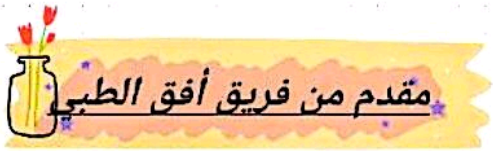
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1412-1991


حركة الشبيبة الفتحاوية
جامعة الأزهر
Fateh Youth Movement

The human chromosom

Dr.ImanNabil

A human chromosome

1- Definition & function of the chromosome.

2- Structure of the chromosome.

3- Telomere: definition & function.

4- Chromosomal changes during the cell cycle& cell division.

5- Types of microtubules during cell division.

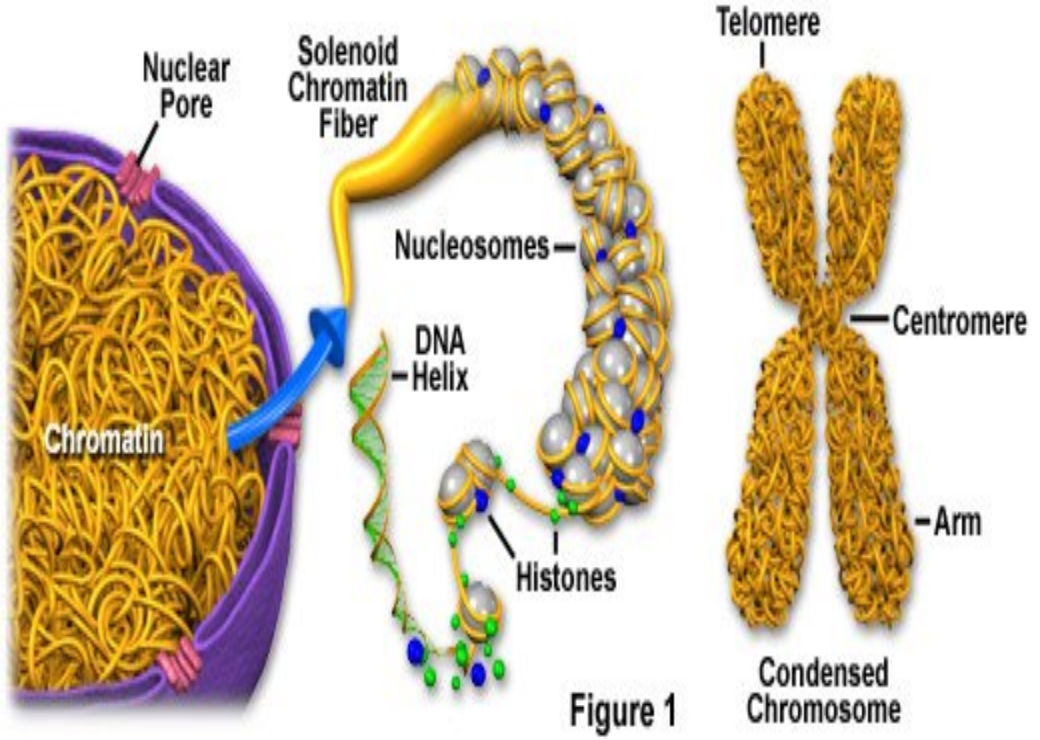
6- Types of chromosomes.

7- Karyotyping: definition, method& importance.

8- Chromosomal abnormalities: causes, risk factors & types.

A human chromosome

- **Definition:** thread-like structures, present in the nucleus.
- **Function:** carries the genetic information in the form of genes.



Structure of the chromosome

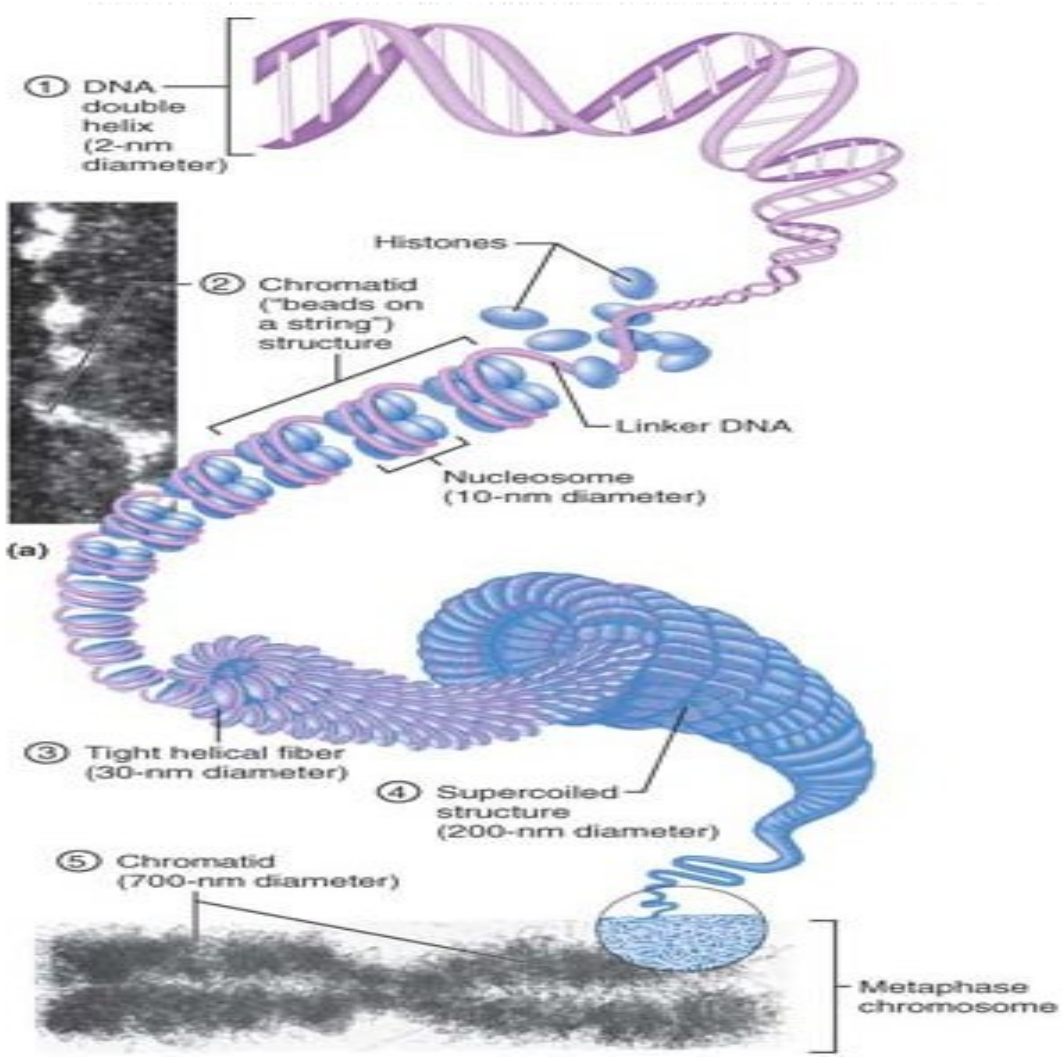
- The DNA is packed in the form of chromatin (DNA + histone proteins).
- DNA is extensively-packaged in chromatin as:

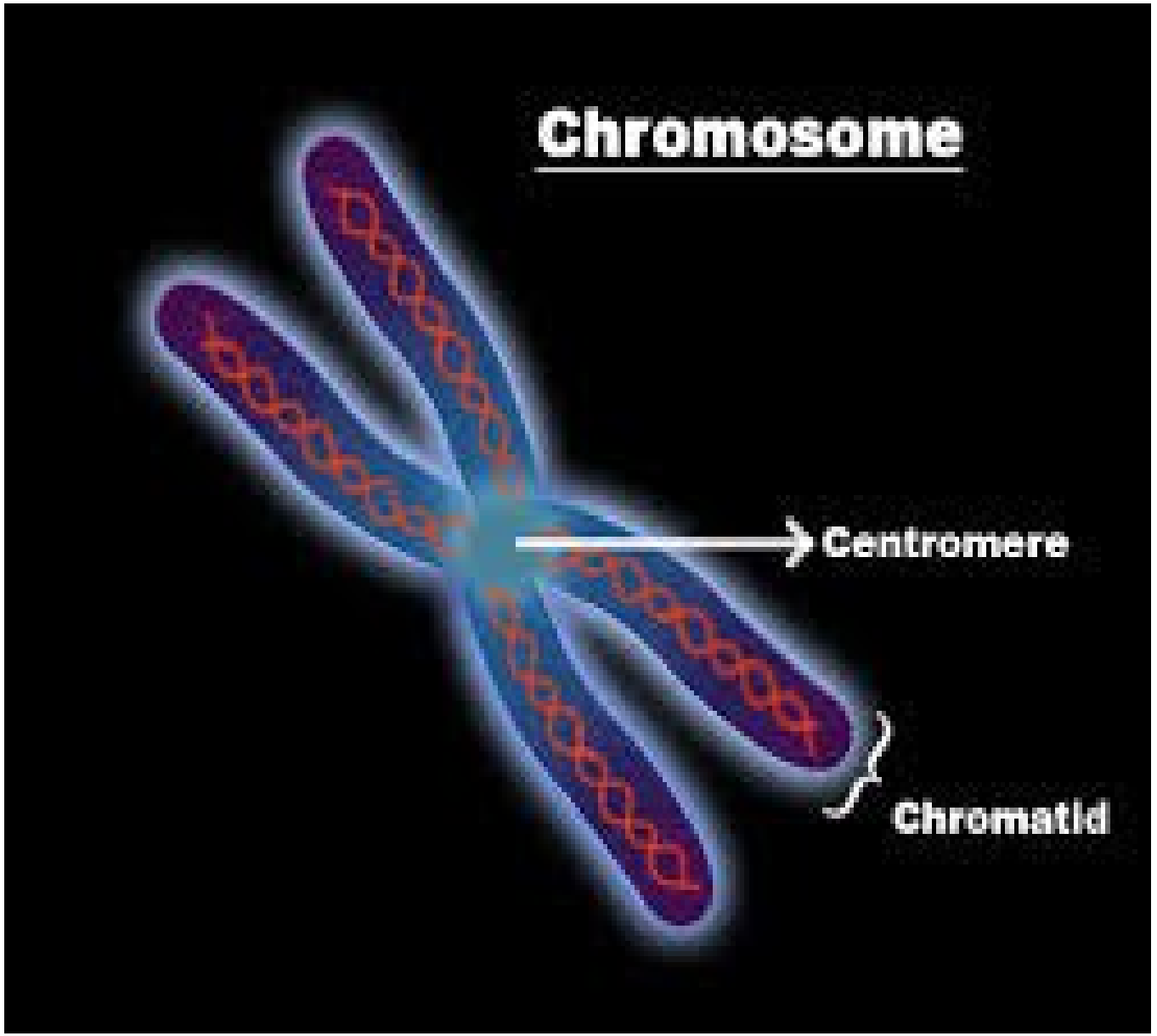
1- A segment of the DNA is wrapped two times around eight histone proteins to form a *nucleosome*. Each nucleosome is separated from the next by a region of linker DNA.

