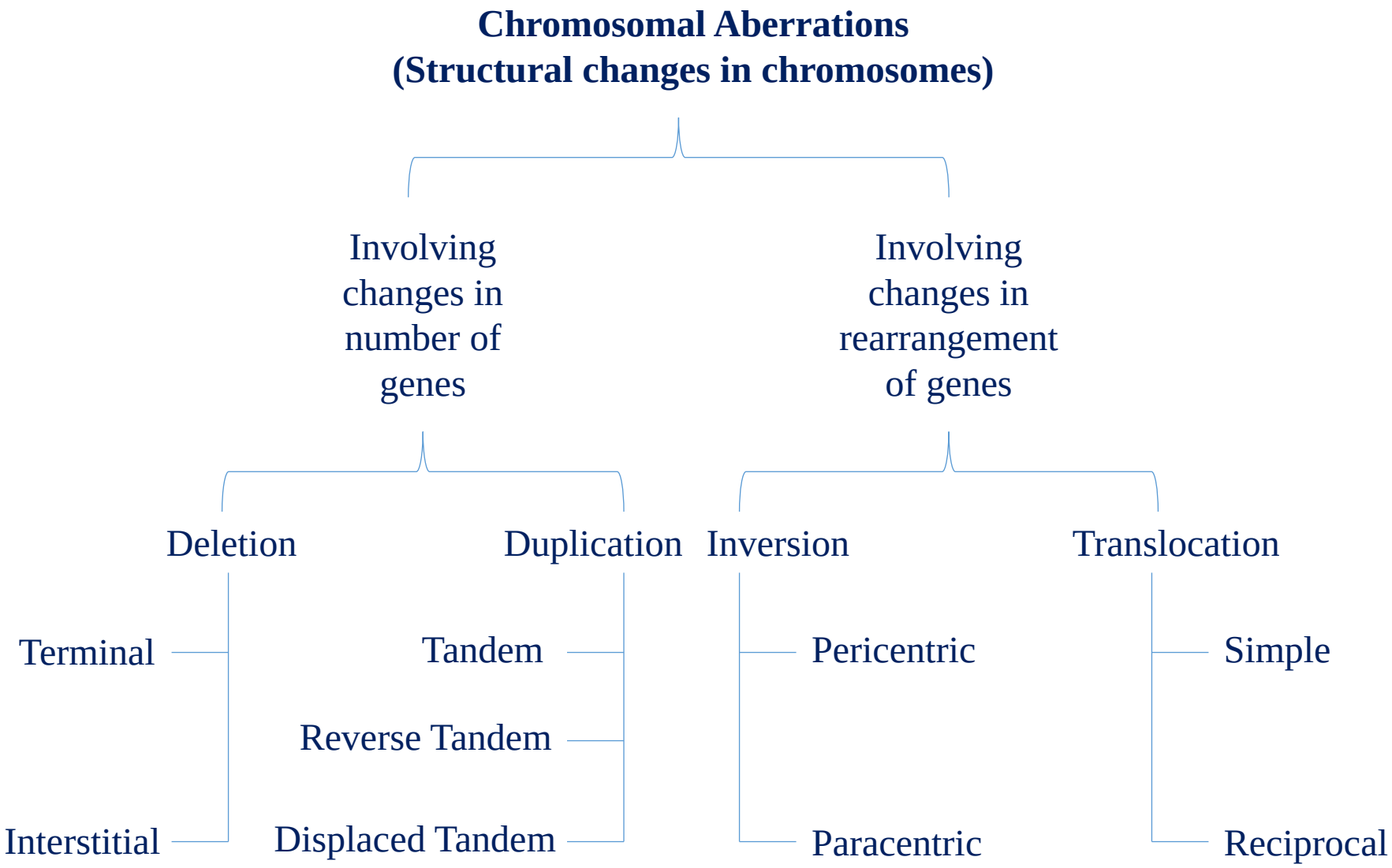
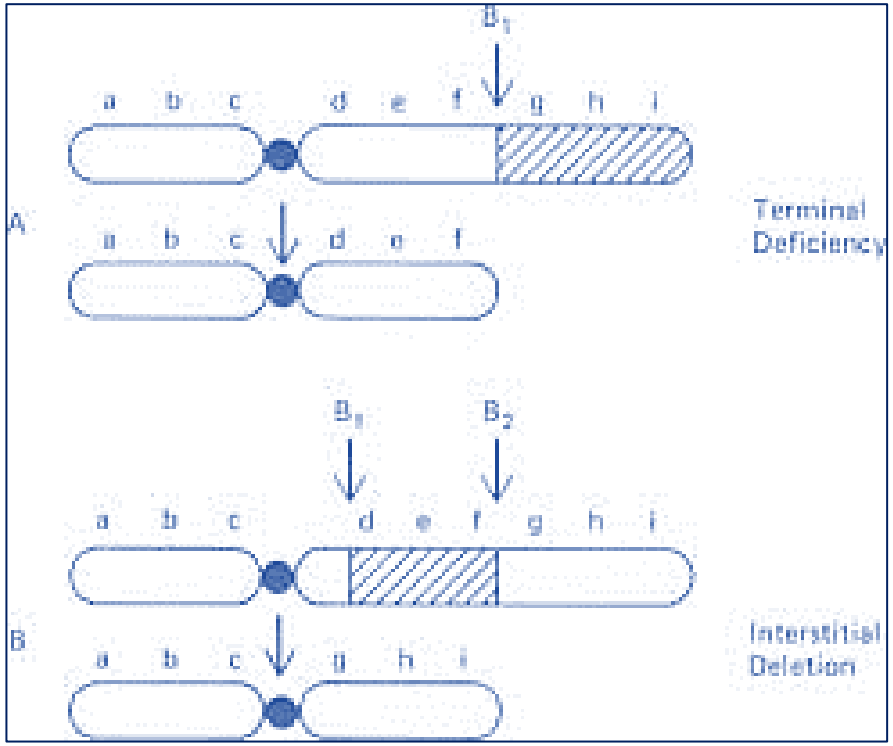
Chromosomal Aberration


