



الوراثة الطبية 1 / الكورس الاول

Chromosomes

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قسم تقنيات المختبرات الطبية

Third Stage

المرحلة الثالثة

محاضرة رقم 5

الجانب النظري : الوراثة الطبية

Lecture No. 5

Theoretical : Medical Genetics

تدريسي المادة : م. د عباس علي

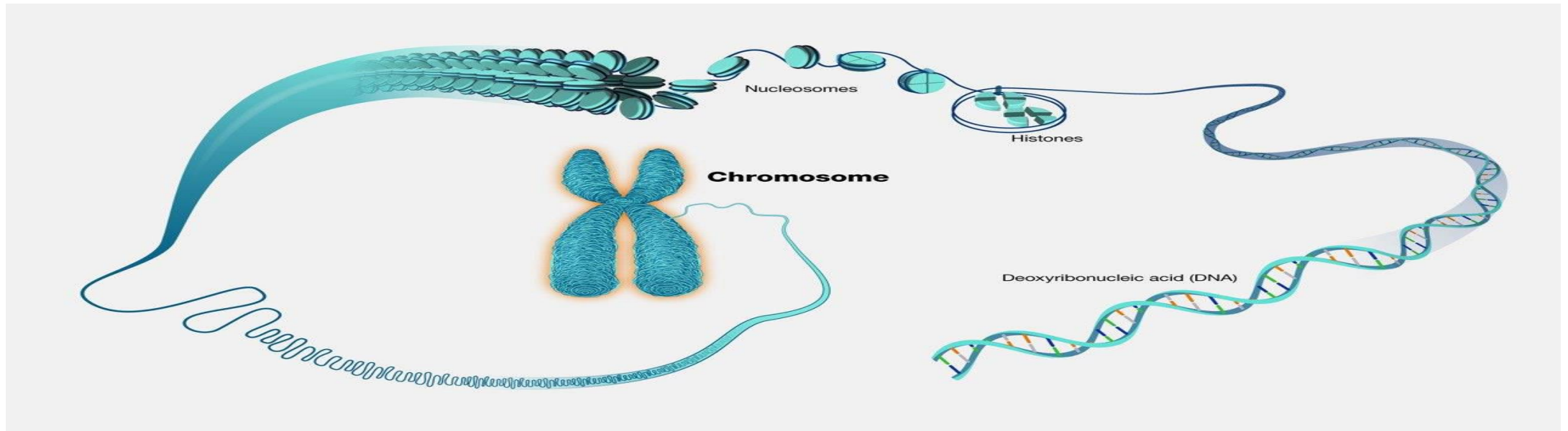
Learning Objectives

By the end of this lecture, you will be able to:

- Define chromosomes and describe their main components.
- Explain how DNA is organized into chromatin and chromosomes.
- Identify the different types of chromosomes based on centromere position.
- Distinguish between autosomes and sex chromosomes.
- Describe the major functions of chromosomes.
- Explain the importance of karyotyping and chromosomal abnormalities.

CHROMOSOMES

- **Chromosomes** are **thread-like structures** located inside the **nucleus** of animal, plant, and human cells. Each chromosome is composed of protein (**histones**) and a single molecule of deoxyribonucleic acid (**DNA**). The DNA and histone proteins together form **nucleosomes**, which represent the basic structural unit of **chromatin**. When chromatin replicates and condenses, it forms **chromosomes**. Chromosomes were first discovered in the late 1800s. Using a light microscope, the biologist Walter Flemming observed many elongated structures inside the cell nuclei.



