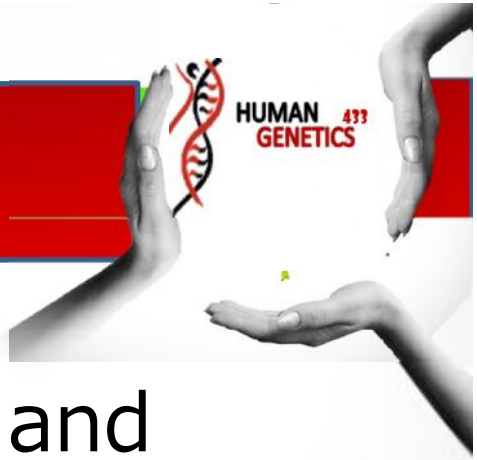
KING SAUD UNIVERSITY
COLLEGE OF MEDICINE
FOUNDATION BLOCK

Human Chromosomes: Genotype/Phenotype

HUMAN GENETICS 433

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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"Genetics is all about showcasing human beauty along with high-quality performance."

Bela Karolyi

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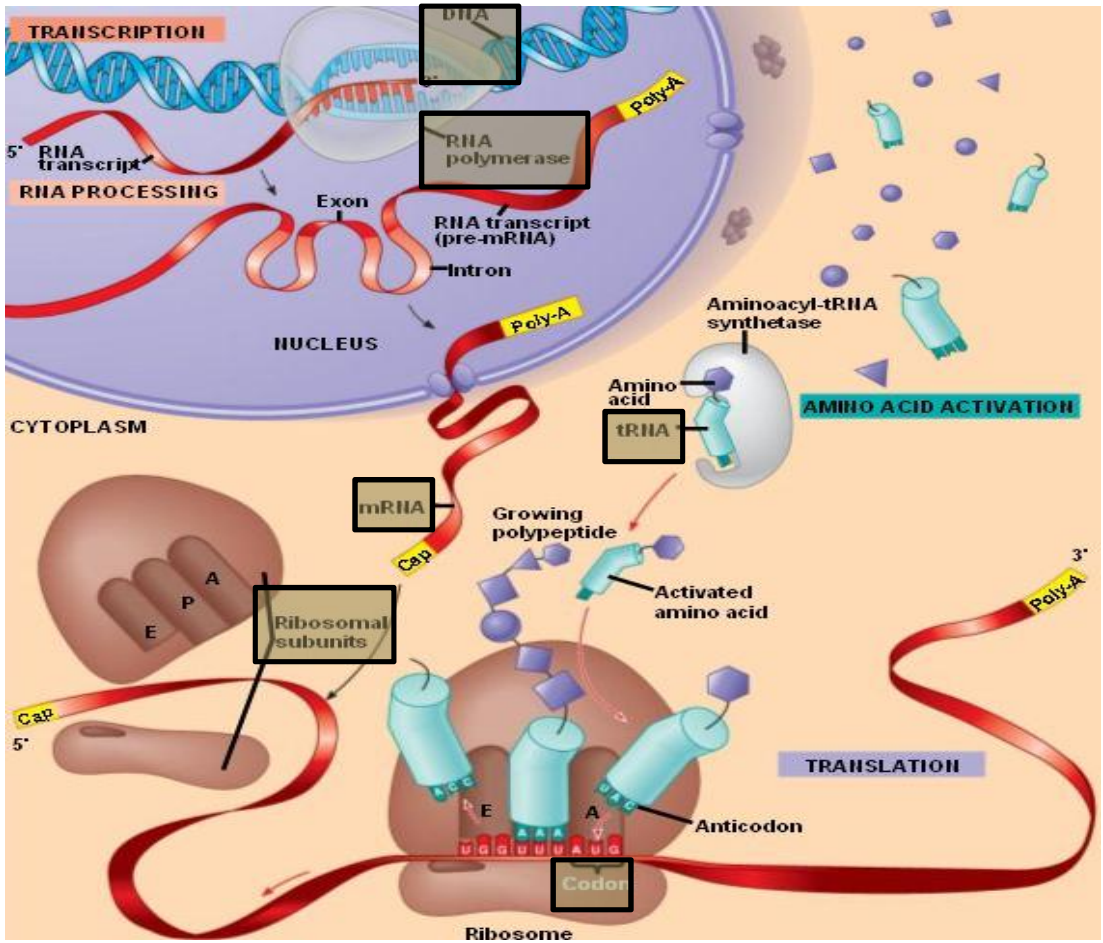
Gene Expression

For a cell to make a protein, the information from a gene is copied, base by base, from a strand of DNA into a strand of messenger RNA. Messenger RNA travels out of the nucleus into the cytoplasm, to cell organelles called ribosomes. There, tansfer RNA directs the assembly of amino acids that fold into a completed protein molecule.



