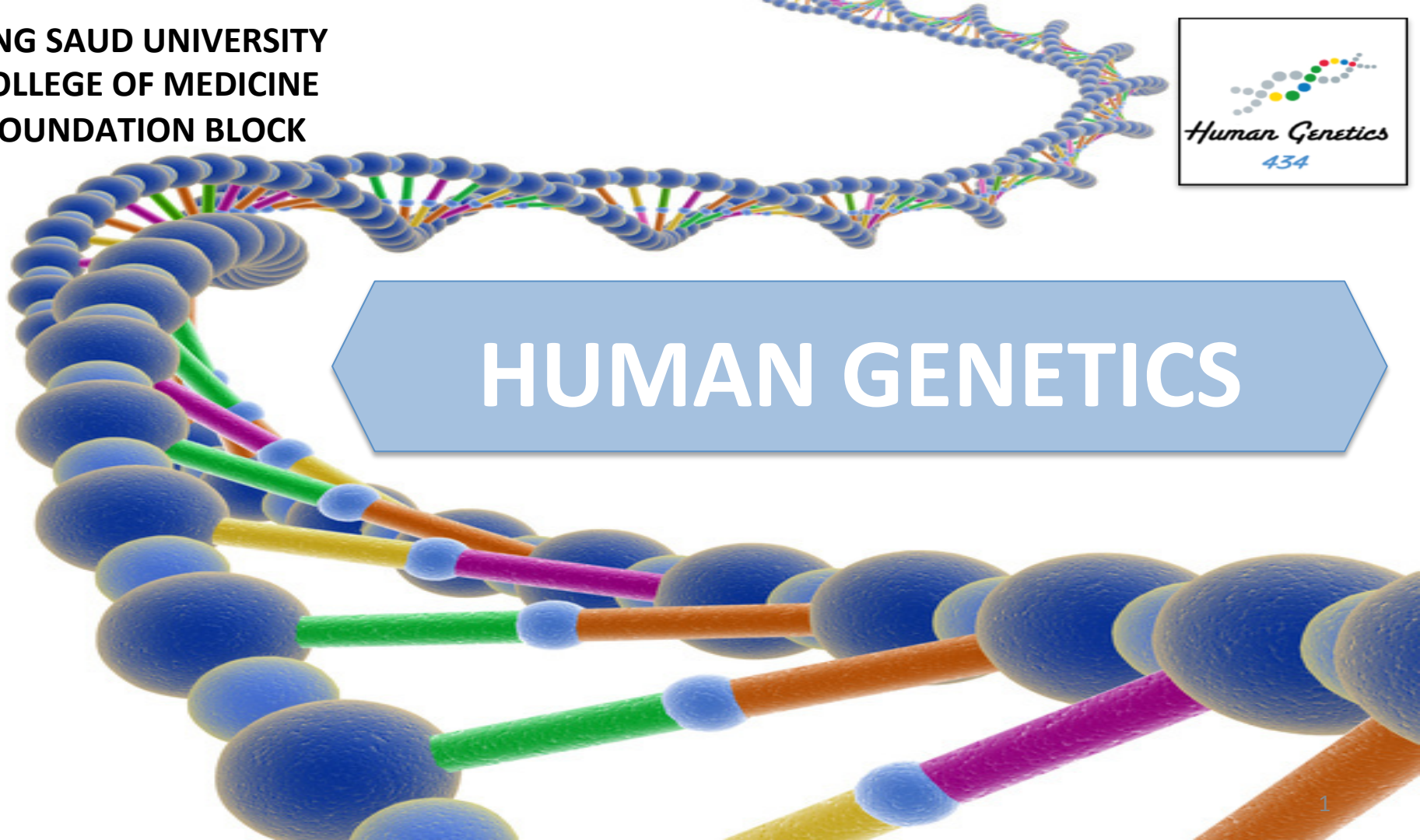
KING SAUD UNIVERSITY
COLLEGE OF MEDICINE
FOUNDATION BLOCK

