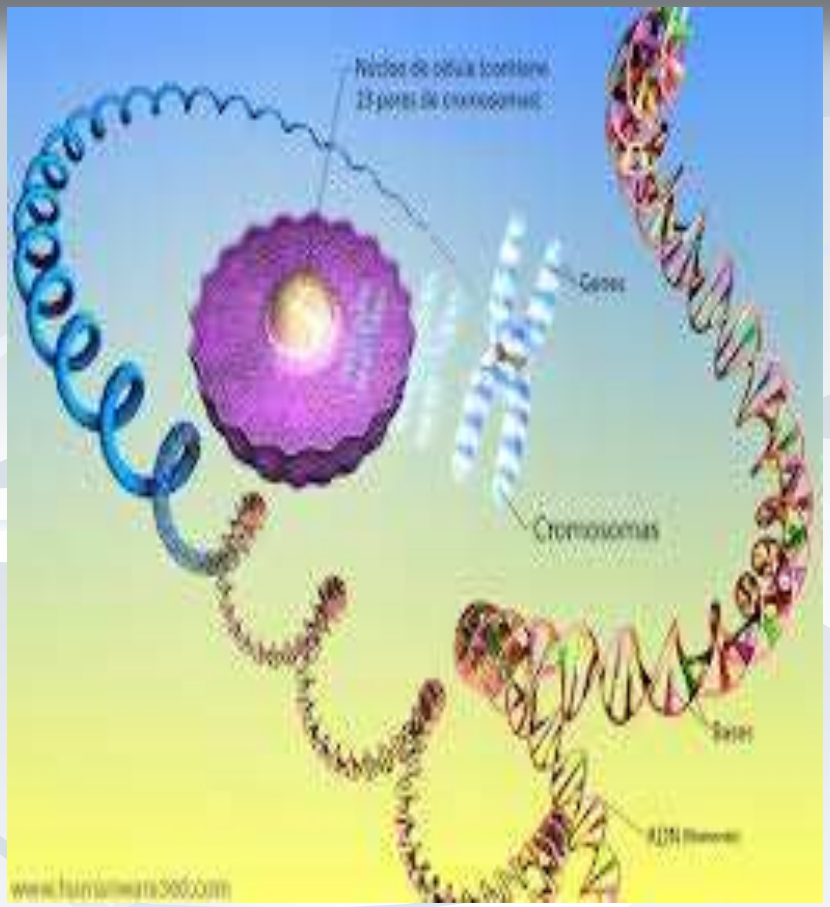


CYTOGENETIC & CHROMOSOME

By;

Lecturer

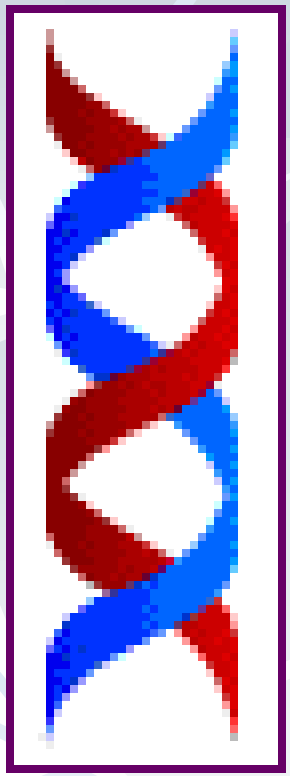
AMAL M. ALI

2016

Cytogenetics

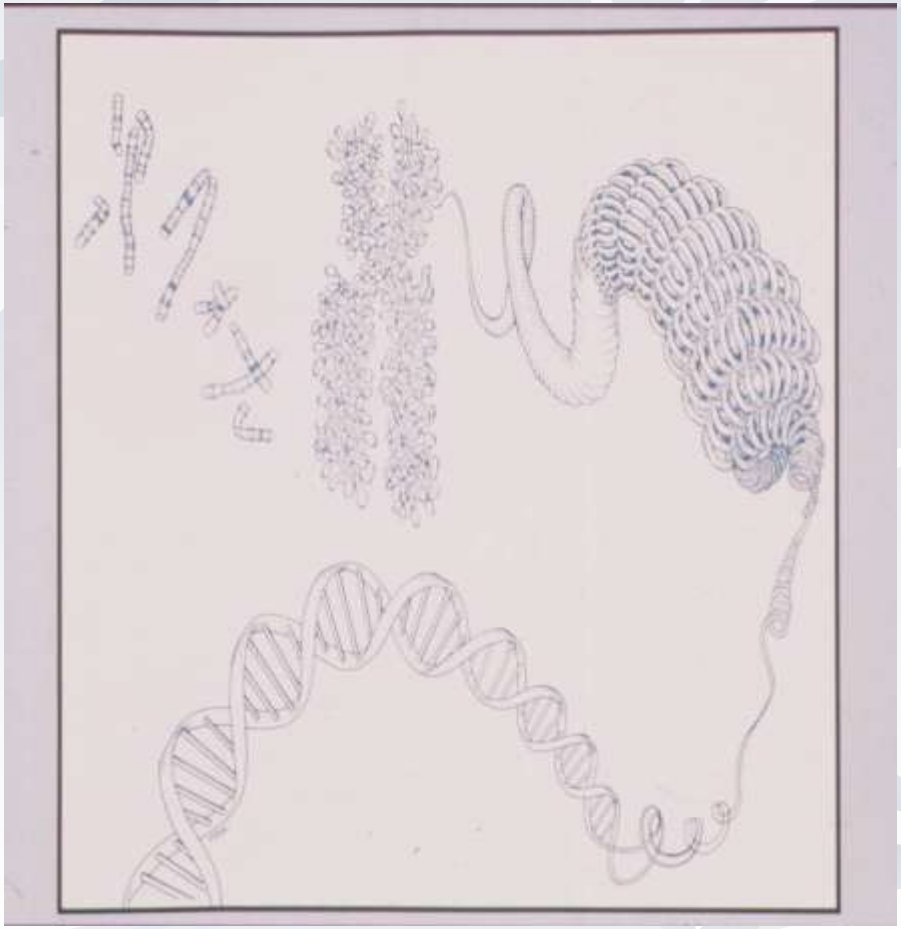
- **Definition**

Cytogenetic is the study of genetic material of cell



Cytogenetics

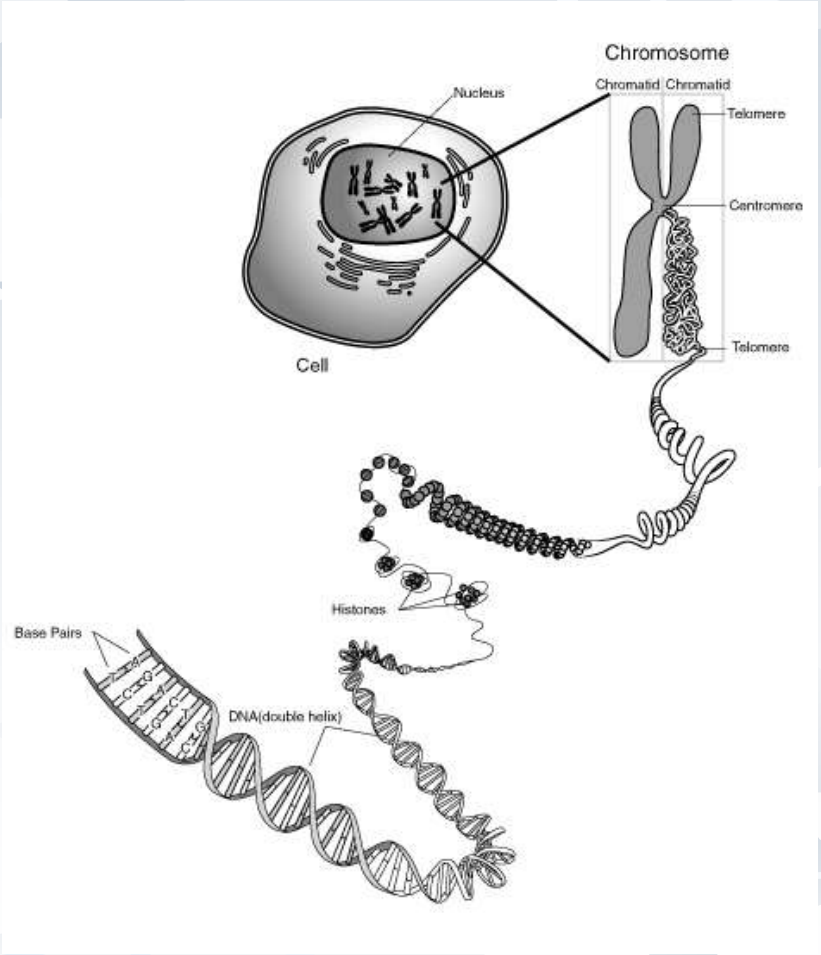
- The study of chromosome and the related disease states caused by abnormal chromosome number and\or structure.



