Human chromosomes

HUMAN GENETICS

Red: important.
Pink: F-slides
Blue: M-slides
Green: doctor's Notes
Gray: extra

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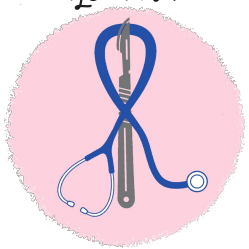


HUMAN GENETICS 441

MED441
KING SAUD UNIVERSITY

Objectives

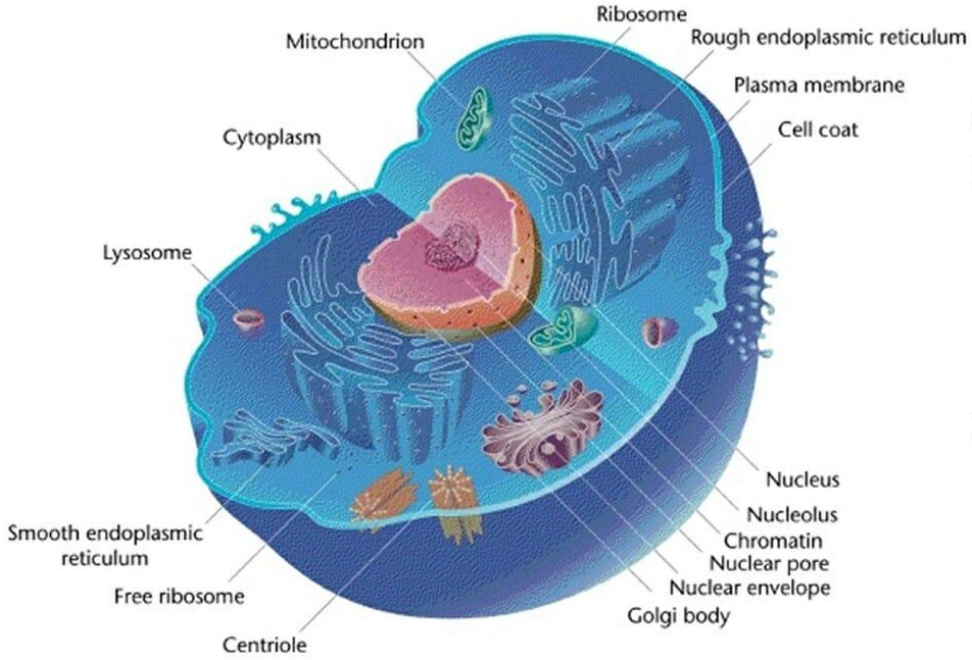
01 Describe the number, structure, and classification of human chromosomes.

02 Explain what a Karyotype is and how it is obtained.

03 Describe chromosomal banding and explain its use.

04 Describe the process of in situ hybridization and the information it provides.

Eukaryotic cell



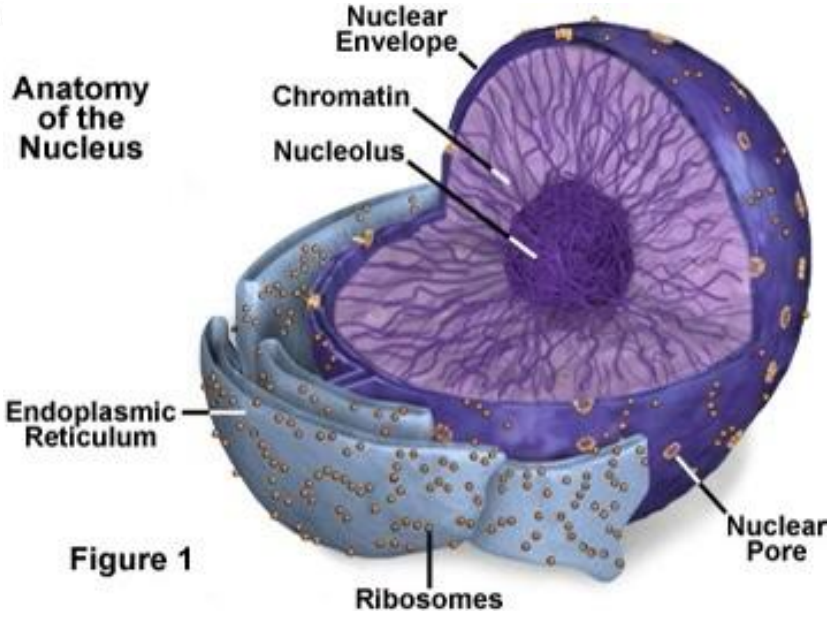


Figure 1

Genetics

Cytogenetics

The study of structure and function of chromosomes and their behaviour during somatic and germline division.