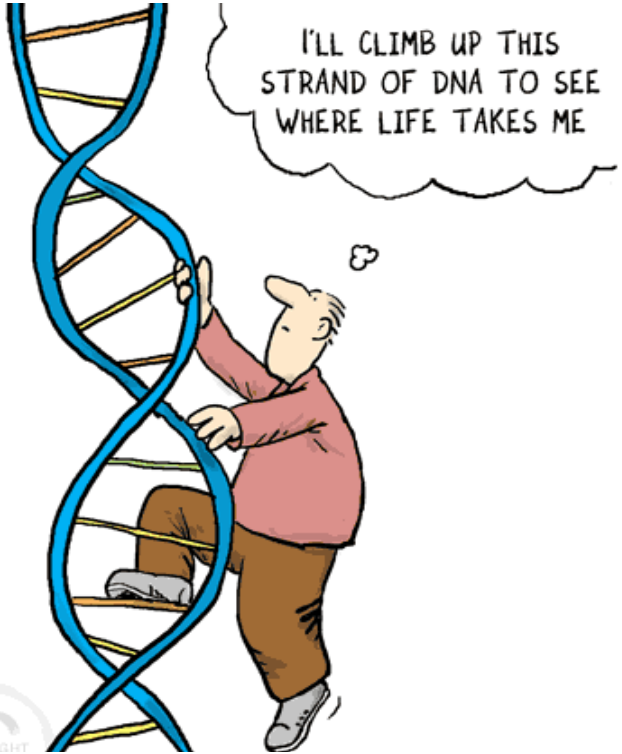
Human Genetics
434



HUMAN GENETICS

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.



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Gene expression:

All nucleotide cells have DNA, and they all have the same gene.

What makes a cell or a tissue different from other cells ?

Some portions of the DNA are active and the rest are inactive.

Note:

Each gene in the DNA makes a protein " if you open a gene → DNA sequence → group of nucleotides → different nitrogen base. "

Note:

* The sequence of nucleotides will differentiate between genes "make different proteins ".

* Making of the protein takes place in the cytoplasm.

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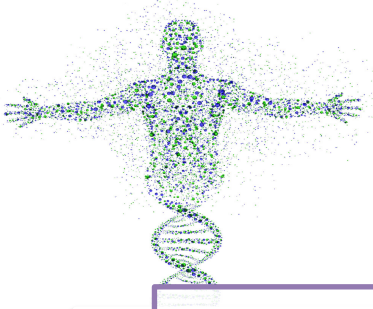

Genetics

Cytogenetics

The study of the structure and function of chromosomes and chromosome behavior during somatic (body related) and germ line (sperm or ova) division.

Human cytogenetic involves the study of human chromosomes in health and disease.

Molecular genetics

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

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Chromosomal studies are important in laboratory diagnostic procedures like:

1- Prenatal diagnosis “diagnose the embryo’s chromosome before birth“

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 patient with malignancies hematological disorders.

Note :

*New techniques allow for increased resolution.

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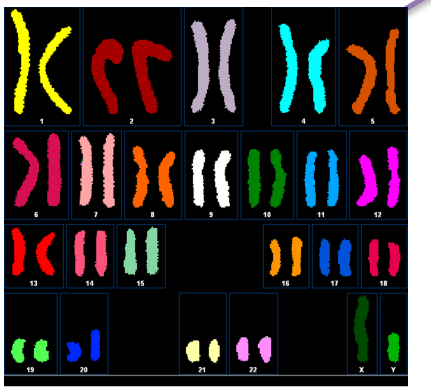
Spectral karyotype:

The preparation and study of karyotype is part of cytogenetic.

It contains 23 pairs of homologue chromosomes:



Karyotype: describe the chromosomes, attention is paid to their

- Presence or absence of Satellites
- Position of centromere
- Length

- One strand of the pair comes from the mother 'maternal' and one from the father 'paternal'
- The intact set is passed to each daughter cell at every mitosis.

