CHROMOSOMES

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Genetics

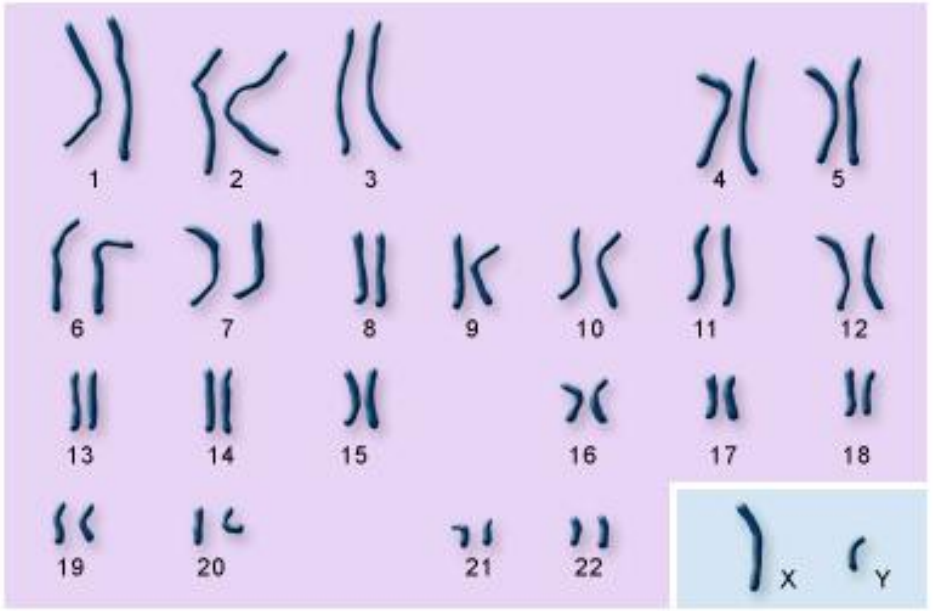
- Genetics is derived from Greek word “gennan”, which means ‘to generate’.

- Genetics is a branch of science dealing with the study of heredity or inheritance and variations.

- It provides a basis for understanding the fundamental make-up of an organism, thus leading to a better understanding of a disease process.

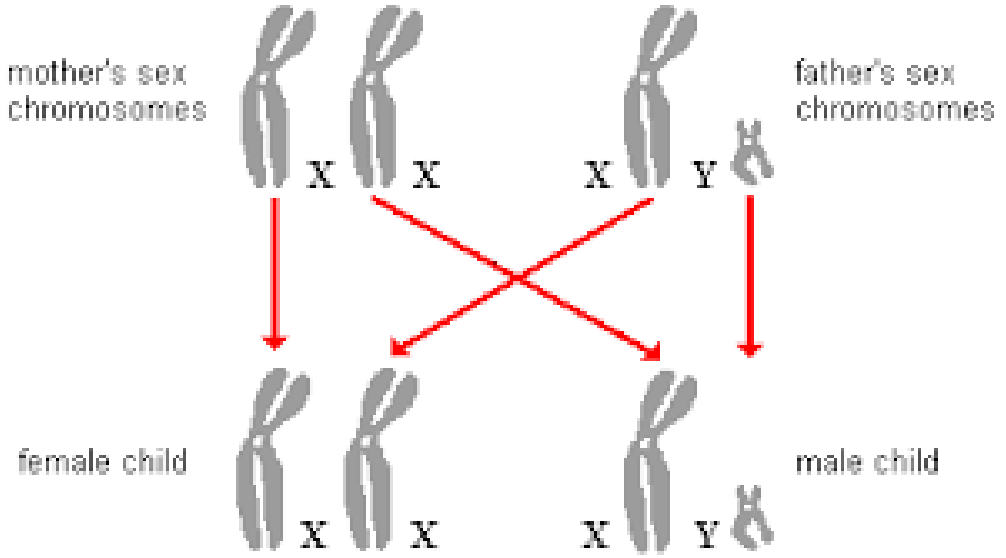
Chromosome

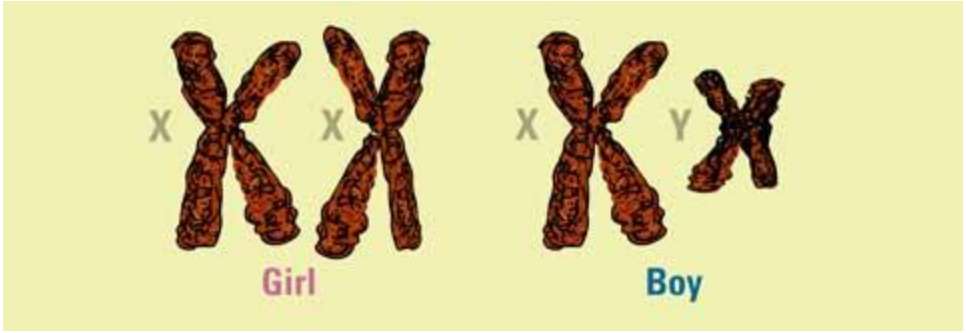
- Genes, the unit of inheritance are located on the chromosomes of the gametes.
- Passed from parents to progeny.
- Chromosome term was introduced by Waldeyer in 1888.
- Number of chromosomes is fixed in each cell but varies from species to species.
- In human beings, the total number is 46 or 23 pairs named as ‘diploid’ (2n).
- While in gametes (male or female) the number is 23 i.e. ‘haploid’ (n).
- 22 pair chromosomes are autosomes while 1 pair is sex chromosome.
- Length: 4-6 µ



