

CELL BIOLOGY, GENETICS AND EVOLUTION

BOT-401 4(3-1)

Chromosomal Mutations -I

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Chromosomal Mutations

- The chromosomes of each species has a characteristic morphology (structure) and number
- But, sometimes due to certain accidents at the time of cell division, crossing over or fertilization some alterations in the morphology and number of chromosomes take place
- The changes in chromosome parts, whole chromosomes, or whole chromosome sets are called chromosome aberrations or chromosome mutations.

Chromosomal Mutations

- **Structural changes in chromosomes**
- **Changes in number of chromosomes**

Chromosomal Mutations

Chromosome mutations are of the following types :

A. Structural changes in chromosomes:

- 1. **Changes in number of genes**
 - ✓ (a) **Deletion**: Loss of gene
 - ✓ (b) **Duplication**: Addition of gene
- 2. **Changes in gene arrangement**
 - ✓ (a) **Inversion**: Rotation of a group of genes within one chromosome
 - ✓ (b) **Translocation**: Exchange of parts between chromosomes of different pairs.

Chromosomal Mutations

B. Changes in number of chromosomes :

- ✓ 1. **Aneuploidy:** Loss, or gain, of a part of the chromosome set.
- ✓ 2. **Euploidy:** Loss, or gain, of whole chromosome set.
 - ✓ (a) **Haploidy:** Loss of an entire set of chromosomes.
 - ✓ (b) **Polyploidy:** Addition of one or more sets of chromosomes.

VARIATIONS IN CHROMOSOME STRUCTURE

- **Deletion**
- **Duplication**
- **Inversion**
- **Translocation**

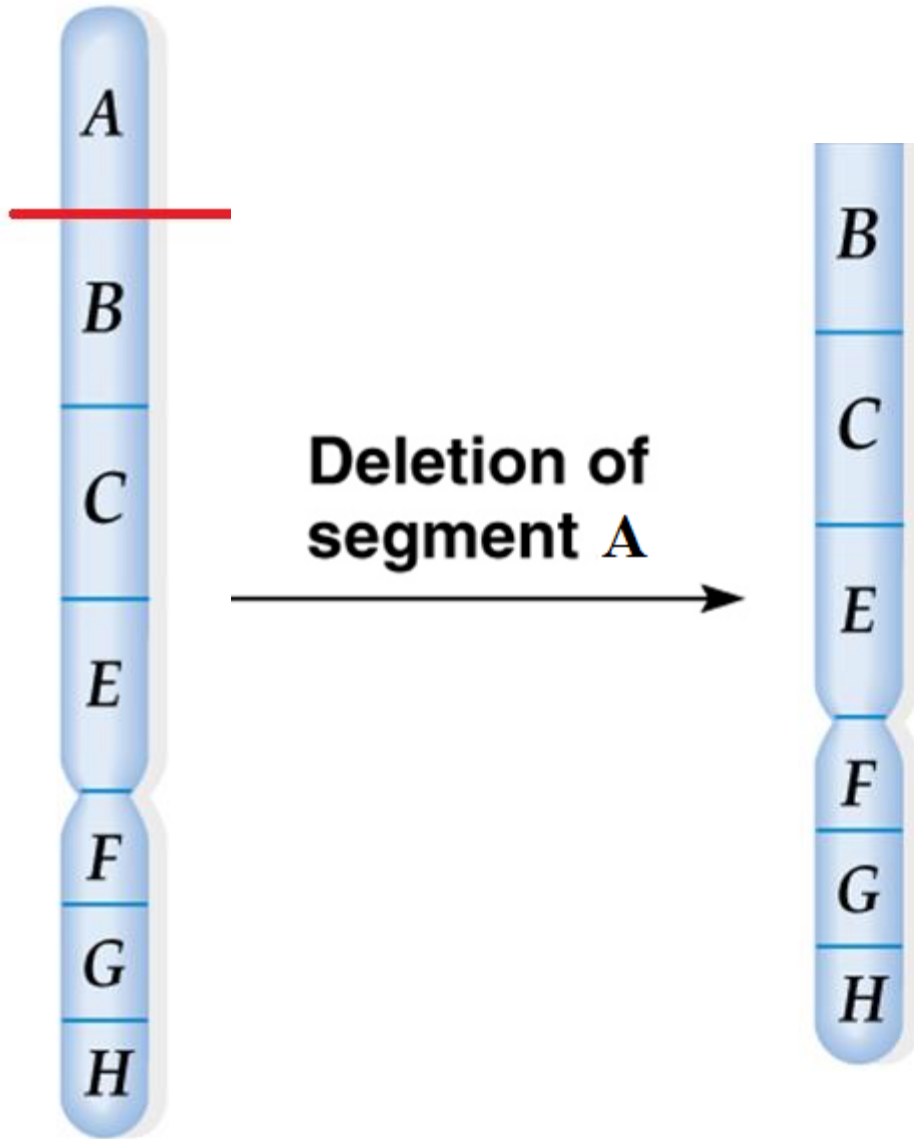
Deletion (or deficiency)