Chemical Composition

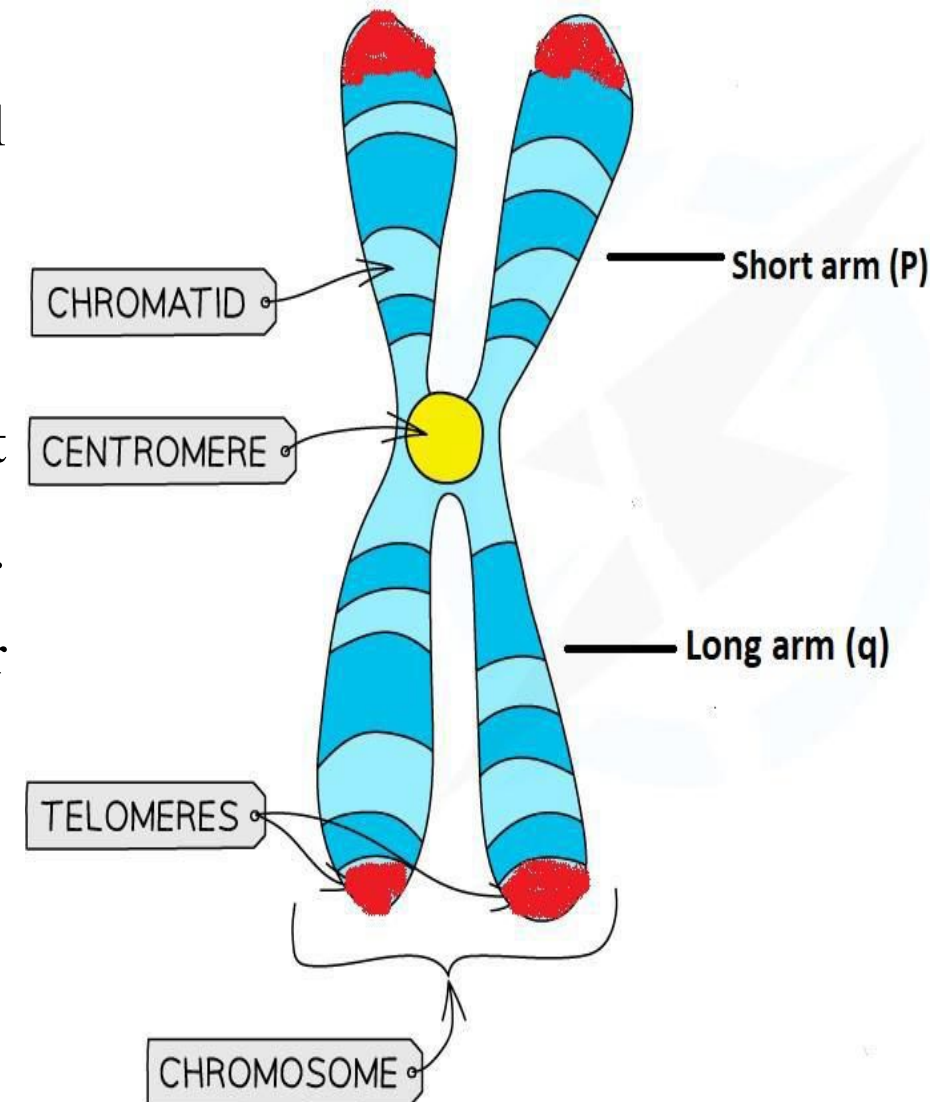
- Each chromosome consists of:
 - 1. DNA (Deoxyribonucleic Acid):** The molecule that stores hereditary information.
 - 2. Histone Proteins:** Basic proteins that help in DNA packaging.
 - 3. Non-histone Proteins:** Involved in gene regulation and chromosome structure.
 - 4. RNA molecules:** Present during transcription and gene activity.

Structure of chromosome

A chromosome has mainly 3 basic parts

1- **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.