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

Molecular genetics

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

- Fluorescent in situ hybridization (FISH) cell is intact and not retrieved

Human cytogenetics

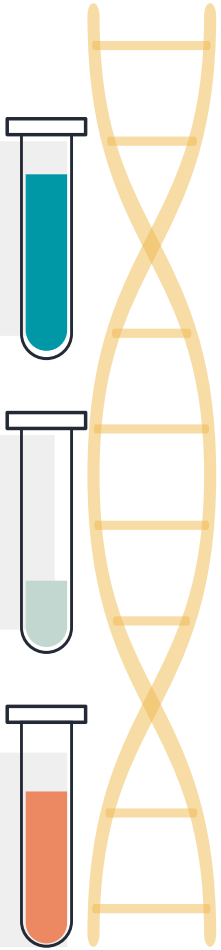
The study of human chromosomes in health and disease.

Chromosome studies are an important laboratory diagnosis in:

- Prenatal diagnosis: Two types noninvasive (NIPT) and invasive
- Certain patients with mental retardation and multiple birth defects.
- Patients with abnormal sexual development.
- Some cases of infertility or multiple miscarriages.
- In the study and treatment of patients with malignancies & hematologic disorders.

Chromosomes

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



Classes

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

Structure of chromosomes

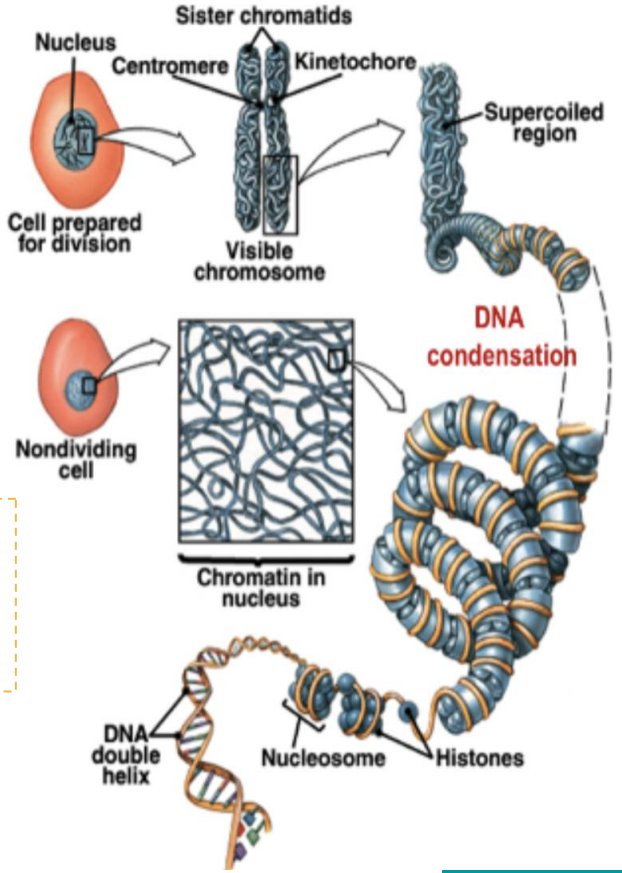
Primary Coiling → Secondary Coiling → Tertiary Coiling → Chromosomes

