# Chromosomal Abnormalities

By :Sahar AlSubaie

# Chromosome Anomaly:

- Abnormal chromosomes' number or structure.

- can be organized into two basic groups:
  - numerical anomalies.
  - structural anomalies.

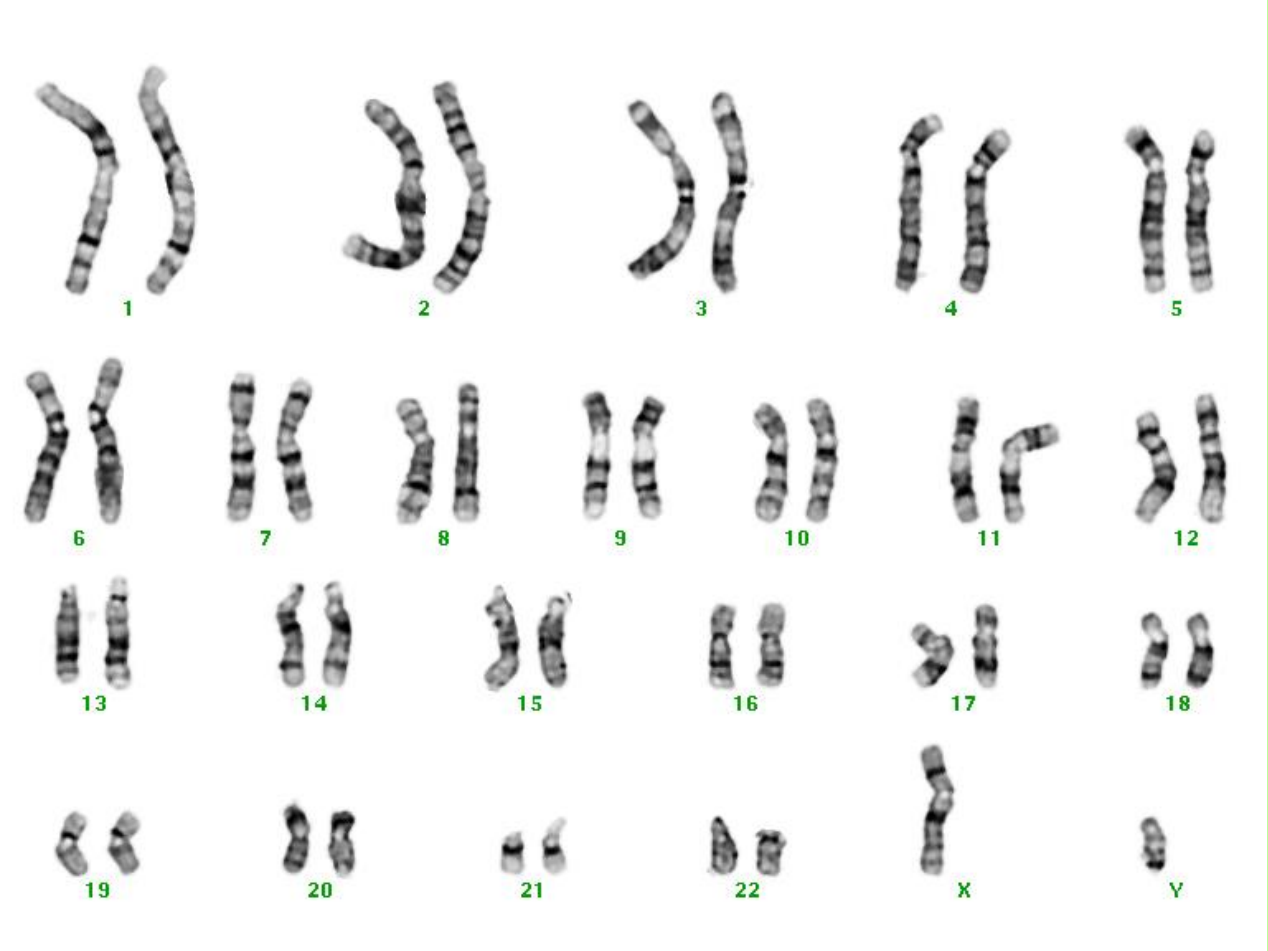
- Occurs due to error in cell division following:

meiosis or mitosis.