2- Repeating nucleosomes with intervening "linker" DNA form a *10nm fiber* (beads on a string).

3- This chain of nucleosomes is packed to form a *30nm fiber*

4- Higher orders of packaging gives the compact structure *700nm* seen in the metaphase of the dividing cell known as the chromatid of a chromosome.




Chromosome
Centromere
Chromatid


You must unwind the DNA . . .
and unwind the DNA . . .
many times to observe the DNA double helix.
Histones - protein DNA wraps around.

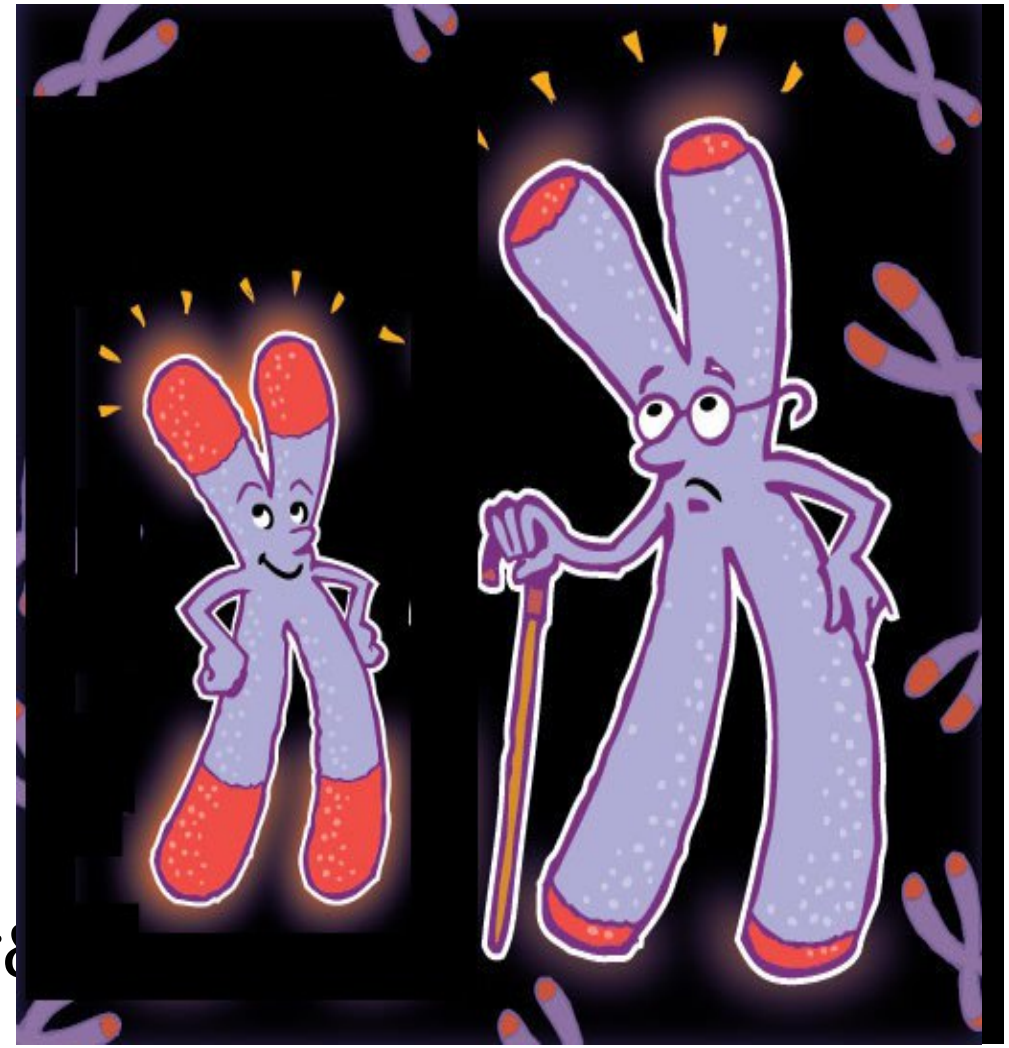
Telomere

- **Definition:** a segment of the DNA present at the end of the chromosomes.
- **Functions:**

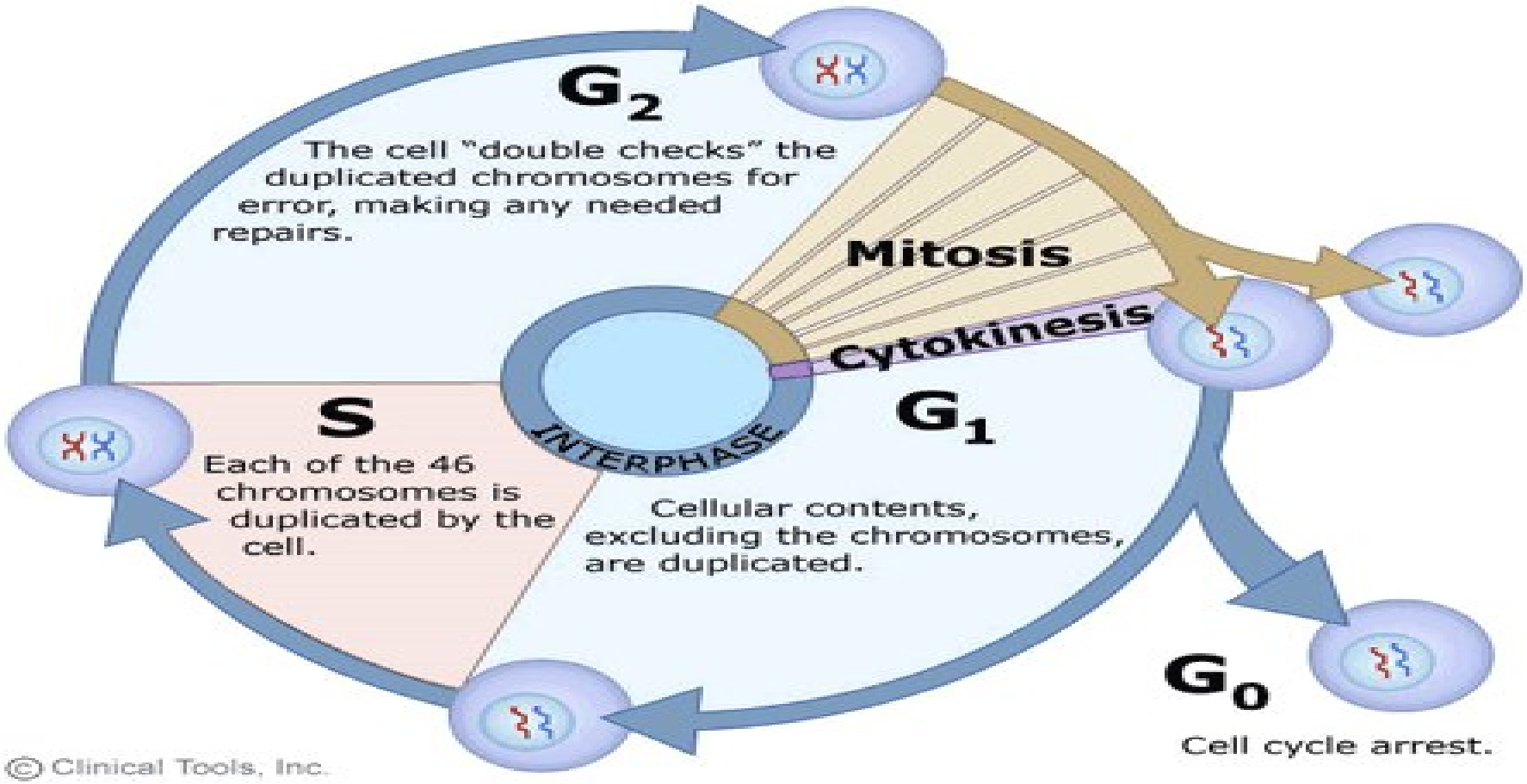
1- Prevent the end of the chromosomes from sticking together.

2- Allow proper dividing of the cells.

