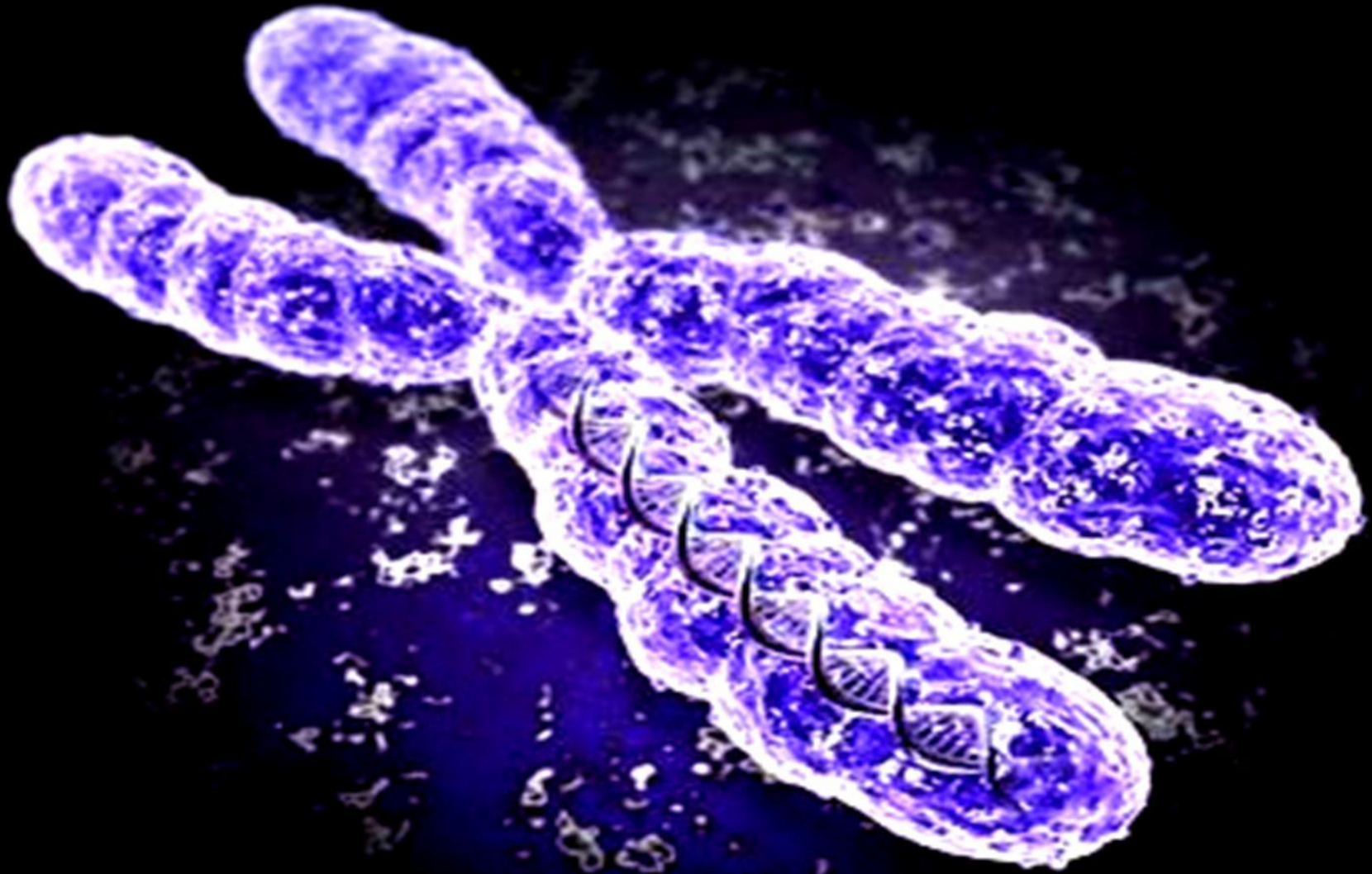
Chapter 6
Section 1

Chromosomes
&
Cell Reproduction



- **Reproduction:**

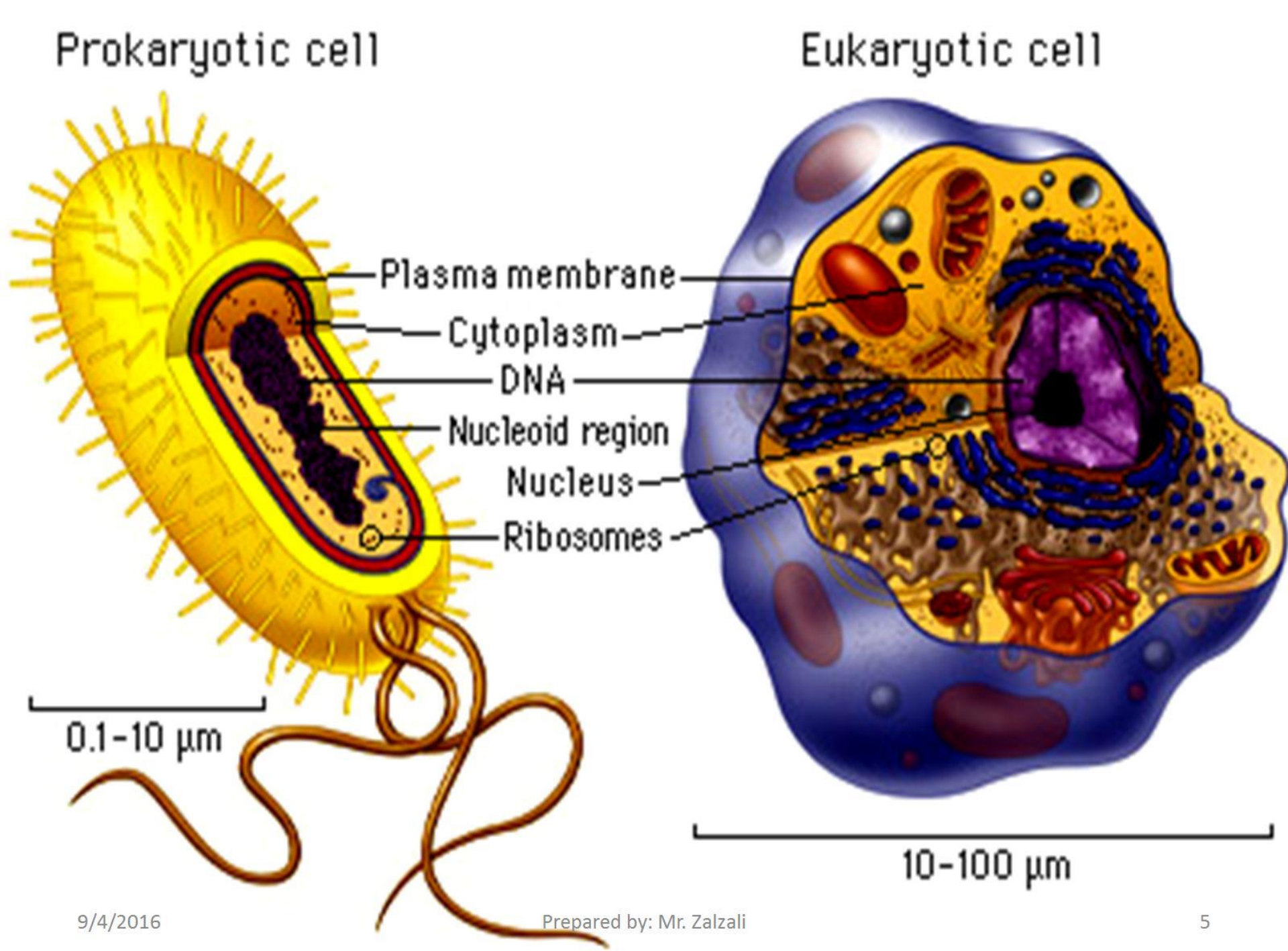
  -Producing new offspring of same species.

  - Reproduction in all organisms depend on the genetic materials (DNA & RNA).
  - DNA: **D**eoxyribo **N**ucleic **A**cid.
  - RNA: **R**ibo **N**ucliec **A**cid
  - Every cell should divide.
  - So cells division is used for:
    - Growth
    - Development
    - Repairing damaged tissues.
    - Asexual reproduction.

    Note: Multicellular organism carry out sexual and Asexual reproduction.

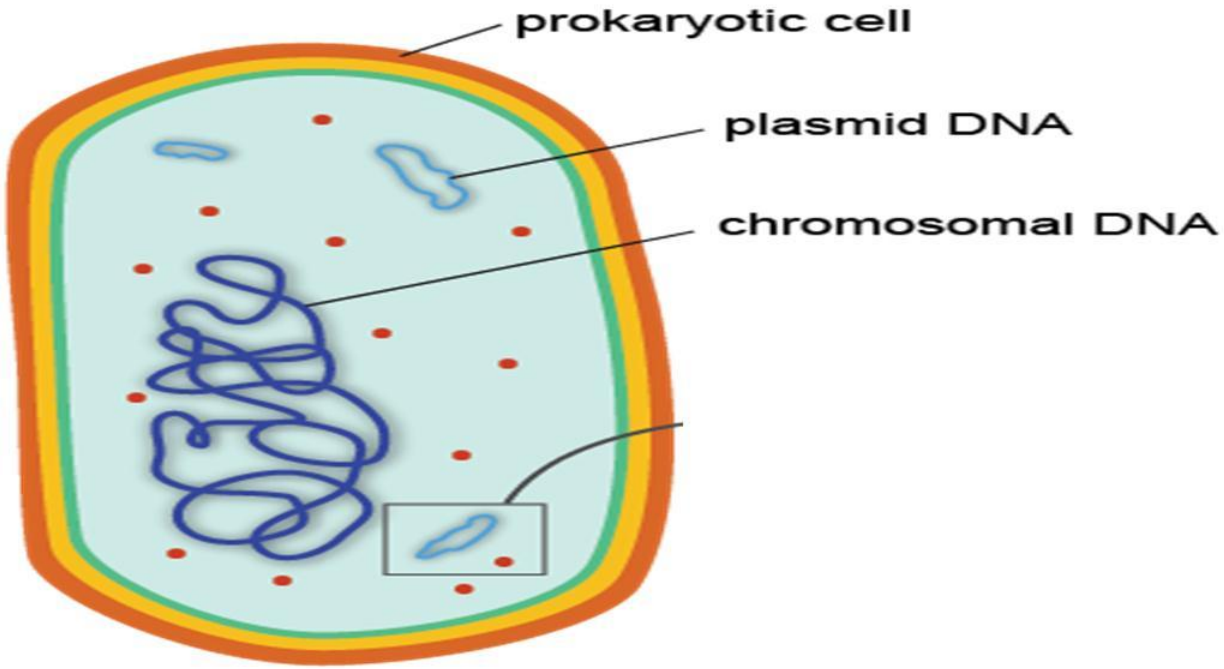
Reproduction

Asexual reproduction	Sexual reproduction
-Involves one single parent only (No male and female)	-Involves 2 parents. (Male and female)
-No formation of gametes -No fertilization	-Involves formation of gametes) -Male and female sex cells fuse during fertilization to combine DNA from male and female
-No genetic variation because all offspring are identical copies of parent and carry same DNA and genes → this will decrease the survival chances of offspring.	-There is great variation because offspring resulted from a new combination of genes from each gametes and this will increase the survival chances of offspring.


Prokaryotic cell
Eukaryotic cell
Plasma membrane
Cytoplasm
DNA
Nucleoid region
Nucleus
Ribosomes
0.1–10 µm
10–100 µm

Living Cells should be:

Prokaryotic cells **(Prokaryotes)**	Eukaryotic cells **(Eukaryotes)**
-Lack Nuclear envelope around DNA or chromosomes (Naked circular DNA)	-Have real nucleus → DNA or chromosomes are surrounded and protected by nuclear membrane.
-Ex: - All species of bacteria. (Kingdom monera)	-Ex: -Human cells - Plant cells -Animal cells -Fungal cells



- **DNA:**
  - Stands for "Deoxyribonucleic Acid
  - Genetic material / complex organic compounds
- **Main functions:**
  - Stores all the genetic information (genetic code) that directs all cell activities and determines its characteristics

- Cell division in prokaryotic cells (Bacteria):

- It's a rapid process→ produces large number of offspring in short time.
- Prokaryotic cell division called "Binary fission"
- Binary fission is a form of "Asexual reproduction".

-Steps of Binary Fission:

1- When the cell reaches a certain size and conditions are favorable → DNA replicates i.e. it copies itself and form 2 exact replicas.

2- Cytoplasm divides.

- Cell membrane constricts and pinches inwards.
- New cell wall starts to form between the 2 parts and each contain one copy of DNA.

3- Two identical daughter bacterial cell are produced and each carries the same characteristics of the parent cell..

- **Cell division in Eukaryotes:**

2- Mitosis

-Form of asexual reproduction in eukaryotes.

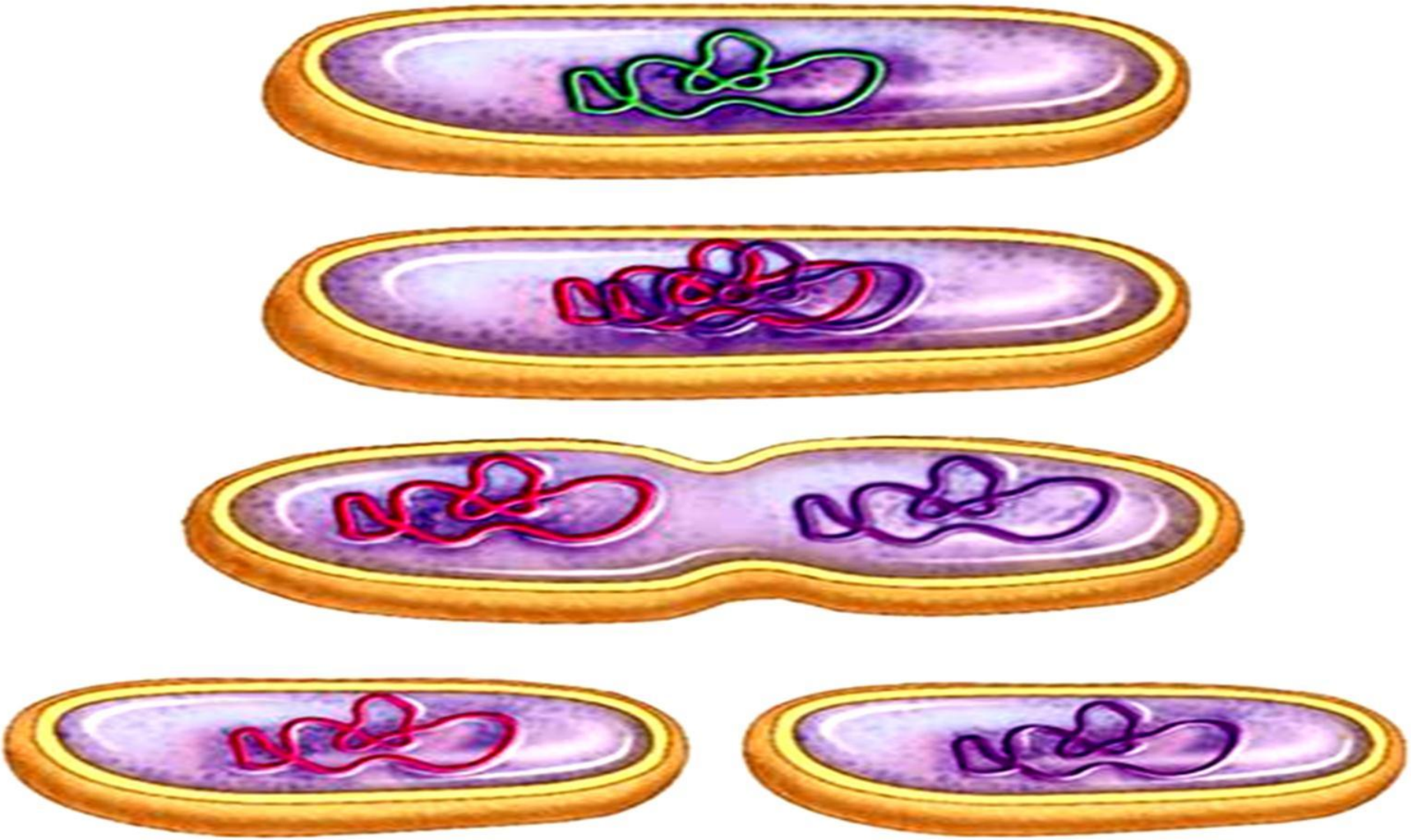
- For growth, development and repairing damaged cells and tissues.

3- Meiosis:

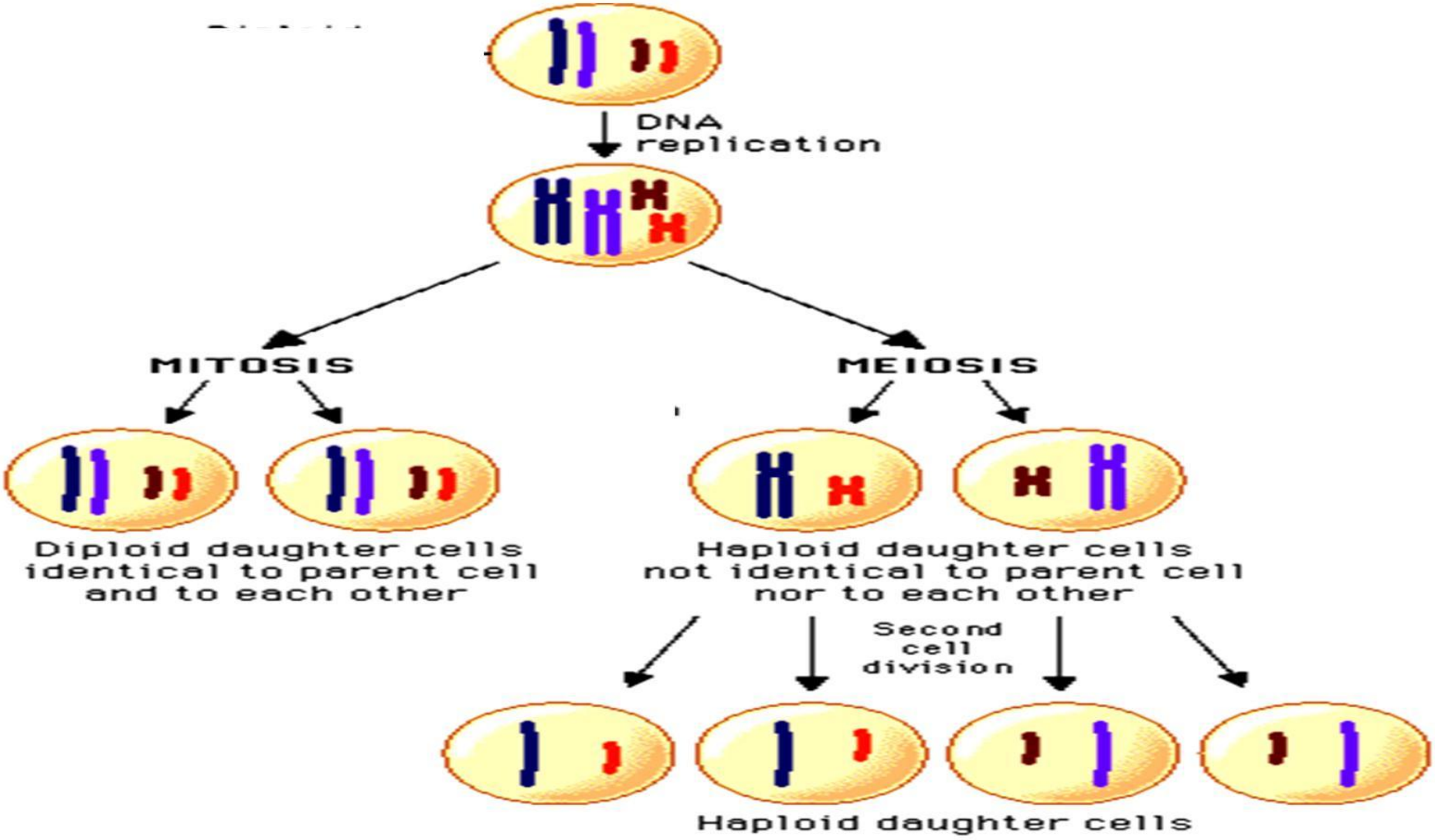
-Form of Sexual reproduction in eukaryotes.