autosomes

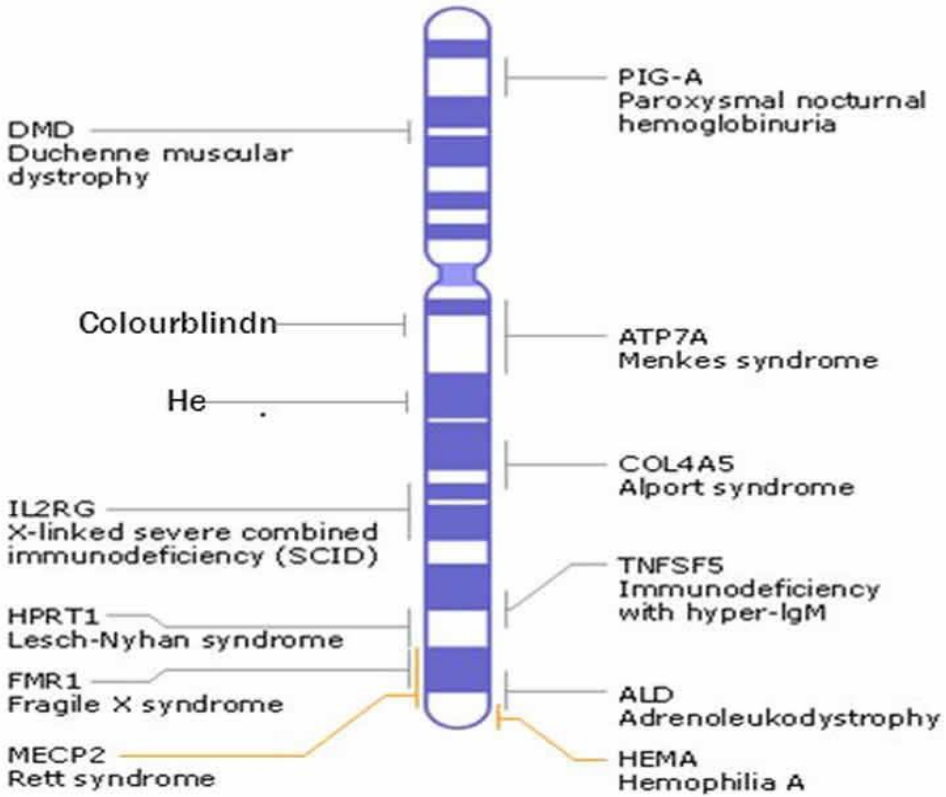
sex chromosomes





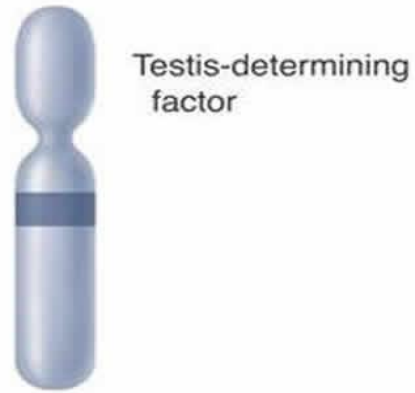
Sex Chromosomes

X chromosome



900-1600 genes

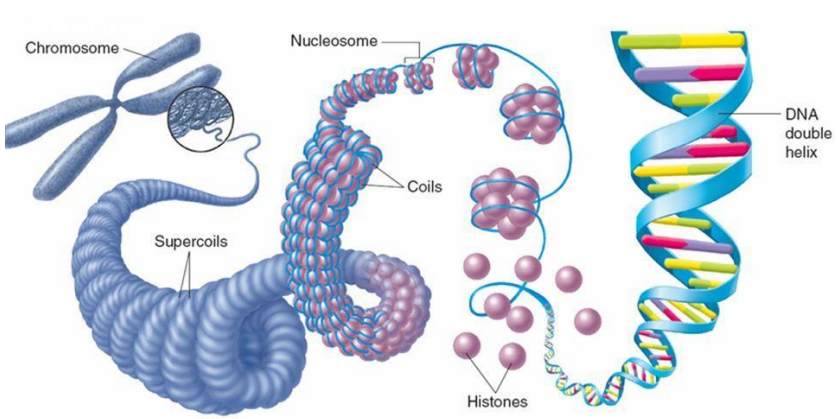
Y chromosome

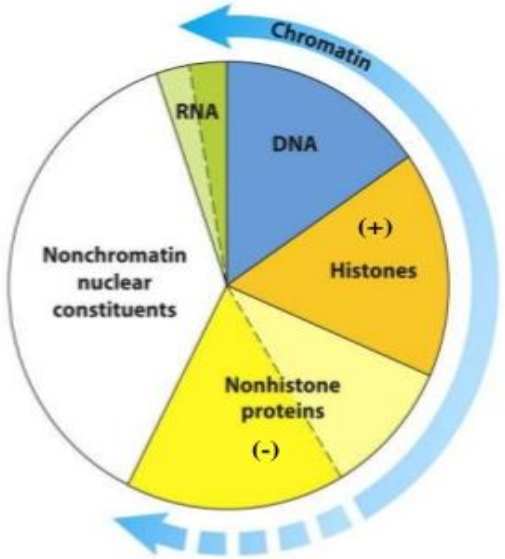


70-200 genes

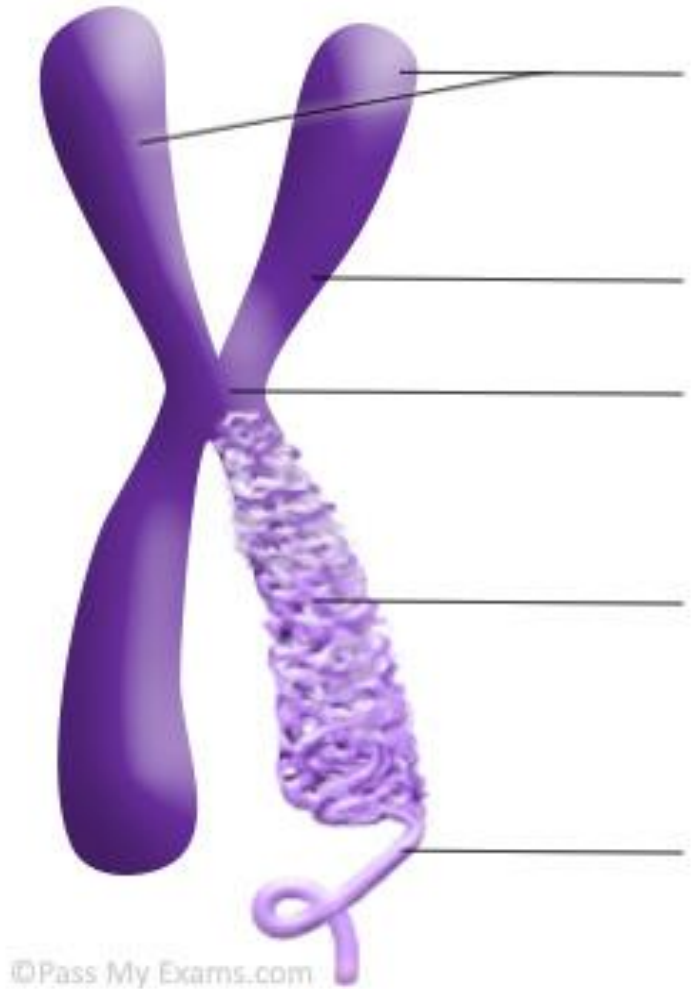
Chemical Constituents

- DNA
- RNA
- Histones (basic in nature)
- Non-histone proteins (acidic in nature)





Structure of one Chromosome



Two Identical Chromatids
One is an exact copy of the other and each contains one DNA molecule.

p arm – short arm structure

Centromere – constricted point of the chromosome

q arm – long arm structure

DNA molecule – long string like DNA molecule formed into a compact structure by proteins called histones.

Centromere- It is a localized region of the chromosome with which spindle fibers attached is known as centromere or primary constriction or kinetochore

Chromatid- One of the two distinct longitudinal subunits of a chromosome is called as chromatid. Chromatids are of two types sister chromatids and non-sister chromatids.

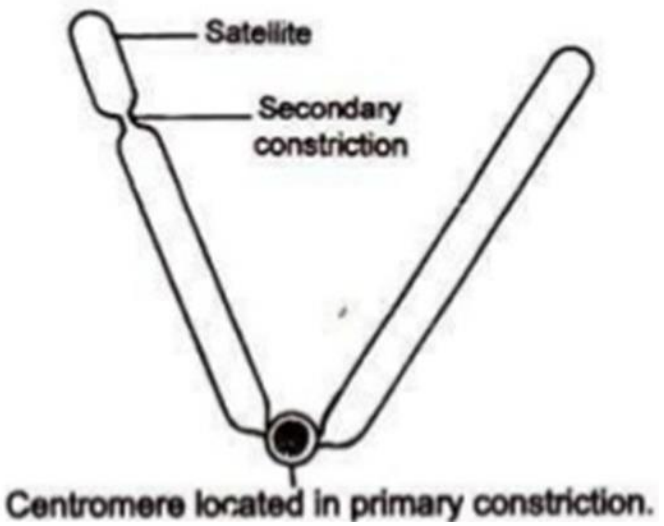
Secondary constriction- Some chromosome exhibits secondary constriction in addition to primary constriction. The chromosomal region between telomere is called as **satellite** or **trapant.** *The chromosome having satellite is called as satellite chromosome.*

Telomere- The two ends of chromosome are called as telomeres. Telomere are highly stable and they do not fuse or unite with telomere of other chromosome.

Chromomere- The chromosomes of some of the species shows small bead like structures called as chromomeres. The structure of chromomeres in chromosome is constant.

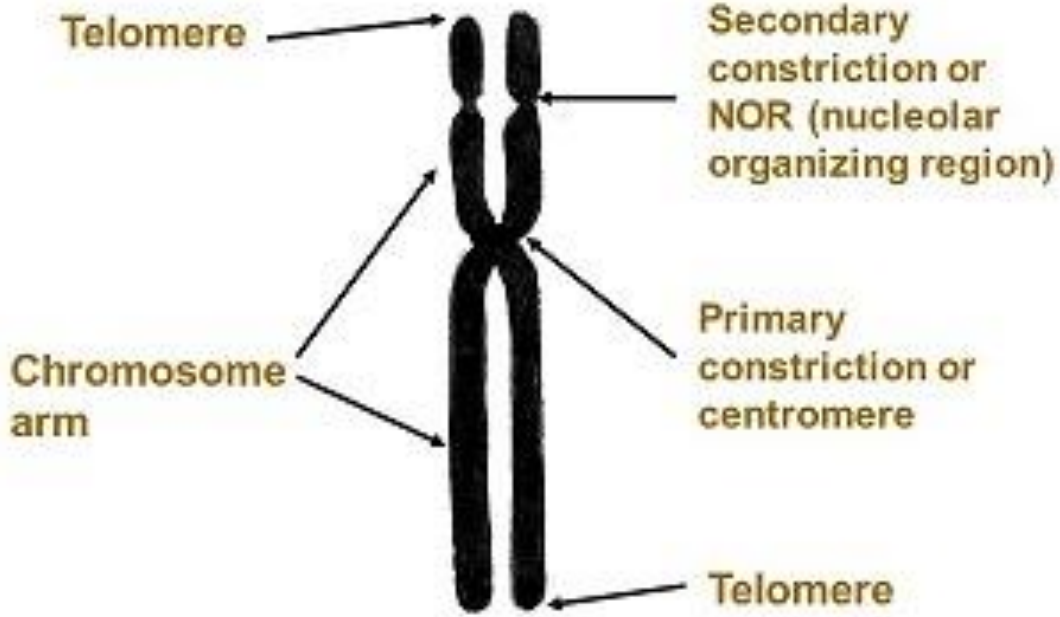
Secondary Constriction

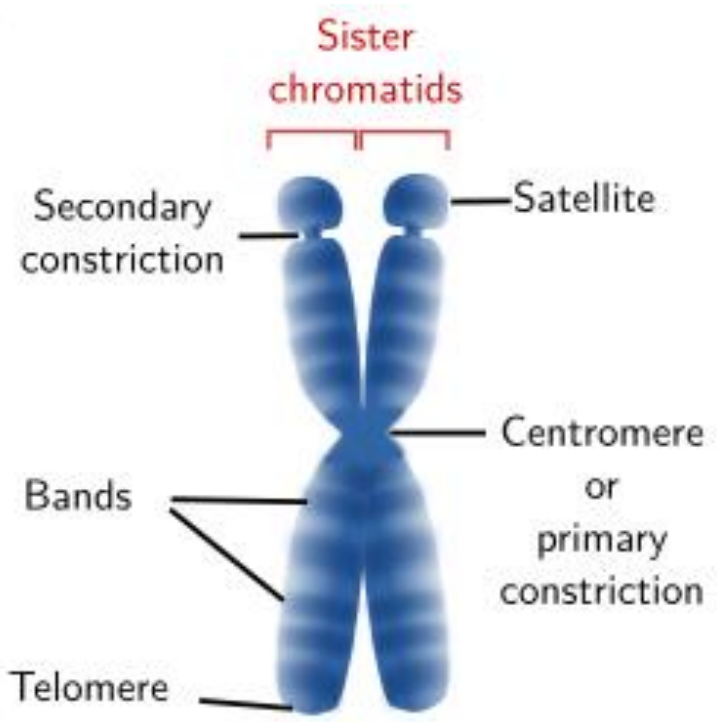
- ✓ In some chromosomes a second constriction, in addition to that due to centromere (primary constriction) is also present. It is known as "Secondary constriction).
- ✓ It is present in short arm near one end, or in many chromosomes they are located in the long arm nearer to the centromere.
- ✓ The region between the secondary constriction and the nearest telomere is known as **satellite**. Therefore, chromosomes having secondary constitution are called " **Satellite Chromosome**" or "**Sat - Chromosomes**.



- the chromosomes number **13, 14, 15, 21 and 22** are examples of SAT chromosomes

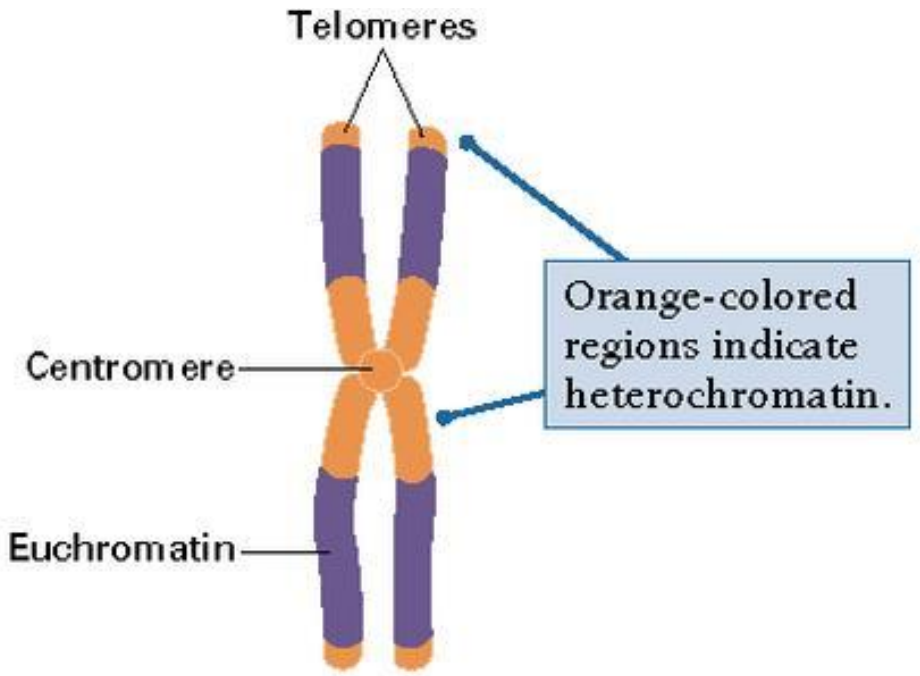
Structure of SAT Chromosome





TELOMERE

- At two ends of chromosome
- Highly stable
- Made up of loops of chromatin fibers
- Maintenance of structural integrity



-osome

A thread-like structure of nucleic acids and proteins found in the nucleus of most living cells, carrying genetic information in the forms of genes.

