

# HUMAN GENETICS

## " LECTURE 1

### Human Chromosomes: Genotypes/Phenotypes

Editing link:

<https://docs.google.com/document/d/1WvdeC1atp7J-ZKWOUSukSLsEcosjZ0AqV4z2VcH2TA0/edit?usp=sharing>

Color index:

- Important
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- Drs' notes
- Extra Information



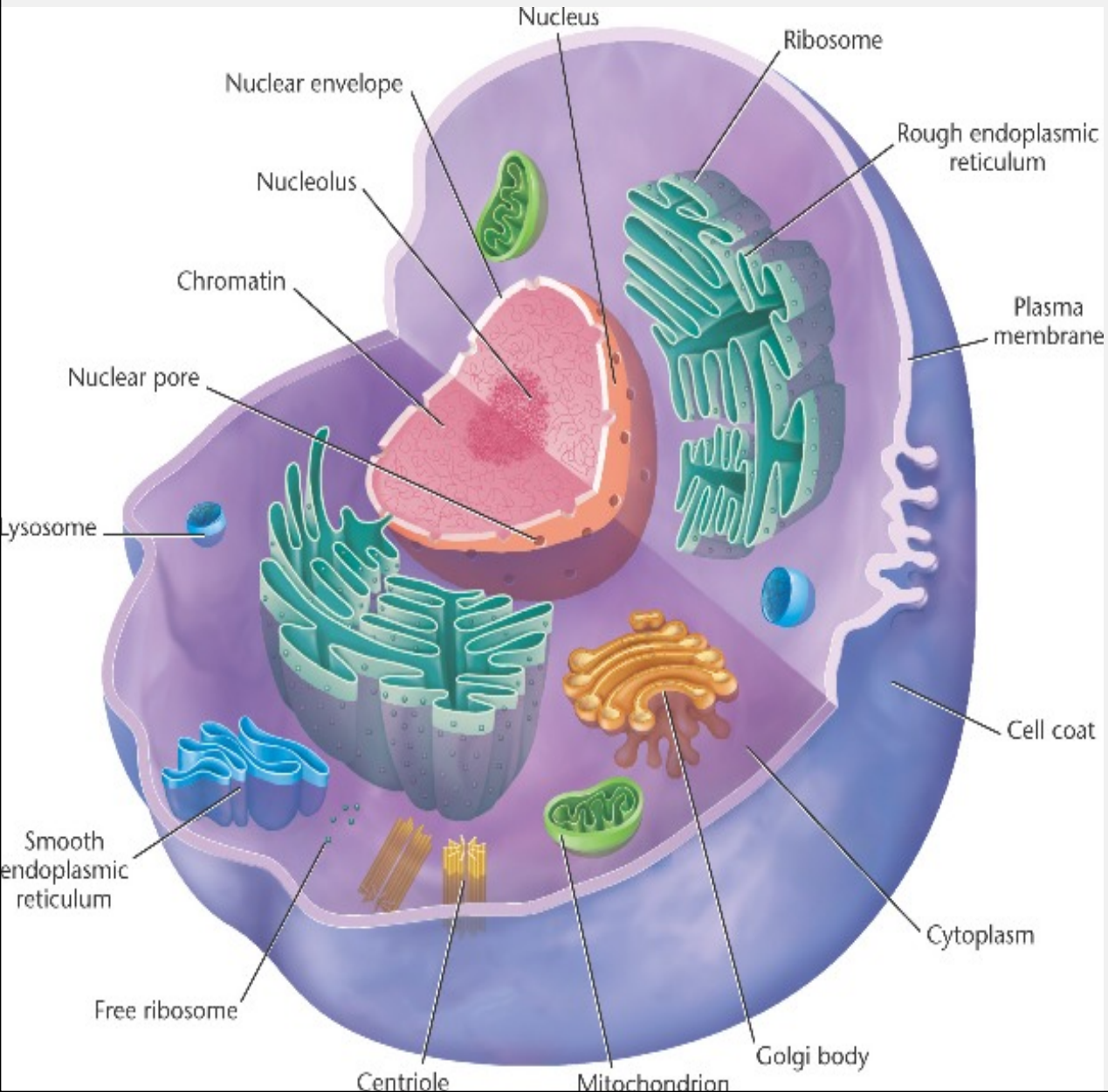
# Objectives:

- Describe the number, structure, and classification of human chromosomes.
- Explain what a Karyotype is and how it is obtained.
- Describe chromosomal banding and explain its use.
- Describe the process of in situ hybridization and the information it provides.





# Eukaryotic cell



# GENETICS :

## ■ Cytogenetics: (Chromatin)

The study of the structure and function of chromosomes and chromosome behaviour during somatic and germline division

## ■ Molecular genetics: ( the sequence i.e. A,G,C,T.)

The study of the structure and function of genes at a molecular level and how the genes are transferred from generation to generation.



# Cytogenetics:

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Human Cytogenetics:  
the study of human chromosomes in health and disease.

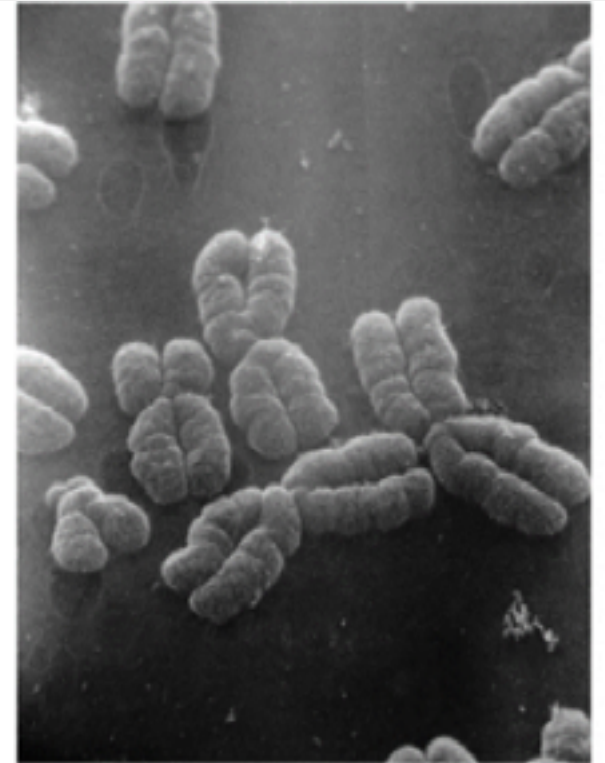
Chromosome studies are an important laboratory diagnostic procedure in:

1. Prenatal diagnosis.
2. Certain patients with mental retardation and multiple birth defects.
3. Patients with abnormal sexual development.
4. Some cases of infertility or multiple miscarriages.
5. In the study and treatment of patients with malignancies & hematologic disorders.