Deficiency or deletion which involves loss of a broken part of a chromosome

- The chromosomes with deletions can never revert to a normal condition

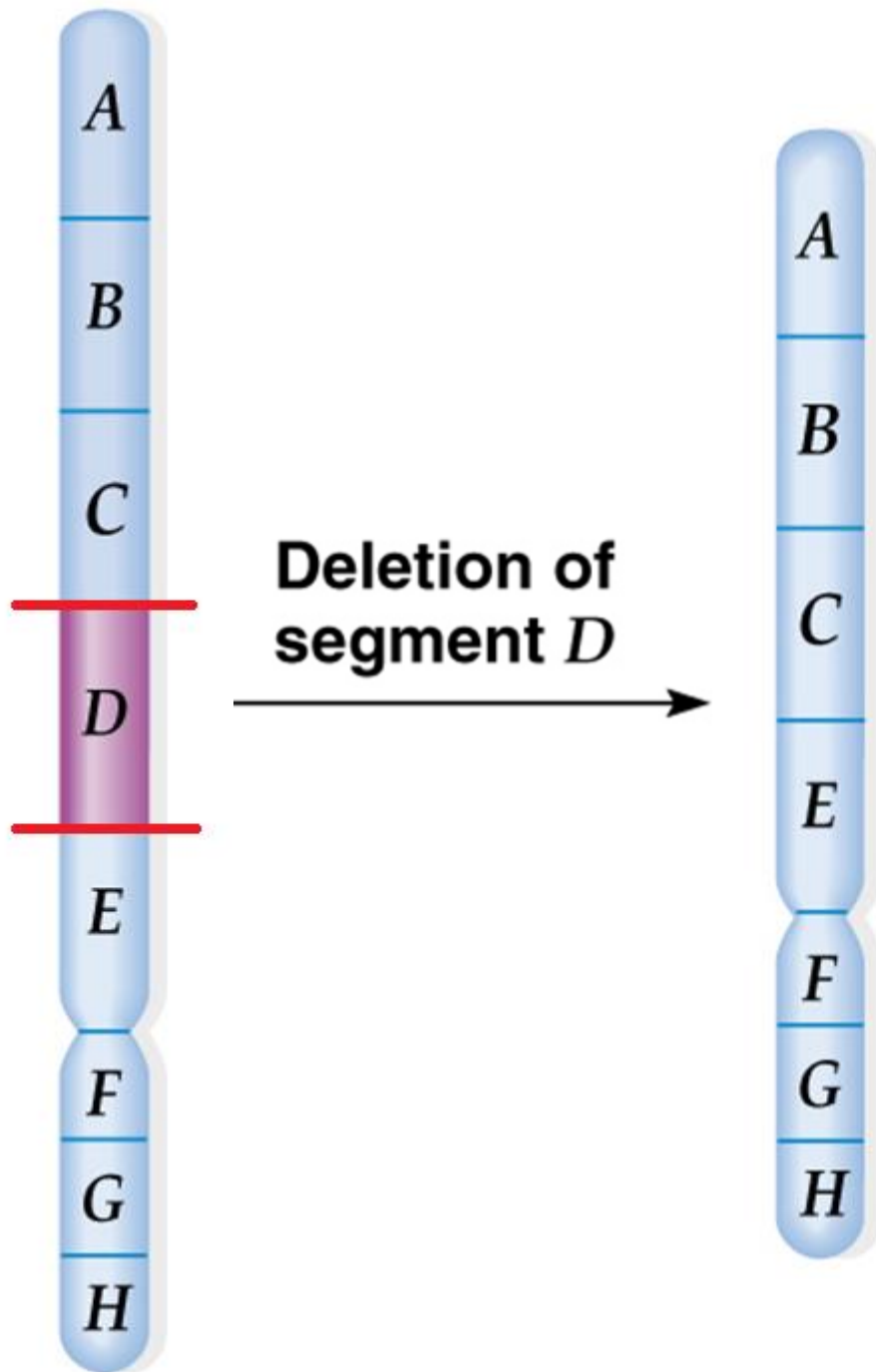
Two types of deletion

- **Terminal deletion**: a terminal section of a chromosome is absent and it is resulted by only one break.
- **Intercalary / interstitial deletion**: an intermediate section or portion of chromosome is lost and it is caused by two breaks — one on either end of the deleted region.



**A deletion of a
chromosome
segment**

**Terminal
deletion**



**A deletion of a
chromosome
segment**

**Intercalary /
interstitial
deletion**

Duplication

- The presence of a part of a chromosome in **excess (duplicate)** of the normal complement is known as **duplication**.
- Thus, due to duplication some genes are present in a cell in **more than two doses**
- Different types of duplication
 1. **Tandem duplication**
 2. **Reverse tandem duplication**
 3. **Terminal tandem duplication**
 4. **Displaced duplication**
 5. **Transposed duplication**

Tandem duplication

- In this case the duplicate region is situated just by the side of the normal corresponding section of the chromosome
- and the sequences of genes are the same in normal and duplicated region.
- For example, if the sequence of genes in a chromosome is ABC.DEFGH (The full stop depicts the centromere)
