Lecture 15. Chromosomal aberration: Variation in chromosome structure – deletion, duplication, inversion and translocation – genetic and cytological implications.

Any change which **alters the normal structure of a chromosome** is known as **structural chromosome change**.

Such changes are called

Chromosomal mutations OR
Chromosomal aberrations

Structural chromosomal changes

- Takes place both in somatic and germ cells
- During interphase or early prophase
- Occur due to breakage and reunion of chromosome
- Breaks may be due to radiations and chemicals
- Two types of breaks- Restituted and Non restituted
- Restituted – reunion restores the original sequence of genes
- Non restituted – various changes in chromosome structure
- Changes can be detected by pairing of chromosomes at pachytene stage or by pollen sterility
- Alters phenotype, fertility, viability and karyotype of an individual

Type of structural chromosomal aberration

- Deletion or deficiency
- Duplication orAdditions
- Inversion
- Translocation

Types of structural chromosomal aberrations

It may be two types

A. Intrachromosomal aberrations

Alter **gene number** in the chromosome

1. **Deletion**
2. **Duplication**

Alter the **sequence of genes** in the chromosome

1. **Inversion**

B. Interchromosomal aberrations.

Alter the **sequence of genes** in the chromosome

1. **Translocation**

Four types of structural changes occur in the chromosome

A. Intrachromosomal Aberrations

When aberrations remain confined to a **single chromosome of a homologous pair**, they are called

intrachromosomal / homosomal aberrations.

1. **Deficiencies/ Deletion**
2. **Duplication/additions**

1. Deficiencies

Loss of a portion of segment from a chromosome

-Deletion / Deficiency

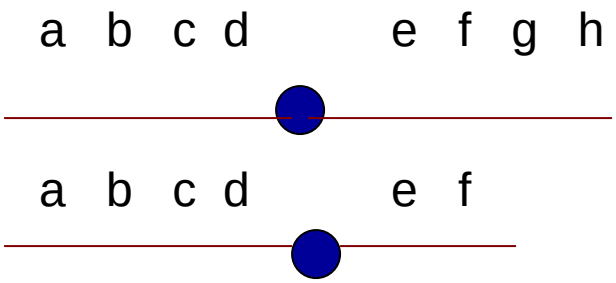
Two types - Terminal deficiency

Intercalary or intersitial deficiency.

1. Terminal deficiency

- **If break occurs near the end of a chromosome, a small piece of the terminal end is lost.**

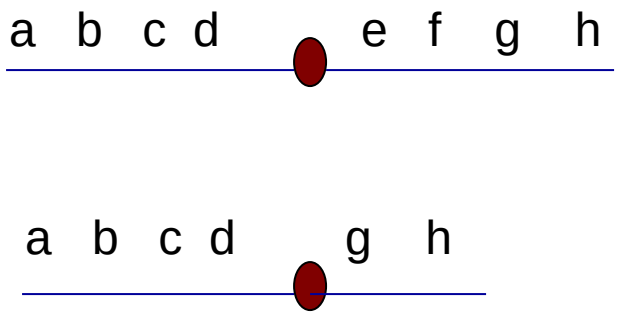
  (The injured end later heals)
- **Two types**
- **Heterozygous – in one chromosome of a homologous pair**
- **Homozygous - in both the chromosomes**