YouTube For Further explanation
http://www.youtube.com/watch?v=JGTbVvVCXQo

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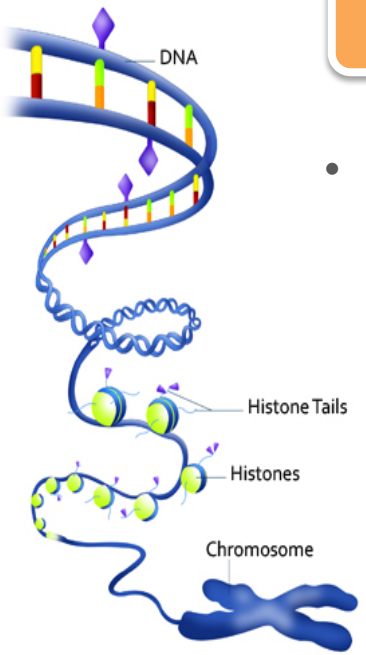
Structure of Chromosomes



- The packaging of DNA into chromosomes involves several orders of DNA coiling and folding.

* Primary coiling: DNA double helix

* Secondary coiling: around histones (basic proteins)→ nucleosomes

* Tertiary coiling → chromatin fiber

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

YouTube For Further explanation

http://www.youtube.com/watch?v=gbSIBhFwQ4s

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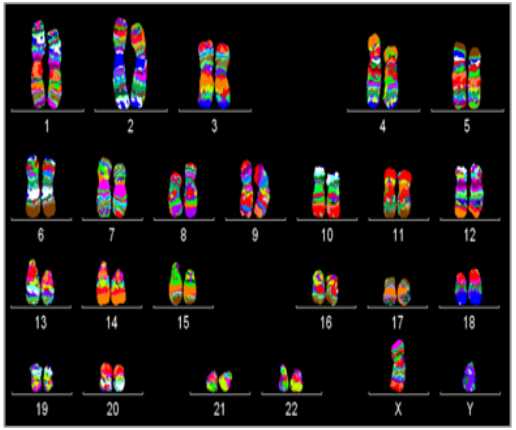
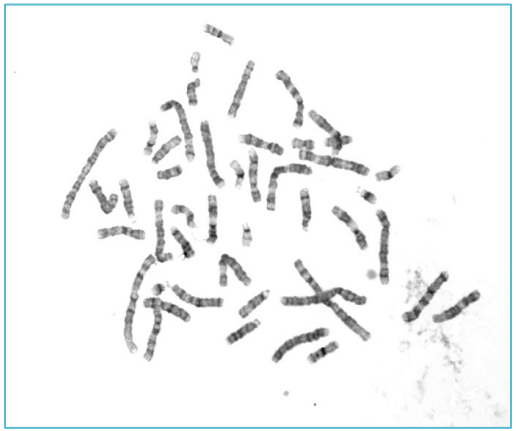
Cytogenetics :