Chromosomal Aberrations
(Structural changes in chromosomes)
Involving changes in number of genes
Involving changes in rearrangement of genes
Deletion
Duplication
Inversion
Translocation
Terminal
Interstitial
Tandem
Reverse Tandem
Displaced Tandem
Pericentric
Paracentric
Simple
Reciprocal

- **Deletion:**
- A missing chromosome segment is referred to either as a **deletion** or as a **deficiency.**
- Large deletions can be detected cytologically by studying the banding patterns in stained chromosomes, but small ones cannot.
- In a diploid organism, the deletion of a chromosome segment makes part of the genome hypoploid.
- This hypoploidy may be associated with a phenotypic effect, especially if the deletion is large.
- A classic example is the ***cri-du-chat*** **syndrome** (French words for “cry of the cat”) in humans:
- This condition is caused by a deletion in the short arm of chromosome 5.
- The size of the deletion varies.
- Individuals heterozygous for the deletion and a normal chromosome have the karyotype 46 del(5)(p14), where the terms in parentheses indicate that bands in region 14 of the short arm (p) of one of the chromosomes 5 is missing.
- These individuals may be severely impaired, mentally as well as physically; their plaintive, catlike crying during infancy gives the syndrome its name.

- Karyotype of a female with the *cri-du-chat* syndrome, 46,XX del(5)(p14):

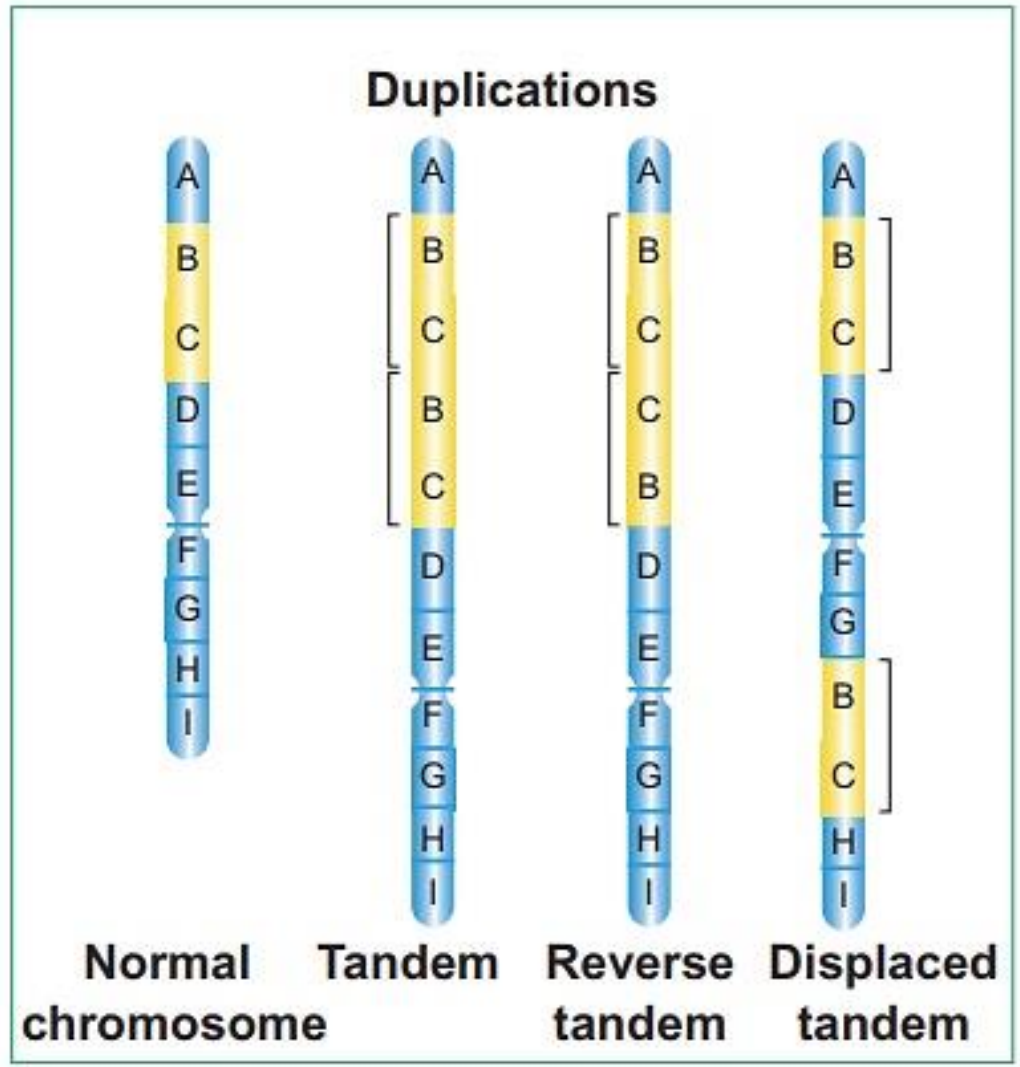

A
B1
a b c d e f g h i
a b c d e f
Terminal
Deficiency
B
B1
B2
a b c d e f g h i
a b c g h i
Interstitial
Deletion

- **Duplication:**
- An extra chromosome segment is referred to as a **duplication.**
- The extra segment can be attached to one of the chromosomes, or it can exist as a new and separate chromosome, that is, as a “free duplication.”
- In either case, the effect is the same: the organism is hyperploid for part of its genome.
- Duplication can be of three types-

