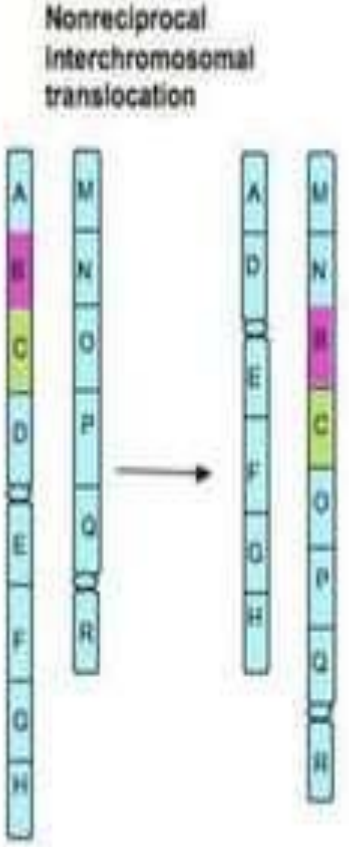
(ii) Interchromosomal Translocation– Transfer of a segment from one chromosome to other

(a) Transpositional translocation- one way transfer of a chromosome segment from one portion to another in the same chromosome

(b) Reciprocal Translocation -mutual exchange of segments between two chromosomes

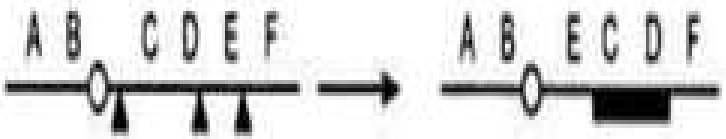
- (i)Intrachromosomal translocation (ii) Interchromosomal Translocation





(i)Intrachromosomal translocation

- **(a)Intraradial Translocation**



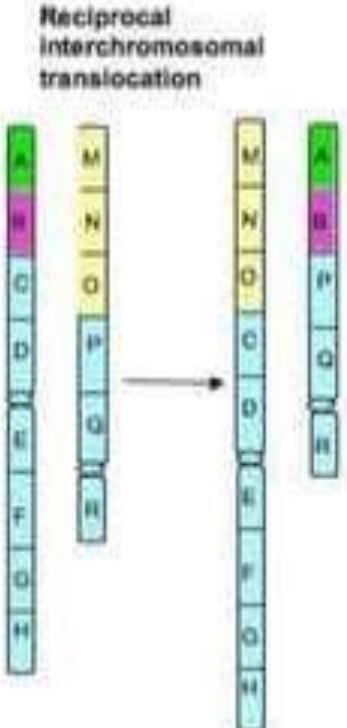
Translocation from one location to another in the same arm

(b)Extraradial Translocation

Translocation from one arm to the other

(ii) Interchromosomal Translocation

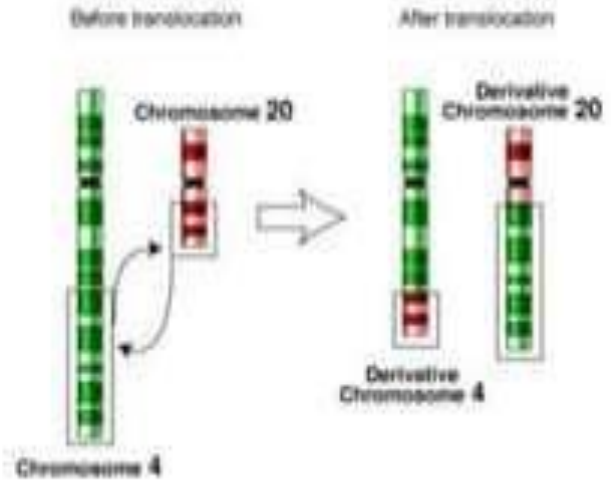
- **Reciprocal Translocation**



Involves two chromosomes

One break in each chromosome

The two chromosomes exchange broken segments

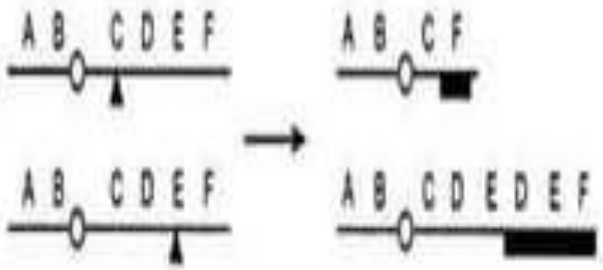


Translocation cont.

