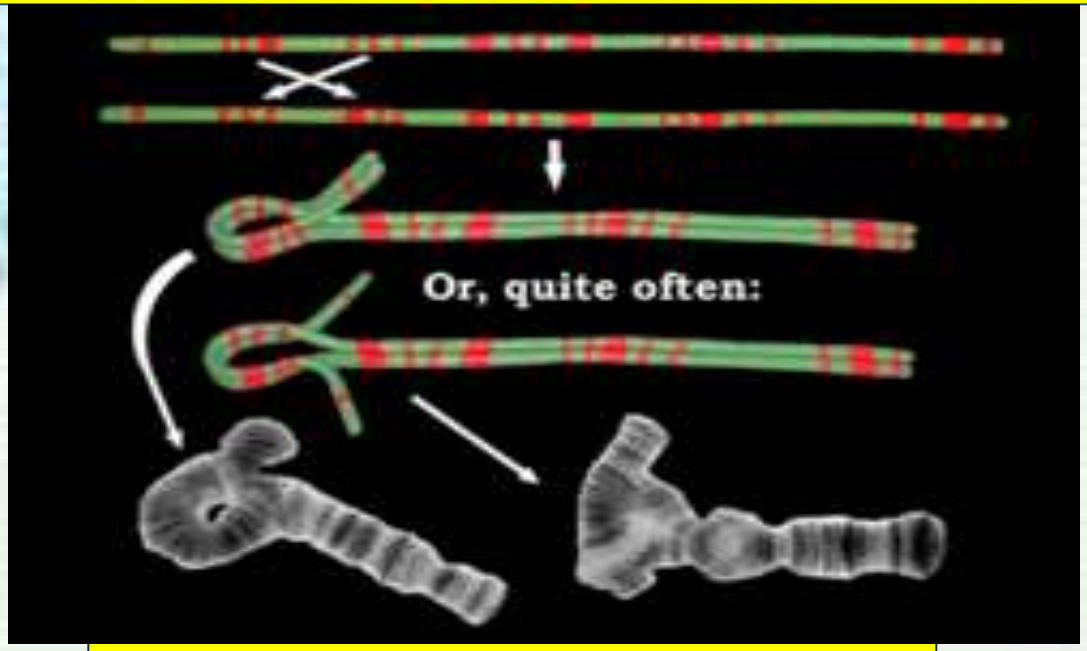
Structural Chromosome Aberration



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Introduction

✓Chromosomes may undergo changes. This is called **structural variations in chromosomes or chromosomal aberration.**

✓The change may occur either in structure of the chromosomes or in the number of chromosomes.

✓Structural chromosome rearrangements are changes in the physical structure of chromosomes that may result in birth defects, mental retardation and increased risk for infertility.

✓Allelic variations are due to mutations in particular genes

✓Chromosomal aberrations are substantial changes in chromosome structure

- These typically affect more than one gene
- They are also called **chromosomal mutations**

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Inversion

Translocation

History

✓Based on these, the chromosomal aberrations are grouped into two major kinds- variation in structure and variation in number.

✓1926 **A.H. Sturtevant** Discovered inversions in chromosomes.

✓1923 **C.B. Bridges** Discovered duplications, deletions and translocations in chromosomes

Deletion

✓A deficiency means deletion of a small portion of a chromosome resulting in loss of one or more genes.

✓A deficiency originates from breakage occurring at random in both chromatids of a chromosome (called chromosome break), or only in one chromatid (chromatid break).

✓The breakage may be caused by various agents such as radiation, chemicals, drugs or viruses at any time during the cell cycle, either in somatic or in germ cells.

✓Depending upon its location, a deletion may be terminal when a single break occurs near the end of the chromosome; or interstitial when two breaks occur in a middle portion of the chromosome.

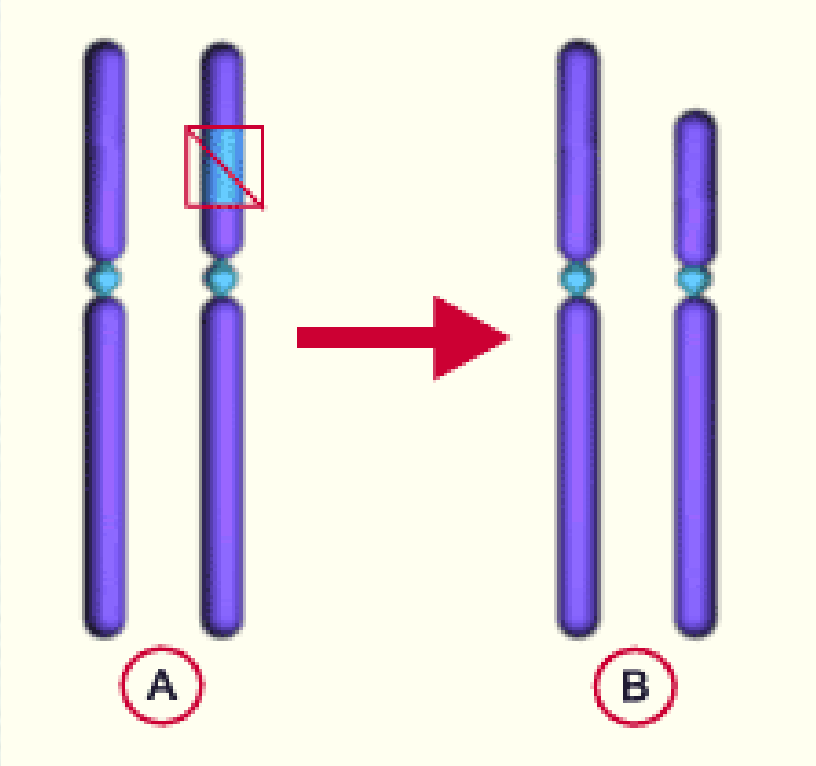
The effect of a deletion depends on what was deleted.