# CHROMOSOMES:

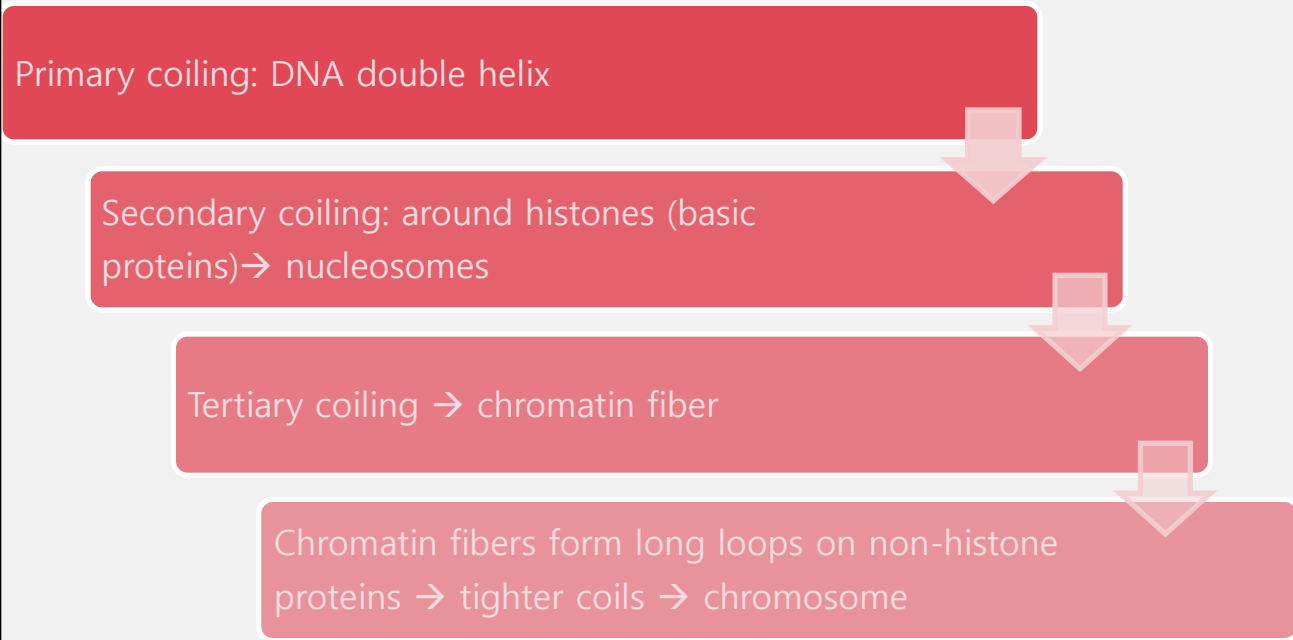
- Carry genetic material
- Heredity: each pair of homologues consists of one paternal and one maternal chromosome.
- The intact set is passed to each daughter cell at every mitosis.

## EM OF HUMAN CHROMOSOMES

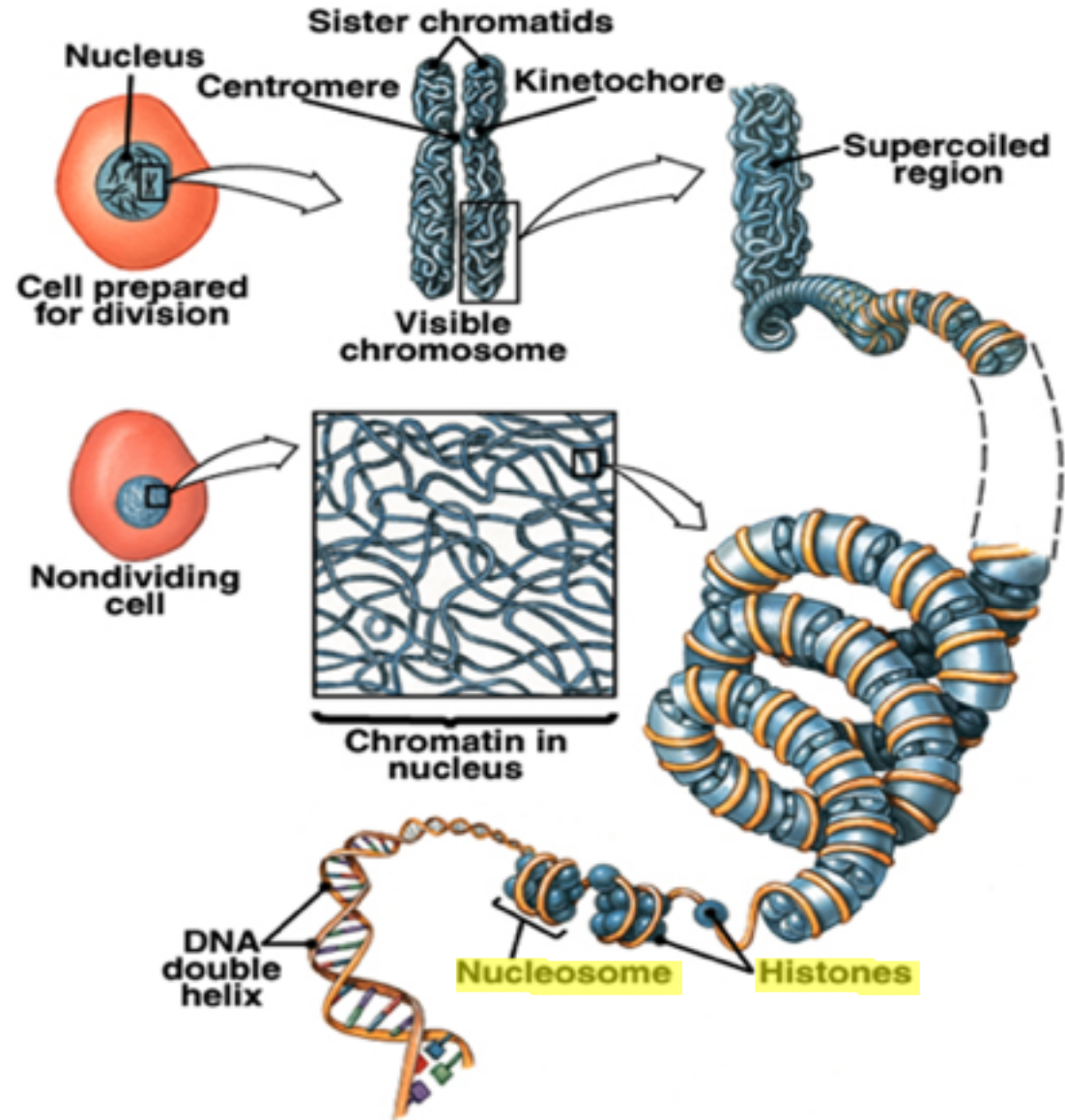


# Structure of Chromosomes

## Orders of DNA coiling and folding:



Important information:  
→ DNA condenses when the cell is ready for chromosomes splitting (division).  
Notice that: the DNA replication need the chromosome to be relaxed (uncondensed) >> if the chromosome condenses the DNA can't replicate.