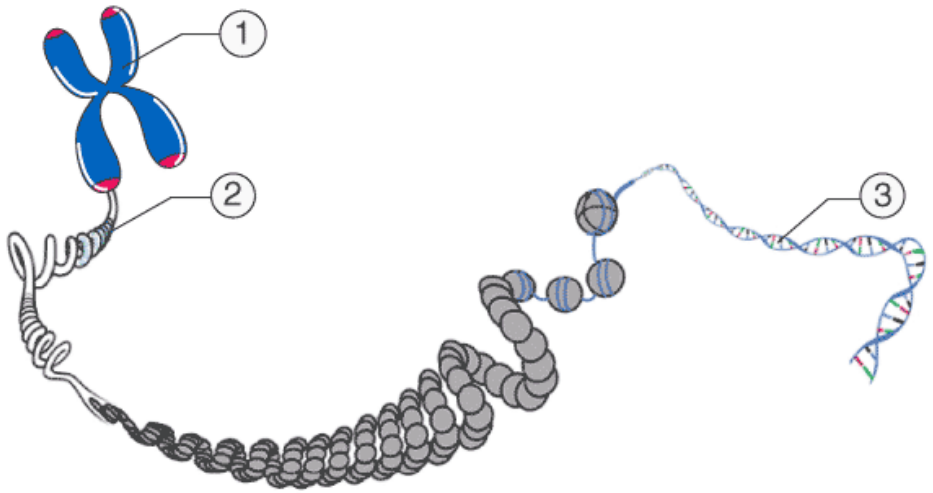
-atid

Each of the two thread-like strands into which a chromosome divides longitudinally during cell division. Each contains a double-helix of DNA.

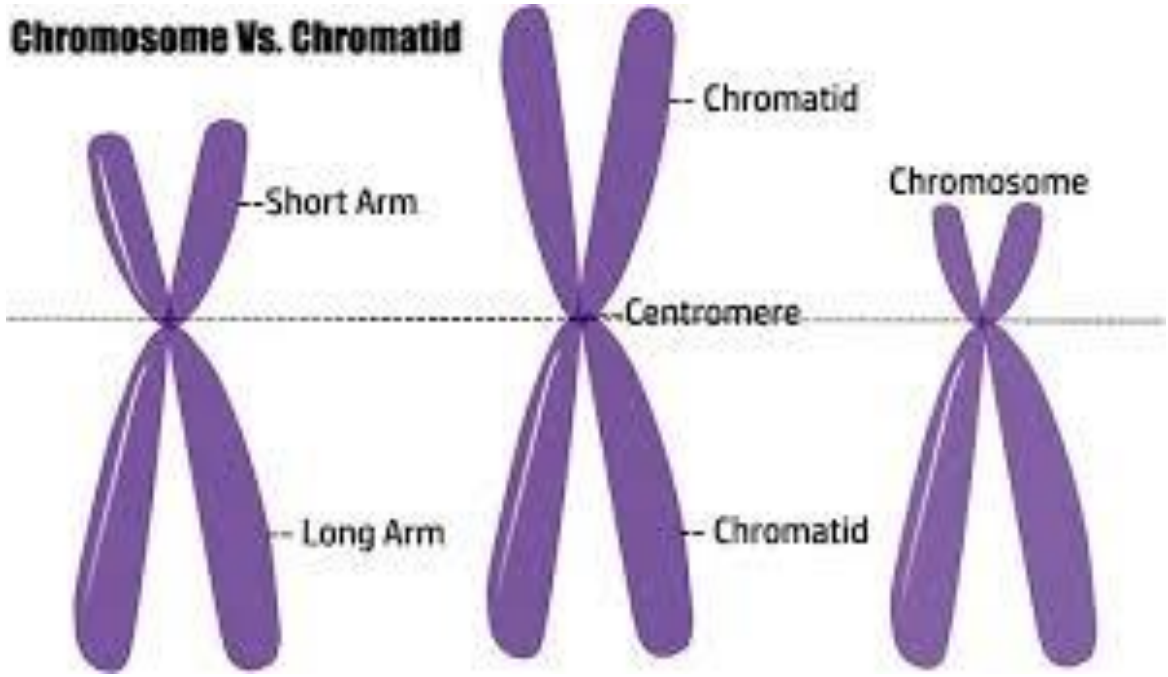
-atin

The material of which the chromosomes of organisms other than bacteria (i.e. eukaryotes) are composed, consisting of proteins, RNA and DNA

Chromatin, Chromosome, Chromatid



Chromatin is a genetic material or a macromolecule comprising of DNA, RNA, and proteins which result in the formation of chromosomes within the nucleus of eukaryotic organisms is termed as chromatin.



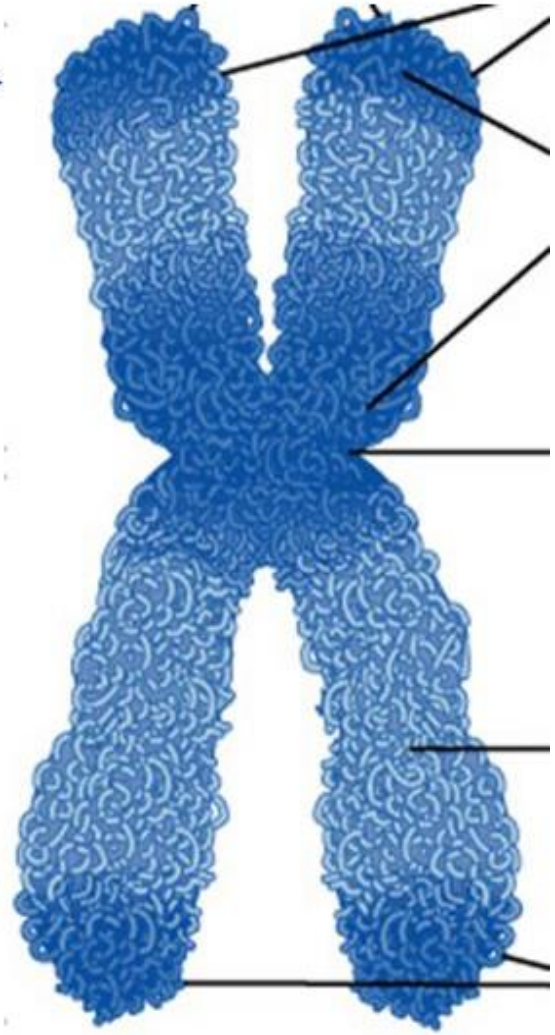
Chromatin

- Coiled strands of DNA bound to basic proteins (histones).
- 2 types:

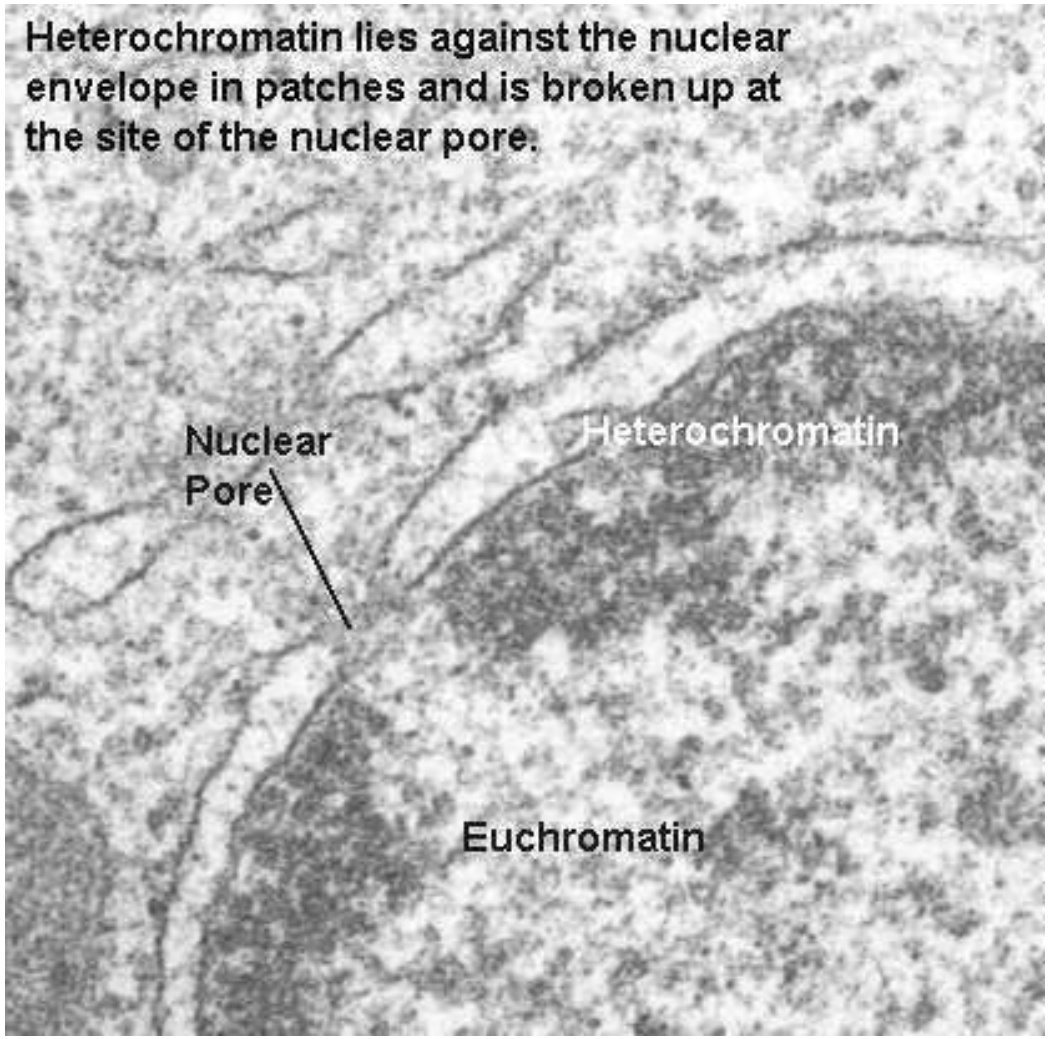
  (1) Euchromatin

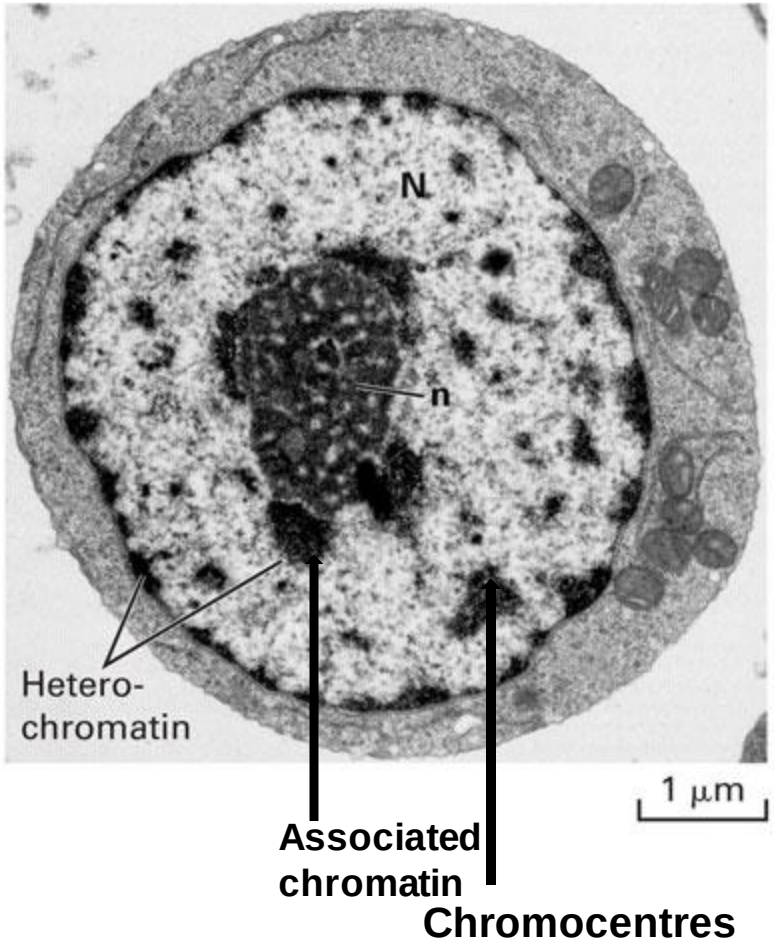
  (2) Heterochromatin
- Basic structural unit: Nucleosome

- Heterochromatin:
  - More condensed
  - Silenced genes (methylated)
  - Gene poor (high AT content)
  - Stains darker
- Euchromatin:
  - Less condensed
  - Gene expressing
  - Gene rich (higher GC content)
  - Stains lighter



HETERO & EUCHROMATIN





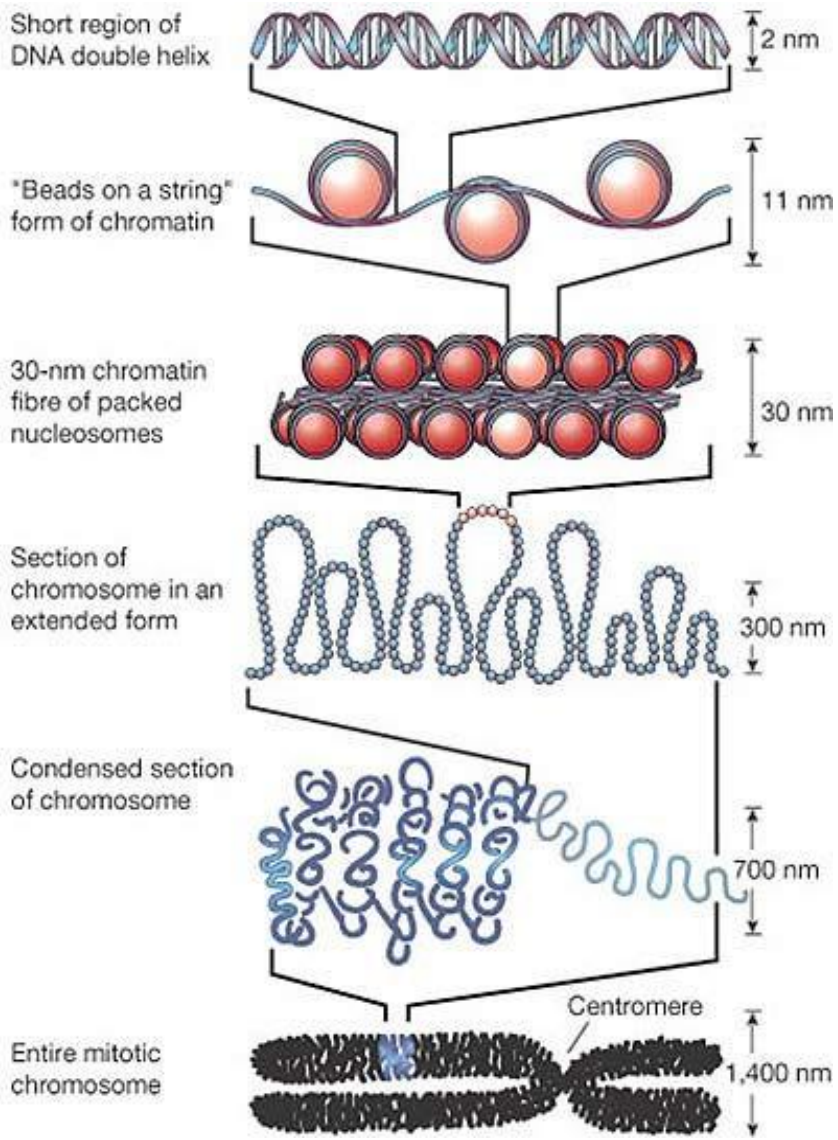
Solenoid model

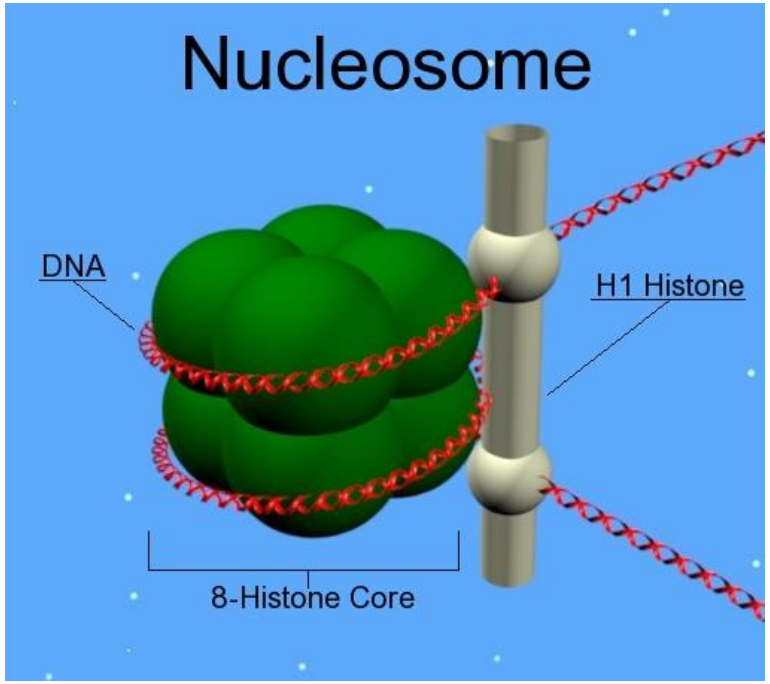
- It was proposed by Kornberg and Thomas in 1974.
- Chromatin composed of a repeating unit called as nucleosome.
- Nucleosome consist of-

  A nucleosome core Linker DNA

  One molecule of 1HI histone

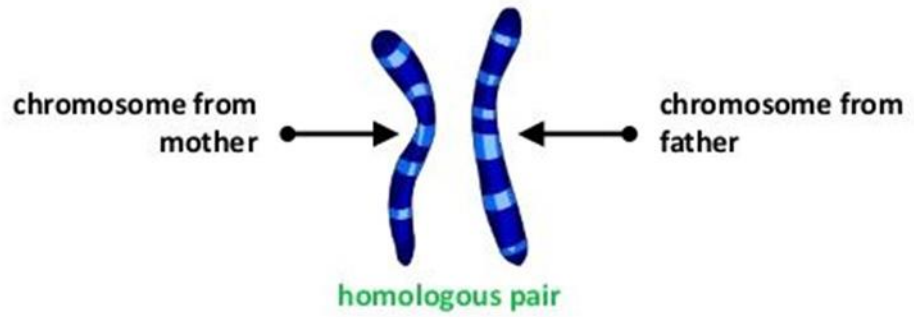
  Other associated chromosomal proteins


Short region of DNA double helix
2 nm
"Beads on a string" form of chromatin
11 nm
30-nm chromatin fibre of packed nucleosomes
30 nm
Section of chromosome in an extended form
300 nm
Condensed section of chromosome
700 nm
Centromere
Entire mitotic chromosome
1,400 nm


Nucleosome
DNA
H1 Histone
8-Histone Core

Types of Chromosomes

- **Homologous chromosomes** pertain to two chromosomes which are same, at least in terms of the gene sequences and loci.



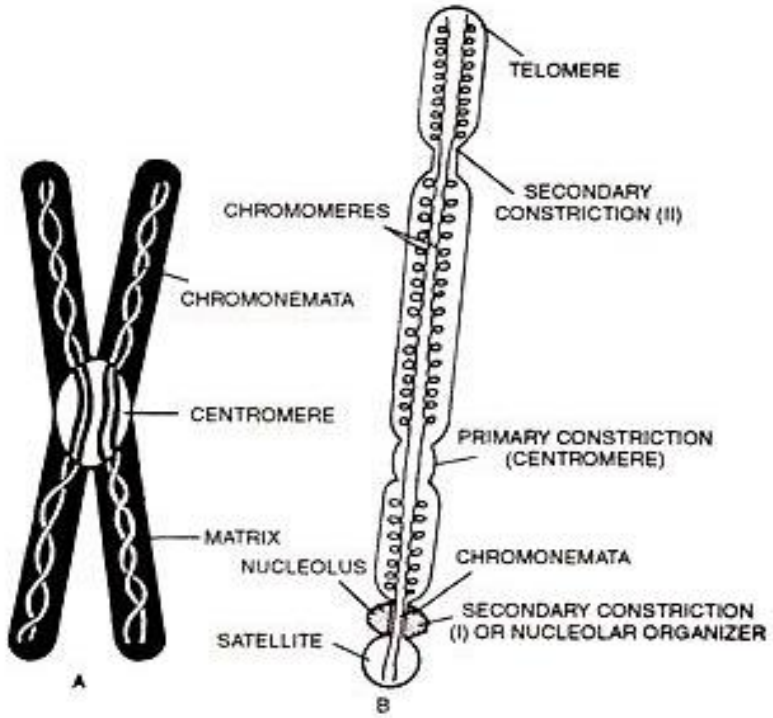
- **Heterologous** (non-homologous) **chromosomes** pertain to any two **chromosomes** that are different, such as in terms of gene sequence and loci.

