Identifying Normal and Abnormal Karyotypes

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Outline

- What is Karyotyping.
- Principle of Karyotyping
- Understanding a Normal Karyotype
- Identifying Abnormal Karyotypes
- Steps in Karyotype Analysis
- Applications of Karyotyping

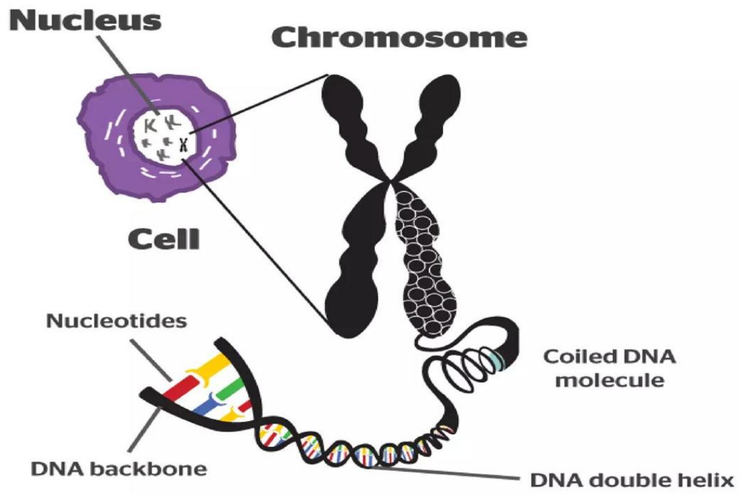
Objectives

- Understand the structure and classification of human chromosomes.
- Identify normal vs. abnormal karyotypes.

❖ Introduction

- Definition: Chromosomes are the rod-shaped, dark-stained bodies seen during the metaphase stage of mitosis.

- It is a tiny structure that look like a thread inside our cells that carry the genetic information.



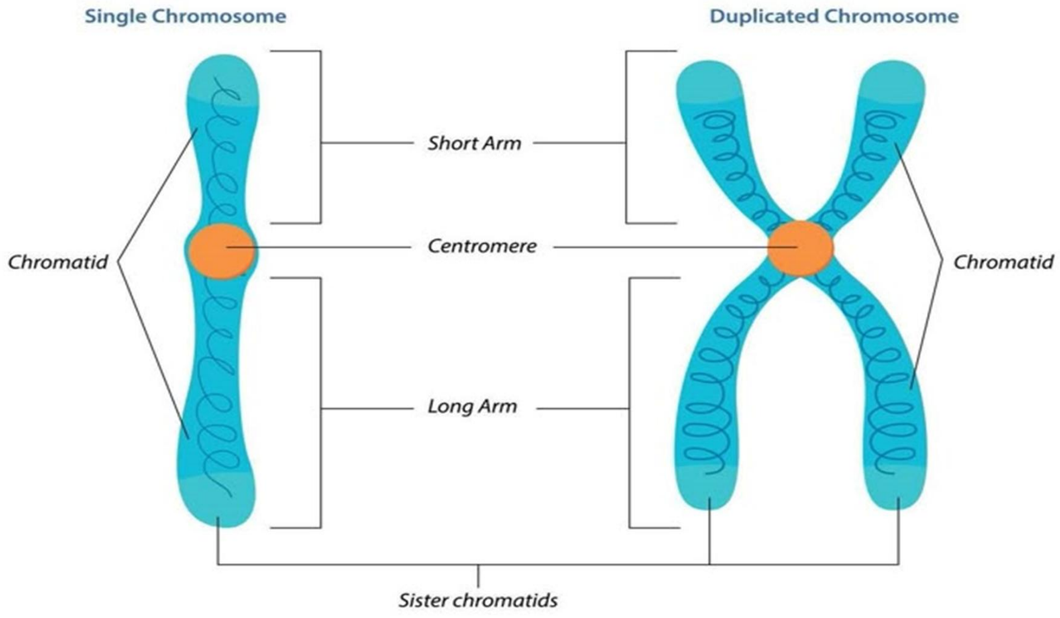
❖ Features of eukaryotic chromosome

- Chromosomes are best visible during metaphase.
- Chromosomes linearly bear genes.
- Chromosomes vary in shape, size, and number in different species of plants and animals.
- Chromosomes have the property of self-duplication and mutation.
- Chromosomes are composed of DNA, RNA, and protein.