What are chromosomes?

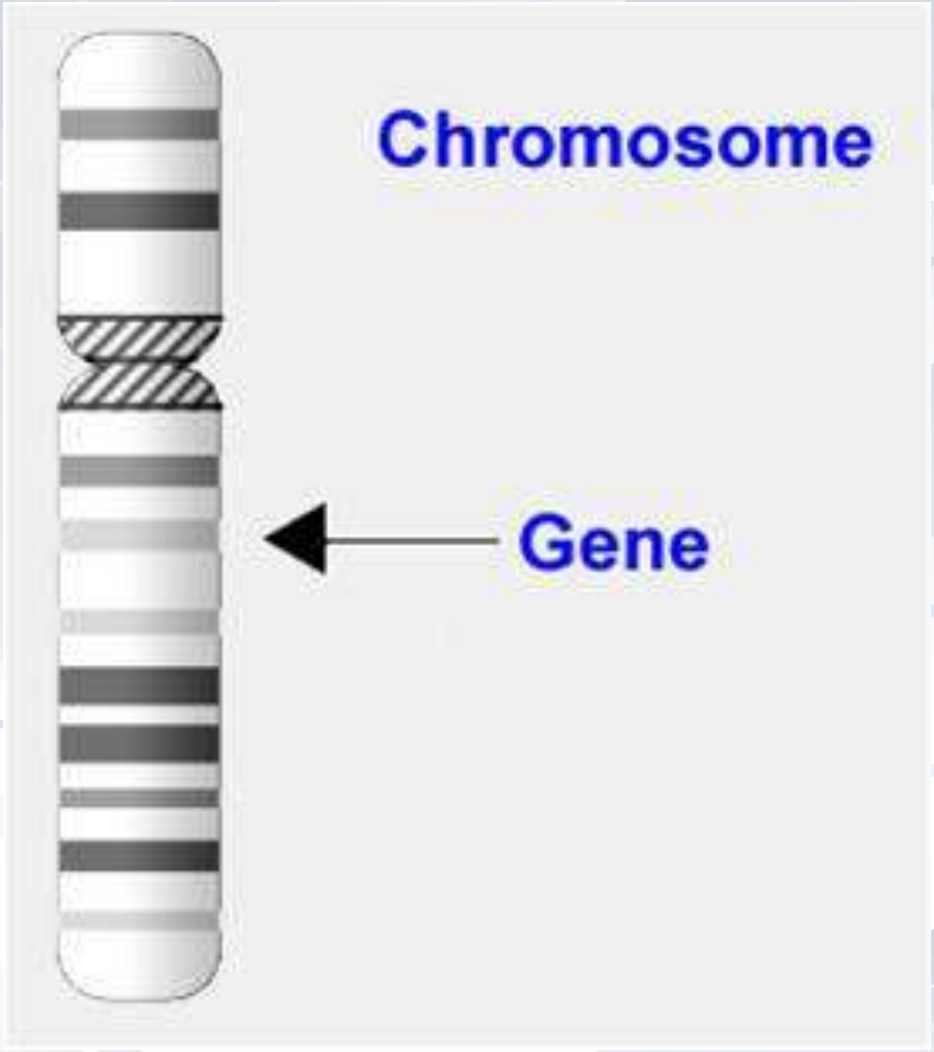
***Chromosomes** are the cellular structures that carry genes*

Chromosomes are distinct dense bodies found in the nucleus of cells , composed of protein and DNA.



What are chromosomes ؟(Cont)

- **The total number of bases in all the chromosomes of a human cell is approximately six billion and individual chromosomes range from 50 to 250 million base.**
- **The DNA sequence for a single trait is called a gene.**
- **Each chromosome contains a few thousand genes.**
- The estimate for the number of genes in humans are thought to be between 30,000 and 40,000 genes.
- Genes can be as short as 1000 base pairs or as long as several hundred thousand base pairs. It can even be carried by more than one chromosome.

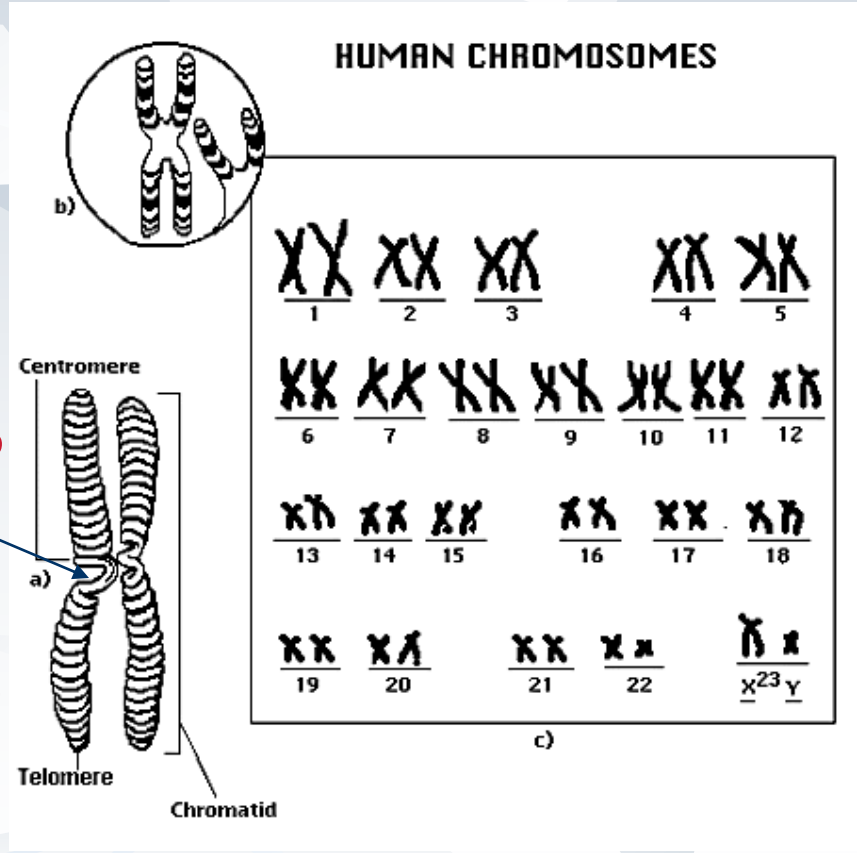

Chromosome
Gene

What are chromosomes ؟(Cont)

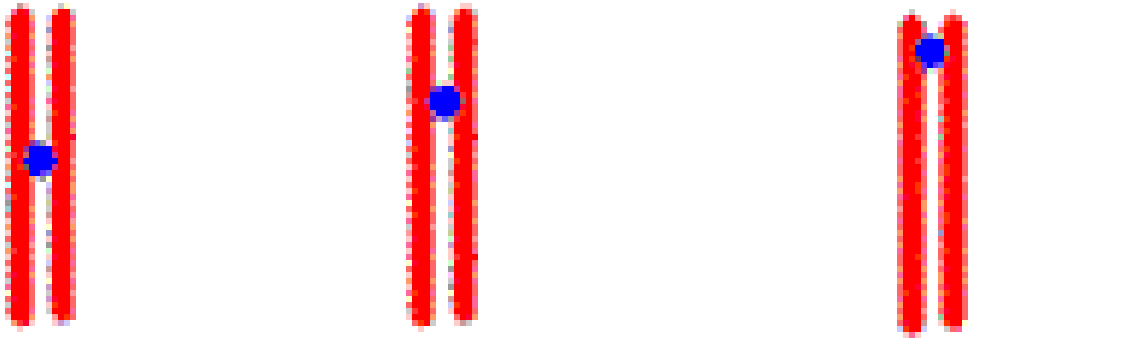
- The number of chromosomes in human cells is 46 with 22 autosomal pairs (one of each type contributed by the mother and one of each type from the father) and 2 sex chromosomes - 2 X chromosomes for females (one from father and one from mother) or an X and a Y chromosome for males (the X from the mother and the Y from the father).