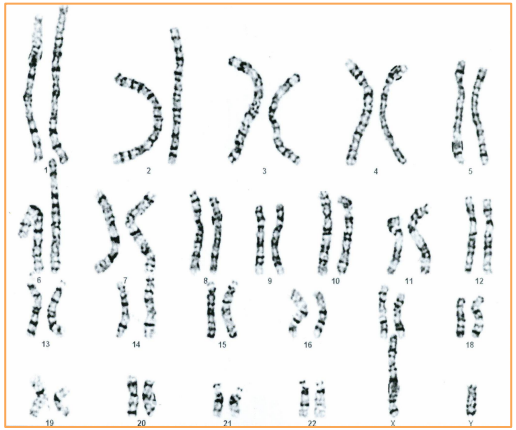
High resolution Karyotype



Non-Banded Karyotype



Banded Karyotype



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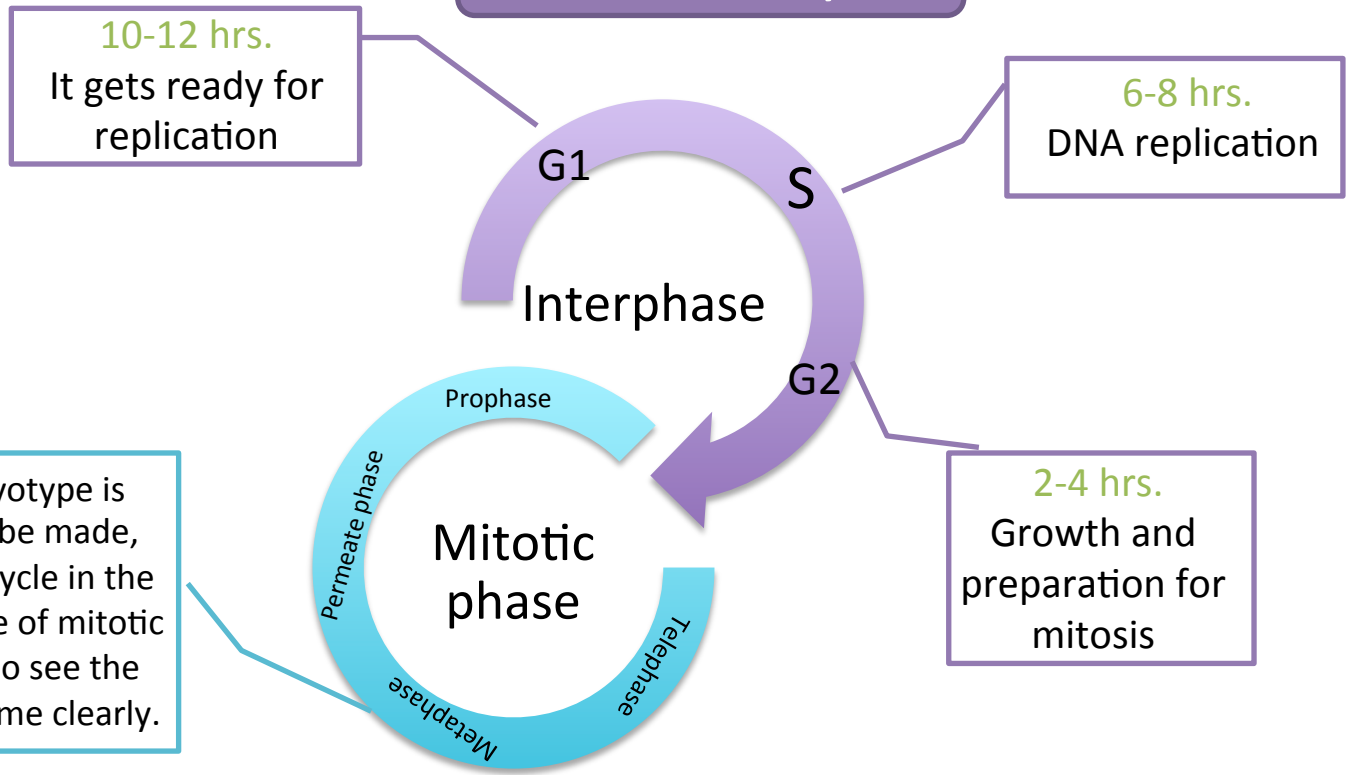
Mitotic cell cycle:



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Procedure of Chromosome Preparation from Peripheral Blood

A series of steps involved :

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1 Culturing

" Culture media contains Phytohemagglutinin to stimulate T lymphocytes to divide. "
Culture at >> 37C for 3 days

2 Harvesting

add colchicine and Hypotonic saline
" Prevents formation "
of the spindle
>>arrest cell division during metaphase.

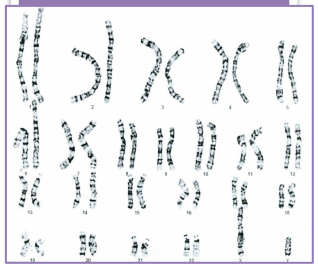
3 Slide-Making

"G banding"
Add trypsin

4 Analyze

"staining"
With Geimsa

5 Karyotype



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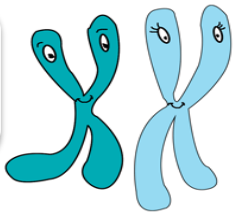
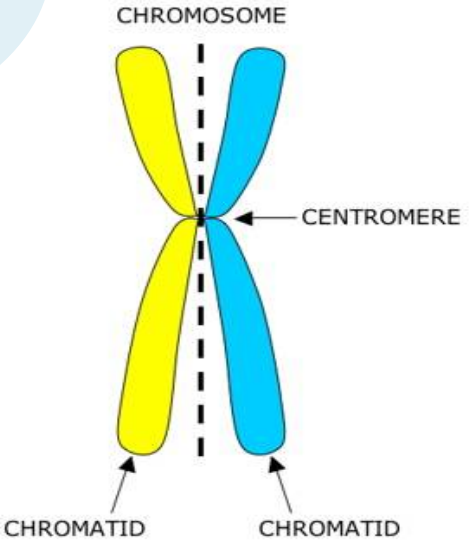
CHROMOSOMES



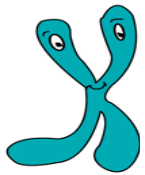
A threadlike structure of nucleic acid and protein "histone" found in the nucleus of most living cells (22 pair + sex chromosomes).