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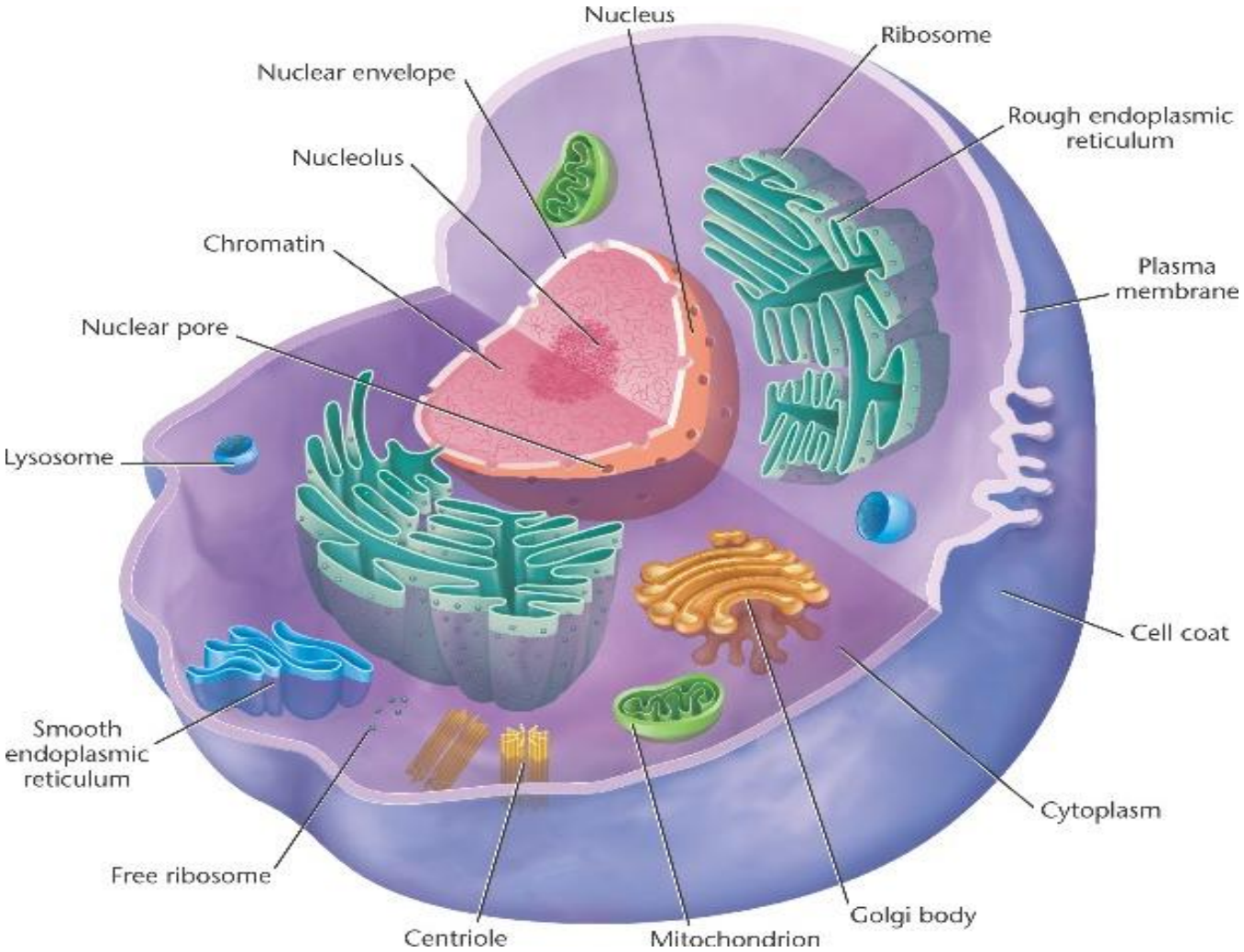
Eukaryotic cell



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GENETICS :

Cytogenetics:

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

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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Comparison of Related Diseases

Human Cytogenetics	Molecular Genetics
Turner's syndrome	Sickle Cell Anemia
Down's syndrom	Hemophillia A
Kleinfelter's syndrome	Colour Blindness

***N.B. Human cytogenetics related to diseases caused by abnormal chromosomal conditions, while molecular genetics is related to diseases caused by mutated genes.**

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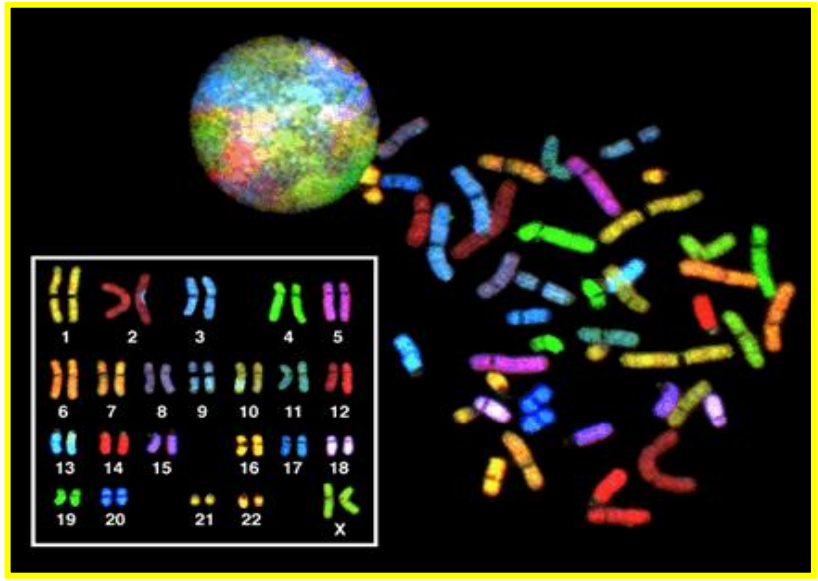
Cytogenetics:

Human Cytogenetics:

Involves the study of human chromosomes in health and disease.

Karyotype:

An arrangement that classifies the chromosomes of an organism based on the number, size, shape, and other characteristics of the chromosomes.



This illustration represents a spectral karyotype, named so for the light marks that identify each chromosome.

