Human chromosome Nomenclature

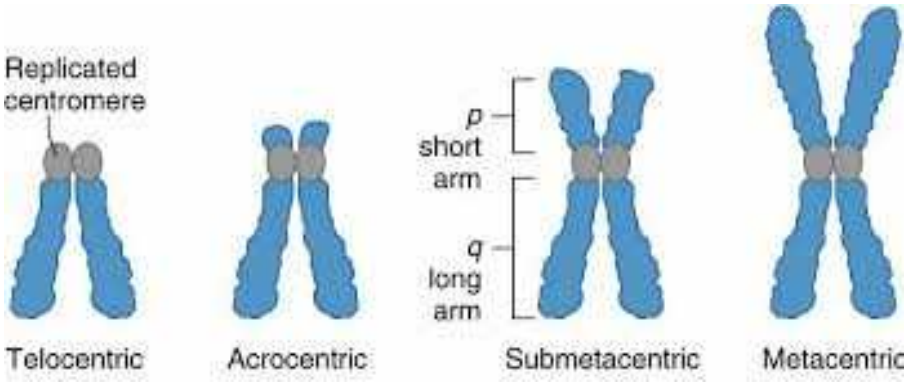
- In humans, each cell normally contains **23 pairs of chromosomes,** Twenty-two (22) of these pairs, called **autosomes**, look the same in both males and females. The 23rd pair, the sex chromosomes, differ between males and females.
- Females have two copies of the X-chromosome, while males have one X and one Y chromosome.
- A chromosome may be:

1- Metacentric, with its centromere in the middle;

2- Sub metacentric, with the centromere closer to one end of the chromosome

3- Acrocentric, in which the centromere is near one end of the chromosome and the short arm is essentially comprised of repetitive DNA that constitutes the satellites and nucleolar organizing regions.

4- Telocentric, a chromosome in which the centromere is terminal (situated at one end), so that there is effectively only one arm.



Chromosome groups:

Group A: chromosome number 1 – 3 , large metacentric

Group B: chromosome number 4 and 5 , Large submetacentric

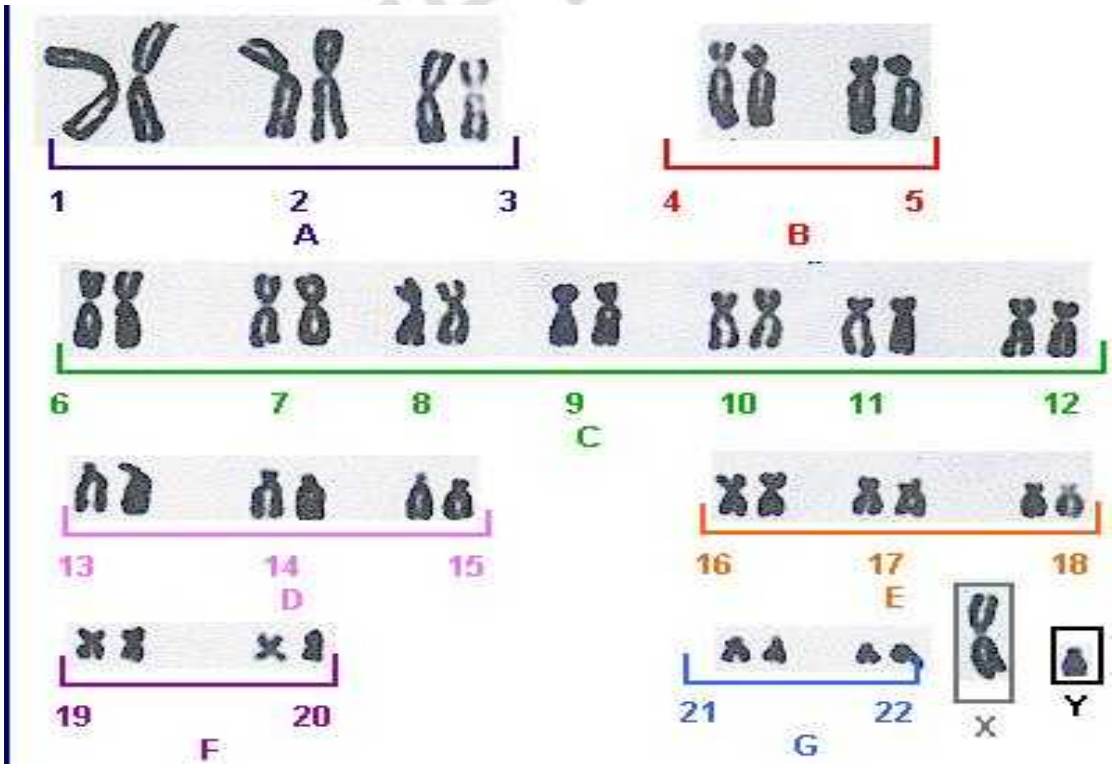
Group C : chromosome number 6-12 and chromosome X , Medium submetacentric