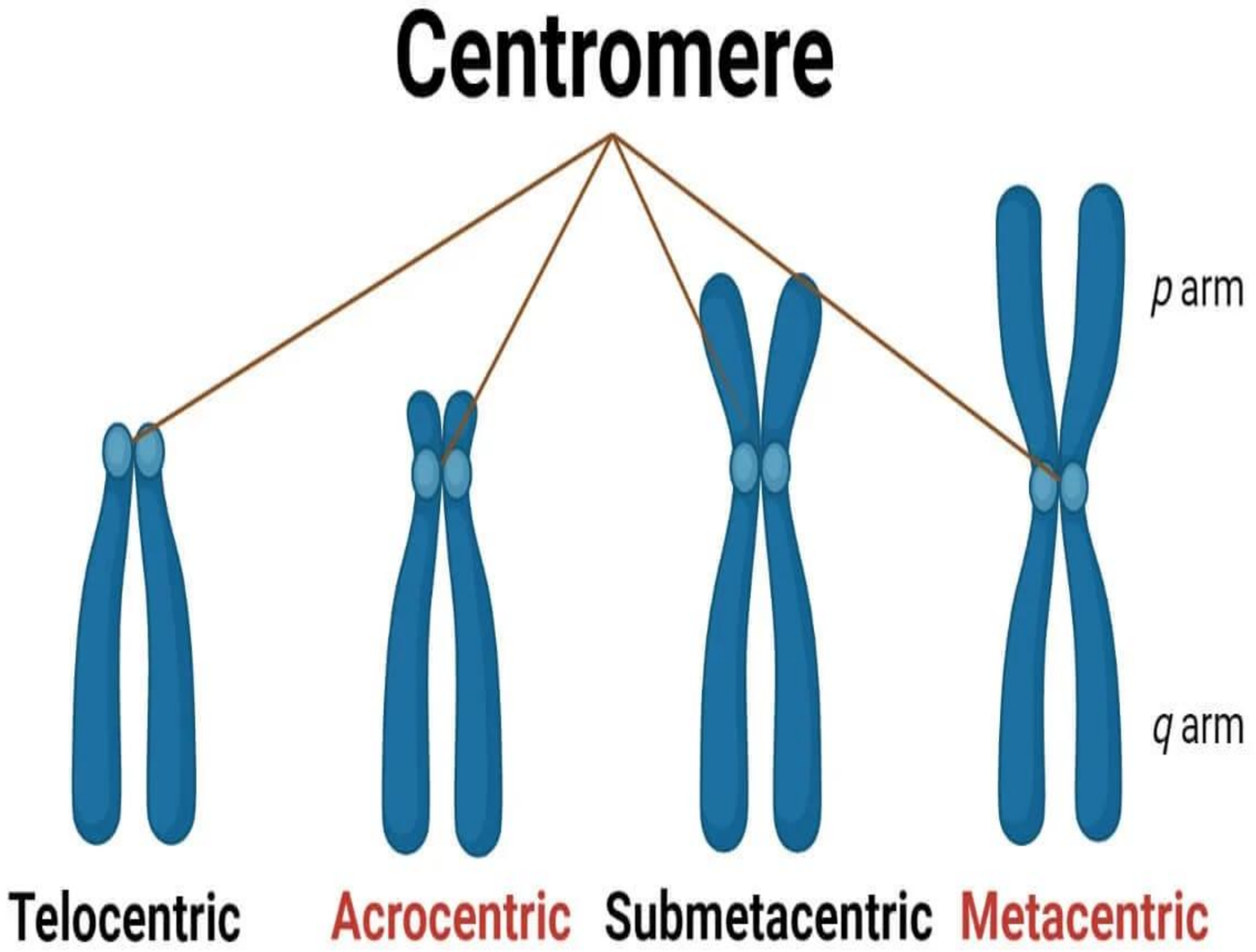
❖ Structure of Chromosomes

- Centromere: It is a part of chromosomes that links sister chromatids.

