

LECTURE (4)

# Human Genetics

**3<sup>rd</sup> stage**

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# CHROMOSOMES

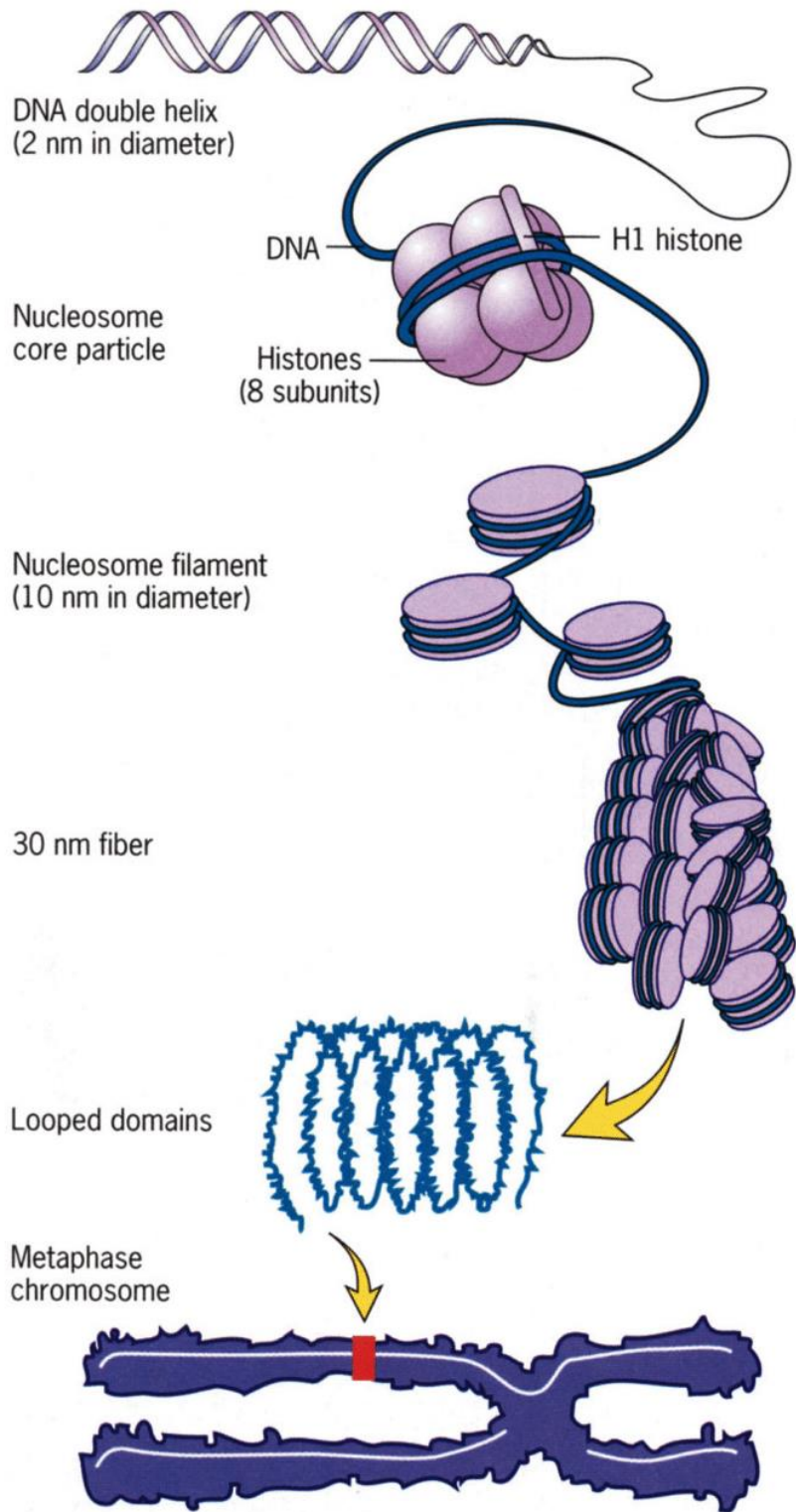
Chromosomes are **thread-like structures** located inside the nucleus of animal and plant cells. Each chromosome is made of **protein** and a **single molecule of deoxyribonucleic acid (DNA)**. First discovered chromosomes in the late **1800s**, by using **light microscope** was, biologist **Walter Flemming** observed many elongated structures in cell nuclei.