Cytogenetic Nomenclature

- Each chromosome is visualized as two chromatids that are joined at a central constriction called the centromere.
- The centromere divides the chromosomes into two arms: a short arm (P) and a long arm (q)



Centromeric position and arm length



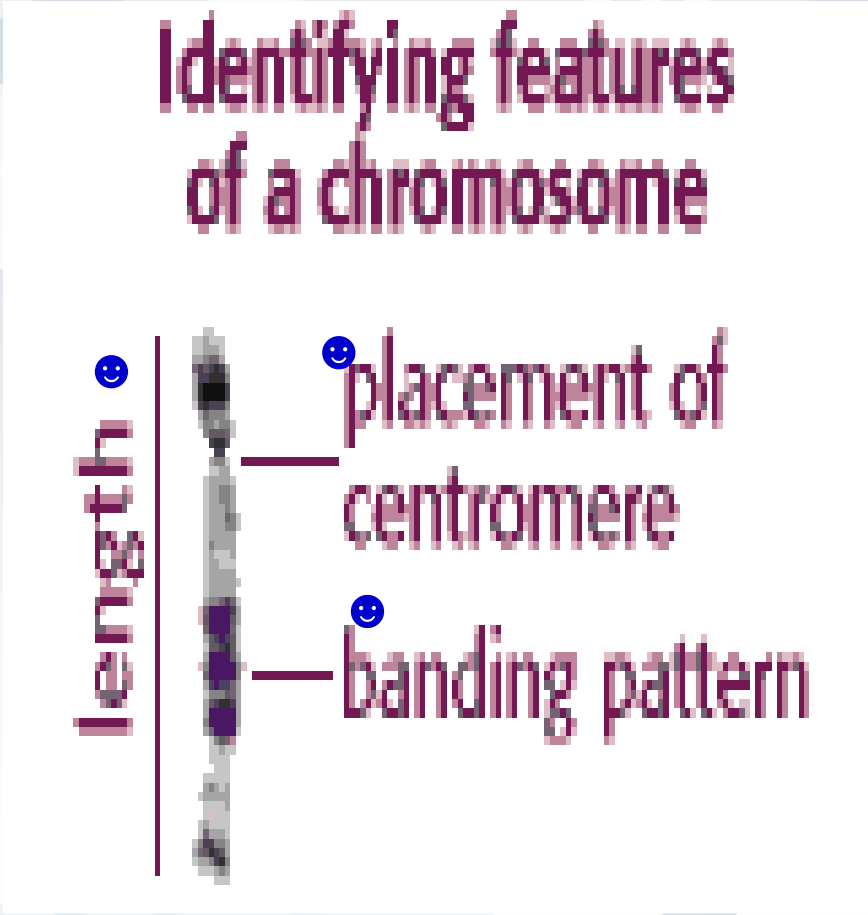
Metacentric Submetacentric Acrocentric

Chromosomes are divided into 7 groups, A……G

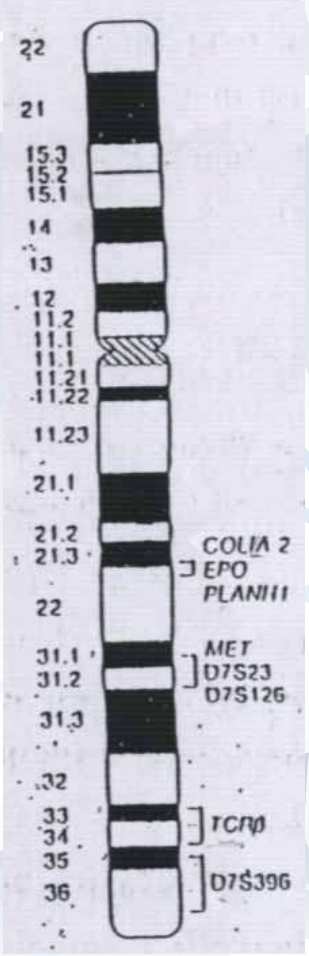
- **Group A: 1,2,3**
- **Group B: 4,5**
- **Group C: 6-12, x**
- **Group D: 13,14,15**
- **Group E: 16,17,18**
- **Group F: 19,20**
- **Group G: 21,22,Y**

Identifying features of a chromosome

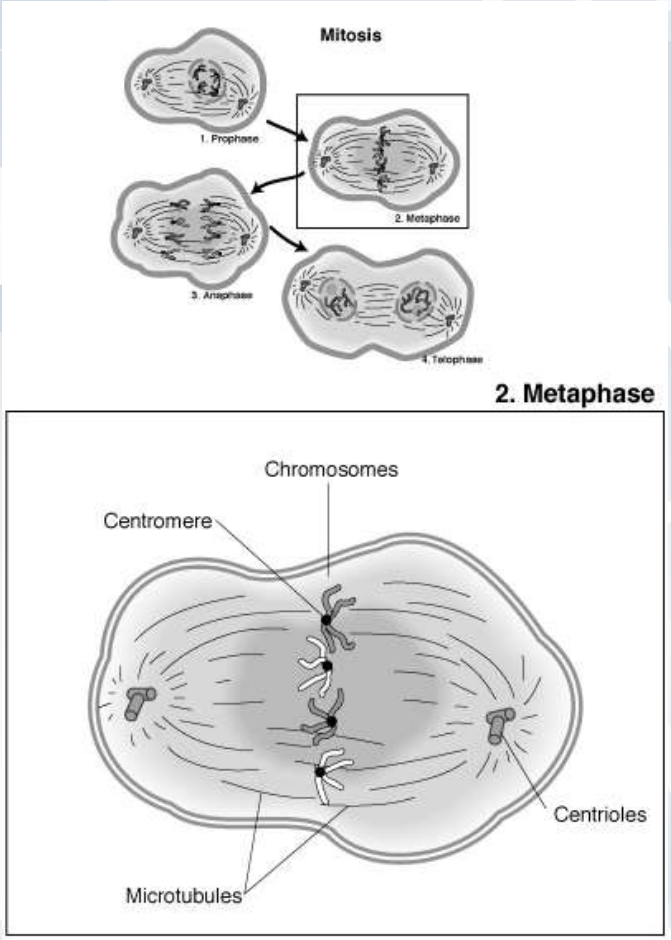
- **Size** (large, medium, small)
- **Position of centromere**
  - metacentric
  - acrocentric
  - submetacentric
- **Banding pattern**



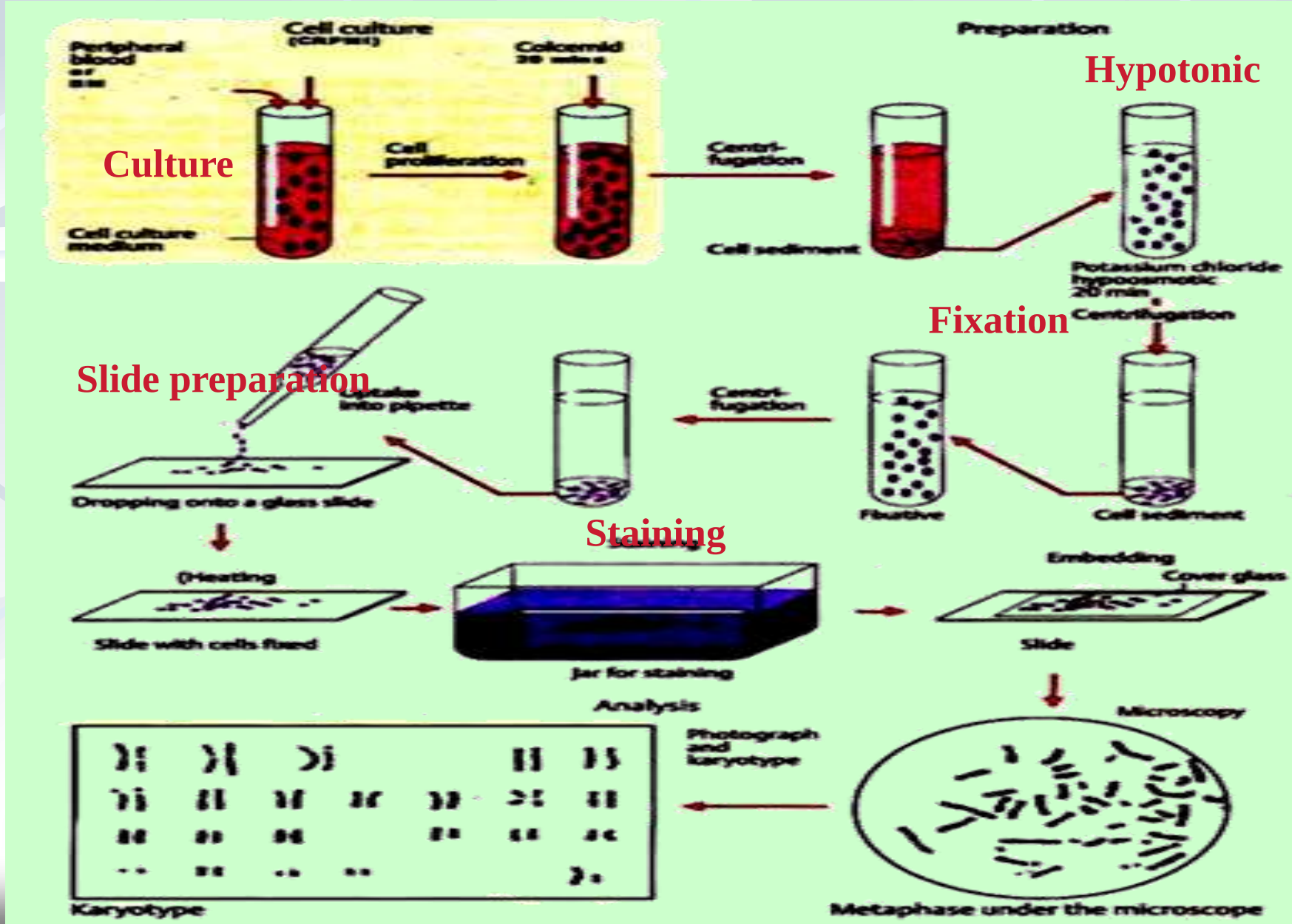
- **Chromosome bands**: part of a chromosome that is distinguished from adjacent parts by appearing **darker** or **lighter** with one or more banding techniques
- In high resolution banding, bands are divided into **sub-bands.** (1q31.1).



