

Chromosomal Aberrations

“ Substantial changes in chromosome structure are called chromosomal aberrations”

*There are TWO primary ways in which the structure of chromosomes can be altered

1-The total amount of genetic information in the chromosome can change ;

- * Decrease : Deficiencies / Deletions
- * Increase : Duplications / Insertions

Chromosomal Aberrations

2-The genetic material may remain the same , but is rearranged;

- * Inversions

- * Translocations

Alterations in Chromosome structure

- **Deletion** : Loss of a chromosomal segment .

- **Duplication** : Repetition of a chromosomal segment .

- **Inversion** : A change in the direction of genetic material along a single chromosome.

Chromosomal Aberrations

- **Translocation** : A segment of one • chromosome becomes attached to a non homologous chromosome

- * **Simple Translocations**

- one way transfer

- * **Reciprocal Translocations**

- Two way transfer

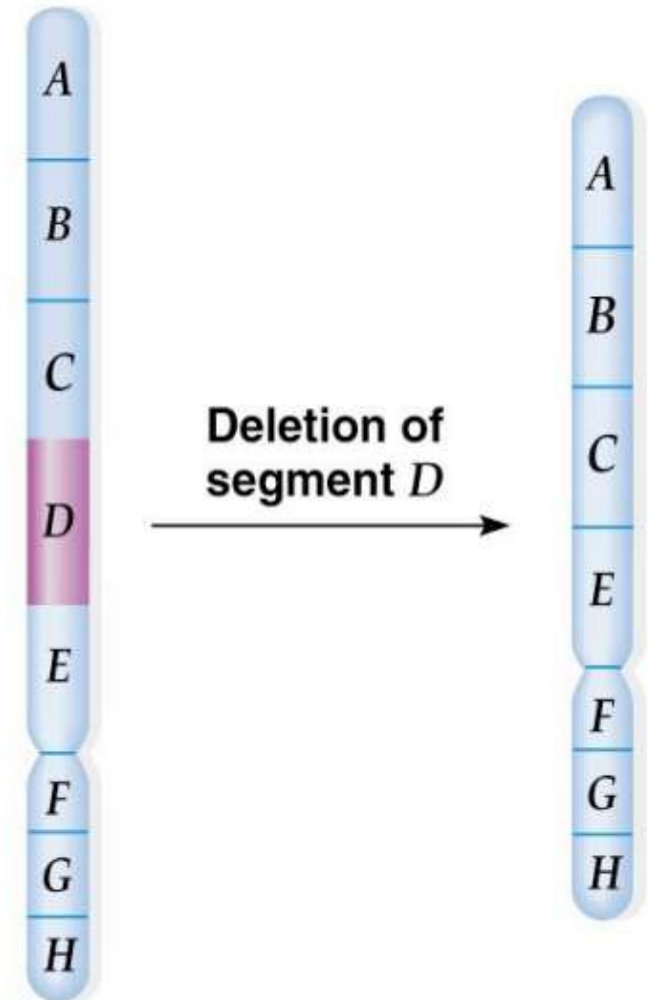
Deletions

Part of chromosome is missing

-Causes Of Deletions

- * Heat or Radiation
(especially ionization)
- * Chemicals
- * Viruses
- * Errors in recombination