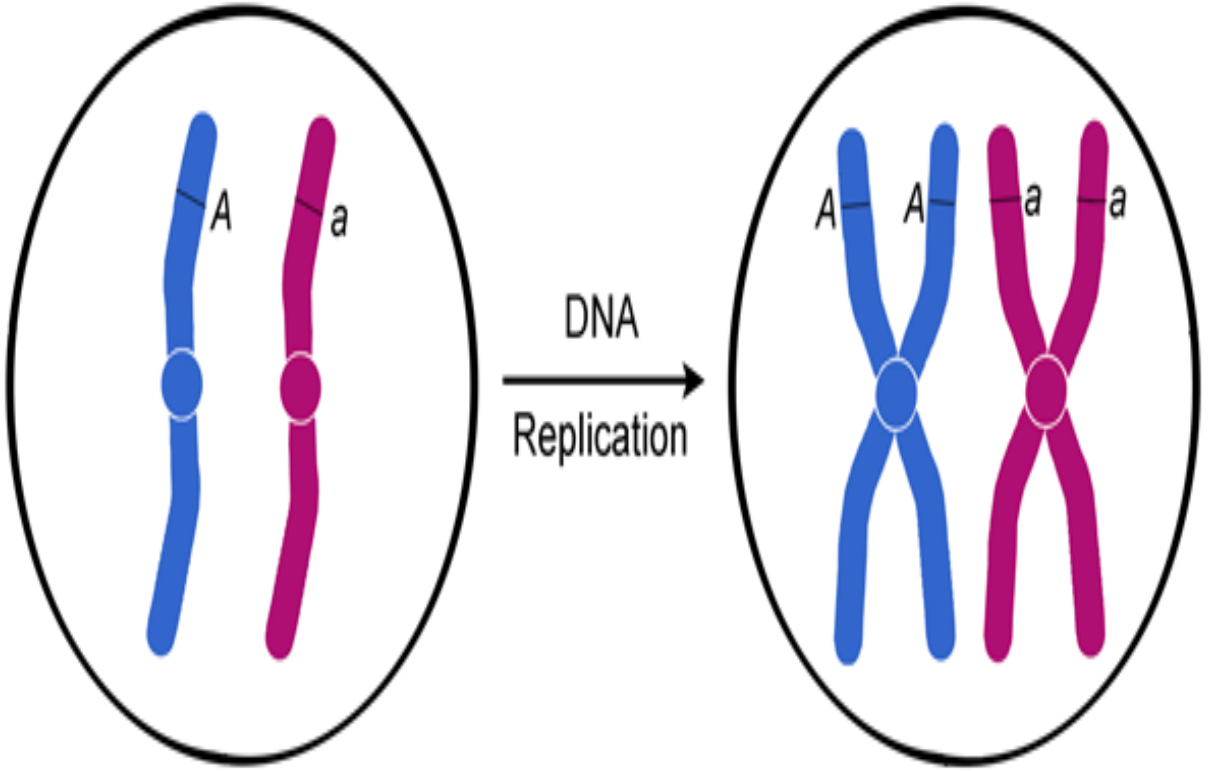
- Each time the cell divides, the telomere get shorter, until reaching certain length, the cell stop dividing& become senescent.



Cell cycle

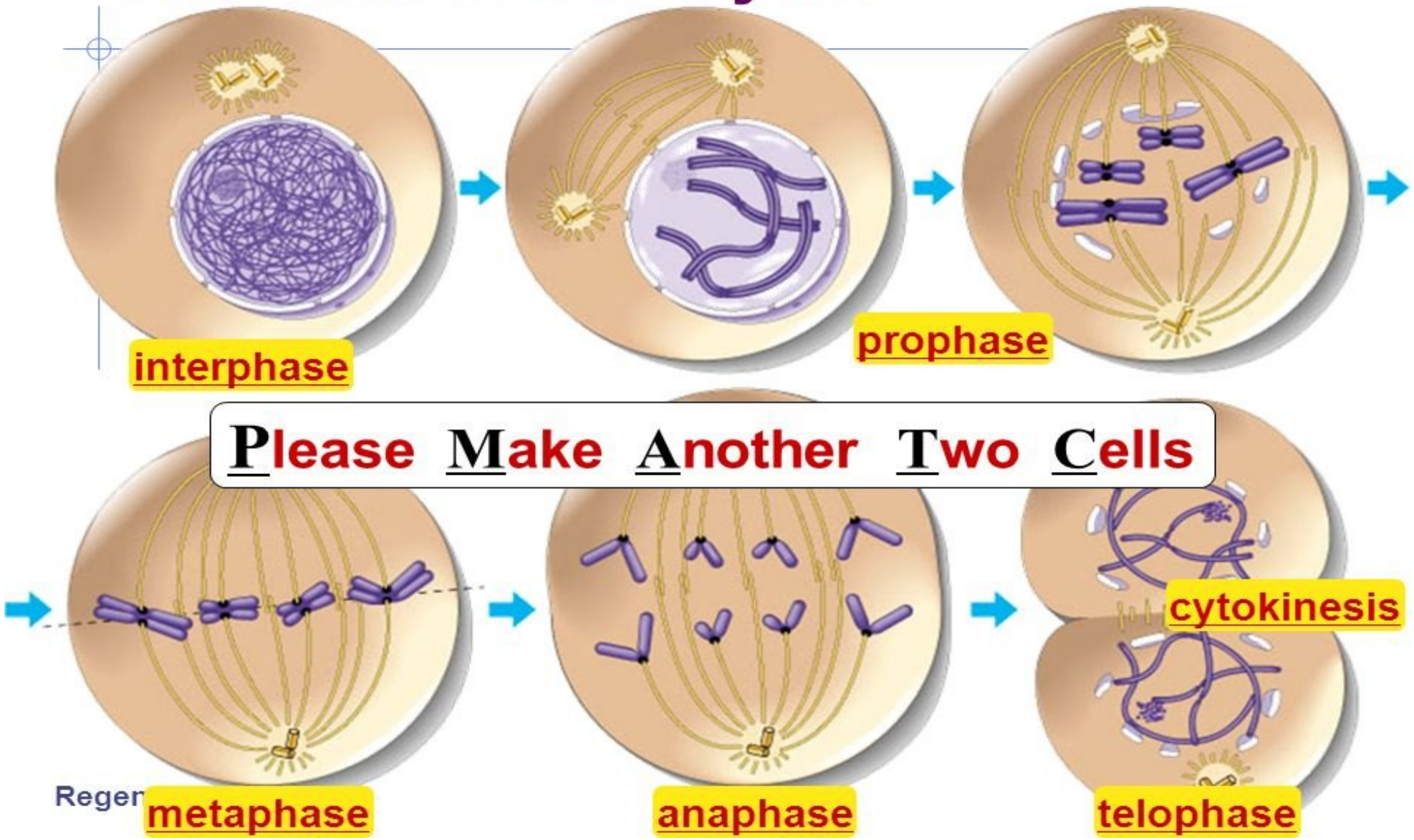

G2
The cell "double checks" the duplicated chromosomes for error, making any needed repairs.
Mitosis
Cytokinesis
INTERPHASE
G1
Cellular contents, excluding the chromosomes, are duplicated.
S
Each of the 46 chromosomes is duplicated by the cell.
G0
Cell cycle arrest.
© Clinical Tools, Inc.

S-phase



Single chromosome

Double chromosome


interphase
prophase
Please Make Another Two Cells
metaphase
anaphase
cytokinesis
telophase

Mitosis

Types of microtubules

1- Astral microtubules:

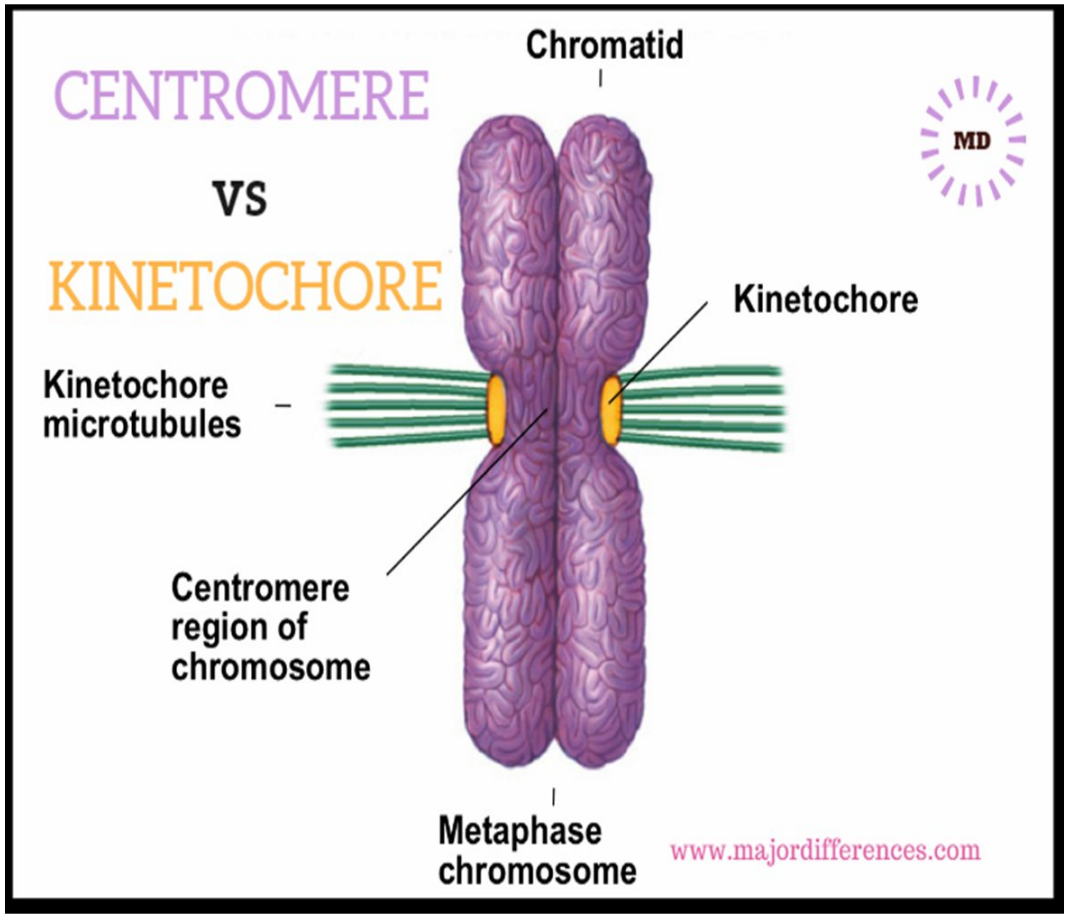
- ★ Originate from the MTOCs in the centrosome & radiate from it toward the cell membrane.
- ★ Function: necessary for proper orientation of the spindle apparatus within the cell.