- and if the chromosomal segment containing the genes **DEF** is duplicated,
- the sequence of genes in tandem duplication will be
ABC.**DEFDEF**GH

Reverse tandem duplication

- Here, the sequence of genes in the duplicated region of a chromosome is just the reverse of a normal sequence
- In the above mentioned example, therefore, the sequence of genes due to reverse tandem duplication will be ABC.DEFFEDGH

Terminal tandem duplication

- Here, the sequence of genes in the duplicated region of a chromosome is the terminal region
- ABC.DEFGH (Normal)
- **AB**ABC.DEFGH (Duplicated)

Displaced duplication

- In this case the duplicated region is not situated adjacent to the normal section.
- Depending on whether the duplicated portion is on the same side of the centromere as the original section
- or **on the other side**, the displaced duplication can be termed either
 1. **Homobranhial** (ABC.DEFGDEFH)
 2. **Heterobranhial** (ADEFBC.DEFGH)

Transposed duplication

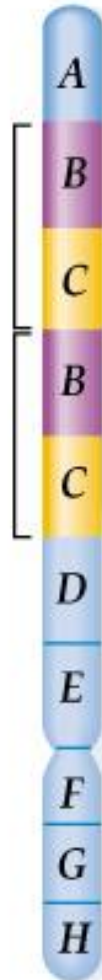
- Here, the duplicated portion of chromosome becomes attached to a non-homologous chromosome.
- For example, if ABC.DEFGH and LMNOPQ.RST represent the gene sequences of two non-homologous chromosomes,
- a transposed duplication will result into chromosomes with gene sequence ABC.GH and LMNDEFOPQ. RST.
- Such a transposed duplication may be either
 1. interstitial (e.g., LMNDEFOPQ. RST)
 2. terminal (i.e., LMNOPQ. RSTDEF).