FUNCTIONS OF CHROMOSOME

- It provides genetic information for cellular functions of organisms.
- It protects genetic material (DNA) from damage during cell division.
- They ensure a precise distribution of DNA to daughter nuclei during cell division.

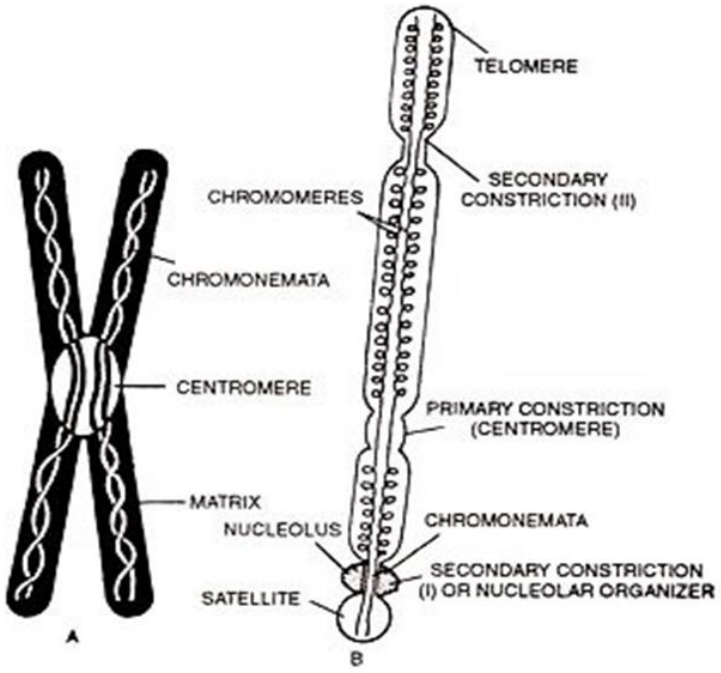
CHROMONEMATA

- The spirally coiled central filament of a chromatid carrying all the inherited instructions, along which the chromomeres are aligned.
- The set of instructions is called as **‘genome’**.



CHROMOMERE

- A chromomere, also known as an idiomere, is one of the serially aligned beads or granules of a eukaryotic chromosome, resulting from local coiling of a continuous DNA thread.



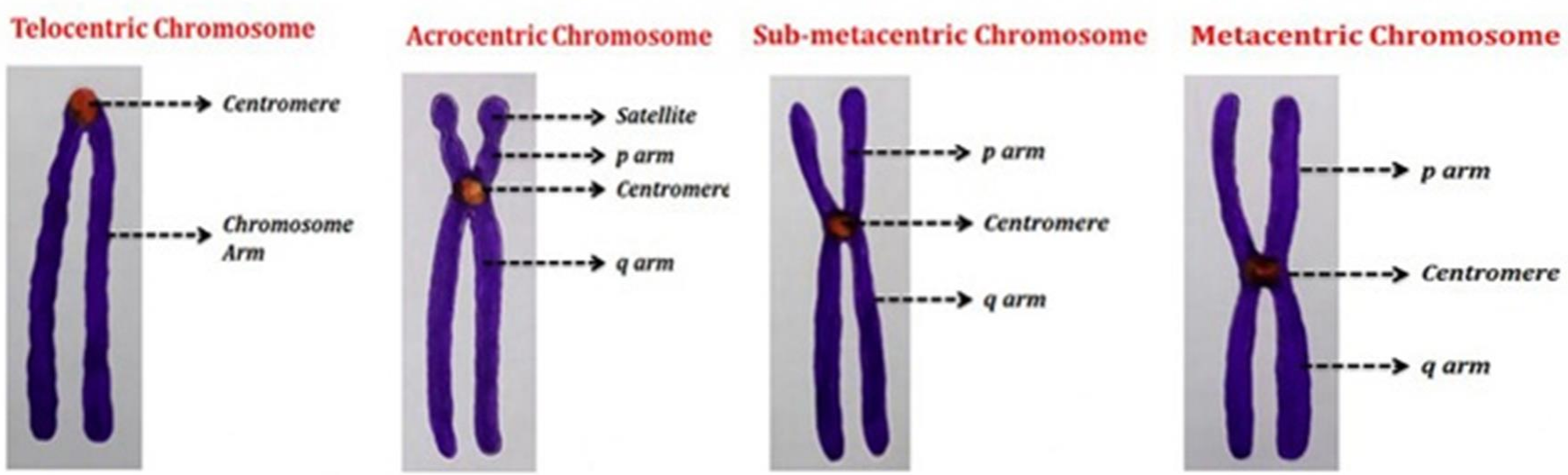
CLASSIFICATION OF CHROMOSOMES

- **According to position of the centromere:**

1. Metacentric
2. Submetacentric
3. Telocentric
4. Acrocentric:

   Satellite chromosomes

   (SAT chromosomes)



- **According to total length of the chromosome (Denver's classification):**

Seven groups (A-G):

A 1,2,3

B 4,5

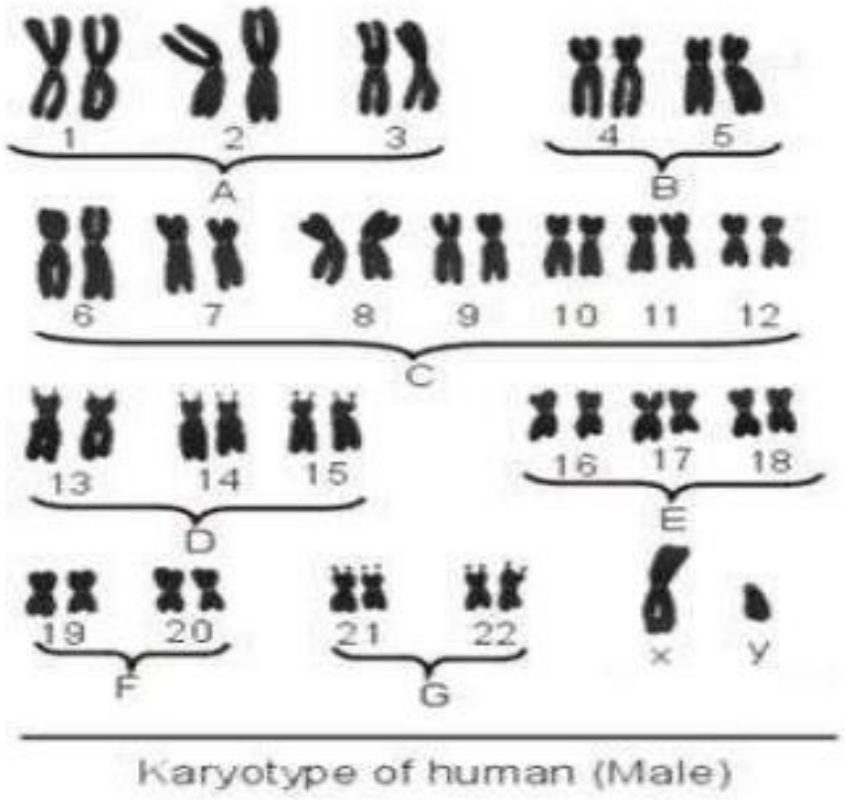
C 6-12

D 13,14,15

E 16,17,18

F 19,20

G 21,22



Karyotype of human (Male)

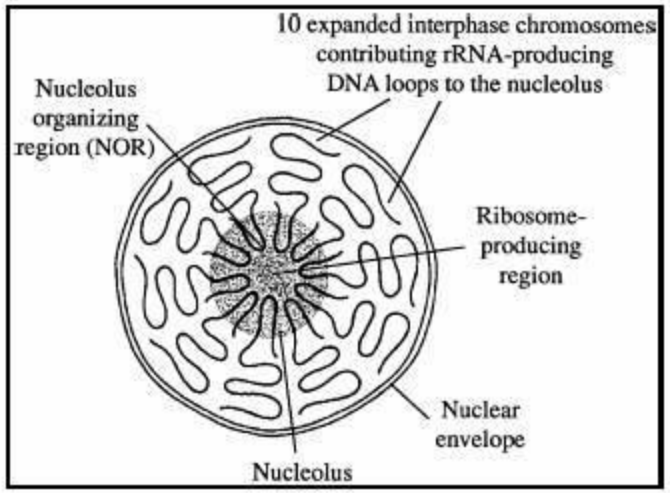
- **According to number of the centromere:**

1. **Monocentric**
2. **Dicentric**
3. **Polycentric**
4. **Acentric**

   (All the 46 human chromosomes are monocentric)

NUCLEOLUS ORGANISER REGION

- **Nucleolus organizer regions** (NORs) are chromosomal regions crucial for the formation of the nucleolus.
- In humans, the NORs are located on the short arms of the acrocentric chromosomes 13, 14, 15, 21 and 22.
- These regions code for 5.8S, 18S, and 28S ribosomal RNA.