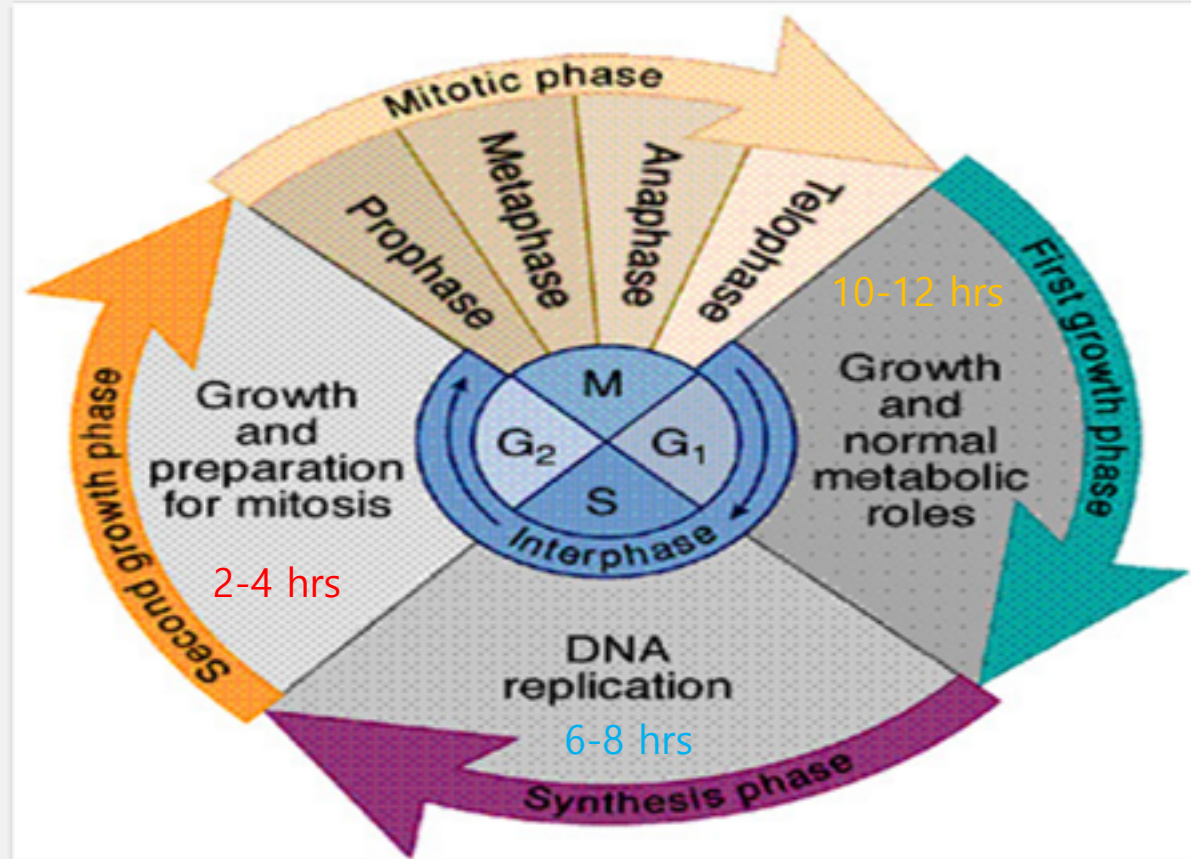
# Mitotic cell cycle

There are two phases in the mitotic cell cycle:

- 1-interphase:
  - GAP1
  - synthesis
  - GAP2
- 2-mitotic phase

Mitotic phase:  
Chromosomes split (segregate)

3-GAP2:  
The chromosomes condense.



1-GAP1:  
Cell's chromosomes don't condense to prepare DNA to replicate.

2-Synthesis:  
DNA replication.



# Karyotype:

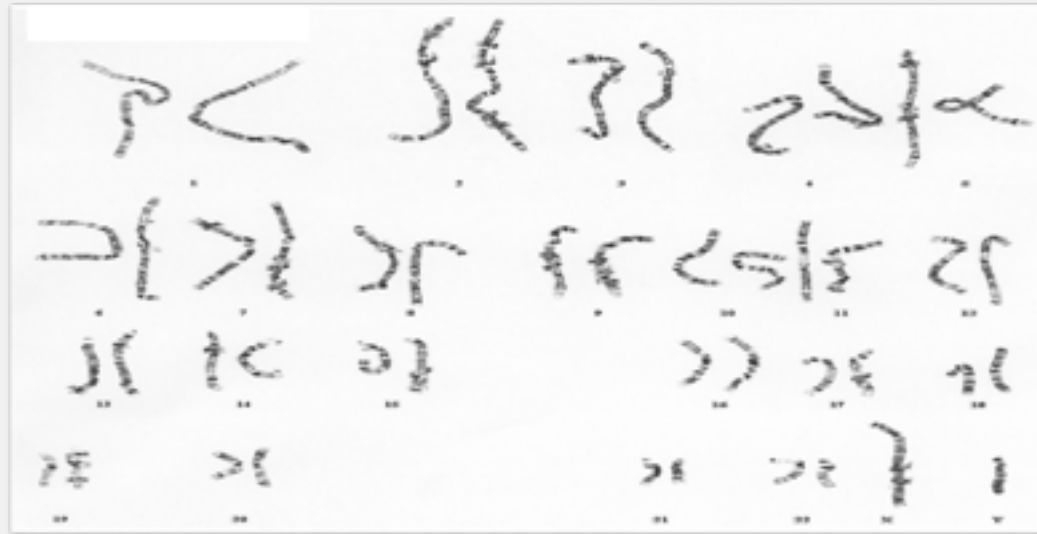
## Cytogenetics:

