THE KARYOTYPE

Chromosomes which are found in a nucleus of a cell are made up of DNA

Each DNA molecule is made molecules up of large number of *GENES*

GENE: Portion of chromosome that is responsible for specific hereditary trait

Cells:

1. Somatic Cells: - 46 chromosomes arranged in pairs

- They are diploid
- Chromosomal no.: 2n=46
- Chromosomal formula: 2n=46, XX or XY
- Divided by mitosis

Example: Skin cells, Muscle cells, etc…

2. Sex Cells (gametes): - 23 chromosomes that are single\

- They are haploid
- Chromosomal no.: n=23
- Chromosomal formula: n=23, X or Y
- Produced by a process, meiosis
- In males, found in testicles, called sperm
- In females, found in ovaries, called oocyte

Germline Cells:

- They are mother cells gametes
- They are diploid (2n=46)
- Make meiosis to produce haploid gametes
- In males, found in testicles, called *Spermatogonia*
-In females, found in ovaries, called *Oogonia*

Definition of Karyotype:

They are arranged set of chromosomes that are found in a nucleus of a cell

Arranged according to the following criteria

The Criteria:

1. Homologous pairs (Diploid cells)
   → Same size
   → Same position of centromere
   → Same banding pattern (same gene, Not necessary same allele)
2. Decreasing order of size

3. Autosomes first then gonosomes

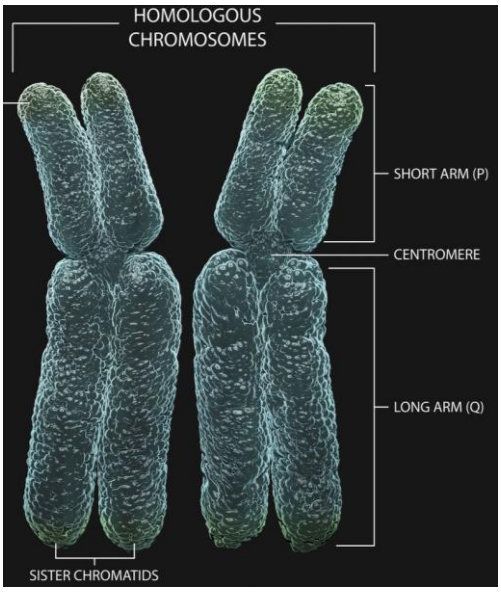
The use of it:

→Determine the gender of the fetus

→Determine the species

→To make sure there is no chromosomal abnormality

(In structure or number)



Paternal Maternal

Title: A schematic diagram showing homologous pair of chromosomes

Gonosomes	Autosomes
→Carry the genetic info that determine the sexual characteristics • Female (diploid) human cell: XX (homologous pair) • Male (diploid) human cell: XY (Non-homologous pair) • Female (Haploid) human cell: only 1 X • Male (Haploid) human cell: only 1 either X OR Y → Thus, The *FATHER* determines the *SEX* of the baby←	→ Carry the genetic info that determines: - Traits - Phenotype • Human (diploid) cell: 44 autosomes or 22 pairs • Human (haploid) cell: 22 autosomes

Explanation of steps of Karyotyping:

- Blood is collected, and WBC (white blood cells) are separated to be used in karyotyping since they have nucleus (has chromosomes)

- WBC are cultured in order to divide (make mitosis)

- Colchine is added in order to stop mitosis at metaphase,

where the chromosomes are arranged in the center of the cell

- The cells are placed in a hypotonic solution, where it enters the cell and it bursts, and the chromosomes are released

- They are stained and observed under the microscope.

REMARKS:

- RBC (red blood cells) can't be karyotyped since they have no nucleus (no chromosomes)
- In mitosis, chromosomes are coiled and condensed so they are easy to observe under a microscope
- Hypotonic solution = Rich in water

Mode of Action of Colchine:

1. Breaks the spindle fiber
2. Locks mitosis at metaphase

TRISOMY: Presence of an extra chromosome

→ Abnormal Karyotype

→ Abnormal Phenotype

Chromosomal number: 2n+1=47

Chromosomal formula: 2n+1=47, XX or XY

MONOSOMY: Absence of a chromosome

→ Abnormal Karyotype

→ Abnormal Phenotype

Chromosomal number: 2n-1=45

Chromosomal formula: 2n-1=45, XX or XY

TRANSLOCATION: The chromosome is present, but it's placed on another chromosome

→ Abnormal Karyotype

→ Normal Phenotype

Chromosomal Abnormality:

Numerical Abnormality:

Autosomal:

Example: Trisomy 21 (Down Syndrome)

Karyotype: extra autosome 21

Gonosomal:

Example 1: Turner syndrome (female)

Karyotype: Missing X gonosome

Example 2: Klinefelter Syndrome (Male)

Karyotype: Extra X gonosome

Structural Abnormality:

Abnormal shape of the chromosome

Mitosis

Definition: Process of division of diploid human cells where it divides to 2 daughter cells that are identical to themselves and the mother cell.