## ➤ **DNA packaging into chromosome**

Specific proteins compact DNA into the microscopic space of the eukaryotic nucleus. These proteins are called **histones**, Proteins are added to DNA to make it more compact and the resulting DNA-protein complex is called **chromatin**. packaging requires **energy** is needed to perform work. This energy comes from (**electrostatic interactions**) between protein and DNA strand. Histones are a family of small, **positively charged** proteins termed **H1, H2A, H2B, H3, and H4**. DNA is **negatively** charged, due to the phosphate groups in its structure, so histones bind with DNA very tightly. As a result, chromatin can be packaged into a much smaller volume than DNA alone.

## ✚ **The Nucleosome**

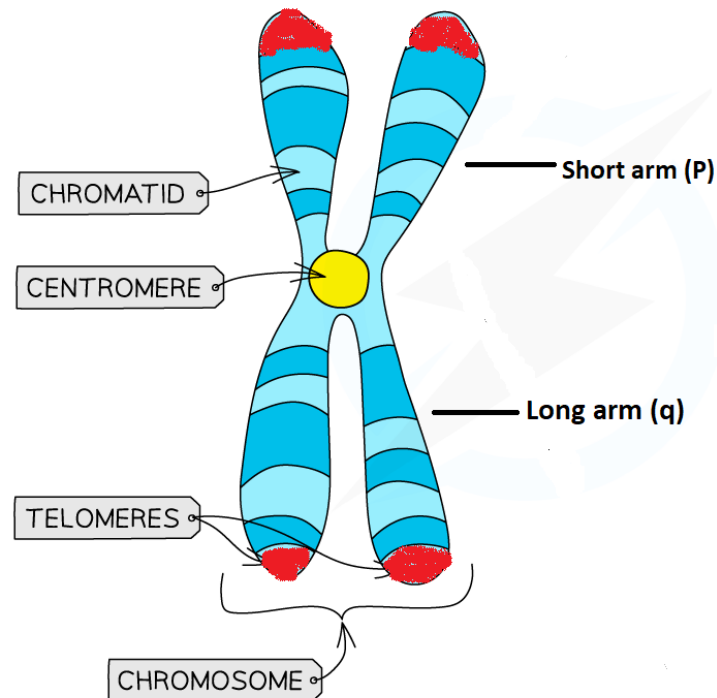
The basic structural (and functional) unit of chromatin is the **nucleosome**, which contains **eight** histone proteins: Two each of the histones **H2A, H2B, H3, and H4** come together to form a **histone octamer**, which binds and wraps approximately **1.7 turns** of DNA and about **146** base pairs. The addition of one **H1** protein wraps another **20** base pairs, resulting in **two full turns** around the octamer, and forming a structure called a **chromatosome**.



## ➤ Structure of chromosome

A chromosome has mainly 3 basic parts

- **Chromatid**: is one of the **two identical** halves of a chromosome that has been replicated in preparation for cell division , its function carrying the genetic material .
- **Centromere** : **constriction** between the chromatids. Its function is to enable spindle fibers attachment and movement of the chromosome during the anaphase stage of cell division. Centromere divides the chromosome into two sections, or “**arms.**” The short arm of the chromosome is labeled the “**p arm.**” The long arm of the chromosome is labeled the “**q arm.**”
- **Telomeres**: structures made from **DNA sequences** and **proteins** found at the **ends of chromosomes**. They cap and protect the end of a chromosome and also telomeres are required for **cell division**.



