Chromosome

The nucleus of a human cell contains all the genetic material necessary to direct all the functions in the body. The genetic material is arranged into chromosomes, which are structures that assist in the transmission of genetic information from one generation to the next. The instructions in each chromosome are contained within genes, which in turn are composed of DNA.

➢ Types of cells in the human body

1. Somatic cells: contain 46 chromosomes (diploid number; 44 autosomal + 2 sex chromosomes).

2. Germ cells (Gametes = sperm & ovum): contain 23 chromosomes (haploid number; 22 autosomal + 1 sex chromosome).

- Haploid: cell has only one set of chromosomes (n) like the gametes (sperm and egg), which contain 23 chromosomes each. These cells are produced through meiosis.
- Diploid: cell or organism that has two sets of chromosomes (2n), one set inherited from each parent. In humans, diploid cells contain 46 chromosomes (23 pairs). Most body cells (somatic cells) are diploid, and they undergo mitosis for growth and repair.

Chromosomes: are thread-like structures present in the nucleus, which carries genetic information from one generation to another. They play a vital role in cell division, heredity, variation, mutation, repair and regeneration. The term chromosome comes from the Greek words for color (chroma) and body (soma). Strausberger discovered chromosome in 1875. The term chromosome was coined by Waldeyer in 1888.

- Humans have 46 chromosomes, which occur in 23 pairs.
- 22 of these chromosome pairs are called autosomes, which are found in both males and females.
- One pair of chromosomes is called the sex chromosomes, because this pair contains the genes that control gender. Males have the sex chromosomes X and Y, and females have two X chromosomes.

- Each human cell contains about 2 meters of DNA; yet the cell nucleus is only 5–8 μm in diameter.
- In eukaryotic cells, very long double-stranded DNA molecules are packaged into chromosomes. The complex task of packaging DNA is accomplished by specialized proteins that bind to and fold the DNA, generating a series of coils and loops that provide increasingly higher levels of organization and prevent the DNA from becoming a tangled, unmanageable mess. Amazingly, the DNA is compacted in a way that allows it to remain accessible to all of the enzymes and other proteins that replicate it, repair it, and control the expression of its genes.

➢ Features of human chromosomes

1. Chromosomes are best visible during metaphase.
2. Chromosomes bear genes in a linear fashion
3. Chromosomes vary in shape, size and length
4. Chromosomes have property of self-duplication and mutation
5. Chromosomes are composed of DNA and protein

Functions of Chromosomes:

1. Chromosomes carry genetic material from one generation to the next.
2. Each chromosome has thousands of genes that code for proteins, which perform various functions.
3. They guide for growth, reproduction, repair, and regeneration, essential for survival.
4. Chromosomes protect DNA from damage and tangling.
5. Histone and non-histone proteins control gene expression by regulating DNA structure.
6. centromere move chromosomes during cell division by attach to Spindle fibers.

➢ Chromosome structures

Structurally chromosomes consists of 7 parts:

1) 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

2. Chromatin: Chromosome is made up of chromatin. Chromatin is made up of DNA, and proteins. At interphase, chromosomes are visible as thin chromatin fibers present in the nucleoplasm. During cell division, the chromatin fibers condense and chromosomes are visible with distinct features.

3. Centromere: It is a localized region of the chromosome with which spindle fibers attached is known as centromere or primary constriction or kinetochore, Centromeres consist of several hundred kilobases of repetitive DNA and are responsible for the movement of chromosomes at cell division.

4. Secondary constriction: some chromosome exhibits secondary constriction in addition to primary constriction. The region between the secondary constriction and the nearest telomere is referred to as a "satellite." Thus, chromosomes containing secondary constriction are known as "satellite chromosomes" or "sat-chromosomes." Chromosomes 13, 14, 15, 21, and 22 are well-known examples of these chromosomes.