- Centromere gives the characteristic shape to the chromosomes and helps describe the location of specific genes.



❖ Types of Chromosomes



Metacentric
Chromosomes Centromere: Middle
Arms: Equal in length (p = q)
Shape: V-shaped
Example: Human Chromosome 1, 3, 16, 19, 20

Submetacentric
Centromere: Slightly off-center
Arms: One shorter (p) and one longer (q)
Shape: L-shaped
Example: Human Chromosome 2, 4-12

Acrocentric
Centromere: Near one end
Arms: Very short (p) and long (q)
Shape: J-shaped
Example: Human Chromosome 13, 14, 15, 21, 22

Telocentric
Centromere: At the terminal end
Arms: Only one visible arm
Shape: Rod-shaped
Example: Found in mice

❖ What is Karyotyping?

- Is a visual representation of an individual's chromosomes, organized in pairs according to **size**, **banding patterns**, and **centromere position**.

- It involves **visualizing**, **organizing**, and **analyzing** the chromosomes to identify **abnormalities in number, size, or structure**.

- It helps in identifying **genetic disorders**, **studying chromosomal abnormalities**, and **diagnosing conditions** related to chromosomal changes.

❖ Principle of Karyotyping:

- Human cells have **46 chromosomes** (**23 pairs**), including **22 pairs of autosomes** and **one pair of sex chromosomes** (XX or XY).

- Chromosomes are stained (commonly with **Giemsa stain**) to produce G-banding patterns, which help in identifying chromosomal abnormalities.

- A normal karyotype will have **two copies of each chromosome**, while an abnormal karyotype may have:

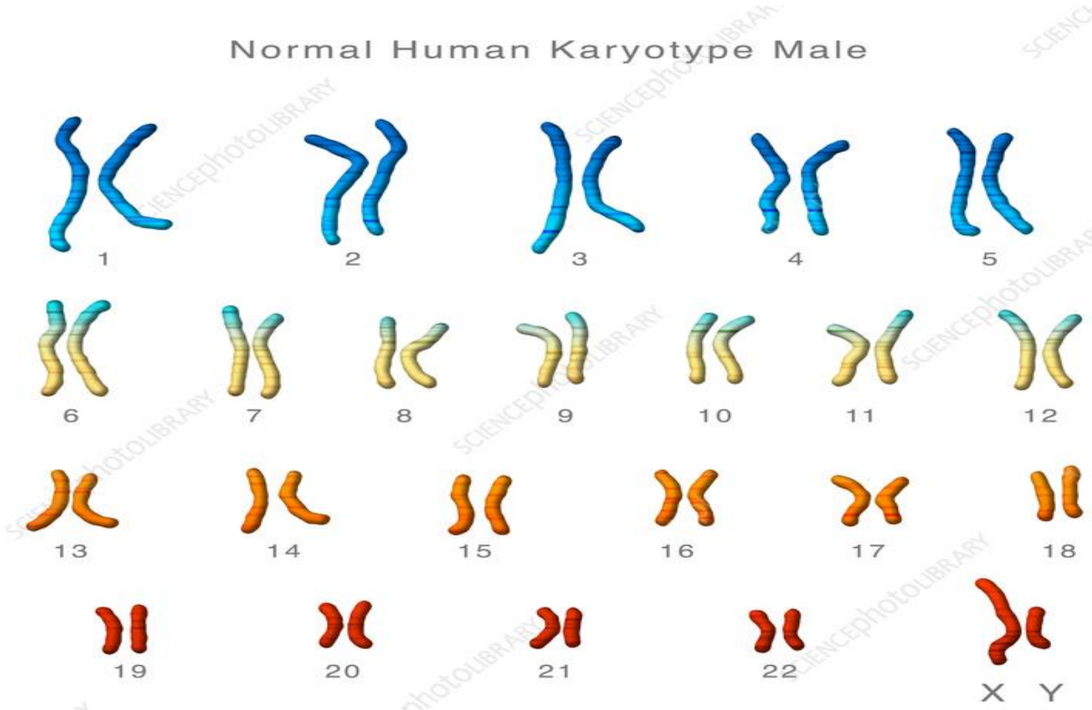
✓ Numerical abnormalities

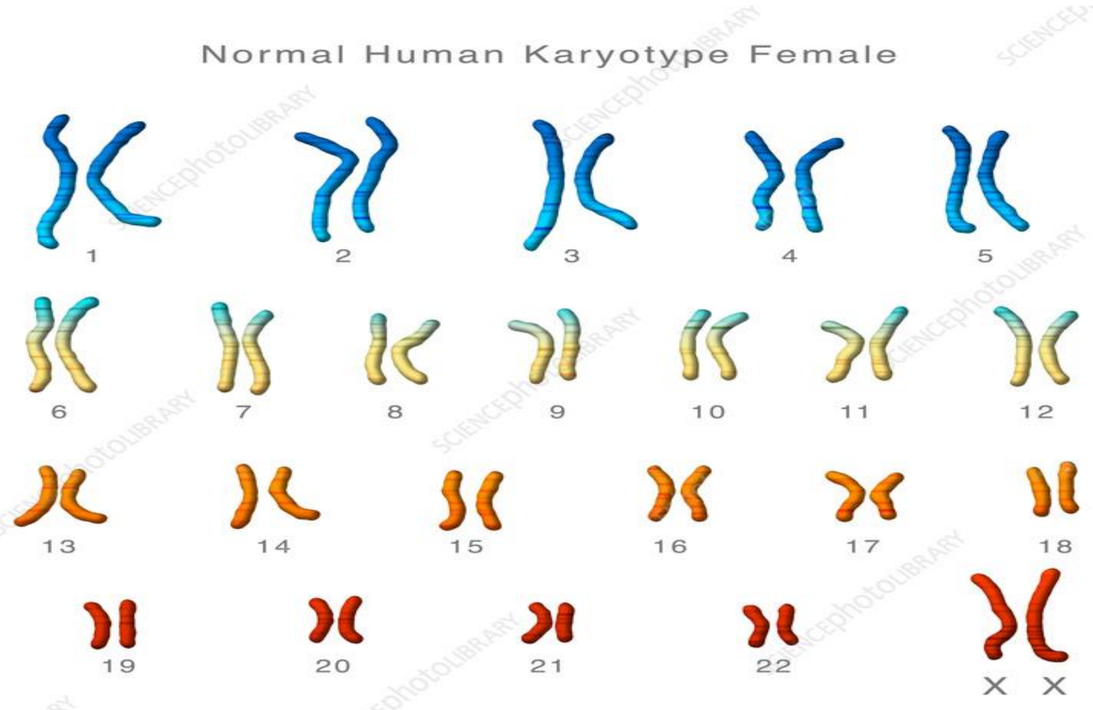
✓ Structural abnormalities

❖ Understanding a Normal Karyotype:

- A normal male karyotype: 46,XY (**44 autosomes** + **XY**).

- A normal female karyotype: 46,XX (**44 autosomes** + **XX**).