DNA double helix

Around histones (basic proteins) → nucleosomes

Chromatin fiber

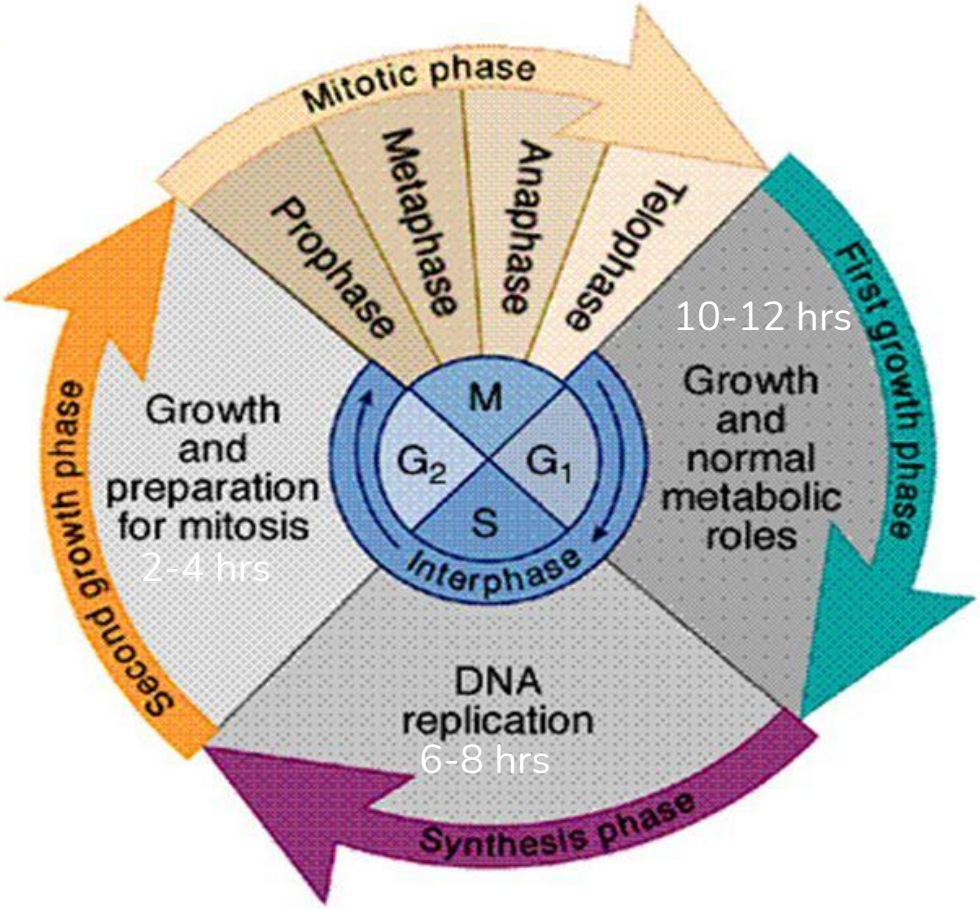
Chromatin fibers form long loops on non-histone proteins → tighter coils



Mitotic cell cycle

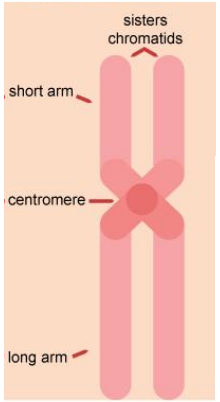
EXTRA NOTE

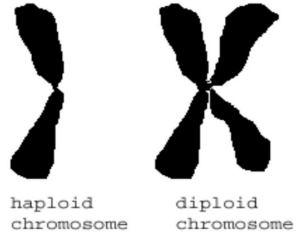
Chromosomes develop in mitotic phase (prophase)



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:
  - short arm (P arm)
  - long arm (Q arm)
- Each arm terminates in a telomere.





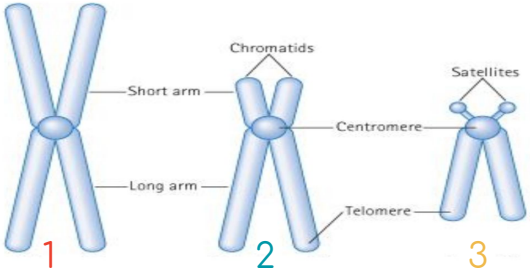
KEEP IN MIND

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

EXTRA NOTE

"Q" arm could never be shorter than the "P" arm

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:

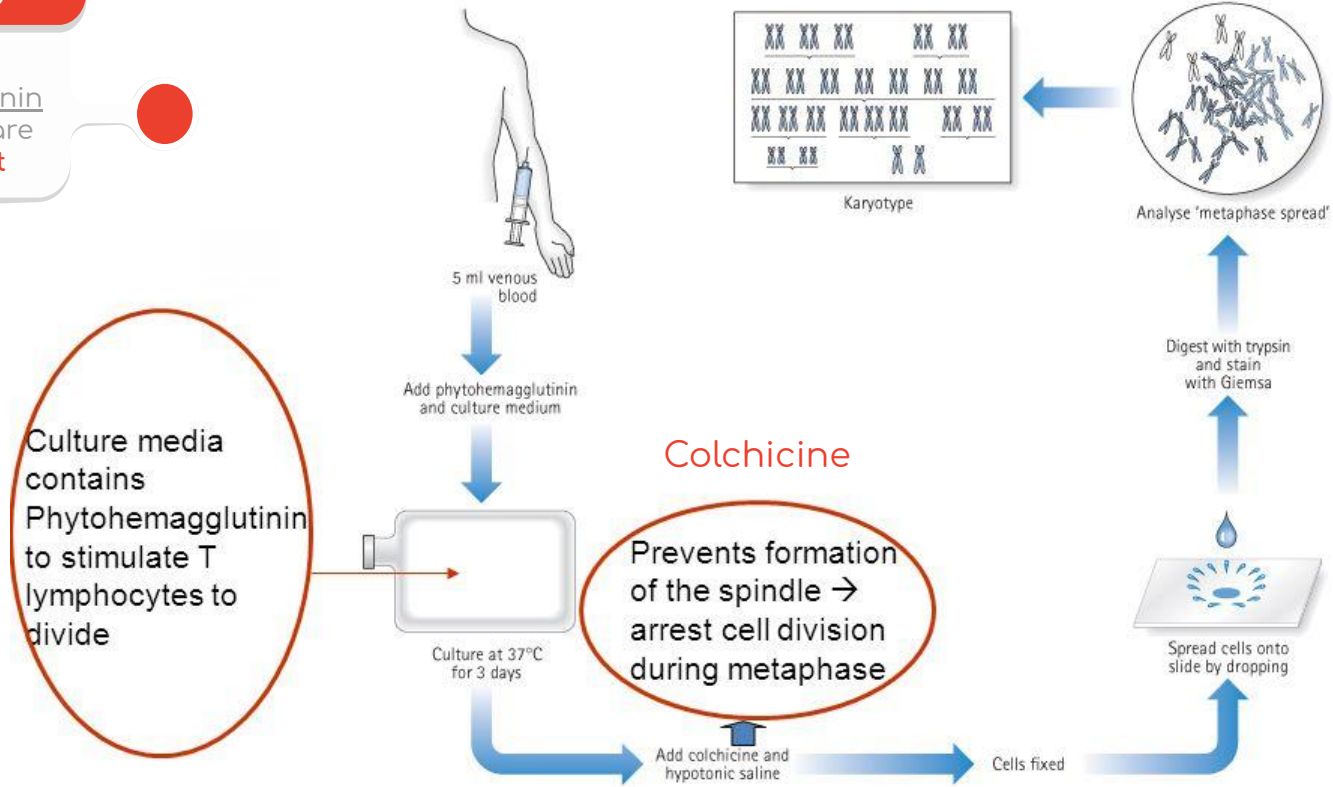
  1-metacentric : Has a longer arm.
  2-sub-metacentric : Has a shorter arm.
  3-acrocentric : Has no arm.

  - In the human karyotype chromosome pairs 13, 14, 15, 21, 22 are acrocentric.

CHROMOSOME PREPARATION FROM PERIPHERAL BLOOD

KEEP IN MIND

Functions of Phytohemagglutinin And Colchicine are very important



Items in the Description Of Karyotype

Normal Karyotypes

46, XY
46, XX

Abnormal Karyotypes

47,XY,+ 21 - Down Syndrome
45,X - Turner Syndrome

Team 439:

Down Syndrome: also called trisomy 21, is a genetic disorder caused by the presence of all or a part of a third copy of chromosome 21.

Turner Syndrome: is a genetic condition in which a female is partly or completely missing an X chromosome.