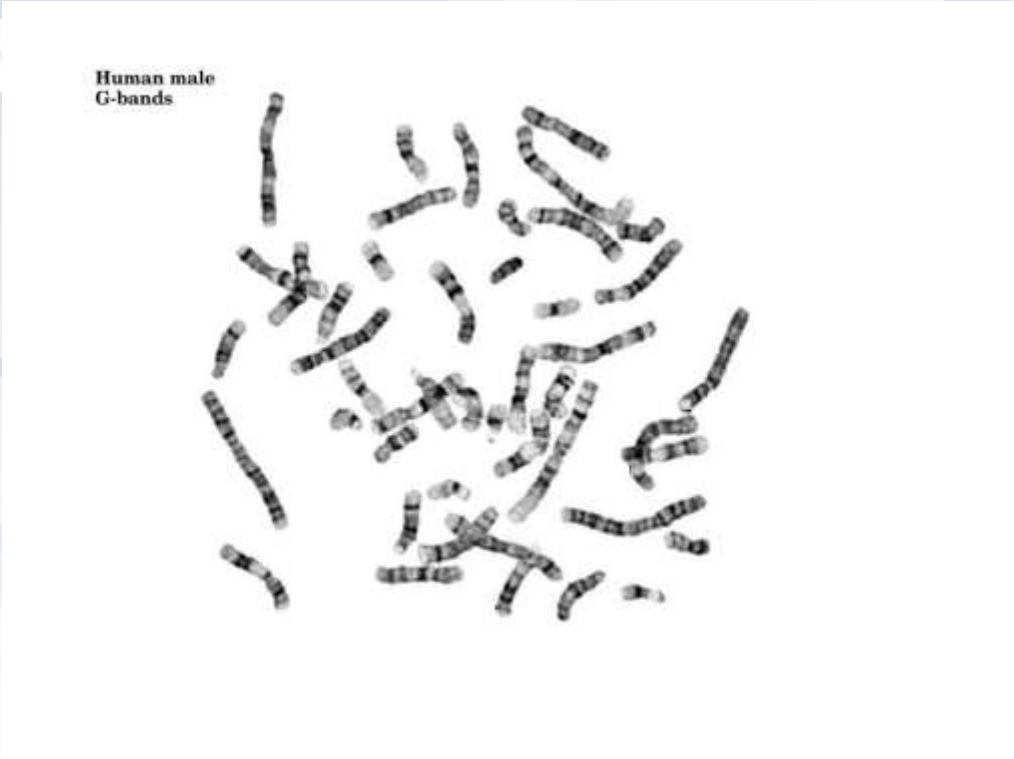
- The chromosomes are most easily seen and identified at the metaphase stage of cell division.