-Deletions do not revert -
because the DNA is degraded



A deletion of a chromosome segment

Deletions

-Loss of several genes can cause lethal consequences ; FOR EXAMPLE ,

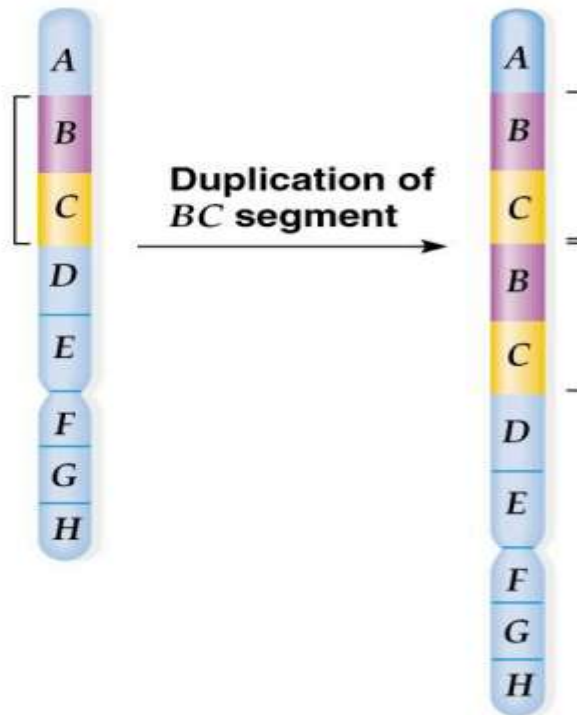
-Human disorders caused by large chromosomal deletions

-**Cry-du-chat** (cry of the cat) syndrome resulting from deletion of part of the short arm of **chromosome 5**.

-The deletion results in several mental retardation and physical abnormalities , For Example ,Microcephaly

Duplications

-Duplications result from doubling of chromosomal segments , and occur in a range of sizes and locations .



Duplication, with a chromosome segment repeated

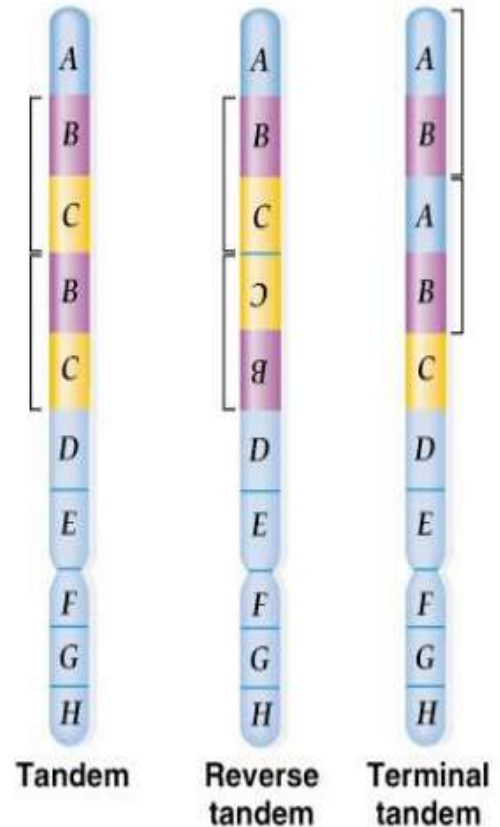
Duplication

- **Tandem Duplications** are adjacent to each other.
- **Reverse Tandem Duplications** result in genes arranged in opposite order of the original.
- Tandem duplication at the end of chromosome is a **Terminal tandem duplication**