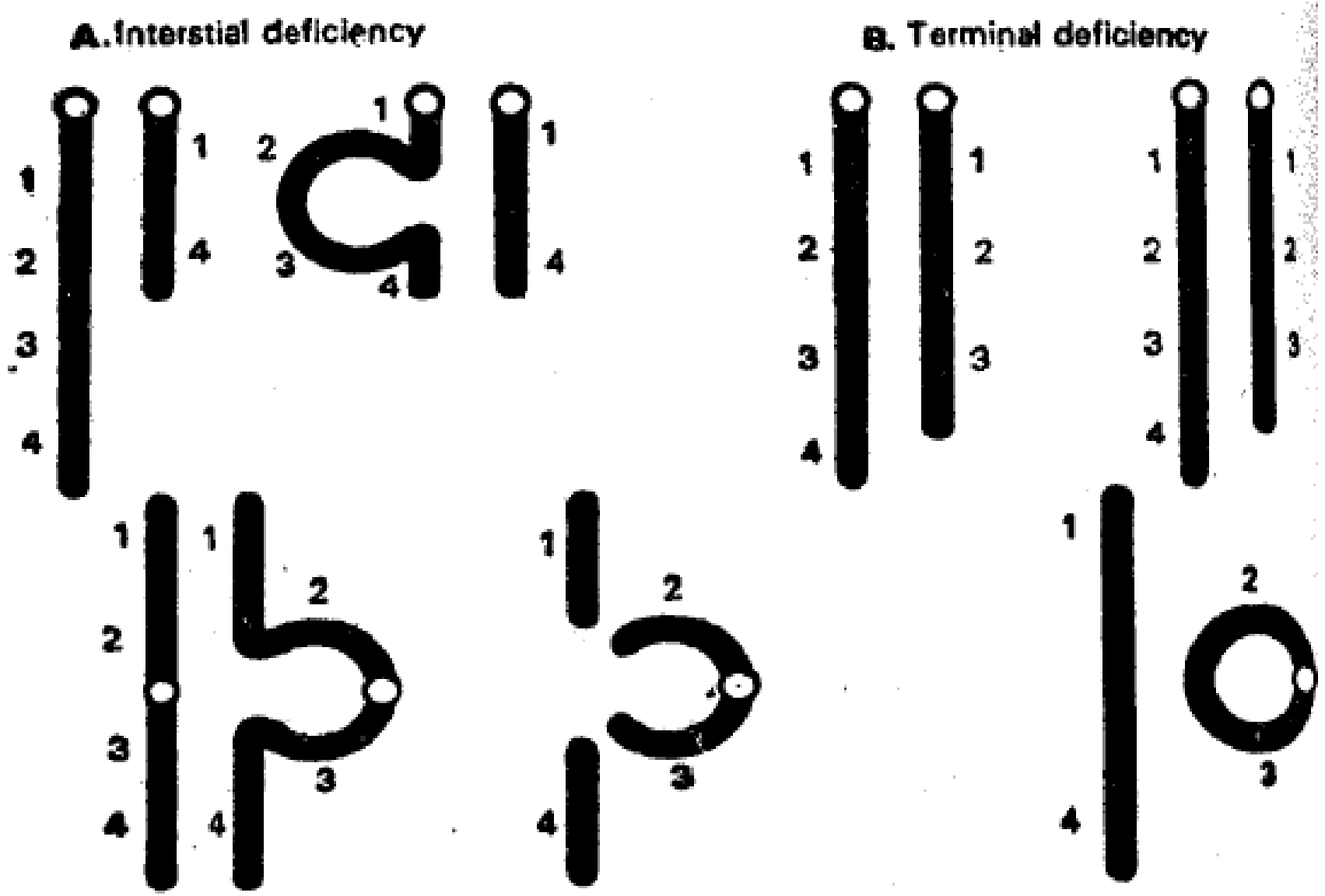
Homozygous deletions - rare and usually lethal

2. Intercalary or interstitial deficiency

Loss of a portion of segment from a chromosome from the intermediate portion or between telomere and centromere.



- **Intercalary deletions are more common than terminal deficiency**
- **The deleted portion may have one / two / several genes**


A. Interstial deficiency
B. Terminal deficiency
C. Formation of an ecentric rod and deletion ring

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Detection of Deletion

- Detected in two ways

1. Cytological method

   Meiotic pairing and chromosome length

2. Genetic method

   Deletion of dominant gene, results in the **expression of recessive gene** – change in the phenotype

Genetic effects due to Deletion/ Deficiency

1. **Fertility : reduced pollen fertility**
2. **Viability: Organisms with homozygous deficiency usually do not survive to an adult stage because a complete set of genes is lacking.**
3. **Crossing over: is suppressed in the region of deficiency**
4. **Phenotype: absence of a dominant gene due to deletion results in the expression of recessive genes, resulting in change in phenotype.**

   Eg: Cat Cry in human (deletion in the short arm of 5th chromosome)

5. **Change in karyotype: Gene number and karyotype of an individual gets changed**

Significance of Deletion

- Important role in species formation and releasing variability through chromosomal mutation
- Cytological tool for mapping genes (for locating genes)

2. Duplications (Additions)

- **Occurrence of a segment twice in the same chromosome**
- It results in **addition of one or more genes** to a chromosome
- Also called as **repeats**
- Reported by **Bridges (1919) in Drosophila**
- Recent reports is on several crops – rice , wheat, maize, Tobacco, Tradescantia, Barley