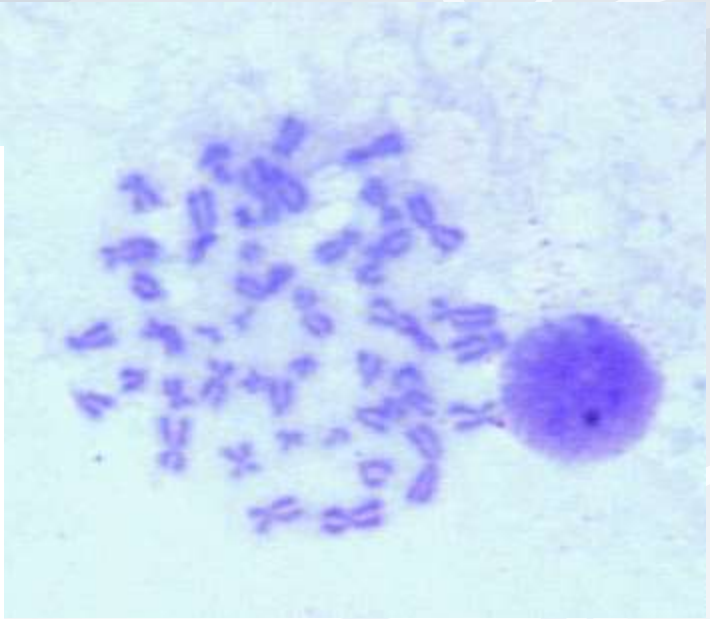


Conventional Cytogenetic Analysis



Metaphase spread



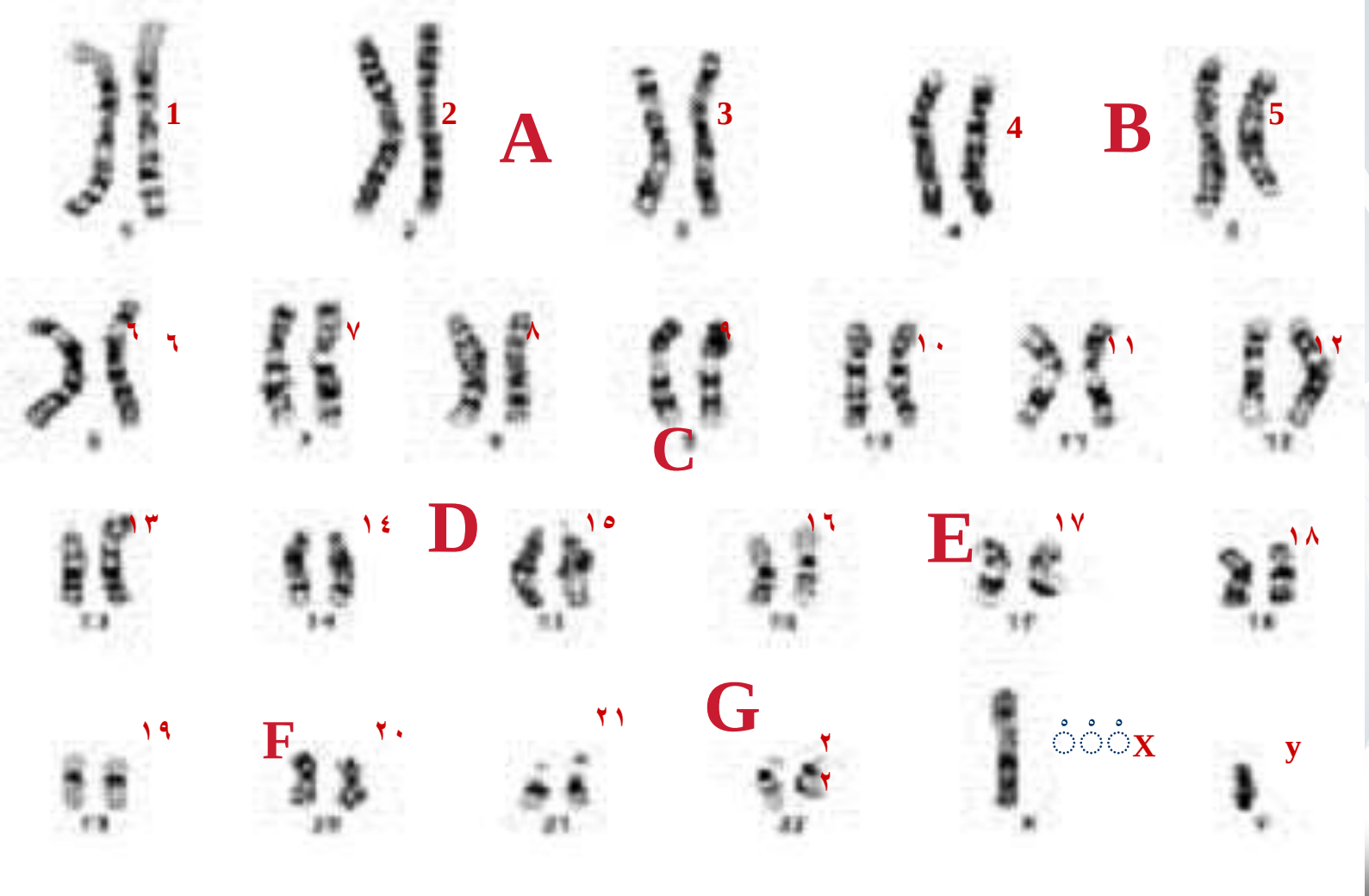


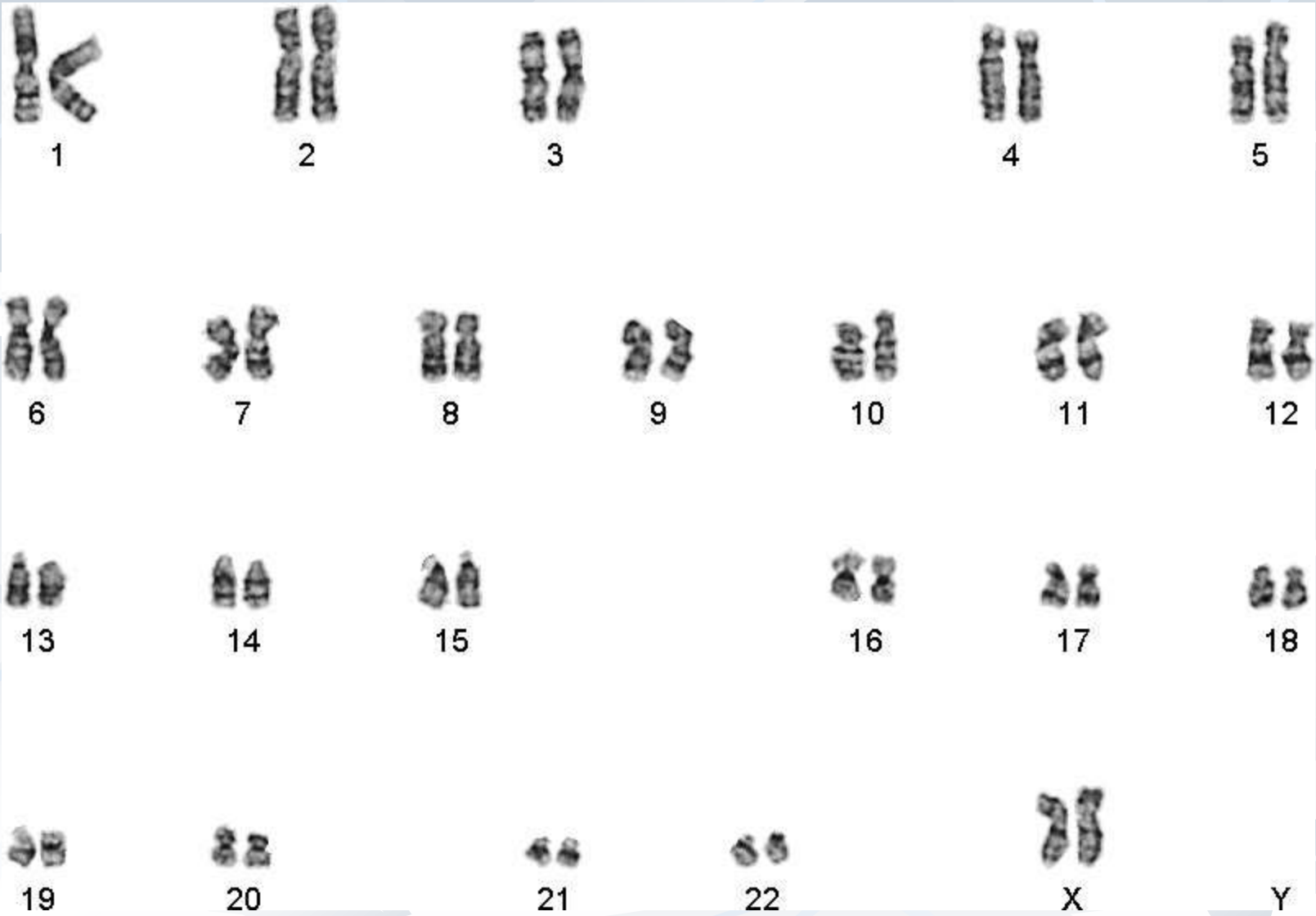
Chromosomes Banding

Type	Stain	Area Stained	Effect
Q-banding	Quinacrine	Chromosome arms; mostly repetitive AT-rich DNA	Under UV light, distinct fluorescent banded pattern for each chromosome.
G-banding	Giemsa	Chromosome arms; mostly repetitive AT-rich DNA	Distinct banded pattern for each chromosome; same as Q-banding pattern except single additional band near centromere of chromosomes 1 and 16
R-banding	Variety of techniques	Chromosome arms; mostly unique GC-rich DNA	Reverse banding pattern of that observed with Q- or G-banding
C-banding	Variety of techniques	Centromere region of each chromosome and distal portion of Y chromosome; highly repetitive, mostly AT-rich DNA	Largest bands usually on chromosomes 1, 9, 16, and Y; chromosomes 7, 10, and 15 have medium-sized bands; size of C-bands highly variable from person to person

A karyotype: **Arrangement of chromosomes from a particular cell, the largest chromosomes are first and the smallest ones are last.**

It is a description of the number and structure of the chromosomes.




1
2
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5
6
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8
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11
12
13
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15
16
17
18
19
20
21
22
X
Y

Chromosomal abnormalities

- **Numerical:** A karyotype with abnormal No. of chromosomes.
- **Structural:** Alterations in the structure of chromosomes.

Changes in number, or sets, of chromosomes

A) Polypoidy – change in complete sets of chromosomes (3n, 4n, etc)
plants > animals.

B) Aneuploidy – change in the no. of chromosomes

- nullisomy 2n-2
- monosomy 2n-1
- trisomy 2n+1
- tetrasomy 2n+2

Gene dosage effect

1- Sex-chromosomal aneuploids .

2- Autosomal aneuploids .

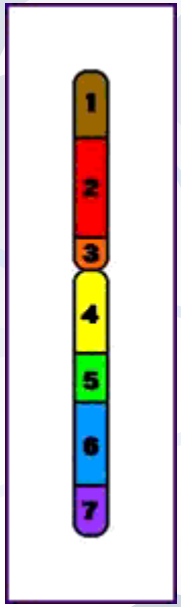
- **Diploid cell:** A cell with a normal complement of structurally normal chromosomes.
- **Pseudodiploid cell:** A cell with 46 chromosomes but with numerical chromosomal abnormality (e.g. loss of one chromosome & gain of another) or structural abnormality.
- **Aneuploid cell:** Cell with abnormal number of chromosomes.

Abbreviations

- del = deletion
- der = derivative
- dup = duplication
- inv = inversion
- ins = insertion
- t = translocation
- add = additional material of unknown origin
- mar = marker chromosome of unknown origin

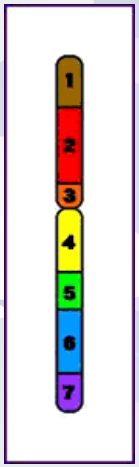
- ***Interstitial deletion*:**

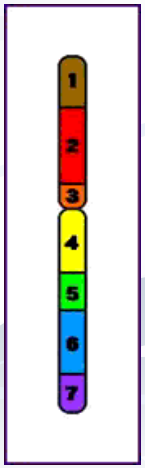
  loss of internal segment.