1. **Fraternal translocation-** mutual exchange between homologous chromosomes
2. **External translocation-** mutual exchange between non-homologous chromosomes
3. **Intercalation or insertional translocation-** intercalary incorporation of a chromosome segment by transposition
4. **Asymmetrical or aneucentric translocation-** Exchanged segments are asymmetrical , produces a dicentric & an acentric chromosome
5. **Symmetrical or Eucentric translocation-** Exchanged segments are symmetrical , produces two monocentric chromosomes

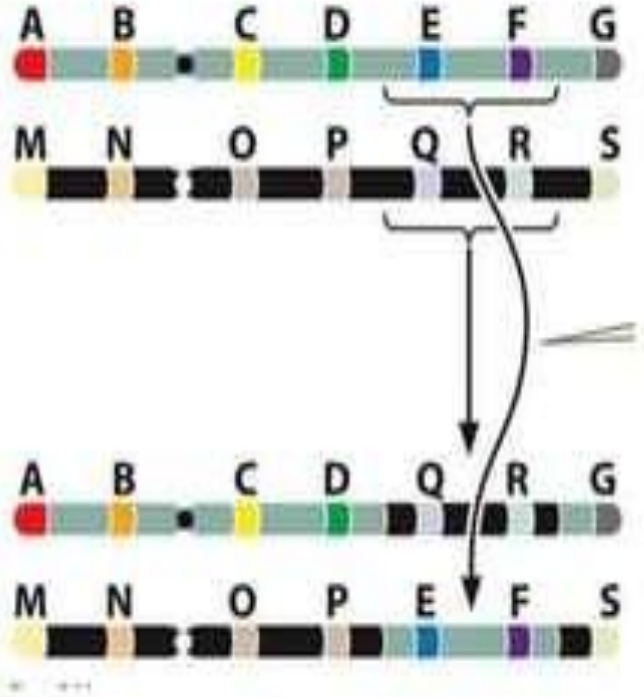
Reciprocal Translocation

Fraternal translocation



Segments are exchanged between homologues

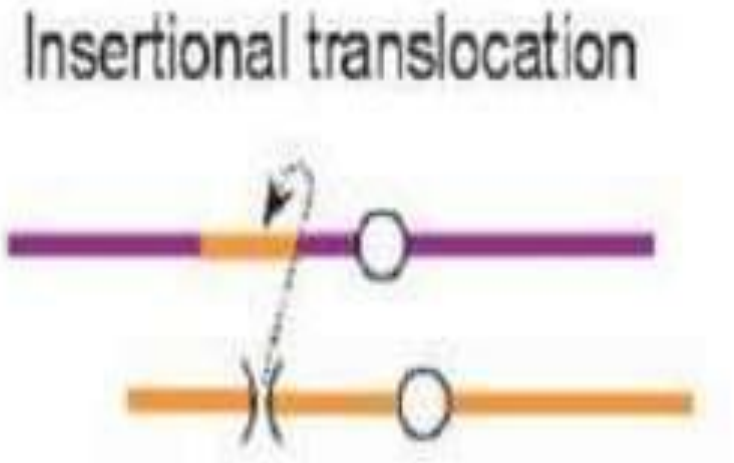
External translocation



Segments are exchanged between non -homologues

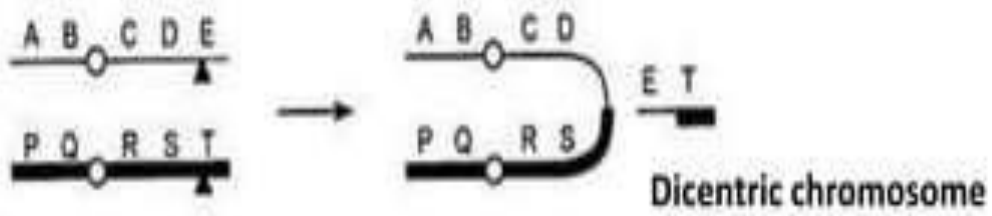
Translocation cont.

- **Intercalation or insertional translocation**
- In the insertional translocation, a segment from the orange (donor) chromosome is transferred to the purple (recipient) chromosome without any reciprocal exchange.

