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Lecture – 10

Structural change in chromosome

by

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WHAT IS CHROMOSOMAL ABERRATIONS

- ❖ A chromosomal disorder occurs when there is a change in the number or structure of the chromosomes.
- ❖ This change in the amount, or arrangement of, the genetic information in the cells may result in problems in growth, development and/or functioning of the body systems.
- ❖ Main feature to identify and classify chromosomes
 1. Size
 2. Location of the centromere
 3. Banding patterns



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CHROMOSOMAL ABERRATIONS

Chromosomal Aberrations

Numerical

aneuploidy
polyploidy

structural

deletion
duplication
inversion
translocation



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Chromosome Structural aberrations

- ❖ The chromosome aberrations in which they alters the structure of chromosome (sequence of genes or kind of genes in chromosome or no. of genes)
- ❖ There are mainly four types which are
 1. **Changes in the numbers of genes**
 - (a) **Deletion/Deficiency**
 - (b) **Duplication**
 2. **Changes in the location of genes**
 - (a) **Inversions :- 180° rotation**
 - (b) **Translocations:- Exchange**



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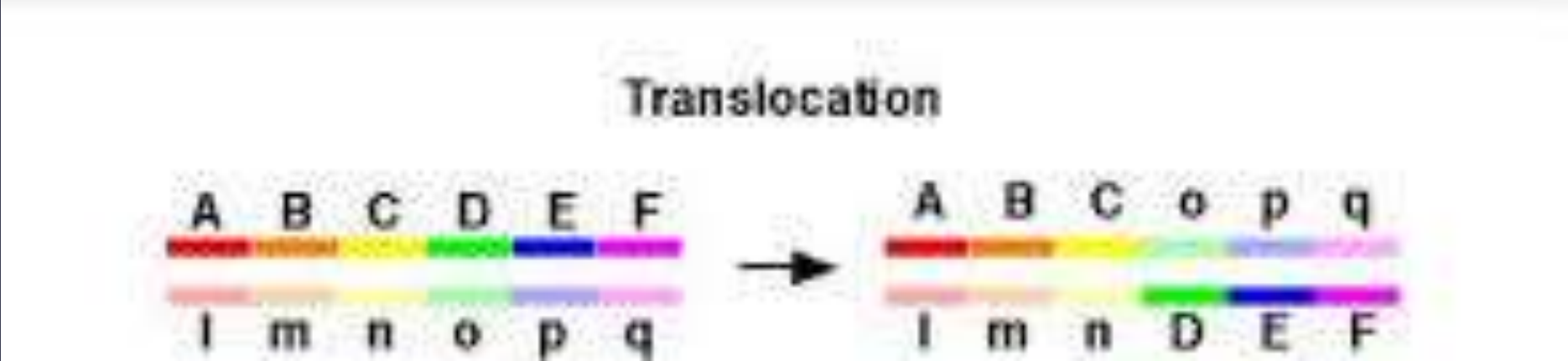
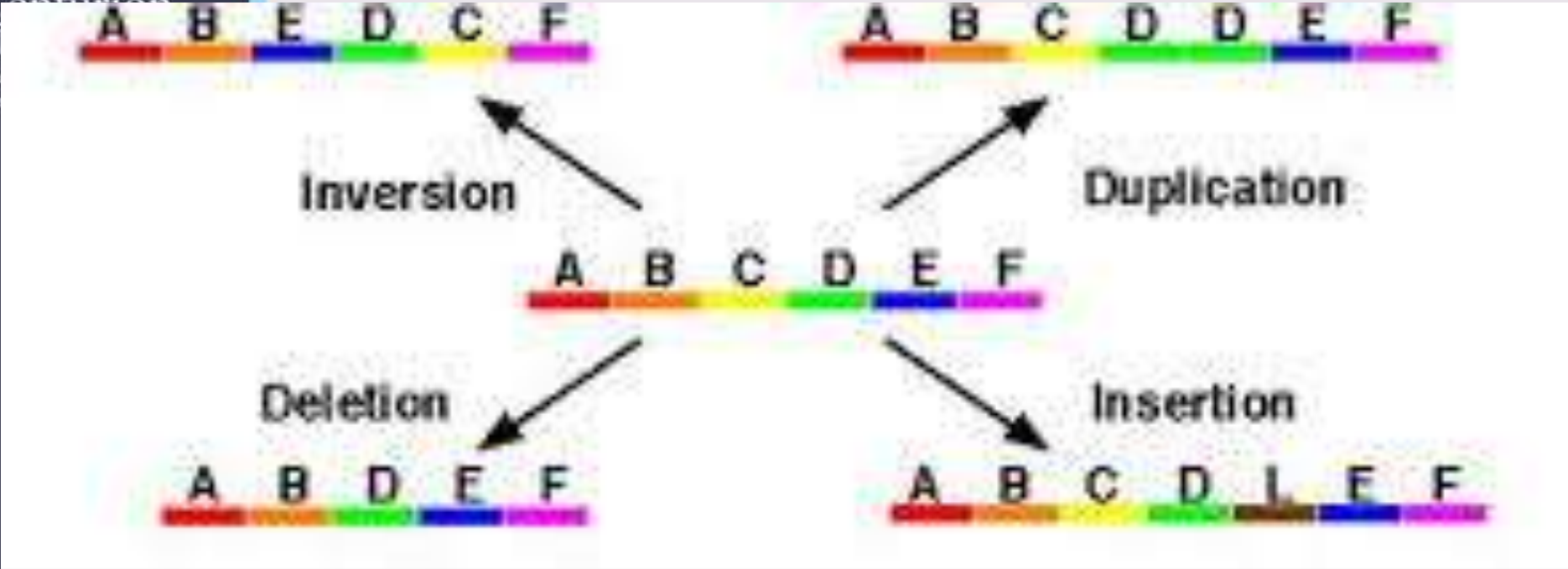
Types of structural chromosomal aberrations:

These aberrations may be confined to a single chromosome or more than one chromosome. Hence, they are of two types.

- I. **Intra -chromosomal aberrations:** When aberrations remain confined to a single chromosome of a homologous pair, they are called intra -chromosomal aberrations.
- I. **Inter-chromosomal aberrations:** When breaks occur in non-homologous chromosomes and the resulting fragments are inter-changed by both the nonhomologous chromosomes, they are known as inter-chromosomal aberrations.