Aim: 1. Regeneration of dead/old/injured cells

2. Maintain biological identity

3. Growth of the organism

RULE: 2^n=---

n: number of mitotic

2^n: number of daughter cells

Cell cycle = Interphase + Mitosis

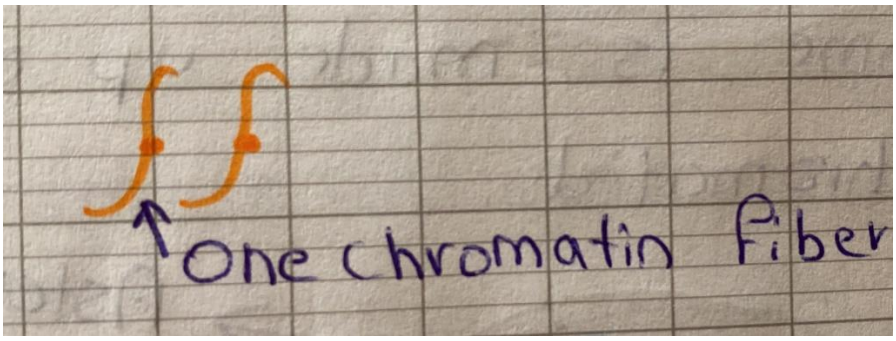
Interphase: G1, S, G2

Before each mitosis, there is interphase

Interphase: It's the phase where the cell prepares itself for division.

G1, Growth/ Gap 1:

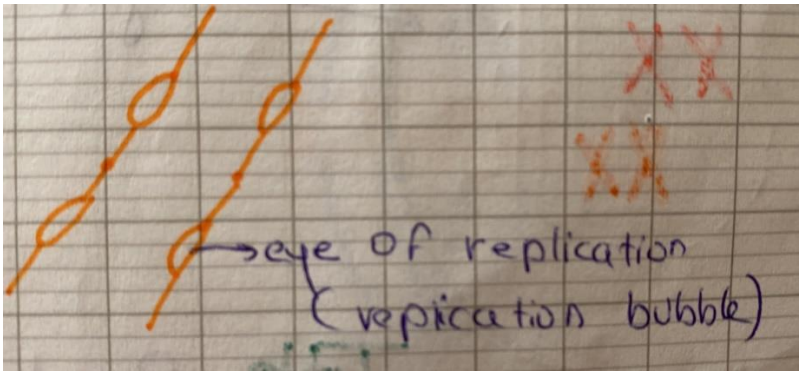
During this phase, the cell begins to grow in size and produce specific protein needed in the next phase.



Title: Aspect of chromatin fiber during G1

S, Synthesis:

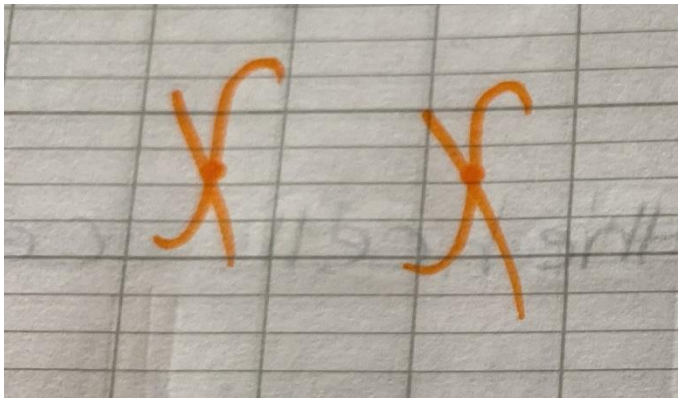
During this phase, the cell replicates its genetic info (Chromatin Fiber) in order to divide equally.