CHROMOSOME NUMBER

Two types:

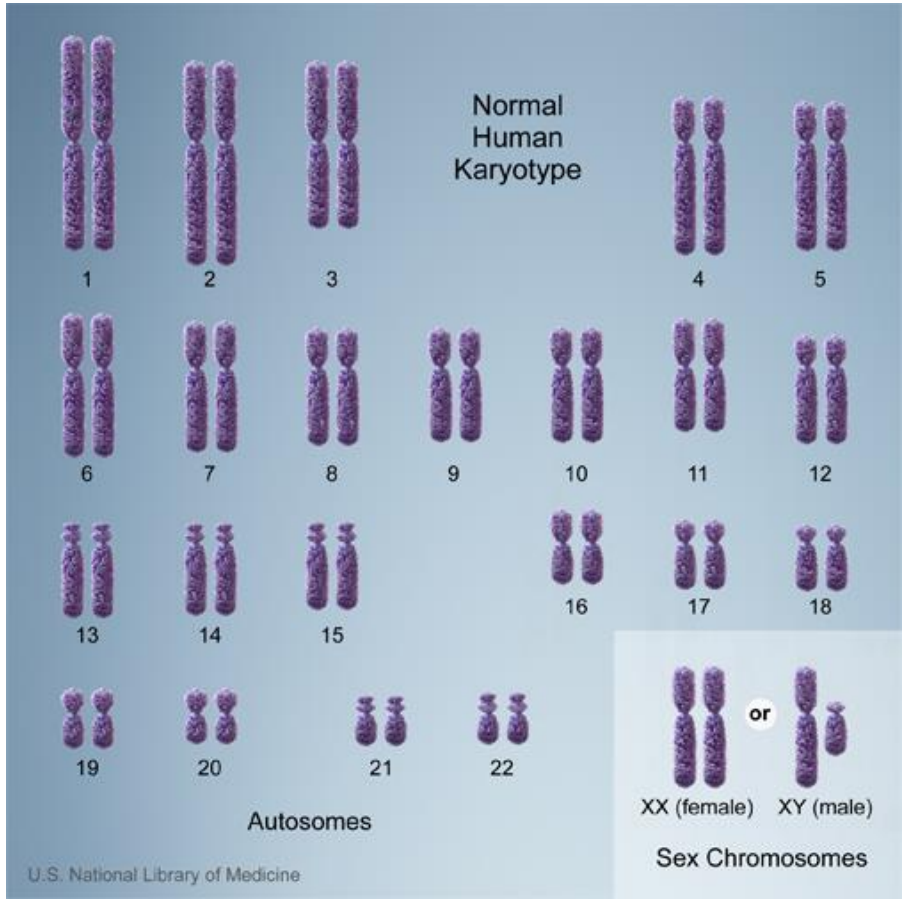
1) **Somatic chromosome number (2n):** any chromosome that is not a sex chromosome; appear in pairs in body cells but as single chromosomes in spermatozoa.
2) **Gametic chromosome number (n):** any of the chromosomes contained in a haploid cell, specifically a spermatozoon or an ovum, as contrasted with those in a diploid, or somatic, cell.

Chromosome size

- Shows variation depending upon stage of cell division.
- Longest and thinnest chromosome seen during interphase.
- In prophase, decreases in length with an increase in thickness .
- Smallest chromosome seen during anaphase.

KARYOTYPE

- General morphology of somatic chromosome complement of an individual.



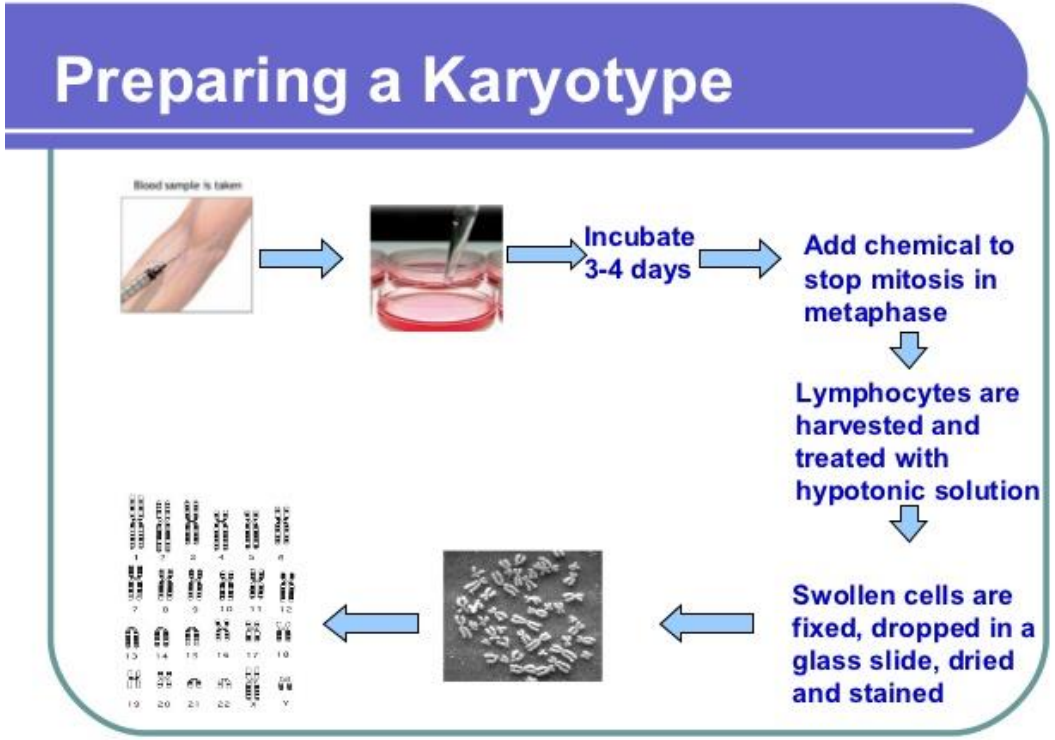
Karyotyping

Karyotyping is the process of pairing and ordering all the chromosomes of an organism, thus providing a genome-wide snapshot of an individual's chromosomes.

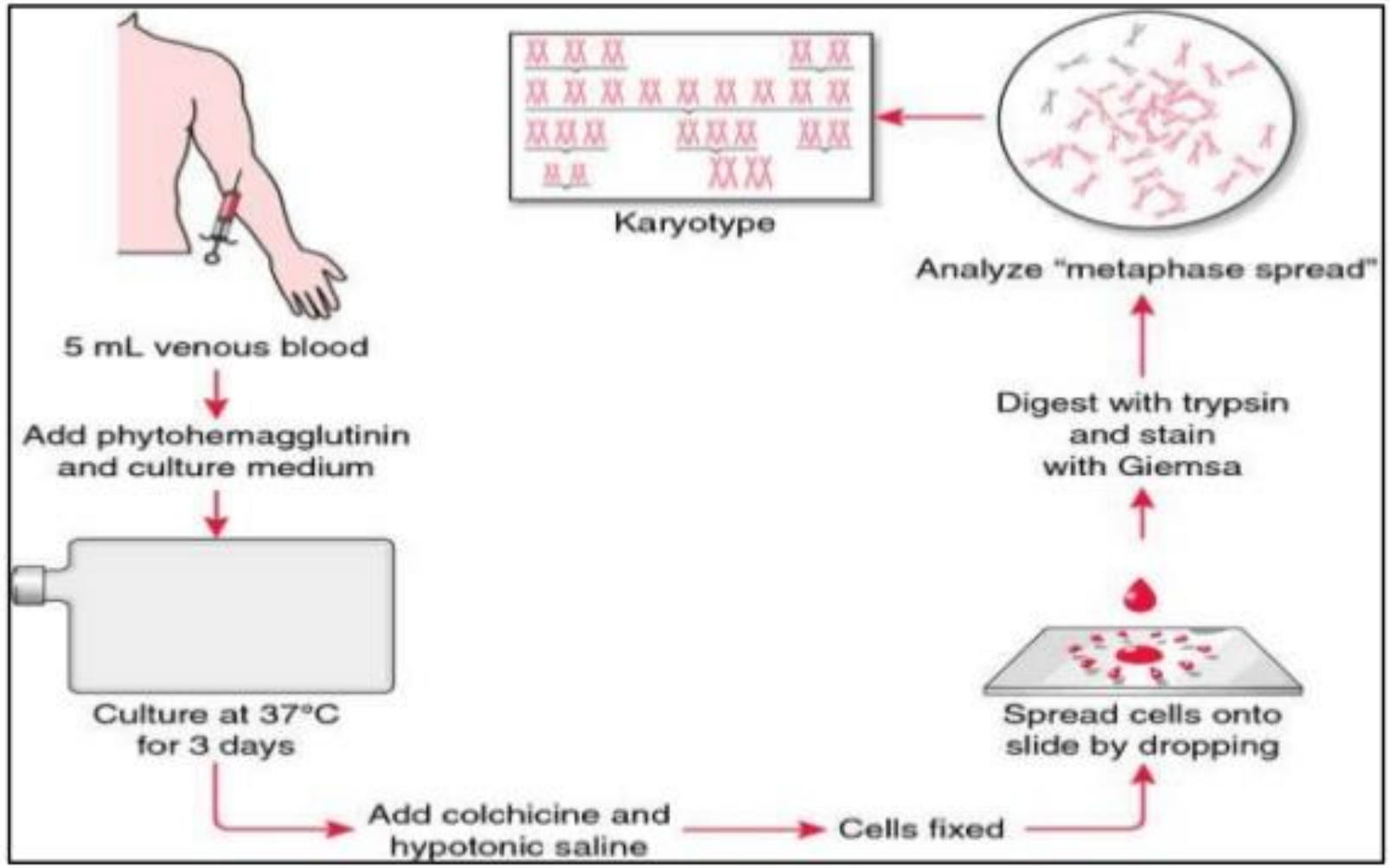
Karyotypes describe the chromosome count of an organism and what these chromosomes look like under a light microscope.

Attention is paid to their length, the position of the centromeres, banding pattern, any differences between the sex chromosomes, and any other physical characteristics.

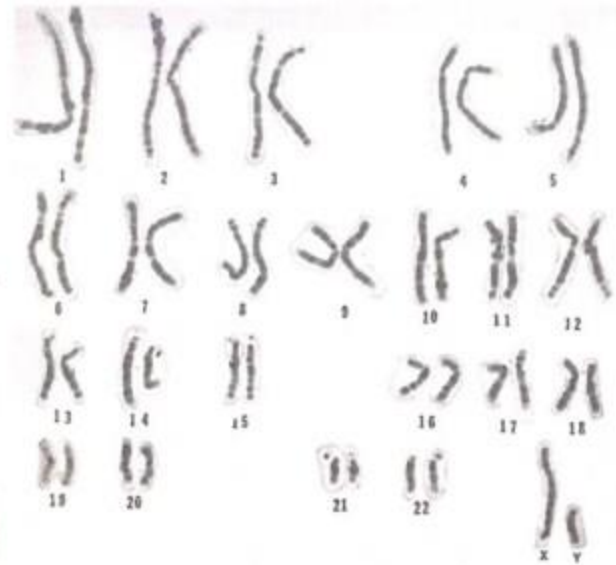
Karyotypes are prepared using standardized staining procedures that reveal characteristic structural features for each chromosome.