Forms of chromosome duplications

Normal chromosome



Duplications



Tandem



Reverse
tandem



Terminal
tandem

Inversion

- Inversion involves a rotation of a part of a chromosome or a set of genes by 180 on its own axis
- It essentially involves occurrence of breakage and reunion
- The net result of inversion is neither a gain nor a loss in the genetic material but simply a rearrangement of the gene sequence
- An inversion can occur in the following way :
 1. Suppose that the normal order of segments within a chromosome is 1-2- 3-4-5-6 ;
 2. Breaks occur in regions 1-2- and -5-6
 3. And broken piece is reinserted in reverse order.
 4. This results in an inverted chromosome having segments 1-2-5-
4-3-6

Inversion

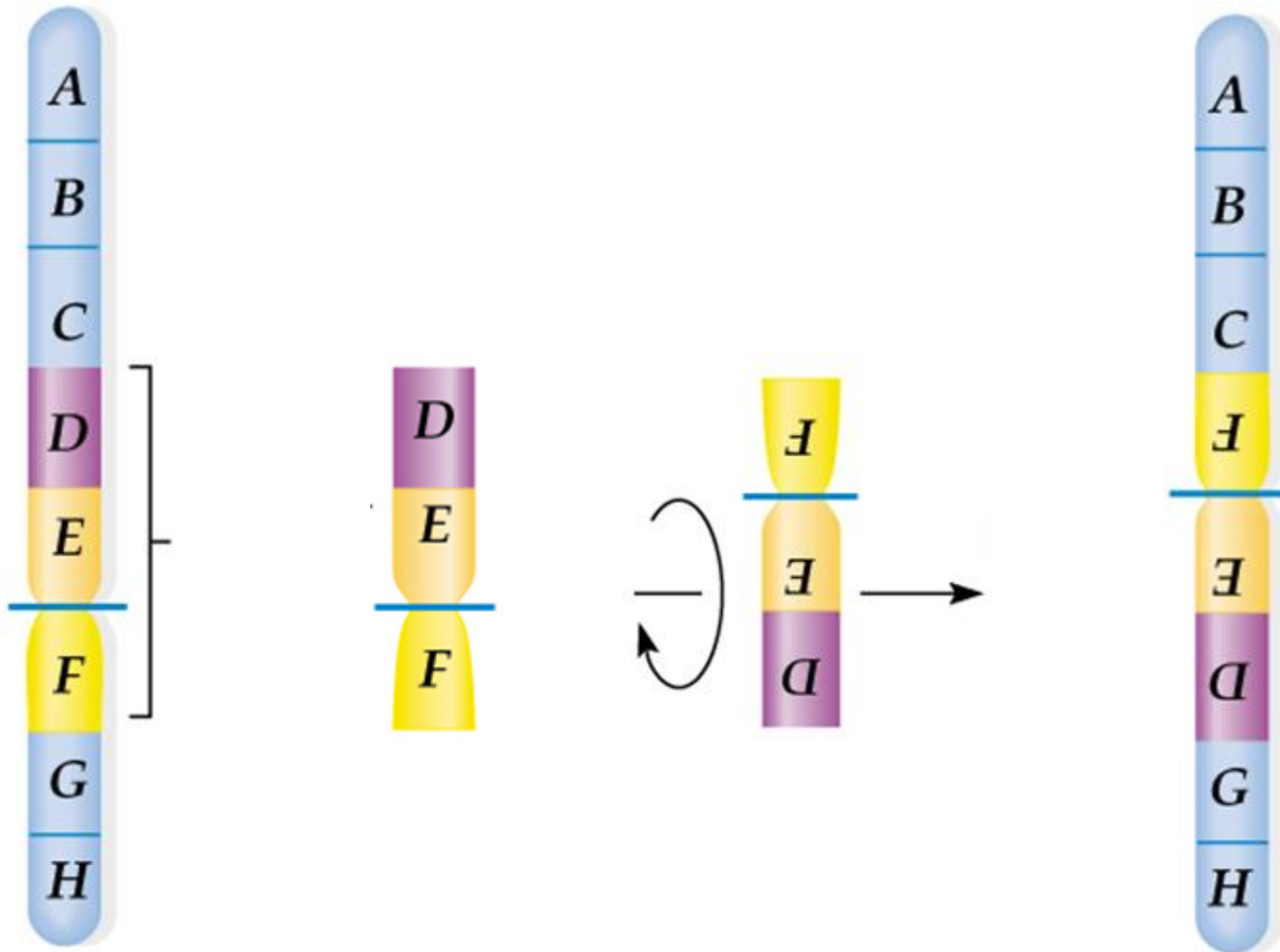
- An **inversion heterozygote** has one chromosome in the inverted order and its homologue in the normal order