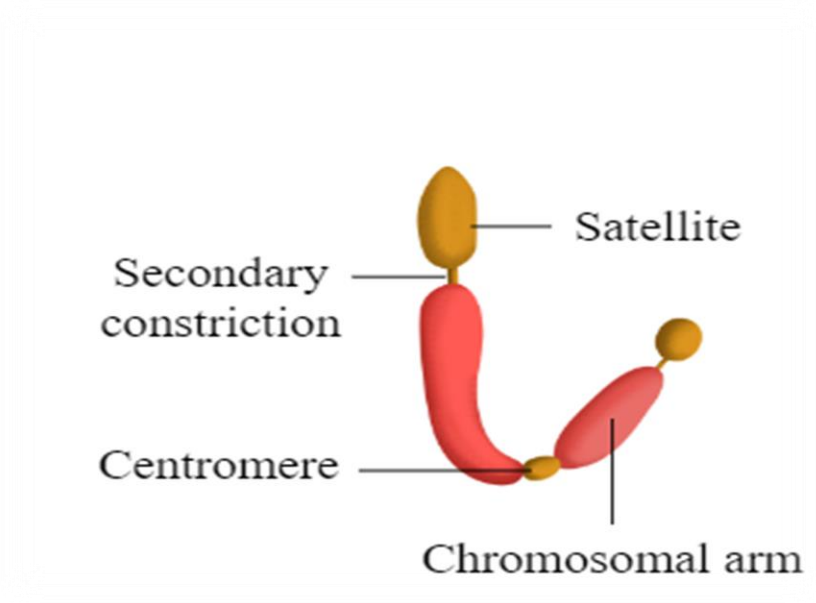


Figure (1) SAT chromosome and second constriction

Secondary constriction: is a phenomenon observed in some chromosomes, where an additional constriction exists in addition to the primary constriction represented by the centromere. This secondary constriction is significant for the organization and function of chromosomes, influencing DNA replication and gene distribution.

5.Telomere: are the ends or the tips of chromosomes , acting as "caps" that protect the chromosome ends and maintain their stability. These telomeres play an important role in preventing chromosomes from sticking together and protect the genetic information contained within the chromosome.

Telomeres are in humans, they consist of repeated sequences of DNA (TTAGGG), which help support their function.

During cell division and DNA replication, there is a natural shortening of telomeres with each division, as the 5′ end of the DNA strand is not fully replicated. This is where the enzyme "telomerase" comes into play, which works to compensate for this shortening by rebuilding the telomeres, allowing the cell to continue dividing. Over time, due to the lack of telomerase activity in most cells, telomeres become shorter and shorter until they reach a critical length. When this happens, the cell loses its ability to divide and enters a phase of senescence, which is a normal part of the aging process.

In some tumors, telomerase activity increases abnormally, allowing cancer cells to continue dividing without limits, bypassing normal aging boundaries. This abnormal activity is one of the reasons cancer cells proliferate rapidly and continuously, making it more difficult to control tumor growth.

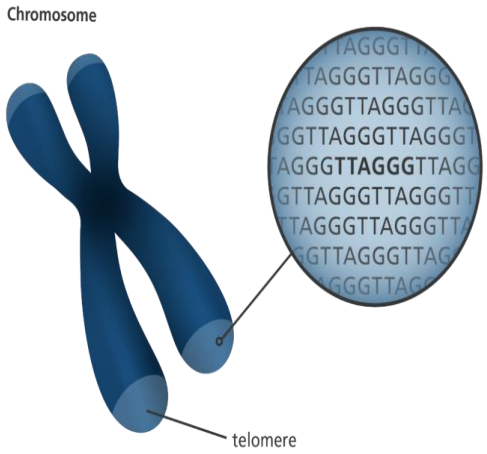


Figure (2) telomere

➢ Classification of chromosome :

Individual chromosomes differ not only in the position of the centromere, but also in their overall length. Based on the three parameters of length, position of the centromere, and the presence or absence of satellitesearly pioneers of cytogenetics were able to identify most individual chromosomes.