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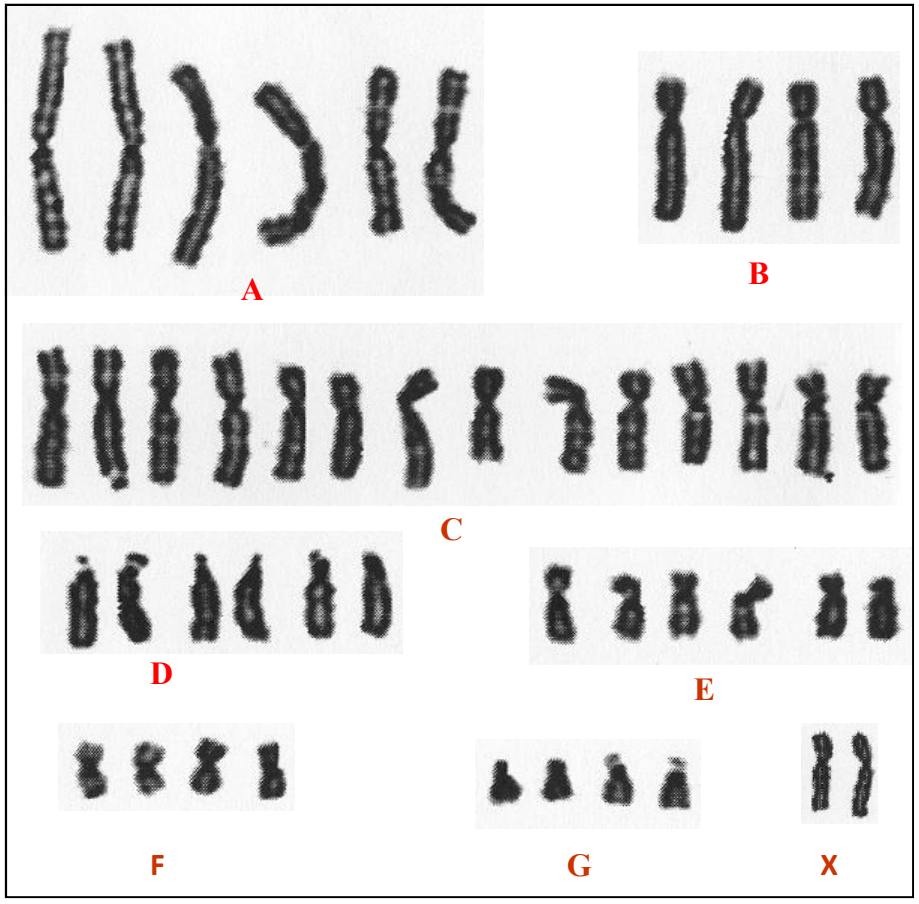
Karyotyping

Based on:

1.the length.

2.the position of the centromere.

3.the presence or absence of satellites.



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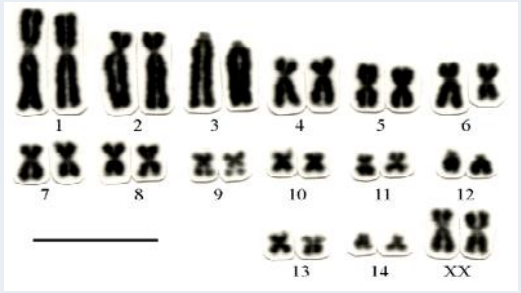
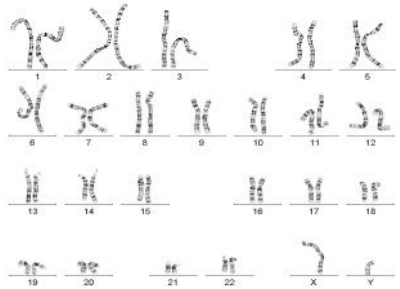
Type	Illustration
Non-Banded Karyotype	
Banded Karyotype	
High Resolution Karyotype	

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Items in the Description Of Karyotype:

- **Normal Karyotypes**

  46, XY
  46, XX

- **Abnormal Karyotypes**

  47, XY, + 21 → **Down Syndrome**

  45, XO, → **Turner Syndrome**

***47,XY , 21 (down syndrome) is due the increase of number in chromosomes " in males "**
***45, XO, (turner syndrome) one X is missing here which causes the abnormality " in females "**

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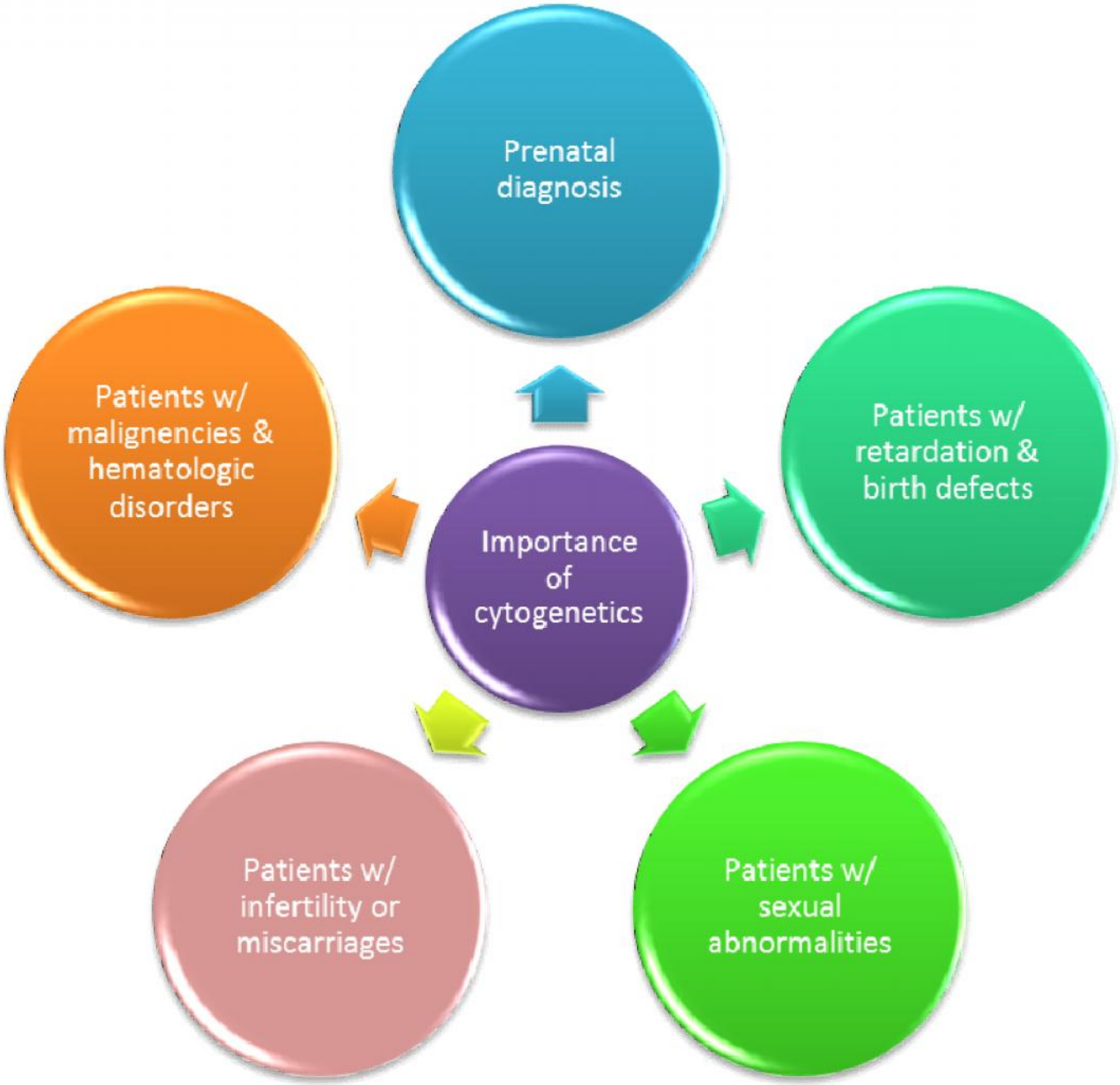