The location of the **centromere** relative to inverted segment determines the genetic behaviour of the chromosomes.

- If **inversion includes** the centromere it is called pericentric inversion
- If the **centromere is not included** in the inversion it is called paracentric inversion

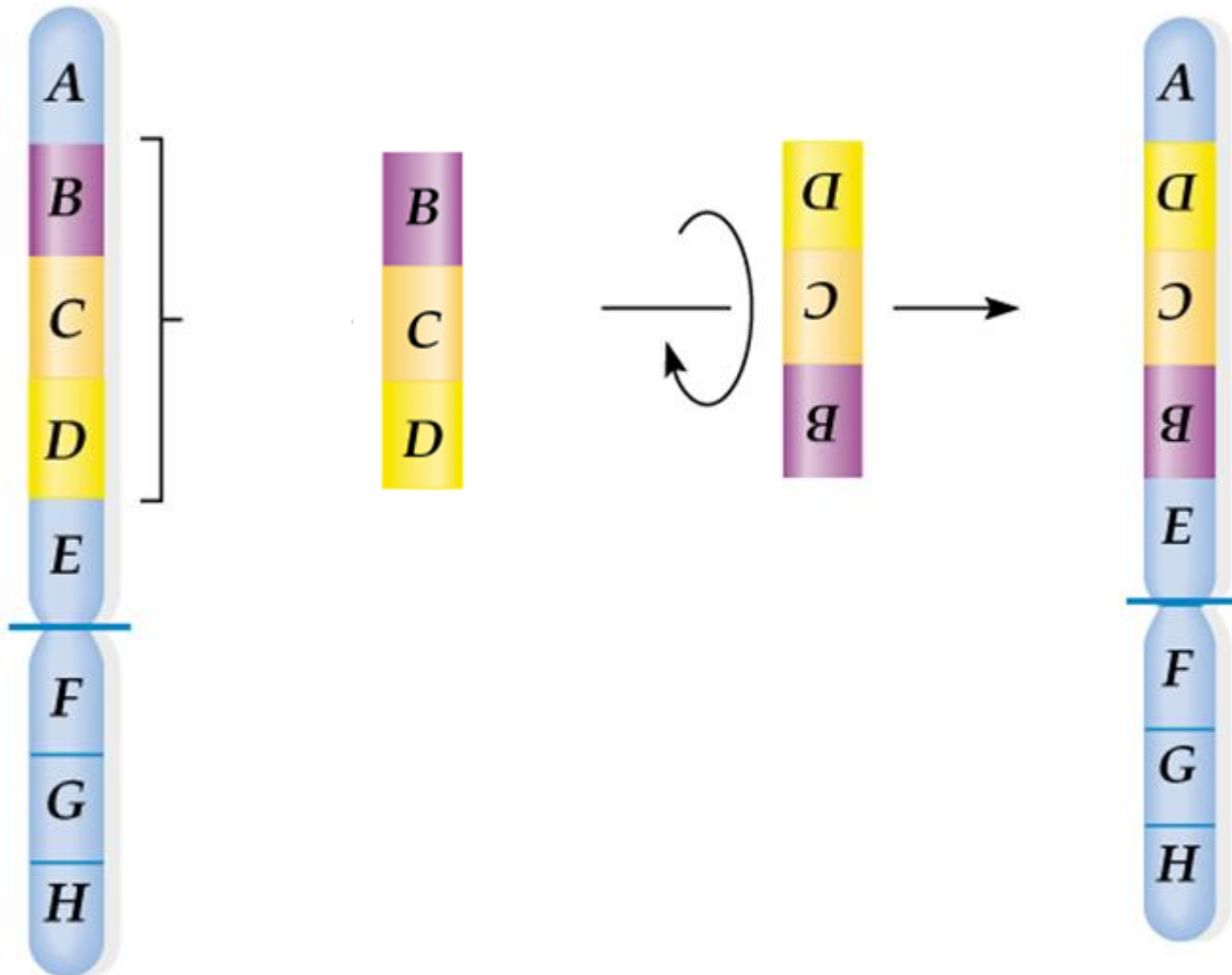
Inversions

a) Pericentric inversion
(includes centromere)



Inversions

b) Paracentric inversion (does not include centromere)



Translocation

- A translocation is a chromosomal mutation in which there is a change in the position of particular chromosome segment