-Involves production of gametes in reproductive organs.

Cell Division in prokaryotes
"Binary Fission"



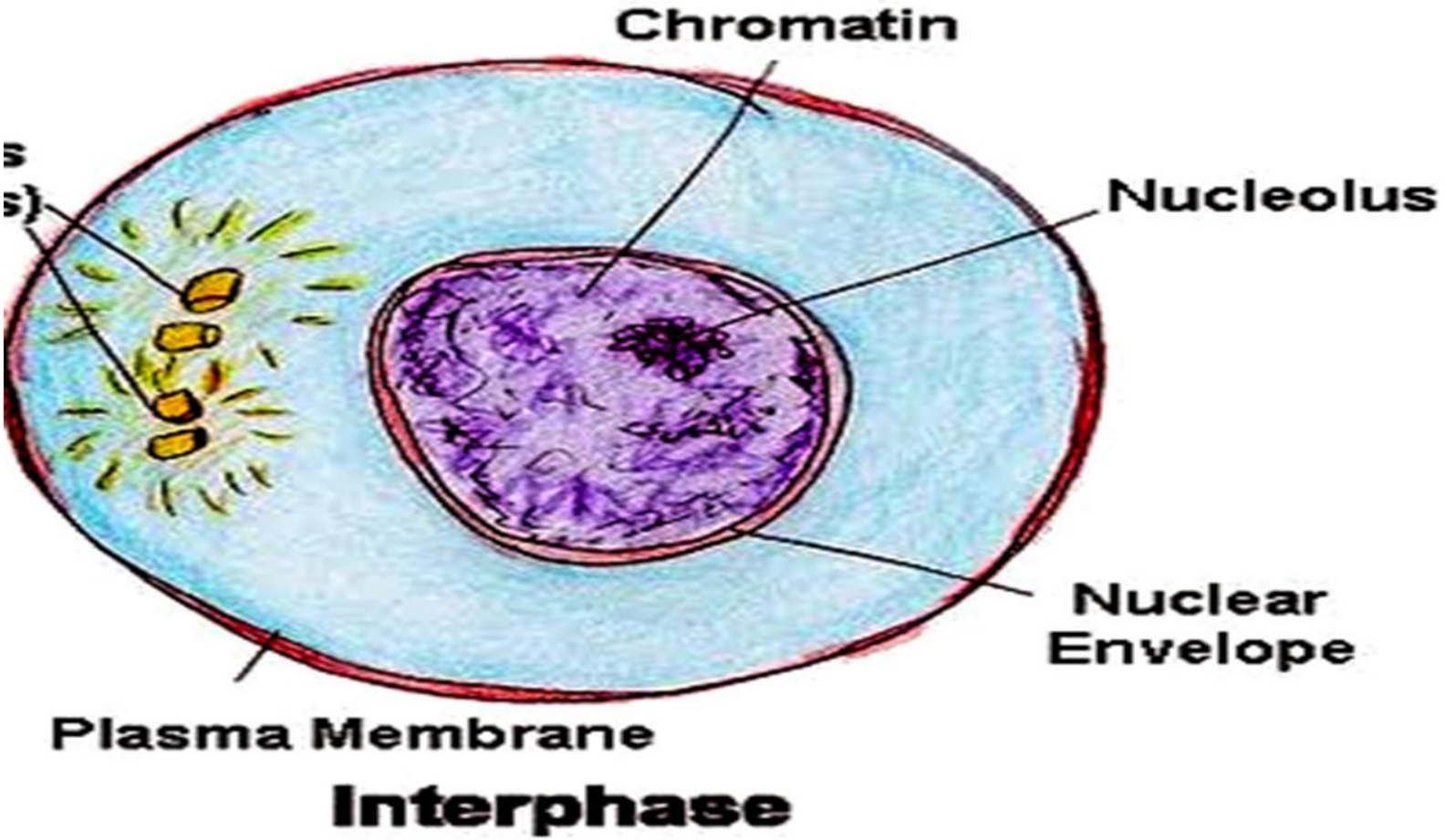
Cell division in Eukaryotes



Eukaryotic cell before division

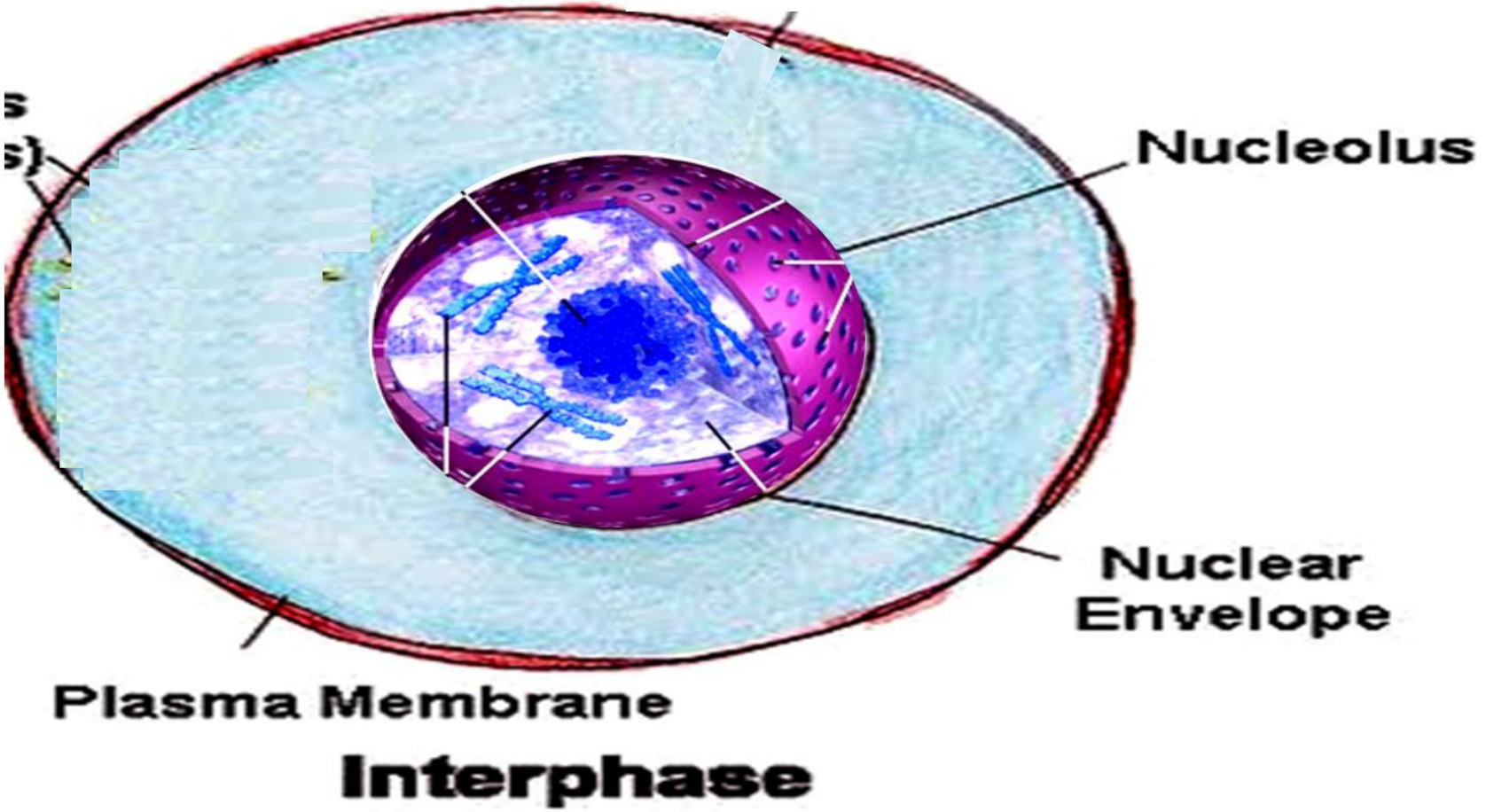
-Chromosomes are made up of genetic material "DNA" .

-Chromosomes cannot be seen under microscope as threadlike structure if the cell is not dividing. (chromosomes are only visible when cell division starts).



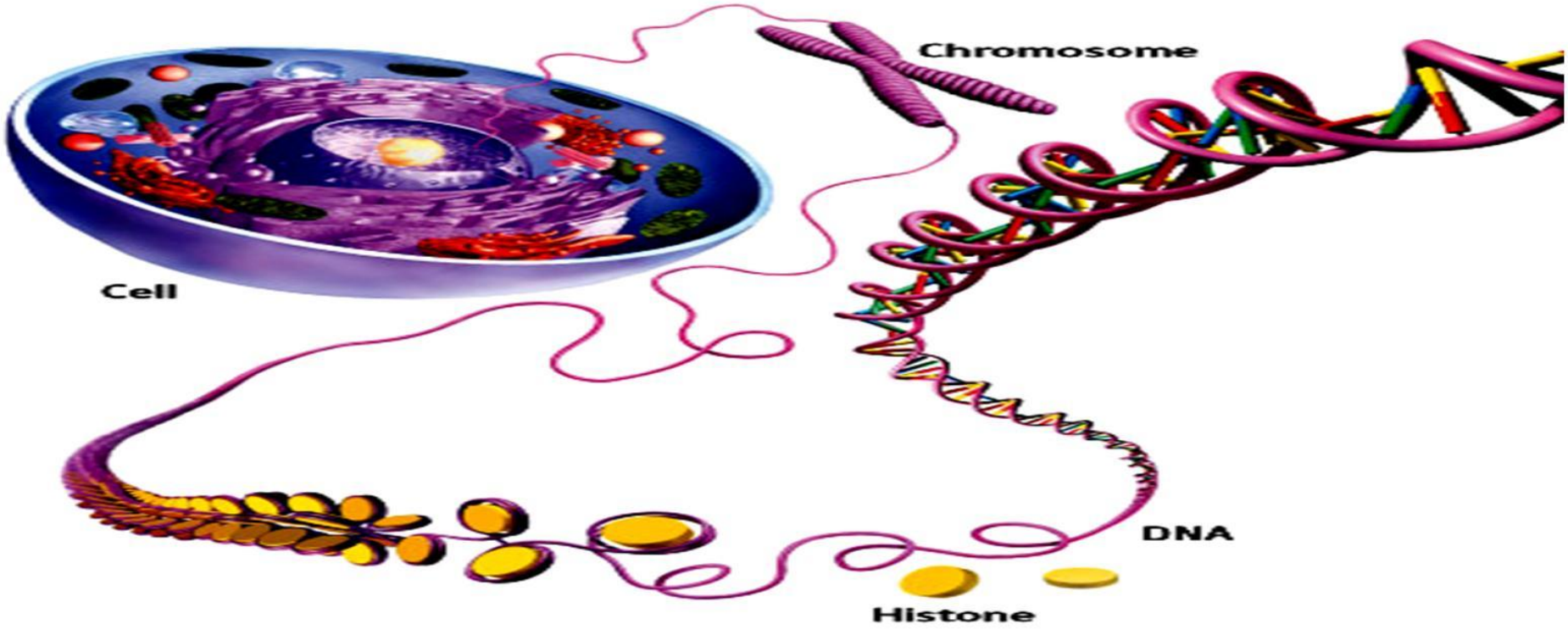
- Eukaryotic cell during division:

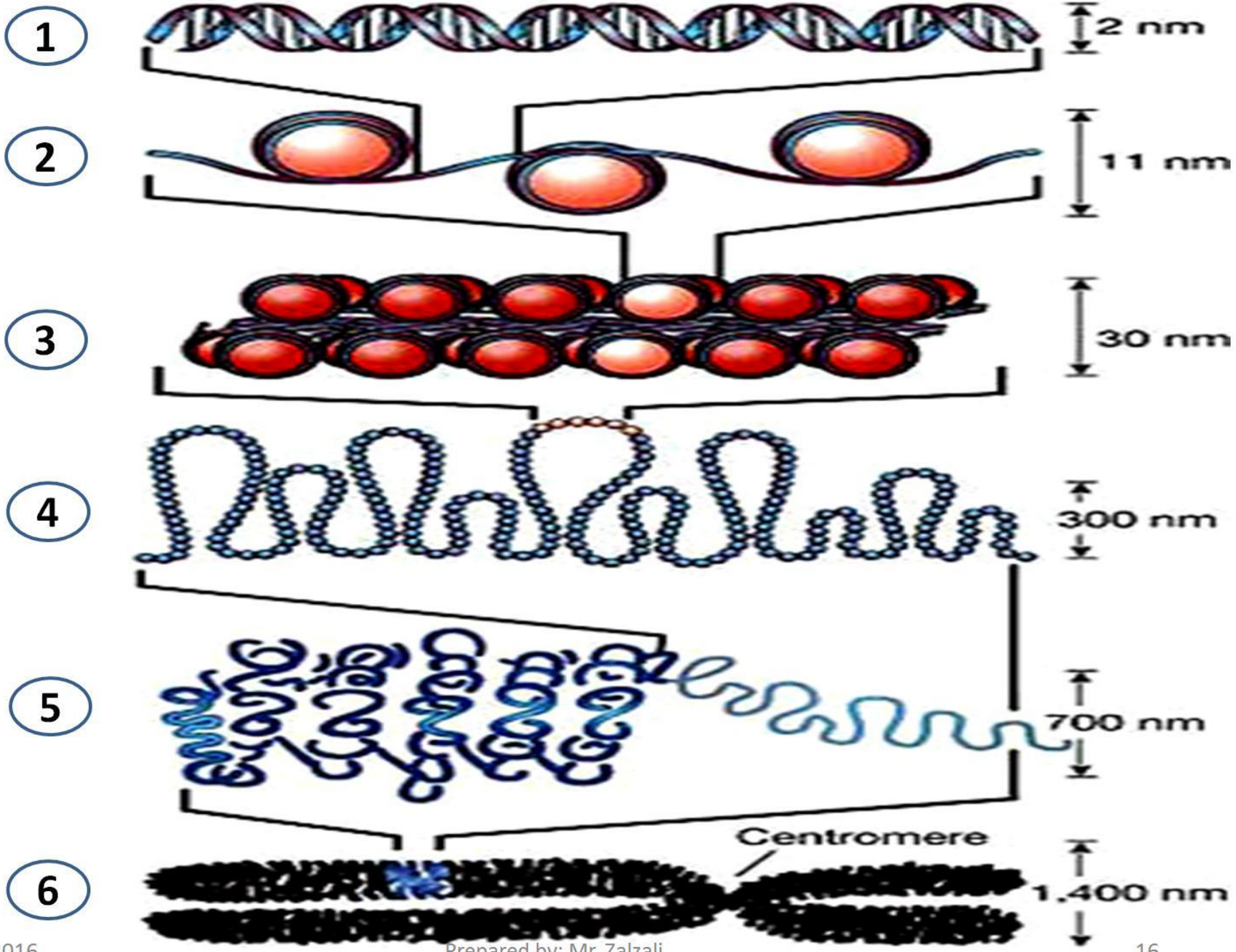
-Chromosomes are visible under light microscope.



Chromosome structure:

- **Before cell division**→ DNA is very long, vey thin, uncoiled and invisible under microscope called "**Chromatin**".
- **When preparing to cell division**:
  - DNA Replicates (copies itself).
  - DNA coils tightly around histone proteins to form shorter, thicker, coiled and visible thread like structure called **"Chromosomes"**

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2 nm
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30 nm
300 nm
700 nm
Centromere
1.400 nm

How do chromosomes form from DNA ?

1- DNA replication occurs 1st when cell begins to divide, making 2 exact copies of itself.

2- DNA coils tightly around histones proteins.

3- Further and further coiling will form sharper, thicker and more visible thread like structures called chromosomes.

* Why is replication important ?

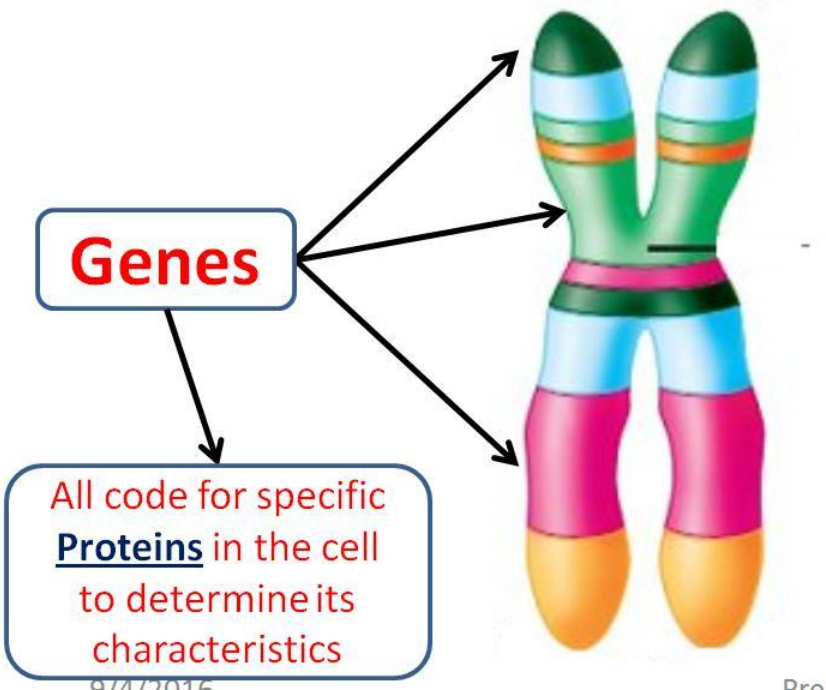
-When the cell begins to divide and DNA replicates. It ensures that one copy of DNA will pass to the new daughter cells, and exact copy of DNA as in the original mother cell.