A. classification of chromosomes according according to the amount of the DNA

1) Single-chromosomes (s-chromosomes): made of one DNA molecule (chromatid) found during interphase (G1 phase)
2) Double-chromosomes (d-chromosomes; mitotic chromosomes): are formed during the S phase. Each d-chromosome is formed of two chromatids, linked at the centromere. Each chromatid is made of a DNA molecule.
3) Homologous chromosome (meiosis-1 chromosomes): Homologous chromosomes are pairs of chromosomes that have the same length, gene position, and centromere location. One chromosome in each pair is inherited from the organism's mother, and the other from the father.

B. classification of chromosomes according to position of the centromere:

*The shape of chromosome is generally determined by the position of centromere

1-metacentric: in which the centromere is located approximately in the middle, and so the chromosome has two arms of equal length.

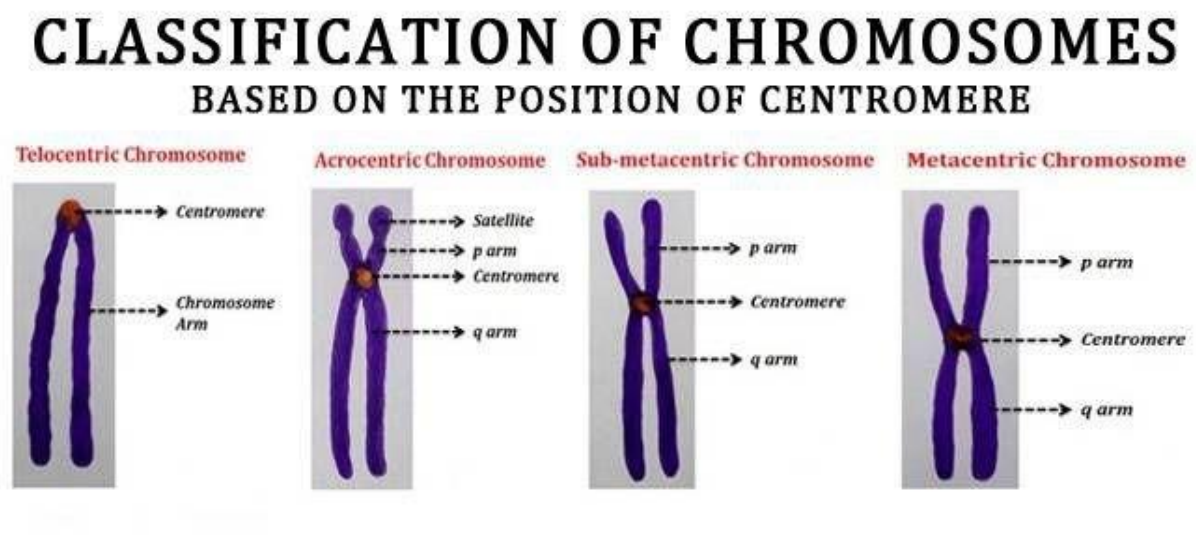
2-submetacentric: in which the centromere is displaced toward one end, creating a long arm and a short arm.

3-acrocentric: in which the centromere is near one end, producing a long arm and a knob, or satellite.

4-telocentric: in which the centromere is at or very near the end of the chromosome.

*On human chromosome, centromere divides into :

- The short arm is designated by the letter p (= petite)
- The long arm is designated by the letter q. (‘g’ = grande),



THE SEX CHROMOSOMES

- The X and Y chromosomes are known as the sex chromosomes because of their crucial role in sex determination.
- The X chromosome was originally labeled as such because of uncertainty as to its function when it was realized that in some insects this chromosome is present in some gametes but not in others.
- In human, the Y chromosome is much smaller than the X and carries only a few genes of functional importance, most notably the testis-determining factor, known as SRY.
- In the female each ovum carries an X chromosome, whereas in the male each sperm carries either an X or a Y chromosome. As there is a roughly equal chance of either an X-bearing sperm or a Y-bearing sperm fertilizing an ovum, the numbers of male and female conceptions are approximately equal. In fact, slightly more male babies are born than females, although during Childhood and adult life, the sex ratio evens out at 1: 1.