Title: Aspect of chromatin fiber during S

G2, Growth/Gap 2:

During this phase, the cell continues to grow in size and replicates all it's organells



2 duplicated chromatin fiber

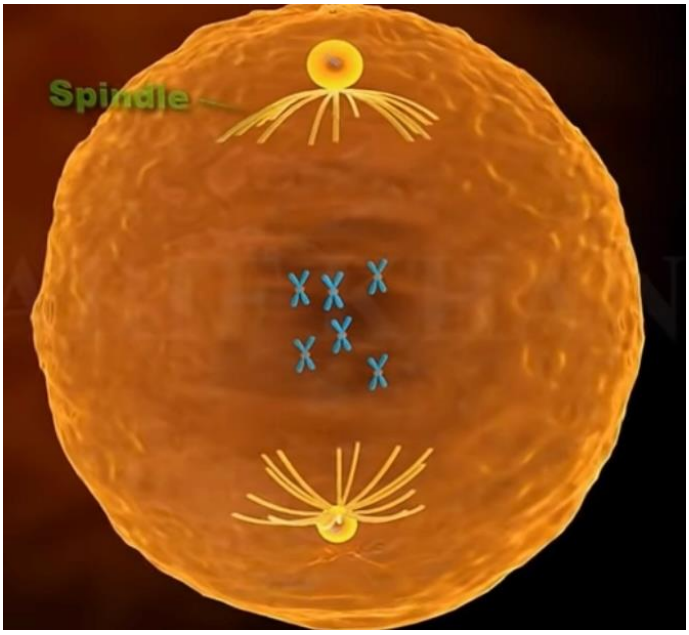
Title: Aspect of chromatin fiber during G2

Mitosis:

Mitosis: -Prophase –Metaphase –Anaphase -Telophase

Prophase (Animal Cell):

- Nuclear membrane starts to disappear
- Centrosome divide into centrioles
- Centrioles turn into aster
- Spindle fiber arise from aster
- Chromatin fiber starts to coil and condense to turn into chromosomes
- Each chromosome is made up of 2 sister chromatids



Title: Prophase in an animal cell.

Prophase (Plant Cell):

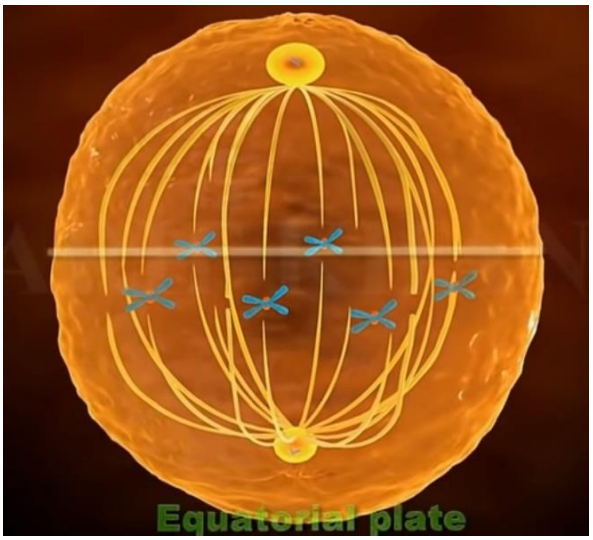
-In a plant cell, same changes occur in prophase, but there is no aster (no centrosome) so spindle fiber arise from cytoplasm

Metaphase (Animal & Plant Cells):

-Chromosomes are attached to the spindle fiber by their centromere

- Each chromosome made up of 2 sister chromatids

- Chromosomes are arranged at the equatorial plate

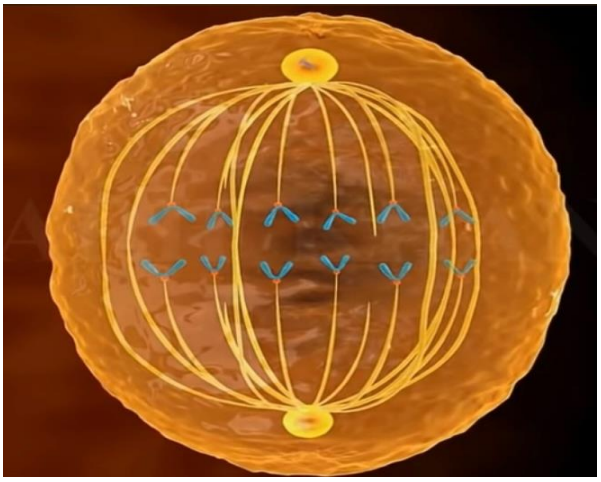


Title: Metaphase in animal cell.