2- Polar microtubules:

- ★ Originate from the MTOCs in the centrosome & begin to polymerize between the centromeres, overlapping each other.
- ★ Function: help to push the spindle apparatus apart away from each other.

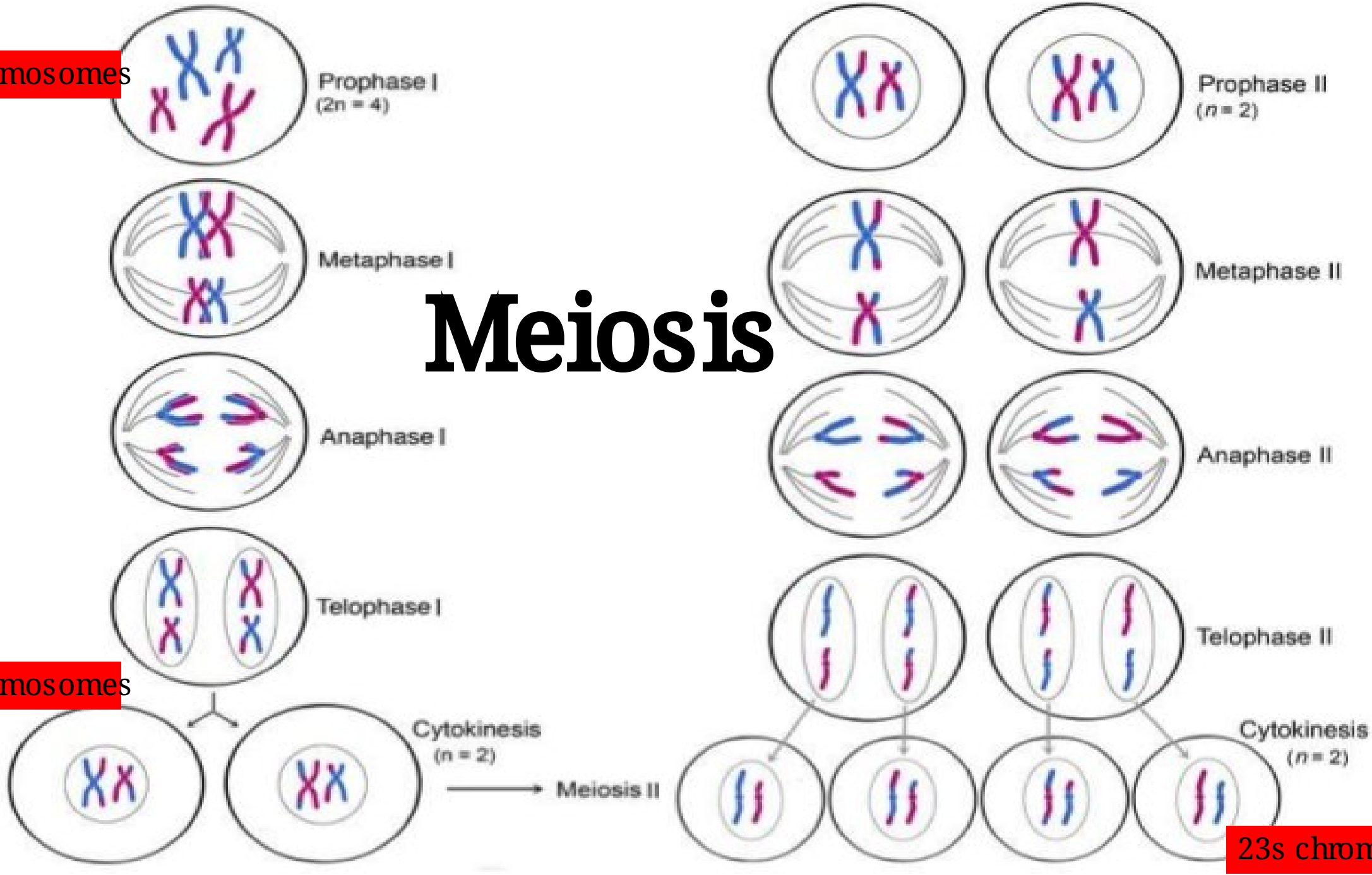
3- Kinetochore microtubules:

- ★ Attach to the kinetochore on each side of the centromeres.
- ★ Function: pull on the chromosomes and move them.



46 d chromosomes

Meiosis

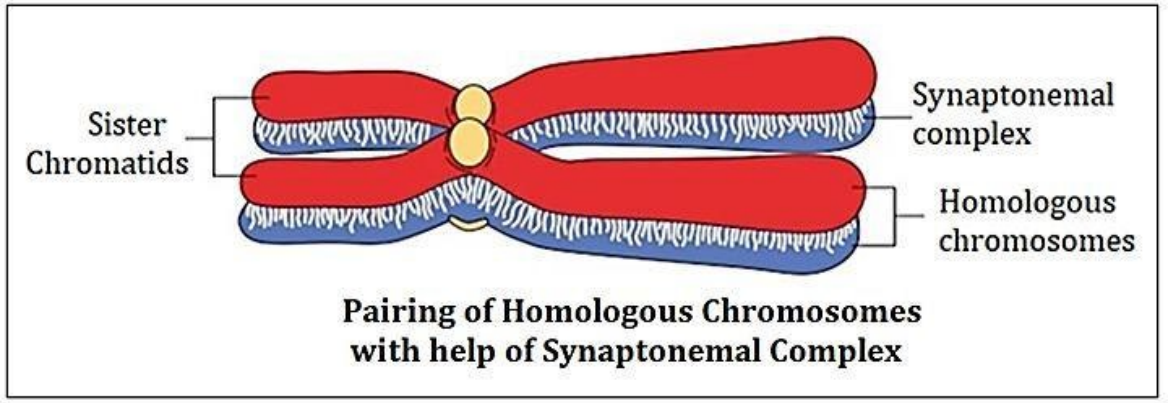


23 d chromosomes

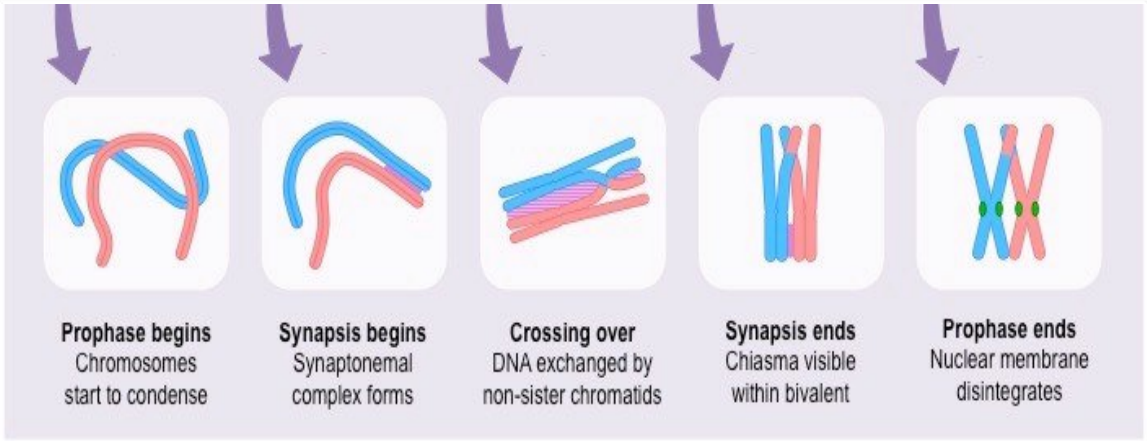
23s chromosomes

Meiosis
Prophase I

- **Leptotene** chromosomes begin to condense appear as *long & thin threads*. Each chromosome consists of 2 chromatids joined together by a centromere.
- **Zygotene** homologous chromosomes *pair up* forming 23 bivalents or tetrads. They begin to make connection (synapsis) by synaptonemal complex.
- **Pachytene** chromosomes becoming *shorter and thicker*. Crossing-over occurs between the chromatids of the homologous chromosomes at sites called chiasmata.
- **Diplotene** the synaptonemal complex dissolves, so the *2 homologous chromosomes* are separated from each other but they are connected at the chiasmata. Each chromosome contains some genes inherited from the mother and other genes inherited from the father.
- **Diakinesis** bivalents *more condensed* & the nuclear membrane and the nucleolus disappear.