- Non-Banded Karyotype
- Banded Karyotype
- High resolution Karyotype

## Molecular cytogenetics:

- Fluorescent In-Situ Hybridization (FISH)

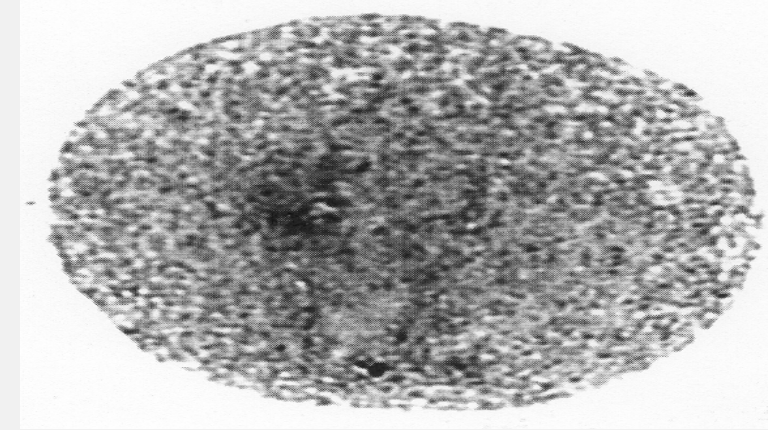
437 note: Technology that is used to examine a specific gene by (Primers) that are made in the lab.



# Karyotype

steps involved :

- CULTURING
- HARVESTING
- Slide-Making
- Banding
- Staining
- Karyotyping
- Chromosome Analysis



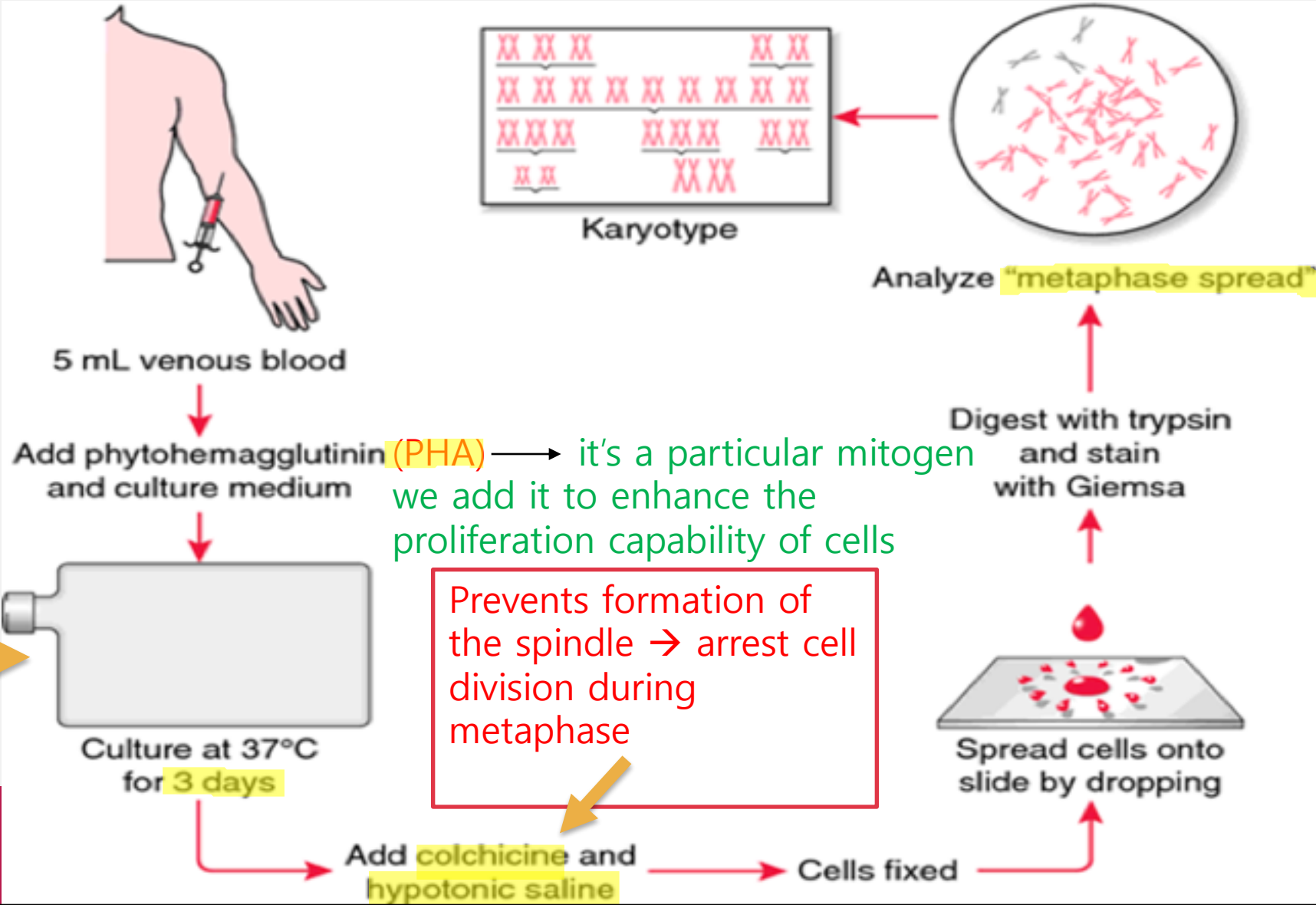
شرحها الدكتورە بشكل بسيط

# Chromosome Preparation from Peripheral Blood

IMPORTANT know the added chemicals and in which they arrest the cell division

Culture media contains Phytohemagglutinin to stimulate T lymphocytes to divide

Important information:  
→The name of the chemical that stops all cells in the same stage is (colchicine or colcemid)



# Metaphase chromosomes:

- The 2 sister-chromatids are principally held together at the **centromeric region**.
- Each chromosome has a centromere (CEN), region which **contains the kinetochore**,
- CEN divides the chromosome into two arms: the **short arm (p arm)** and the **long arm (q arm)**.
- Each arm terminates in a **telomere**.