Structural chromosomal aberrations



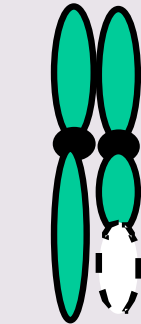


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Chromosome Structure Abnormalities



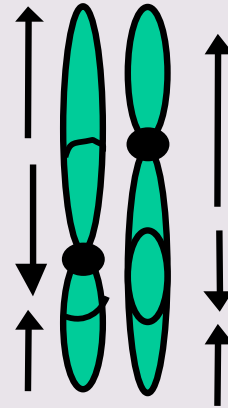
Translocation



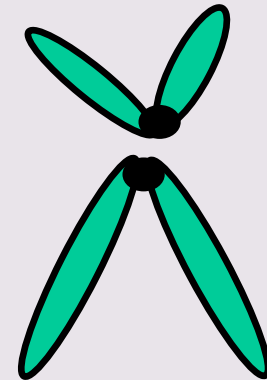
Deletion



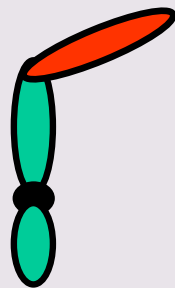
Insertion



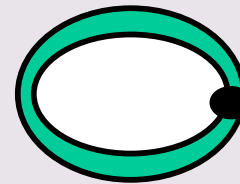
Inversion



Isochromosome



Derivative
chromosome



Ring
chromosome



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1. Deletion/ deficiency

- First chromosome aberration discovered by the Bridges in 1917 in X- chromosome of Drosophila
- Loss of a (generally small) segment of chromosome
- Usually detected by based on unpaired region of normal chromosome produces a loop during pachytene stage

Source of deletion

1. Natural deficiency
2. Small repetitive DNA segment may have capacity to get deleted and moving to other chromosome.
3. Segregation product of pericentric inversion
4. Induction through physical and chemical mutagens



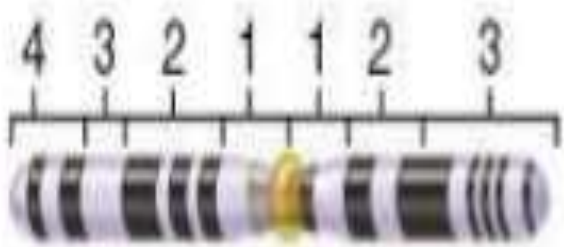
Types

Terminal deletion: If a break occurs near the end of chromosome and a small piece of terminal chromosome is lost, it is called terminal deletion.

- In 1938 Muller postulated that loss of telomere makes chromosome unstable so it is not commonly observed Ex: maize

Interstitial or intercalary deletion: Sometimes two breaks may occur at any two points and the broken ends of the original chromosome get fused or reunited and as a result, an interstitial deletion is formed.

- It is commonly observed Ex: Drosophila
- If the chromosome has a centromere, it will persist. Otherwise it will be lost during cell division.

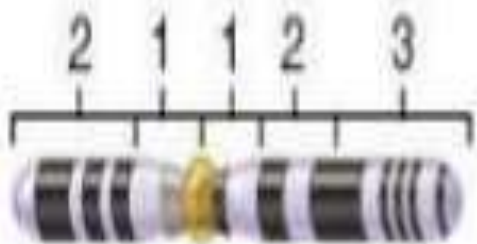


Single break



(Lost and degraded)

+



(a) Terminal deficiency



Two breaks and reattachment of outer pieces



(Lost and degraded)

+



(b) Interstitial deficiency



Cytological detection of deletion

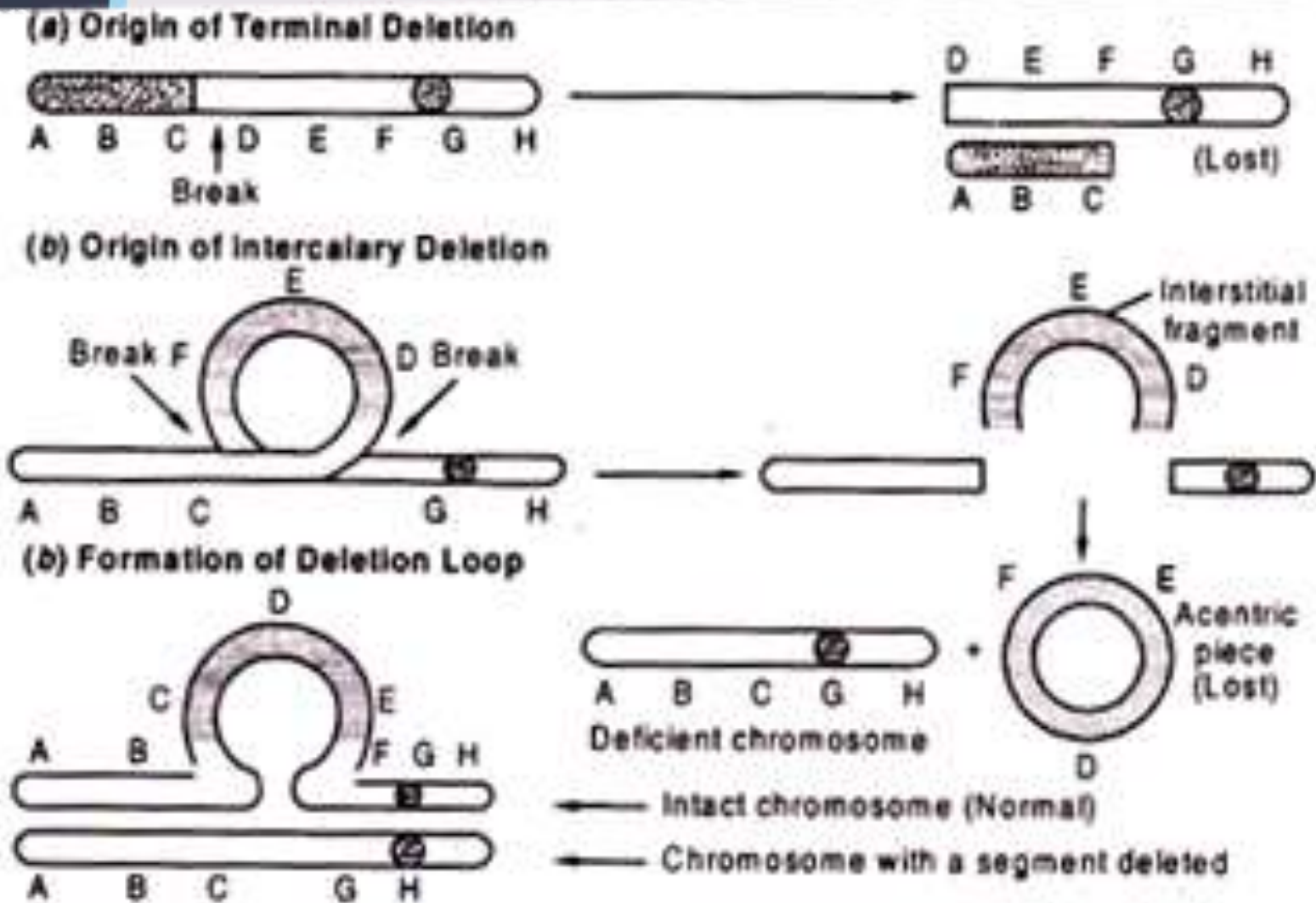


Fig. 22.3 Terminal (a), and interstitial (b) deletions, (c) Formation of loop in deletion heterozygote.



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Genetic significance / effects of deletions:

1. Organisms with homozygous deletion do not survive to an adult stage because a complete set of genes is lacking (**lethal effect**).
2. Pseudo dominance e.g.. in *Drosophila*, maize and other organisms
3. Crossing over not there
4. Harmful effect on diploid organisms
5. Most are as recessive Lethals
6. Morphological effects
7. Deletions play an important role in species formation and creating variability through chromosomal mutations.