- ✓ A deletion in one allele of a homozygous wild- type organism may give a normal phenotype, while the same deletion in the wild-type allele of a heterozygote would produce a mutant phenotype.
- ✓ Deletion of the centromere results in an acentric chromosome that is lost, usually with serious or lethal consequences.

Examples of human disorders caused by large chromosomal deletions:

✓Cri-du-chat (“cry of the cat”) syndrome, resulting from deletion of part of the short arm of chromosome 5

✓The deletion results in severe mental retardation and physical abnormalities.



Duplication

✓A duplication involves attachment of a chromosomal fragment resulting in addition of one or more genes to a chromosome.

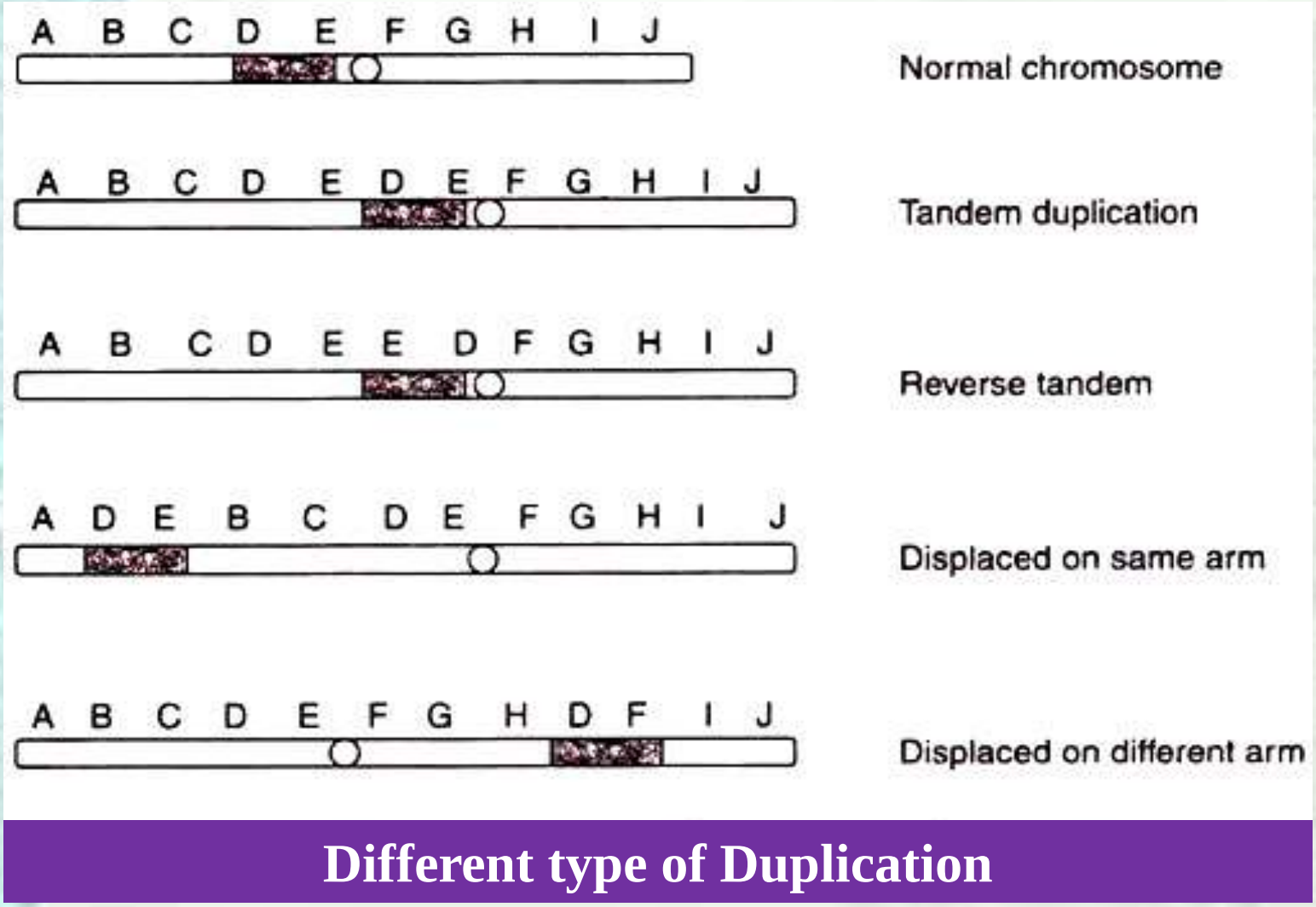
✓Whenever there is a duplication in a chromosome, there is a corresponding deletion in another chromosome.