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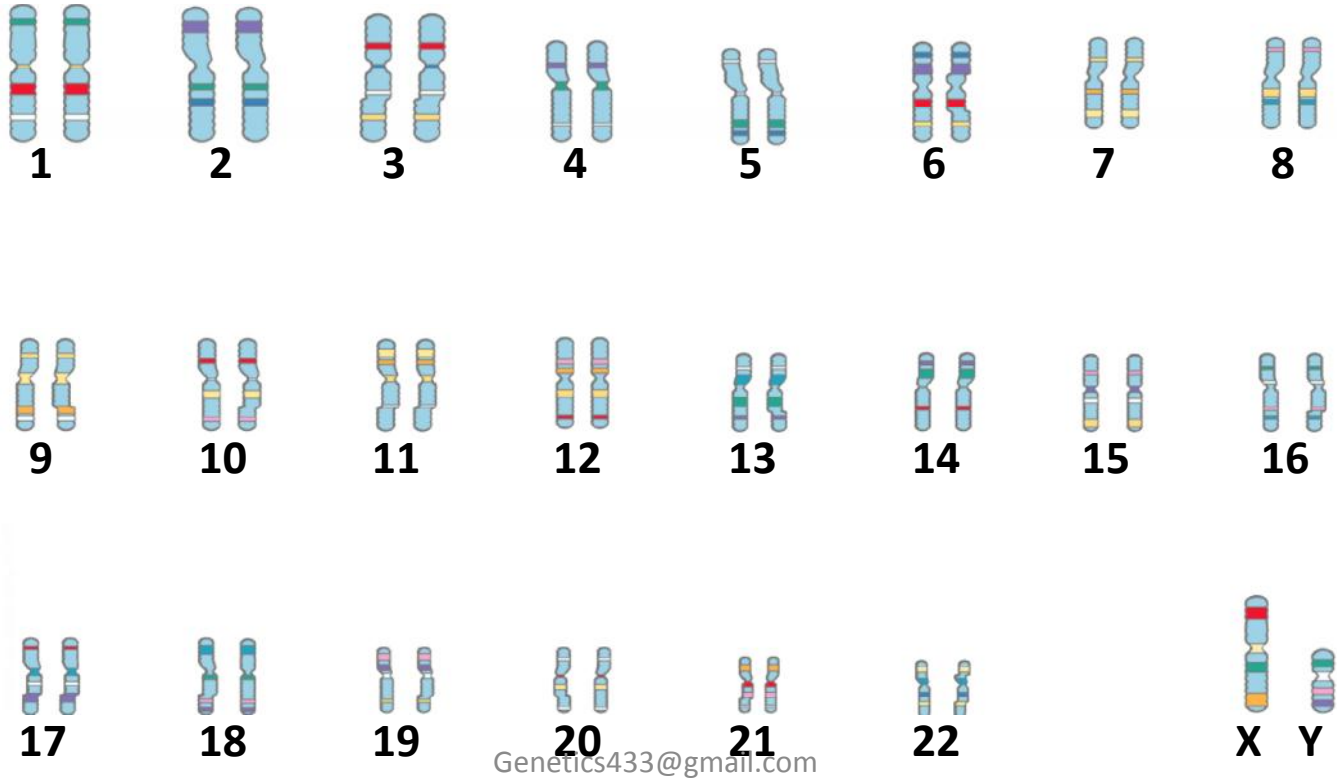
Application of Cytogenetics in Laboratory Diagnostic Procedures

Type of Application	Example/notes
Prenatal Diagnosis	Sample can be taken from • Amniotic fluid • Chorionic villi • Cord blood
Mental Retardation	_____
Birth Defects	_____
Abnormal Sexual Development	Kleinfelte's syndrome Turner's Syndrome
Infertility/Miscarriages	_____
Hematologic Disorders	Malignancies: Leukemia Anemia: Sickle Cell Anemia

Human Chromosomes

-Human cells contain two sets of chromosomes, one inherited from the mother and one from the father.

-Each set has 23 single chromosomes--22 autosomes and a sex-determining chromosome, either X or Y. The set shown here is from a male, since it contains an X and a Y chromosome; if the chromosome set were from a female, it would contain an X and an X.