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Deletion in human

Produces distinct sets of symptoms i.e. **SYNDROM**

1. **Chronic myelogenous leukemia:** deletion in chromosome 22 (Philadelphia 22)
2. **Cri-du-chat (cry of cat) syndrome:** deficiency in small arm of chromosome 5
3. **Wilm's tumor:** deficiency of 7th and 14th autosome.
4. **Angelman Syndrome:** deletion in chromosome 15. uncontrolled laughter, no speech development



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2. Duplication/ Repeat

- ❖ Duplication was first reported in *Drosophila* by C.B. Bridges in 1919.
- ❖ Duplication occurs when a segment of chromosome is represented two or more times in a chromosome of a homologous pair.
- ❖ The extra segment may be a free fragment with a centromere or a chromosomal segment of the normal complement. As a result, in one chromosome of the homologous pair, there will be deletion, while in other there will be a duplication



Source of duplication

Naturally

- A chromosomal duplication is usually caused by abnormal events during recombination

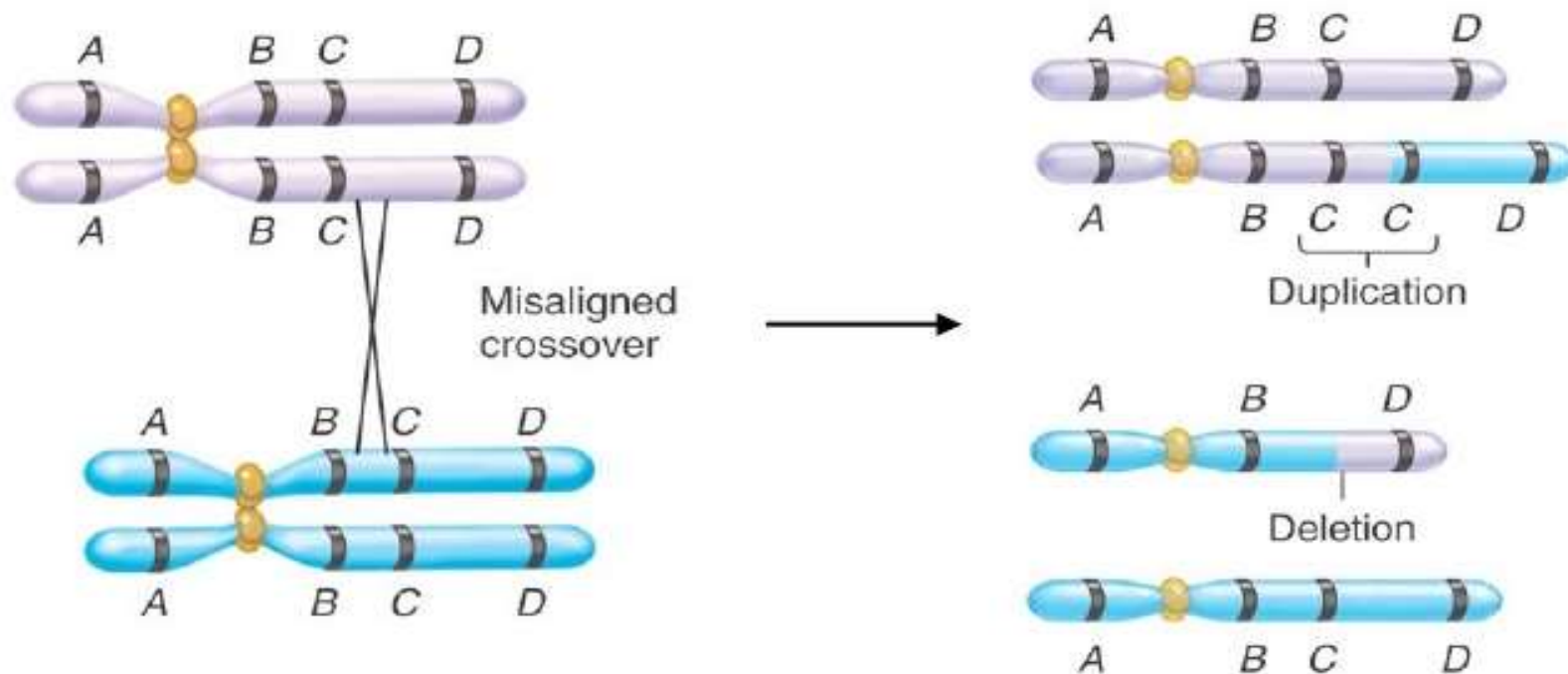


Figure 8.5



Mainly four types

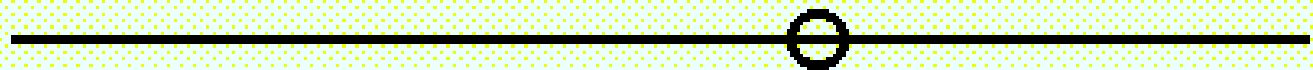
Normal chromosome

A B C D E F G H



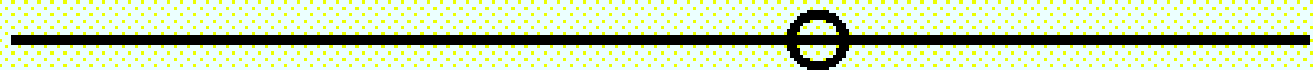
Tandem duplication

A B C D B C D E F G H



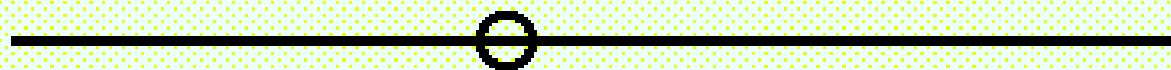
Reverse tandem duplication

A B C D D C B E F G H



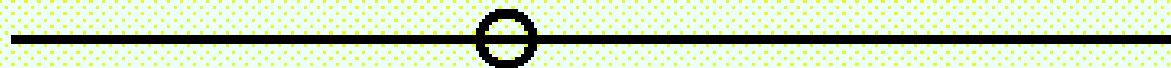
Duplication on a different arm

A B C D E B C D F G H



Non-homologous (displaced) duplication

L M N O P Q B C D R S





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Cytological detection of duplication

- It is detected by presence of loop formation during pachytene stage in duplicated region or cross pairing between segments of non-homologous chromosomes.