Normal chromosome

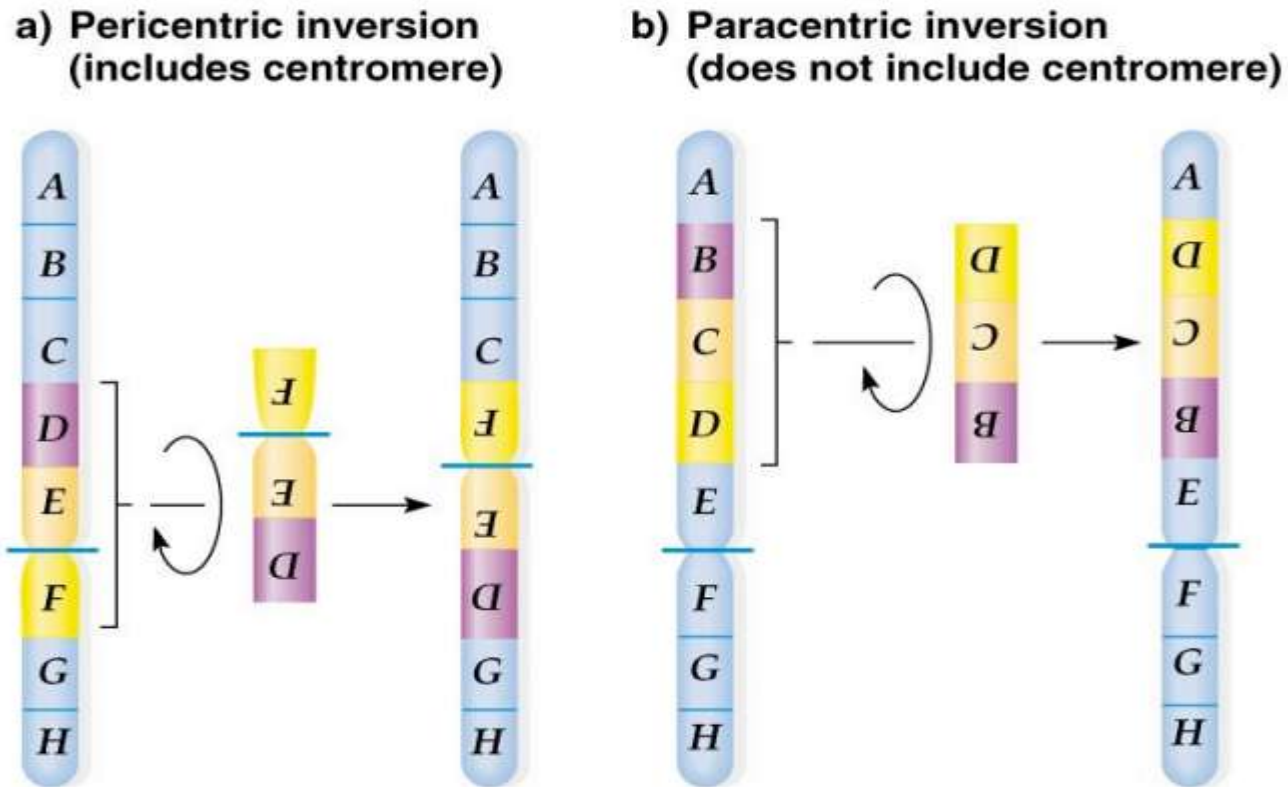


Duplications



Inversions

-Inversions results when a chromosomal segment excises and reintegrates , oriented 180 degree from the original orientation .



Inversion

*In an Inversion , the total amount of genetic information stays the same. •

-Therefore ,the great majority of inversions have no phenotypic consequences •

*In rare cases, inversions can alter the phenotype of an individual, •

-Break point effect :

-The breaks leading to the inversions occur in a vital gene •

•

Inversions

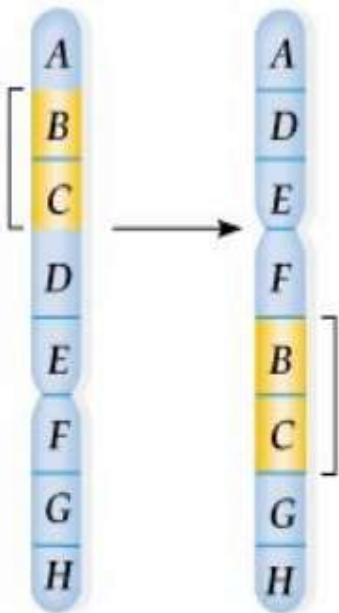
- Position effect : •
- A gene is repositioned in a way that alters its gene expression •
- About 2 % of the human population carries inversions that are detectable with a light microscope. •
- most of these individuals are phenotypically normal •
- However , a few produce offspring with genetic abnormalities •