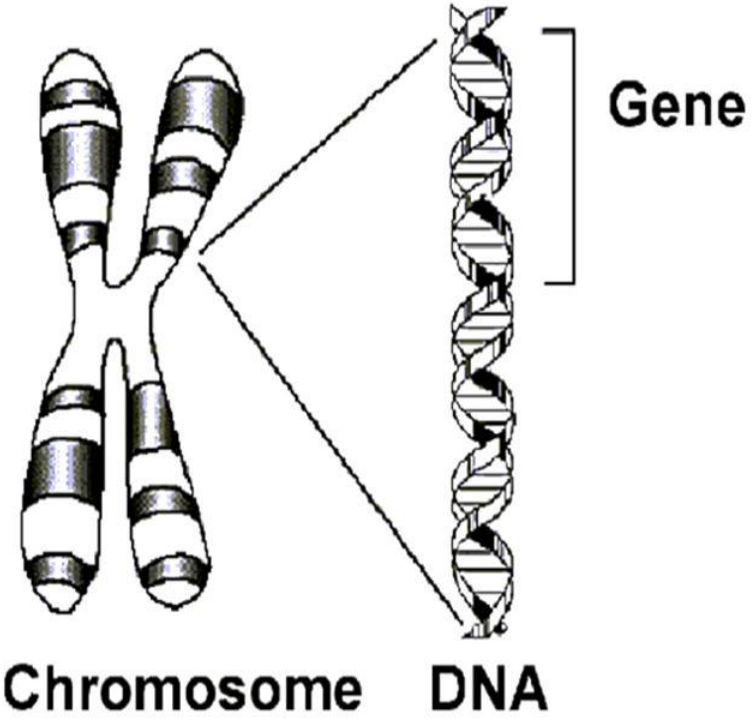
-Genes:

- Is a segment of DNA that codes for specific proteins in each cell.

-Proeins:

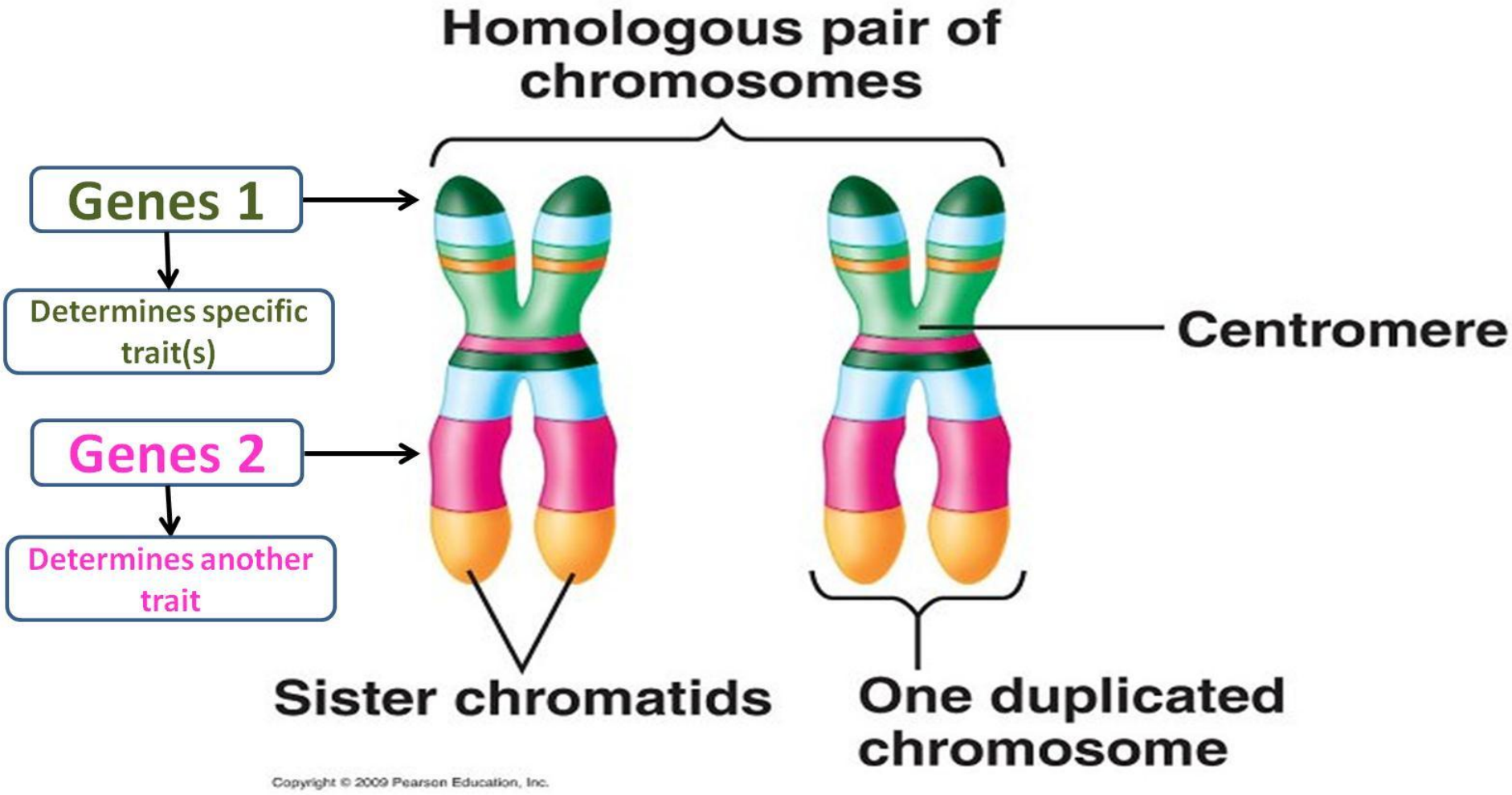
- Are the building blocks of cells to determine the function and the shape of each cell → So the combination of proteins in each cell determines the features of the living organism..



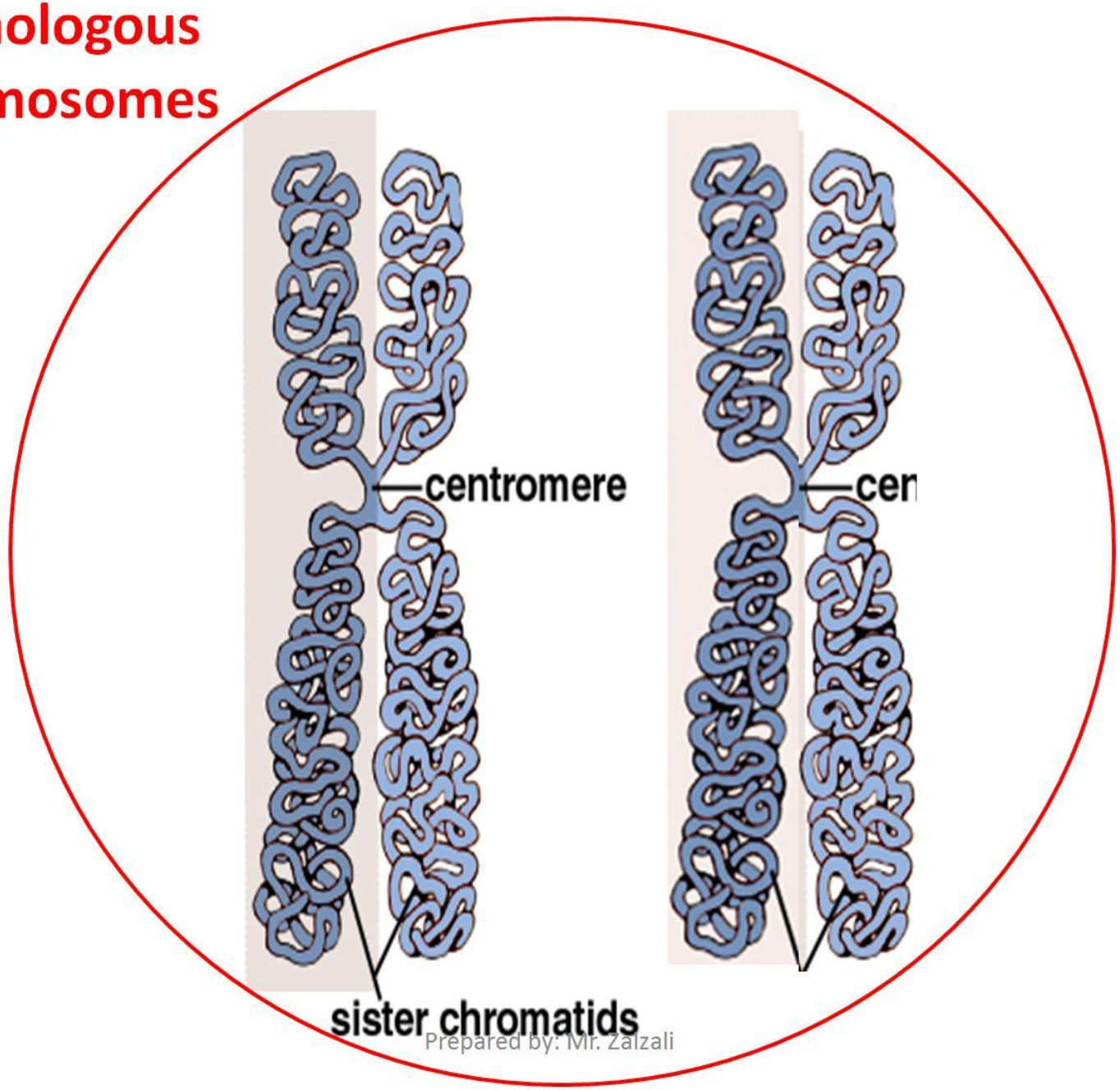


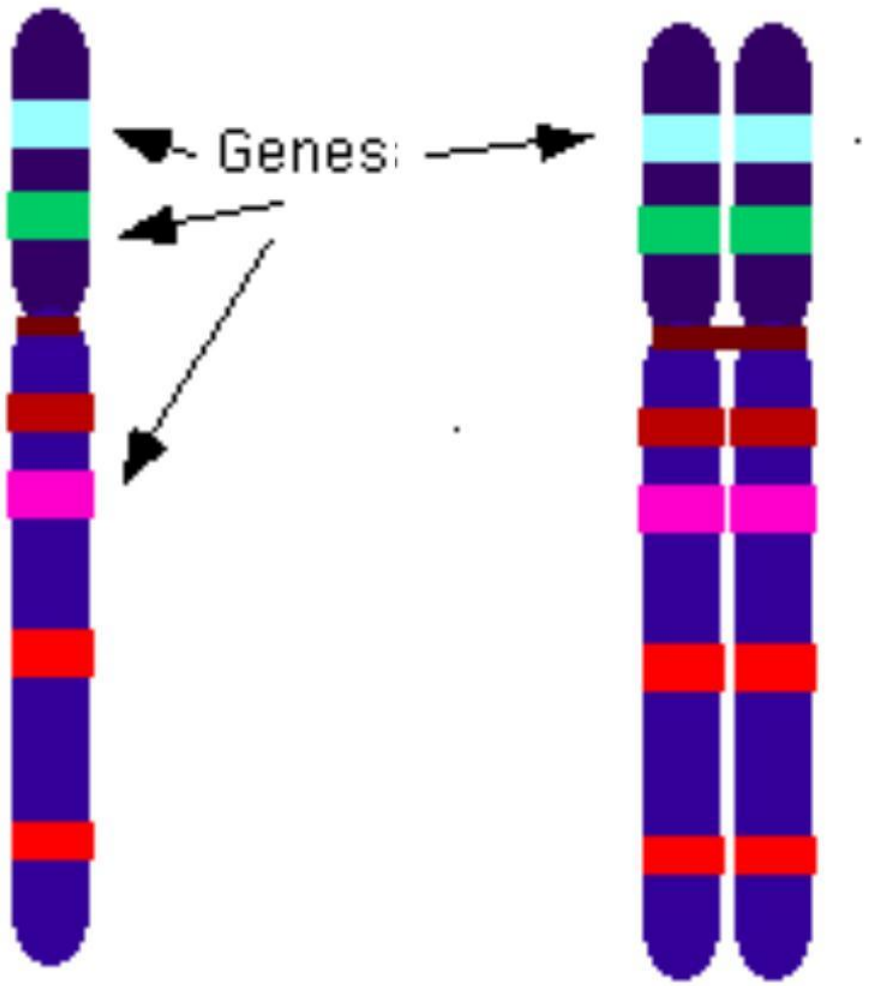
-Homologous pair → **a pair of chromosome. Both have:**

- Same shape
- Same length
- Same genetic content
- Same number of genes
- Same position of centromere.