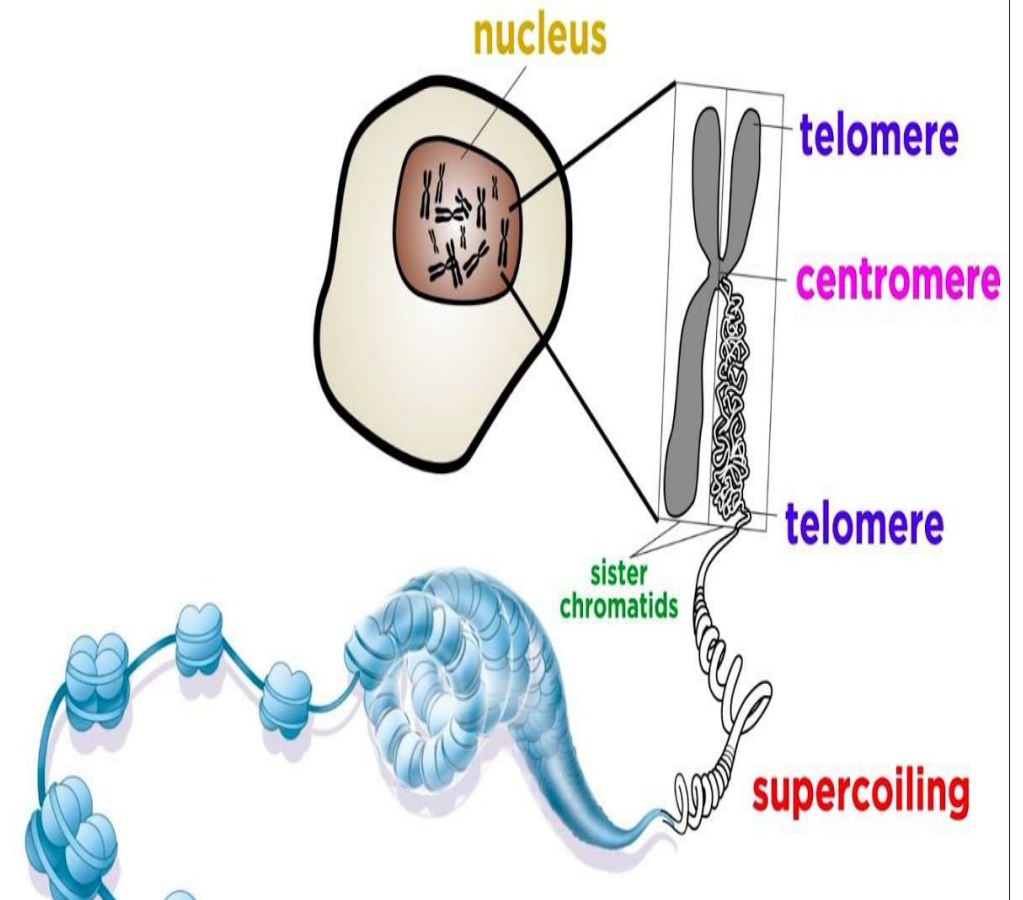
2- **Centromere** : **constriction** between the chromatids. Its function is to enable spindle fibers attachment and movement of the chromosome during the anaphase stage of cell division. Centromere divides the chromosome into two sections, or “**arms**.” The short arm of the chromosome called **petit** is labeled the “**p arm**.” The long arm of the chromosome is labeled the “**q arm**.” come from alpha beta



Structure of chromosome

3- **Telomeres**: structures made from DNA sequences and proteins found at the ends of chromosomes. They cap and protect the end of a chromosome and they do not fuse or unite with telomere of other chromosome.