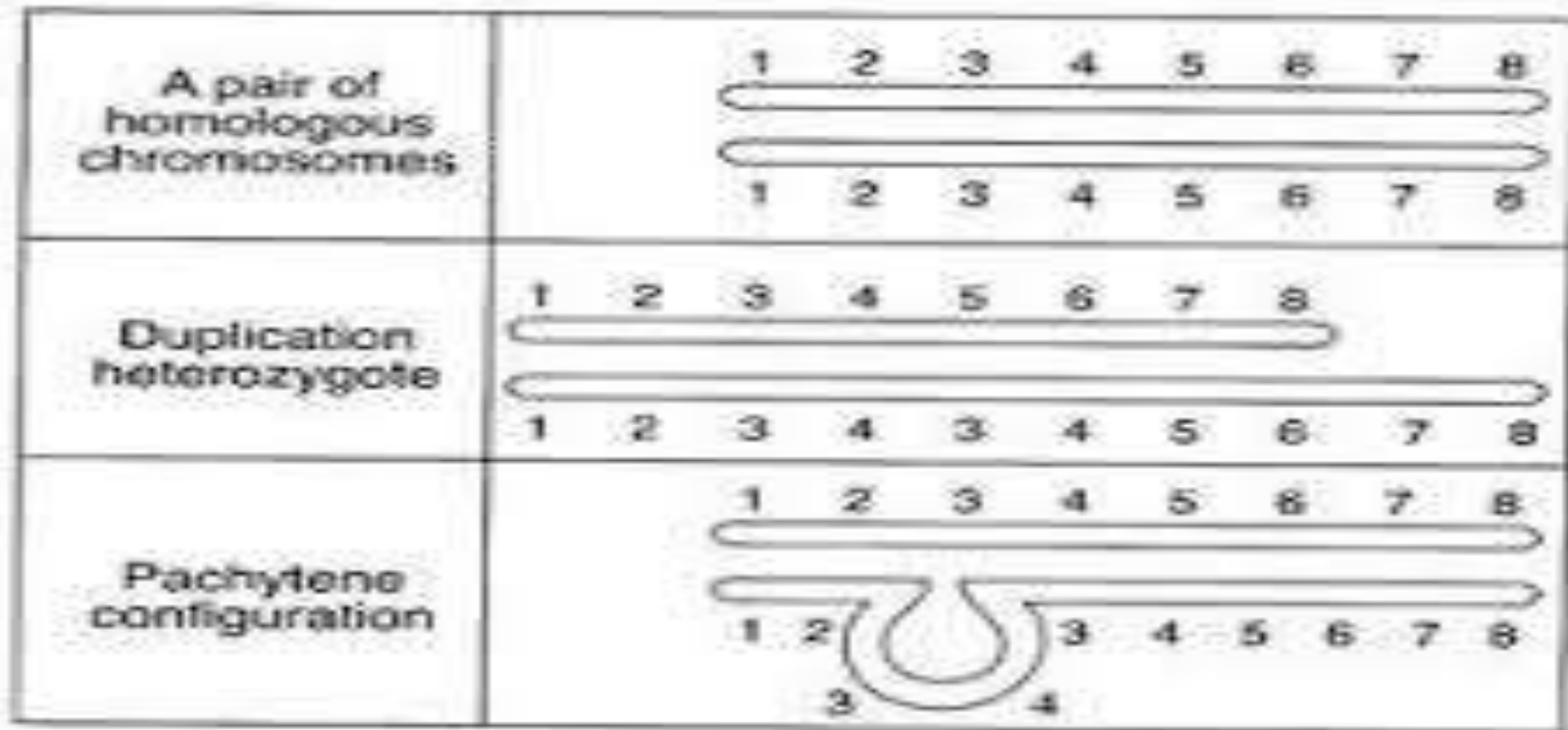


Fig. 12.7: Chromosome pairing in a duplication heterozygote



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Genetic significance / effects of duplications:

1. Duplications are not as harmful as deletions.
2. According to Ohno(1970) origin of new genes mainly due to duplication resulted the evolution
3. Large duplications can reduce fertility.
4. **Position effect.** Ex: Bar eye is result of duplication of 16A region of X chromosome of Drosophila
5. Assume multiple factors (polygenes) have originated
6. Activity of certain enzymes may be doubled
Ex:chromosome 6 of barley increased the activity of α -amylase (hagberg in 1965)



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3. Inversion

- ❖ It is the structural aberration in which the change in the sequence of chromosomal arrangement
- ❖ Two breaks in one chromosome
- ❖ The fragment generated rotates 180° and reinserts into the chromosome
- ❖ No loss or gain of genetic materials involved.
- ❖ First observed by Sturtevant & Plunkett (1926) by comparing linkage map in *Drosophila melanogaster* (*Se, St, P, DI, H, Ca*) and *D. simulans* (*Se, St, H, DI, P, Ca*) at chromosome no. 3

Origin

A/C to definition inverted segment rotate by 180° involving two break and reunion of interstitial segments.



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Detected by

- ❖ **Phenotypically Detection:** Inversion heterozygote exhibit partial male sterility
- ❖ **Genetically Detection:** comparing with the normal strain by using linkage map
- ❖ **Cytologically Detection:** presence of inversion loop during pachytene



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Types of inversion

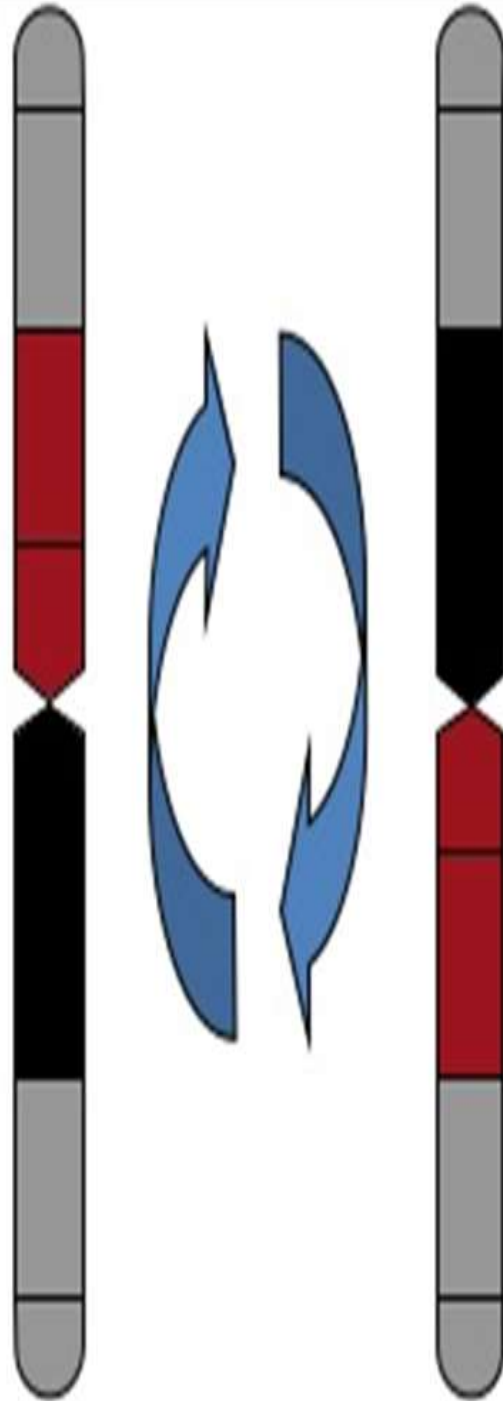
- **Paracentric:-** inverted segment does not include centromere and confined to one arm.
- **Pericentric:-** included the centromere

However, Paracentric don't alter the morphology of chromosome but Pericentric do so.