- No genetic material is lost or gained (in a population of chromosomes) due to translocation
- **Intra-chromosomal translocations** involve a change in the position of a chromosome segment within the same chromosome

Translocation

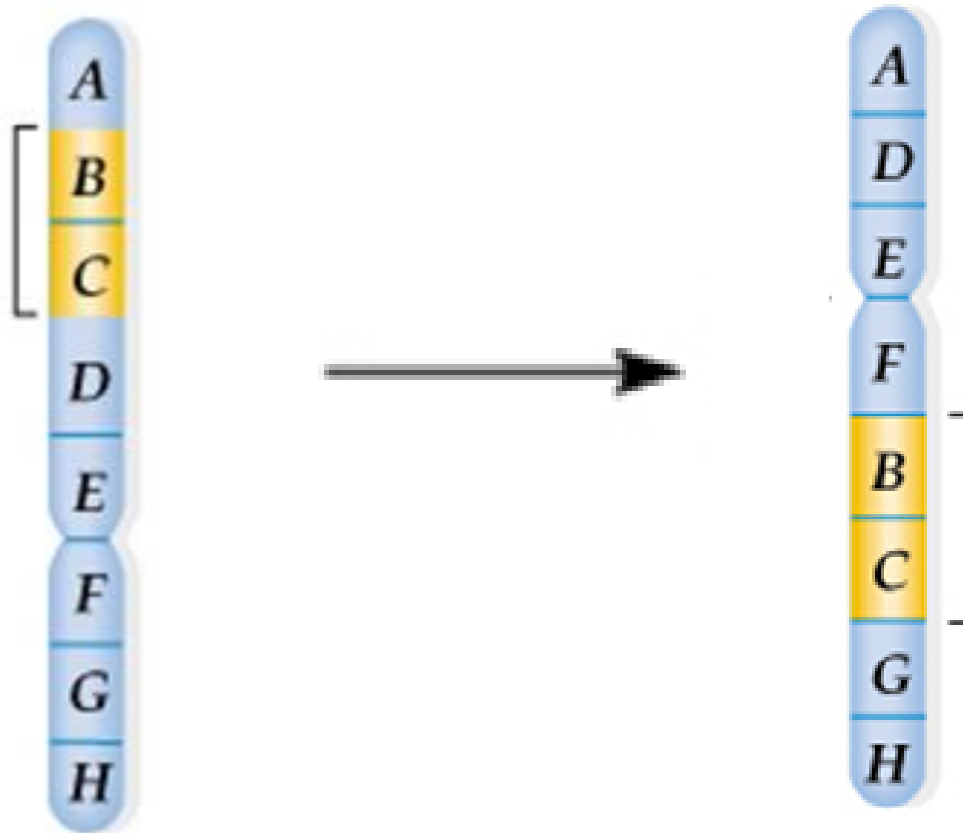
- Types of Translocation
- 1. **Intra-chromosomal translocations**
involve a change in the position of a chromosome segment within the same chromosome
- 2. **Inter-chromosomal translocations**
involve more than one chromosome

