

KARYOTYPING
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∞ The Study Of
Chromosomes And Related
Diseases That is caused by
changes in the number of
chromosomes and their
structure

KARYOTYPING



- ❧ Karyotype is an organized profile of an individual's chromosomes
- ❧ Karyotyping is a technique that is use to examine chromosomes in a sample of cells which can help identify genetic problems as the cause of disorder or a disease

MAIN PURPOSE OF KARYOTYPING



- ☞ Main purpose of the karyotyping is to locate or visualize the changes in the number of chromosomes and abnormality in the structure
- ☞ Also to locate the evolution

Basis of karyotyping



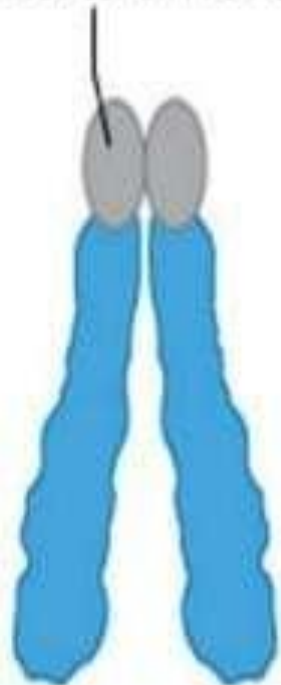
Basically karyotyping is based on three patterns

On the basis of size of chromatids

On the basis of bending patterns

On the basis of centromeric positions

Replicated
centromere



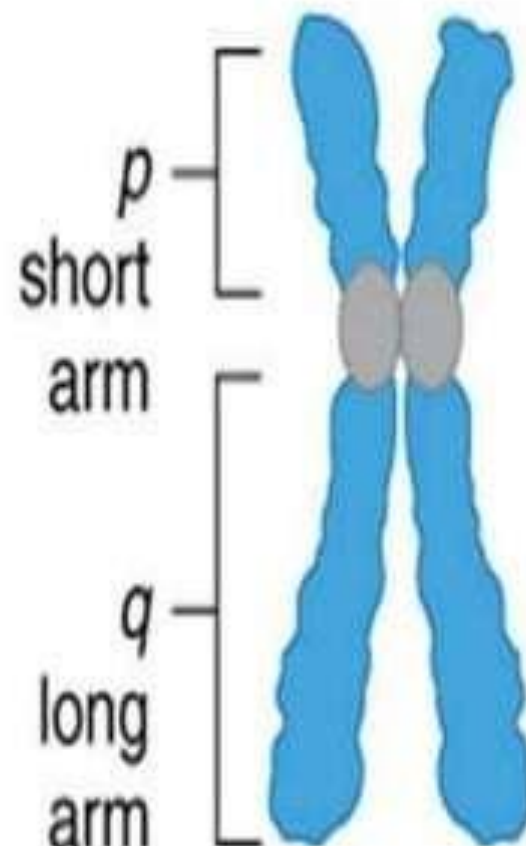
Telocentric

a.



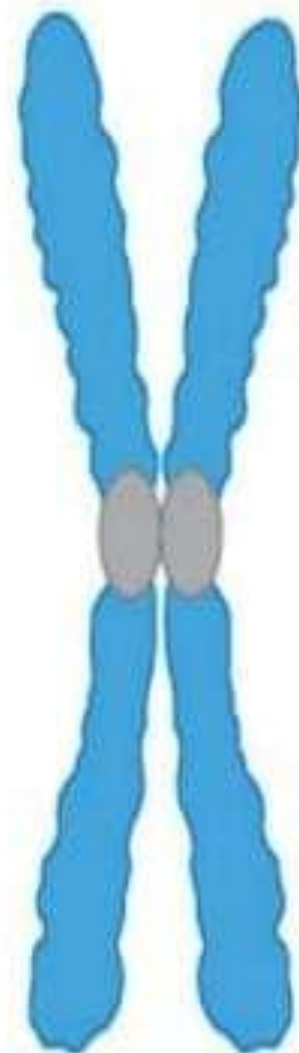
Acrocentric

b.