* They carry genetic material

Each chromosome has a centromere (CEN), region which contains the kinetochore



CHROMATIDS:



During cell division each DNA strand is duplicated, and the chromosomes condense to become visible as distinct pairs of chromatid (or sister chromatid) joined at the centromere. (metaphase)

Note :

- 22 pairs of autosome
- 1 pair of sex (by order of decreasing end) (largest to smallest)
- chromosomes [XX female, XY male]

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The ration of the length of the two arms is constant for each chromosome.

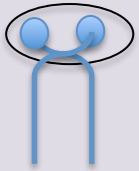
Centromeric Position and arm length

3 types of chromosomes based on the location of the centromeres:

- Meta centric
- sub-meta centric
- acrocentric

CEN divides the chromosome into two arms:
the short arm (p arm) and the long arm (q arm).
Each arm terminates in a telomere.





satellite

In human: karyotype chromosomes pairs 13,14,15,21,22 are acrocentric.

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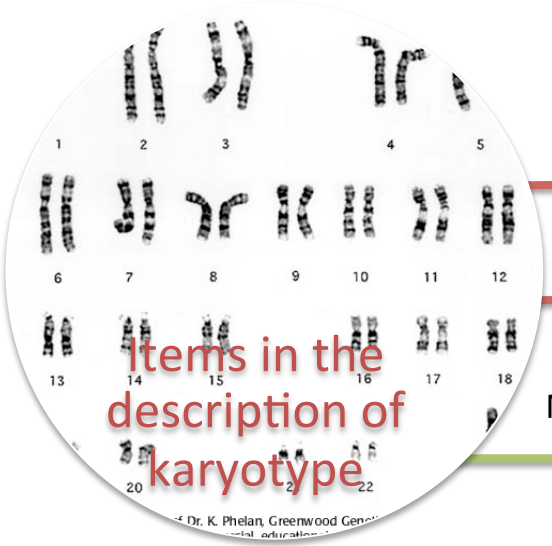
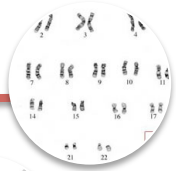
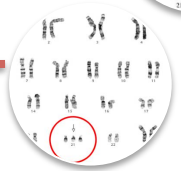


Abnormal



45 XY, T(D;G)

Abnormal



47, XY + 21

[Down syndrome]

Normal

46, XX

46, XY

T = Translocation

In the karyotype: the X chromosome is larger than the Y chromosome.

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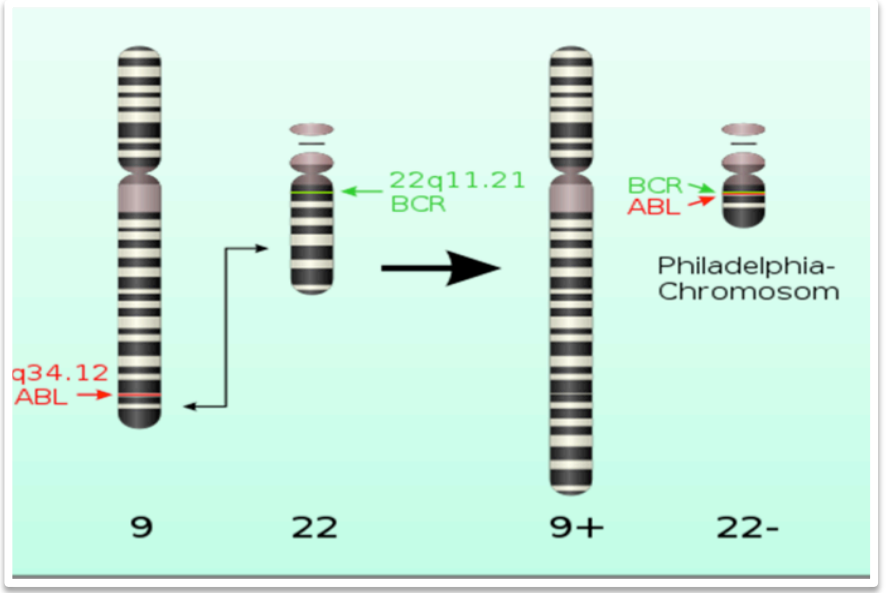
Philadelphia – Chromosome Translocation