## ➤ Chromatin

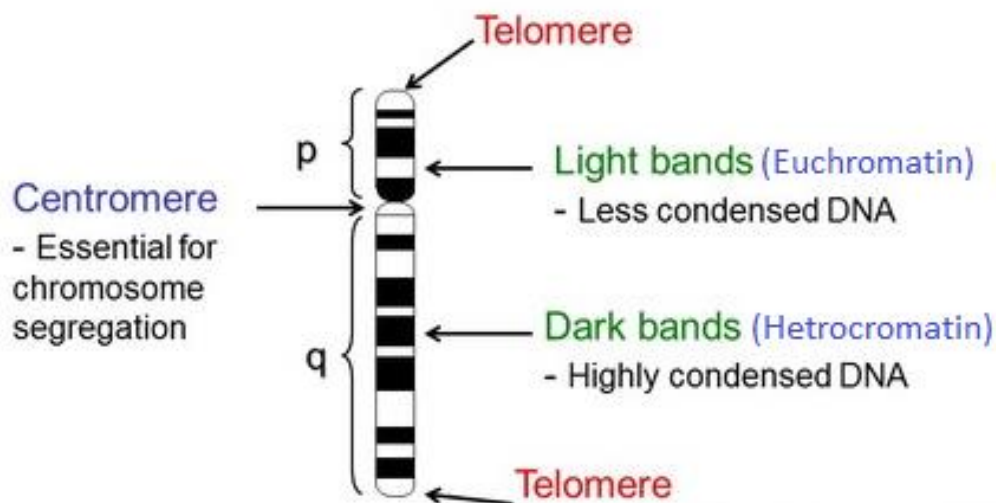
There are **two types** of chromatin within chromosome

### ✓ Euchromatin

Form of chromatin is **lightly** packed (DNA, and protein) that is enriched in **genes**, and is often under active transcription , 92% of the human genome with euchromatin

### ✓ Heterochromatin

Form of chromatin is **tightly** packed form of (DNA and protein), it was thought to be **inaccessible** to protein and therefore **not transcribed**

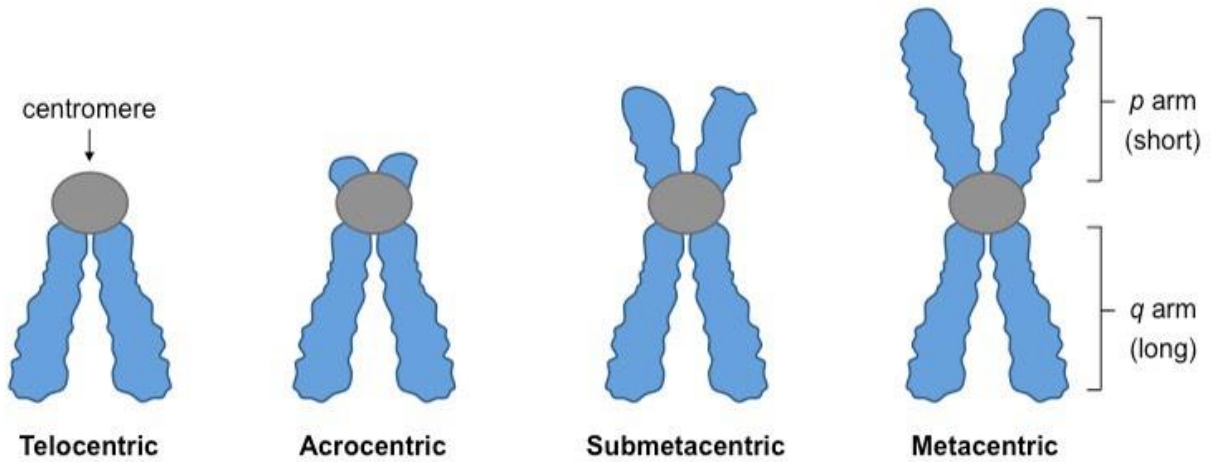


## ➤ Classification Of Chromosomes depending on centromere:

Based on the **position of the centromere**, chromosomes are divided into **four** categories. **Metacentric**, **sub-metacentric**, **telocentric**, and **acrocentric**.