Duplications are of 4 types

1. Tandem
2. Reverse tandem
3. Displaced
4. Reverse displaced

Duplications are of 4 types

1. Tandem

Sequence of genes in the duplicated segment is **similar** to that of the sequence of genes in the original segment

a b c d e f g h I j k

a b c b c d e f g h I j k

2. Reverse tandem

Sequence of genes in the duplicated segment is **reverse** to that of the sequence of genes in the original segment

also called as adjacent duplication

a b c d e f g h

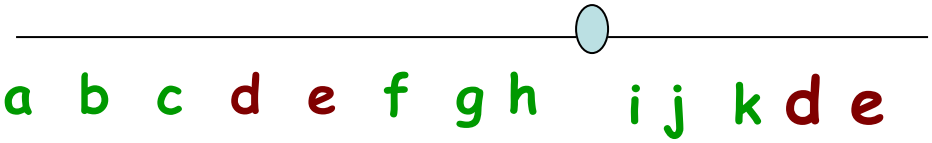
a b c c b d e f g h

3. Displaced

When duplication is found **away from the original segment** but on the same arm of the chromosome

4.Reverse displaced (Non adjacent)

When duplication is found **away from the original segment** but on the other arm of the chromosome



- Duplications arise due to unequal crossing over during meiosis
- Always chromosomes pair with its corresponding identical loci
- Sometimes mis alignment leads to unequal crossing over between non sister chromatids
- This gives rise to two types of chromatids
    - one with duplication
    - other with deletion

- Can be detected by cytological and genetic methods
    - Extra chromosome length
    - Suppression of two recessive alleles by a single duplicated dominant gene

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Genetic significance of Duplications

1. The duplications of chromosomes are not deleterious to the organism like the deficiency, but, they usually protect the organism from the effect of a deleterious recessive gene or from an otherwise lethal deletion.
2. some duplications are useful in the evolution of new genetic material. In an organism with duplications, because the old genes can continue to provide for the present requirements of the organism, the superfluous genes may be free to mutate to new forms without a loss in immediate adaptability.
3. Large duplications can reduce the fertility as a result of meiotic complication, and in this way reduce their own probability of survival (Sybenga, 1972).
4. Relocation of chromosomal material without altering its quantity may result in an altered phenotype, this is called position effect.