Procedure of karyotyping

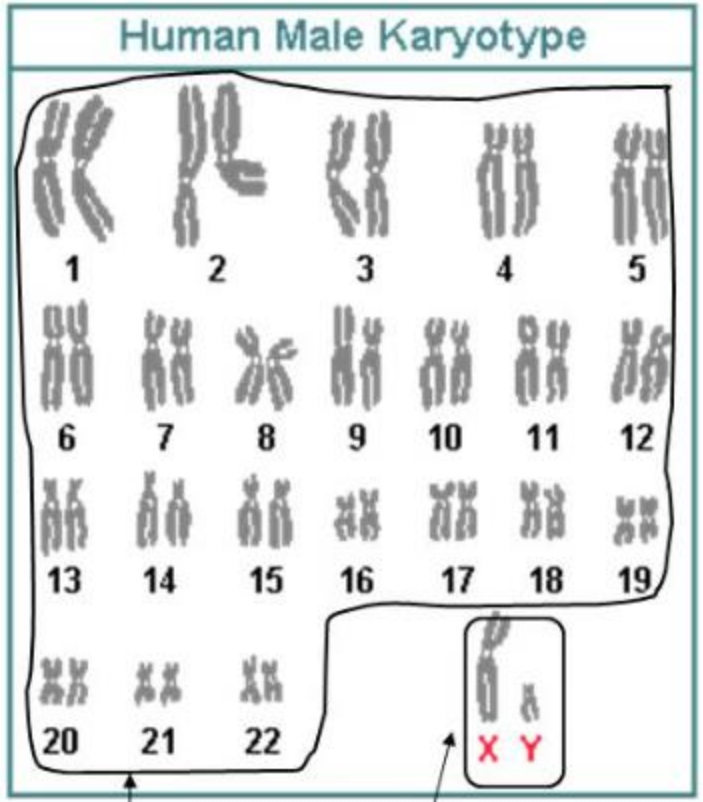


Metaphase Chromosomes



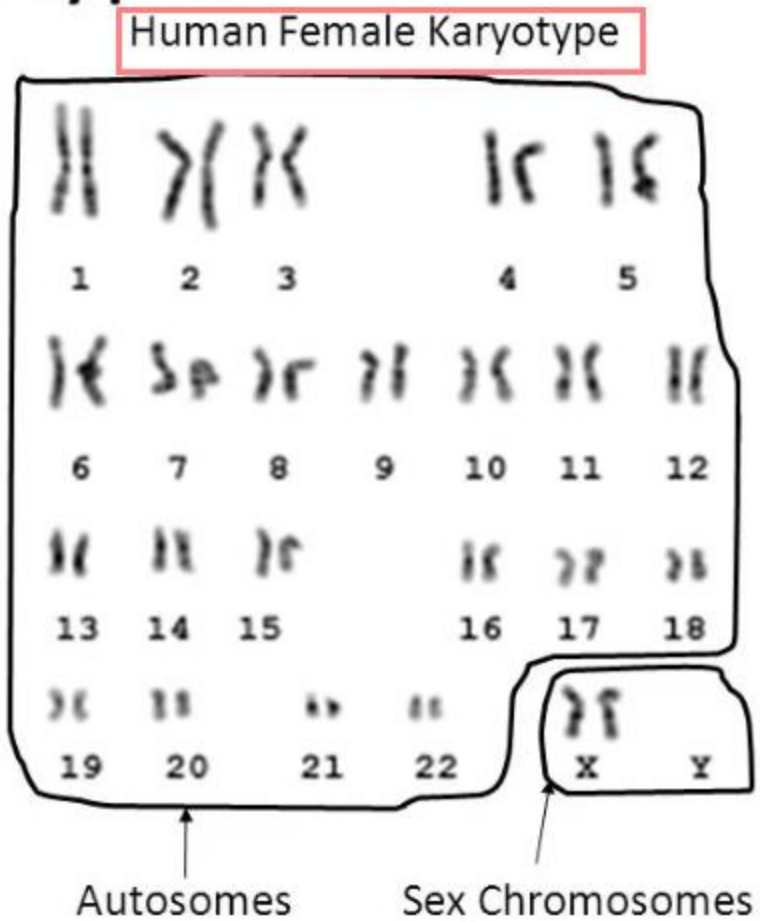
Chromosomes arranged

Karyotypes



Notation: **46,XY**

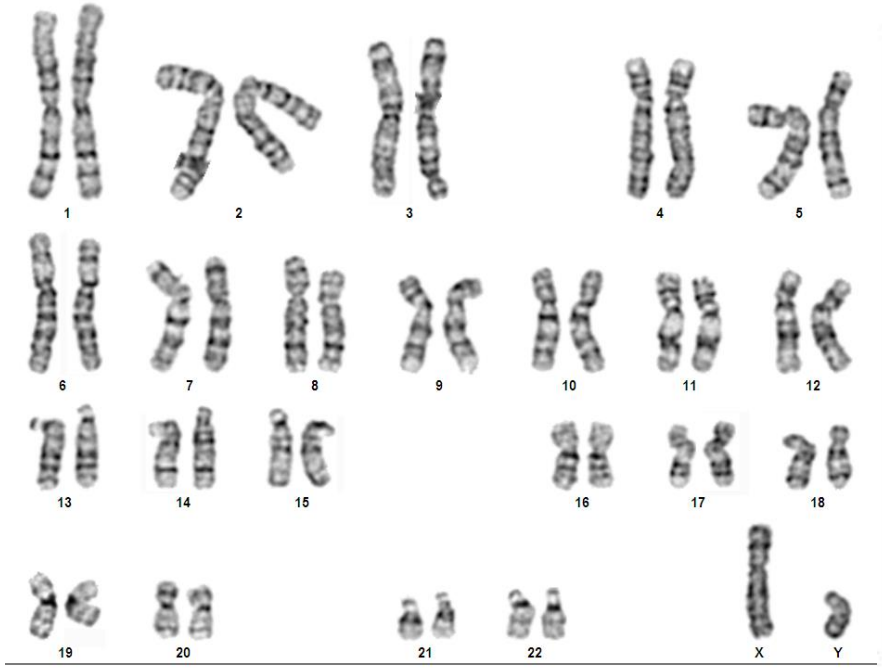
Diagnosis: Normal male



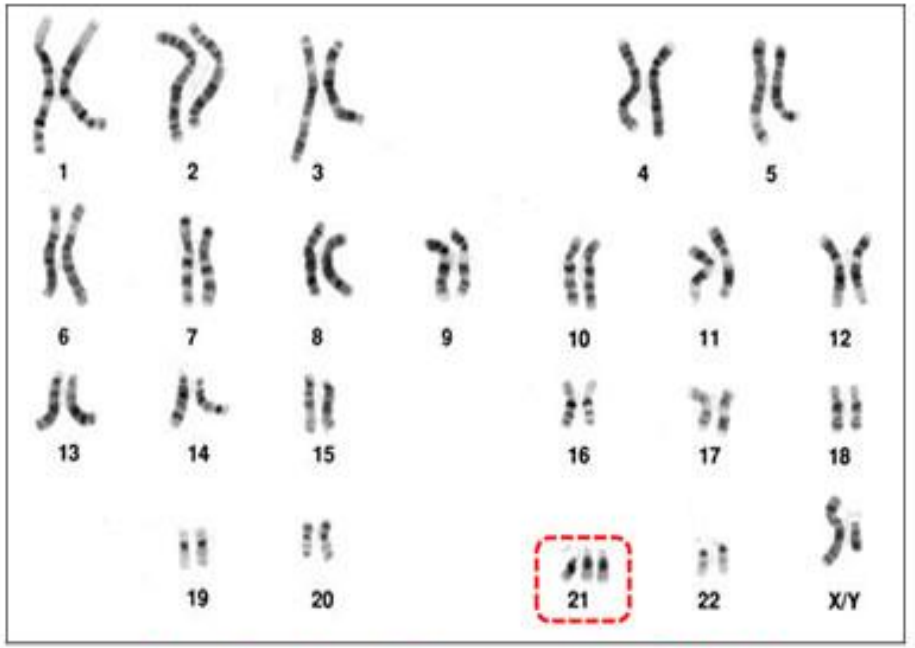
Notation: **46,XX**

Diagnosis: Normal female

Karyogram: a diagram or photograph of the chromosomes of a cell, arranged in homologous pairs and in a numbered sequence, also called idiogram.



Normal Karyotype

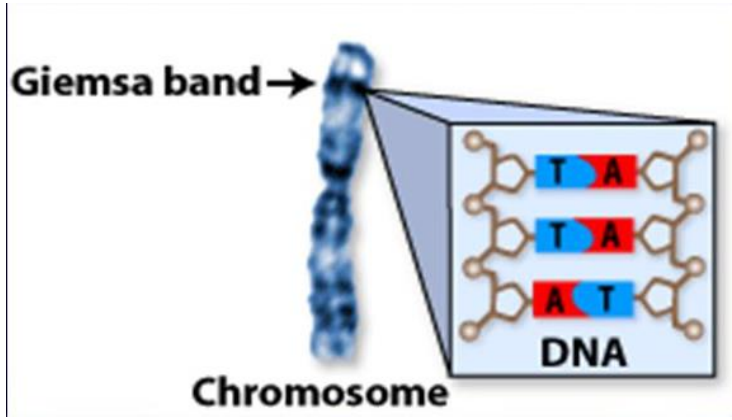


Down Syndrome Karyotype, 45 XY (Male)

G-banding

- **G banding**, or **Giemsa banding** is a technique used in cytogenetics to produce a visible karyotype by staining condensed chromosomes.

- It is useful for identifying genetic diseases through the photographic representation of the entire chromosome complement.



Applications of Karyotyping

- Detection of chromosomal aberration
- Identify loss and addition of chromosome
- Determine the risk of individual
- Detect aneuploidy
- Prebirth diagnosis of genetic diseases

Contd……..

- **Karyotyping** can be used to detect a variety of genetic disorders. For example, a woman who has premature ovarian failure may have a chromosomal defect that **karyotyping** can pinpoint.