Submetacentric

c.

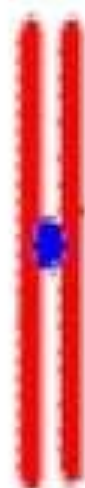


Metacentric

d.

...and centromere position.

Centromeric position and arm length



Metacentric



Submetacentric



Acrocentric



Telocentric

Continued.....



- ☞ Through karyotype analysis , you will obtained the following information about your cell lines
- ☞ Species identification .
- ☞ Index of genome stability
- ☞ Validation of normal diploid karyotype
- ☞ Numerical chromosomes abnormalities
- ☞ Monoploidy , poliploidy
- ☞ Monotomy , trisomy
- ☞ Structural abnormalities

Continued.....



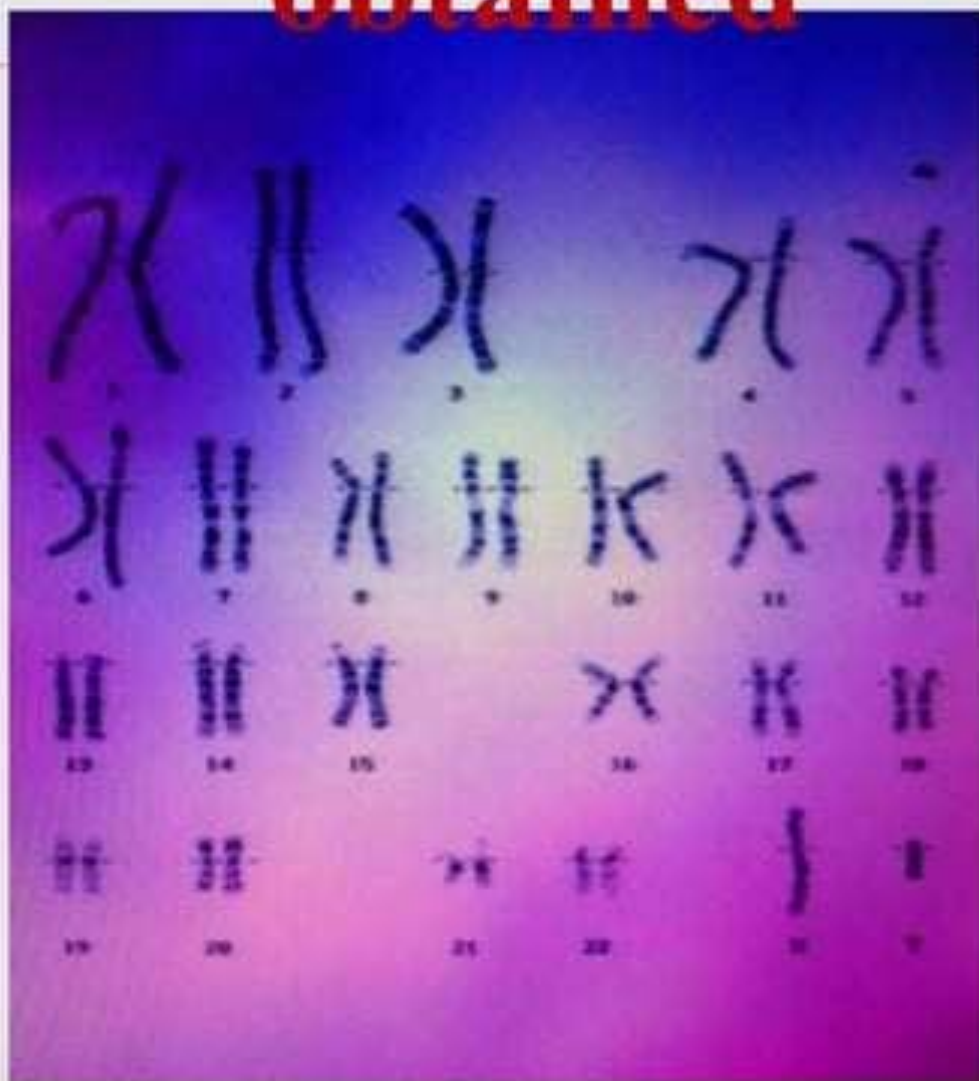
Deletion

Duplication

Translocation

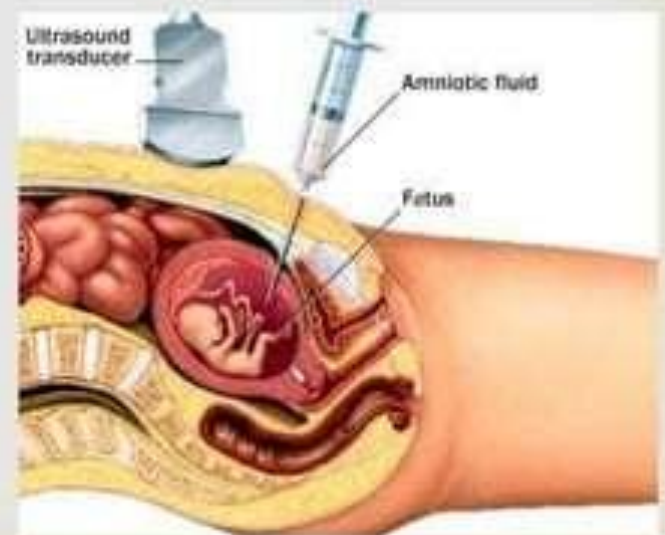
Inversion

The karyotype is obtained



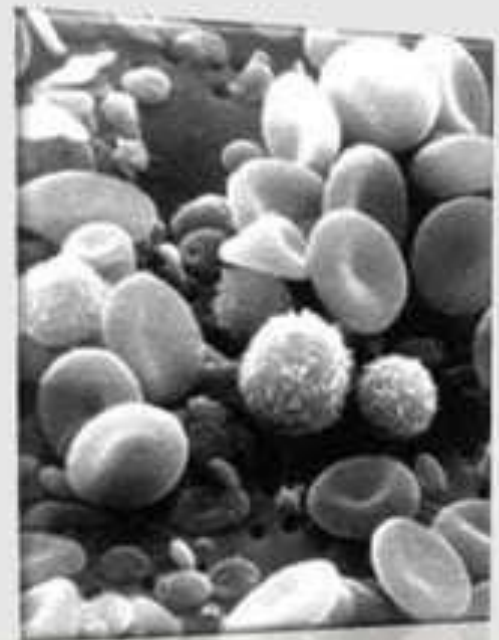
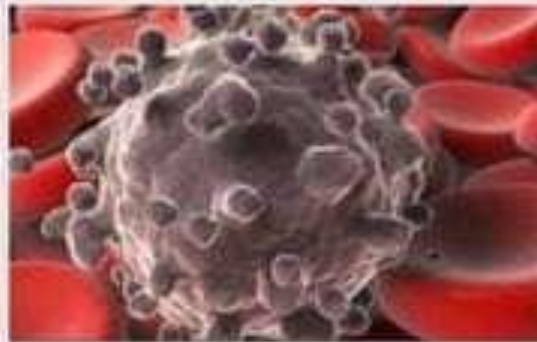
1. Sample collection

The first step in performing a karyotype is collecting the “sample”. The sample can either be collected from the amniotic fluid through amniocentesis or a piece of the placenta collected during a chorionic villi sampling test, or simple 5ml of venous blood.