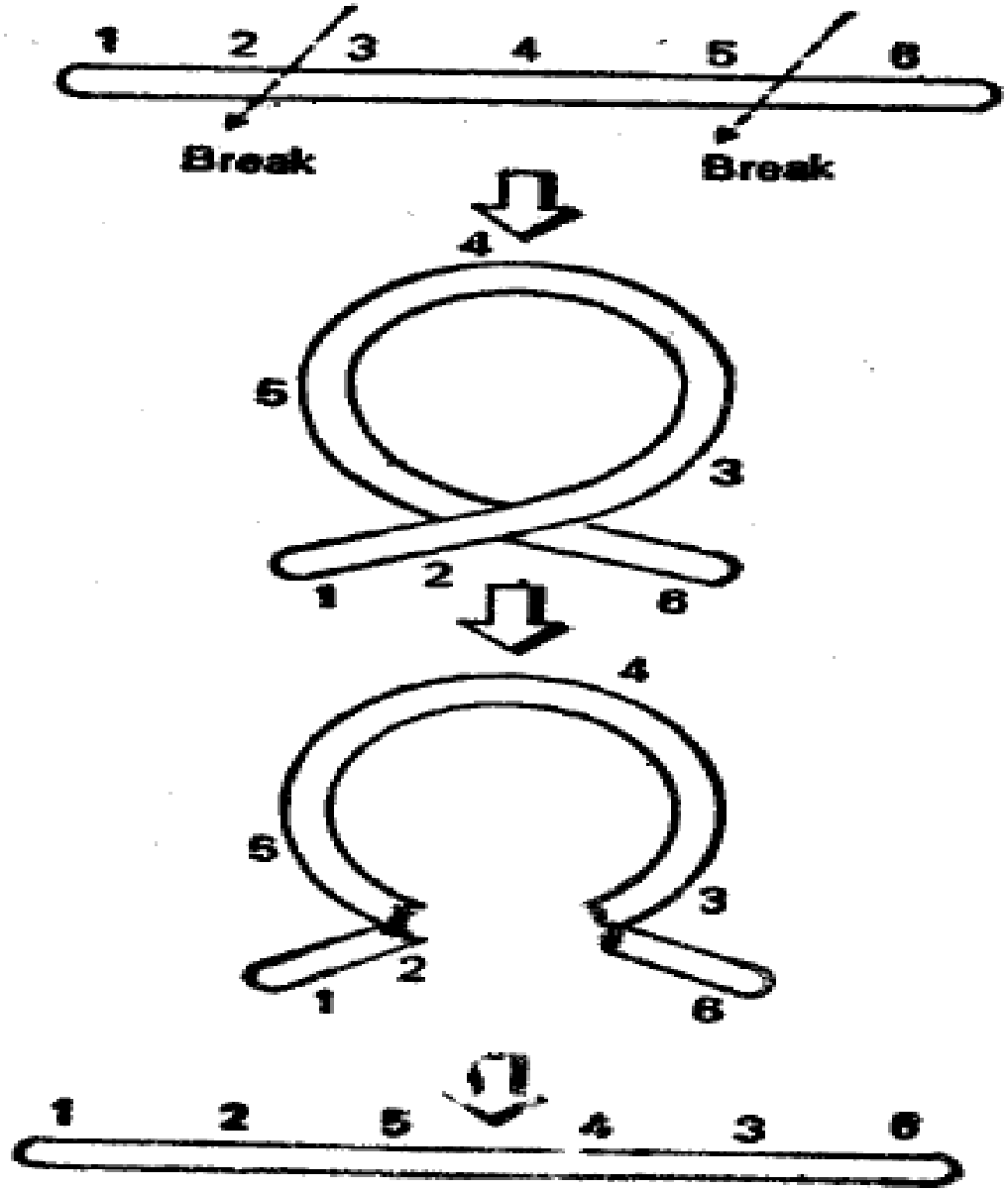
B. Intra-chromosomal aberration

- **INVERSIONS**

  An inversion is an intra-chromosomal aberration in which a segment is inverted 180 degrees.

For example

If a chromosome has segments in the order of 1-2-3-4-5-6 and breaks occur in regions 2-3 and 5-6 and the broken piece (3-4-5-) is reinserted in reverse order, then the inverted chromosome will have segments in order of 1-2-5-4-3-6,



The origin of an inversion (after Stansfield, 1969).

- In a diploid organism, out of two homologous chromosomes, one chromosome undergoes the inversion, then, it is called inversion heterozygote.
- During synapsis of such a homologous pair having inversion heterozygoe, the synapsis configuration attempts to maximize the pairing between homologous regions in the two chromosomes.
- This is usually accomplished by a characteristic inversion loop in one of the chromosome.

Types of inversions

The inversions are of following types:

i) Pericentric inversions

- When the inverted segment of chromosome includes or contains centromere, then such inversions are called heterobranchial or **pericentric inversions**.

- **ii) Paracentric inversions**
- **When the inverted segment includes no centromere and the centromere remains located outside the segment, then such type of inversion is called homobranchial or paracentric inversion.**

Genetic significance of inversions

i) Simple inversions do not have primary phenotypic effects other than on chromosome shape. Frequently, however, some DNA at a break point has been damaged and this may result in an observable mutation, often recessive (e.g., c 1B lethal mutation in Drosophila).

ii) Due to inversion a peculiar kind of position effect occurs. The position effect is caused by the transfer of a gene from a euchromatic segment to the vicinity of heterochromatic segment. Heterochromatinization may then extend into a displaced, originally euchromatic region and suppress the transcription of the gene in it.

iii) Normal linear pairing is not possible in inversion heterozygotes. The difficulties encountered with pairing cause a reduction of exchange (crossing over) in and around the inversion.

iv) They maintain heterozygosity from generations to generations.