✓The origin of duplications can be traced to unequal crossing over during meiosis. Normally homologous chromosomes are paired in a perfect manner so that identical loci lie exactly opposite each other.

✓The mechanism ensures that after crossing over between non-sister chromatids, equal exchange products are formed.

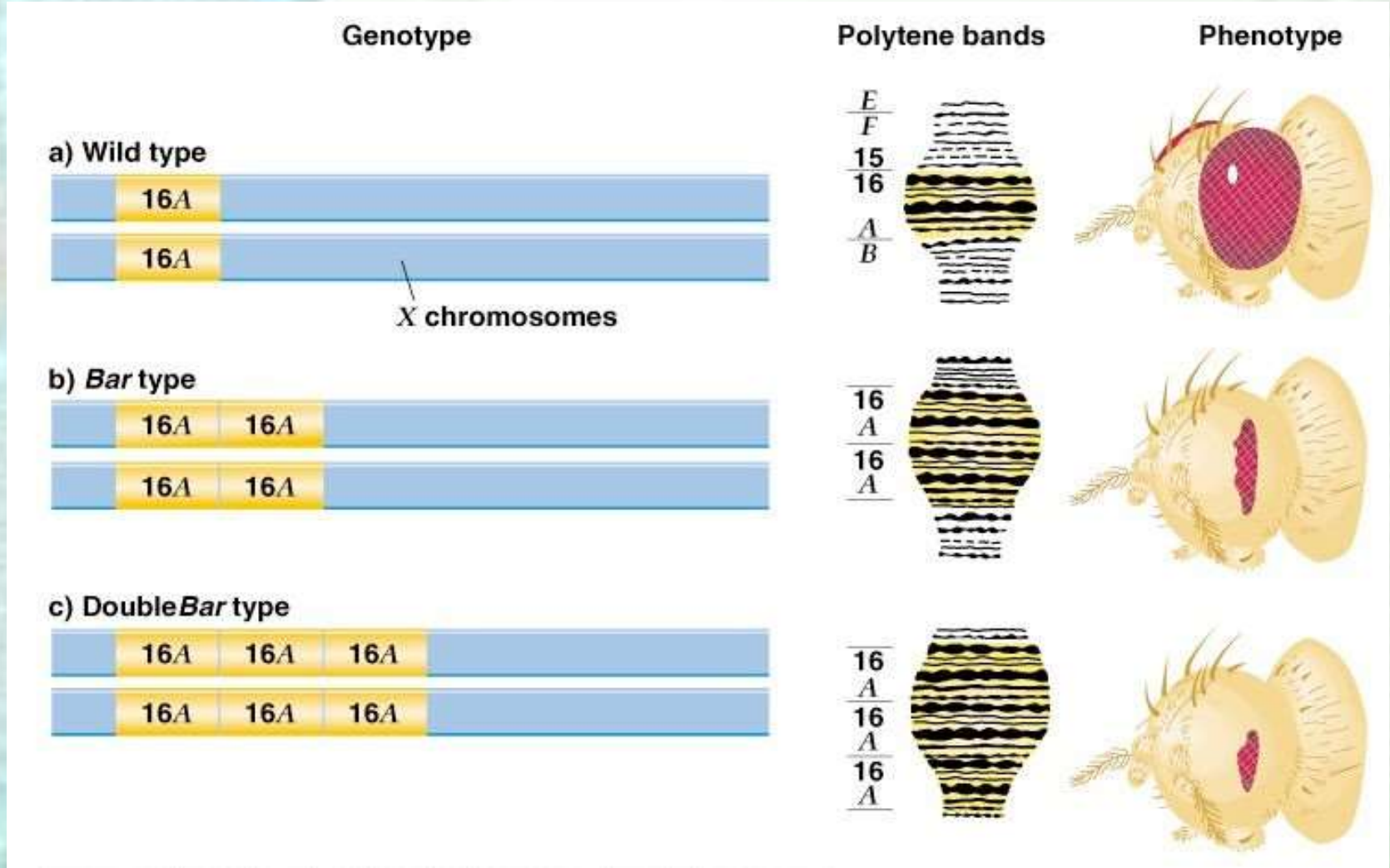
✓If paired chromosomes are misaligned, it is not possible for exchange to take place between exactly opposite locations on two chromatids.