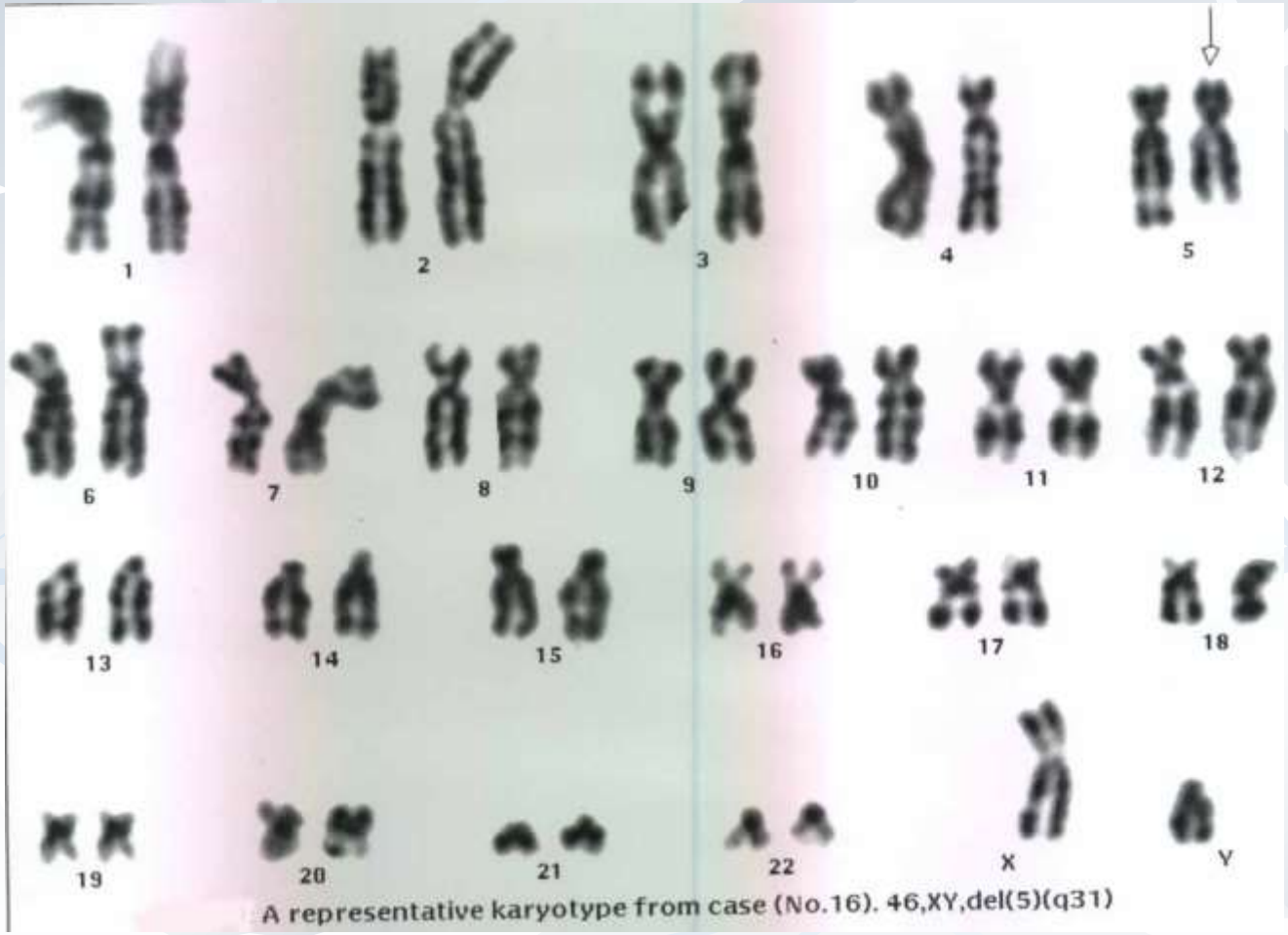
Terminal deletion

loss of tip of chromosome





46,xy,del (5)(q31)



A representative karyotype from case (No.16). 46,XY,del(5)(q31)

46,xy,del(7)(q22;q32)

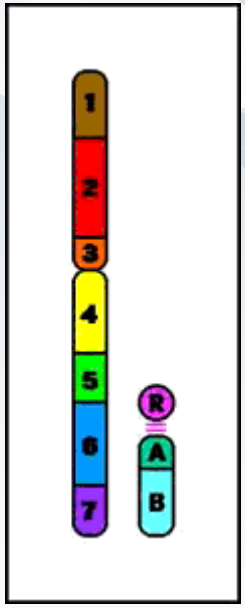
A representative karyotype from case (No.25). 46,XY,del(7)(q22q32)

- **Translocation (t): relocation of material from one chromosome to a different chromosome.**

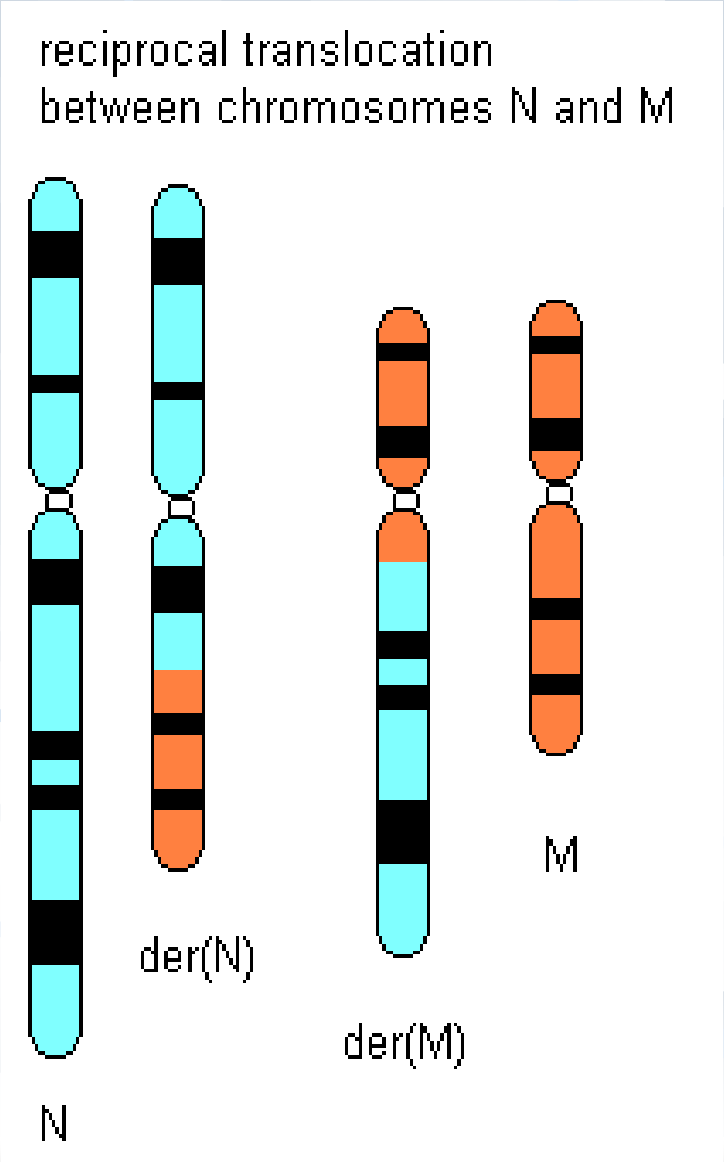
  ***Reciprocal*: exchange of material between different chromosomes. t(9;22)(q34;q11)**