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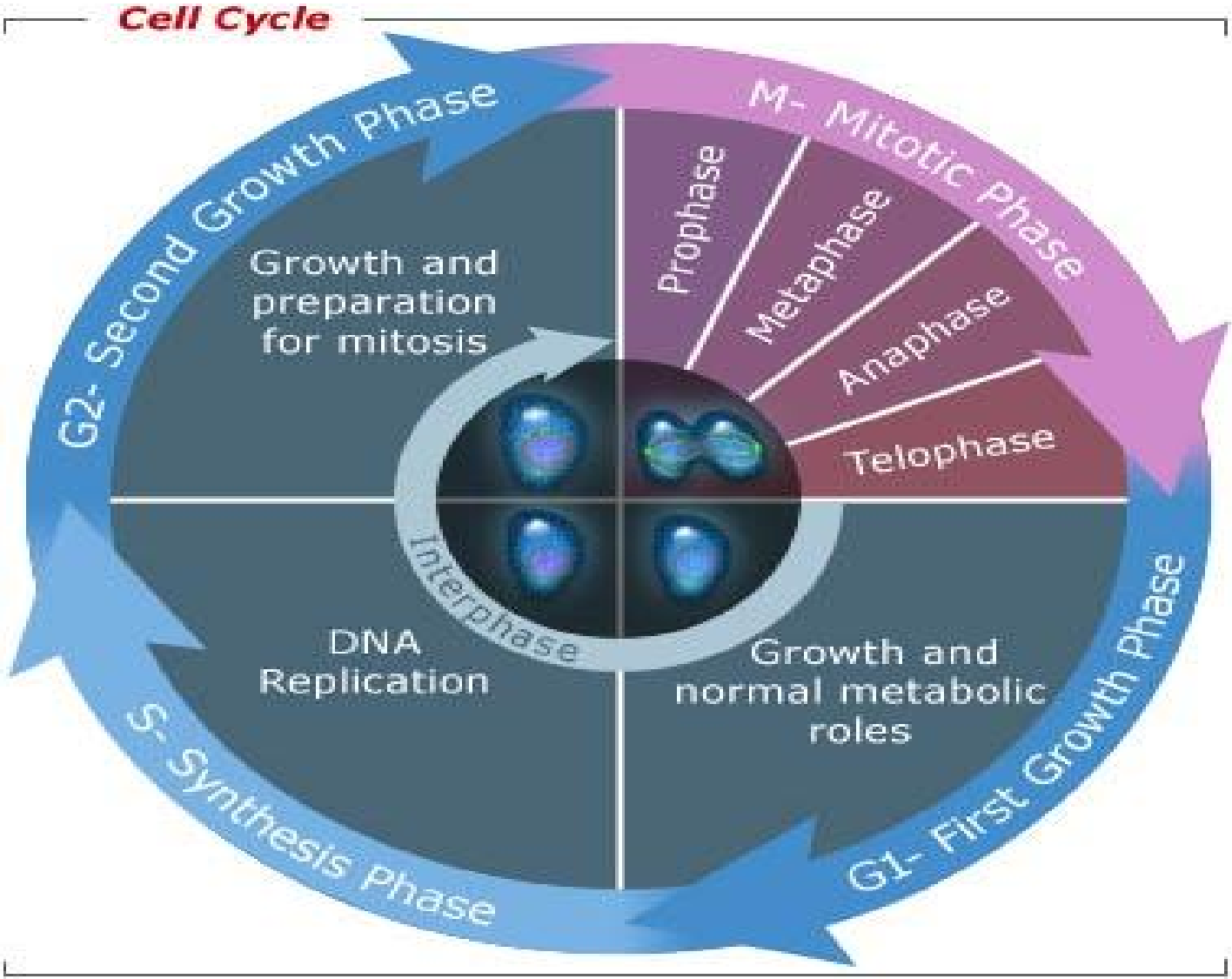
Mitotic cell cycle



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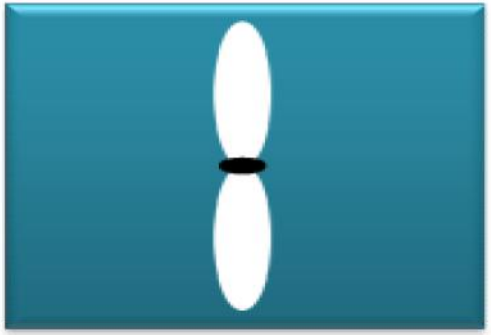
Chromosome Morphology

Metacentric

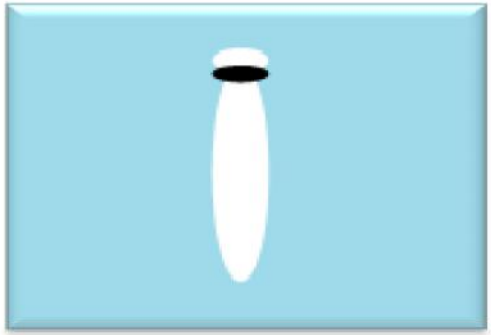


- Centromere: ***in*** the centre
- Divides into: two equel arms

Submetacentric

- Centromere: ***near*** the center
- Divides into: long arm and short arm

Acrocentric



- Centromere: very close to telomere
- Divides into: very small arm (satellite)and long arm

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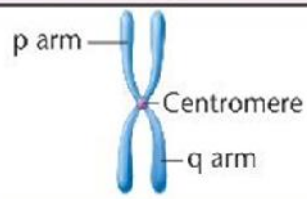
Centromeric position and arm length:

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:
i-*metacentric* ii-*sub-metacentric* iii-*acrocentric*

Centromere location	Designation	Metaphase shape
Middle	Metacentric	
Between middle and end	Submetacentric	
Close to end	Acrocentric	