Karyotype

KEEP IN MIND

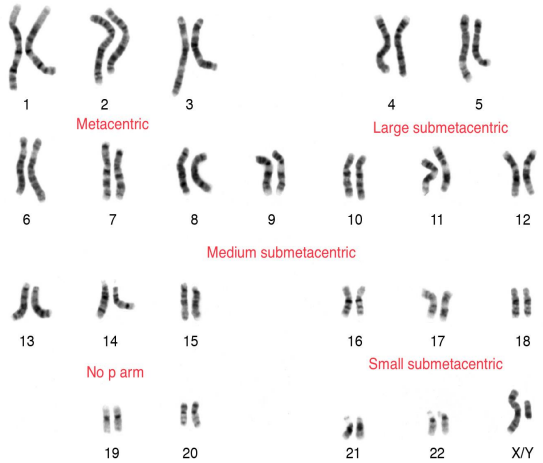
Bases of karyotyping are VERY IMPORTANT

steps involved in karyotyping:

- Culturing
- Harvesting
- Slide-Making
- Banding
- Staining
- Karyotyping
- Chromosome Analysis

Karyotyping is based on:

- The length
- The position of the centromere
- The presence or absence of satellites



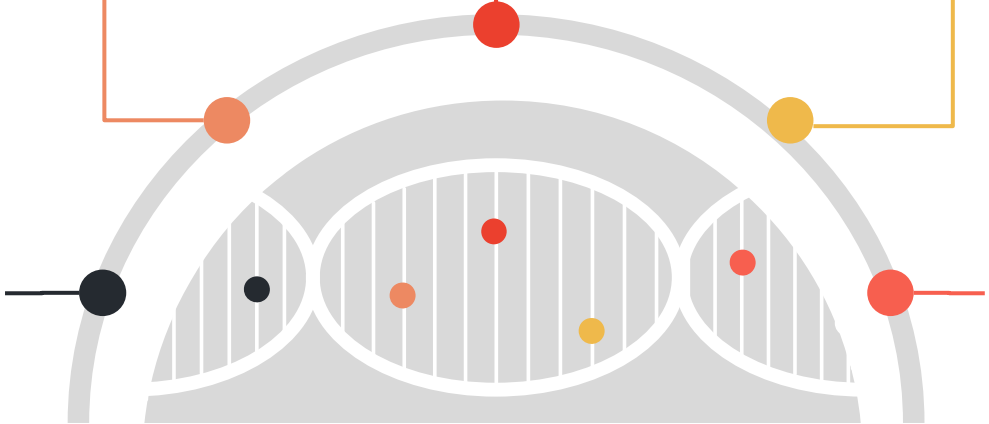
Banding

Allowing accurate identification and longitudinal mapping for locating gene positions and characterising structural changes.

Patterns are specific and repeatable for each chromosome.

Each arm presenting a sequence of dark and light bands.

Dark; non- active
Light ; active
R-bonding is the opposite



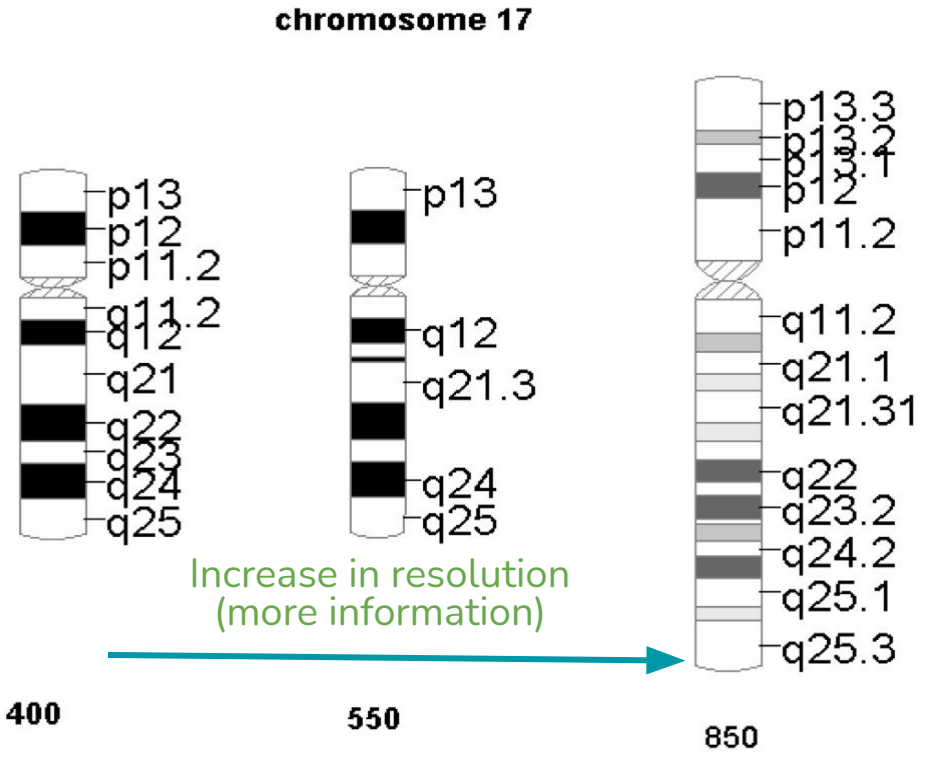
Patterns, and the nomenclature for defining positional mapping have been standardised.