- The test is also useful for identifying the Philadelphia chromosome. Having this chromosome can signal chronic myelogenous leukemia (CML)

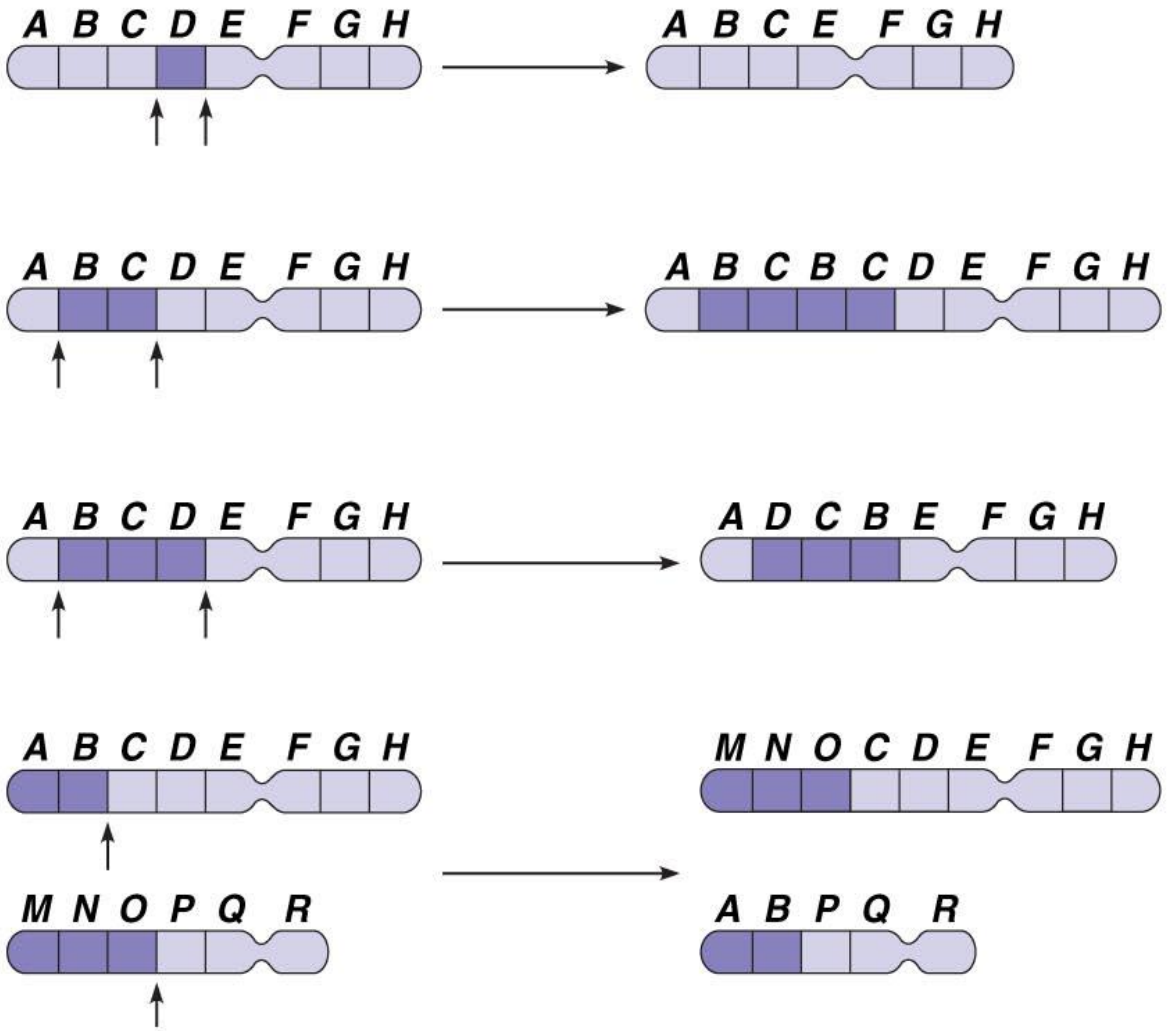
Alteration of chromosome structure

(a) A deletion removes a chromosomal segment.

(b) A duplication repeats a segment.

(c) An inversion reverses a segment within a chromosome.

(d) A translocation moves a segment from one chromosome to another, nonhomologous one.



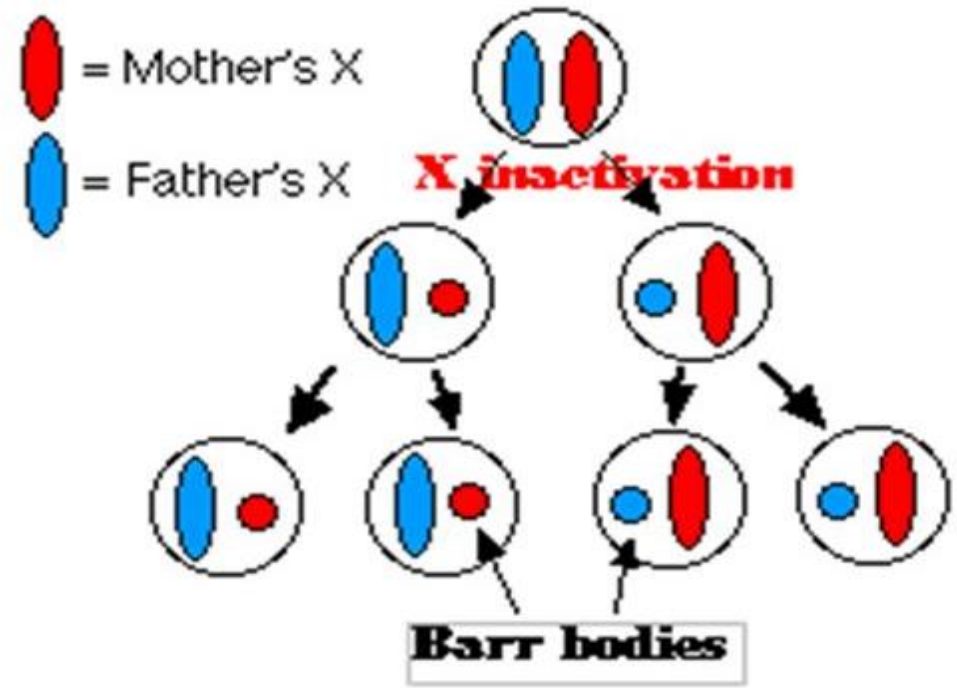
Lyon hypothesis

- In somatic cells of a female only one of the X chromosomes is active
- X-inactivation
  - Occurs early in embryonic life
  - Is random
    - either paternal or maternal X
  - Is complete
  - Is permanent
  - Is clonally propagated through mitosis

Mary Lyon

X Chromosome Inactivation (Lyonization)

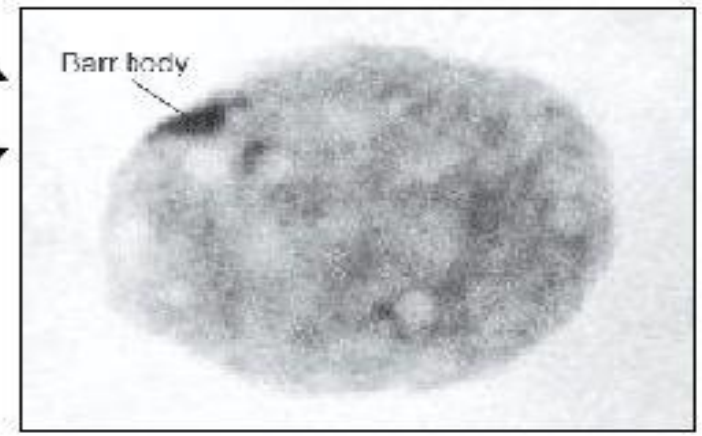
- In mammals only one X chromosome is expressed in somatic cells
- Second X condenses to become a **barr body**
- Barr bodies are reactivated during gamete formation



Barr body

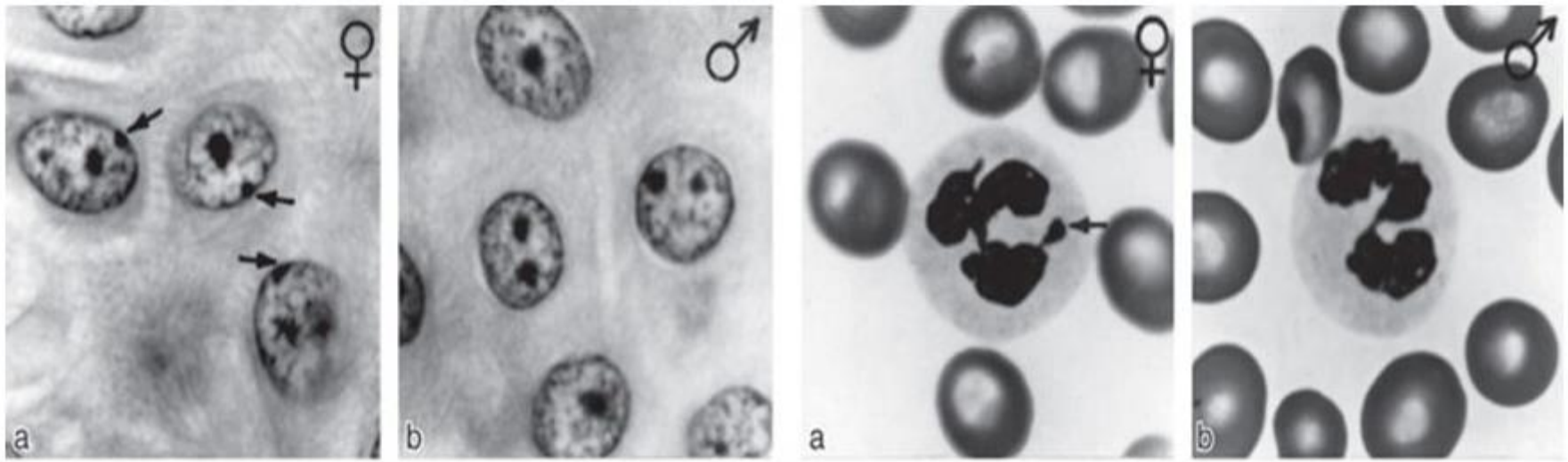
- = sex-chromatin
- Inactivated X chromosome
- Female XX
  - 1 Barr body
- Male XY
  - no Barr hody
- Klinefelter syndrome XXY
  - 1 Barr body

Murray L. Barr



Sex Chromatin

The inactive X chromosome condenses and can be seen in various types of cells, usually near the nuclear membrane, as the **Barr body,** also called sex chromatin



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- In humans with more than one X chromosome, the number of Barr bodies visible at interphase is always one fewer than the total number of X chromosomes.
- For example, people with Klinefelter syndrome (47,XXY karyotype) have a single Barr body, and people with a 47, XXX karyotype have two Barr bodies.

Condition	Sex chromosomes	Number of Barr bodies
Normal male	XY	None
Normal female	XX	One
Trisomy X	XXX	Two
Turner's syndrome	X	None
Kleinefelter's syndrome	XXY	One

Turner’s Syndrome

Thank You