# Numerical Abnormalities:

- Aneuploidy (an abnormal number of chromosomes).
- missing either a chromosome from a pair (monosomy)
- has more than two chromosomes of a pair (Trisomy, Tetrasomy, etc).
- Euploidy it is of three types : monoploidy (1n), diploide (2n), polyploidy.

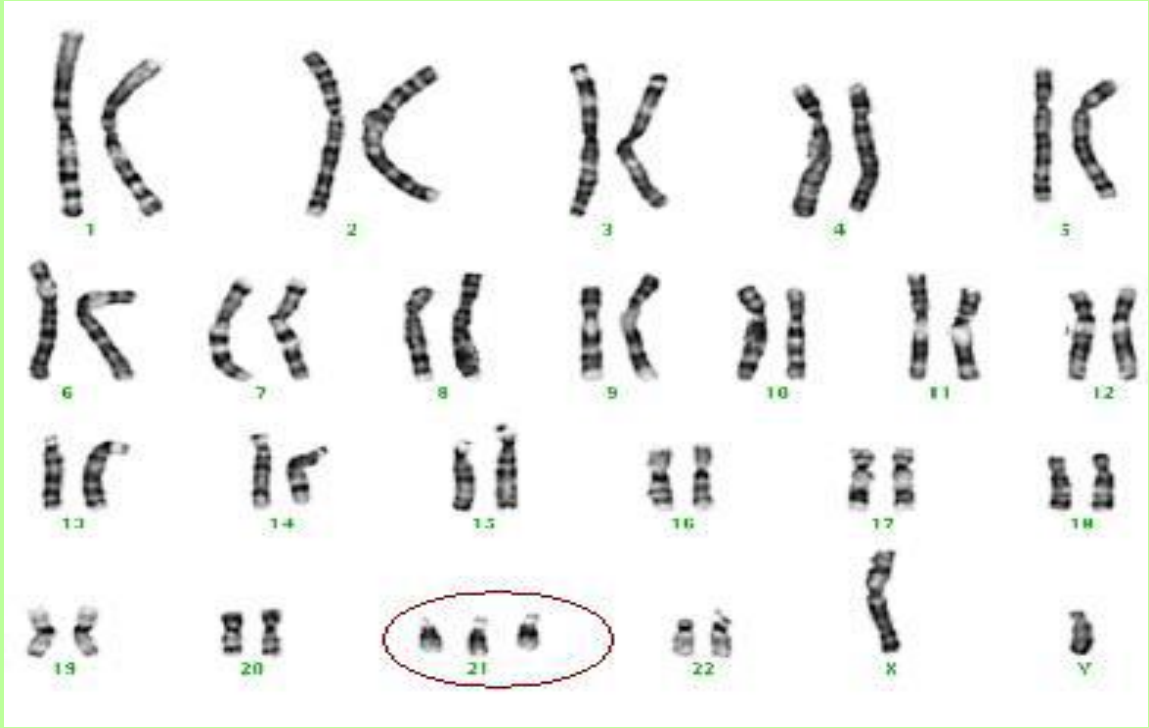
# Normal Karyotype :