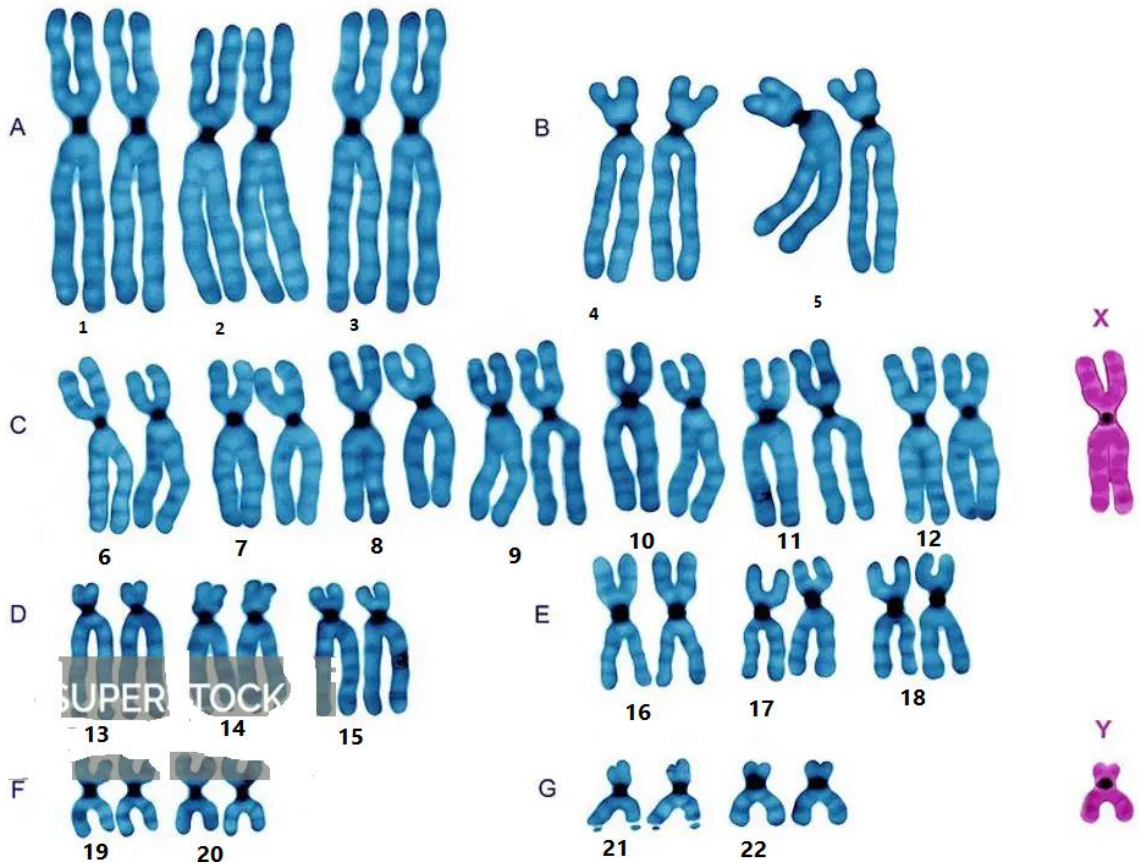
- **Metacentric** chromosome is one in which the centromere sits in the **center** of the chromosome and separates it into **two equal** arms.
- **Sub-metacentric**: a chromosome's centromere is located **distant** from the central region. One arm is **much longer than** the other in this situation
- **Acrocentric**: a chromosome is one in which the centromere is **near to one of the ends**. One arm is too long, while the other is too short.

- **Telocentric:** A chromosome is one in which the centromere is positioned at ends within telomere.



➤ **The Denver system of chromosome**

Denver classification, established in 1959, identified the chromosomes by their length and the position of the centromeres.



Group	C- Class	Size	Pcentromere
A	1-3	Large	Metacentromeric
B	4-5	Large	Submetacentric
C	6-12, X	Medium	Submetacentric
D	13-15	Medium	Acrocentric
E	16-18	Relatively short	Meta/ Submetacentric
F	19-20	Short	Submetacentric
G	21-22, Y	Short	Acrocentric