2. Separating the cells

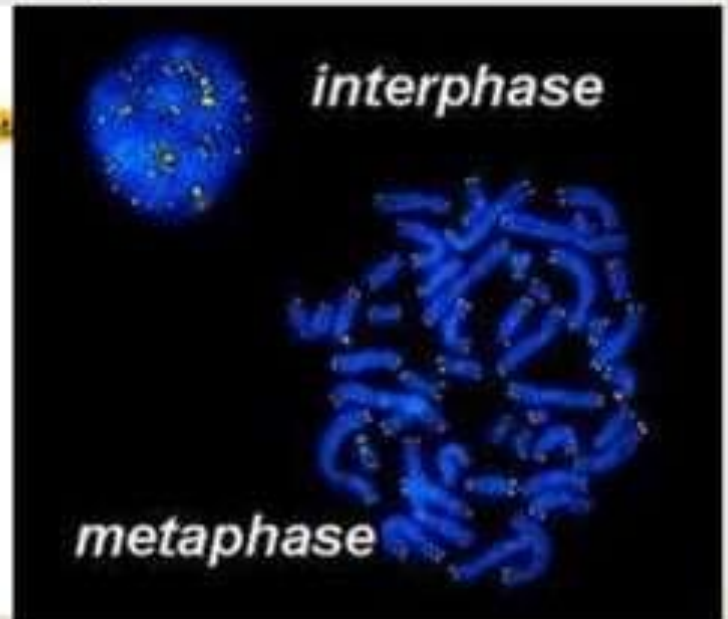
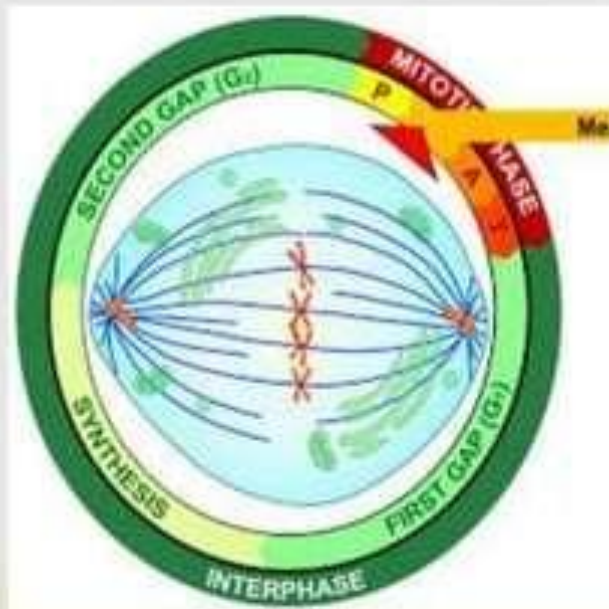
In order to analyze chromosomes, the sample must contain cells that are actively dividing (or in mitosis). In blood, the white blood cells are actively dividing cells. Once the samples reaches the cytogenetics lab, the non divided cells are separated from the dividing cells using special chemicals. The red blood cells are separated and the collection of white blood cells take place because mature red blood cell no longer have a nucleus. Therefore you can't karyotype.