# Numerical Abnormalities:

- Down Syndrome

  Trisomy 21

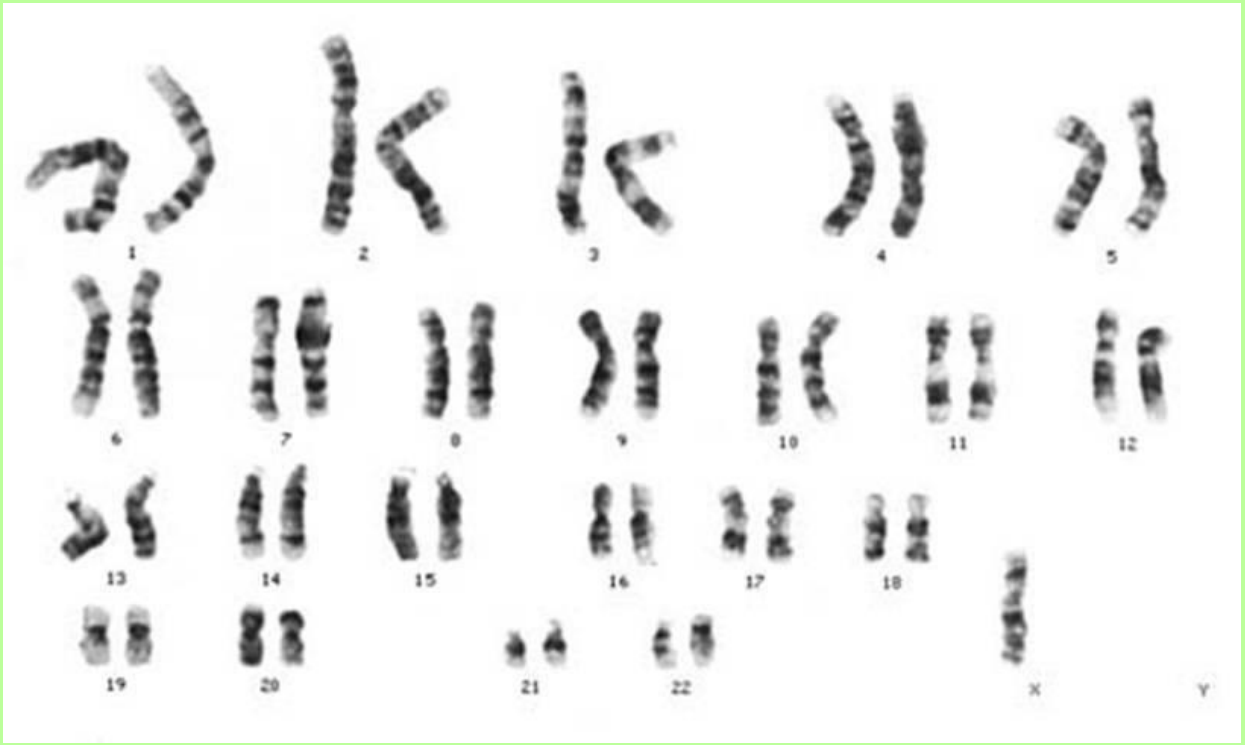



47,XY,+21 Down syndrome karyotype

# Numerical Abnormalities:

- Turner Syndrome

  monosomy ( one sex chromosome, an X )