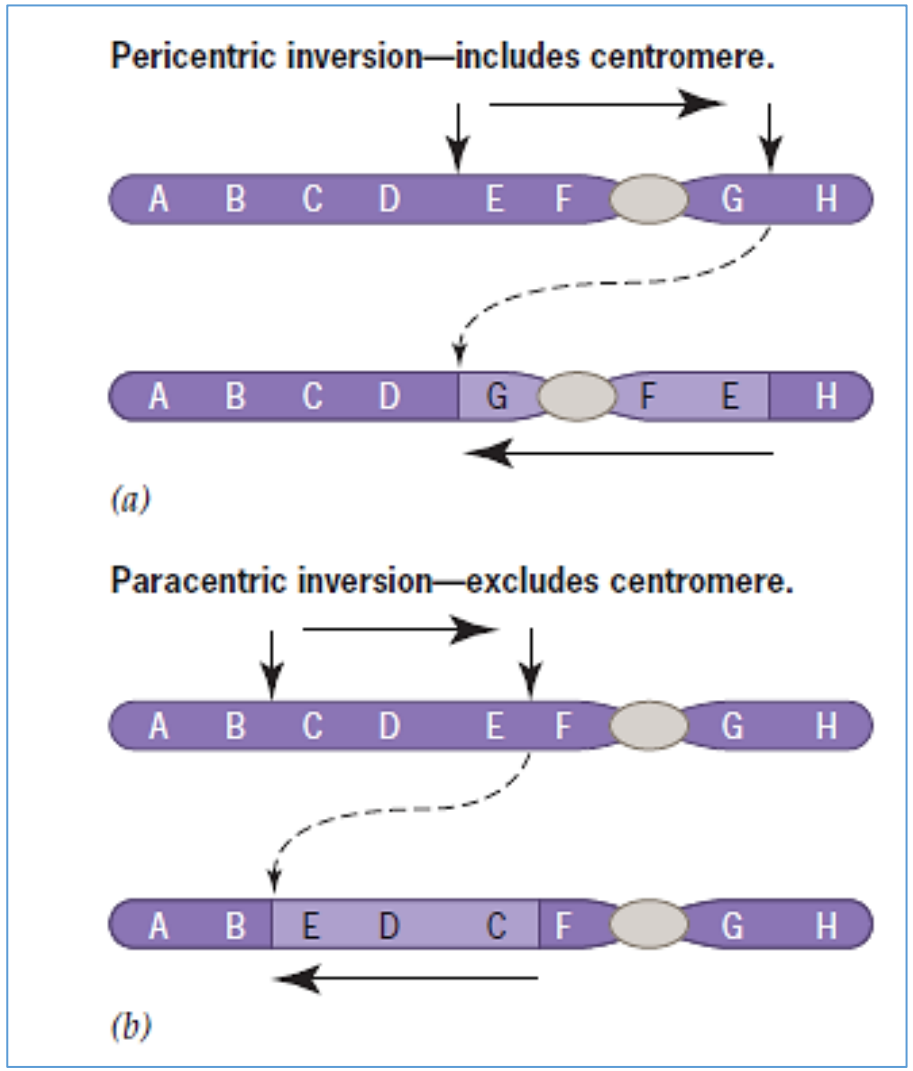
i. Tandem

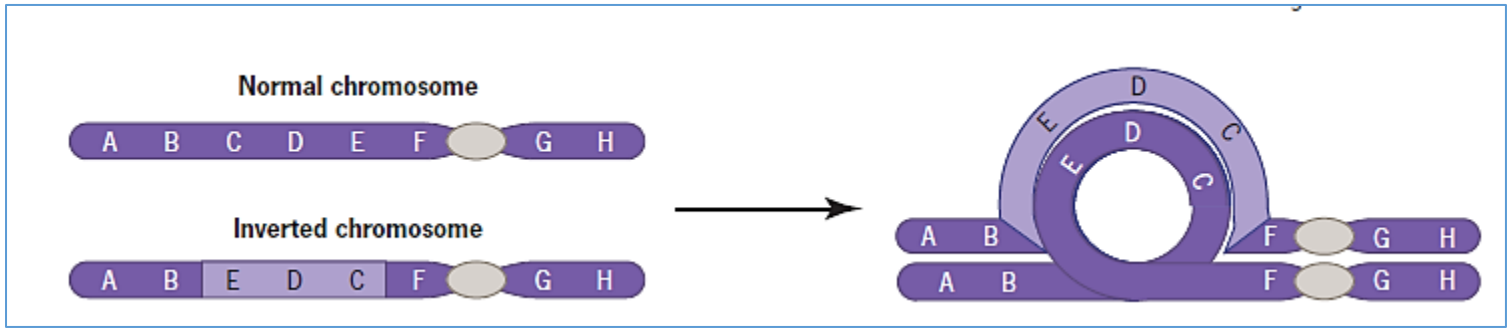
ii. Reverse Tandem &

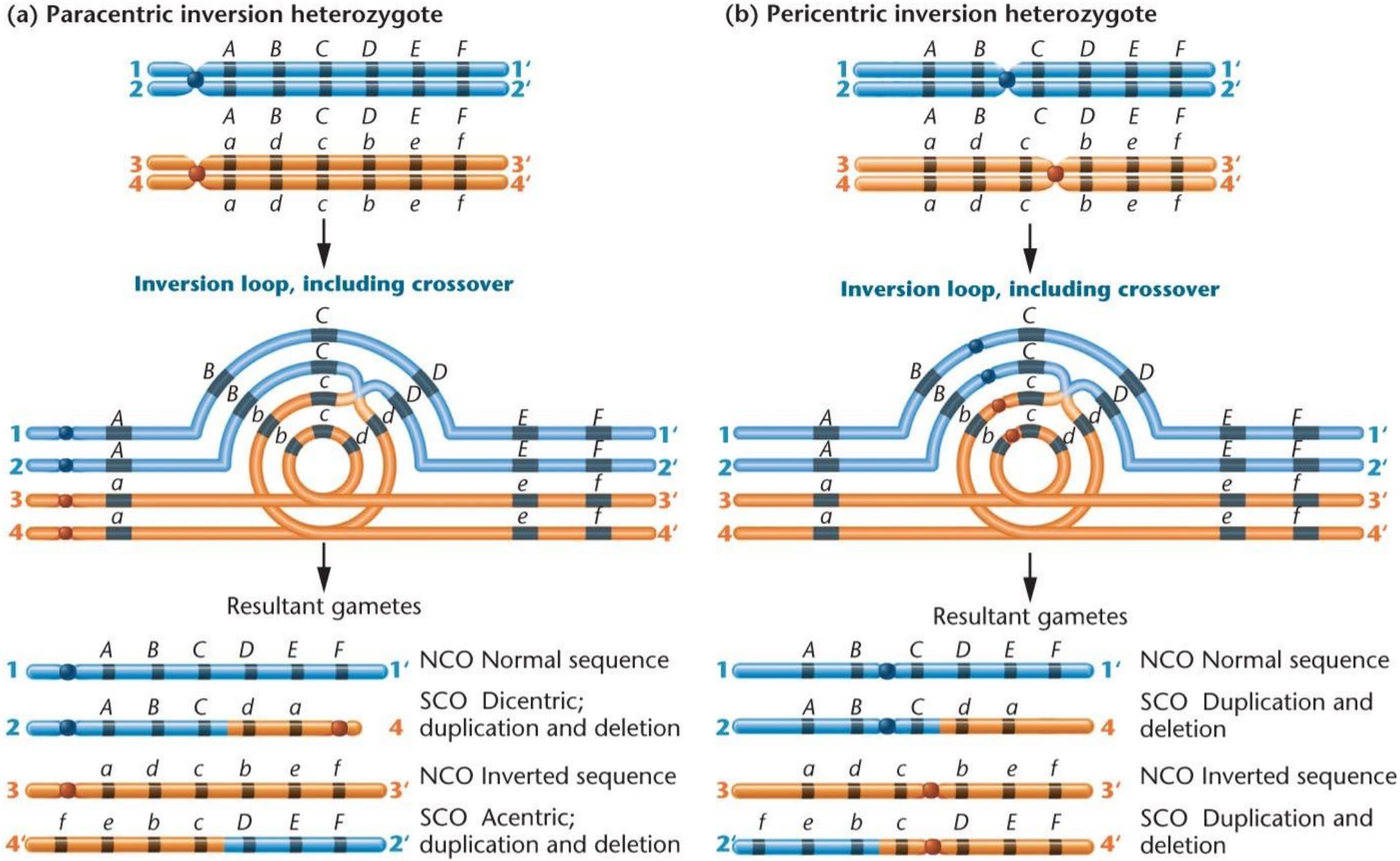
iii. Displaced Tandem.


Duplications
A
B
C
D
E
F
G
H
I
Normal chromosome
Tandem
Reverse tandem
Displaced tandem

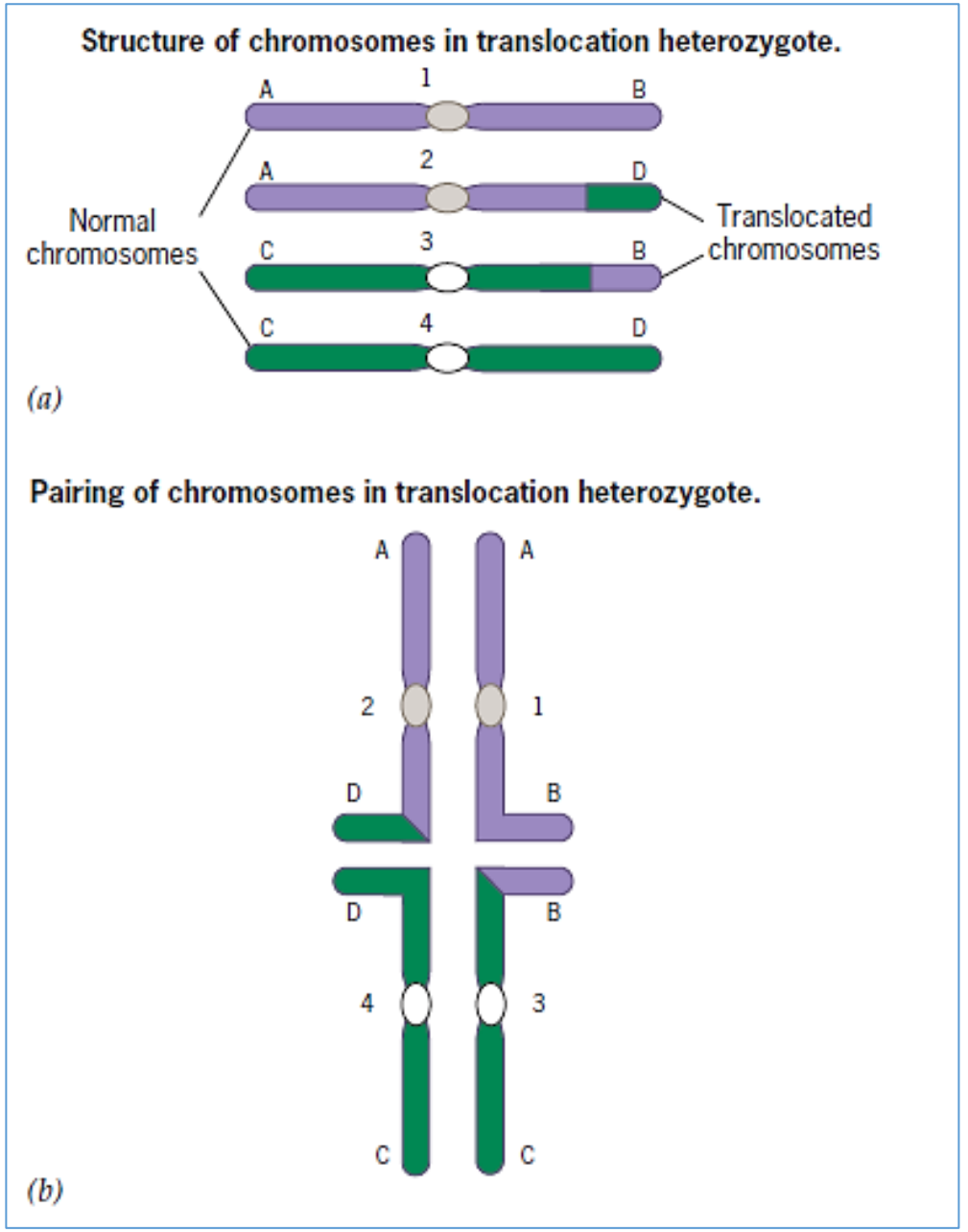
- **INVERSIONS**
- An **inversion** occurs when a chromosome segment is detached, flipped around 180°, and reattached to the rest of the chromosome.
- As a result, the order of the segment's genes is reversed.
- Such rearrangements can be induced in the laboratory by X-irradiation, which breaks chromosomes into pieces.
- Sometimes the pieces reattach, but in the process a segment gets turned around and an inversion occurs.
- There is also evidence that inversions are produced naturally through the activity of transposable elements.
- Cytogeneticists distinguish between two types of inversions based on whether or not the inverted segment includes the chromosome's centromere.
- **Pericentric** inversions include the centromere, whereas **paracentric** inversions do not.
- The consequence is that a pericentric inversion may change the relative lengths of the two arms of the chromosome, whereas a paracentric inversion has no such effect.
- Thus, if an acrocentric chromosome acquires an inversion with a breakpoint in each of the chromosome's arms (that is, a pericentric inversion), it can be transformed into a metacentric chromosome.
- However, if an acrocentric chromosome acquires an inversion in which both of the breaks are in the chromosome's long arm (that is, a paracentric inversion), the morphology of the chromosome will not be changed.