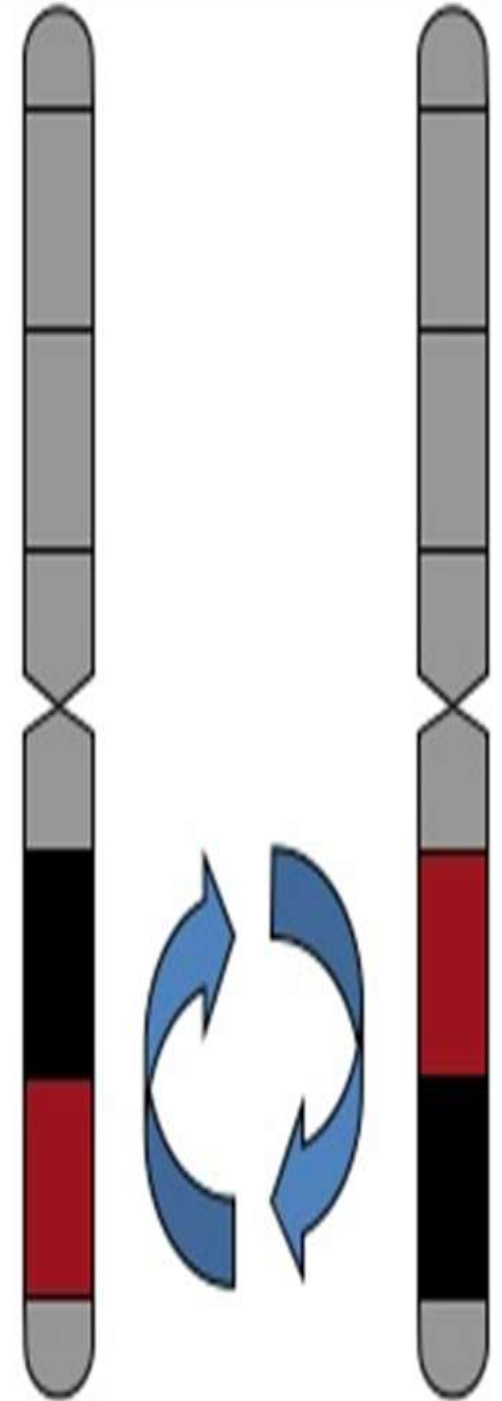
- ❖ **Overlapping Inversion**
- ❖ **Adjacent Inversion**

chromosome

Pericentric -
involves p and q
arm



Paracentric -
involves only
one arm

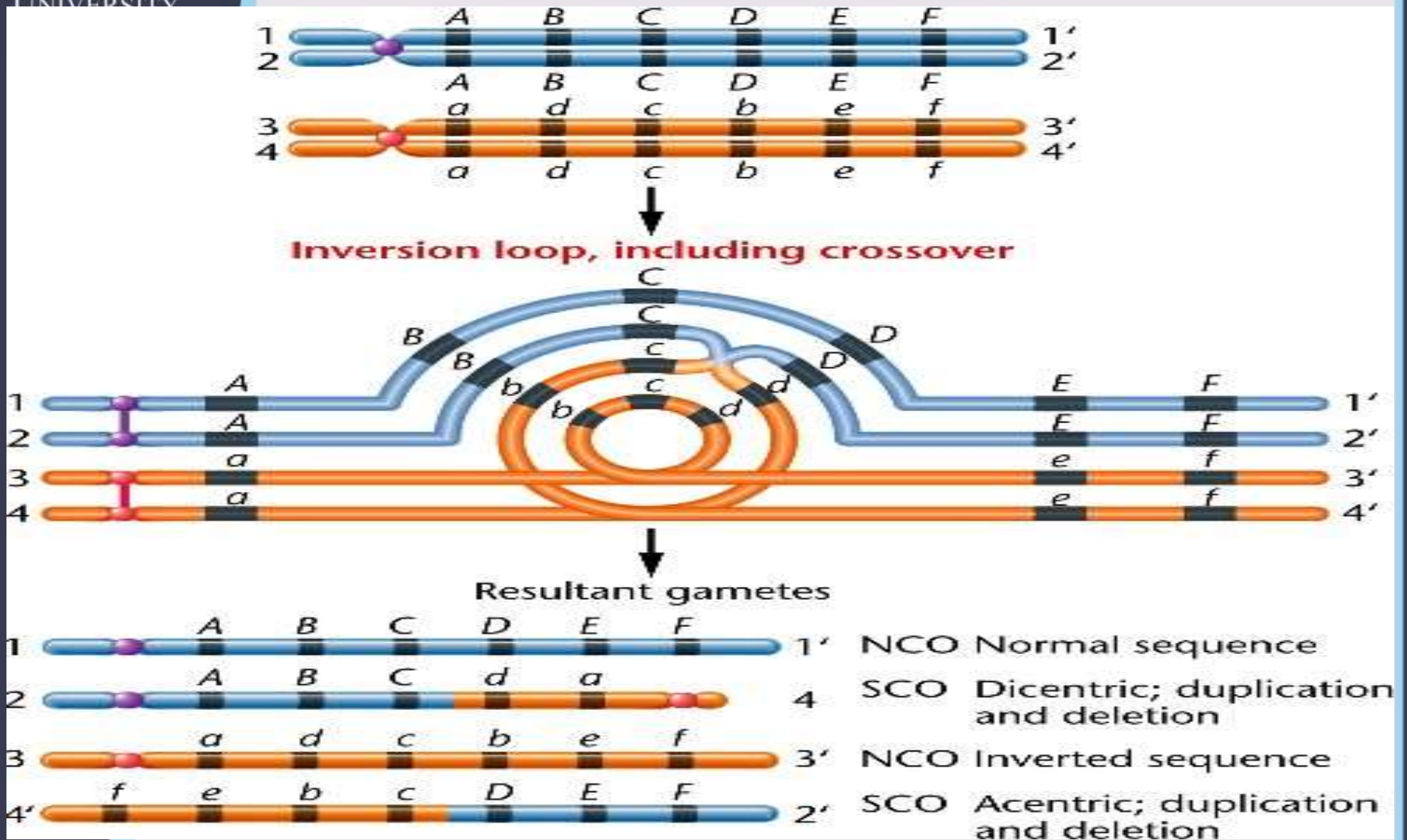




Cytological behavior of paracentric inversion

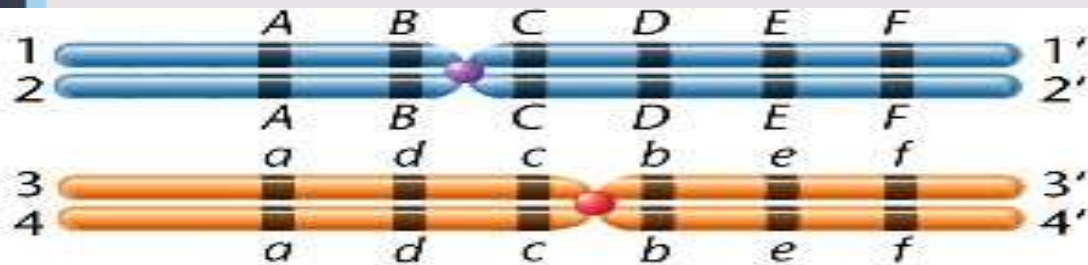
1. Paracentric Inversion- crossover suppressor (C)