  ***Non-reciprocal*: rare**

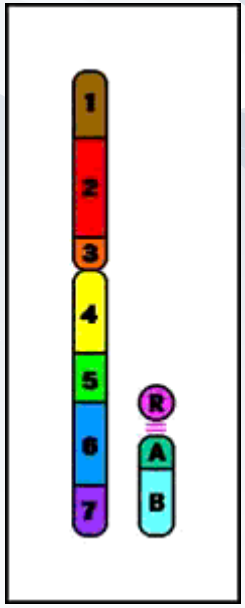
Reciprocal Translocation



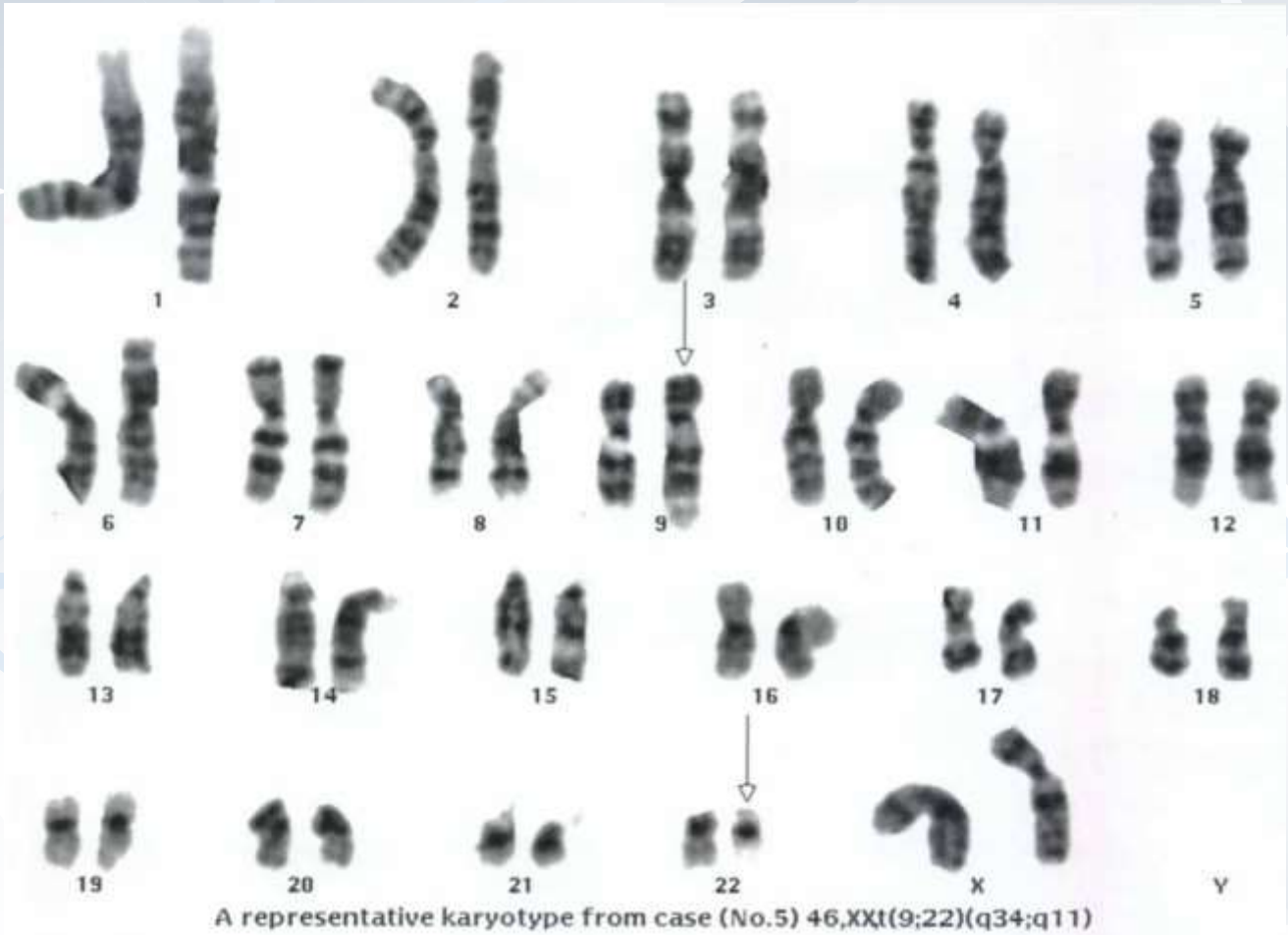
Reciprocal Translocation



Non-reciprocal translocation

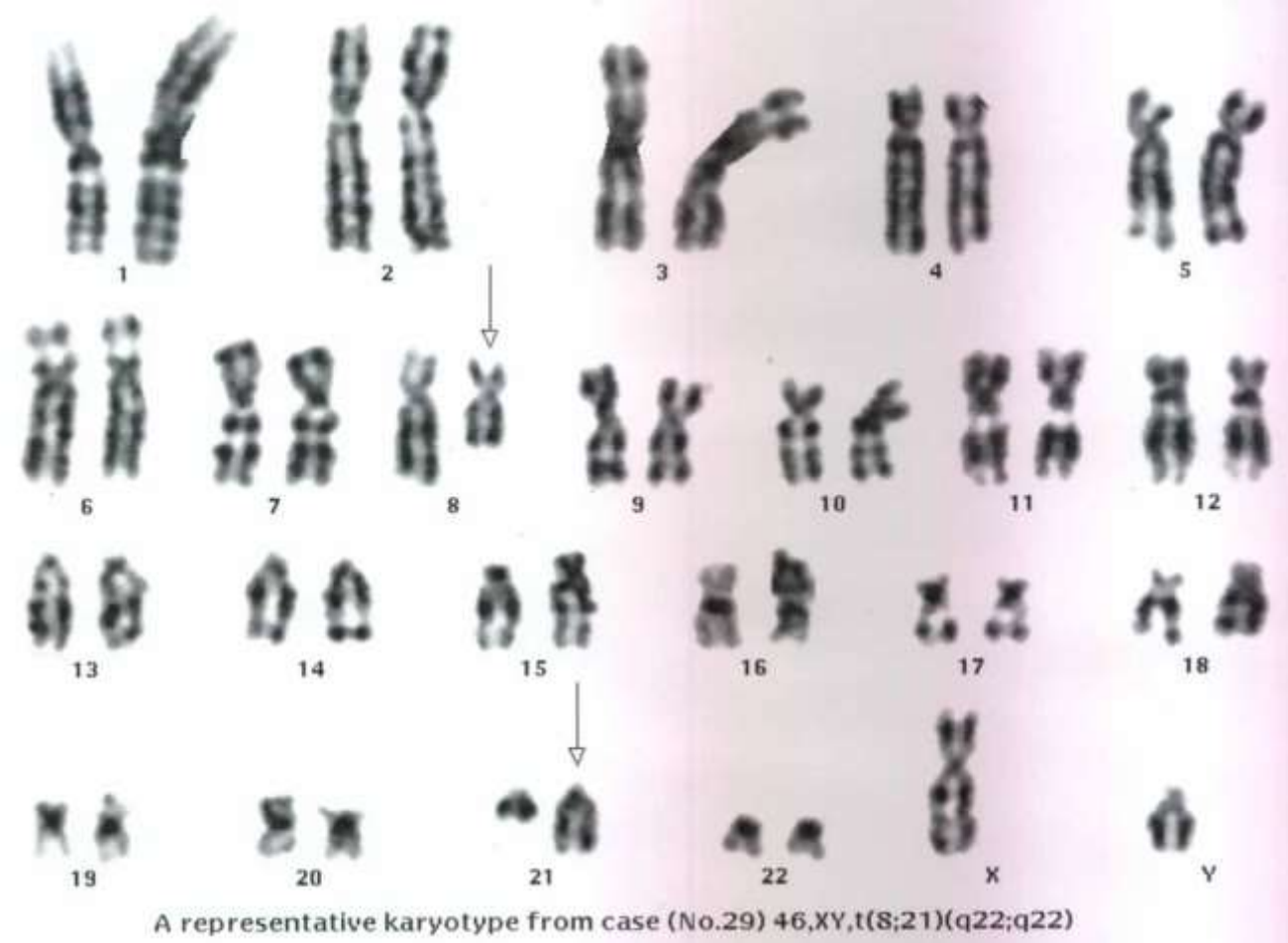


46,xx,t(9;22)(q34;q11)



A representative karyotype from case (No.5) 46,XXt(9;22)(q34;q11)

46,xy,t(8;21)(q22;q22)



A representative karyotype from case (No.29) 46,XY,t(8;21)(q22;q22)

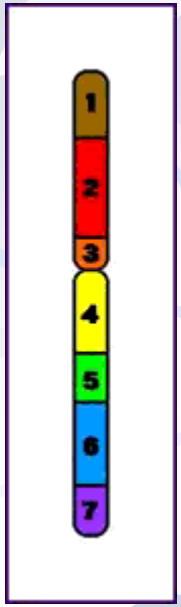
- **Inversion (inv): it is 180 rotation of a chromosome segment**
- ***Pericentric***

  Paracentric

Pericentric Inversion

- **Pericentric:**

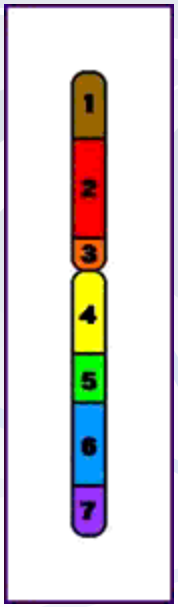
  The inverted segment include the centromere inv (16)(p13;q22)