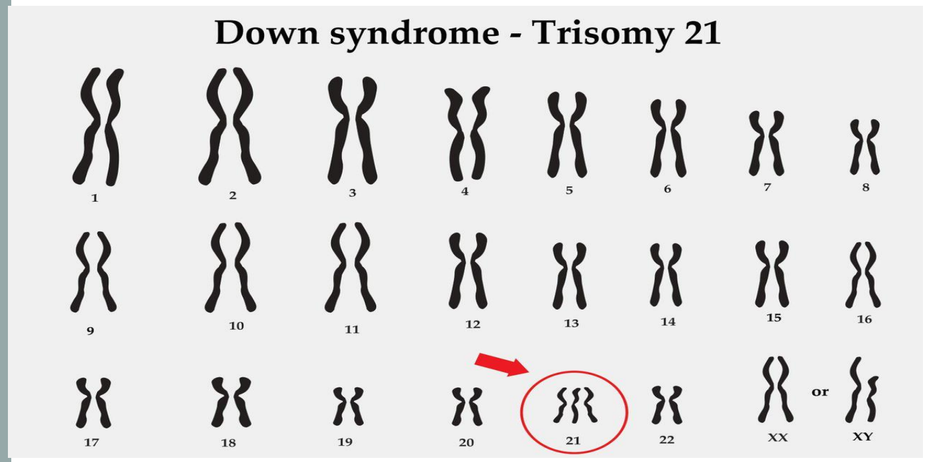


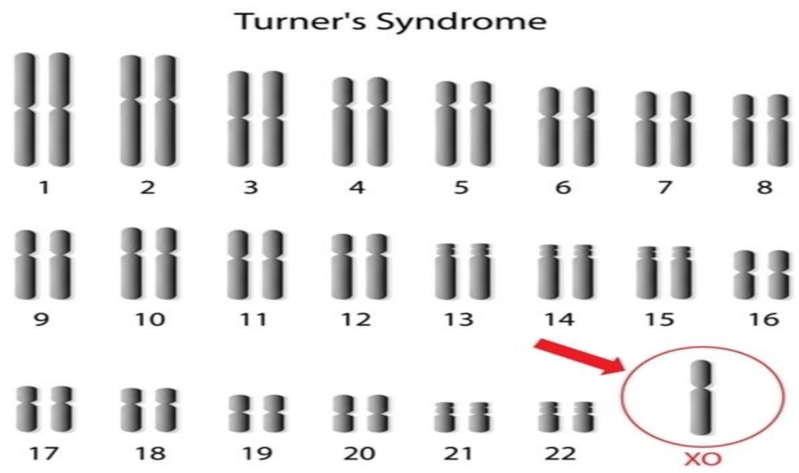


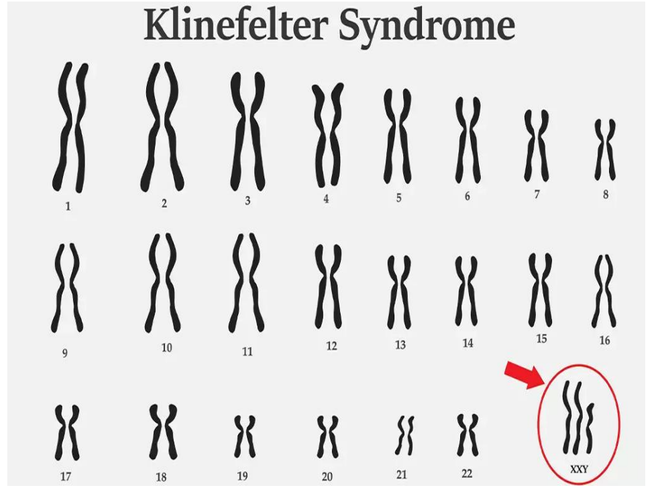
❖ Identifying Abnormal Karyotypes:

- **Numerical Chromosomal Abnormalities (Aneuploidy):**

- These involve a gain or loss of whole chromosomes, leading to conditions such as:

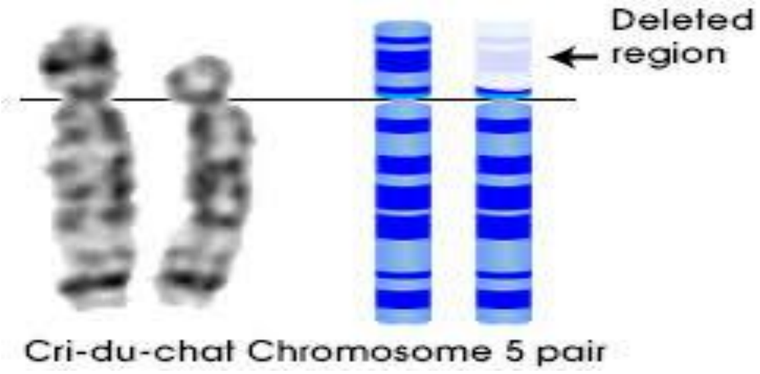


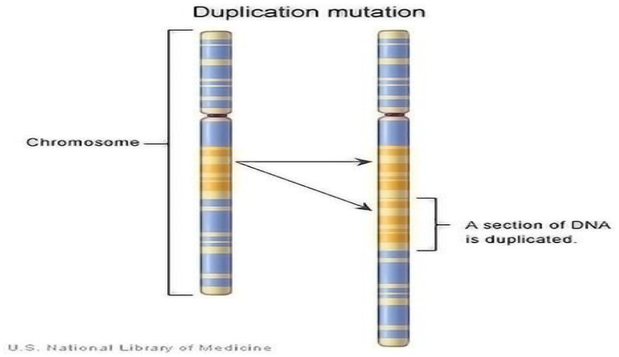


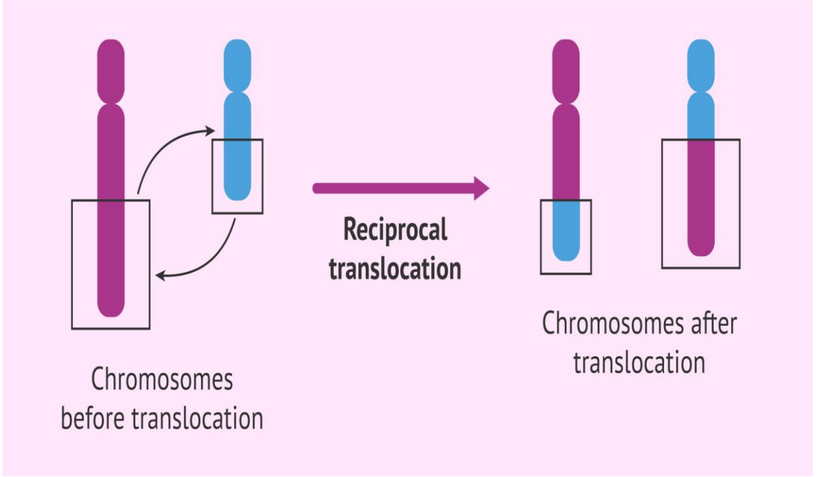


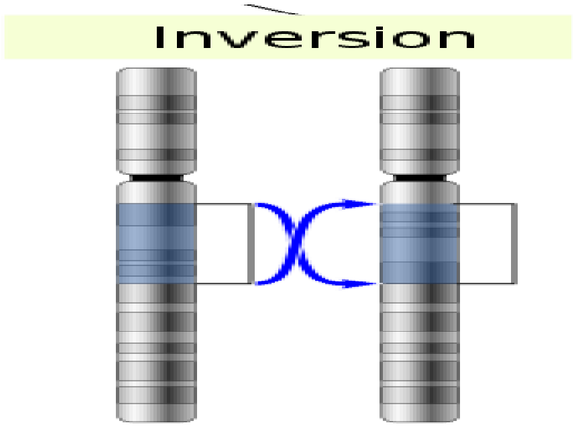
❖ Structural Chromosomal Abnormalities:

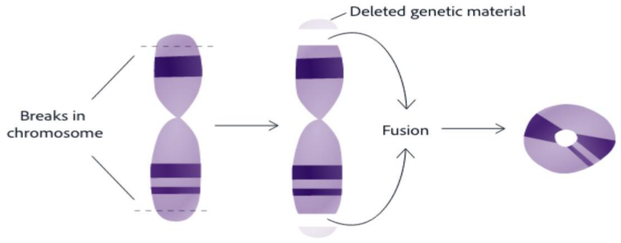
- These involve changes in chromosome structure without affecting the number.











❖ Important Steps in Karyotype Analysis:

- Arrange chromosomes into homologous pairs based on size and banding pattern.
- Check the total chromosome number (46 in normal karyotypes, abnormal in aneuploidies).
- Observe the shape and structure for deletions, duplications, or translocations.
- Determine the sex chromosome composition (XX for females, XY for males).
- Compare the karyotype with normal references to identify any abnormalities.

❑ Applications of Karyotyping:

- Prenatal Diagnosis.
- Cancer Diagnosis.
- Infertility Studies.
- Evolutionary Genetics.

References

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Thanks