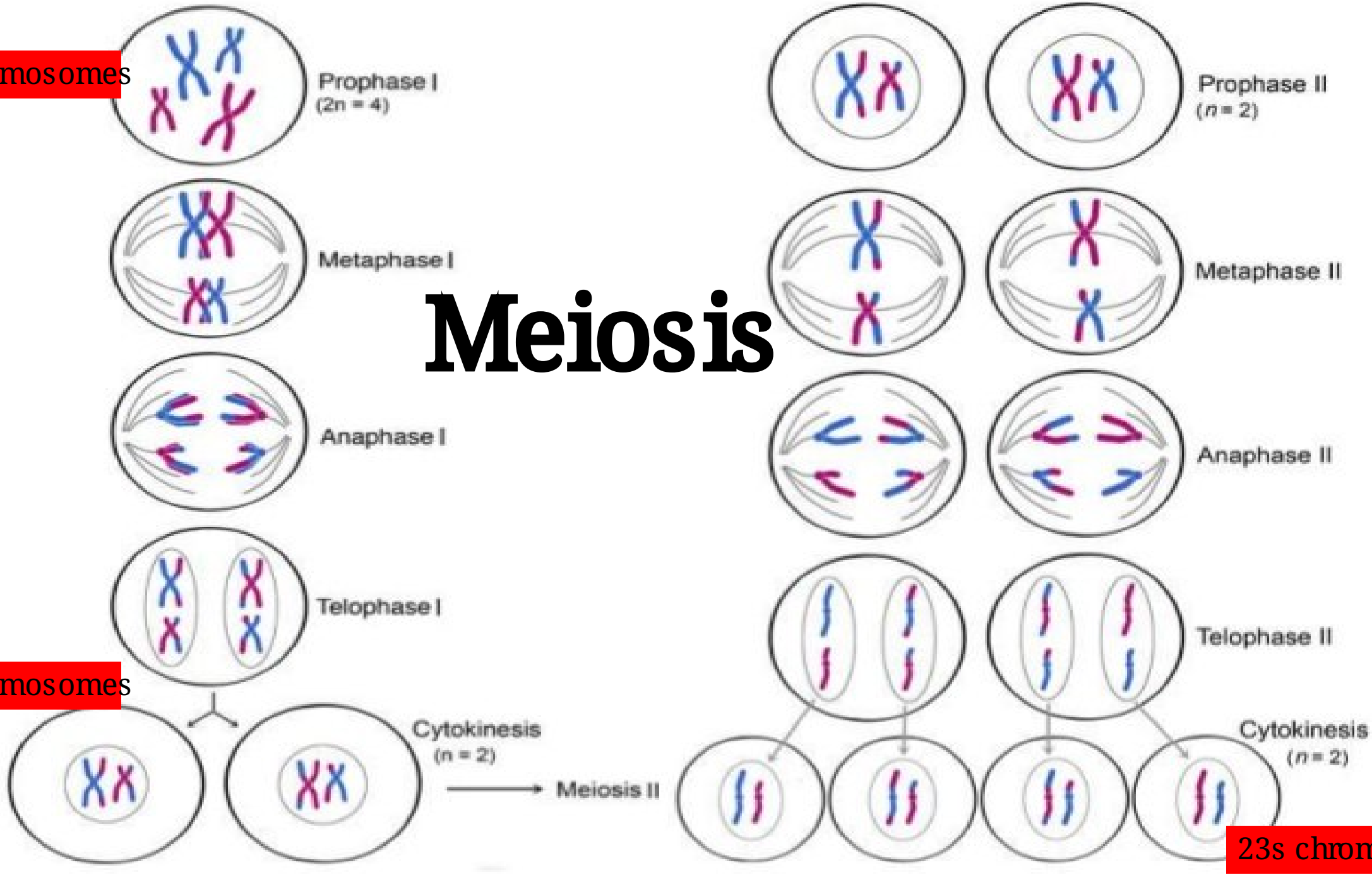


Pairing of Homologous Chromosomes with help of Synaptonemal Complex



46 d chromosomes

Meiosis



23 d chromosomes

23s chromosomes

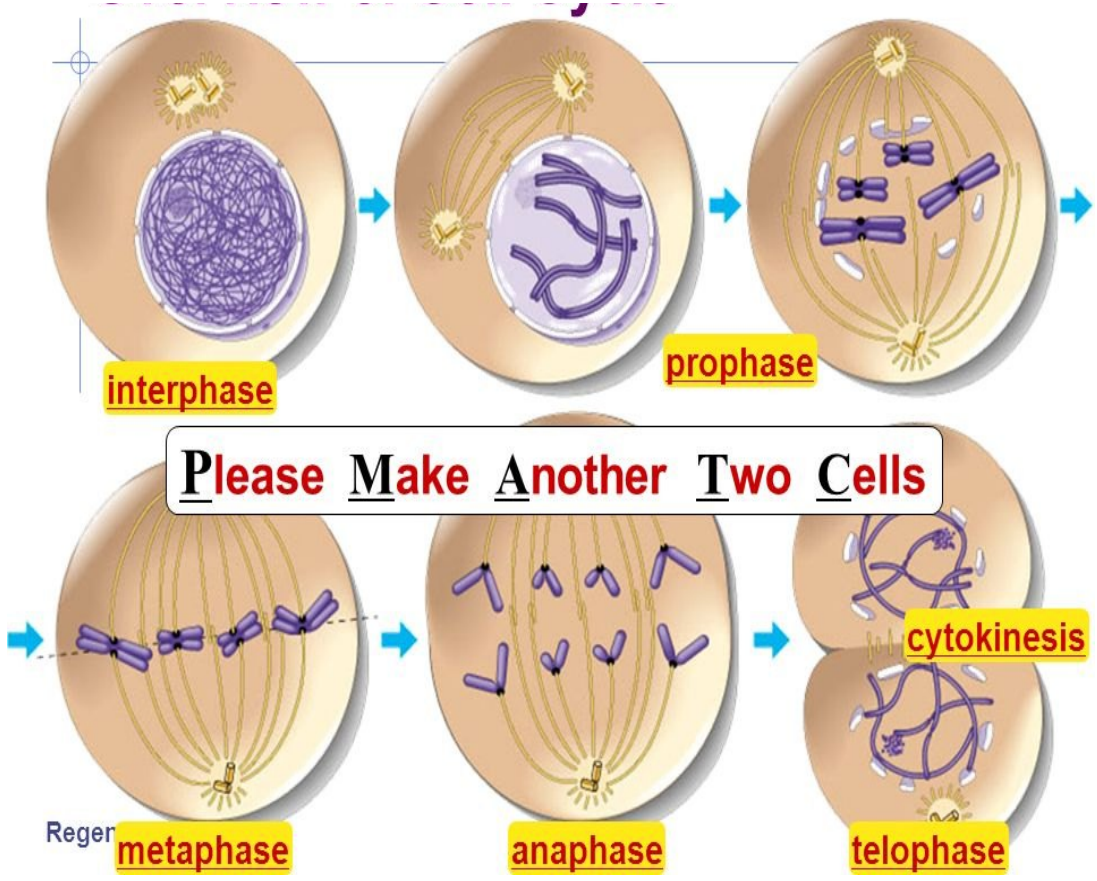
Size of the chromosome

1- Interphase: long & thin.

2- Division:

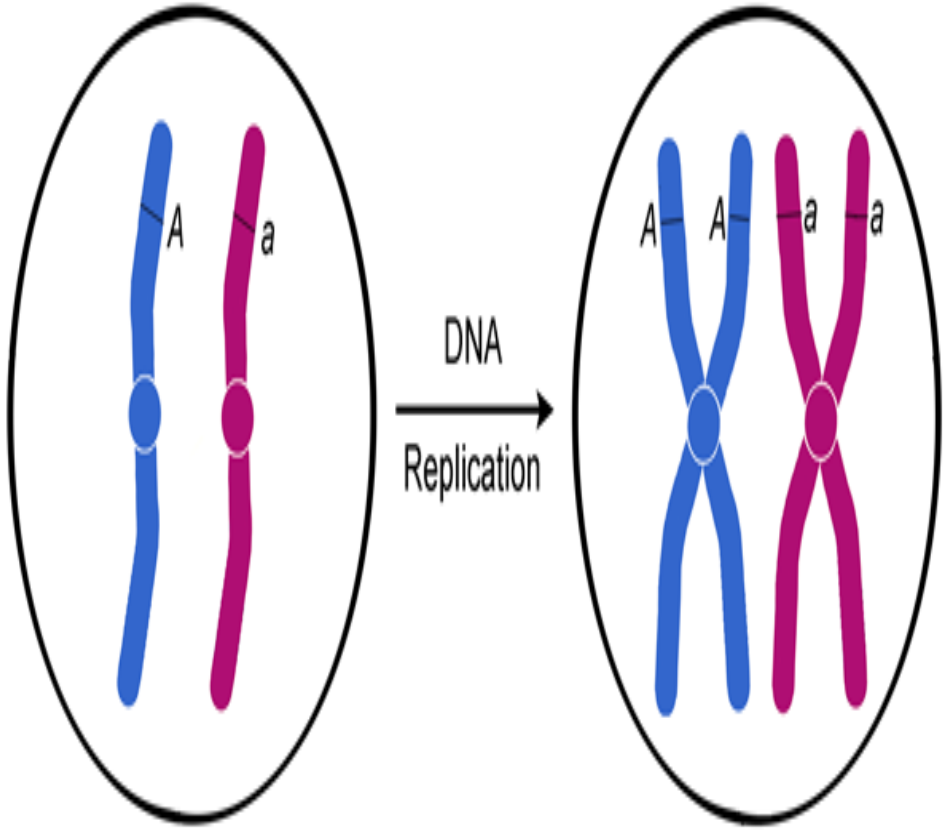
★ Prophase: progressive decrease in its length accompanied by increasing its thickness.

★ Metaphase: very thick & quite short (easily recognized).



Types of chromosomes
A- According to the amount of the DNA

- **Single-chromosomes (s-chromosomes)** made of one DNA molecule (interphase chromosomes = chromatin or chromatids).
- **Double-chromosomes** (d-chromosomes; mitotic chromosomes): are formed during the S phase. Each d-chromosome is formed of two chromatids, linked at the centromere. Each chromatid is made of a DNA molecule.