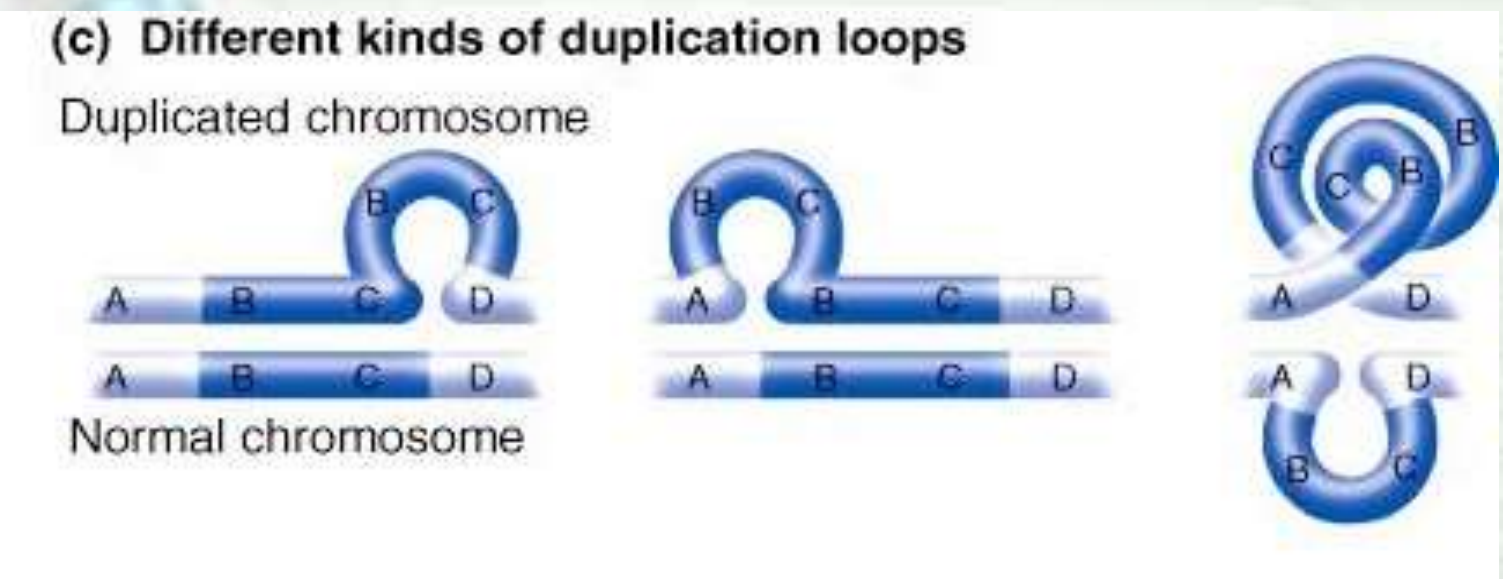
✓Duplications do not produce any drastic consequences as like deletion in terms of phenotype and survival.



Different type of Duplication

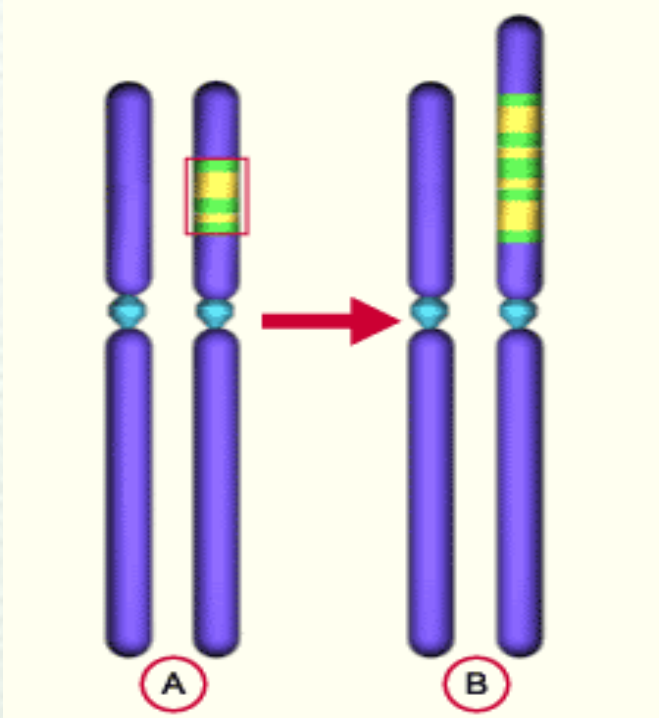
Example is the *Drosophila* eye shape allele, *Bar*, that reduces the number of eye facets, giving the eye a slit-like rather than oval appearance