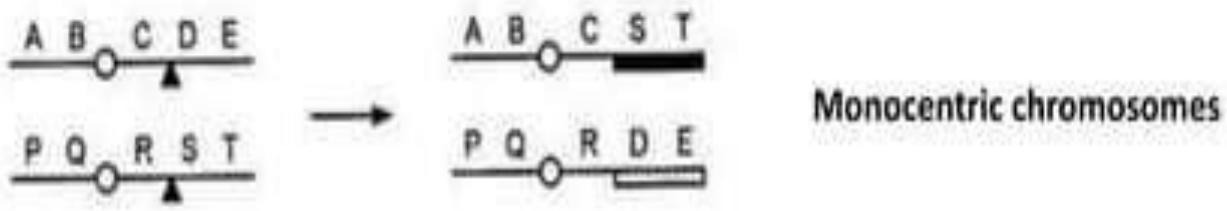


Reciprocal Translocation in monocentric chromosomes

- **Asymmetrical or aneucentric translocation**



- **Symmetrical or Eucentric translocation**



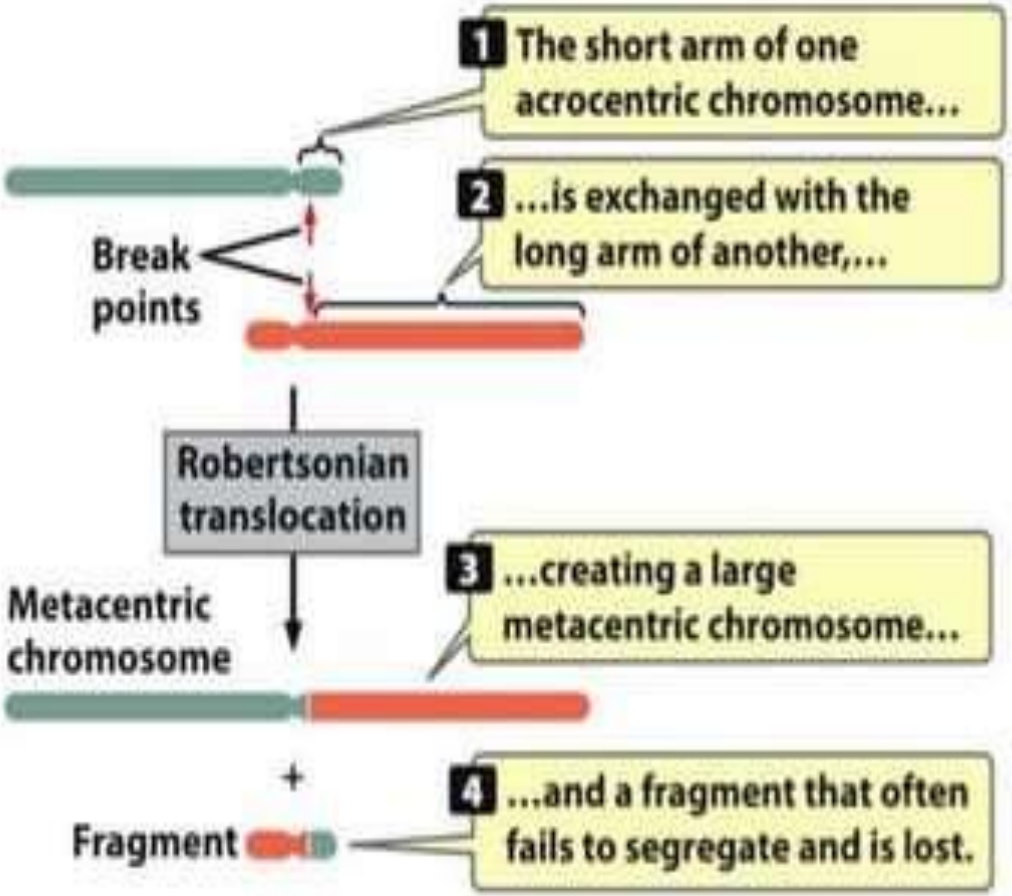
Whole arm translocations

- Translocation which involve the transfer of a whole chromosome arm

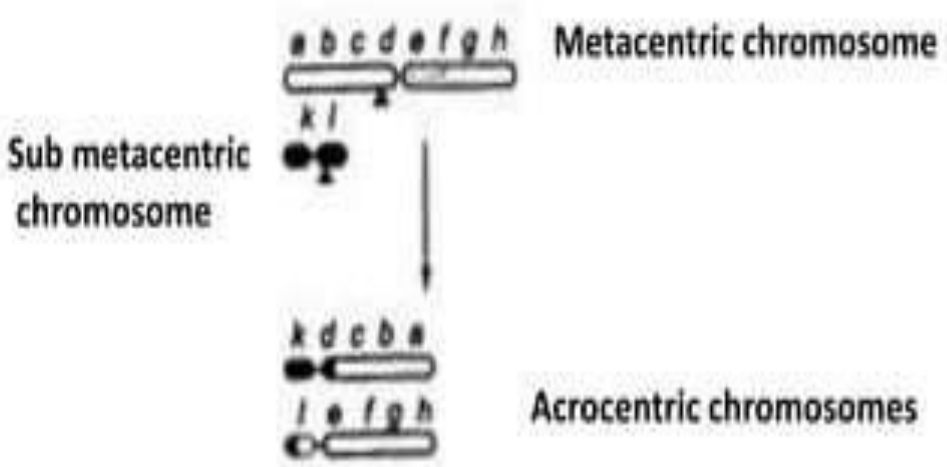
They take place by

- **Centric fusion-** Two breaks in two acrocentric chromosome. One break is in the short arm of a chromosome and the other one is in the long arm of the other chromosome. Their subsequent fusion results in a metacentric chromosome
- **Dissociation**- It is the reverse process in which a metacentric chromosome and a sub metacentric chromosome give rise to two acrocentric chromosomes
- **Tandem fusion** – it is the end to end fusion between an acrocentric chromosomes and a metacentric chromosome with the loss of a centric segment It produces a double length acrocentric chromosome.