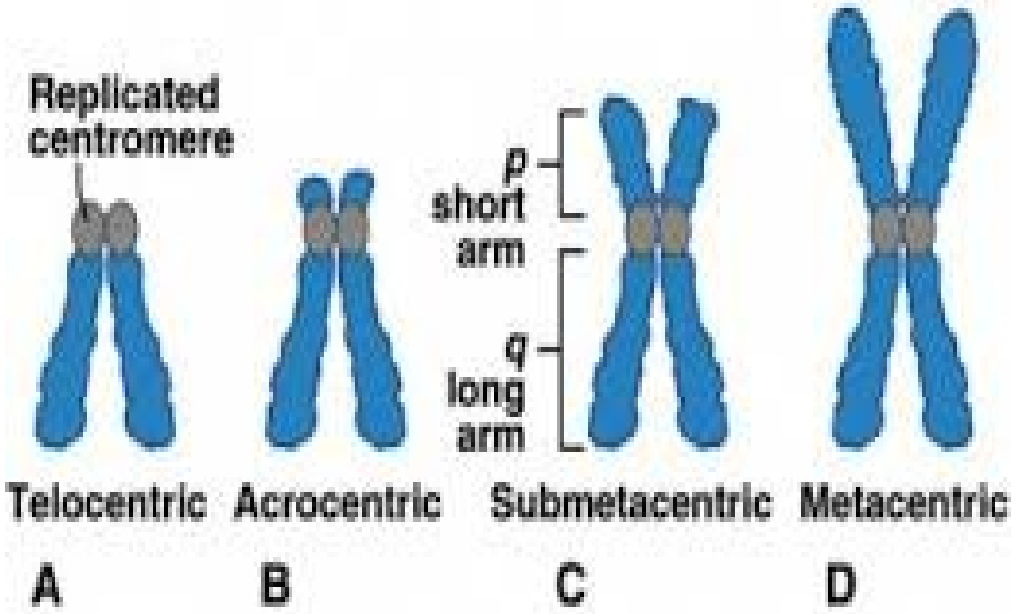
Types of chromosomes
B- According to position of centromere

1- Metacentric: the centromere *in the middle.*

2- Submetacentric: the centromere is displaced *slightly away* from the center.

3- Acrocentric: the centromere is displaced *away* from the center.

4- Telocentric: the centromere is positioned *at the very end* of the chromosome.



Types of chromosomes
C- Autosomal or sex chromosome

1- Autosomal chromosomes:

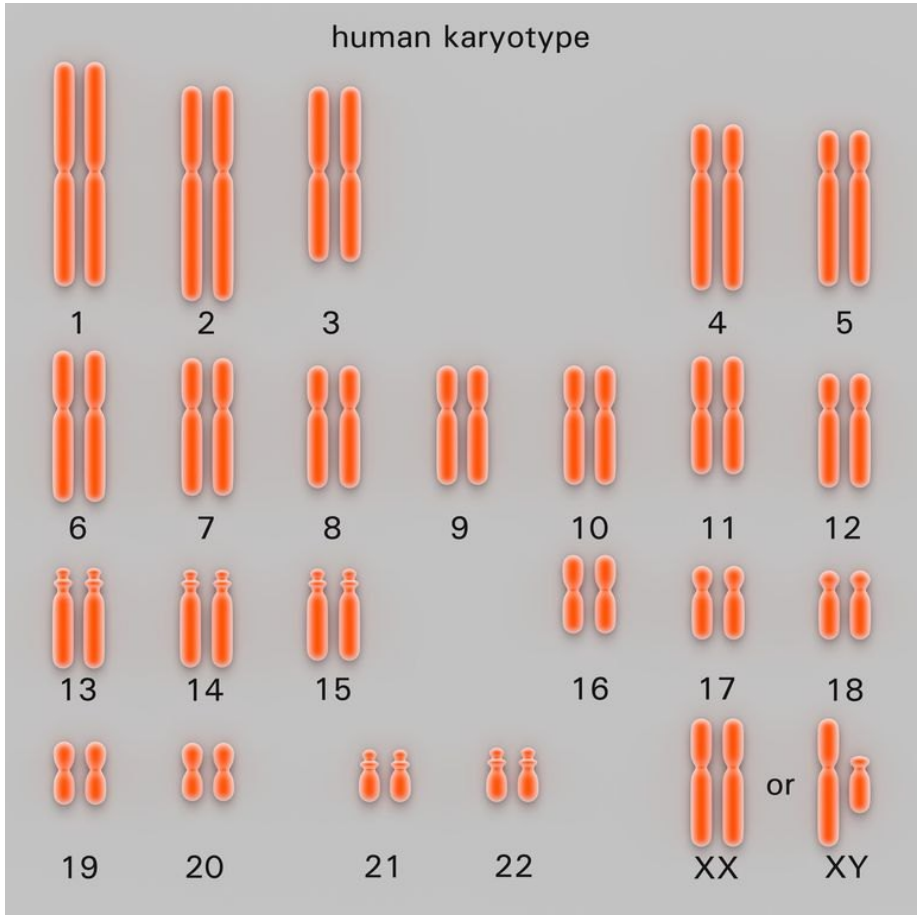
★ Paired chromosomes with the same length, shape, position of the centromere & genetic information.

★ Carry the genes for *somatic characteristics*.

2- Sex chromosomes:

★ Paired chromosomes differ between males & females.

★ Carry genes for *sex characteristics*.

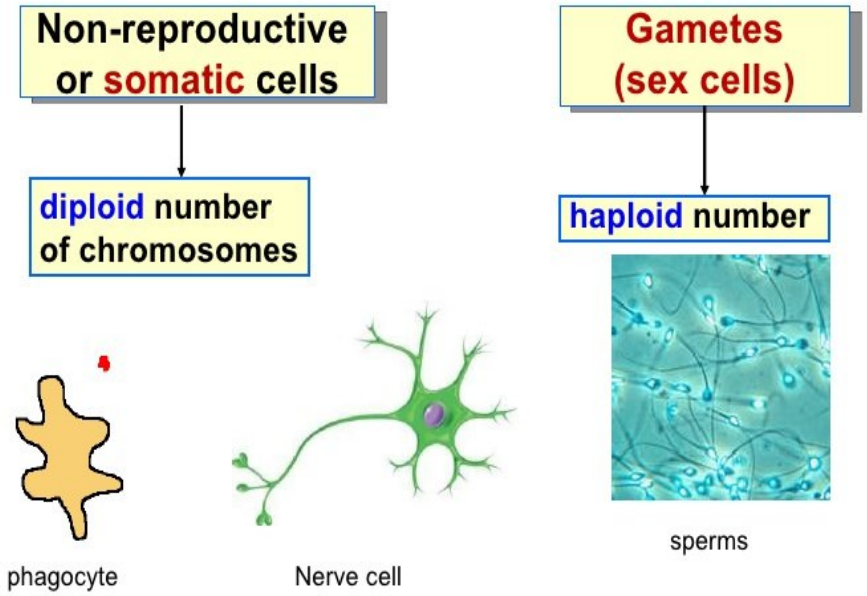


X chromosome	Y chromosome
It is a sex chromosome, that occurs paired in female& single in males	It is a sex chromosome, that is presentnormallysingle in males
In female: one X chromosome is inherited from the mother& the other	In male: One chromosome i inherited from the father.
Contains genes for female sex determination.	Contains genes for male sex determination.
Bigger in size. Contains about 1000 genes.	Smaller. Contains about 50-60 genes.
Represents 5 % of the entire human genome.	Represents 2% of the entire human genome.

Types of cells in the human body

1- Somatic cells: contain 46 chromosomes (44 autosomal+ 2 sex chromosomes).

2- Germ cells (Gametes= sperm & ovum):contains 23 chromosomes (haploid number; 22 autosomal+ 1 sex chromosome).



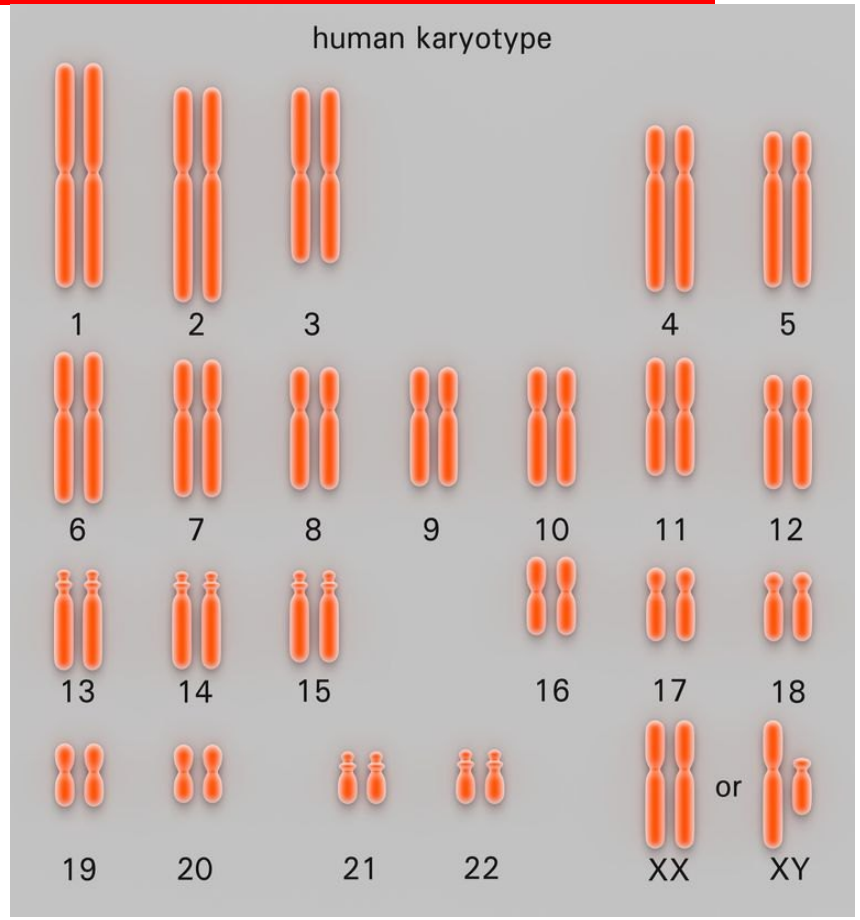
Karyotyping

- The *somatic cell* contains 46 chromosomes.
- **Karyotyping:** is the arrangement of the chromosomes of a person during metaphase into groups of homologous pairs:

★ 22 homologous pairs of *autosomal chromosomes*; the same in length, position of centromere & genetic information,

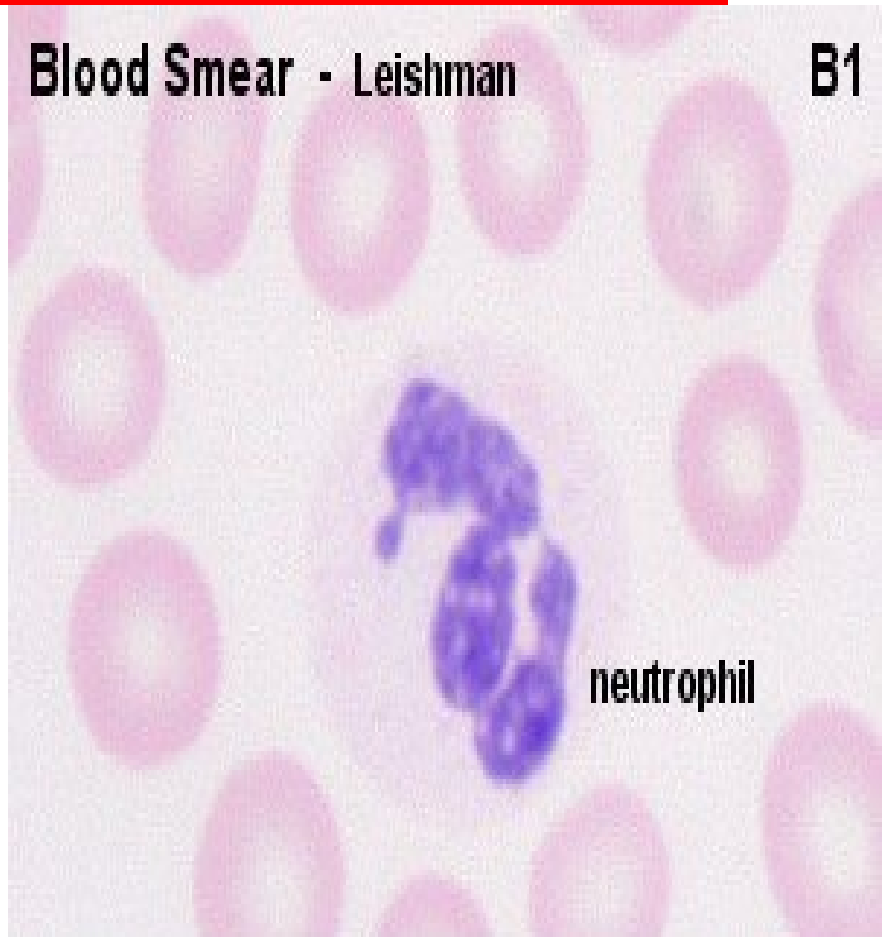
★ One pair of *sex* chromosomes.

★ Each pair contains one chromosome originally derived from the mother & the other chromosome derived from the father.



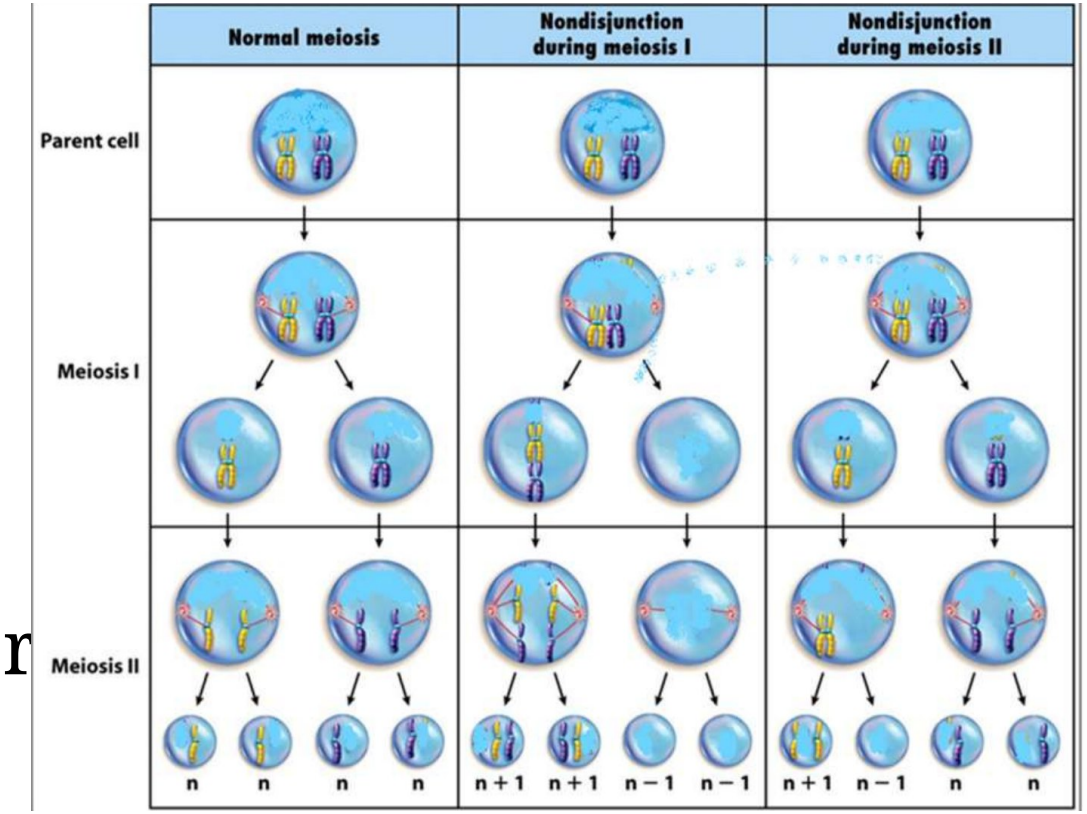
Karyotyping

- In females(44 autosomes +XX) :oneX chromosomeis heterochromatic(*Barr body*), it can be identified in neutrophils, attached tothe nucleusin the form of a drumstick mass.
- Importance: regulates the amount of X-linked gene products being transcribed.



How do chromosome abnormalities happen?

- Chromosome abnormalities usually occur when there is an *error in cell division* in mitosis & meiosis: the correct number of chromosomes is supposed to end up in the resulting cells. However, errors in cell division can result in cells with too few or too many copies of a chromosome.
- Errors can also occur when the DNA are being *duplicated*



How do chromosome abnormalities happen?

- **Factors that can increase the risk of chromosome abnormalities are:**

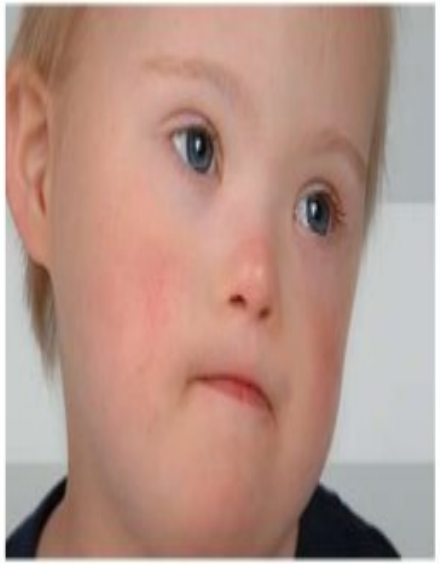
1- Maternal Age: .

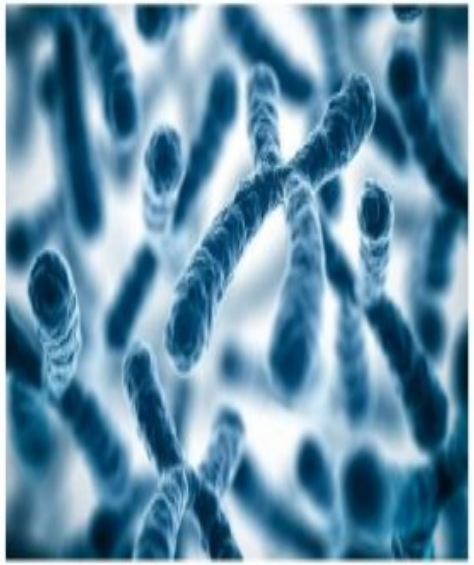
★ *Older women* are at higher risk of giving birth to babies with chromosome abnormalities than younger women because they are born with all the eggs they will ever have.

★ Some researchers believe that errors can crop up in the eggs' genetic material as they age.

★ Because men produce new sperm throughout their lives, paternal age does not increase risk of chromosome abnormalities.

2- Environment it is still possible that the environment may play a role in the occurrence of genetic errors.