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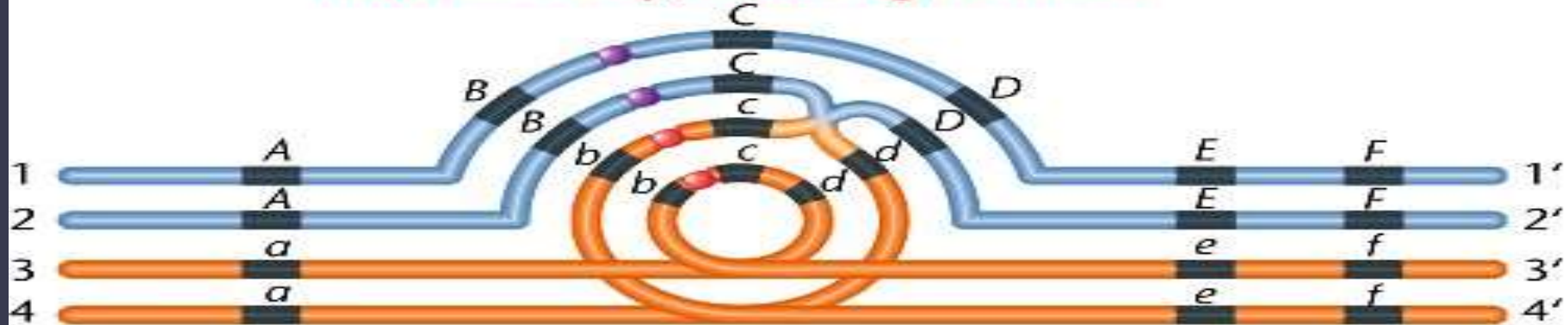




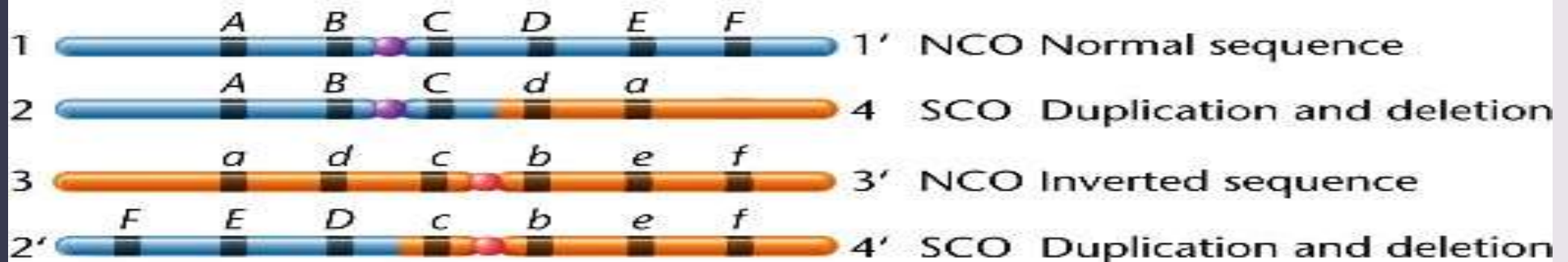
2. Pericentric Inversion-



Inversion loop, including crossover



Resultant gametes





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Comparative summary of cytological behavior of C.O. at pachytene in Paracentric and Pericentric inversion in higher plant and animals

Types of Inversion	Cytological C.O. in inverted region
1. Pericentric	1. No acentric fragments and dicentric bridge observed 2. Crossover chromatids have duplication and deficiency
2. Paracentric	1. acentric fragments and dicentric bridge observed 2. Crossover chromatids have duplication and deficiency



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Genetic significance / effects of inversion

- ✓ Partial male sterility
- ✓ Formation of Recessive mutation
- ✓ Position effect
- ✓ move active genes to sites generally inactive; lose gene function and vice versa
- ✓ May change in the Karyotype symmetry and morphology of chromosome.
- ✓ Available evidence indicates that evolution of human X and Y chromosome from a pair of autosome was due to inversion.



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4. Translocation

- ❖ Integration of a chromosome segment into a non – homologous chromosome is called translocation.
- ❖ It involves shifting of one part of chromosome to another non homologous chromosome.
- ❖ The phenomenon of translocation was discovered by C.B. Bridges in 1923 in *Drosophila* and by Hugo de Vries in *Oenothera lamarckiana*.



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Types

Based on number of breaks involved

1. Simple translocation

- In simple translocation, the terminal segment of chromosomes is integrated at one end of a non-homologous chromosome. However, they are rare.
- It involves one break
- Reported by painter and muller in 1929 in *Drosophila*

2. Shift translocation

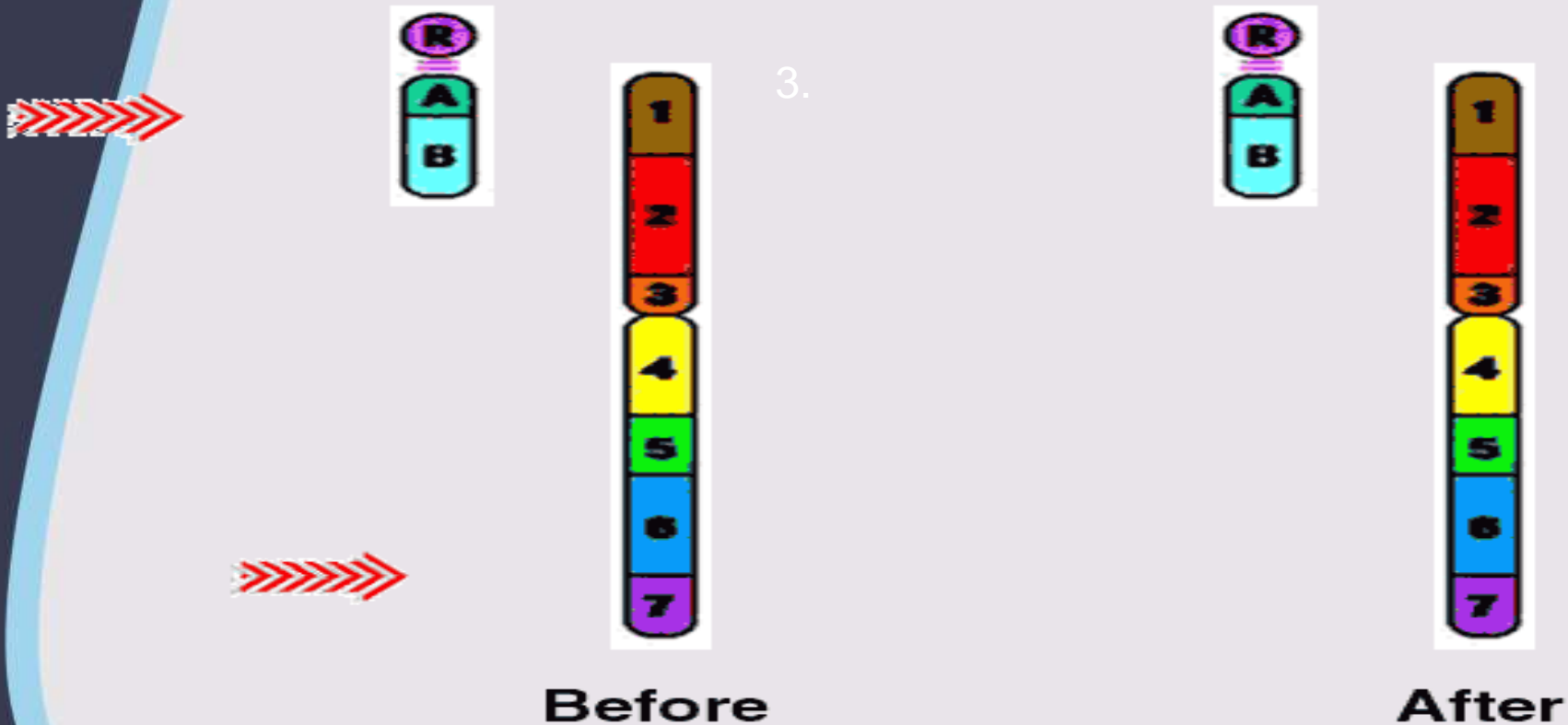
- It requires three breaks
- Intercalary segment of a chromosome is integrated within a non-homologous chromosome
- Commonly seen in *Drosophila*, *Neurospora*