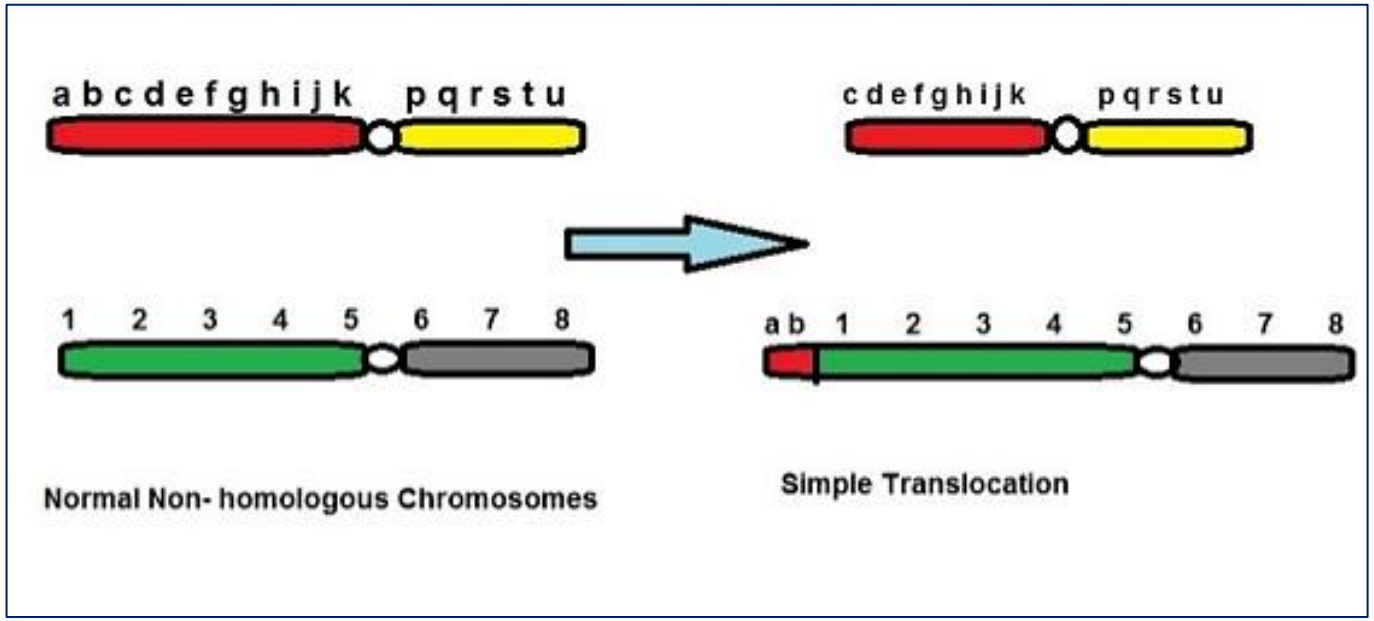

Pericentric inversion—includes centromere.
A B C D E F G H
A B C D G F E H
(a)
Paracentric inversion—excludes centromere.
A B C D E F G H
A B E D C F G H
(b)