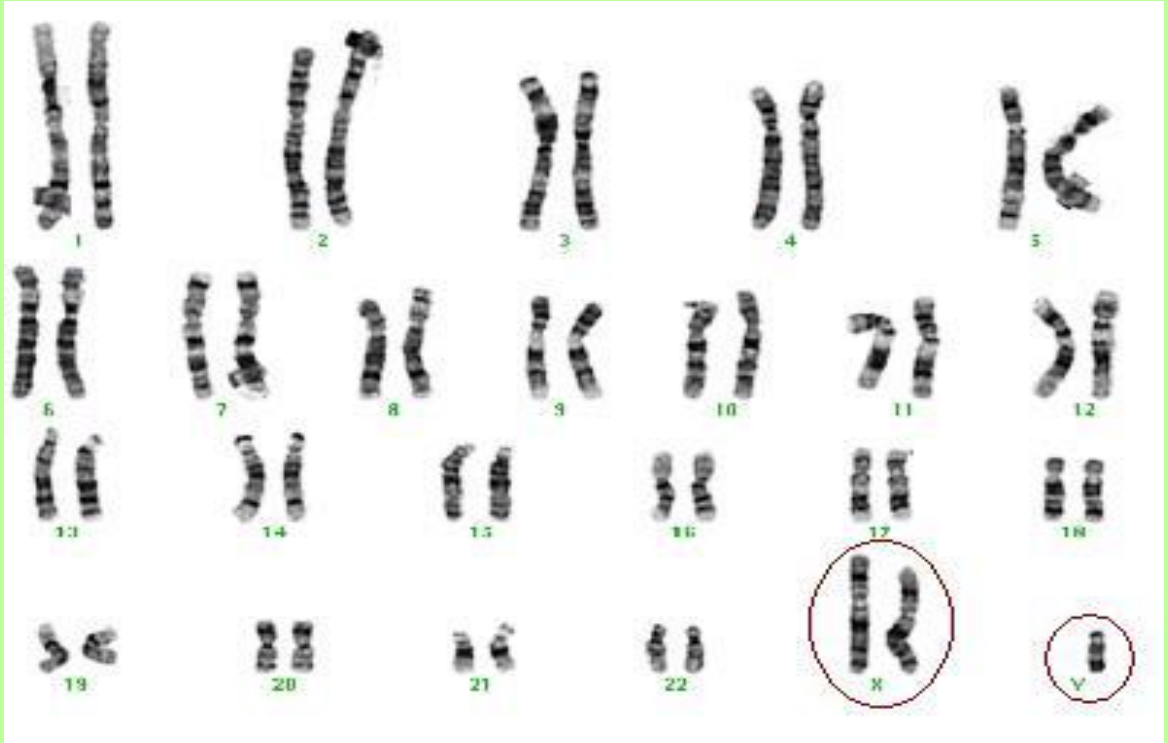


45,X Turner syndrome karyotype.

# Numerical Abnormalities:

- Klinefelter's syndrome.

  47, XXY, or XXY syndrome



47,XXY Klinefelter syndrome karyotype

# Structural Abnormalities:

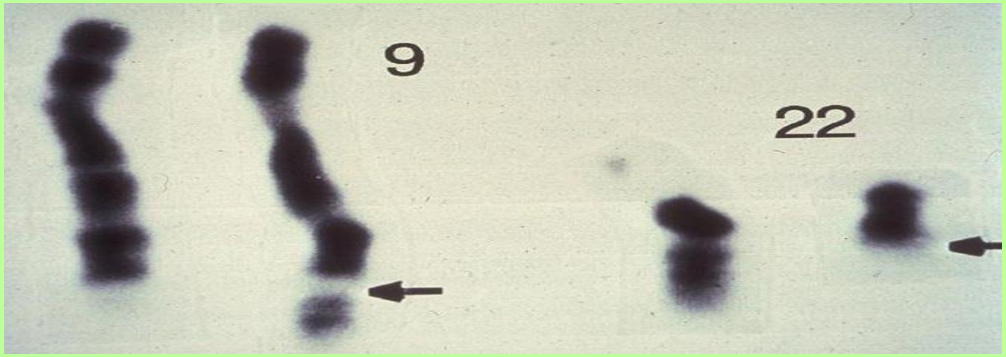
- Translocation
- Inversion
- Deletion
- Duplication
- Ring
- Isochromosomes

# Translocation:

- portion of one chromosome is transferred to another chromosome.
- Two types:
  - Reciprocal translocation.
  - Robertsonian translocation.

# Reciprocal translocation:

- segments from two different chromosomes have been exchanged.
- More common than Robertsonian translocation
- Occurs in  any chromosome at any point
- Phenotypically normal .

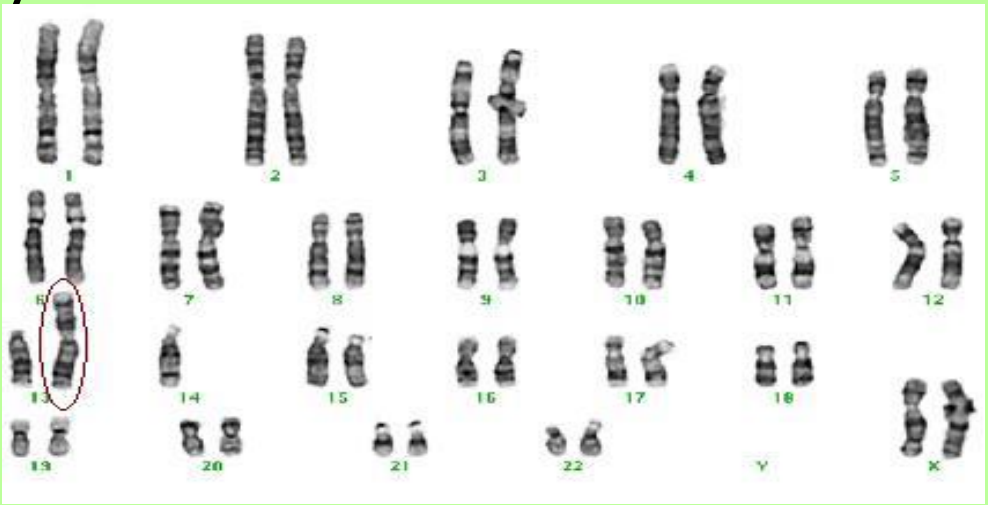



Philadelphia chromosome

PeterC.Nowell,MD,DepartmentofPathologyandClinicalLaboratoryoftheUniversityofPennsylvaniaSchoolofMedicine,1960