✓Duplication loops form when chromosomes pair in duplication heterozygotes

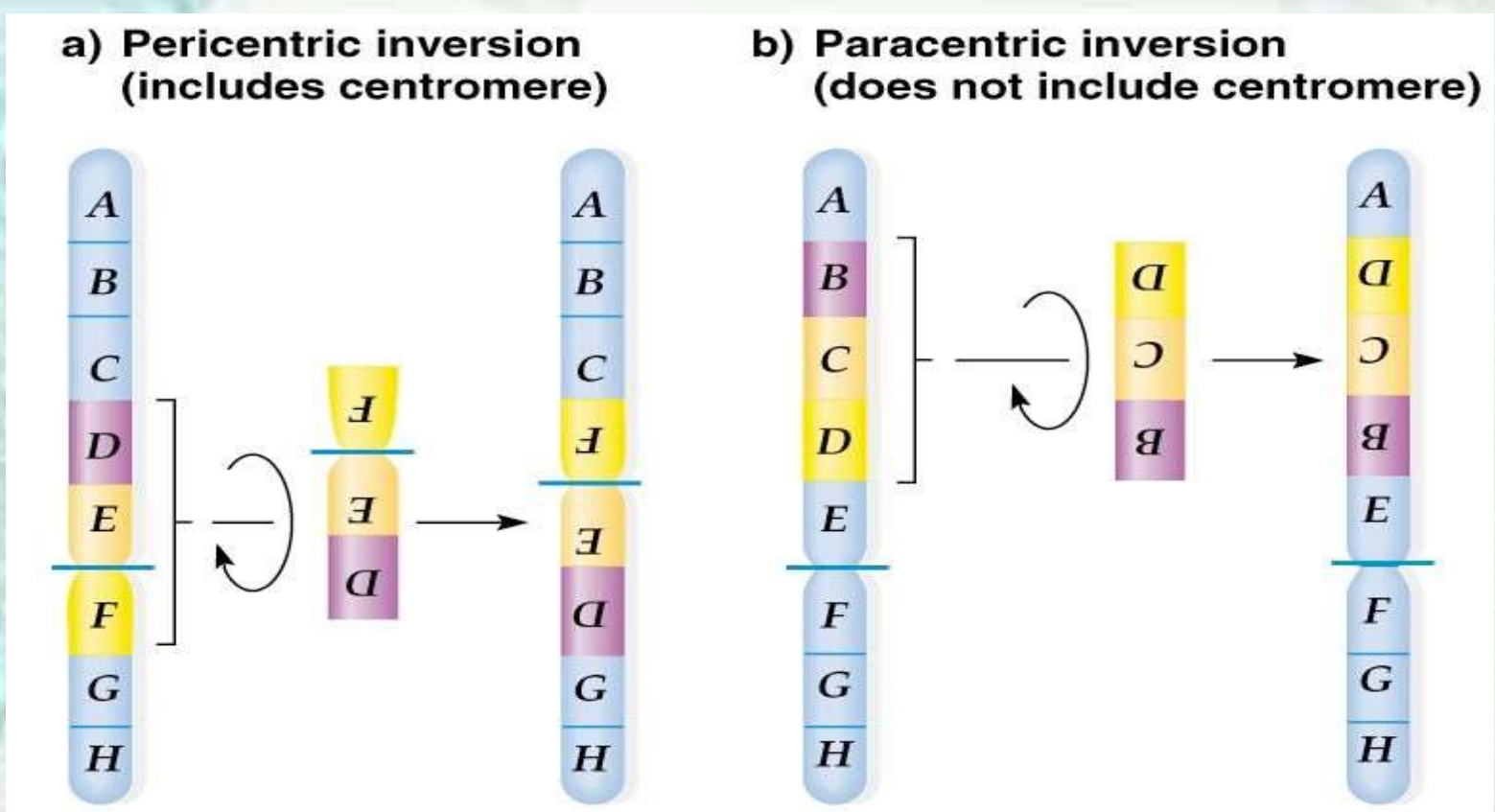
✓In prophase I, the duplication loop can assume different configurations that maximize the pairing of related regions



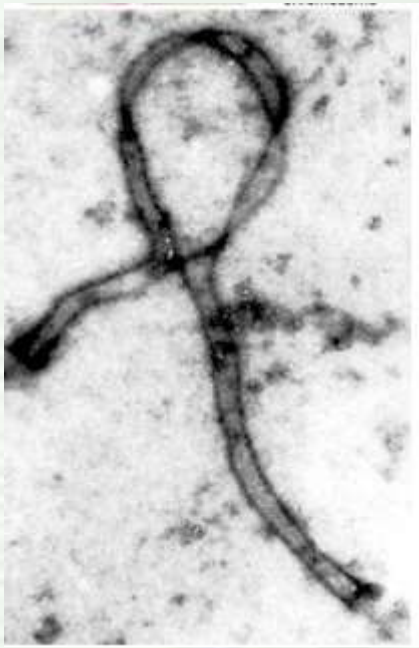
Inversion

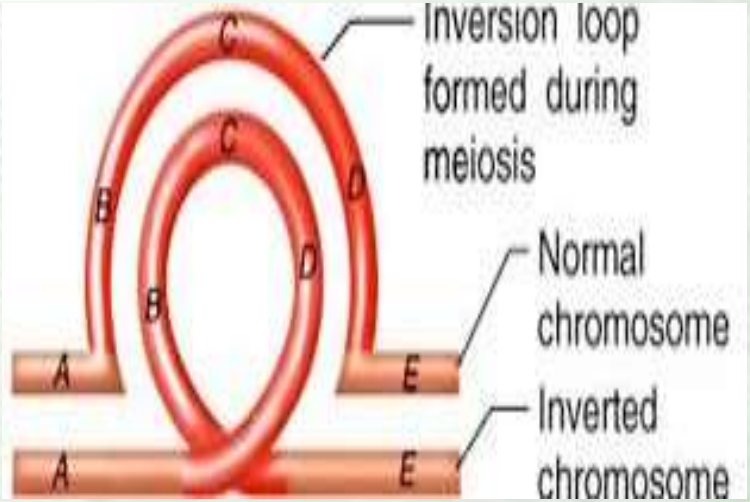
Inversion results when a chromosome segment excises and reintegrates oriented 180° from the original orientation.