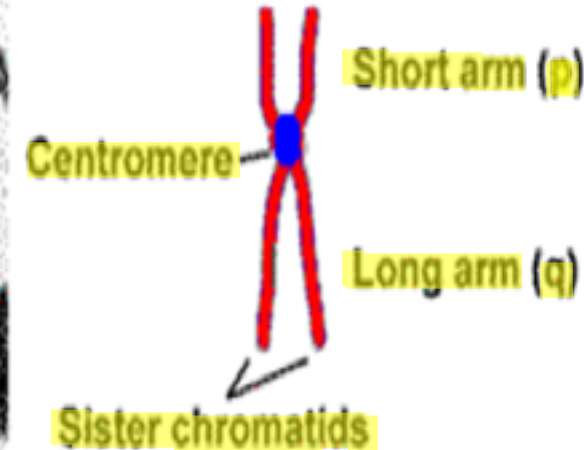
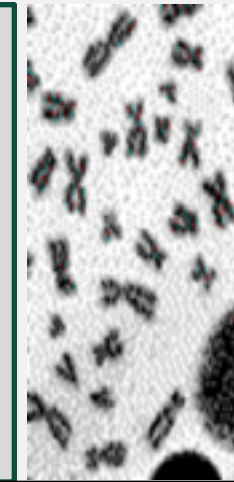
A single complete set of chromosomes. (N=23 for humans)



Important information:  
→to identify the chromosomes:  
1-the position of the centromere as it's in the (middle,terminal,not p arm)  
2-the length of the chromosome  
3-the ratio of the (p to q) or (q to p)



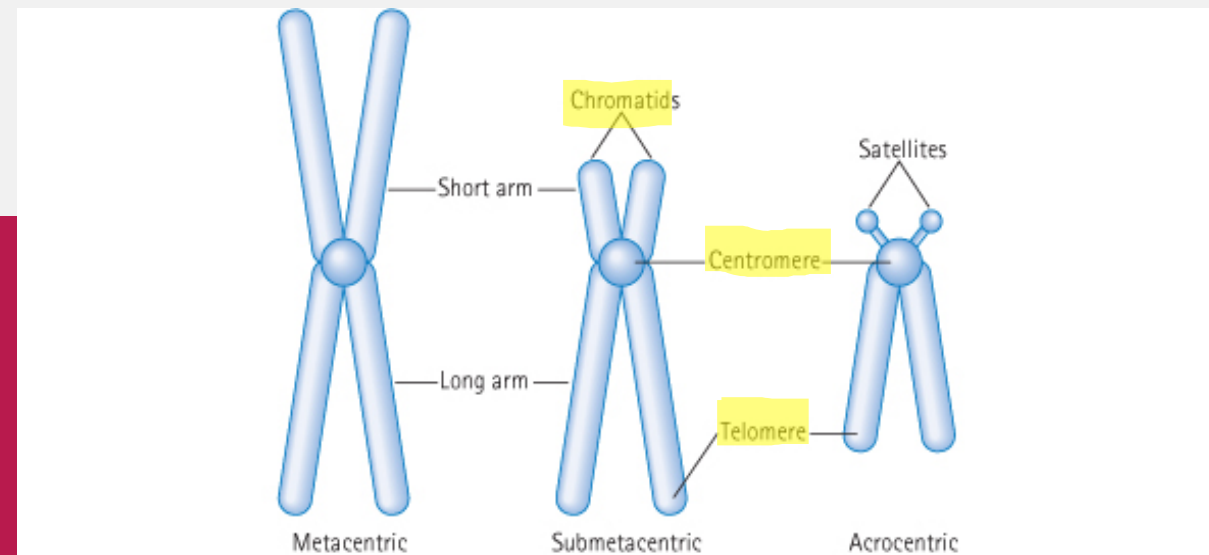
Doctor's notes:  
Telomeres function to protect the chromosomes.  
- (They do that by keeping the ends of chromosomes from sticking together, they seal off the ends of chromosomes)  
- The telomeres get shorter as a person ages, hence representing a degenerative process.  
- Females have longer telomeres than males.





# Centromeric position and arm length:

- The ratio of the lengths of the two arms is constant for each chromosome.
- This ratio is an important parameter for chromosome identification and allows classification of chromosomes into several basic morphologic types:
  - metacentric*
  - sub-metacentric*
  - acrocentric*
- In the human karyotype chromosome pairs 13, 14, 15, 21, 22 are *acrocentric*



Important information: (you have to know why we have the telomeres and where are they.)

- at the end of the each chromosome there are terminals caped with (telomeres).
- their function is to protect the chromosomes from getting affected. If the telomeres got affected, the chromosomes also will be affected >> and from that, we will have a structural abnormality.

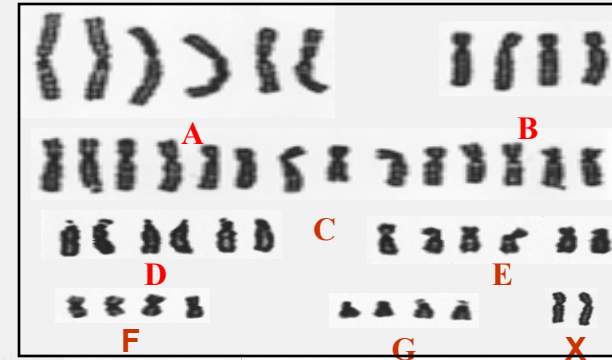
# Chromosomal classification

- 22 pairs of autosomes, numbered from 1 to 22 by order of decreasing length
- 1 pair of sex chromosomes:  
XX in the female,  
XY in the male.

## Karyotyping

Based on:

- 1-the length
- 2-the position of the centromere
- 3-the presence or absence of satellites



Important information:

→karyotyping is based on:

- 1-the length
- 2-the position of the centromere
- 3-the presence or absence of satellites