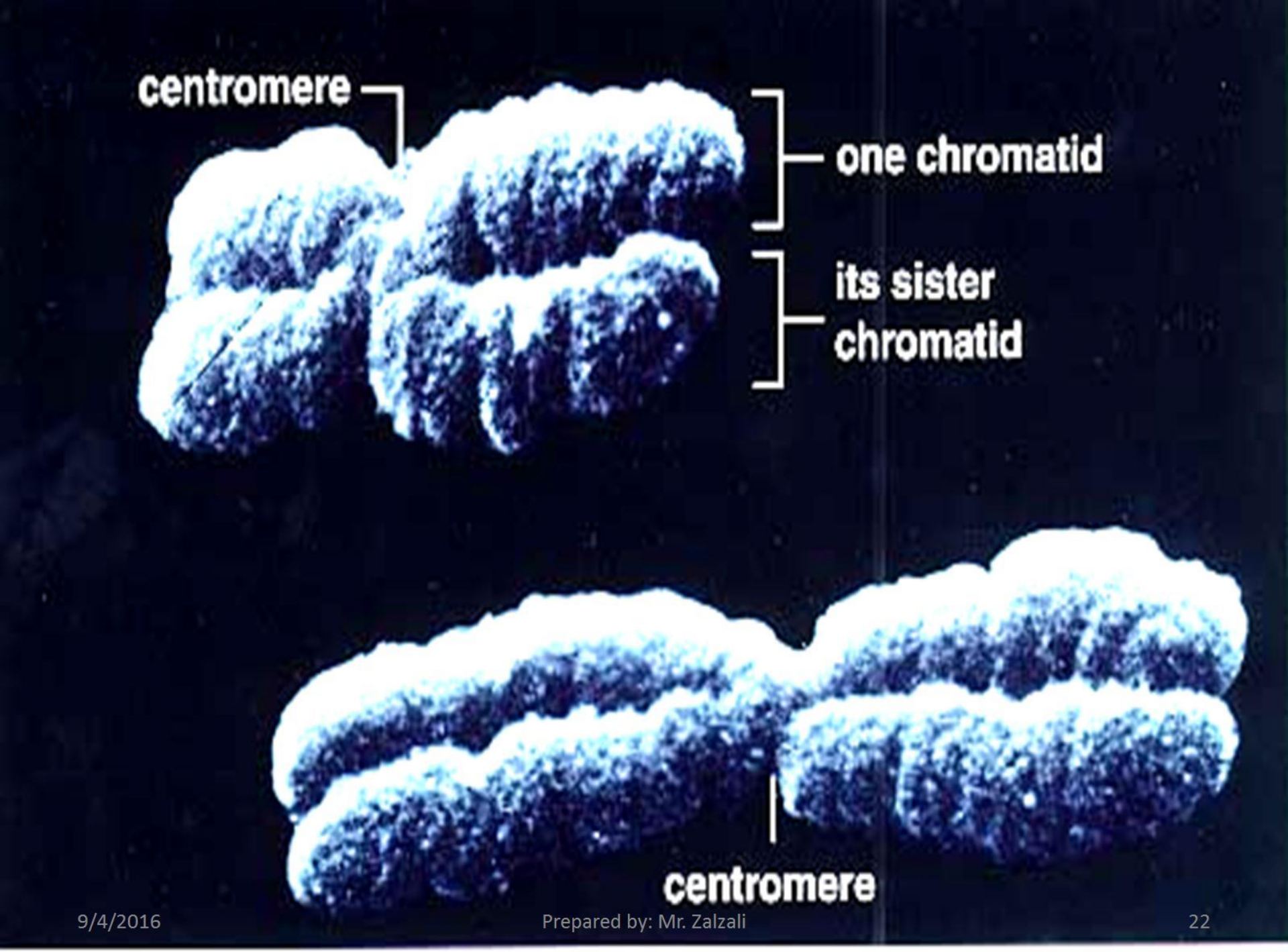


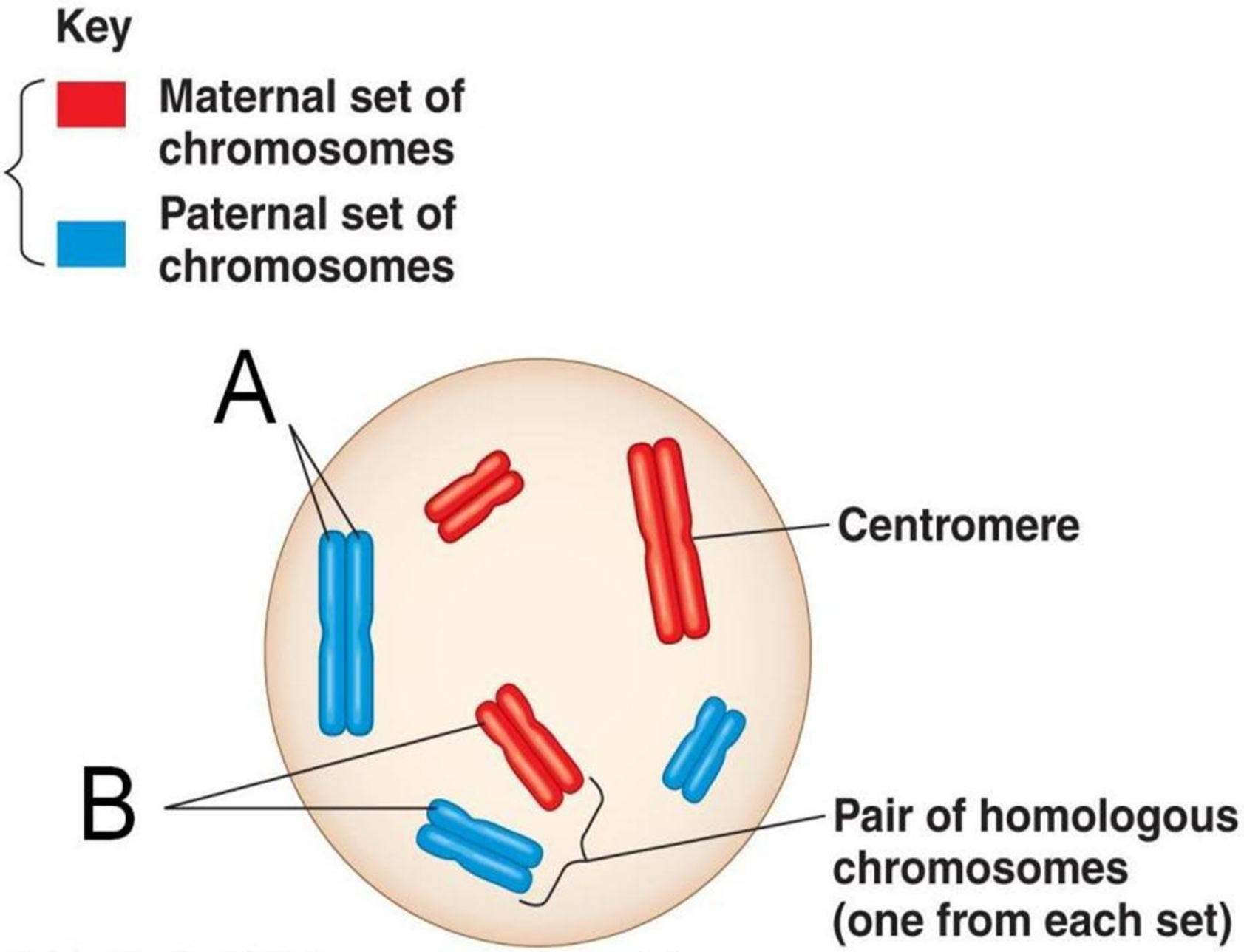
Homologous chromosomes





Unduplicated Duplicated


centromere
one chromatid
its sister
chromatid
centromere



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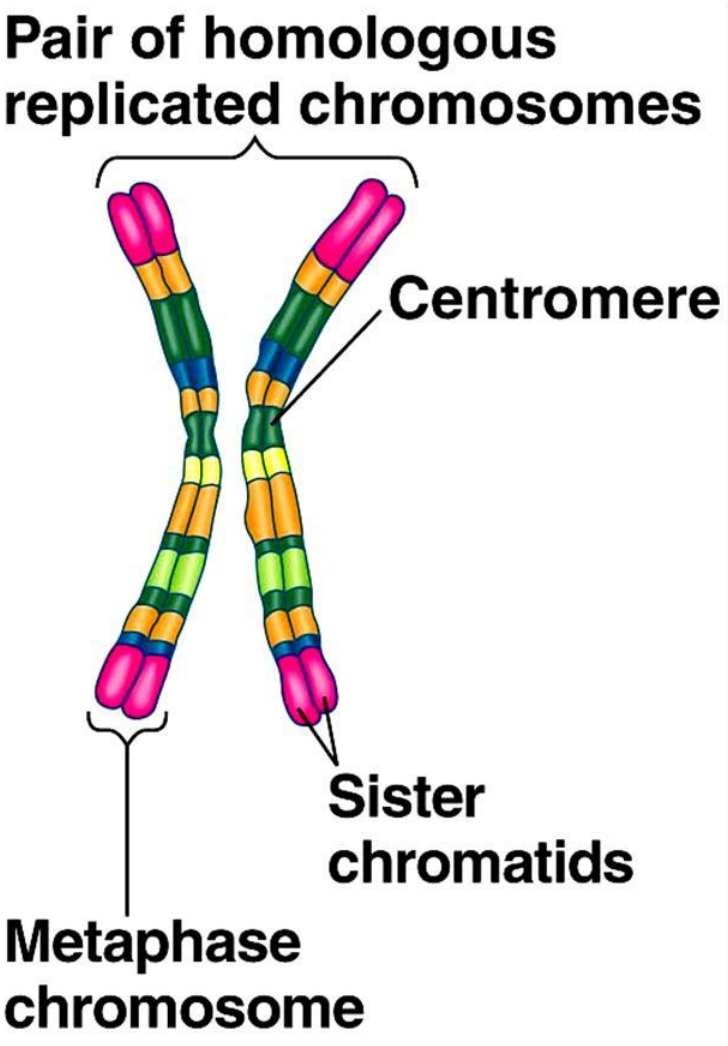
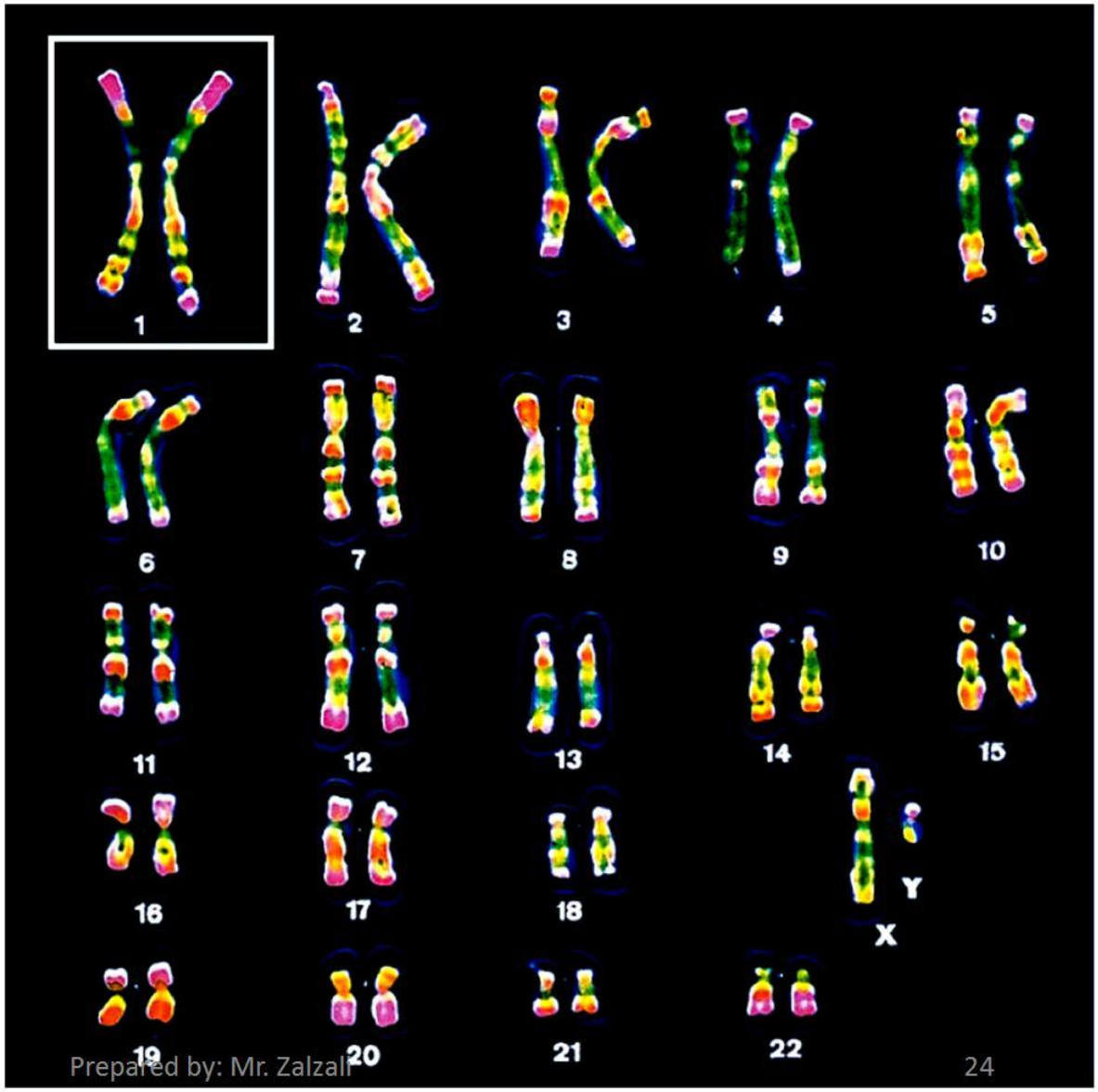
TECHNIQUE





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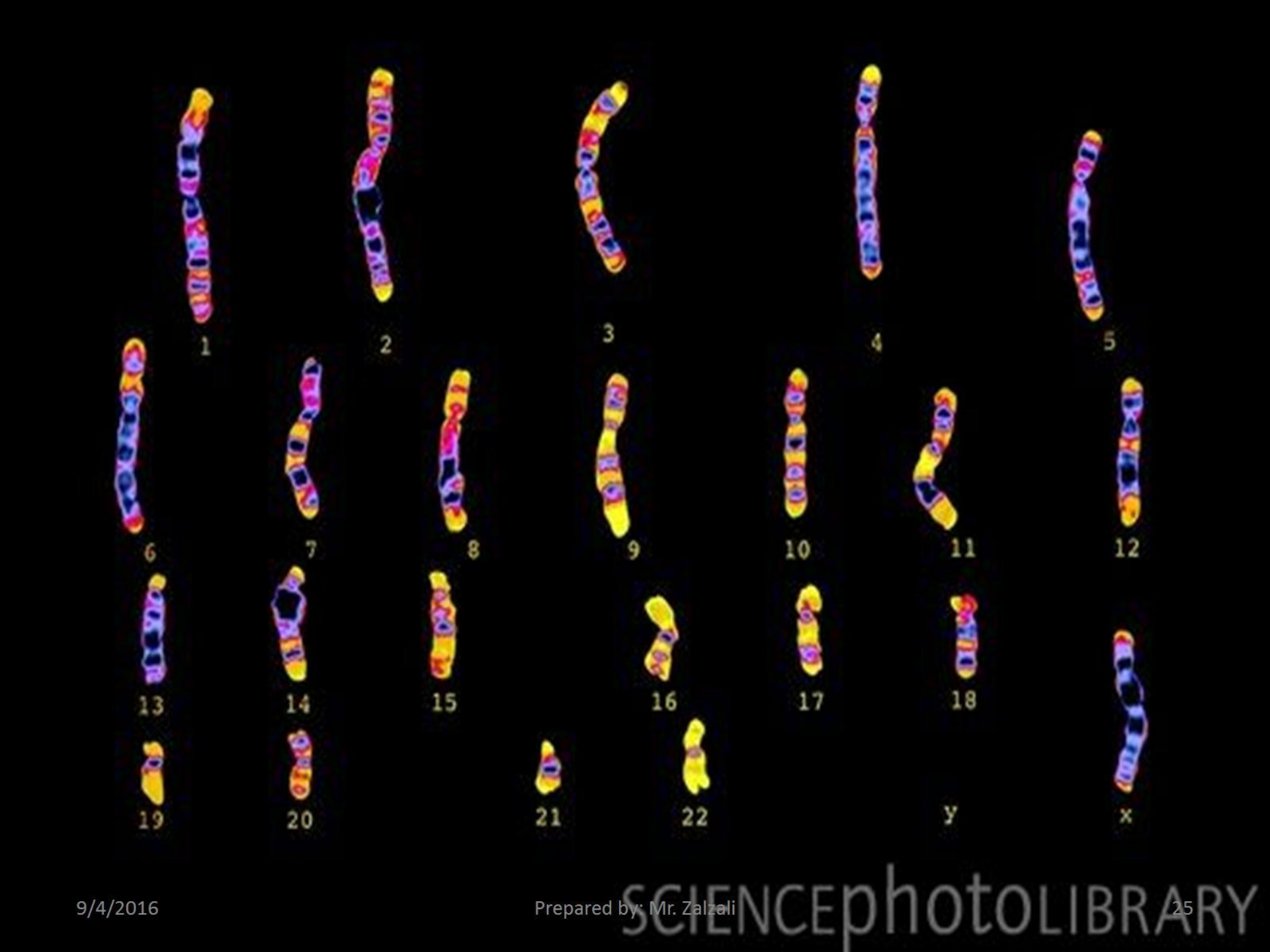
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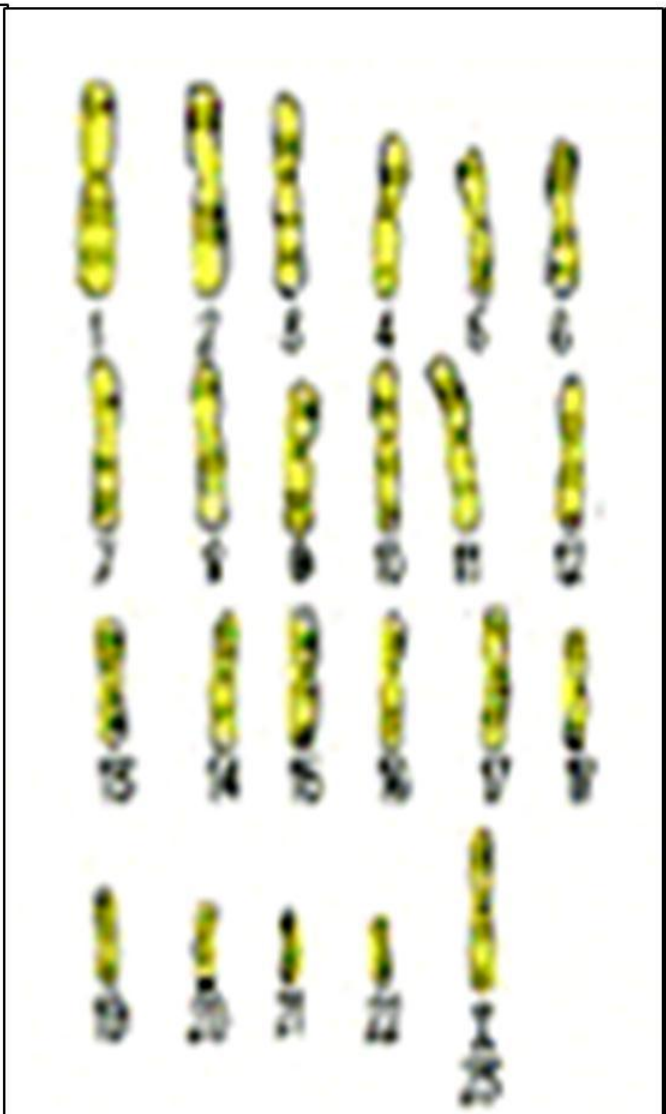
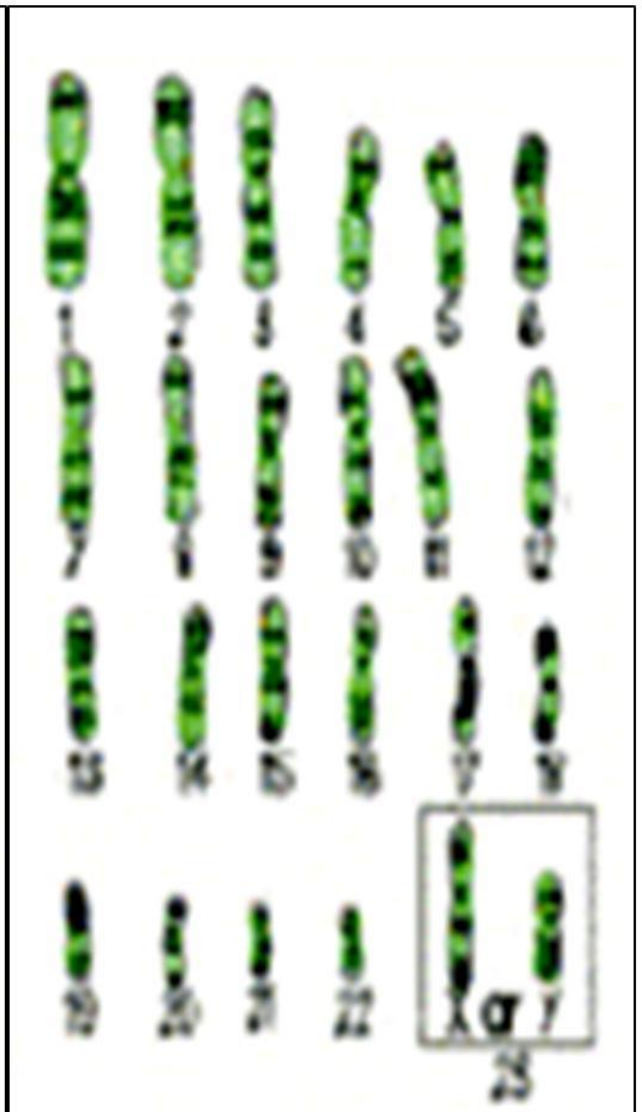
Normal Cell
46 Single Chromosomes
23 Chromosome Pairs



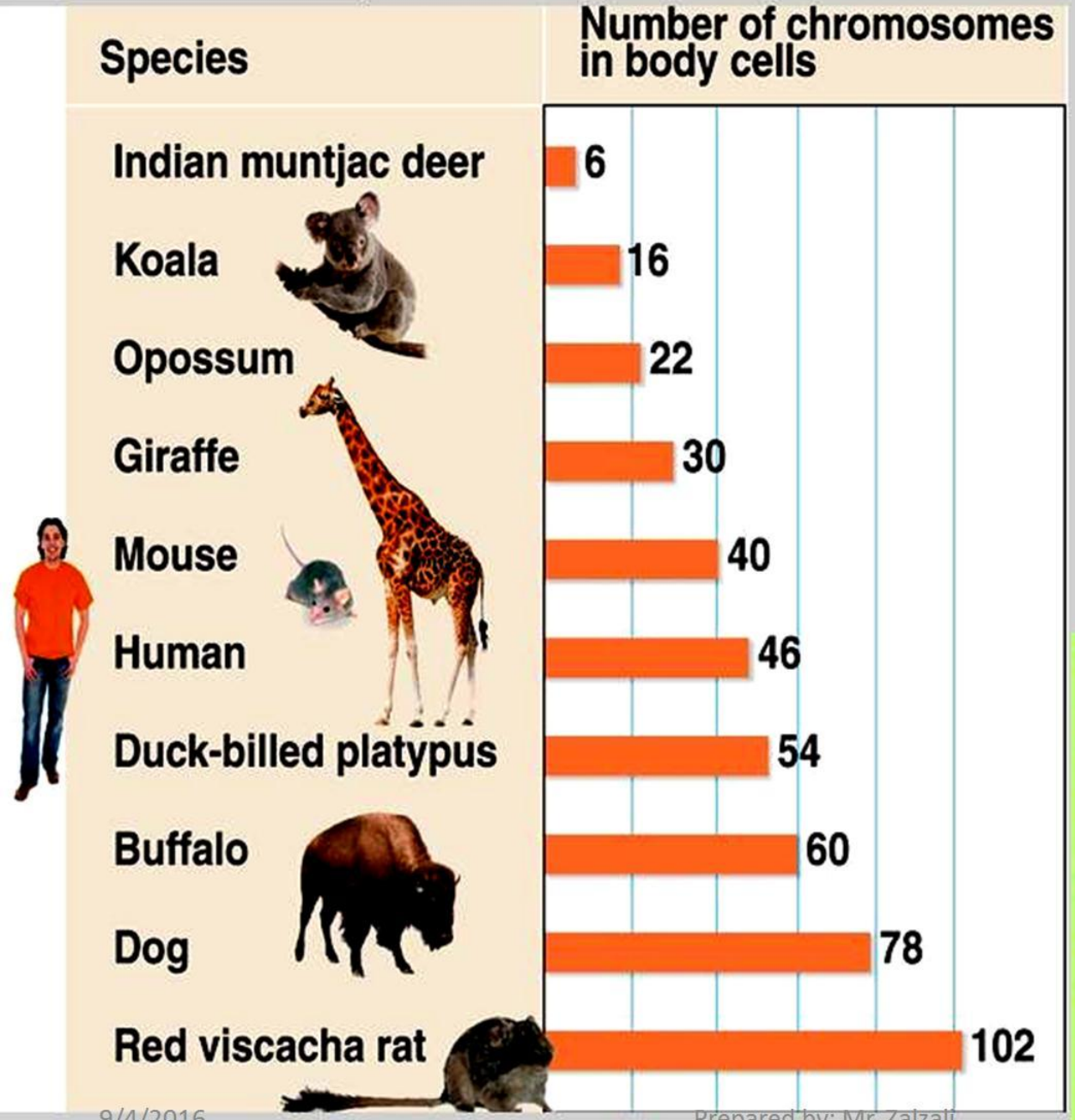
Egg
23 Single Chromosomes



Sperm
23 Single Chromosomes

Eukaryotic cells	
Somatic cells	**Gametes**
-Al body cells that contain chromosomes in pairs (homologous chromosomes)	-All sex-cells that contain one set of chromosomes (no homologous pair)
- Called: **" Diploid cells" (2n)**	- Called: **"Haploid cells " (1n)**
-There are **2 copies from each chromosome**. One from male gamete and other from female gamete.	**-One copy of each chromosome** in each gamete.
-Ex : skin cells, liver cells, stem cells.....	-Ex. : Sperm and egg cells→ in animals.