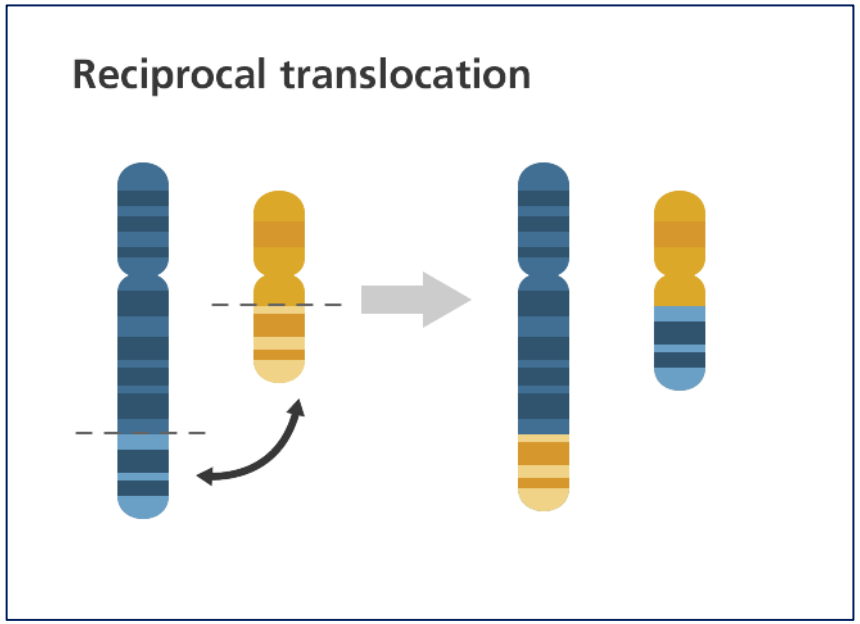

Normal chromosome
A B C D E F G H
Inverted chromosome
A B E D C F G H
A B F G H
A B F G H

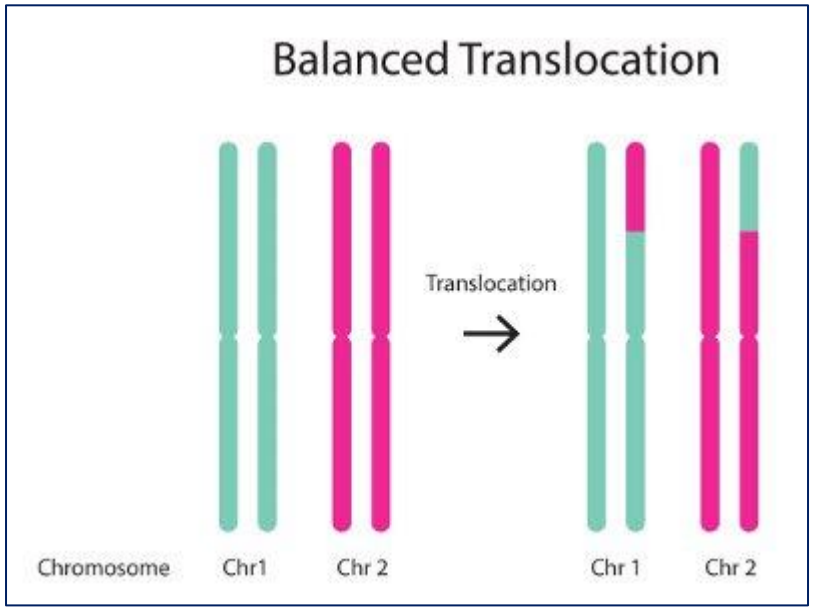

(a) Paracentric inversion heterozygote
(b) Pericentric inversion heterozygote
Inversion loop, including crossover
Resultant gametes
NCO Normal sequence
SCO Dicentric; duplication and deletion
NCO Inverted sequence
SCO Acentric; duplication and deletion
SCO Duplication and deletion

- **TRANSLOCATIONS**
- A **translocation** occurs when a segment from one chromosome is detached and reattached to a different (that is, nonhomologous) chromosome.
- The genetic significance is that genes from one chromosome are transferred to another.
- When pieces of two non-homologous chromosomes are interchanged without any net loss of genetic material, the event is referred to as a reciprocal translocation.
- During meiosis, these translocated chromosomes would pair with their untranslocated homologues in a cruciform, or crosslike, pattern.
- Translocation can be of two types:
- Simple translocation or
- Reciprocal translocation