3. Synchronizing cells

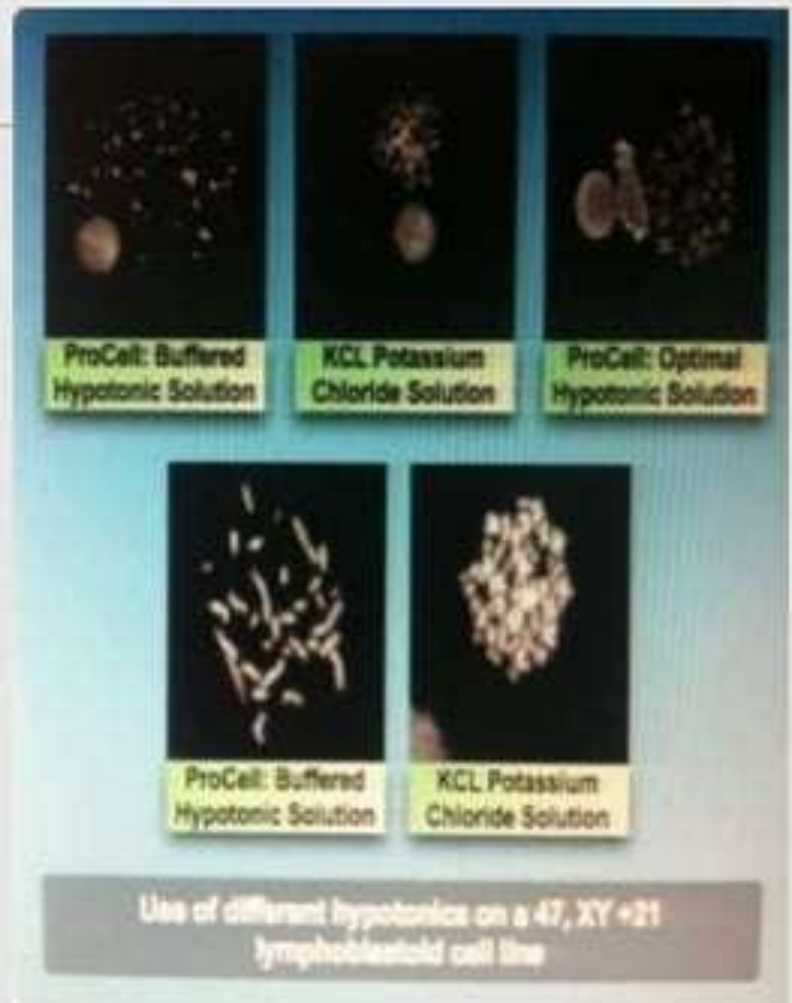


In order to see chromosomes clearly under a microscope, chromosomes have to be their most compact form. This compact form occur at the specific stage of mitosis called metaphase. In order to get all the cells to this specific stage of cell division. The cells are treated with a chemical called colchicines which stops cell division metaphase.



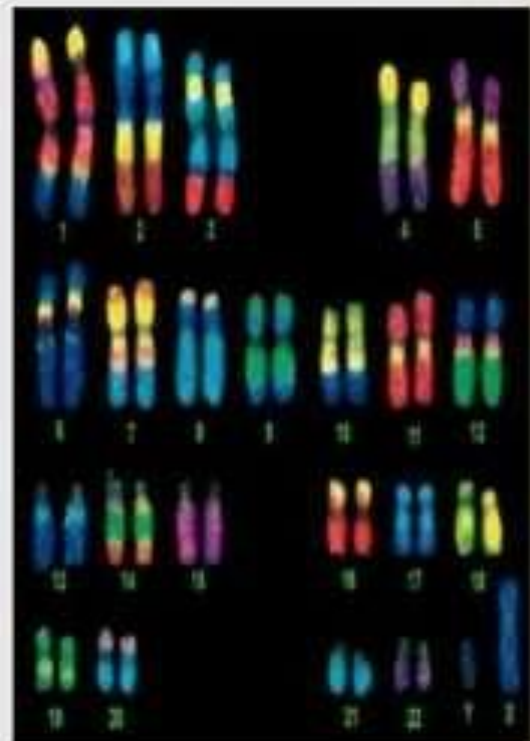
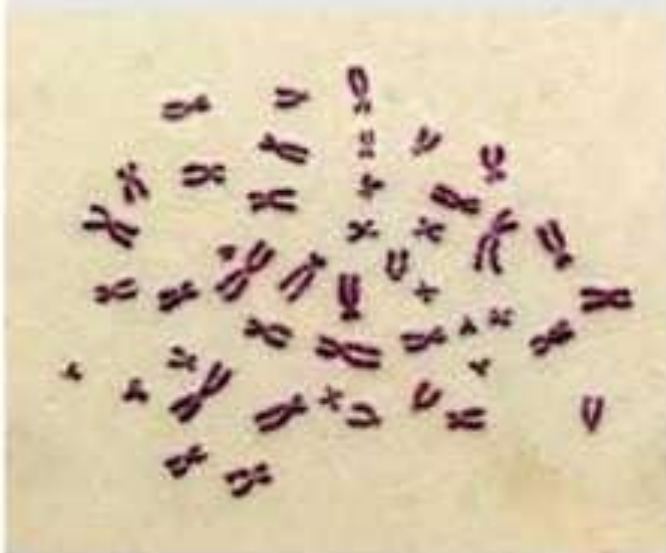
4. Releasing the chromosomes from their cells

In order to see these compact chromosomes under a microscope, the chromosomes have to be out of the white blood cells. Moreover, this is done by the treating the white blood cells with a special solution which causes the swelling and lysing (busting-breaking down) of lymphocytes. Consequently this provides a better spreading of chromosomes.