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

NOTE : **Karyotype (def) : is the arrangement of chromosomes in size and shape

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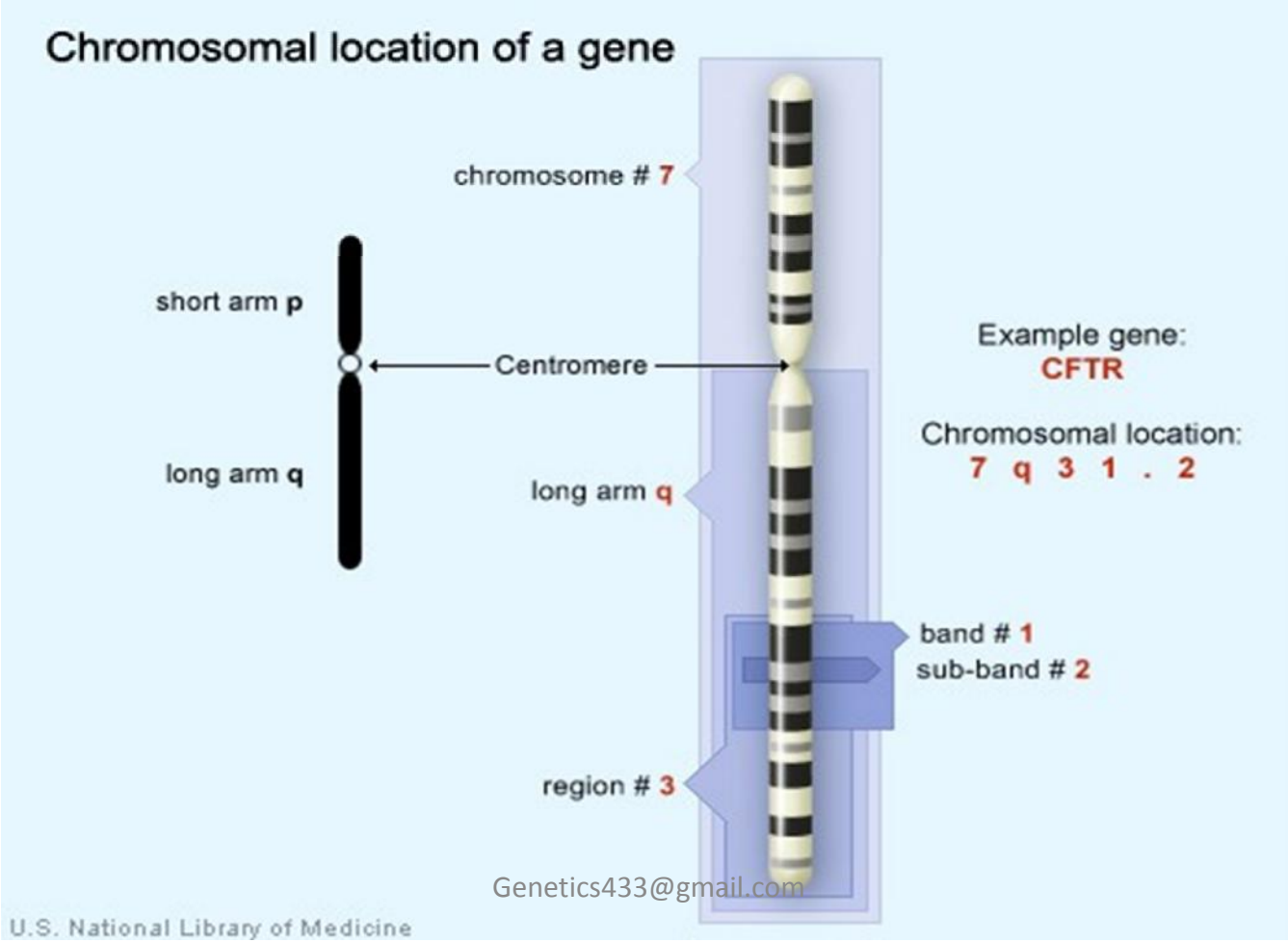
Defining Chromosomal Location

How to read and name a specific chromosome : "7" the chromosome number , Q is the arm's letter, 3 is the region, 1 is the band, the last number is the subband.
So the Overall definition of this gene's position is 17q31.2



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How to describe a gene's location?

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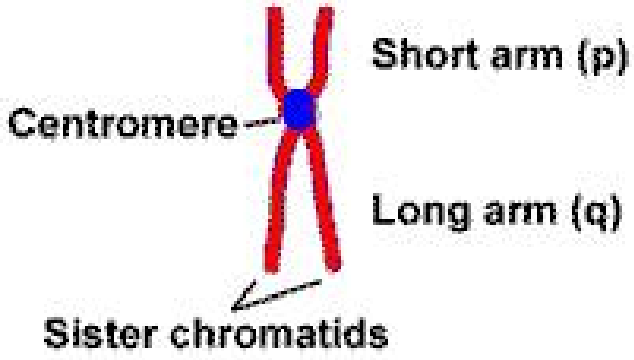
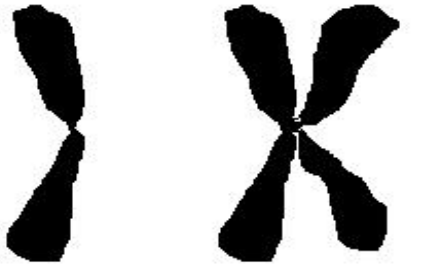
Metaphase chromosomes:



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



- 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.