3. Reciprocal translocations

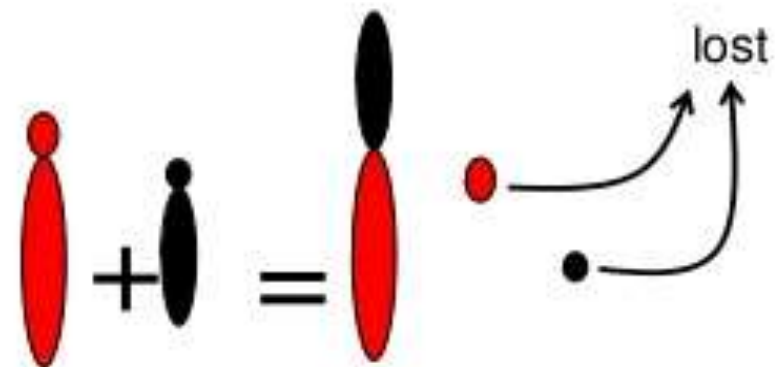
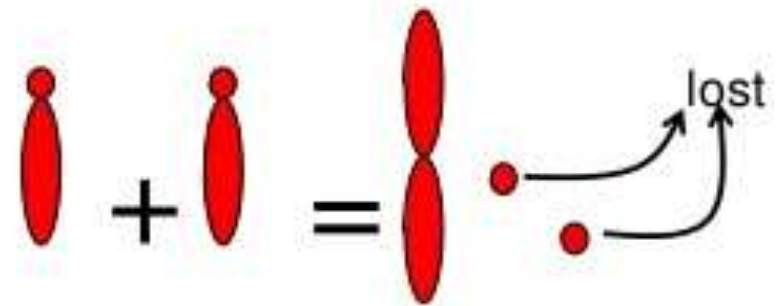
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- It requires one break in each of the chromosomes
- Segments will be exchanged between non homologous chromosomes
- Designated by prefix 'T'



Robertsonian Translocation

- Named after W. R. B. Robertson who first identified them in grasshoppers in 1916
- Most common structural chromosome abnormality in humans
 - Frequency = 1/1000 livebirths
- Involves two acrocentric chromosomes
- Two types
 - Homologous acrocentrics involved
 - Non-Homologous acrocentrics involved





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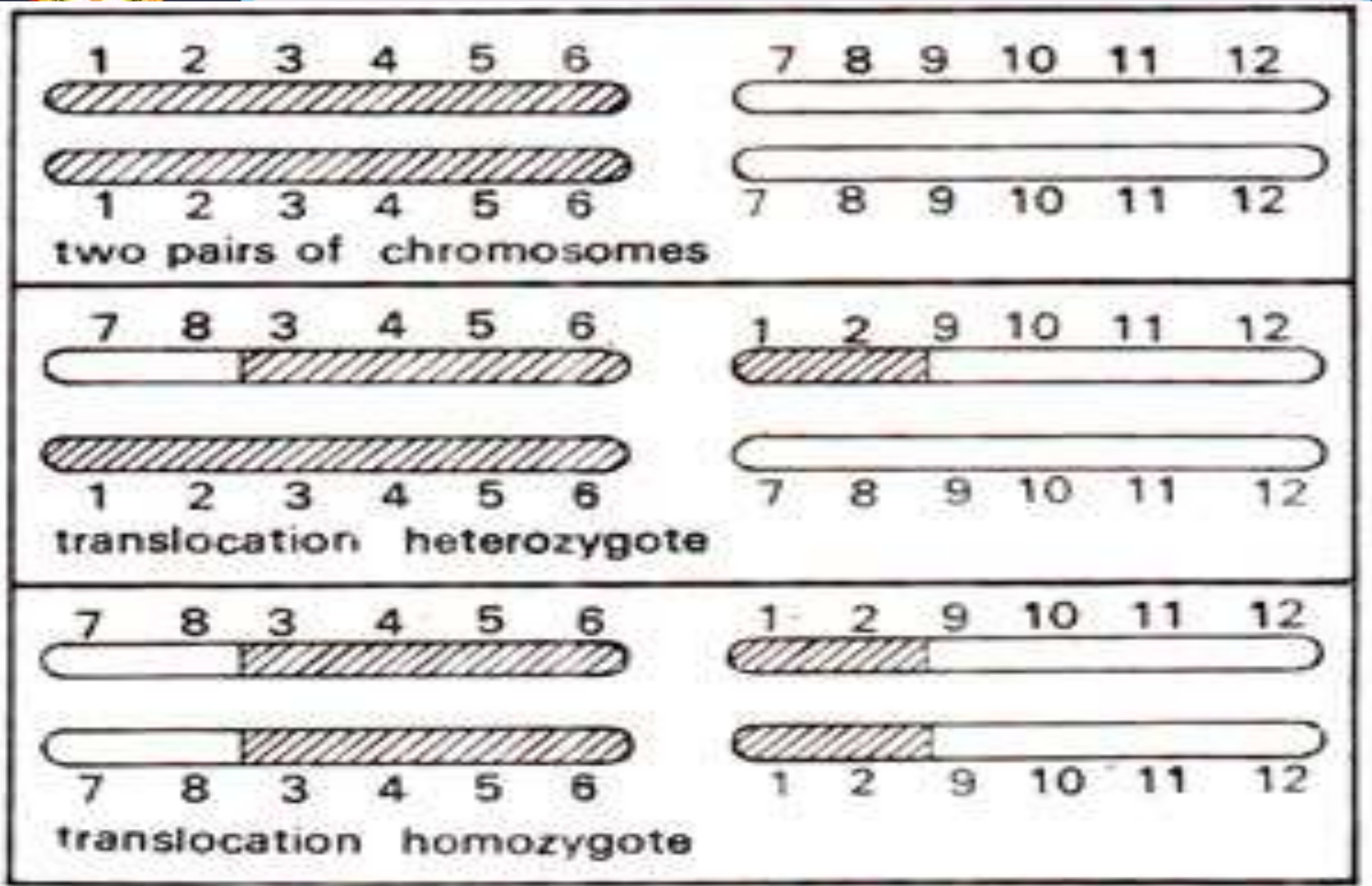
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Cytological behavior of translocation

- Alters the chromosome morphology
 - Position effect
 - Damage to DNA may result in formation of recessive lethals
 - Lead to impaired fertility
 - complications to synopsis and segregation