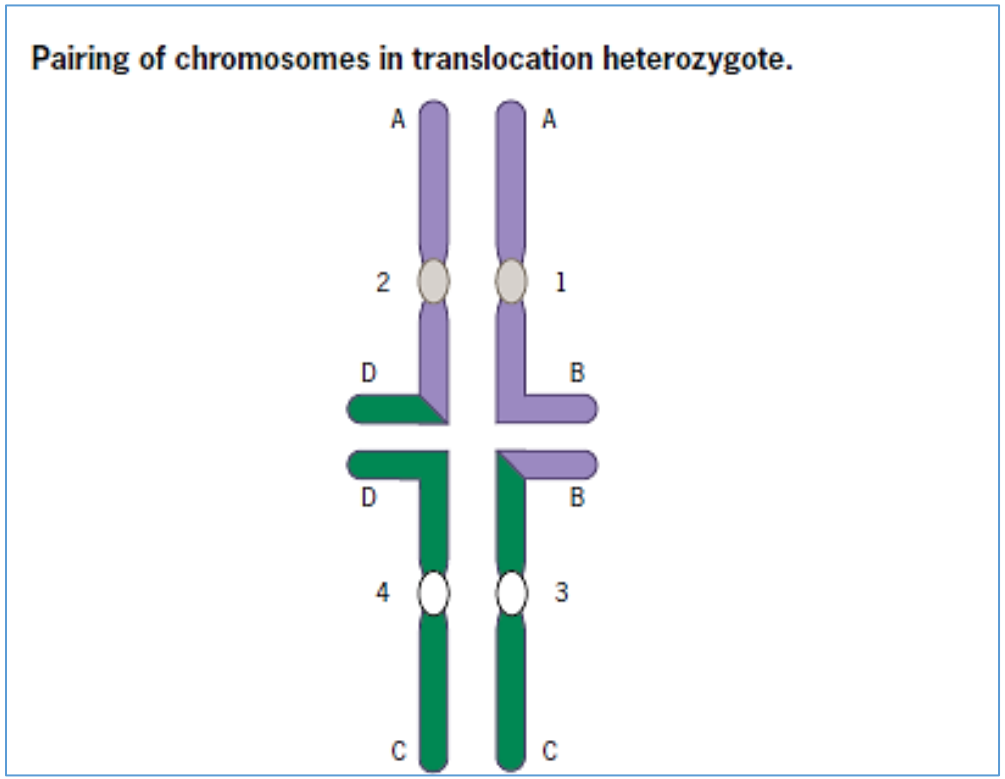


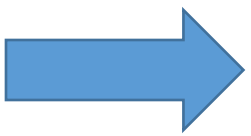

a b c d e f g h i j k
p q r s t u
c d e f g h i j k
p q r s t u
1 2 3 4 5 6 7 8
a b 1 2 3 4 5 6 7 8
Normal Non- homologous Chromosomes
Simple Translocation