Anaphase (Animal & Plant Cells):

-Spindle fiber shortens, separating the 2 sister chromatids into the opposite poles of the cell

- Each chromosome is made up of 1 chromatid



Title: Anaphase in an animal cell

Telophase (Animal Cell):

- Spindle fiber disappears

- Aster turn back to centrosome

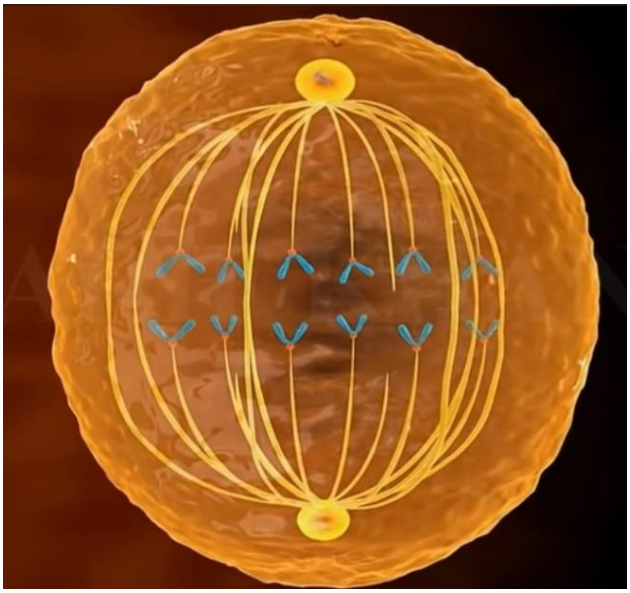
- Chromosomes de-condense and uncoil to become to become long thin chromatin fiber

- Each chromosome made up of 1 chromatid

- Nuclear membrane reappears around each set of genetic info

- The cell begins to constrict in the middle forming cleavage-furrow, in order to divide equally into identical daughter cells

(Process called ***CYTOKINESIS***)



Title: Telophase in an animal cell

Telophase (Plant Cell):

-In plant cell, there is no constriction due to the hard cell wall. So, a new cell wall appears in the middle of the cell, where it grows in order to divide the cell into 2 daughter cells.

NOTES:

1) Q → Quantity of DNA (chromosomes)

2) Quantity of chromosomes varies but the number remains the same (2n)

3) Duration of interphase is longer than that of mitosis

4) Less DNA → Faster Mitosis

5) Different types of cell in the body, have different durations of cell cycle

6) If duration of cell cycle → Long

=> Cell divides slowly

7) If duration of cell cycle → Short

=> Cell divides rapidly

8) The duration of skin cell cycle is faster than that of bone

=> Cell division of skin is fast

Meiosis

Definition: Process where the diploid mother cells divides to 4 haploid daughter cells which known as gametes

In Males: Found in testicles

In Females: Found in ovaries

Aim:

-Production of male gametes (sperm)
→ Its process: ***SPERMATOGONIA***

-Production of female gametes (oocyte)
→ Its process: ***OOGENESIS***

Sexual Reproduction: Meiosis (production of gametes)

Fertilization: Union of sperm and oocyte to give fertilized egg

NOTES:

- Oocyte ➔ Ovum (➔: Fertilization)

- Mitosis: 2n → 2n

Meiosis: 2n → n

Fertilization: n → 2n

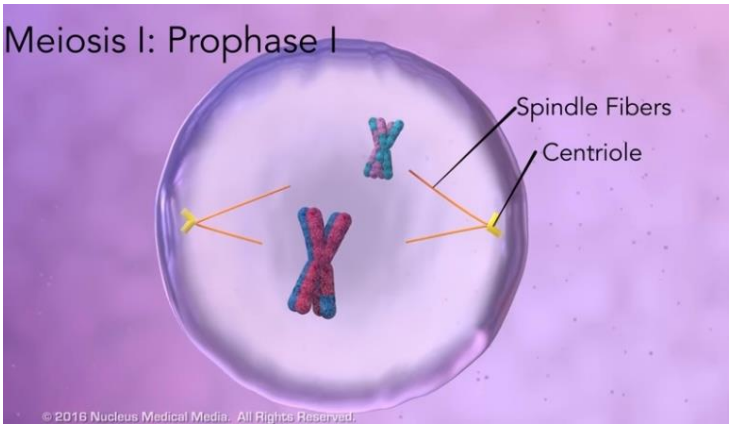
IMPORTANT: Meiosis leads to the production of gametes that are genetically different, this causes the genetic diversity.