Structure and Organization of Chromosomes



Chromatin

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

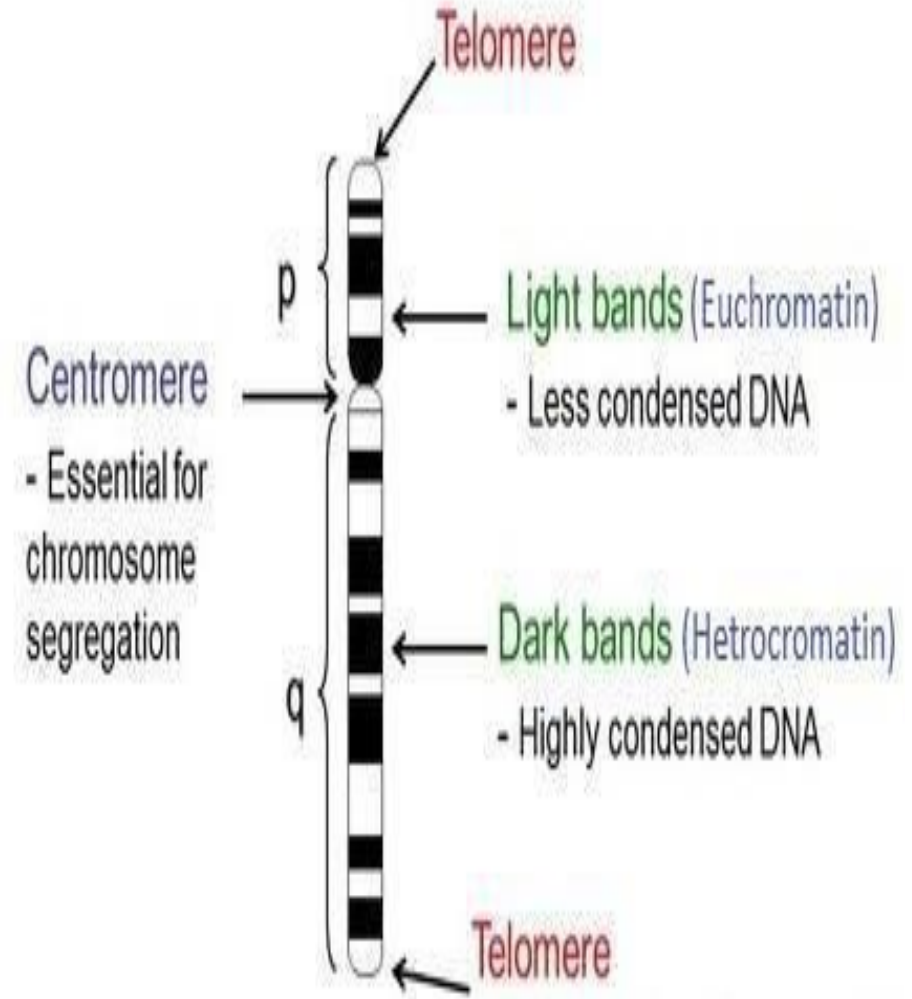
1- Euchromatin

It is a **lightly** packed form of chromatin (DNA and protein) that is enriched in genes and is often under active transcription.

About **92% of the human genome** consists of euchromatin.

2- Heterochromatin

It is a **tightly** packed form of chromatin (DNA and protein). It was once thought to be inaccessible to proteins and therefore **not actively transcribed**.



Classification Of Chromosomes depending on centromere:

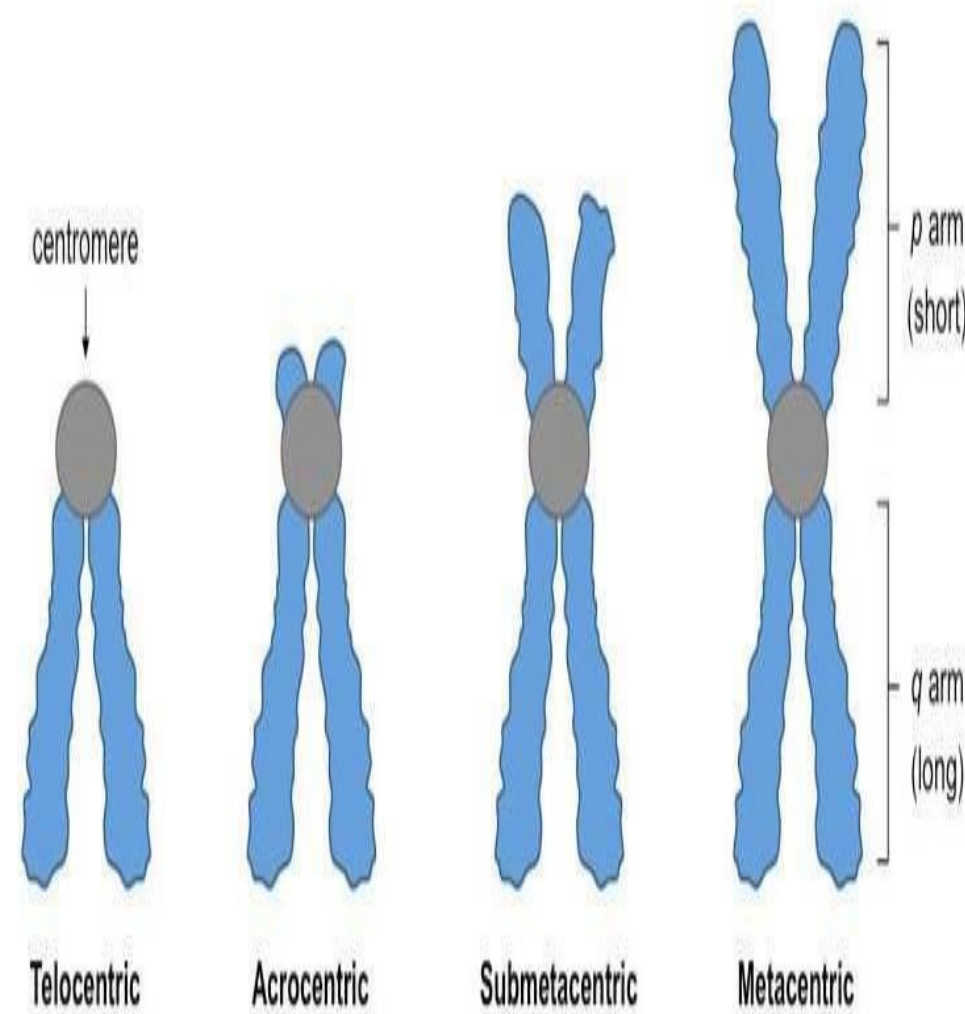
Based on the **position of the centromere**, chromosomes are divided into **four** categories.