# Items in the Description Of Karyotype:

## Normal Karyotypes

46, XY Males  
46, XX Females

## Abnormal Karyotypes

47,XY,+ 21 Down Syndrome

45,X Turner's syndrome → Loss of y or x



# Banding

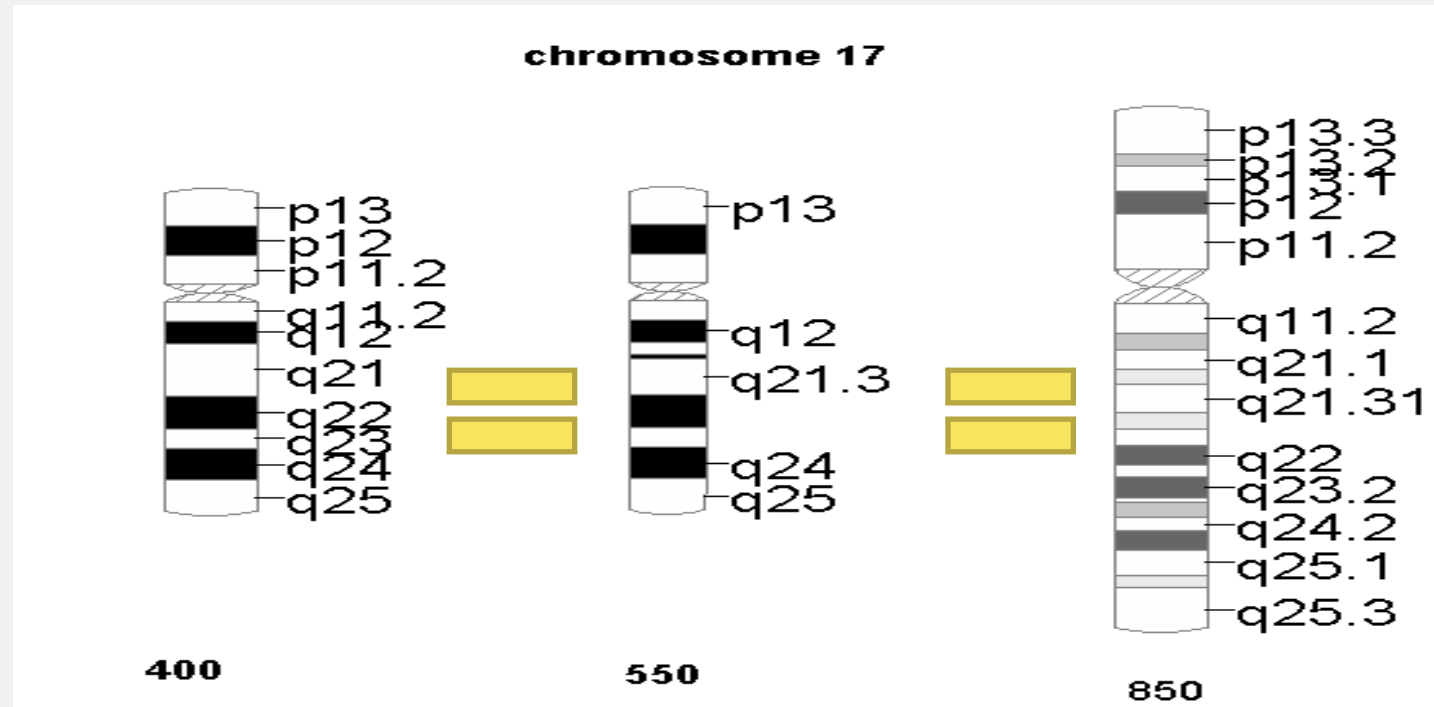
- ❖ **Certain staining techniques cause the chromosomes to take on a banded appearance,**
- ❖ **Each arm presenting a sequence of dark and light bands.**
- ❖ **Patterns are specific and repeatable for each chromosome,**
- ❖ **Allowing accurate identification and longitudinal mapping for locating gene positions and characterising structural changes.**
- ❖ **Patterns, and the nomenclature for defining positional mapping have been standardised**

Doctor's notes:  
dark chromosomal  
regions are "inactivated",  
light regions are  
"activated".



# Chromosome Banding

- Band resolution = estimate of number of light + dark bands per haploid set of chromosomes
- 400 → 850+



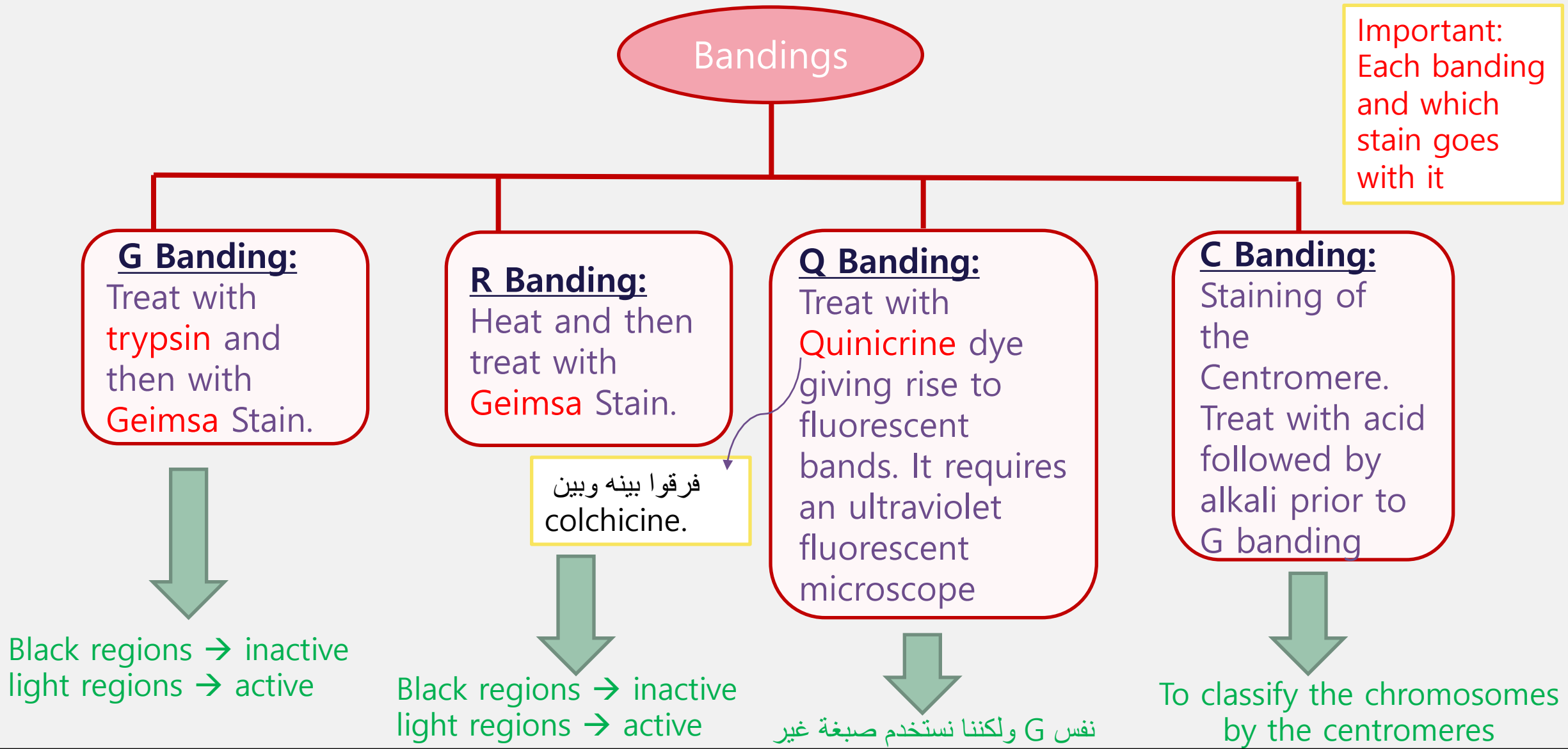
أهم شيء نعرف ان هذا هو شكل الباندينق يكون اجزاء سوداء واجزاء بيضاء + انه يفيدنا في معرفة الاجزاء الناقصة في الكروموسوم