Translocations

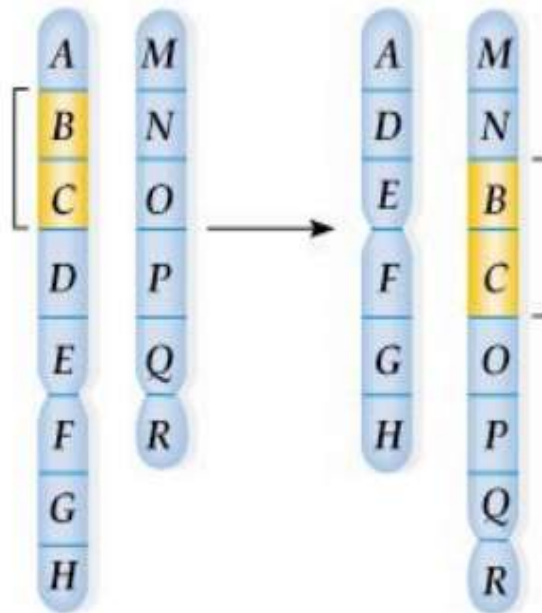
- Translocation occurs when a segment of one chromosome becomes attached to another. •
- There are two main types of translocations ;
 - * **Reciprocal** (balanced) **translocations**
 - * **Robertsonian**(unbalanced) **translocations**
- Both types of translocations can cause disease in humans.

Translocations

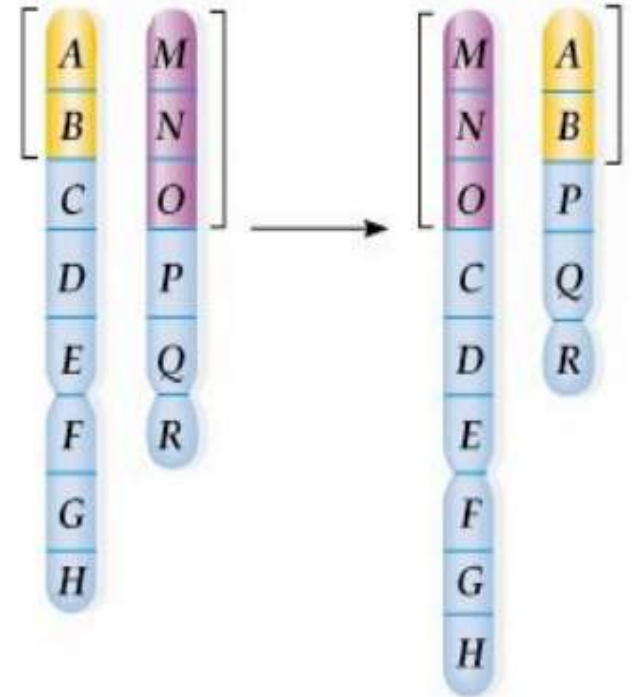
a) Nonreciprocal intrachromosomal translocation



b) Nonreciprocal interchromosomal translocation



c) Reciprocal interchromosomal translocation



Reciprocal Translocations

-In Reciprocal translocations two non-homologous Chromosomes exchange genetic material.

-Reciprocal translocation arise from two different mechanisms ;

1-chromosomal breakage and DNA repair

2-Abnormal crossovers

-Reciprocal translocations lead to the re-arrangement of the genetic material,

-not a change in the total amount

-thus , they are also called **balanced translocations**

Robertsonian Translocations

- In **Robertsonian translocations** the transfer of genetic material occurs in only one direction. •
- Robertsonian translocations are associated with phenotypic abnormalities or even lethality.
- EXAMPLE :Familial Down Syndrome
- in this condition , the majority of chromosome 21 is attached to chromosome 14 .
- The individual would have three copies of genes found on a large segment of chromosome 21

Robertsonian Translocations

- Robertsonian translocations are confined to • chromosomes 13 , 14 , 15 , 21
- Acrocentric Chromosomes

TAUQEER AHMAD

BS-Zoology

Govt. Murray College

Sialkot