Figure 8.2



3e

- Fruit fly 8
- Earthworm 36
- Cat 38
- Elephant 56
- Goldfish 102
- Snail 24
- Horse 64
- Cow 60
- Sheep 54
- Pig 38

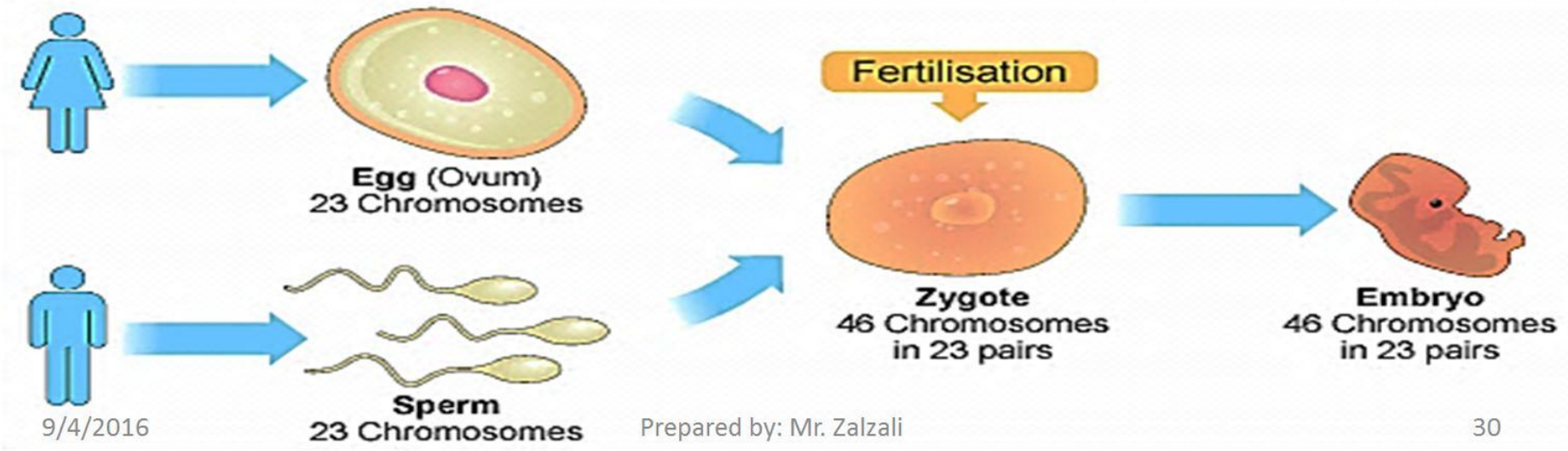
- Rye 14
- Corn 20
- Wheat 42
- Tobacco 48
- Fern 1262
- *Arabidopsis* 10
- Wild potato 24
- Cultivated potato 48

48 chromosomes

In Humans	
Somatic cells	**Gametes**
-All body cells have 46 chromosomes → 23 Homologous pairs.	-All sex cells have only 23 chromosomes. No pairs. No homologous chromosomes.
- Called: **" Diploid cells" (2n)**	- Called: **"Haploid cells " (1n)**
-Ex : skin cells, liver cells, stem cells.....	-Ex.: - Sperm - Egg cells

Fertilization:

-Fusion of male and female gametes to form diploid embryo.



-Importance of reducing NO. of chromosome into ½ in gametes →

-To restore the diploid number of chromosome in the embryo and to maintain a stable constant number of chromosomes passing from one generation to next generation in a certain species.

-Mutation:

- Any sudden change in chromosome that may lead to abnormalities and defects in the new offspring.

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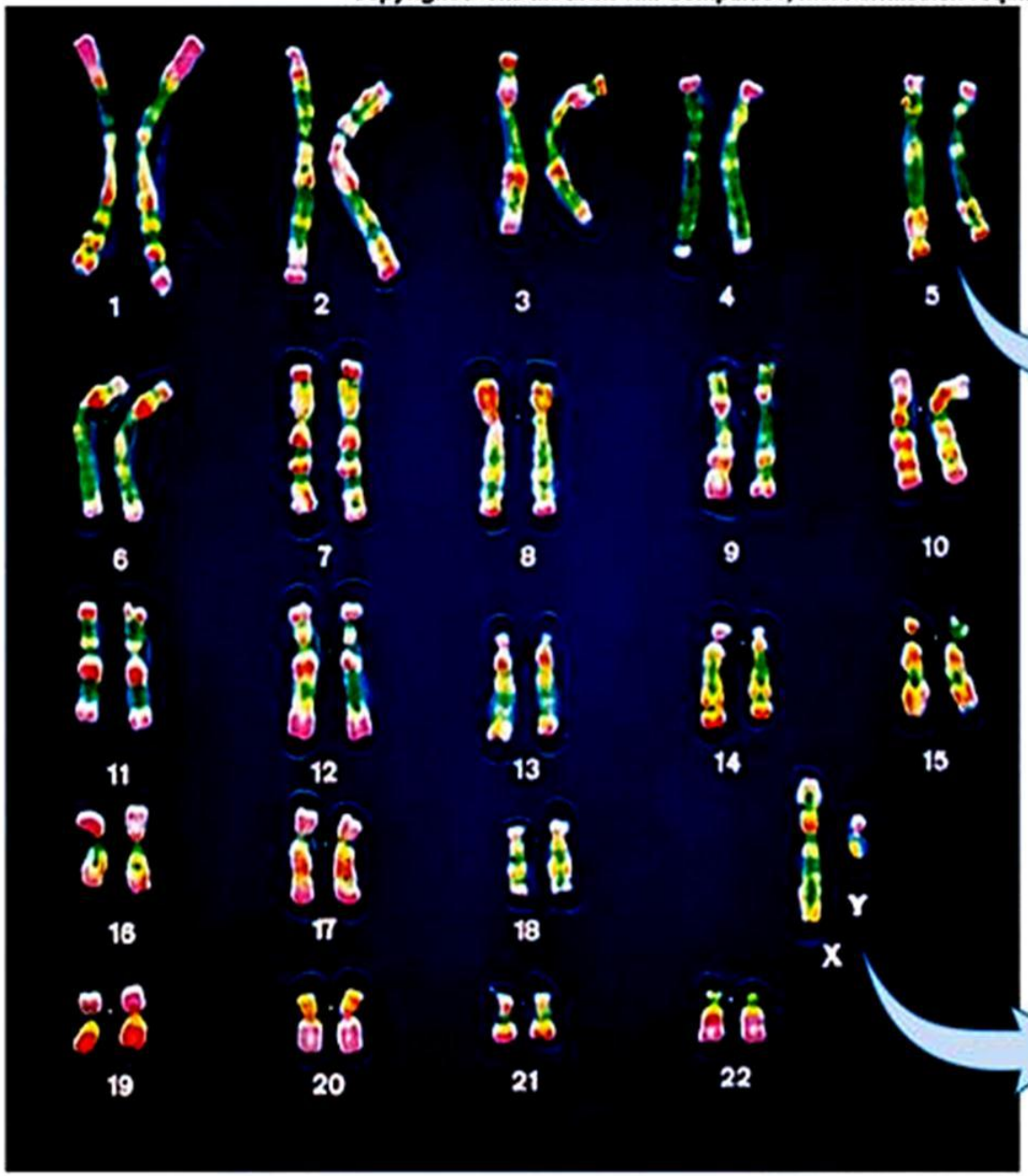

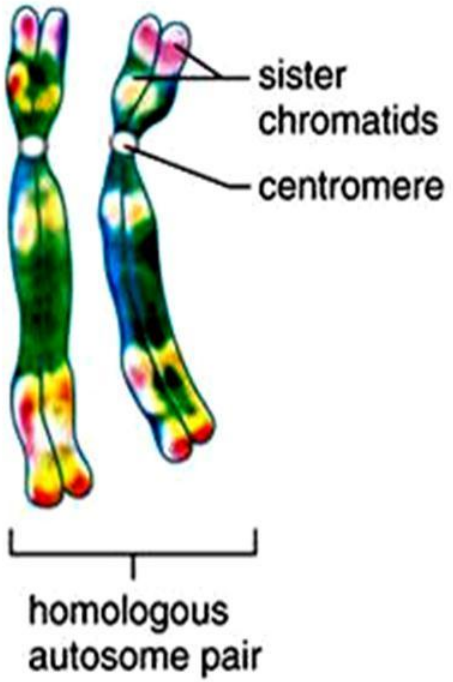

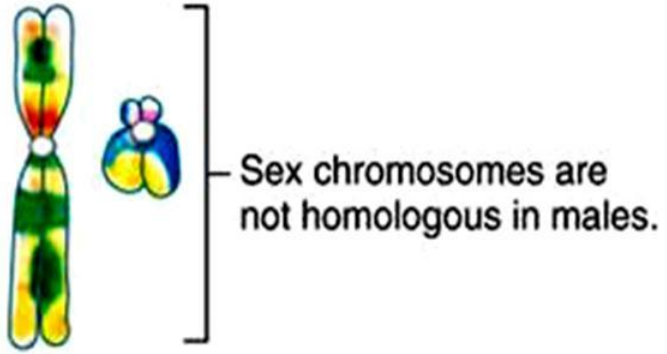

The 46 chromosomes of a male

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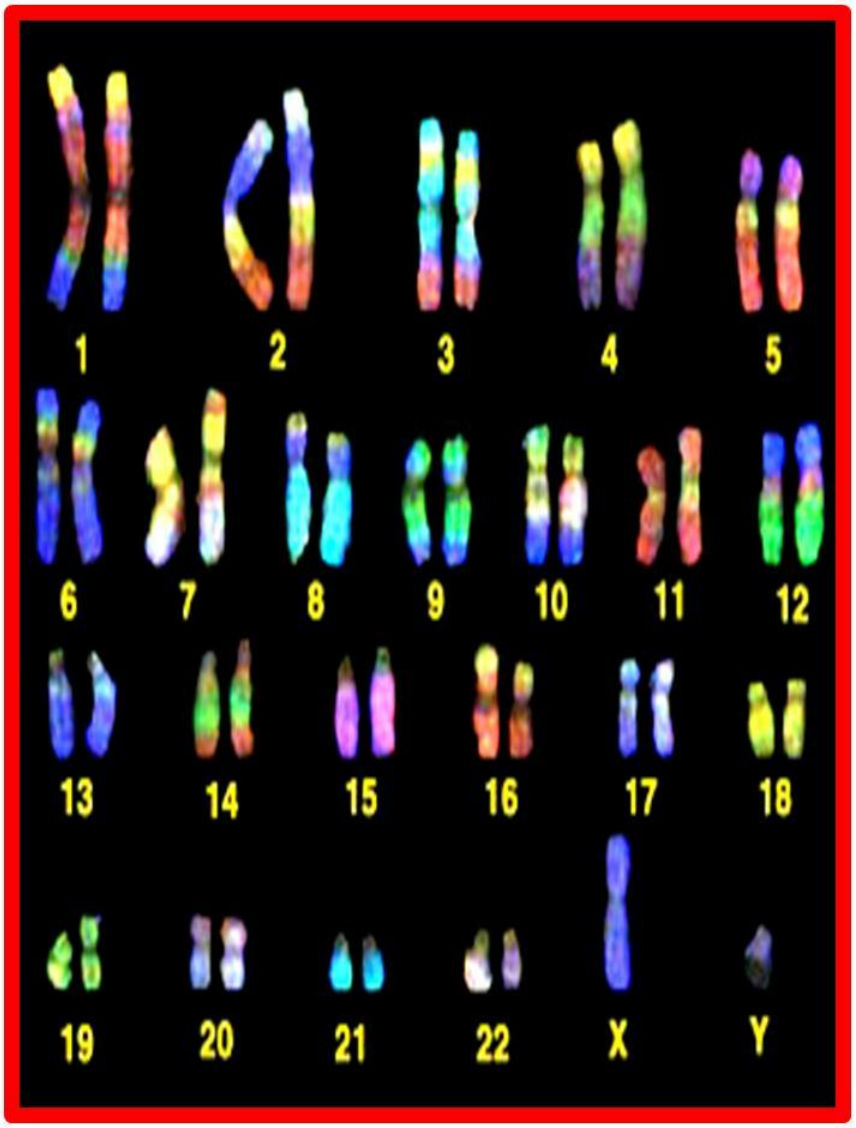
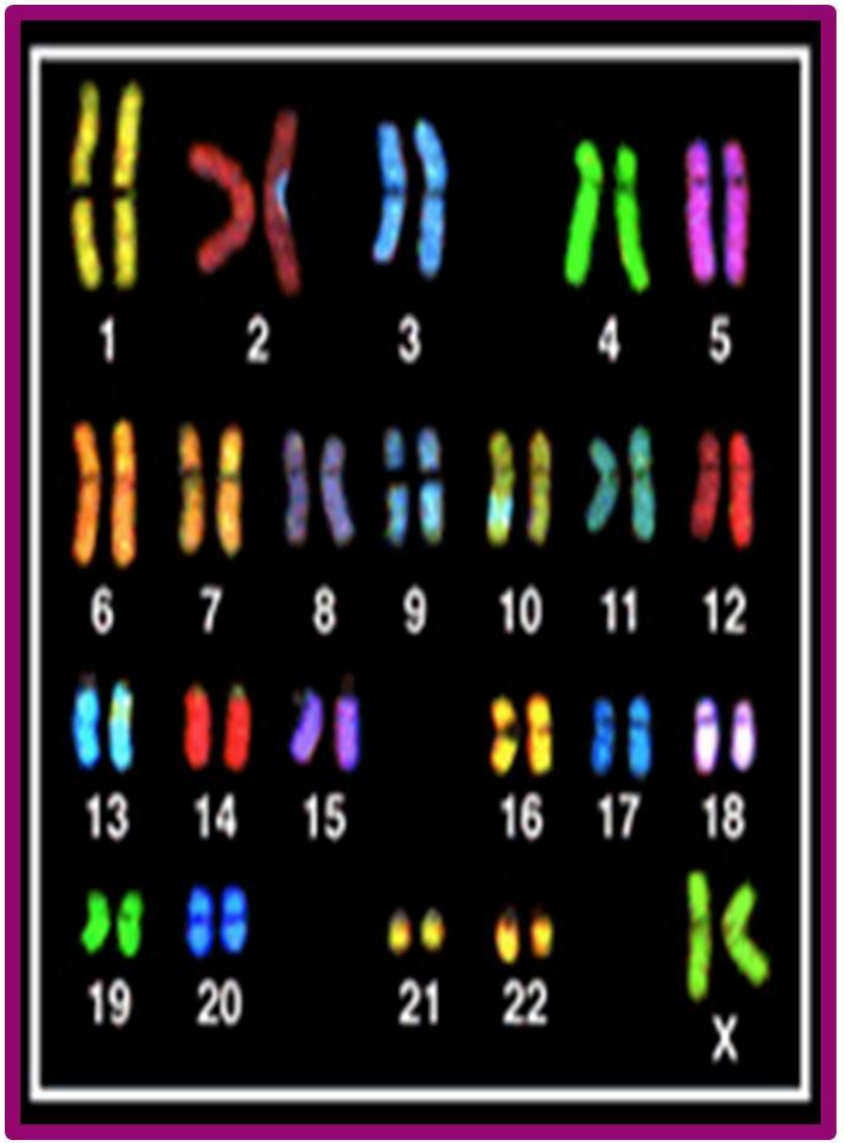
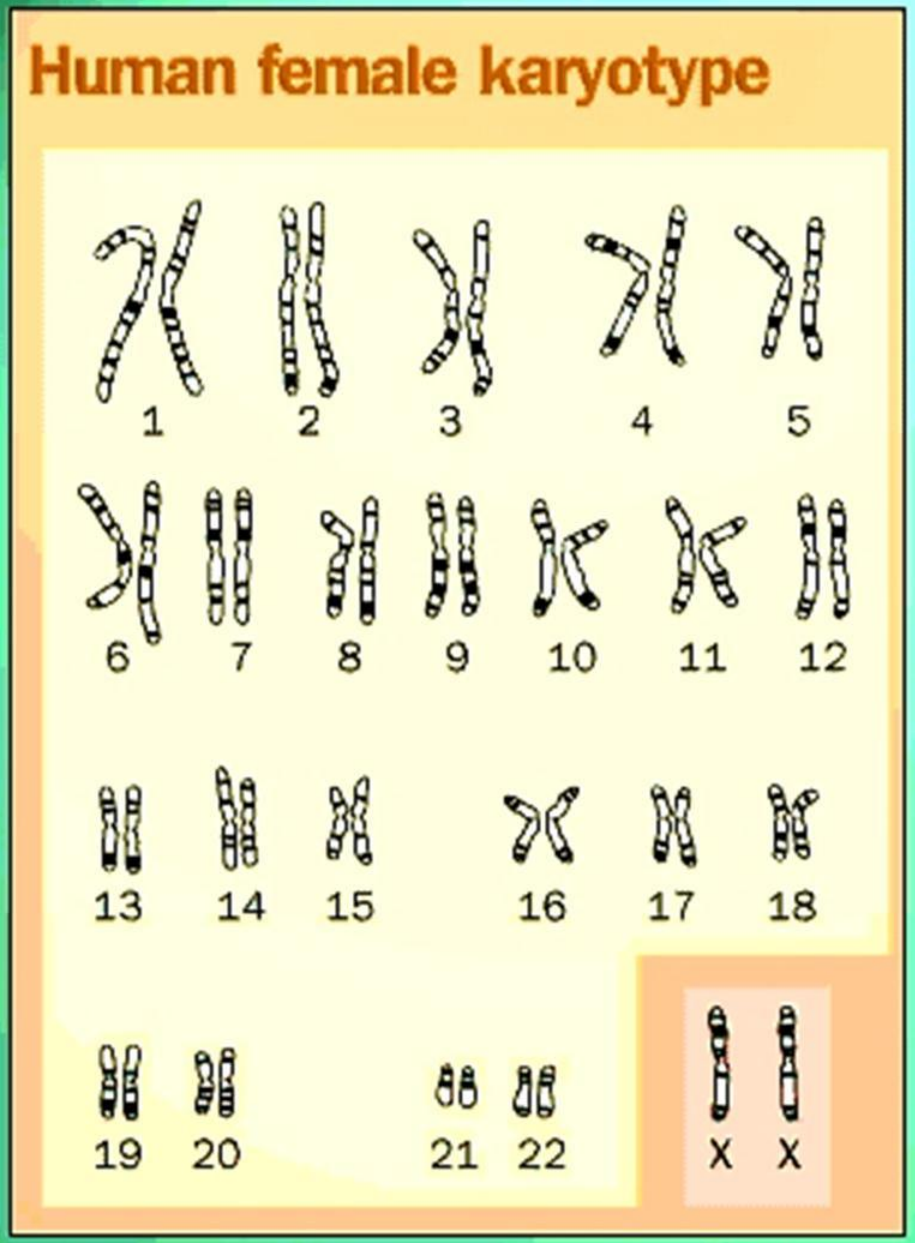
Male Karyotype