Translocation

- **Types of Inter-chromosomal Translocation**
- **A) Non-Reciprocal Translocation :**
- Inter-chromosomal translocations that result in a one-way transfer of genetic material from one chromosome to another are called **non-reciprocal translocations**
- **B) Reciprocal Translocation:**
- If segments are exchanged it is a **reciprocal translocation**

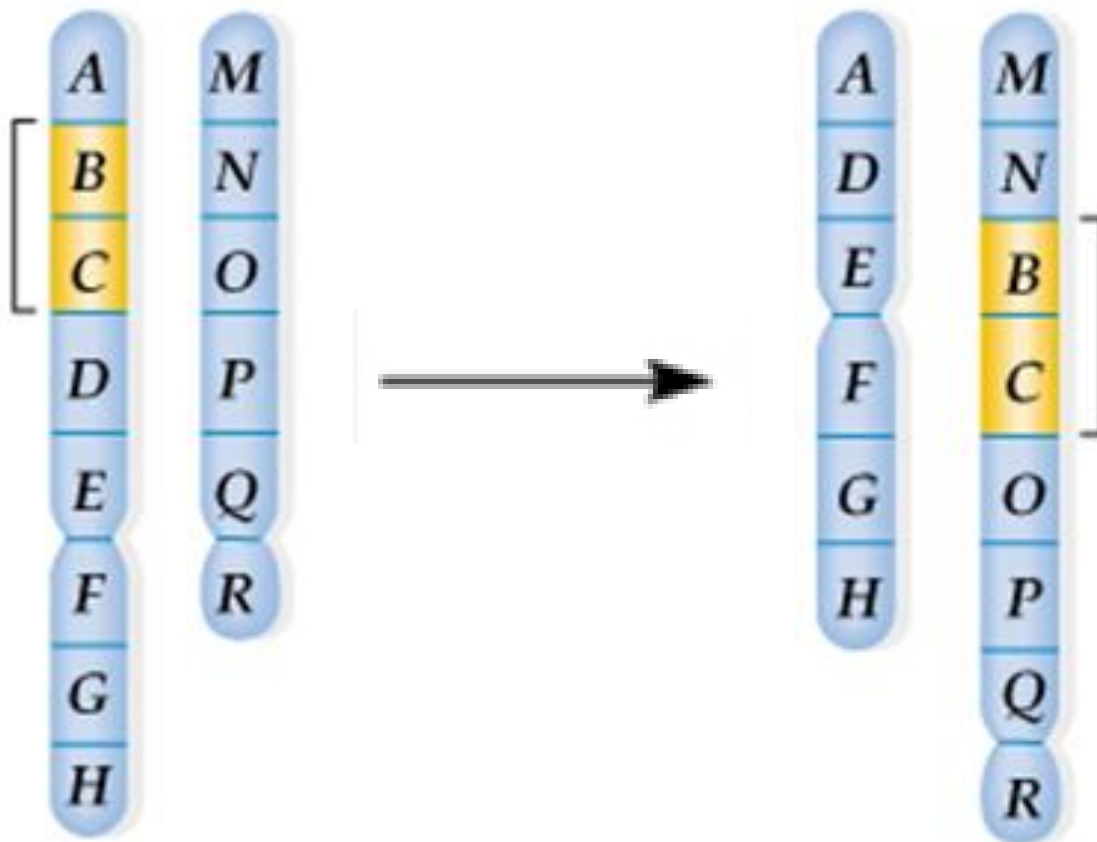
Chromosomal Translocations

a) Nonreciprocal
intrachromosomal
translocation



Chromosomal Translocations

b) Nonreciprocal interchromosomal translocation



Chromosomal Translocations

c) Reciprocal interchromosomal translocation