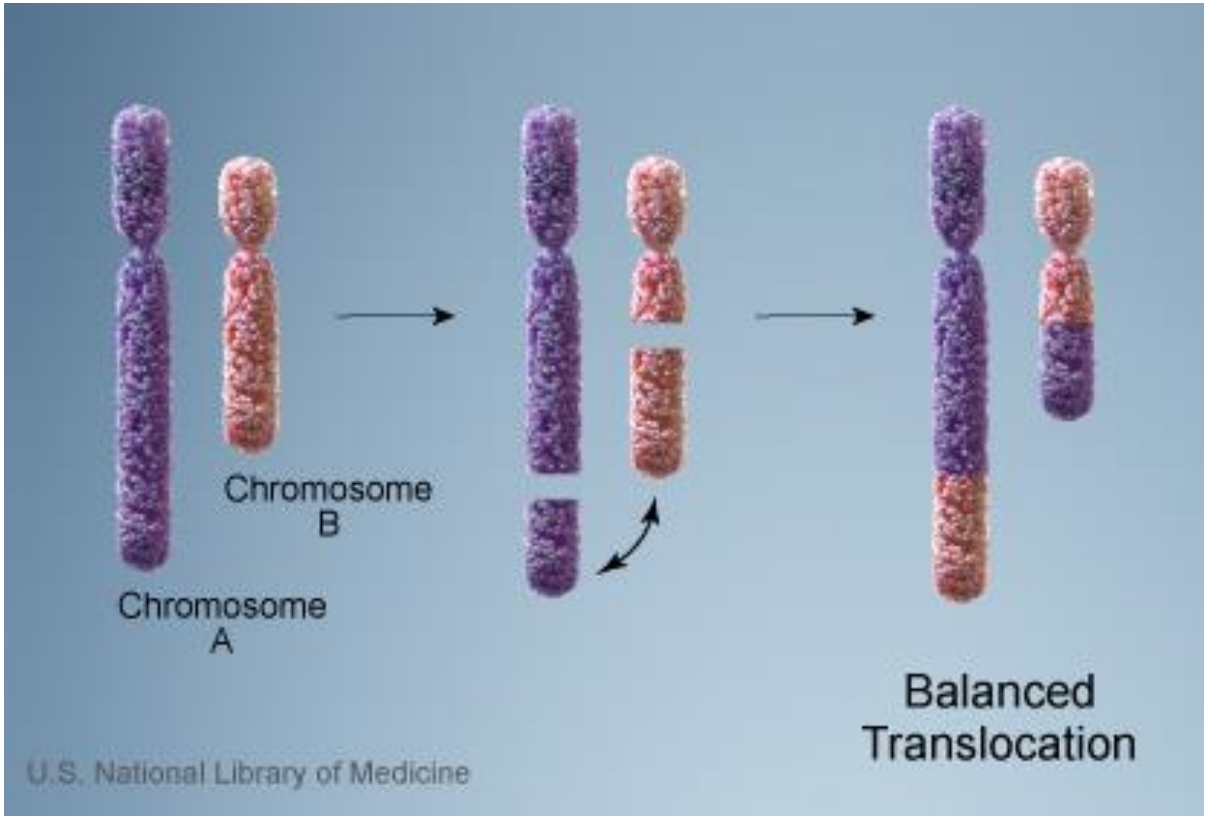
B. Interchromosomal aberrations

When breaks occur in non-homologous chromosomes and resulting fragments are interchanged by both of the non-Homologous chromosomes, the inter-chromosomal or heterosomal aberrations occur.

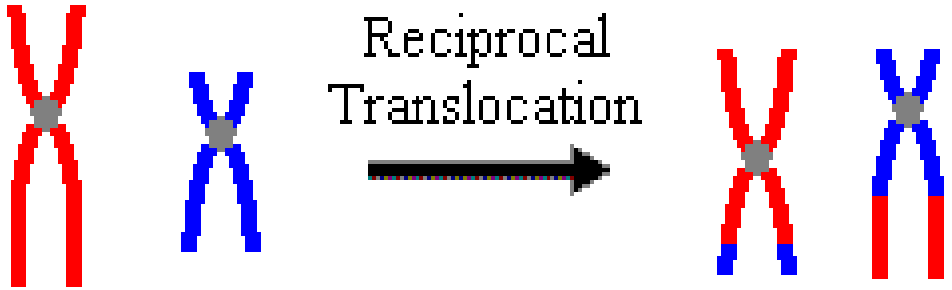
TRANSLOCATION

- **Translocation : It is an inter-chromosomal abberation where in exchange of chromosomal segments occurs between non-homologous chromosomes**
- Translocation involves the shifting of a part of one chromosome to another non homologous chromosome. If two non-homologous chromosomes exchange parts, which need not be of the same size, the result is a **reciprocal translocation**.
- The reciprocal translocation may be of following types:

Translocation

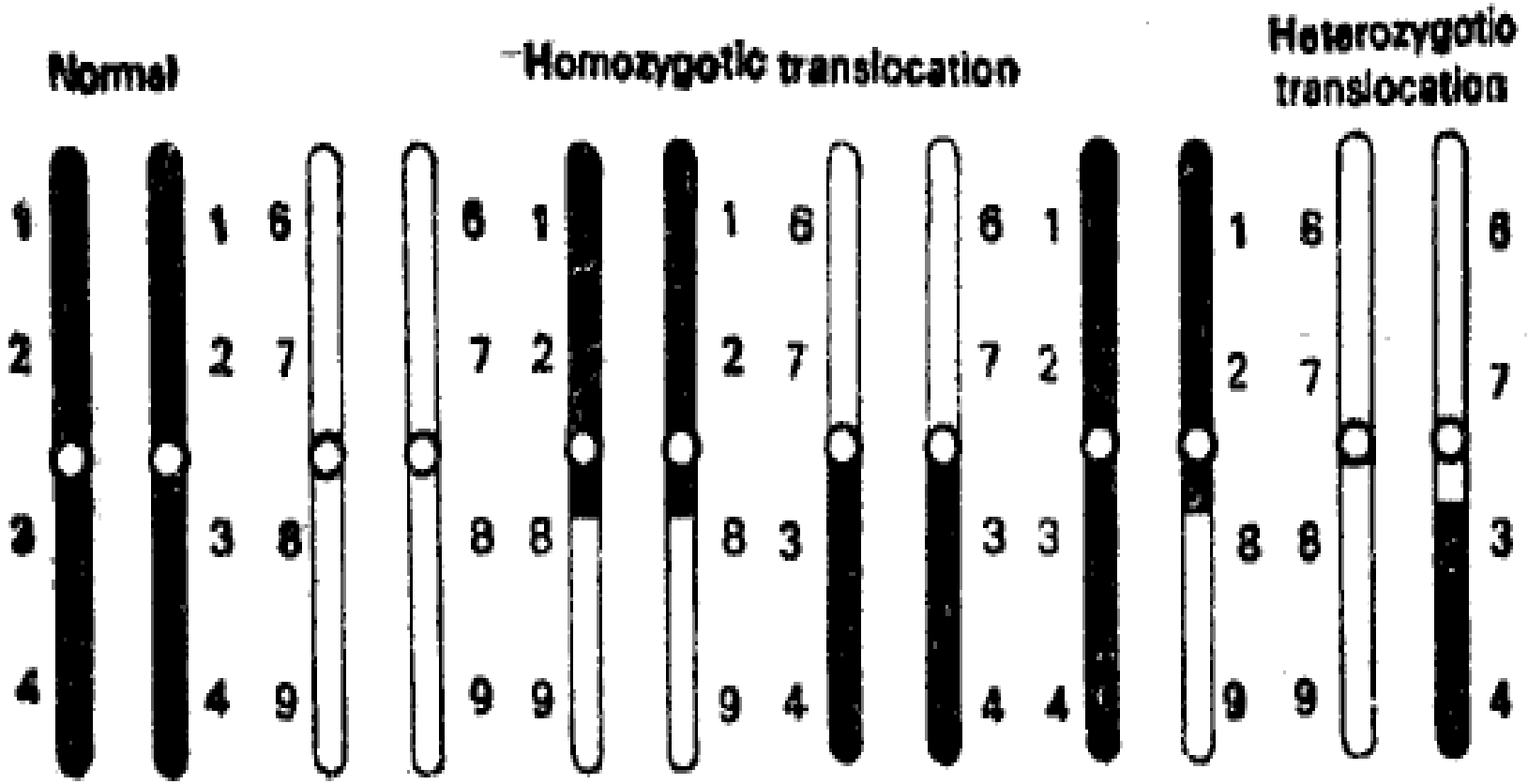


- **Reciprocal translocation:** A type of chromosome rearrangement involving the exchange of chromosome segments between two chromosomes that do not belong to the same pair of chromosomes.



1. Homozygotic translocation

- In homozygotic translocation normal meiosis occur and cannot be detected cytologically.
- Genetically they are marked by altered linkage group by the fact that a gene with new neighbours may produce a somewhat different effect in its new location (position effect).



Homozygotic and heterozygotic translocations
(after De Robertis, Saez and Nowinski 1970)

2. Heterozygotic translocation

- In heterozygotic translocation a considerable degree of meiotic irregularity occur.
- During meiosis, an individual which is heterozygous for a reciprocal translocation must form a **cross-shaped configuration**
- In order to affect pairing of all homologous segments. This cross-shaped configuration often opens out into a ring as chiasmata terminalize.
- The meiotic products (gametes) are of three types – normal, balanced and unbalanced gametes

Genetic significance of Heterozygotic Translocation

1. The heterozygous translocation produce semi-sterile organisms because between half and two third gametes fail to receive the full complements of genes required for normal development of sex.
2. Alter the linkage relationships of genes
3. The phenotypic expression of a gene may be modified when it is translocated to a new position in the genome (position effect).