NOTE : *Here * You have to know the structure of the chromosome

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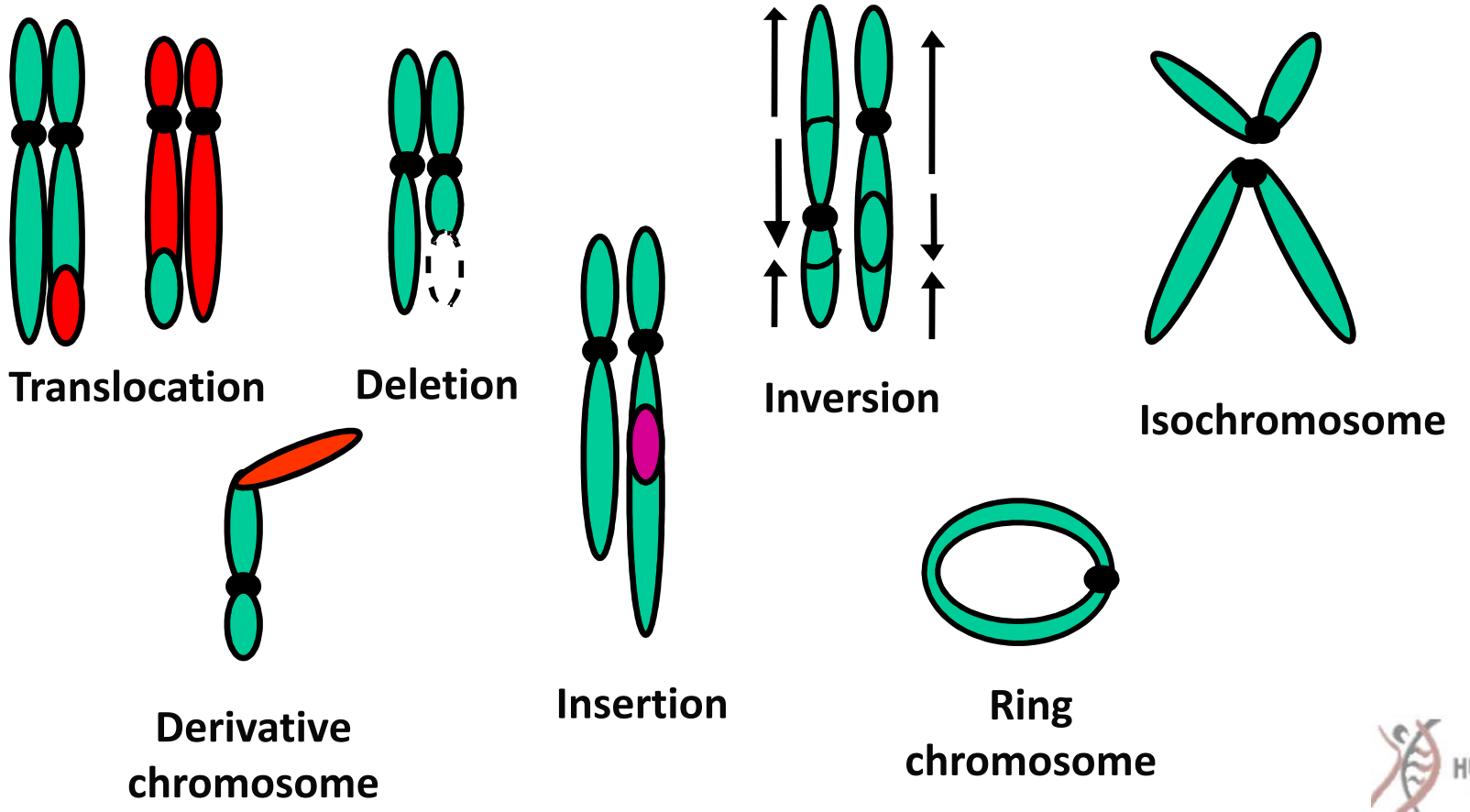
Chromosome Structure Abnormalities



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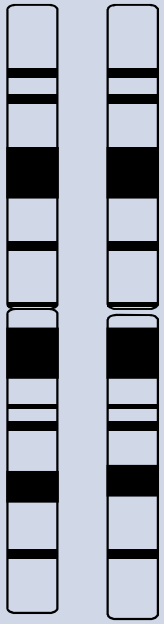
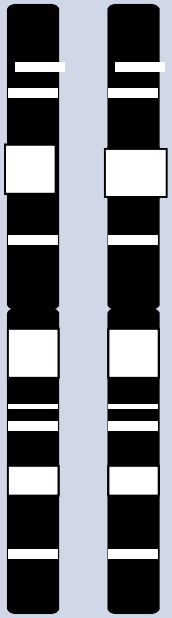
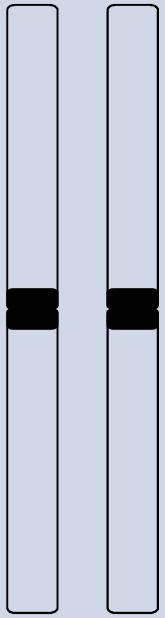
Visualizing Metaphase Chromosomes (Banding)

G-Bands (Giemsa)	R-Bands (Reverse)	C-Bands (Centromere)
		
A type of stains	**stains regions that were not stained by giemsa**	**Stains the centromere region.**