- Each meiosis is preceded by interphase
- Meiosis has 2 divisions:
    - Meiosis I
    - Meiosis II

Meiosis I (**REDUCTIONAL** Division):

Prophase I:

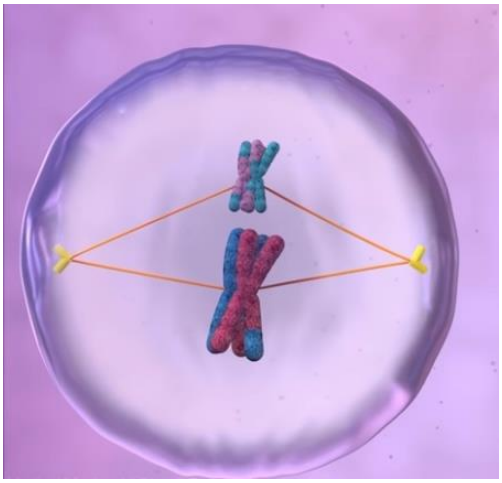
- Same changes occur just like Prophase in Mitosis, but with a major difference: Homologous pair of chromosomes join together forming a tetrad
- Tetrad: 4 chromatids of paired homologous chromosomes.



Title: Prophase I in a cell.

Metaphase I:

-Tetrads are arranged at the equatorial plate (attached to the spindle fiber by their centromere)



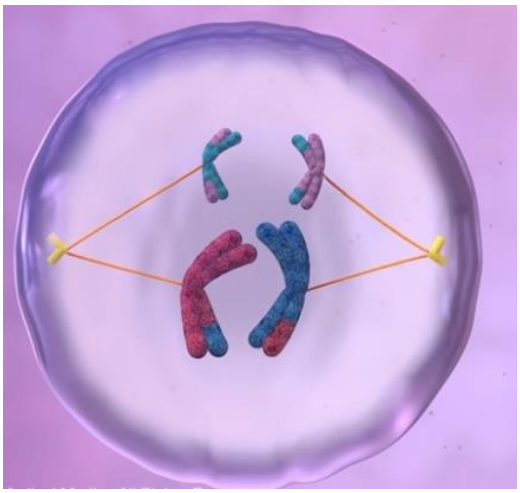
TITLE: Metaphase I in a cell.

Anaphase I:

- The spindle fiber shortens separating the homologous airs to the opposite pokes of the cell

- Each chromosome made up of 2 chromatids

- The chromosomal number is reduced from 2n → n



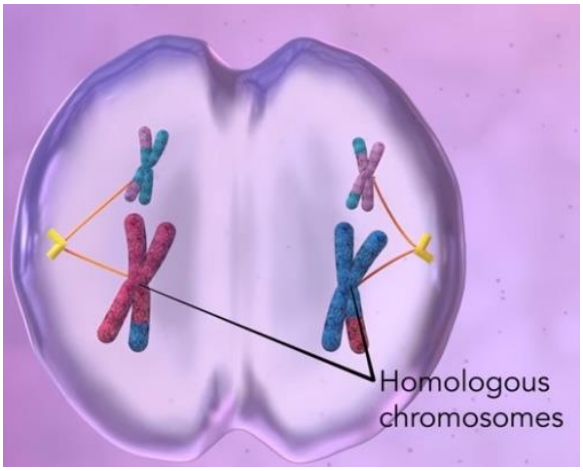
Title: Anaphase I in a cell.

NOTE:

If there is an error in the separation of the homologous pair of chromosomes (non-disjunction) this will lead to having gametes either missing or extra genetic info

Telophase I:

- The 2 daughter cells are haploid
- Each chromosome is made up of 2 chromatids
- Number of chromosomes is n

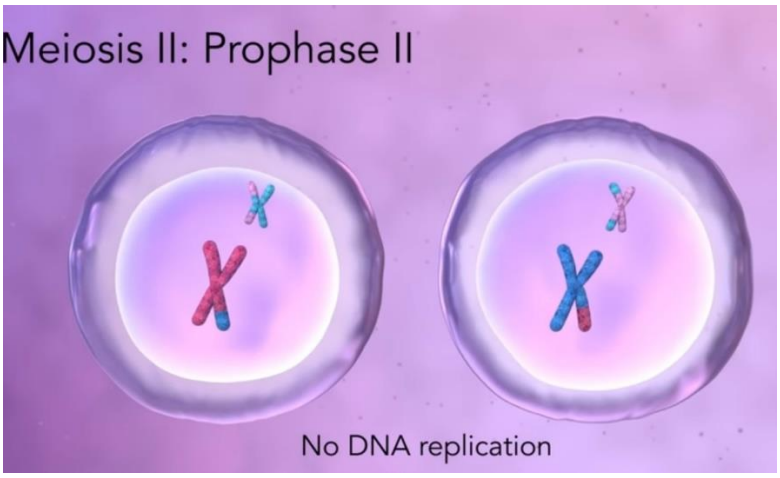


Title: Telophase I in a cell.