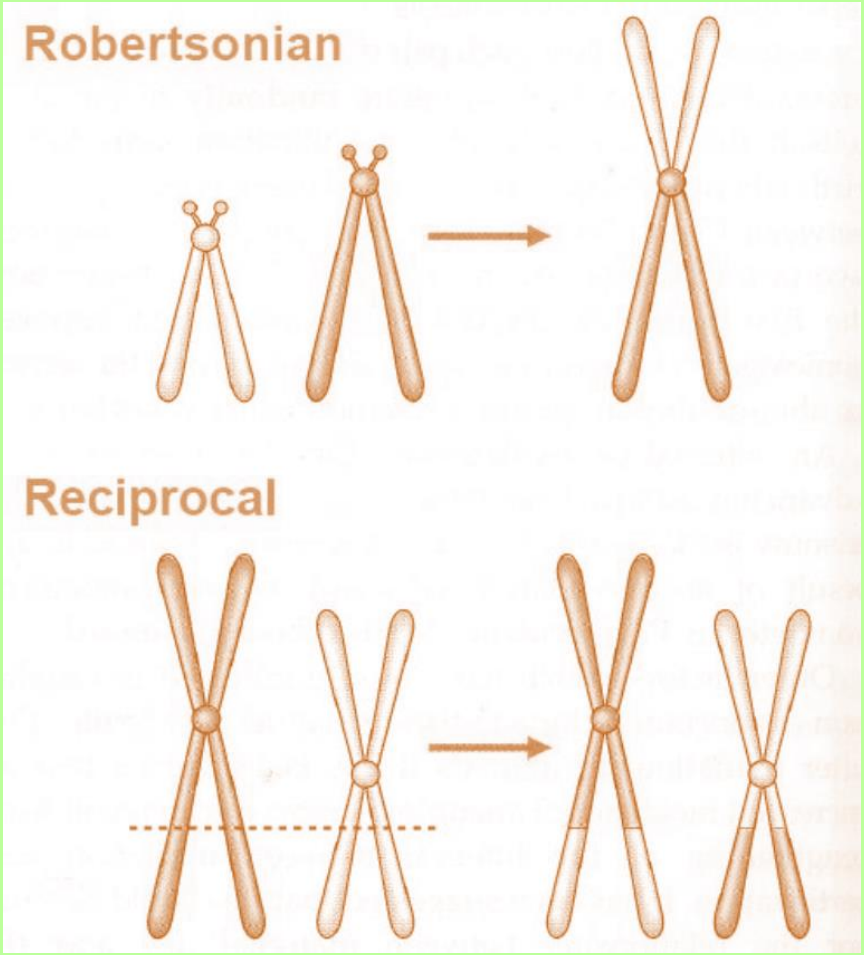
# Robertsonian translocation:

- an entire chromosome has attached to another at the Centromere
- in humans these only occur with chromosomes 13, 14, 15, 21 and 22.
- Phenotypically normal .



Robertsonian translocation 45,XX,der(13;14)(q10;q10).