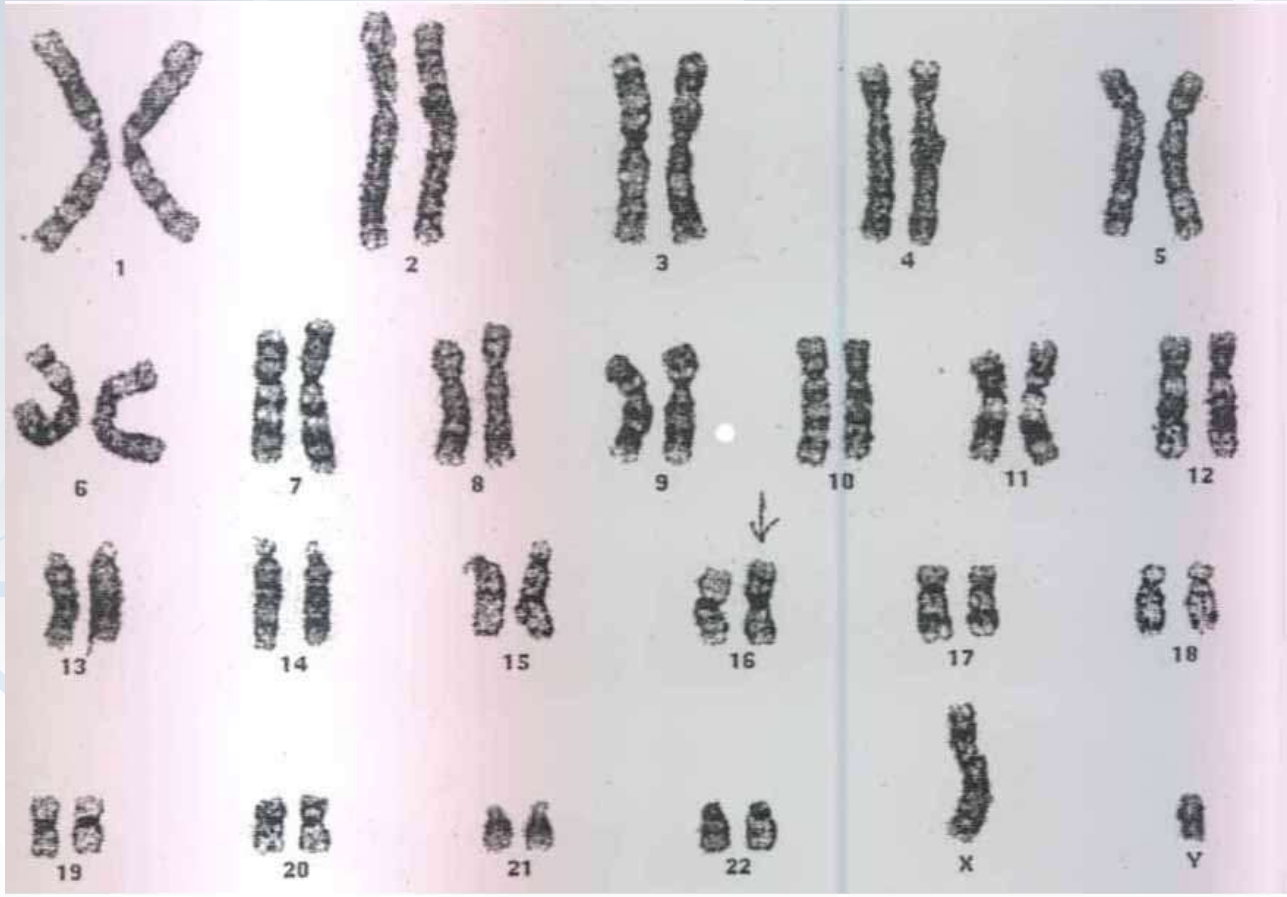
Paracentric inversion

- **Paracentric:**

  The inverted segment within either the short or long arm
  inv(3)(q21;q26)

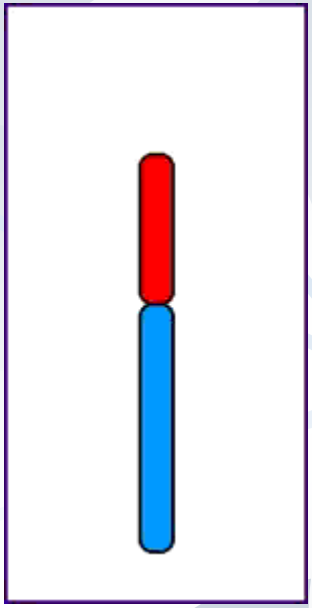


46,xy,inv(16)(p13;q22)

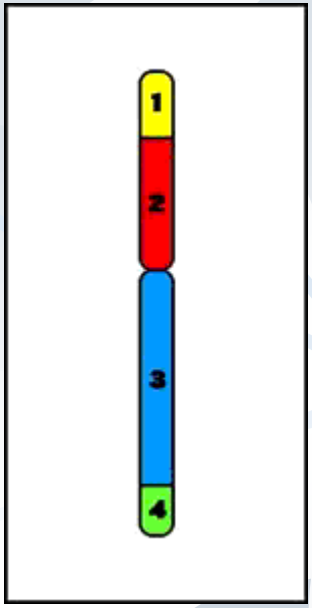


A representative Karyotype from case (No.13) showing 46,XY, inv(16)(p13q22)

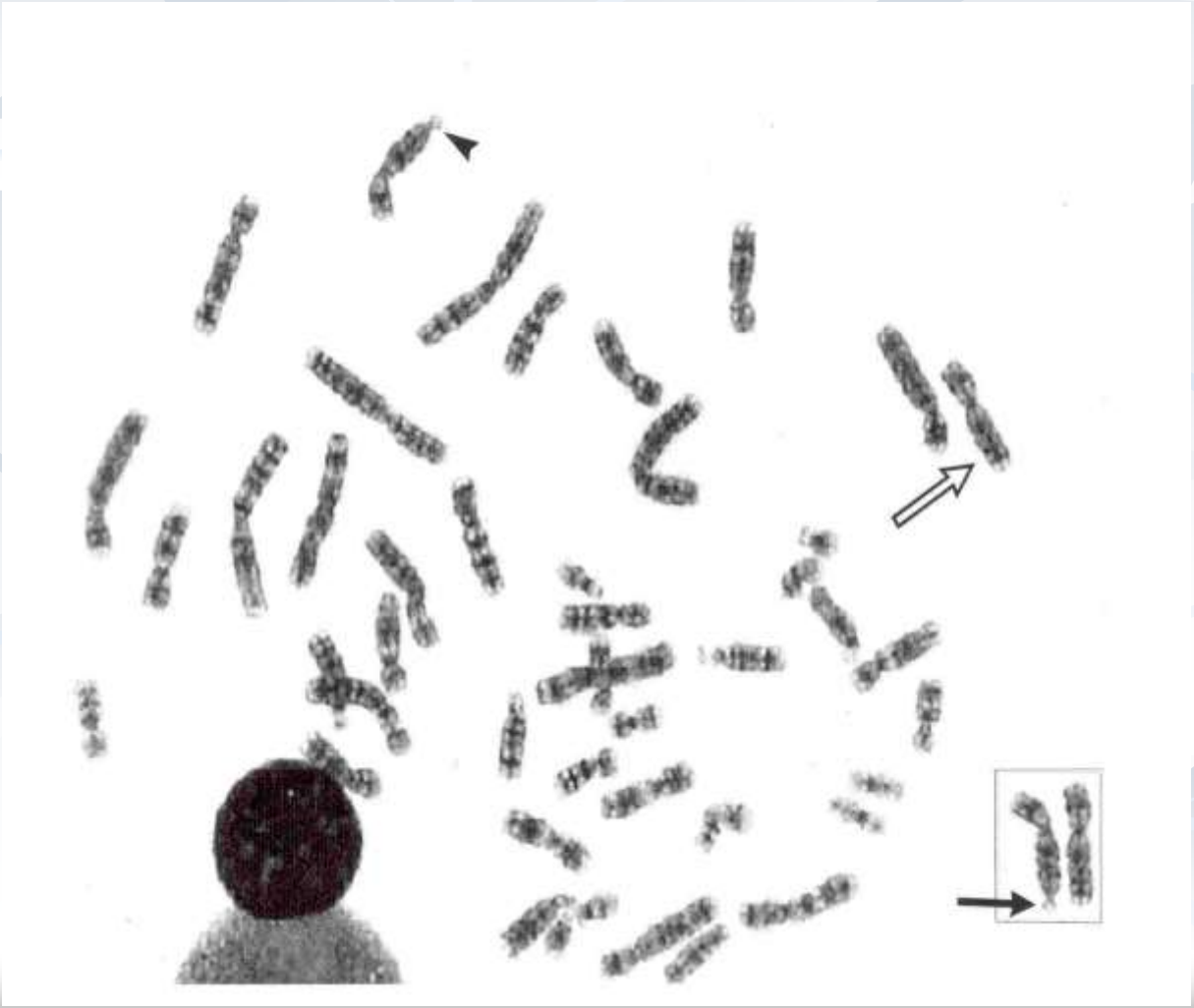
- **Isochromosome (i):** Two identical chromosome arms positioned as mirror images of each other i(17q)

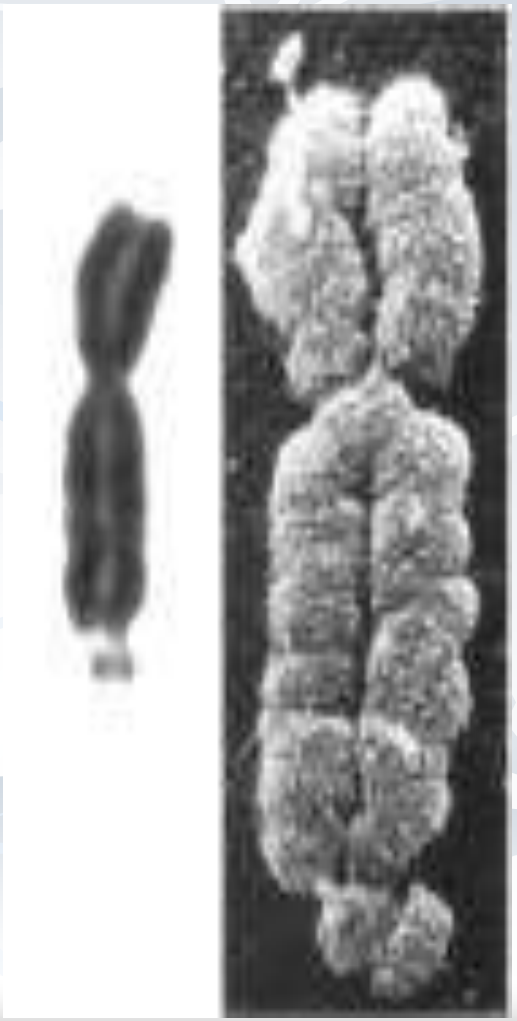


- **Ring chromosome(r):** breaks have occurred in both the short and the long arms and the break points have joined together …… closed ring



Fragile-X-Syndrome





Thank you