Female Karyotype




Human female karyotype
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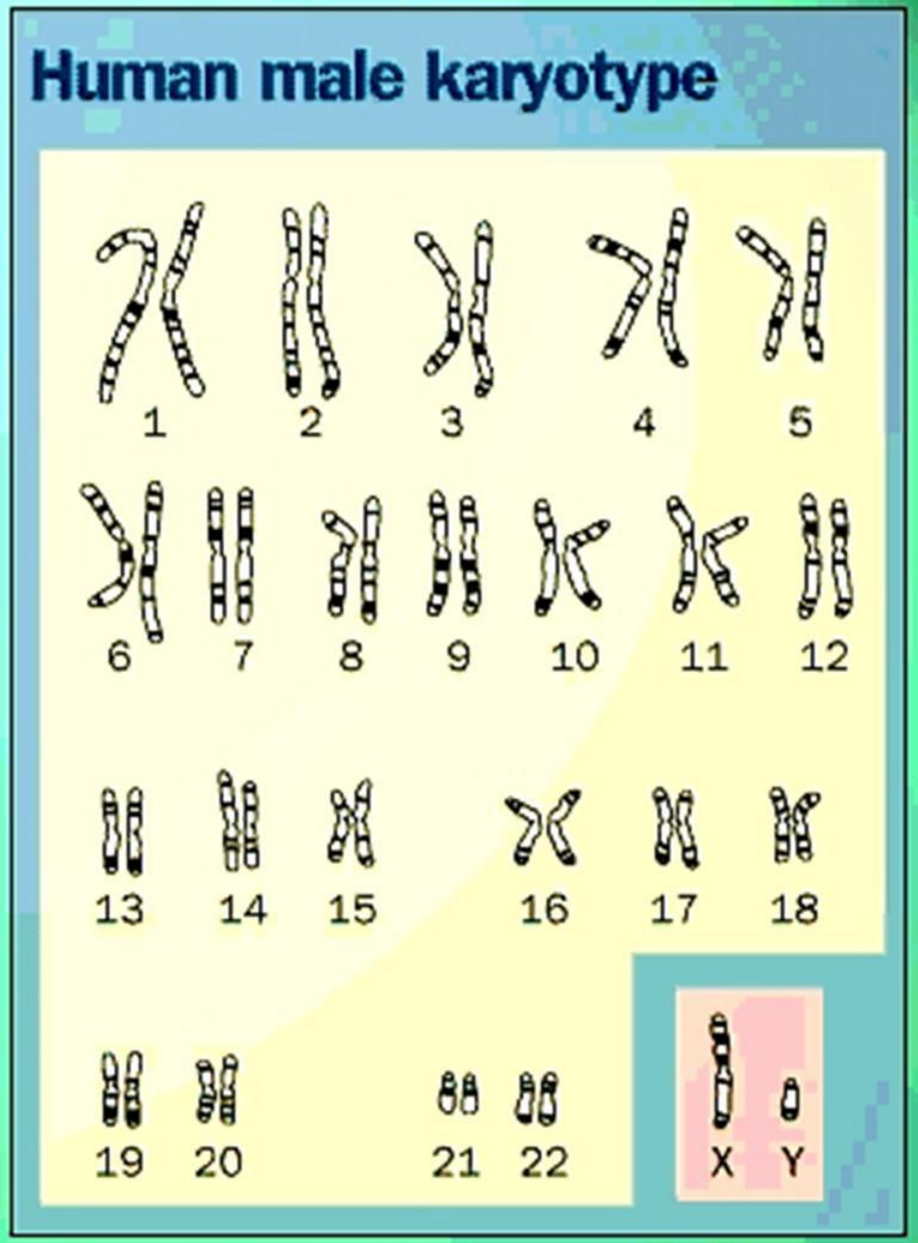
Autosomes
Sex chromosomes


Human male karyotype
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Autosomes
Sex chromosomes


X-chromosome
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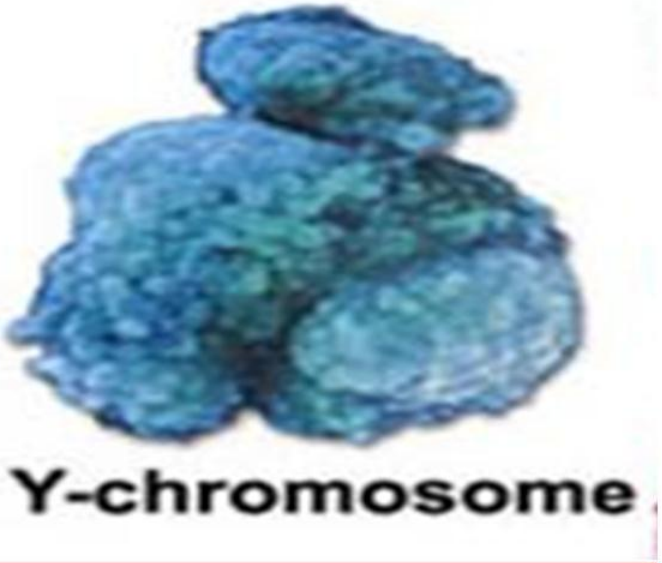
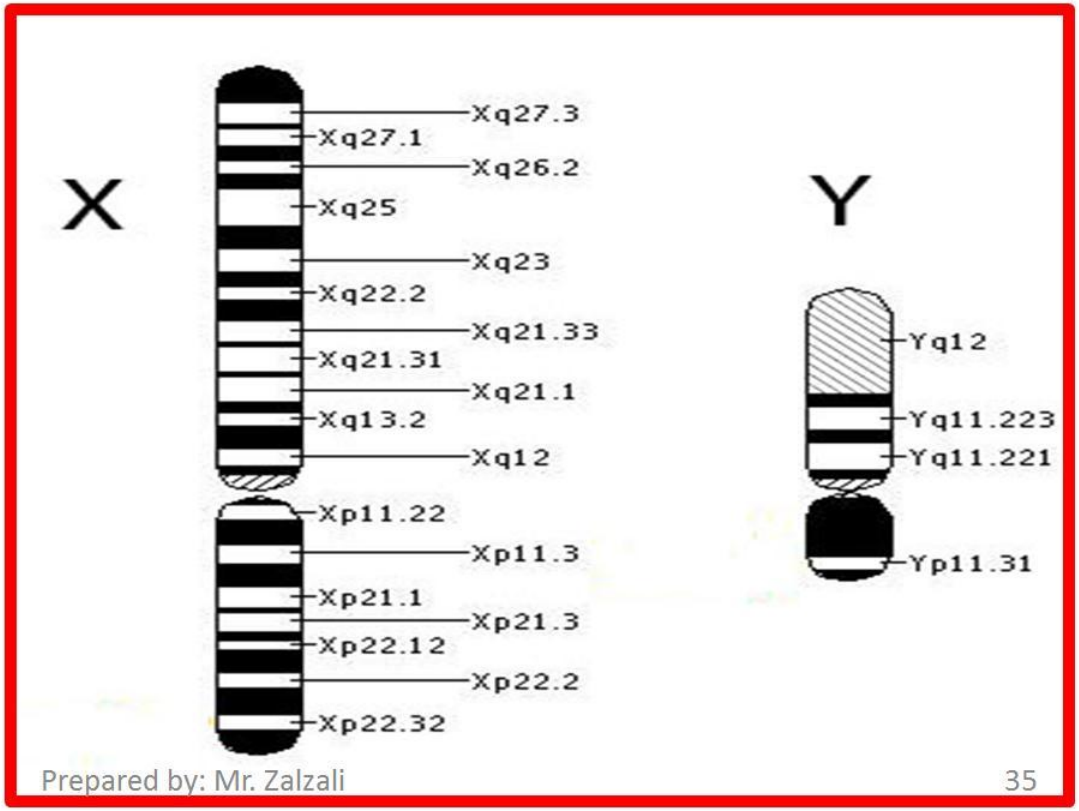

Y-chromosome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Xq12
Xp11.22
Xp11.3
Xp21.1
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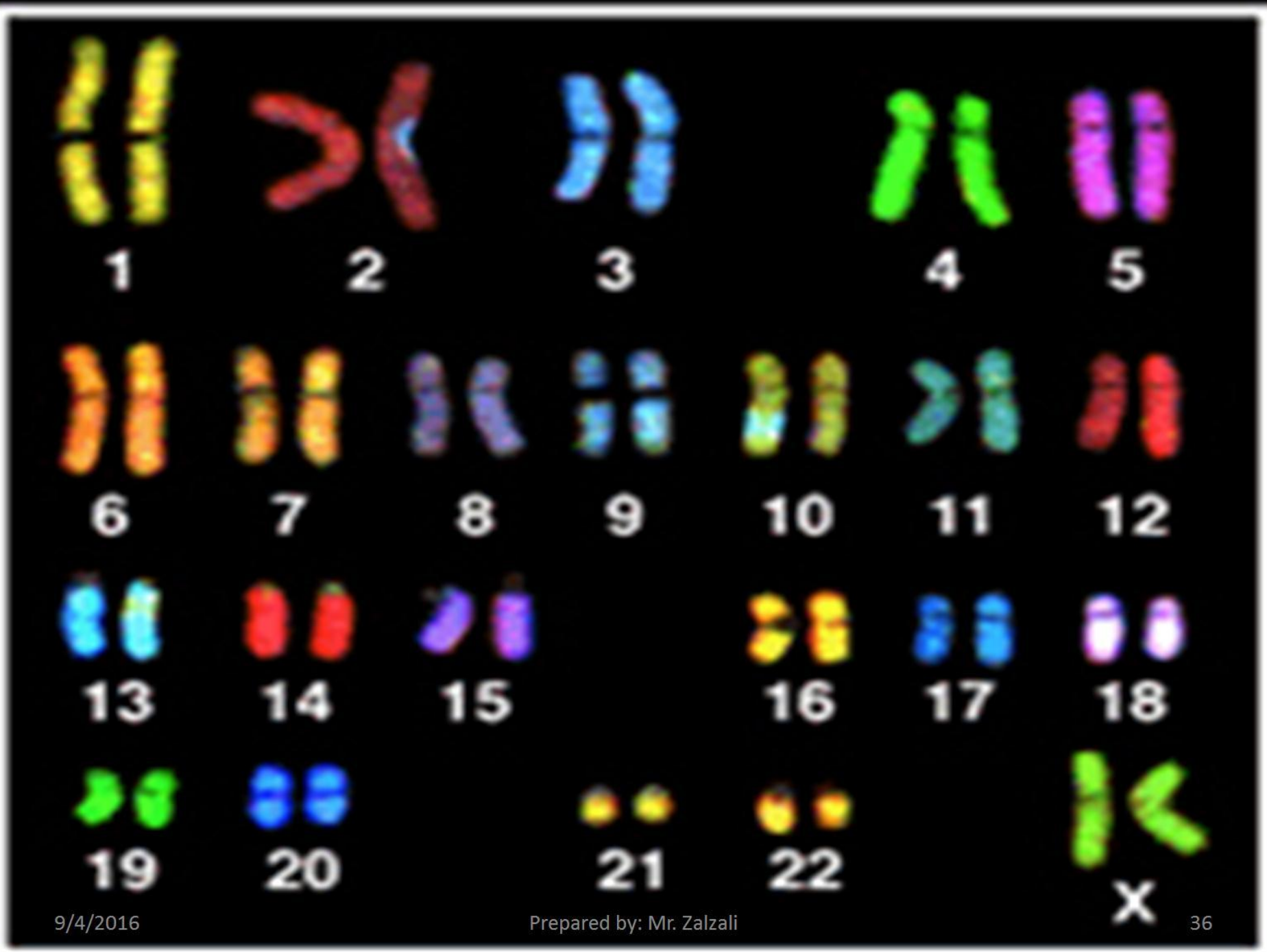
Yq11.221
Yp11.31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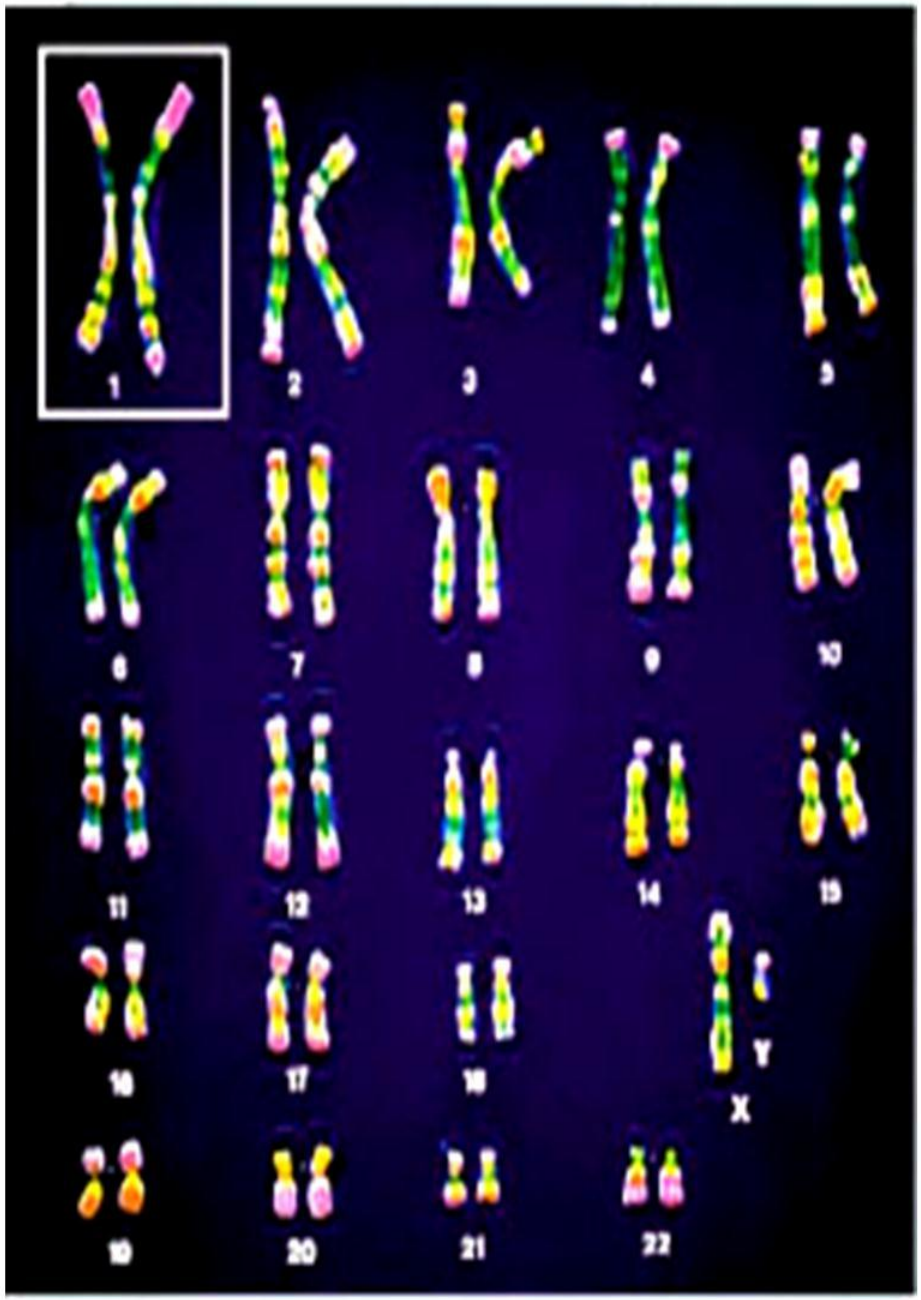
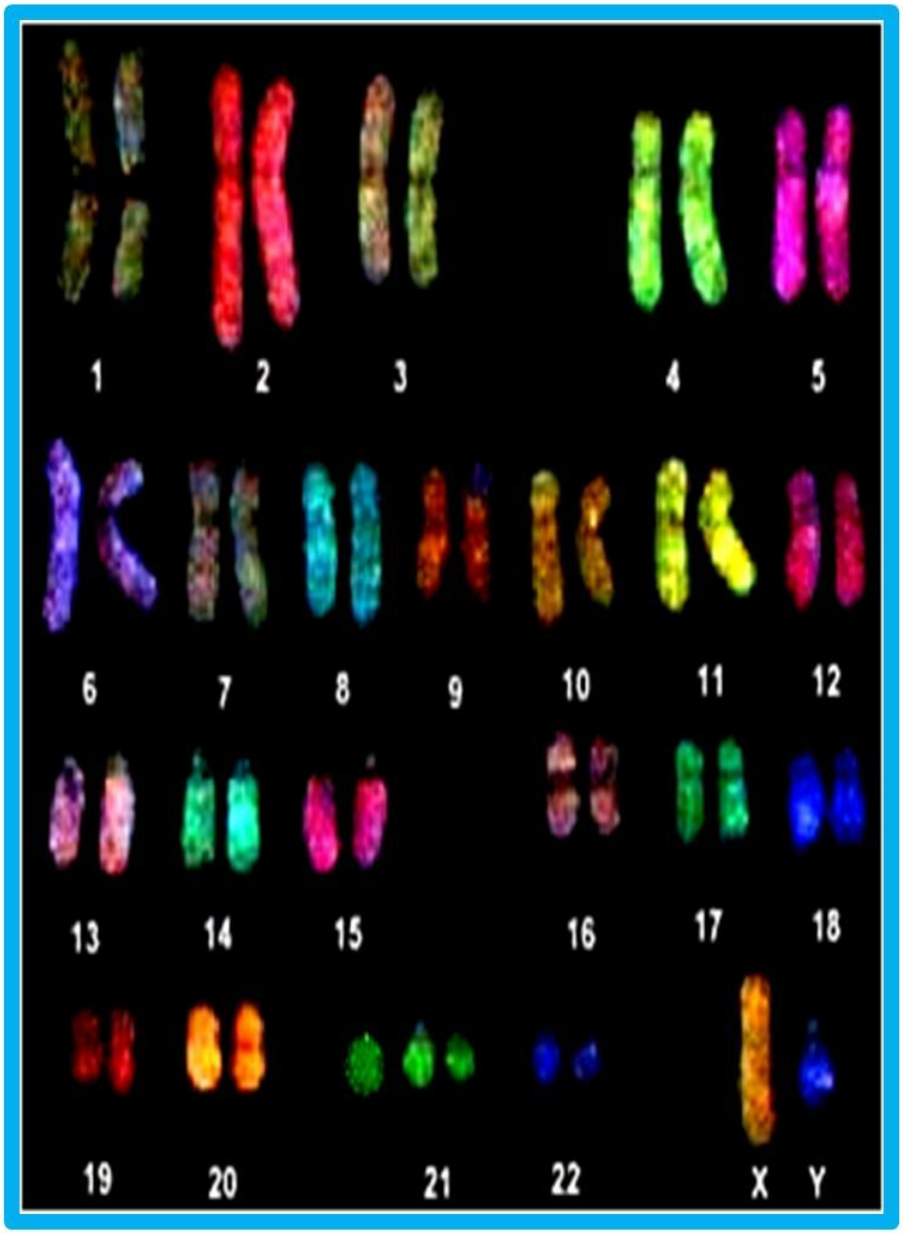
In sexually reproducing organisms, Chromosomes:

Autosomes	Sex- chromosomes
-All chromosomes not involved in determining the sex of the individual -They are 22 pairs = 44 chromosomes in humans.	-Chromosomes that determine sex of individuals -In humans pair 23 is the sex chromosome - Females → XX– two identical chromosomes - Males→ XY → Non-identical chromosomes.