# Translocation:

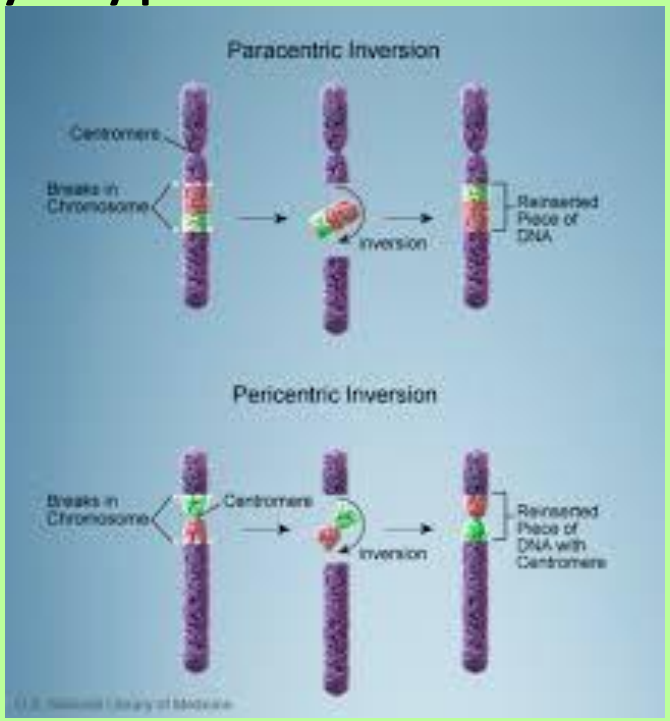


# Inversion:

- portion of the chromosome has broken off, turned upside down and reattached, therefore the genetic material is inverted
- If too small, cant detected by karyotype
- Very rare in humans
- Paracentric:

  segments of the same arm
- Pericentric:

  around the centromere



# Deletion:

A portion of the chromosome is missing or deleted.

Wolf-Hirschhorn syndrome

partial deletion of the short arm of chromosome 4

Jacobsen syndrome

also called the terminal 11q deletion disorder.

# Duplications:

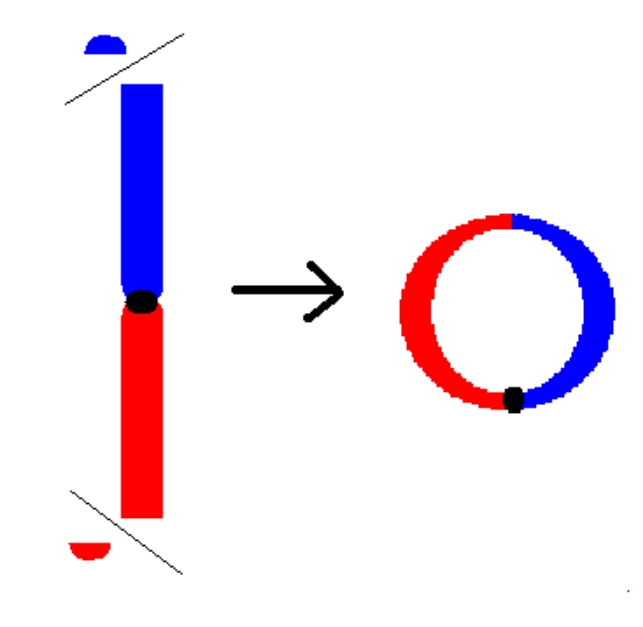
A portion of the chromosome is duplicated, resulting in extra genetic material.

Charcot-Marie-Tooth disease type 1A

duplication of the gene encoding peripheral myelin protein 22 (PMP22) on chromosome 17.

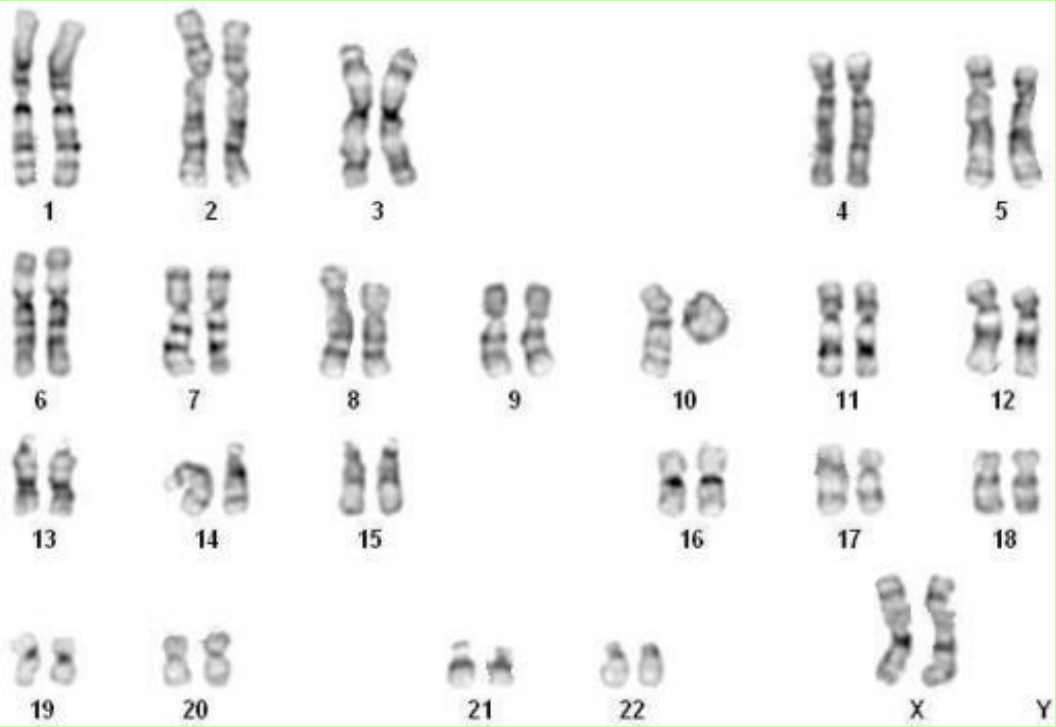
# Ring chromosome:

- A portion of a chromosome has broken off and formed a circle or ring .
- with or without loss of genetic material.



Formation of a ring chromosome.

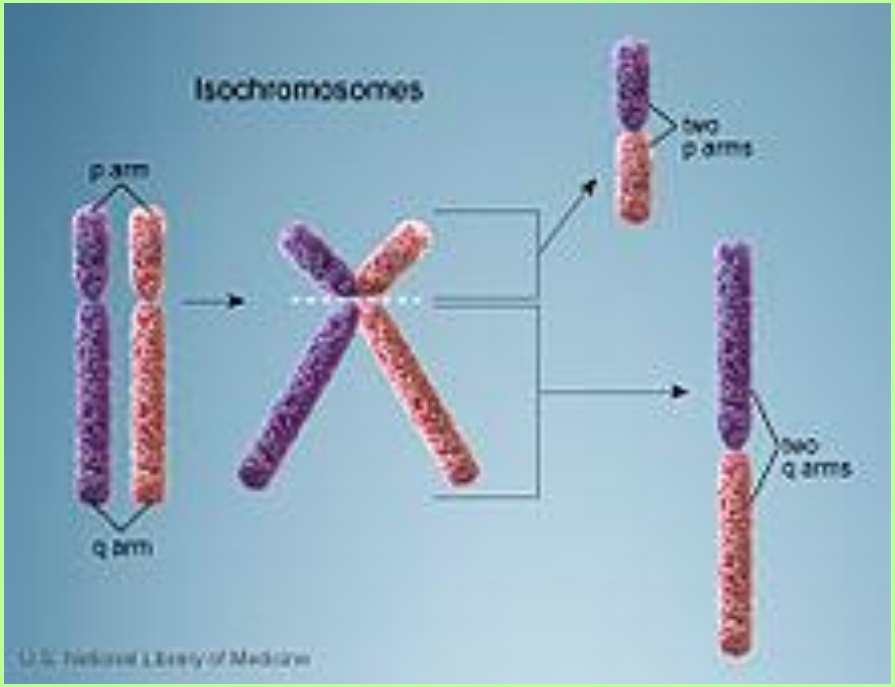
# Ring chromosome:




46,XX,r(10)

# Isochromosome:

Formed by the mirror image copy of a chromosome segment including the centromere.



# Chromosome instability syndromes:

a group of disorders characterized by chromosomal instability and breakage. They often lead to an increased tendency to develop certain types of malignancies.

# Chromosome Nomenclature:

- International System for Human Cytogenetic Nomenclature (ISCN):
  - Number of chromosomes.
  - Structural abnormality of chromosomes.
  - Numerical abnormality of chromosomes.

# Chromosome Nomenclature:

- del - deletion
- dic - dicentric
- fra - fragile site
- i - isochromosome
- inv - inversion
- p - short arm
- r - ring
- der - derivative
- dup - duplication
- h - heterochromatin
- ins - insertion
- mat - maternal origin
- q - long arm
- t - translocation