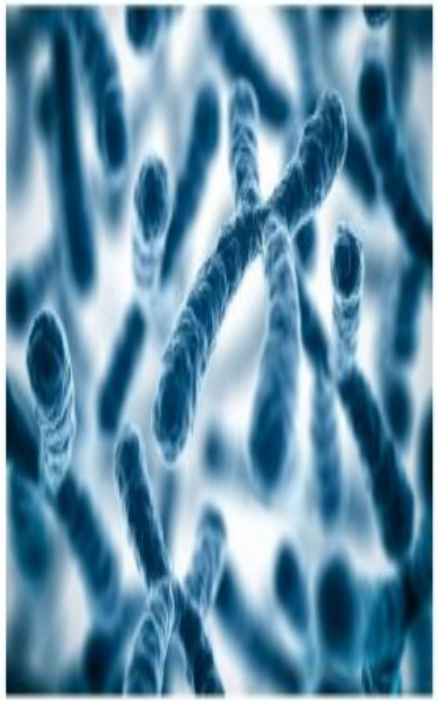




Types of chromosomal abnormalities

1- Numerical abnormalities (aneuploidy): when a whole chromosome from a pair either *missing* (monosomy) or *extra* to the normal pair (trisomy).

2-Structural abnormalities: when part of an individual chromosome is missing, extra, switched to another chromosome, or turned upside down.



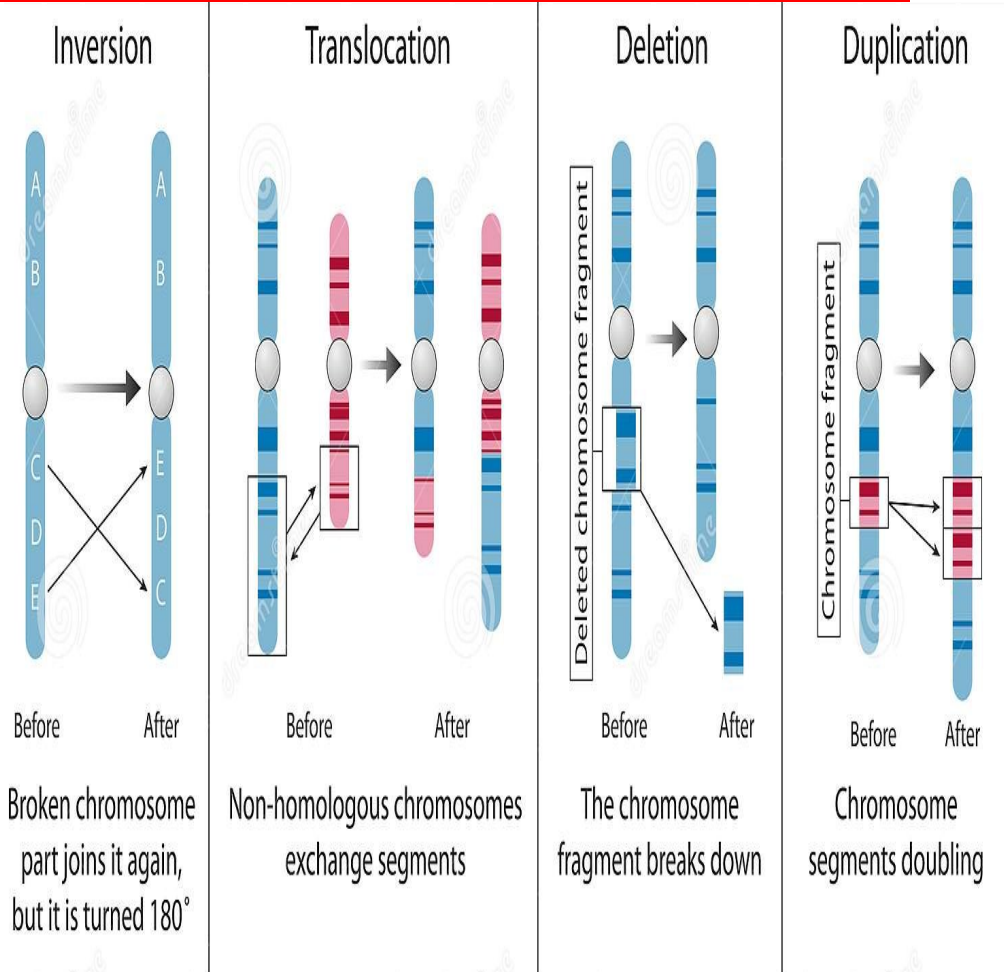
Structural abnormalities

1- Deletion: When a chromosome *breaks* with loss of some genetic materials.

2- Duplication: When a part of the chromosome is *duplicated* (2 copies which means having extra genetic materials)

3- Translocation: When a piece of one chromosome *breaks off & attaches to another chromosome*.

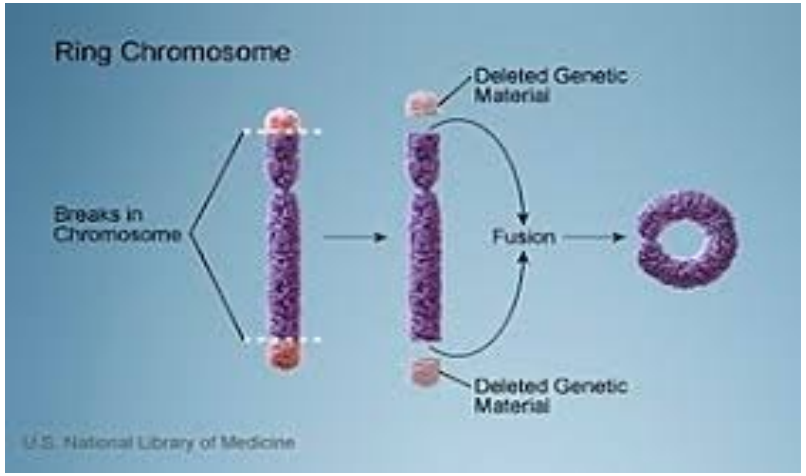
4- Inversion When a piece of one chromosome *breaks off& turned upside down, and reattached to the same chromosome*. As a result, the genetic material is inverted

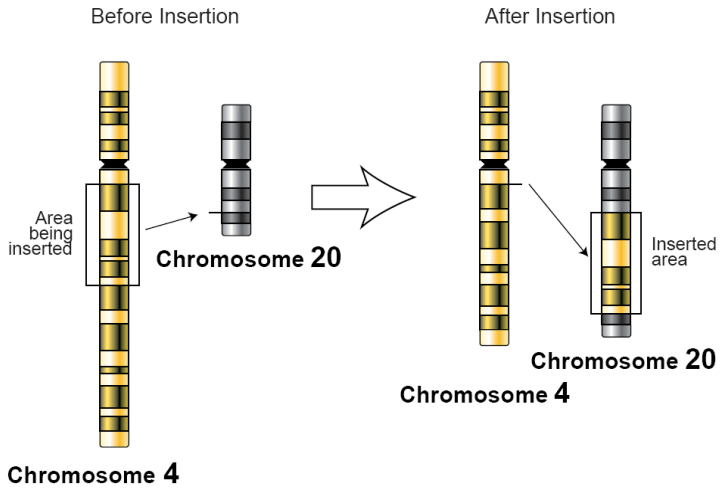


Structural abnormalities

5- Rings: when a chromosome breaks in two places and *its broken ends fuse together.*

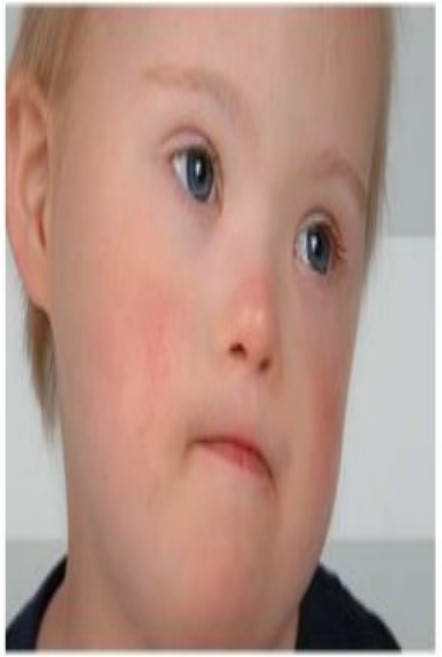
6- Insertions: whena segment of one chromosome *is translocated and inserted* into another non-homologous chromosome (inter-chromosomal insertion), or into a different region of the same chromosome (intra-chromosomal insertion)

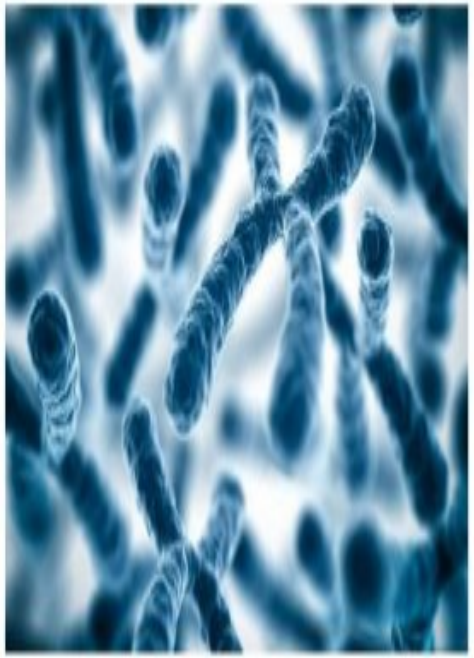




Chromosomal abnormalities

- Most chromosome abnormalities occur as an *accident in the egg or sperm*. In these cases, the abnormality is present *in every cell* of the body.
- Some abnormalities, however, happen *after conception*; then *some cells* have the abnormality and some do not.
- Chromosome abnormalities can be *inherited* from a parent or be "*de novo*" (new to the individual). This is why, when a child is found to have an abnormality, chromosome studies are often performed on the parents.






Thank You