Centric fusion



Dissociation



- a metacentric chromosome and a sub metacentric chromosome give rise to two acrocentric chromosomes

- In this process an acentric segment is transferred from the former to the later

Tandem fusion

Metacentric chromosome

Acrocentric chromosome

Double length acrocentric chromosome

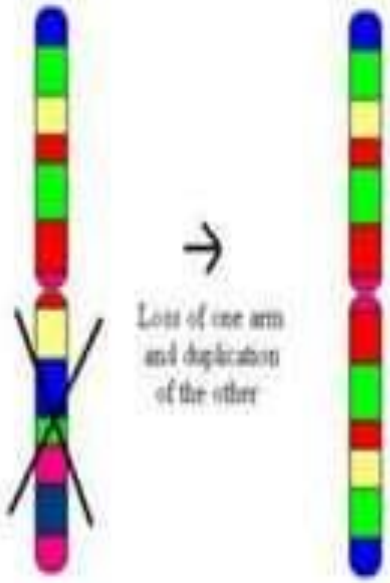
- end to end fusion between an acrocentric chromosomes and a metacentric chromosome with the loss of a centric segment
- Produces a double length acrocentric chromosome

EFFECTS OF DUPLICATION

- Traanslocations & inversions change the linkage relationships of the genes involved in the affected segments
- Translocation results in unusual linkage, reduced meiotic recombination, low viability of chromosomes, familial patterns of trisomy
- It may causes changes in chromosomes & also in gene combinations, promoting variation, specialtion, & evolution

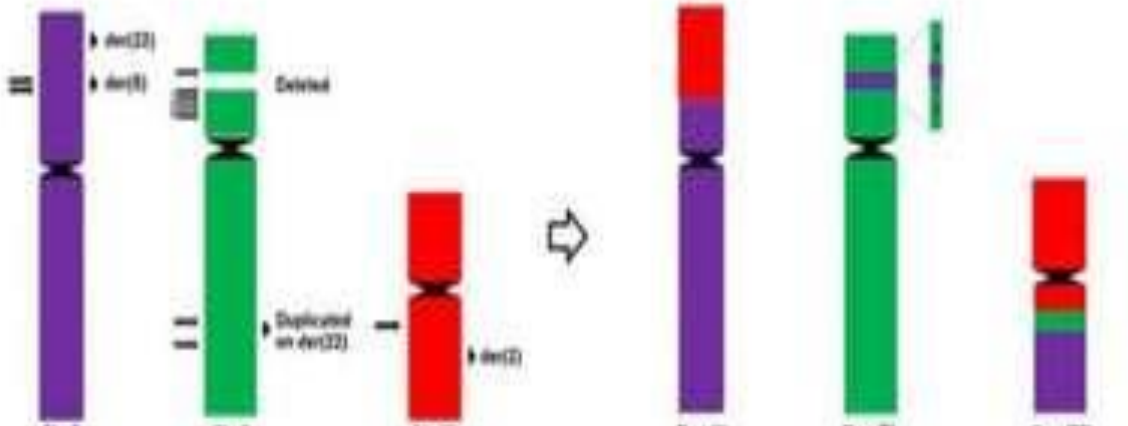
Isochromosome

- An **isochromosome** is an unbalanced structural abnormality in which the arms of the **chromosome** are mirror images of each other. The chromosome consists of two copies of either the long (q) arm or the short (p) arm because isochromosome formation is equivalent to a simultaneous duplication and deletion of genetic material.



Derivative chromosome

- A **derivative chromosome** is a structurally rearranged **chromosome** generated either by a rearrangement involving two or more **chromosomes** or by multiple aberrations within a single **chromosome** (e.g. an inversion and a deletion of the same **chromosome**, or deletions in both arms of a single **chromosome**)

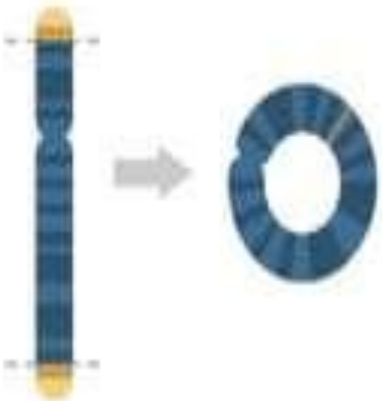


Ring chromosomes

- A ring chromosome is a chromosome whose arms are fused together to form a ring.

or

- A structurally abnormal chromosome in which the end of each chromosome arm has been lost and the broken arms have been reunited to form a ring



I'm through

THANKYOU.