Important information:

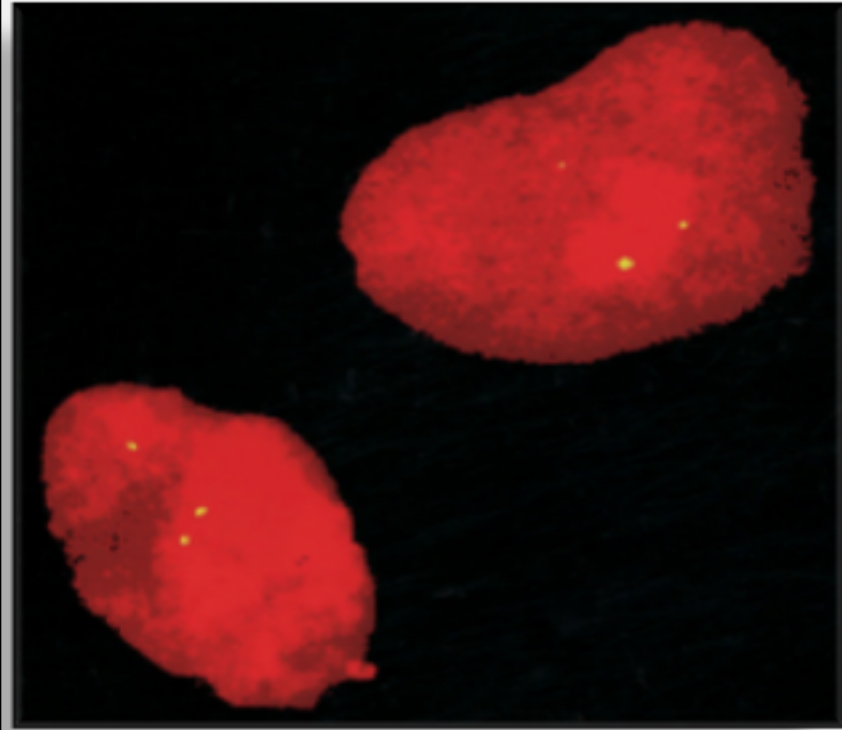
→the light regions are "activated"

→the dark regions are "inactivated"

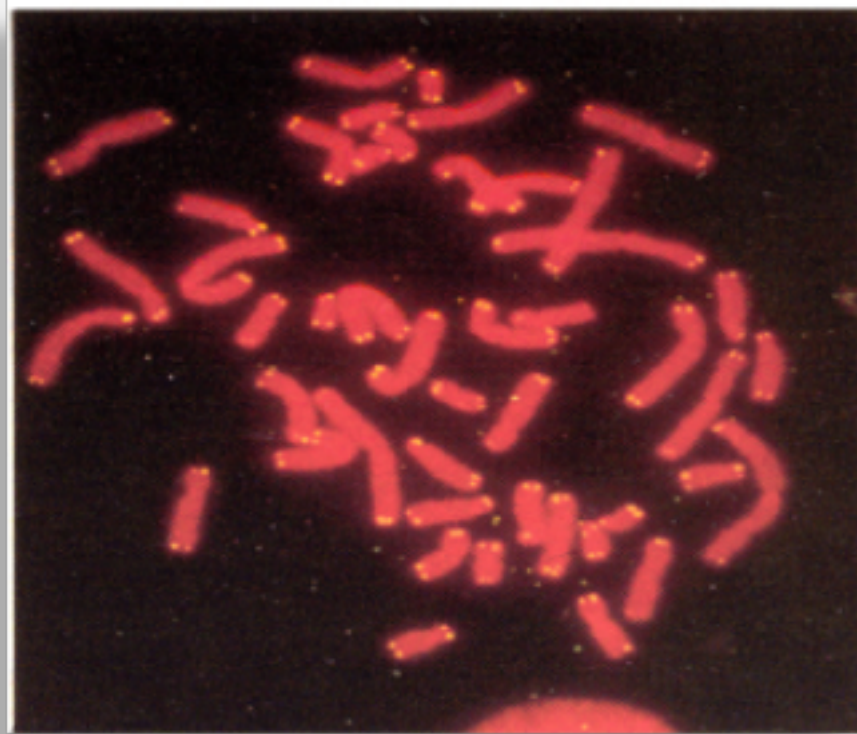
# bandings



# Fluorescence In-Situ Hybridization (FISH)



FISH of interphase nuclei with a chromosome 21 centromeric probe showing 3 signals consistent with trisomy 21



FISH of metaphase with a probe for telomere showing signals at the end of each chromatid

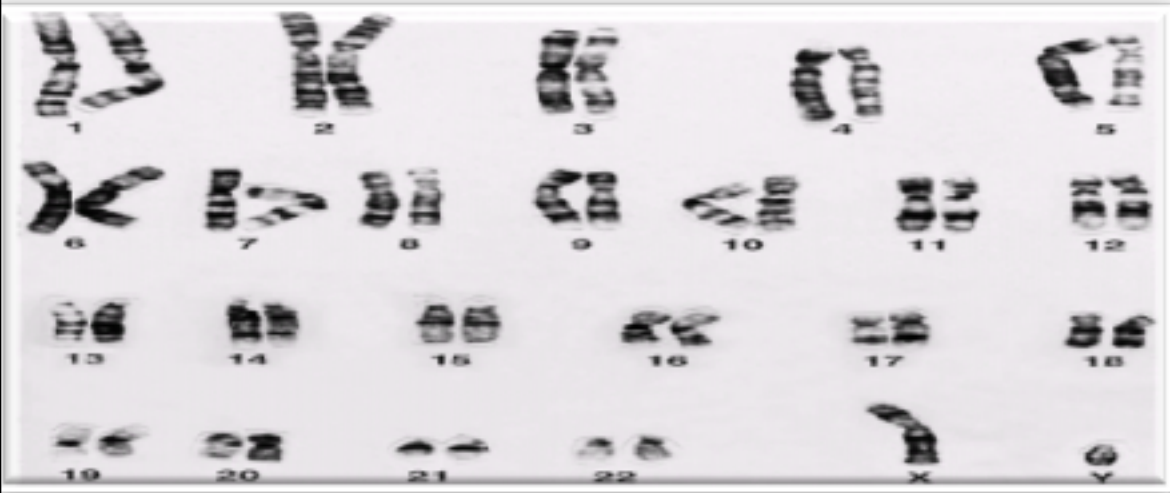
فكرته باختصار انه عبارة عن تسلسل معين من القواعد راح يرتبط بالقواعد المكملة له ويعطيني اللون عشان اركز على هذا التسلسل