5. Staining the chromosomes

Chromosomes are colorless. In order to be able to tell one chromosome from another, a special dye called Giemsa dye is applied to the chromosomes on the slide.

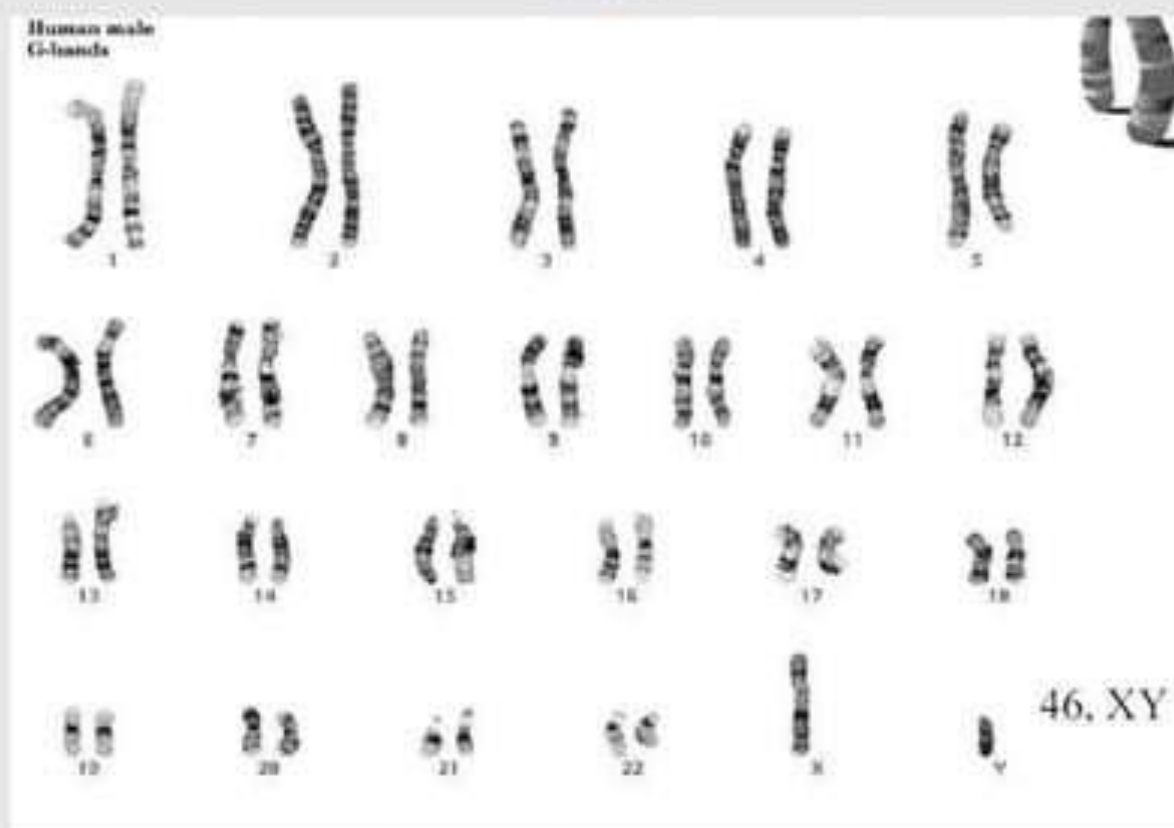


Smear

Human male
G-bands



Human Karyotype



46,XY

Abnormal results may be due to a genetic syndrome or condition, such

as:

- ⌘ Down syndrome
- ⌘ Klinefelter syndrome
- ⌘ Philadelphia chromosome
- ⌘ Trisomy 18
- ⌘ Turner syndrome



Thanks



The end