Translocation homozygote and heterozygote





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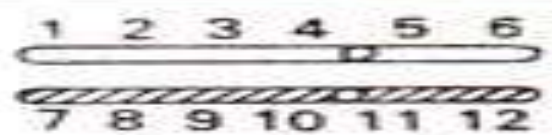
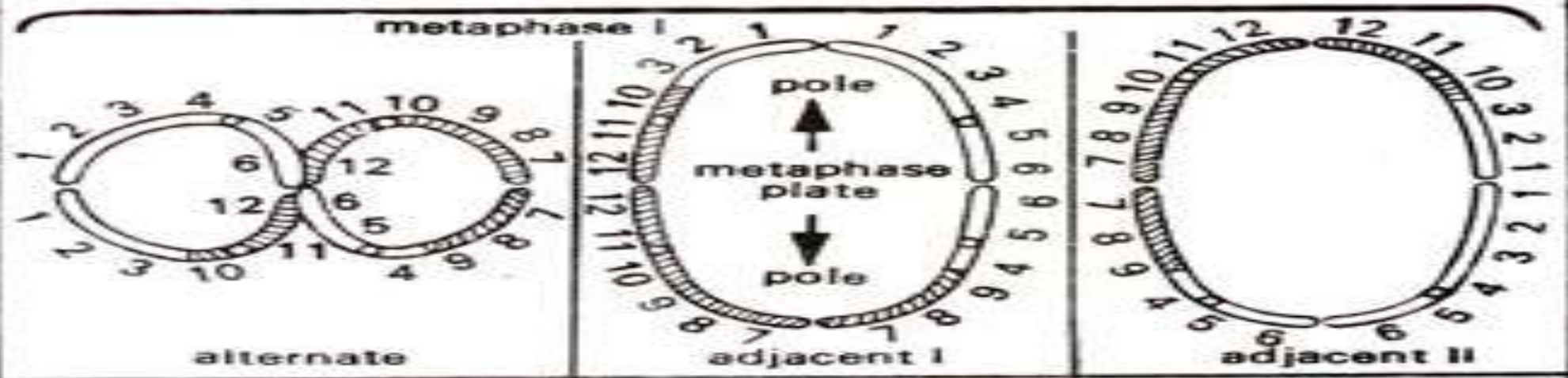
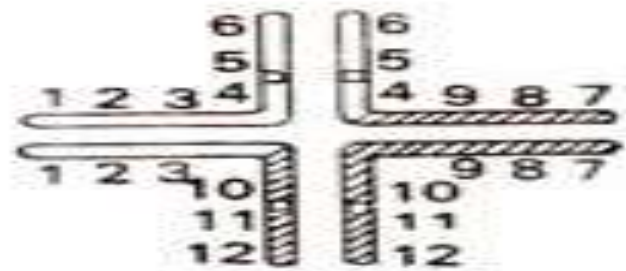
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- ❖ In a translocation homozygote, the two homologues of each of the two translocated chromosomes are identical in their gene content. As a result, they form normal bivalent and there is no detectable cytogenetic aberration (peculiarity).
- ❖ In a translocation heterozygote, one member from each of two homologous pairs is involved in reciprocal translocation, while the remaining chromosomes of the two concerned pairs are normal. Due to the pairing between homologous segments of chromosomes, a cross-shaped (+) figure involving four chromosomes will be observed at pachytene.
- ❖ These four chromosomes at metaphase I will form a quadrivalent, which may exhibit any one of the following three orientations. (1) Alternate (2). Adjacent I (3) Adjacent II

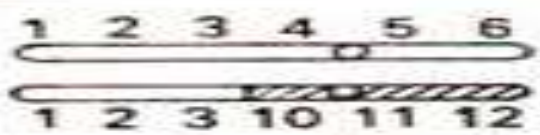


a translocation heterozygote

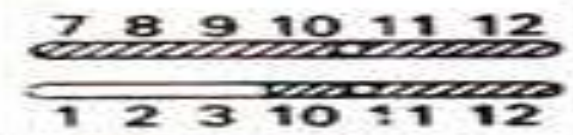
pachytene configuration



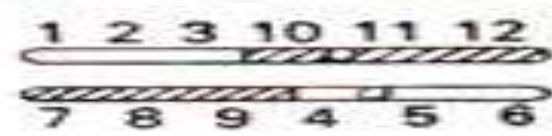
functional



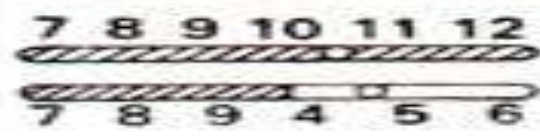
nonfunctional



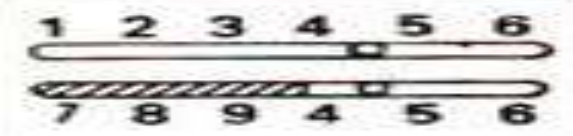
nonfunctional



functional gametes



nonfunctional



nonfunctional



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1. Adjacent-I

- In adjacent I orientation, adjacent chromosomes having nonhomologous centromeres will orient towards the same pole. In other words, the chromosomes having homologous centromeres will orient towards the opposite poles.
- Thus, a ring of four chromosomes will be observed.

2. Adjacent – II

- In adjacent II orientation, the adjacent chromosomes having homologous centromeres will orient towards the same pole.
- In this case also a ring of four chromosomes will be observed.

In both Adjacent-I and adjacent-II disjunctions, one normal and one translocated chromosome move to the opposite poles. The gametes produced from such types of disjunction have duplication and deficiency of one or other genes and hence become non-viable.



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3. Alternate

- ❖ In this case, the centromeres lying alternate to each other in the cross shaped figure move to the same pole. In other words, the adjacent chromosomes will orient towards opposite poles.
- ❖ As a result, the two normal chromosomes move to one pole, while the two translocated chromosomes move to the opposite pole.
- ❖ Such a segregation can take place only when the cross shaped figure of four chromosomes is twisted to form a figure of '8'.
- ❖ E.g. *Oenothera sp.* and *Rheo discolor*



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Adjacent-I and adjacent-II disjunctions will form gametes which would carry duplications or deficiencies and as a result would be non-functional or sterile. Therefore, in a plant having translocation in heterozygous condition, there will be considerable pollen sterility.



Genetic significance of translocation heterozygotes

1. They produce semi sterile plants with low seed set.
2. Some genes which earlier assorted independently tend to exhibit linkage relationship.
3. The phenotypic expression of a gene may be modified when it is translocated to a new position in the genome.
4. The presence of translocation heterozygosity can be detected by the occurrence of semi-sterility and low seed set. This can then be confirmed at meiosis by quadrivalent formation. Functional gametes will be formed only from alternate disjunction, which will give rise to three kinds of progeny *viz.*, normal, translocation heterozygotes and translocation homozygotes in 1:2:1 ratio.



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Role of structural chromosomal aberrations in plant breeding

1. They are useful in the identification of chromosomes
2. Utilization of vigour as in case of duplication.
3. Useful in genome analysis.
4. Useful for the transfer of desirable characters through translocation.
5. They have evolutionary significance.
6. Construction of linkage map
7. Evolution of new genes



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