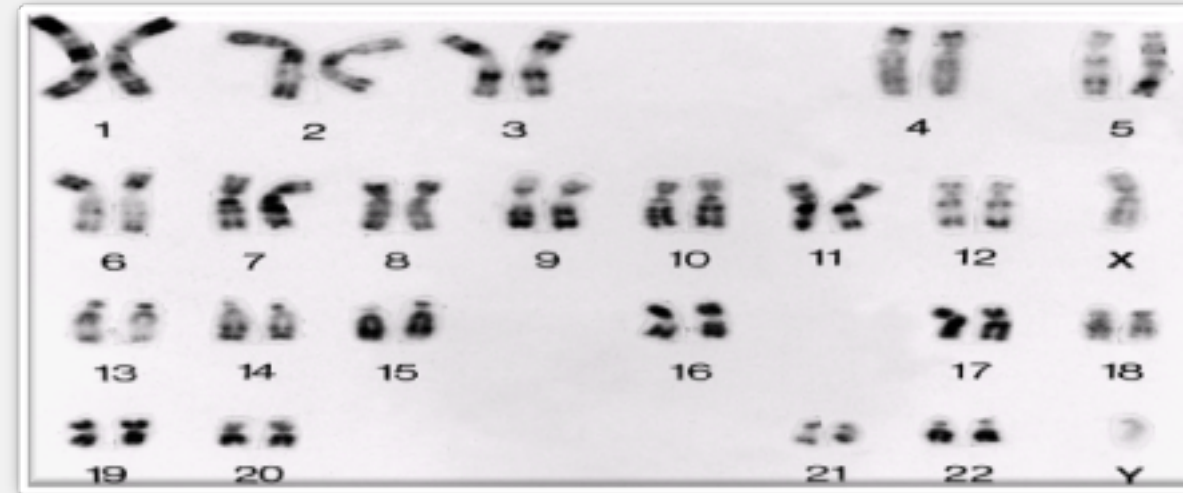
Important information:

ماكانت واضحة..راح نسأله ان شاء الله بالمحاضرة الجايه ونبليكم

# Banded Karyotype: Normal Banded Karyotypes



A normal **G-banded**  
male Karyotype



A normal **R-banded**  
male Karyotype



# Take Home Message

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- The packaging of DNA into chromosomes involves several orders of DNA coiling and folding.
- The normal human karyotype is made up of 46 chromosomes consisting of 22 pairs of autosomes and a pair of sex chromosomes, XX in the female, and XY in the male.
- Each chromosome consists of a short (p) and a long (q) arm joined at the centromere.
- Chromosomes are analyzed using cultured cells and specific banding patterns can be identified using special staining techniques.
- FISH is based on the ability of a single-stranded DNA probe to anneal to its complementary target sequence. It can be used to identify and study genes on chromosomes in metaphase or interphase.

# Important information

Important information:

→DNA condenses when the cell is ready for chromosomes splitting (division).

Notice that: the DNA replication need the chromosome to be relaxed (uncondensed)>>if the chromosome condenses the DNA can't replicate.

Important information:

→The name of the chemical that stops all cells in the same stage is (colchicine or colcemid)

Important information:

→to identify the chromosomes:

- 1-the position of the centromere as it's in the (middle,terminal,no p arm)
- 2-the length of the chromosome
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Important information: (you have to know why we have the telomeres and where are they.)

→at the end of the each chromosome there are terminals caped with (telomeres).

→their function is to protect the chromosomes from getting affected.

If the telomeres got affected, the chromosomes also will be affected>> and from that, we will have a structural abnormality.

Important information:

→the light regions are "activated"  
→the dark regions are "inactivated"

Important information:

ماكانت واضحة. راح نسأله ان شاء الله بالمحاضرة الجايه ونبلغكم

Important information:

→karyotyping is based on:

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- 2-the position of the centromere
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# Quiz

key answer:1-D-2-B-3-C 4-A 5-B

1-Prevents formation of the spindle (stop the cell division in metaphase)?

- |                   |                       |         |                                    |
|-------------------|-----------------------|---------|------------------------------------|
| A) Culture medium | B) phytohemagglutinin | C) 37°C | D) Colchicine and hypotonic saline |
|-------------------|-----------------------|---------|------------------------------------|

2-two sister-chromatids held together by?

- |                |               |          |          |
|----------------|---------------|----------|----------|
| A) kinetochore | B) centromere | C) q arm | D) p arm |
|----------------|---------------|----------|----------|

3-The study of the structure and function of genes?

- |              |                 |                       |               |
|--------------|-----------------|-----------------------|---------------|
| A) Karyotype | B) Cytogenetics | C) Molecular genetics | D) Chromosome |
|--------------|-----------------|-----------------------|---------------|

4- Which one of these stains goes with Q banding?

- |               |           |            |               |
|---------------|-----------|------------|---------------|
| A) Quinacrine | B) Geimsa | C) Trypsin | D) Colchicine |
|---------------|-----------|------------|---------------|

5- The genotype of turner's syndrome is

- |             |         |           |          |
|-------------|---------|-----------|----------|
| A) 47,XY+21 | B) 45,X | C) 47,XXY | D) 46,XX |
|-------------|---------|-----------|----------|

# Thank You



## TEAM LEADERS:

- محمد المطيري
- جود العتيبي

## GIRLS TEAM MEMBERS:

- طيف الشمري
- سفانا العمر
- ريم القرني
- ريناد الكنعان
- في البقمي
- لمى الدخيل
- مي بابعير
- نجود عبداللطيف

## BOYS TEAM MEMBERS:

- عبدالعزيز الفهيد
- عمر العماري
- نايف السبير
- فيصل العمر
- البراء السيف

Special thanks to **Team 437**