Certain staining techniques cause the chromosomes to take on a banded appearance.

Chromosome Banding

Band resolution = estimate of number of light + dark bands per haploid set of chromosomes

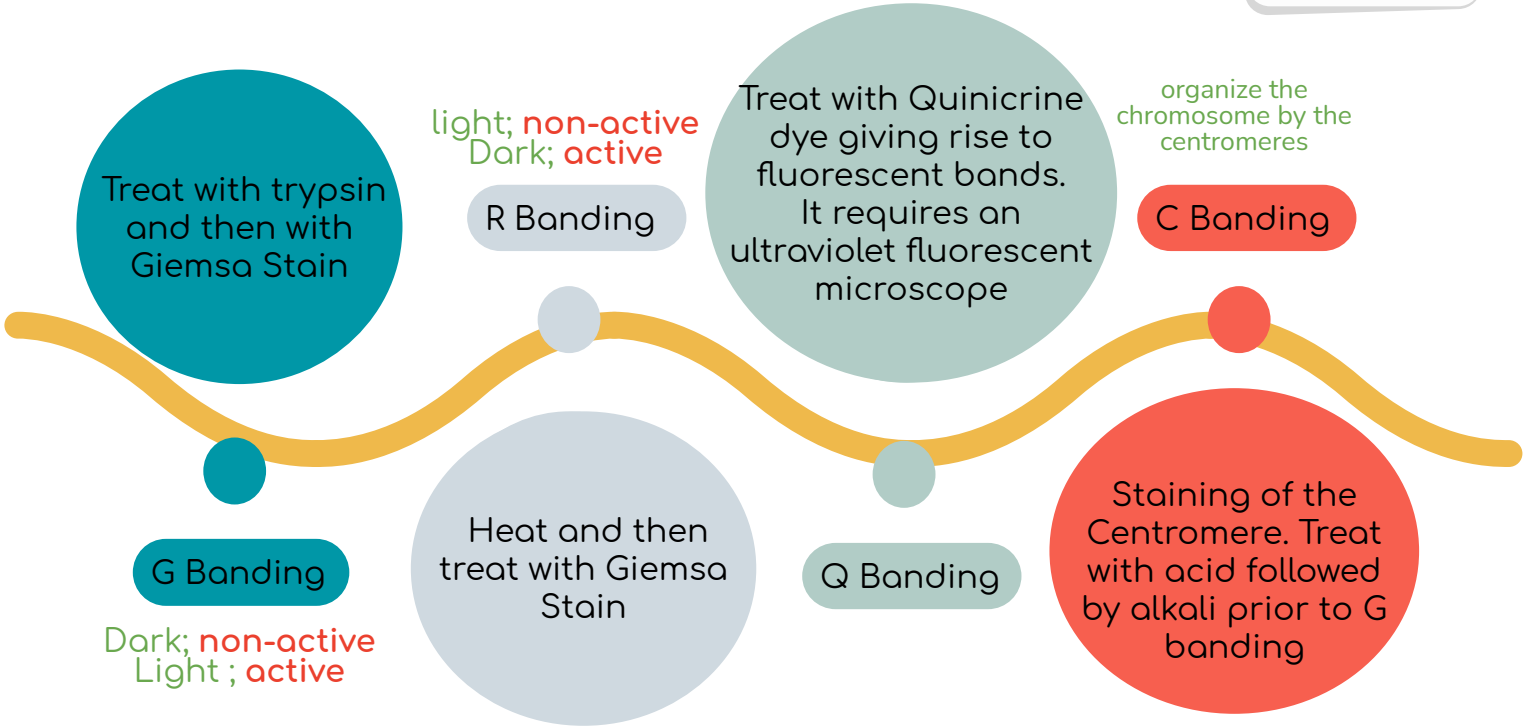
400 → 850+



Types of Banding

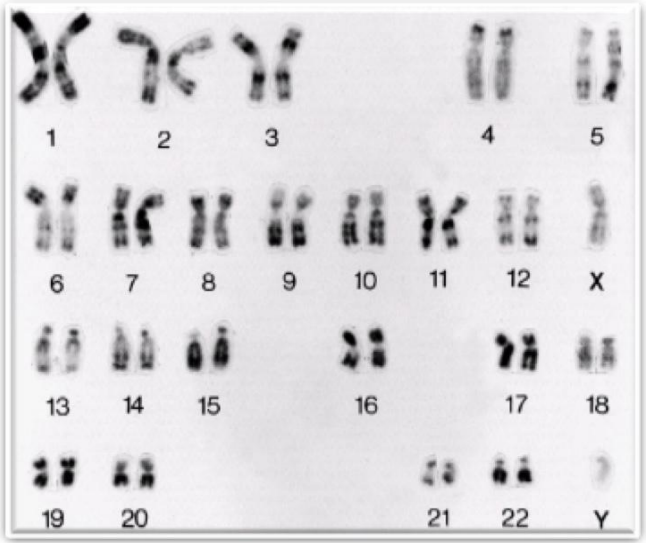
KEEP IN MIND

Types of Banding and banding in general are very important, Especially G-Banding.



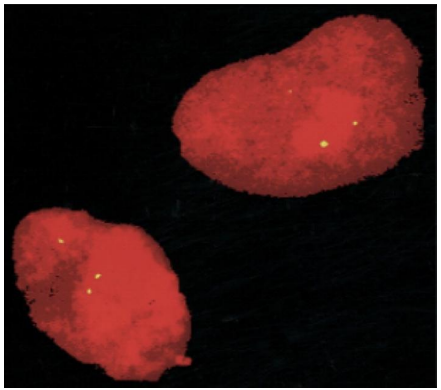
Banded Karyotype – Normal Banded Karyotypes

A normal G-banded male Karyotype

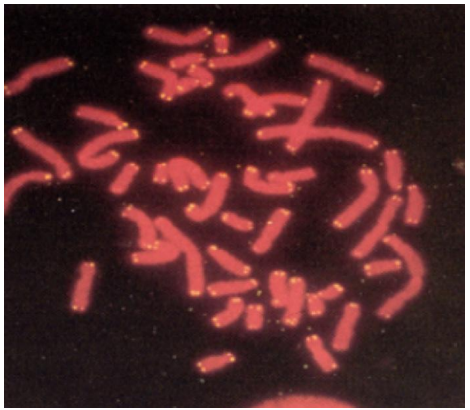


A normal R-banded male Karyotype

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

ببساطة: هذه التقنية تستخدم لتحديد وإظهار بروتينات أو مواد جينية
،أخرى ، ولكنها محدودة لإظهار جزء مرغوب معين
وهذه من السلبيات حيث لا نستطيع إكتشاف مرض محدد إلا بعد استخدام
التقنية المحددة لهذا المرض. أو الجزء المرغوب كيف؟
هذه العملية سريعة نسبياً وتكون عن طريق دمج أعواد مضيئة مصنفة،
مع شريط واحد من الذي أن أي
مثلاً لتحديد مناطق معينة أو مناطق حذف او تبديل سيتم شرح هذه
المناطق في المحاضرة القادمة
Team : 437

-in-situ= in its place, determines the chromosomes and its position Extremely accurate detection happens at the intermolecular level: finding the centromere of a chromosome.(Thanks 439)

It's like a biological clock of chromosome

Take home massage

- 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.

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MCQs

Q1: It stimulates T lymphocytes to divide:			
A.Phytohemagglutinin	B.Colchicine	C.Trypsin	D.Hypotonic saline
Q2: Karyotyping is based on:			
A.The length	B. The position of centromere	C.The presence or absence of satellites	D. All are correct
Q3: In which active parts of chromosomes are represented in white and inactive in black (dark)			
A. G banding	B. Q banding	C. R banding	D. C banding
Q4:The genotype of turner's syndrome is			
A.47,XY,+ 21	B.45,X	C.47,XXY	D.46,X

Answer Key: 1A - 2D - 3A - 4B

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Editing file

“Live as if you were to die tomorrow.
Learn as if you were to live forever.”

—Mahatma Gandhi