Meiosis II (**EQUATIONAL** Division):

Prophase II:

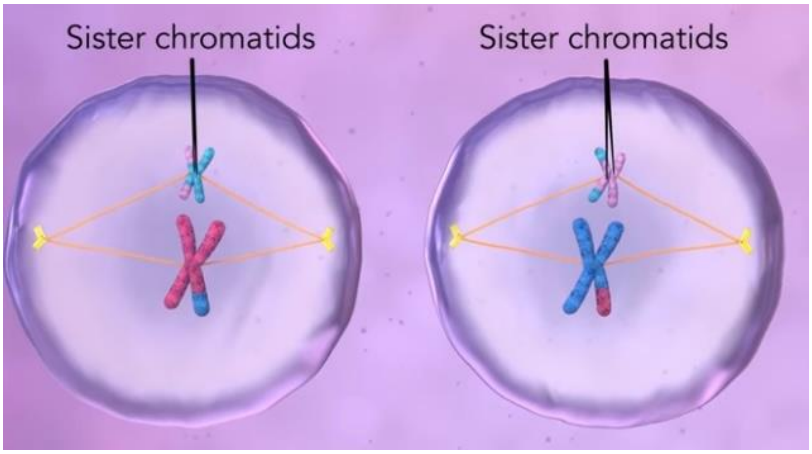
Same changes as mitosis



Title: Prophase II in a cell.

Metaphase II:

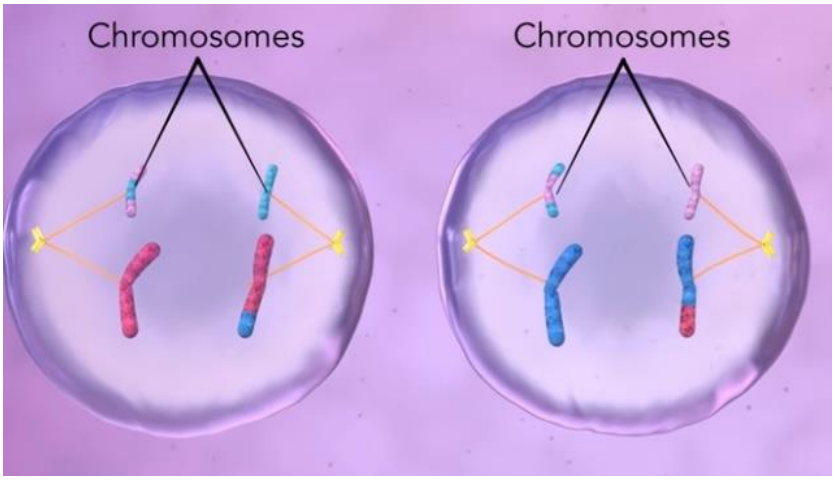
Chromosomes arranged at the equatorial plate (attached to the spindle fiber by their centromere)



Title: Metaphase II in a cell.

Anaphase II:

The spindle fiber shortens, separating the sister chromatids to the opposite poles of the cell



Title: Anaphase II in a cell.

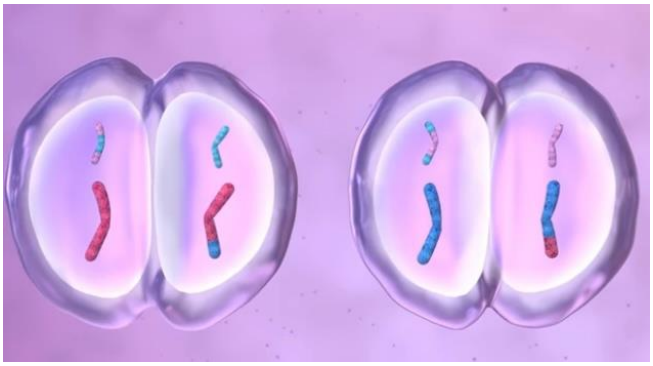
NOTE:

If there is an error in Anaphase II of Meiosis II, → Non-disjunction of chromatids, it leads to having 2 cells either missing or extra chromosome.

Telophase II:

- Each cell will divide to give 2 daughter cells resulting 4 daughter cells (haploid) gametes that are genetically different

- Each chromosome is made up of 1 chromatid



Title: Telophase II in a cell.