Karyotype



- **Karyotype**
  - A photo that shows all chromosomes in a human cell arranged by size.
  - Useful technique because it detects the abnormalities or **defects in the chromosome number** in any cell.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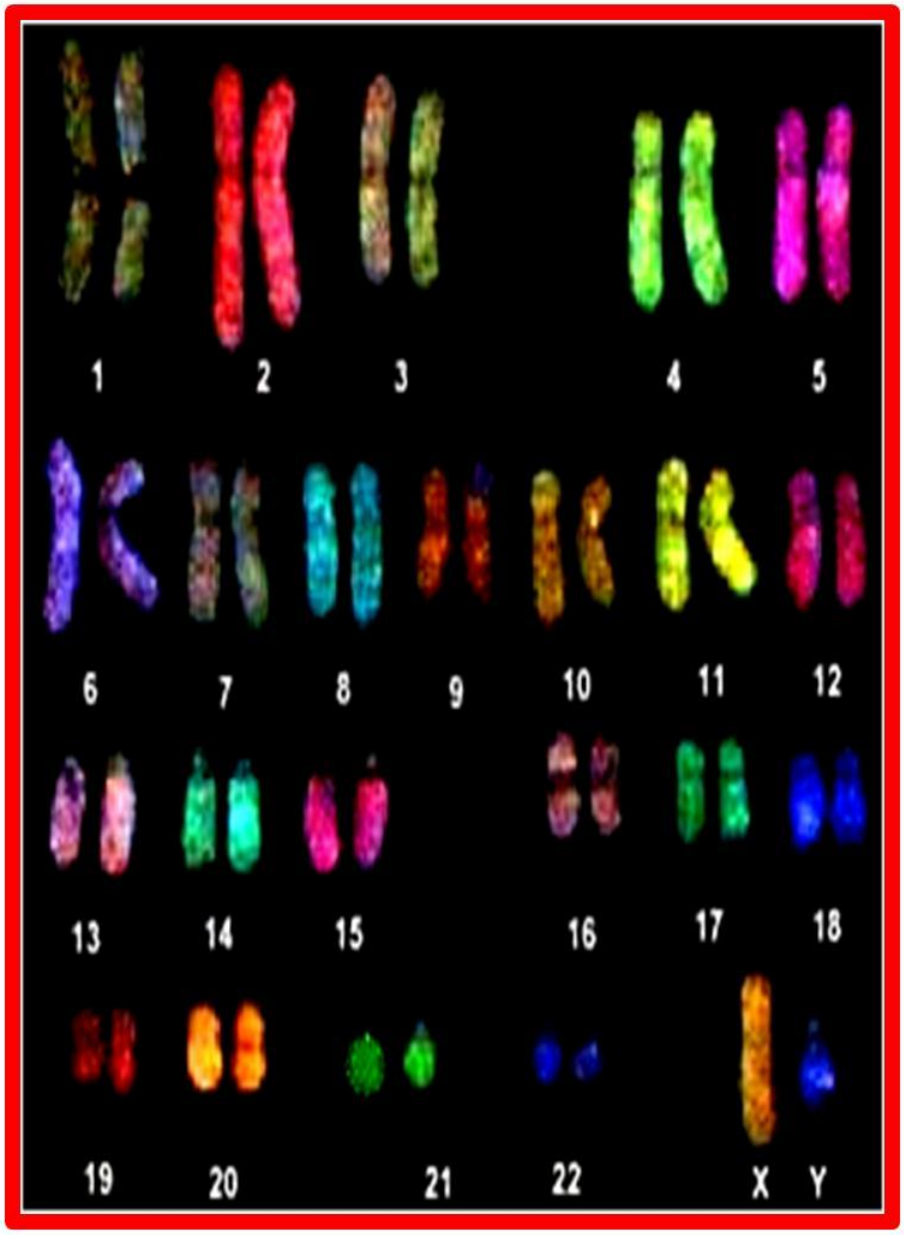
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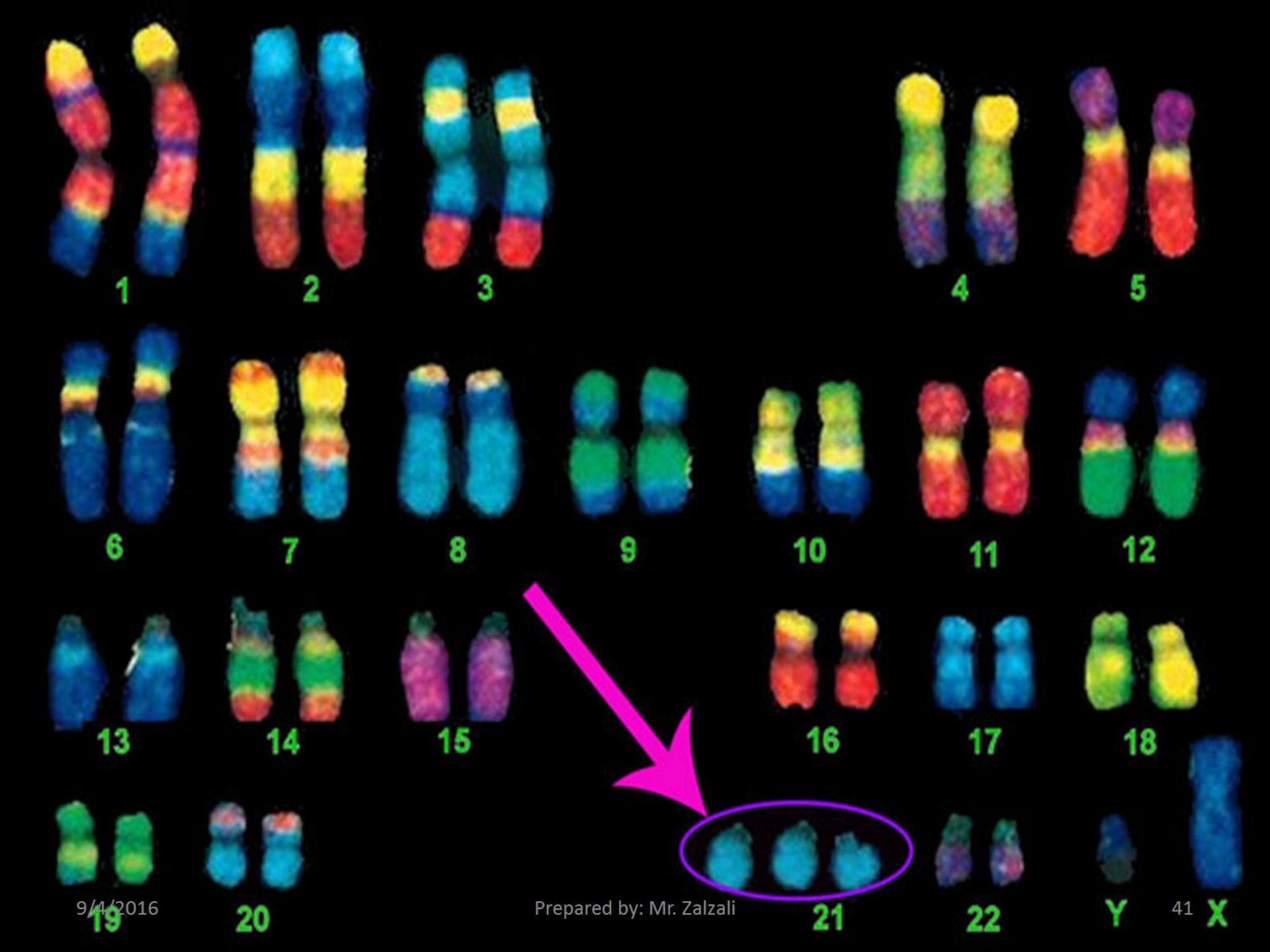
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Down's Syndrome

we should all see the world from their point of view

-Syndrome:

-Disorder /problem that's permanent and has no cure.

-Trisomy:

- Any genetic condition when human cells have more than 2 copies of a chromosome and this person develops abnormally.

-Down syndrome / (Trisomy 21):

- A genetic condition in which chromosome **21 has 3 copies** instead of two. It means every cell in the body contains 47 chromosomes instead of 46.

-Symptoms of down syndrome:

-Round face with thick folded eyelids
-Varied degrees of mental retardation
-Susceptible to infections and heart diseases
- Weak muscles. Short stature.

-Chances of having D.S. with:

- Mothers younger than 30 → 1/1500 births
- 37 years old mother→ 1/290 births
- Mothers Over 45 → 1/46 births

-Why is the ratio of down syndrome increasing in older mothers (above 40 years old) ?

- In older mother, in ovaries during cell division, to form haploid egg cells, there is accumulate increase in the DNA damage leading to produce egg cells with extra of lesser chromosomes → this increase rate of mutations to take place.

-Because of this risk→ a pregnant woman over the age of 35 may be advised to undergo **prenatal test** and fetal karyotyping.

-Chromosomal mutations:

- Mutations could be:

-Chromosome Number.

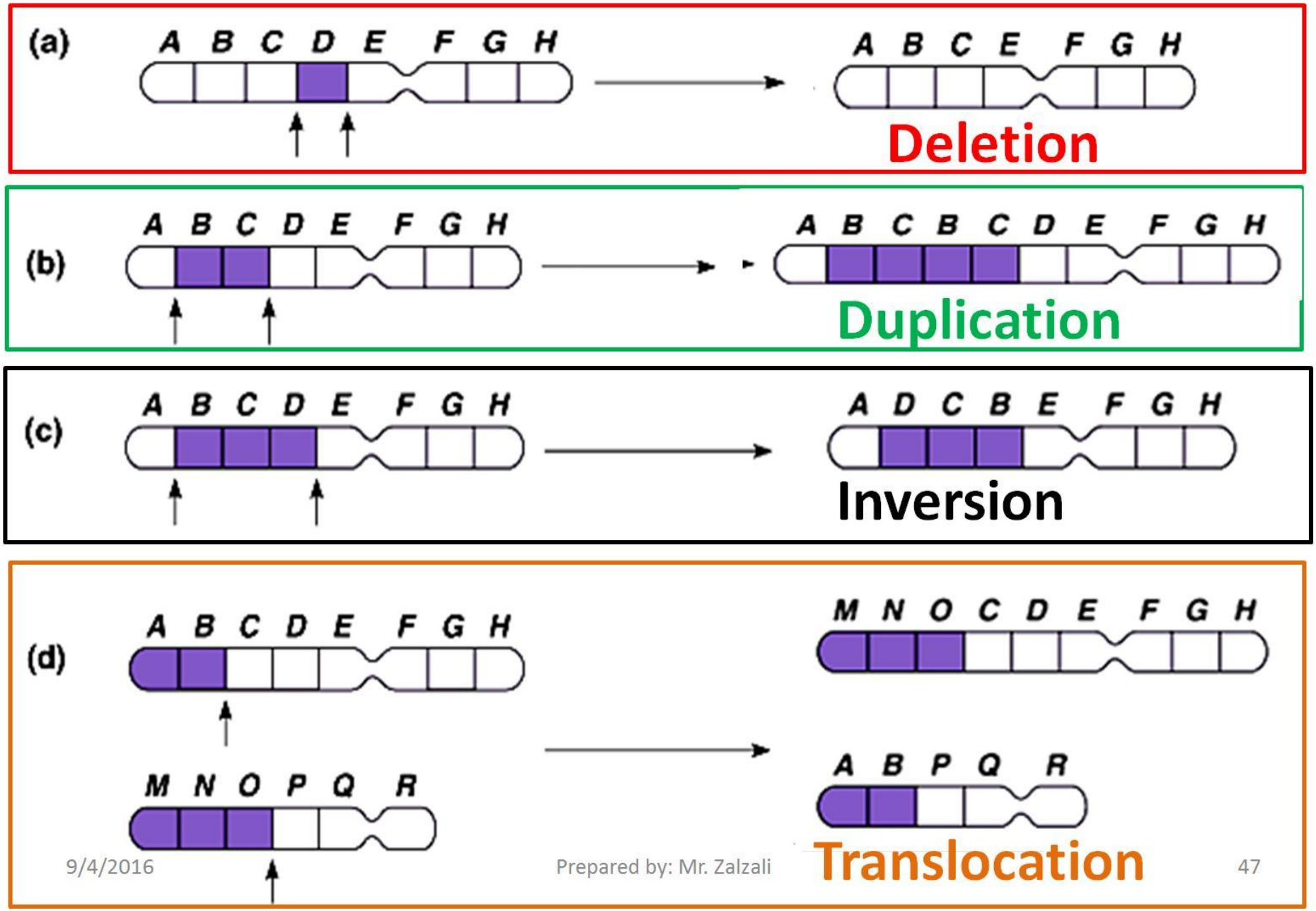
- Chromosome structure.

- Gene mutation → damaged DNA within the chromosome.

-Karyotype:

- An enlarged photo or picture for all chromosomes in a cell arranged as homologous pairs and according to size and shape.
- Can detect chromosomal mutations, but cannot detect gene mutation.
- Useful to detect some genetic disorders or defects in the fetus.

Types of chromosomal mutations:



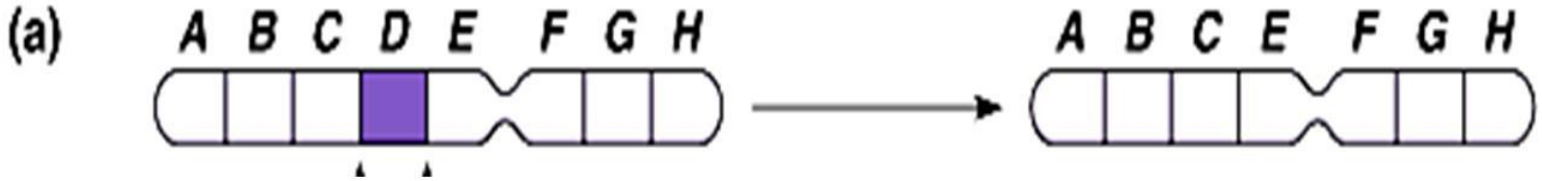
Mutation:

- Sudden change in organism's chromosome(s) structure.

-Any event that changes genetic structure.

-Types of mutations:

1- Deletion: a piece/segment of chromosome breaks off and Is lost completely . So the cell lacks some genes.



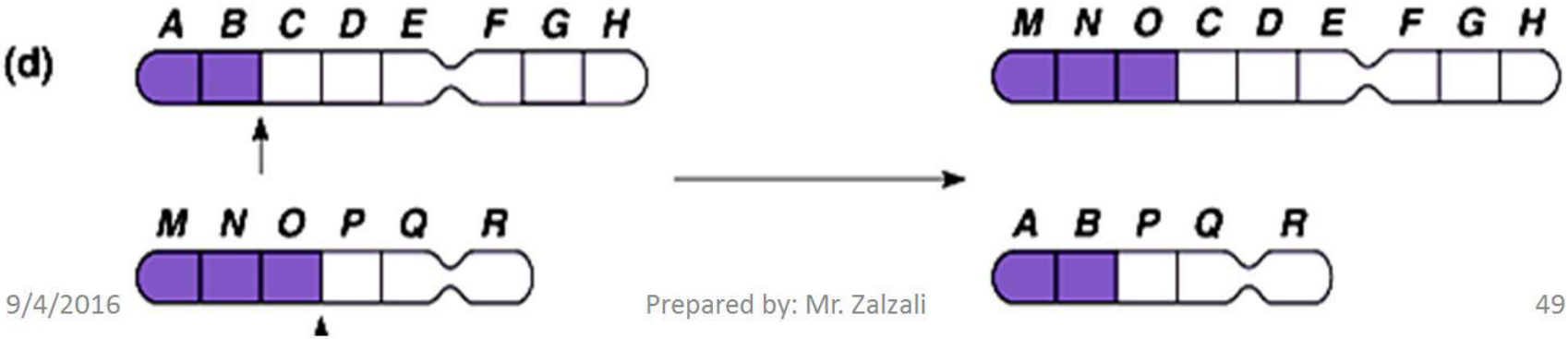
2- Inversion : A piece/ segment of chromosome breaks off and reattaches to original chromosome but in reverse position.

3- Duplication:

- A piece or segment of chromosome attaches to its homologous. So the homologous will carry 2 copies of certain genes.

4- Translocation:

- A piece of chromosomes is cut off and reattaches itself to a non-homologous chromosome.



Prenatal test

Prenatal tests :

-Medical techniques to detect abnormalities in fetus (during pregnancy).

1- Karyotyping :

- A technique of taking enlarged photo of all chromosomes, arranged by size and shape into pairs.