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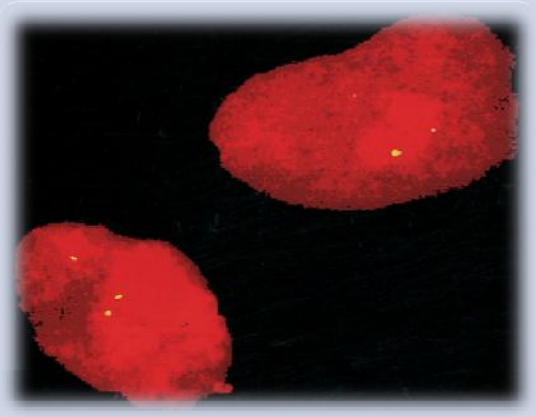
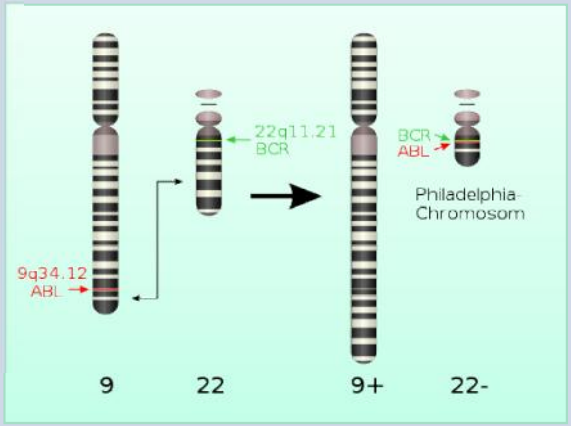
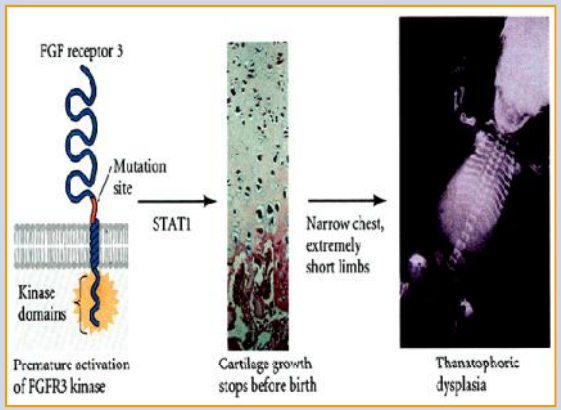
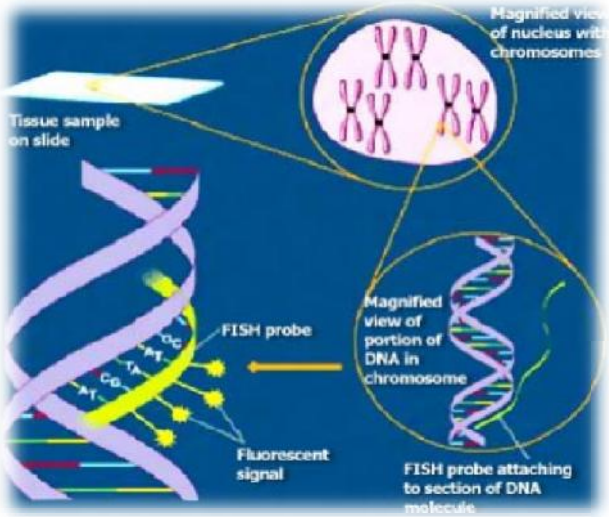
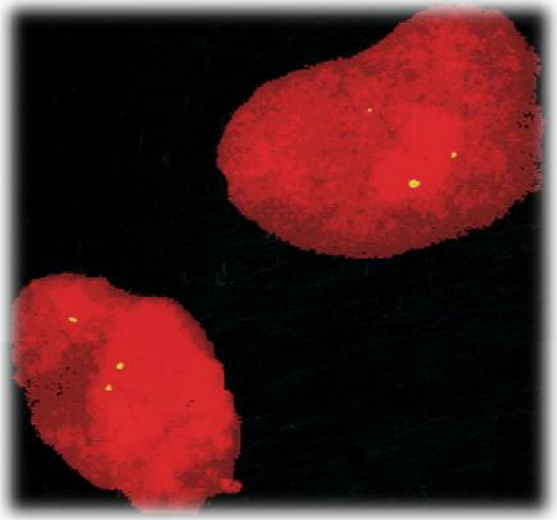
Chromosome 21 trisomy (Down's Syndrome)	Philadephia's Chromosome	Achondroplasia
		
Caused by an a extra copy of chromsome 21	**Caused by a translocation between chromosome 9 and 22**	**Caused by a dominant mutaion in FGF receptor 3.**
47 XX+21	**This translocation can be described as:** **<u>46, XY, t(9;22)(q34;q11)</u>**	**FGF (Fibroblast growth factor)-instruct cells not to divide**

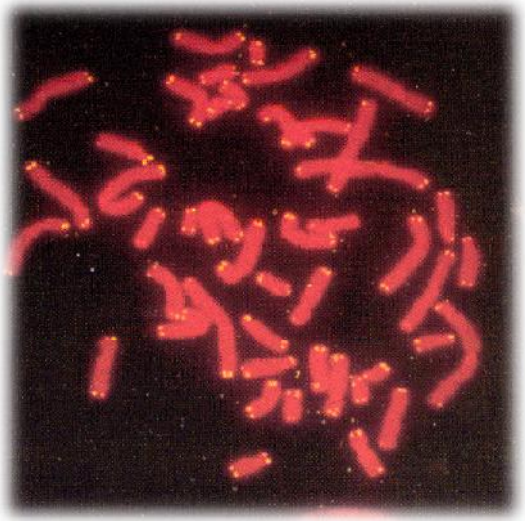
Fluorescence In-Situ Hybridization (FISH)



Hybridization of complementary gene- or region-specific fluorescent probes to chromosomes. Thereby identifying gene or region.



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.

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MCQs

- **The chromosomal number is 17 , region 2 , short arm , band 1 and 2 sub band. What is the formula of the chromosomal locati on?**

  A)17p21.2 **B)17q21.2**

  C) 17p22.1 **D) 17q22.1**

- **The morphologic type which presents satellite is:**

  A) Metacentric **B) submetacentric**

  C) acrocentric

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MCQs

- What do we called the chromosome 22 when it had translocat ion with 9 chromosome?

  A) Isochromosome B) Philadelphia chromosome

  C) X chromosome D) ring chromosome

- When the chromosome loses one of it's arms and replaces it with an exact copy of the other arm, this abnormal structure called:

  A) Inversion B) translocation

  C) derivative chromosome D) isochromosome

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MCQs

- **Down syndrome can be simplified as:**
  A) XY+21 B) X0
  C) XX D) XY+23

- **The two sister chromatids are held together by:**
  A) Centrioles B) Centromeres
  C) Kinetochores D) Chromosomes

- **Karyotoping is based on:**
  A) Width of the chromosomes B) The number of genes
  C) Centromere position D) Both A and B

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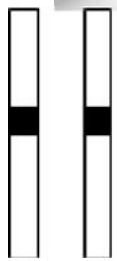
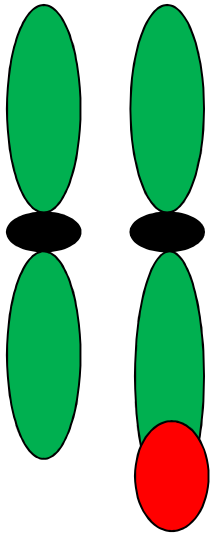
MCQs

- **These bands of chromosomes are:**
  - **A) Giemsa bands B) Reverse bands**
  - **C) Normal bands D) Centromere stained**



- **The topic that focuses on the study of the structure and function of chromosomes is:**
  - **A) Molecular Genetics B) Histology**
  - **C) Cytogenetics D) Microbiology**

- **What phrase describes this abnormality:**
  - **A) Translocation B) Deletion**
  - **C) Inversion D) insertion**



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