*Critical genetic event in the development of CHL(Chronic Myelogenous Leukemia)

*Due to a cytogenetic abnormality

(translocation between 9 and 22)



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Banding

Certain staining techniques cause the chromosome 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 characterizing structural changes.

Patterns, and the nomenclature for defining positional mapping have been standardized. (All Around The World)

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Chromosome Banding

G Banding	R Banding	Q Banding	C Banding
" Standard " - Treat with trypsin and then with Geimsa Stain.	" Reverse Banding " - Heat and then treat with Geimsa Stain.	- Treat with Quinicrine dye giving rise to fluorescent bands. It requires an ultraviolet fluorescent microscope	- Only staining of the centromere. Treat with acid followed by alkali prior to G banding.

Band resolution =

estimate of number of light + dark bands per haploid set of chromosomes

" More Numbers >> More Resolution. "

" 400 >> 550 >> 850+ ".

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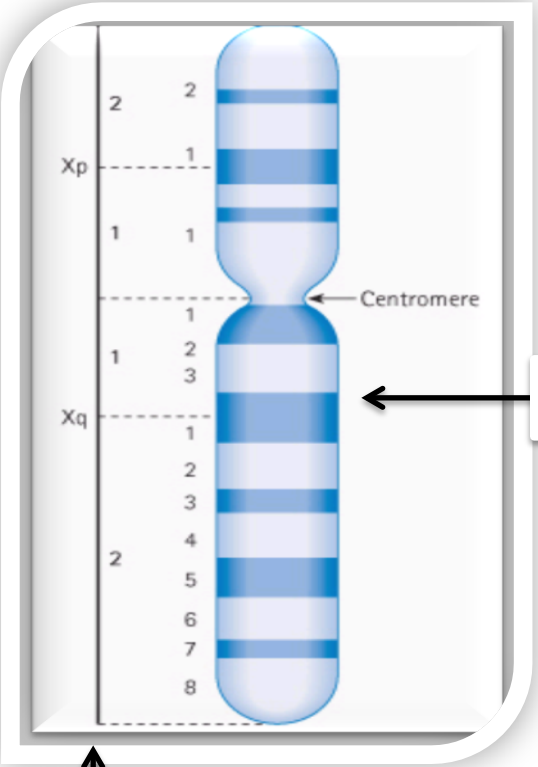
Nomenclature



An X chromosome showing the short and long arms each subdivided into regions & bands.

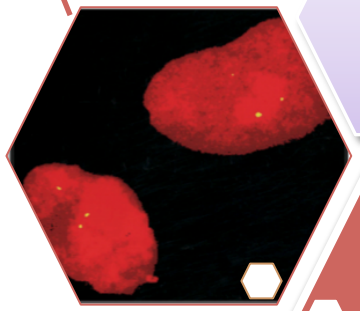
Ex. 9q13.14

Chromosome #9

Large arm

Fluorescence In-Situ Hybridization (FISH)

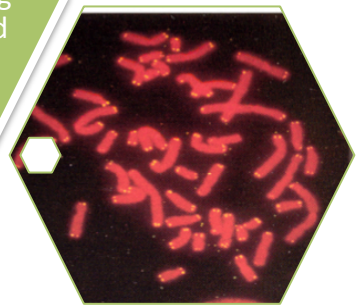
Using fluorescent probe that binds with its complementary target sequence.



They can be used to study chromosomes in metaphase or interphase.

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.



Molecular cytogenetic technique used to detect and localize the presence of a specific DNA.

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MCQS

Quiz yourself!

https://www.examtime.com/p/1378928

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