1-Metacentric chromosome is one in which the centromere sits in the **center** of the chromosome and separates it into **two equal** arms.

2-Sub-metacentric: a chromosome's centromere is located **distant** from the central region. One arm is **much longer than** the other in this situation

3-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.

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



Classification chromosome according to the **number** of the centromere:

- Acentric : **without** any centromere
- Mono centric: **with one** centromere
- Dicentric : **with two** centromeres
- Polycentric: **with more** than two centromeres

Classification of Chromosome



Acentric



Monocentric



Dicentric



Polycentric

Types of Human Chromosomes

Type	Number	Function	Example
Autosomes	44 (22 pairs)	Control body traits and functions	Chromosomes 1–22
Sex Chromosomes	2 (1 pair)	Determine biological sex	XX (female), XY (male)

➡ Total = 46 chromosomes (23 pairs) in human somatic cells.

Functions of Chromosomes

1. Carry genetic information in the form of genes.
2. Ensure accurate distribution of DNA during cell division.
3. Regulate gene expression and cellular activities.
4. Protect DNA ends using telomeres.
5. Maintain genetic stability.

Karyotyping

- When the sets of chromosomes from a human are lined up according to size it can be seen that they exist in pairs. These are called **homologous pairs** because they are similar in structure. An image of such an arrangement of chromosomes is called the **karyotype**. In humans there are 23 **pairs** of chromosomes. The reason the chromosomes are in pairs is because one set of chromosomes comes from the female parent via the egg and the other set of chromosomes comes from the male parent by way of the sperm.

Karyotyping

Karyotyping is the process of **arranging and analyzing the chromosomes** of a cell according to their **number, size, shape, and banding pattern**. It is used to detect **chromosomal abnormalities**, such as **extra, missing, or structurally altered chromosomes**, and to determine **the sex of an individual**.

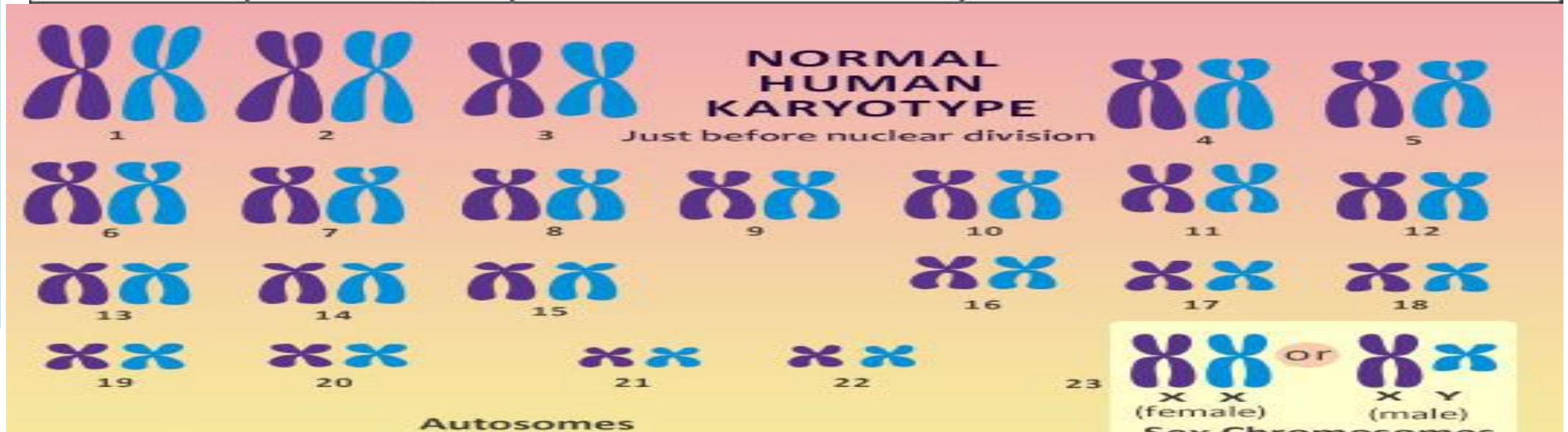
In karyotyping:

Number of autosome chromosome = 44 xx

Number of sex chromosome = 2 , xy in male and xx in female

Complete set of chromosome = 46

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



Any questions?

*Thank
You*