– There are two types

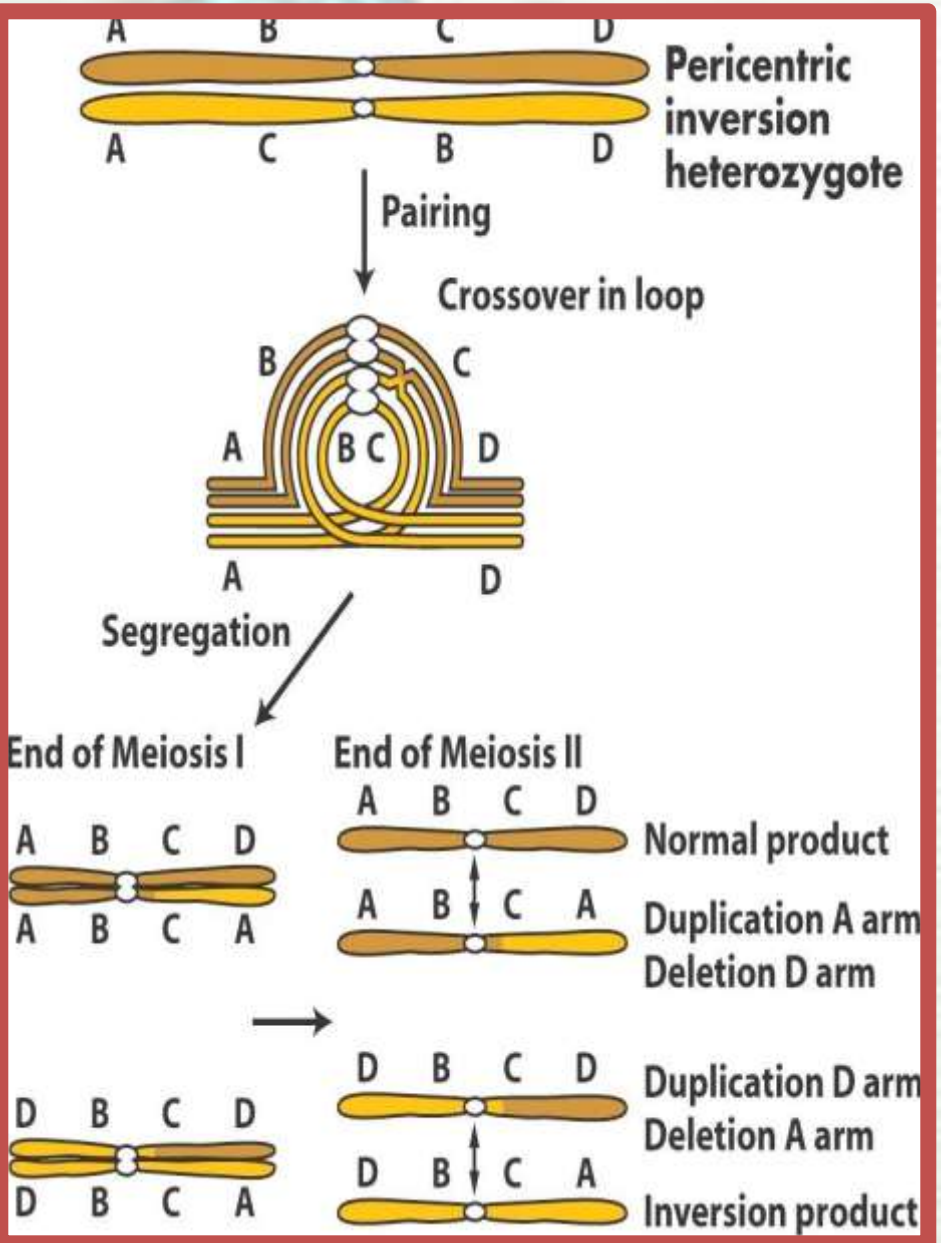


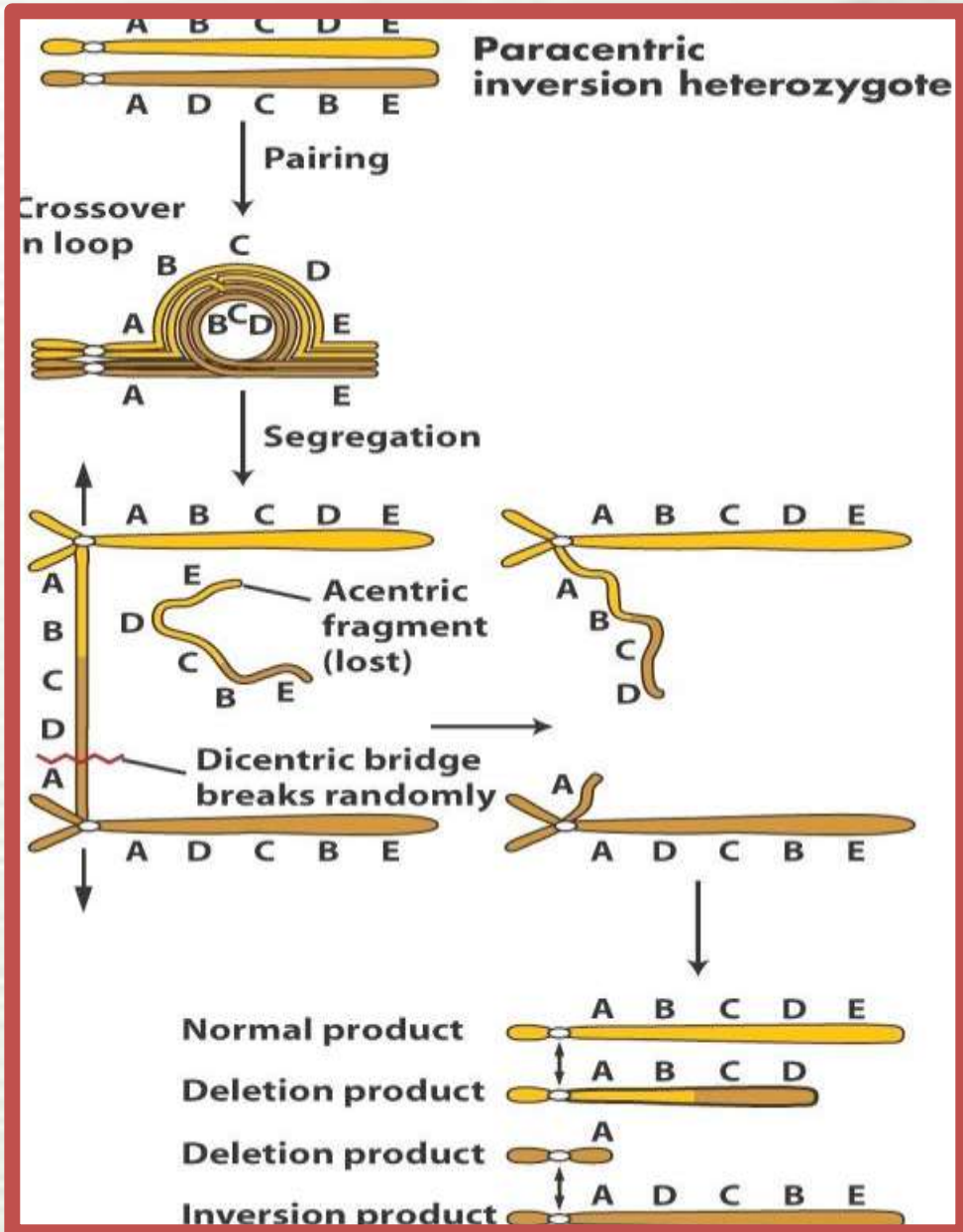
- ✓ Linked genes are often inverted together.
  - ✓ The meiotic consequence depends on whether the inversion occurs in a homozygote or a heterozygote.
- ✓ A homozygote will have normal meiosis.
- ✓ The effect in a heterozygote depends on whether crossing-over occurs.
  - ✓ If there is no crossing-over, no meiotic problems occur.
  - ✓ If crossing-over occurs in the inversion, unequal crossover may produce serious genetic consequences.
  - ✓ Inversion loop in heterozygote forms tightest possible alignment of homologous regions