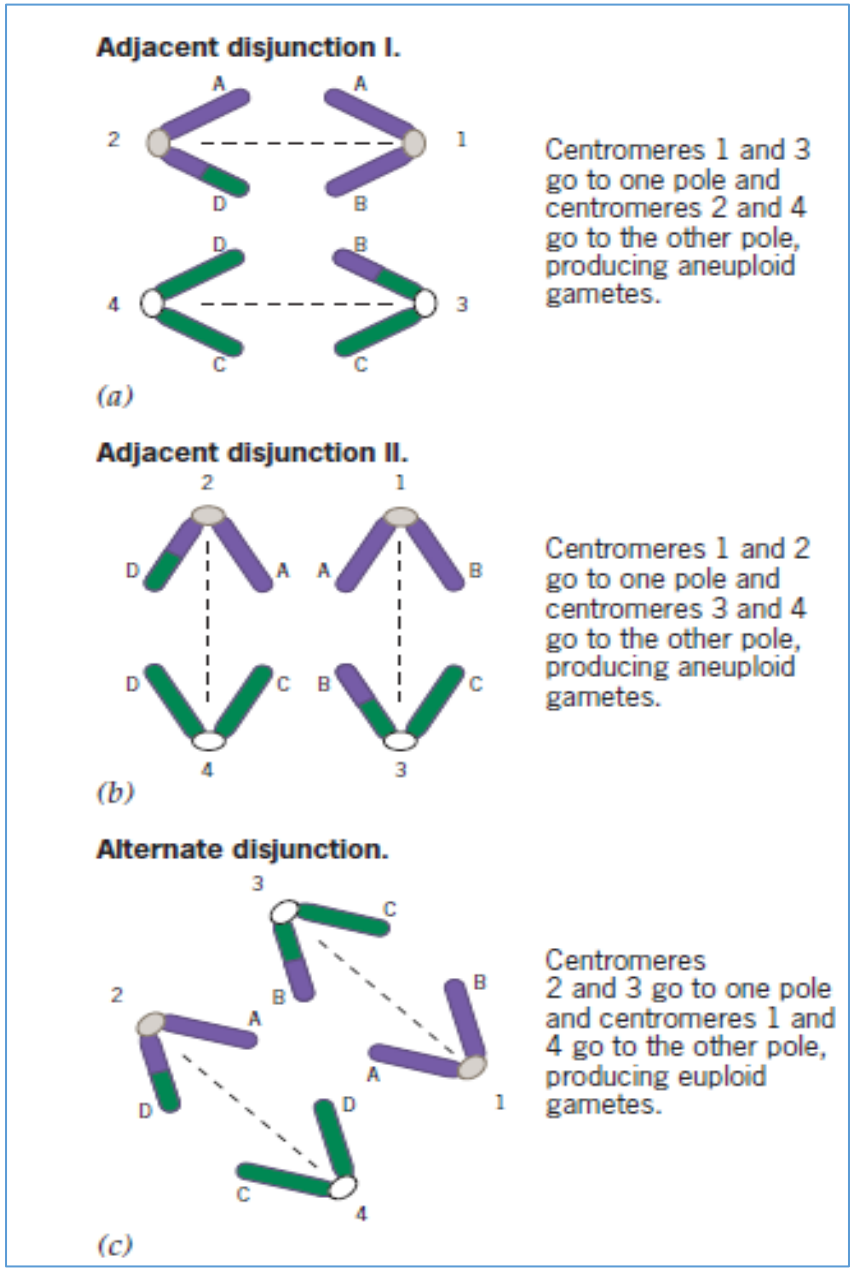

Reciprocal translocation

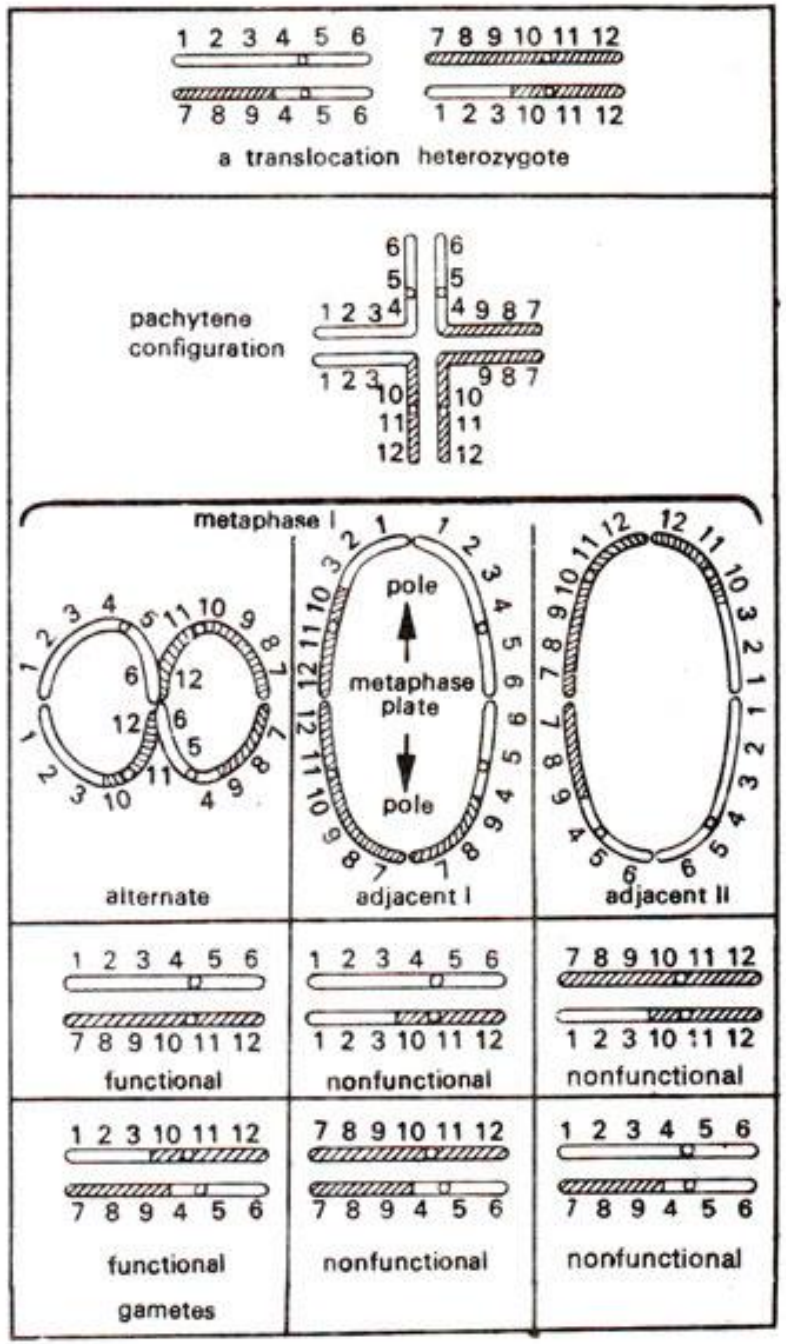

Balanced Translocation
Translocation
Chromosome
Chr1
Chr 2
Chr 1
Chr 2

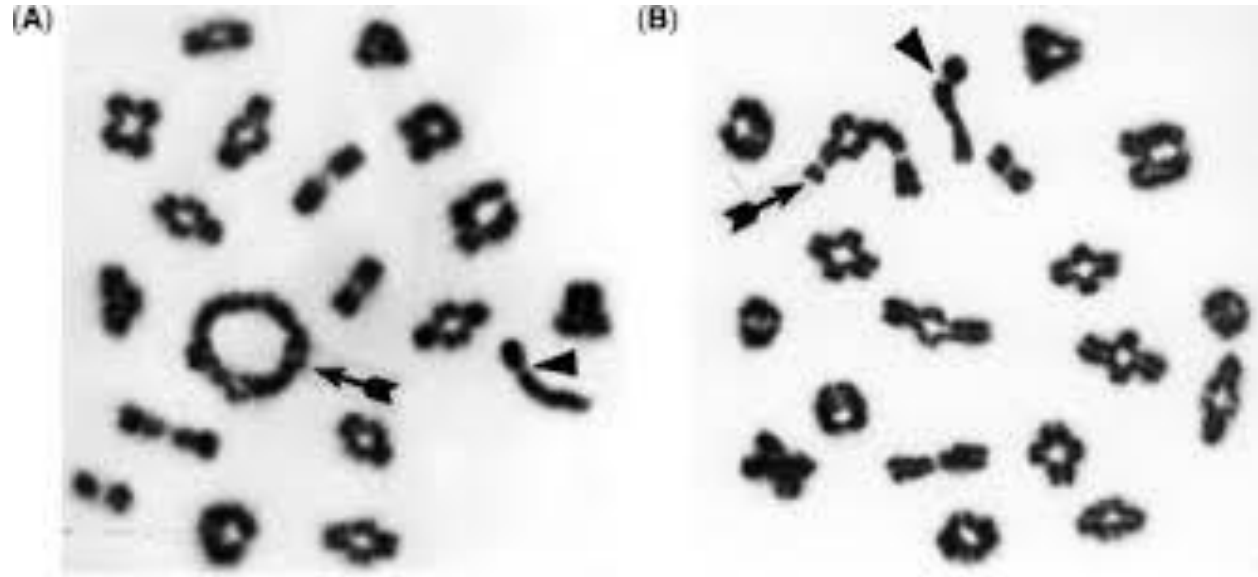
- Types of disjunction in a translocation heterozygote during meiosis:








1 2 3 4 5 6
7 8 9 10 11 12
7 8 9 4 5 6
1 2 3 10 11 12
a translocation heterozygote
pachytene configuration
metaphase I
pole
metaphase plate
pole
alternate
adjacent I
adjacent II
functional
nonfunctional
nonfunctional
functional gametes
nonfunctional
nonfunctional


(A)
(B)

- Nonhomologous chromosomes can also fuse at their centromeres, creating a structure called a **Robertsonian Translocation.**
- It was named after the cytologist F. W. Robertson.
- For example, if two acrocentric chromosomes fuse, they will produce a metacentric chromosome; the tiny short arms of the participating chromosomes are simply lost in this process.