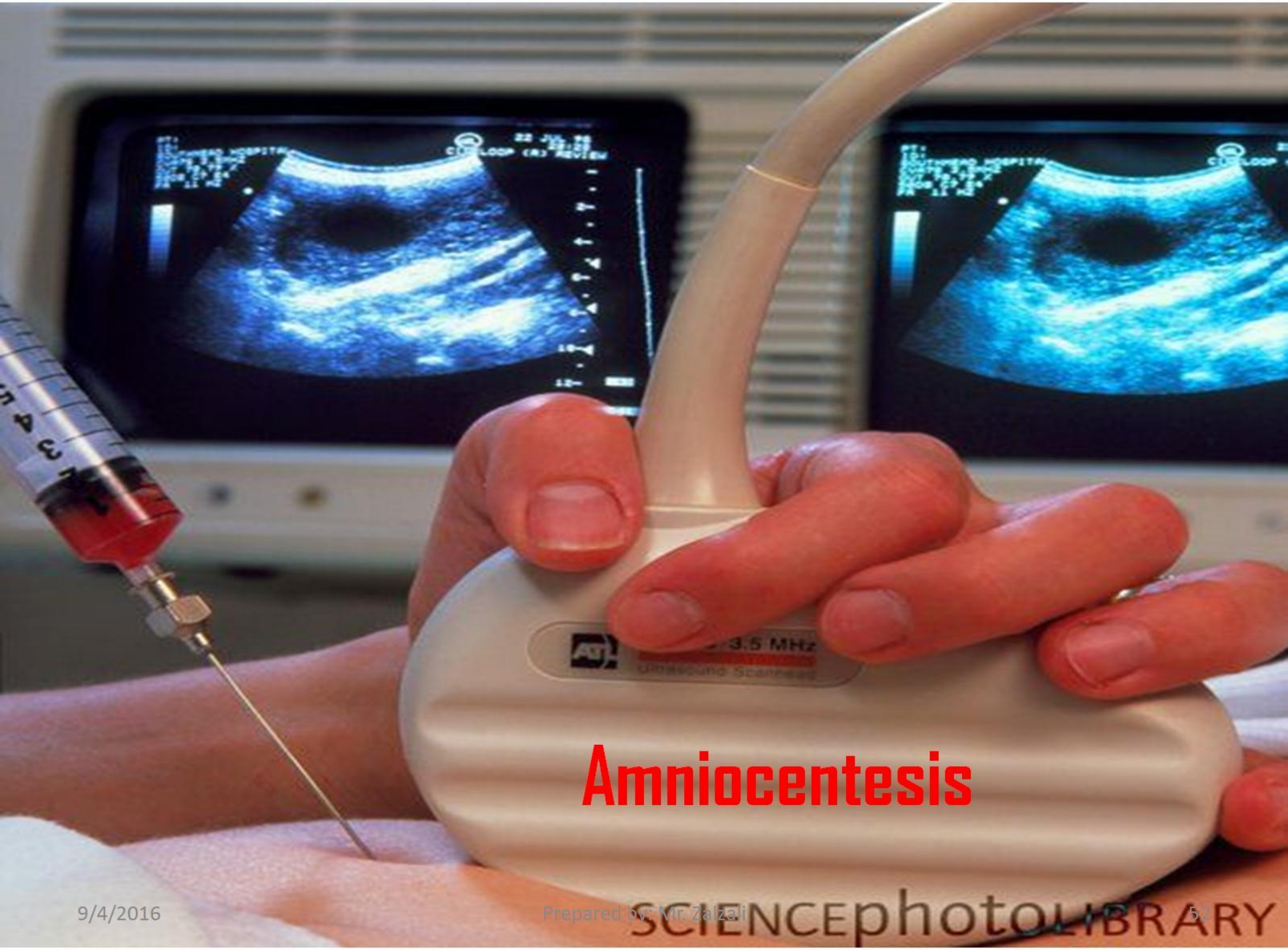
* Its advantages:

-Useful to inform doctors about chromosomal abnormalities in the fetus (e.g. down's syndrome)

- Useful to know the sex of the fetus.

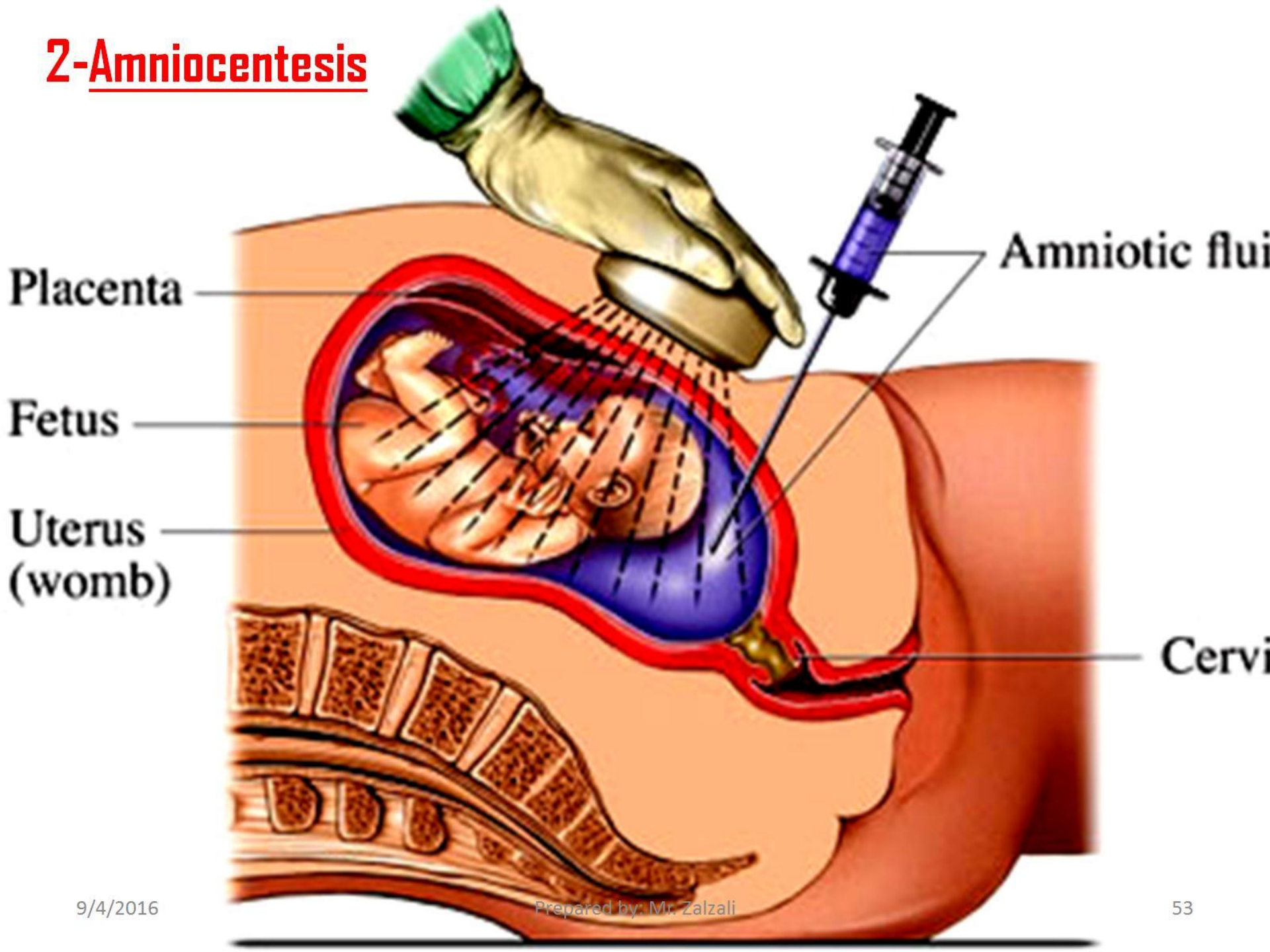
* Disadvantages:

-Could be inaccurate because it detects abnormalities in the number, size and shape of chromosomes but cannot detect gene mutations.



Amniocentesis

2-Amniocentesis



2- Amniocentesis :

-A procedure when a long needle is used to take a sample from the amniotic fluid around the fetus, then the amniotic fluid is cultured in lab to find fetal cells (about 2-4 weeks) → then fetal cells will be karyotyped to detect any abnormalities in fetal cells.

-It detects chromosomal mutations and abnormalities in fetal cells.

-Sample is taken thru abdomen.

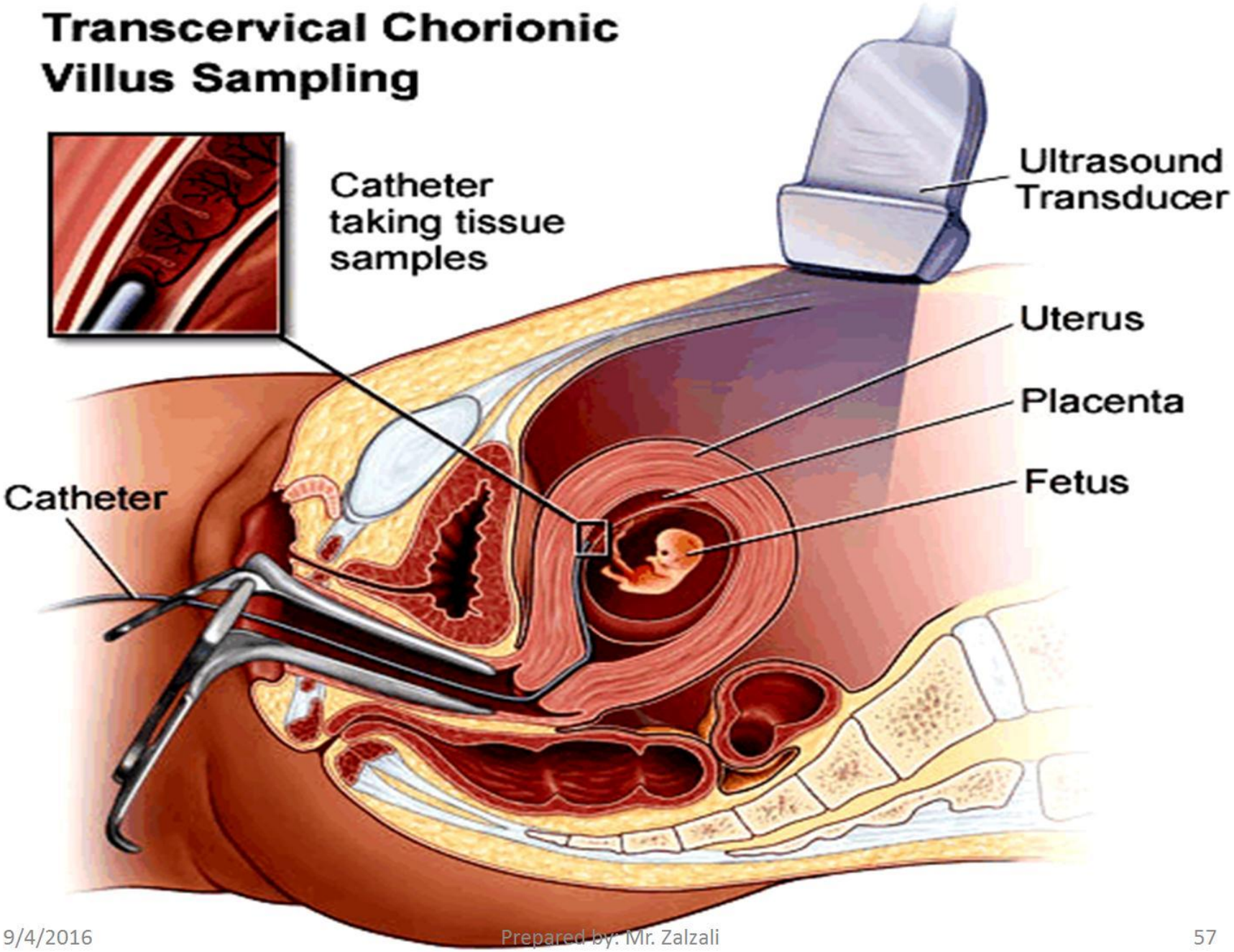
-Possible disadvantages:

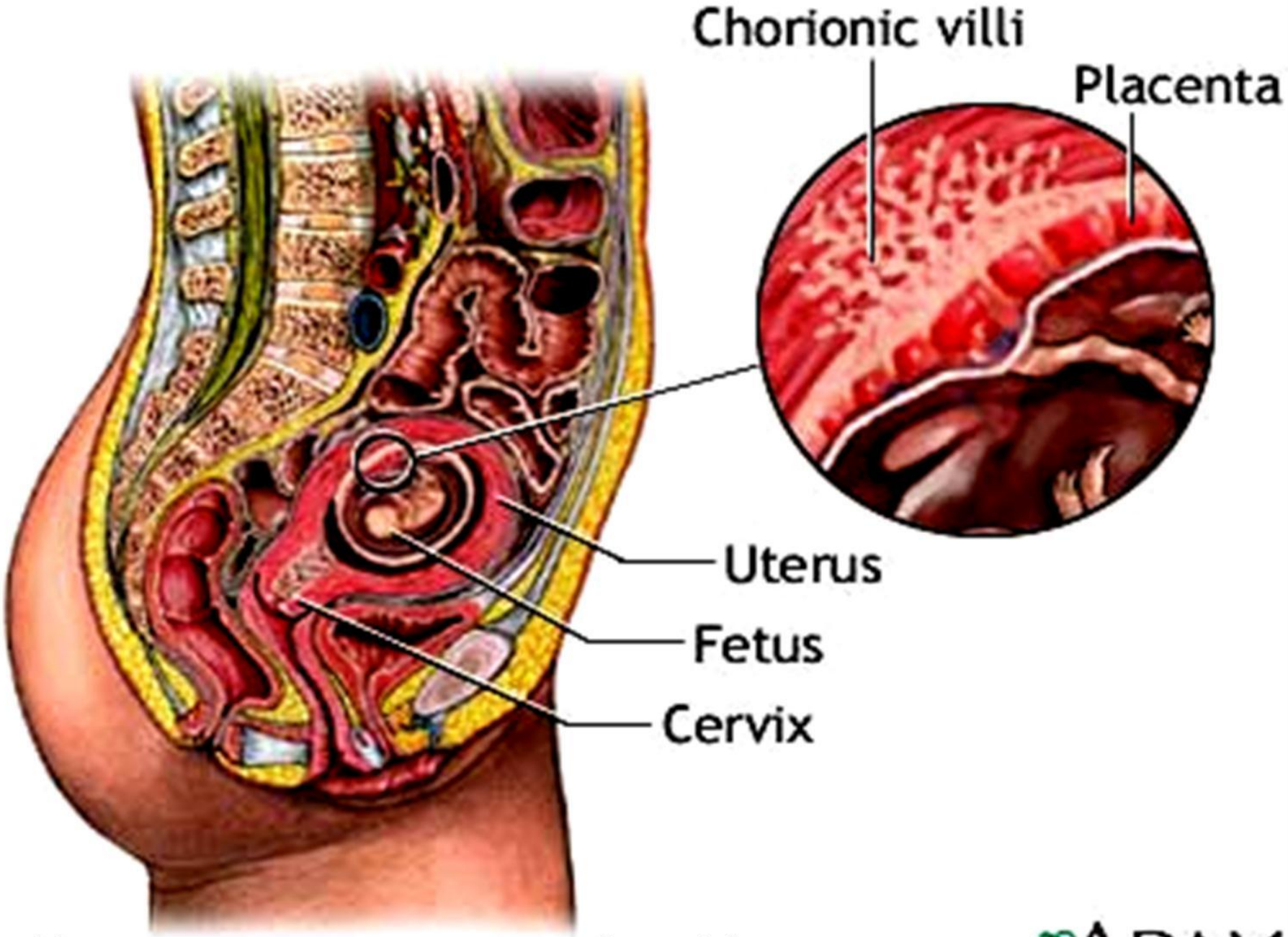
1- A small risk of miscarriage (0.5 – 1 %) regardless of the genetic status of the fetus.

2- It's carried out relatively in late pregnancy (around week 12-16). so termination of pregnancy will be a difficult decision for parents.

3- It takes a long time to get the results of karyotyping.

(about 2-4 weeks) → Puts parents under emotional stress.

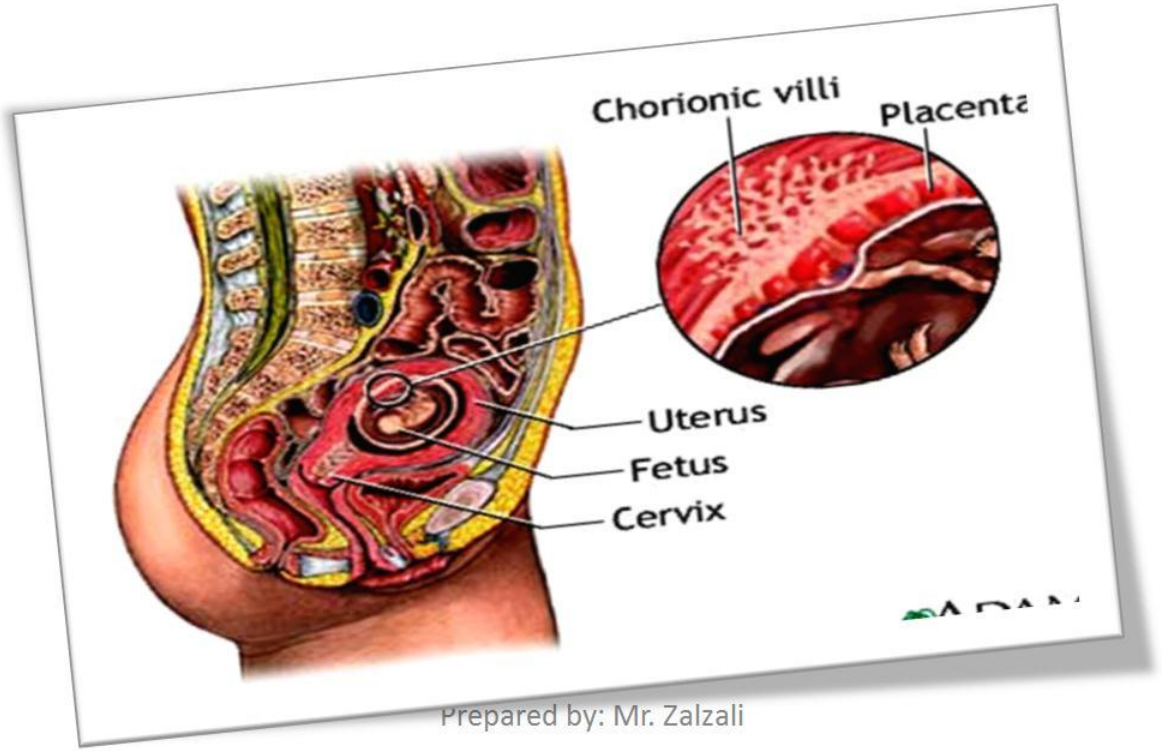

Transcervical Chorionic Villus Sampling
Catheter taking tissue samples
Ultrasound Transducer
Uterus
Placenta
Fetus
Catheter


Chorionic villi
Placenta
Uterus
Fetus
Cervix

3- Chorionic Villi Sampling (cvs):

-A technique of taking a sample from the finger like projections of placenta to obtain dividing cells from chorion villi → make a karyotype for the chromosome in the fetal dividing cells. (chorionic-villi cells carry the same genetic make-up as the fetus).

- Sample taken thru vagina or abdomen.



- **Advantages of CVS :**

1-Cells are karyotyped without culturing them in lab

2- A bigger sample of fetal cells can be examined because this sample is taken usually through the vagina.

3- Its usually carried out earlier than amniocentesis (8-12 weeks) of pregnancy → so if parents decide to terminate this pregnancy. The decision is less traumatic

4- The results of the test are obtained within few days and its less stressful for parents

- **Possible disadvantages:**

1- Higher risk of miscarriage than amniocentesis (2.5- 5 %)

2- CVS is more expensive than amniocentesis

3- All paternal test (chromosomes from the father are inactivated during CVS test, so if there is a mutation in these chromosomes, this can't be detected.