Gametes produced from pericentric and paracentric inversions are imbalanced

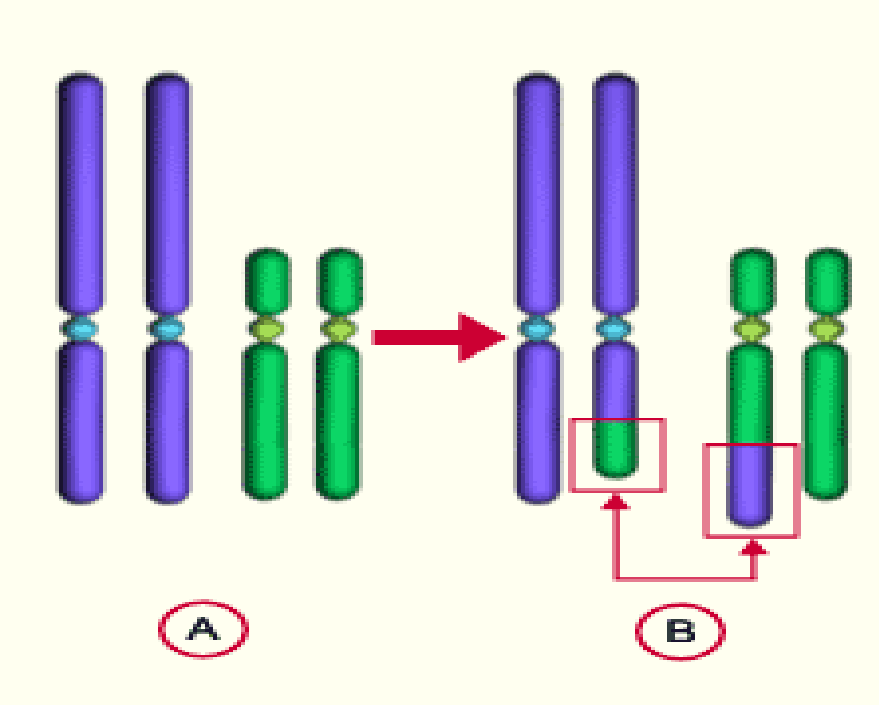




Translocation

✓Integration of chromosome segment into a non-homologous chromosome is known as translocation.

✓It involves the transfer of a segment of chromosome to a different part of the same chromosome or a different chromosome.



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the breakage takes place in two non-homologous chromosomes, and is followed by the reunion of broken segment to the wrong partners.

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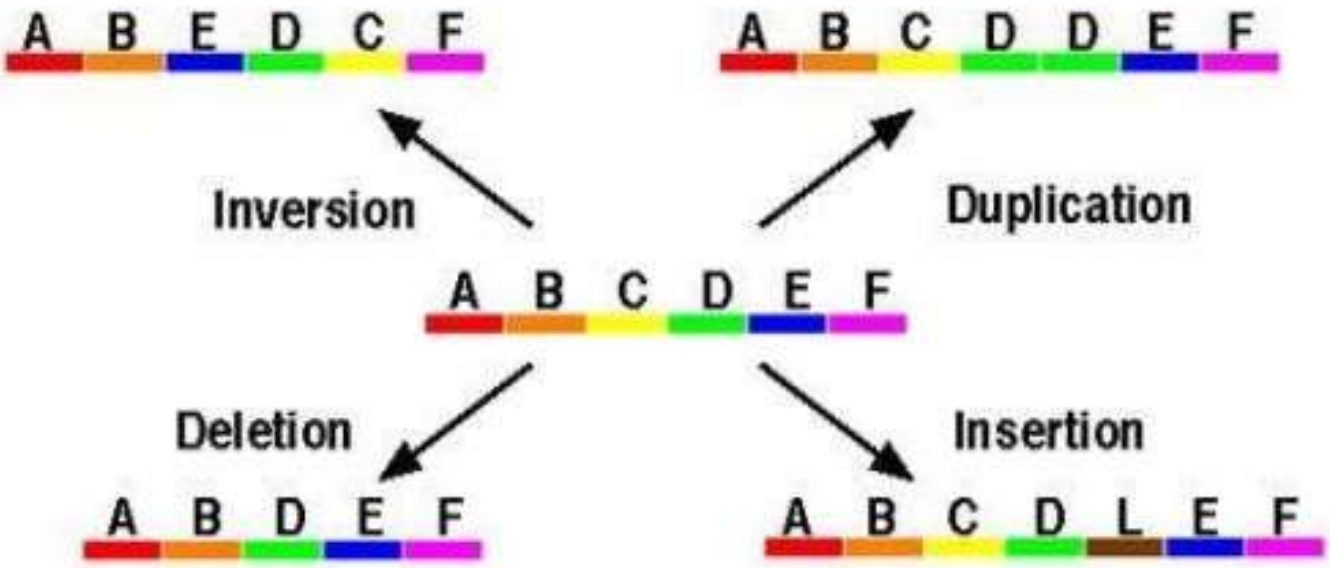
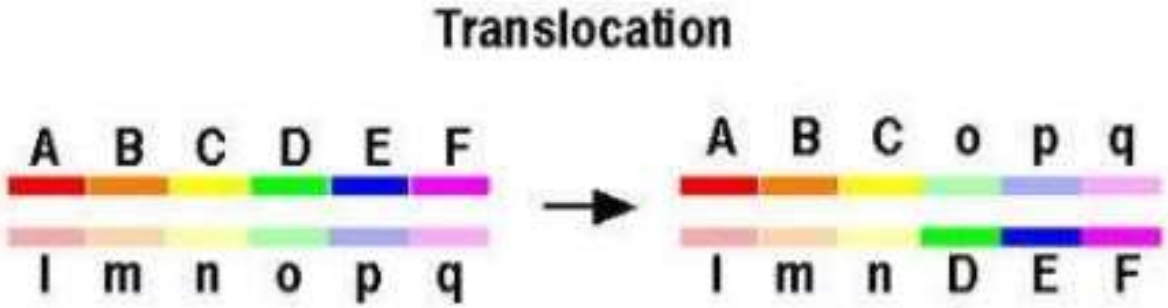
Shift

It requires at least three breaks in the chromosome. Among these, Reciprocal, and shift are the most common types.


A B E D C F
A B C D D E F
Inversion
Duplication
A B C D E F
Deletion
Insertion
A B D E F
A B C D L E F


Translocation
A B C D E F
l m n o p q
A B C o p q
l m n D E F

THANK YOU