Group D : chromosome number 13, 14 , 15 , Medium acrocentric

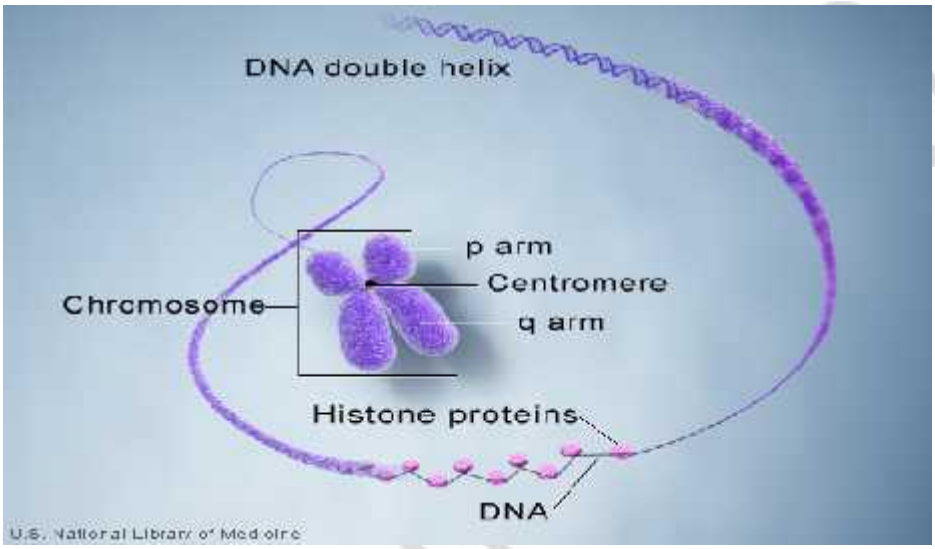
Group E : chromosome number 16 Short metacentric and 17 , 18 , Short sub metacentric

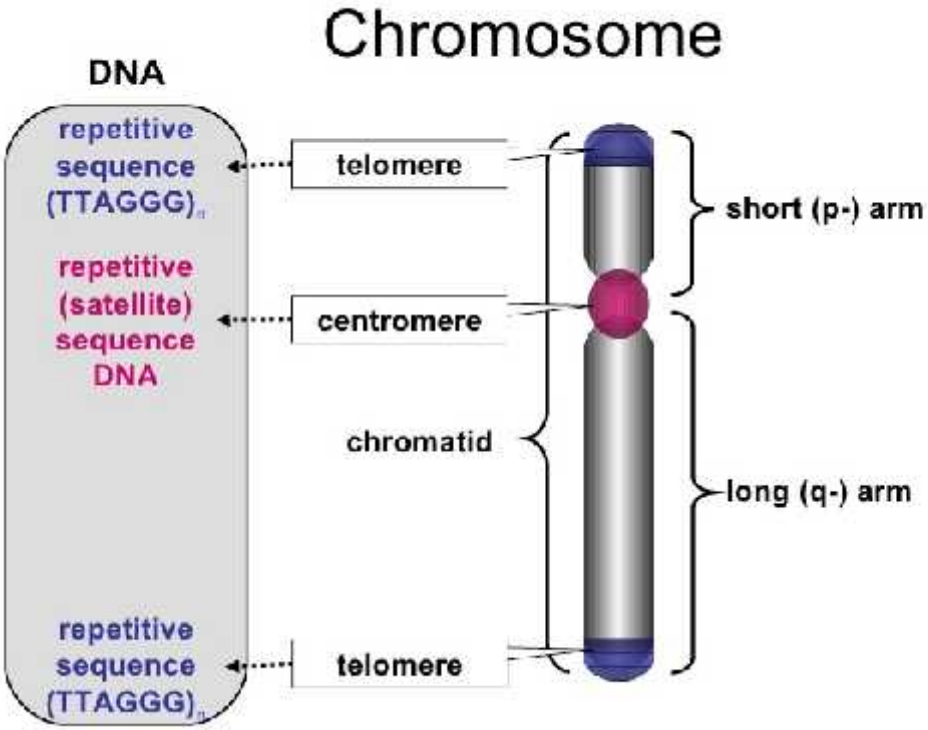
Group F : chromosome number 19 , 20 Short metacentric

Group G : chromosome number 21 , 22 and chromosome Y , Short acrocentric



- Each chromosome has a constriction point called the **centromere**, which 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.”

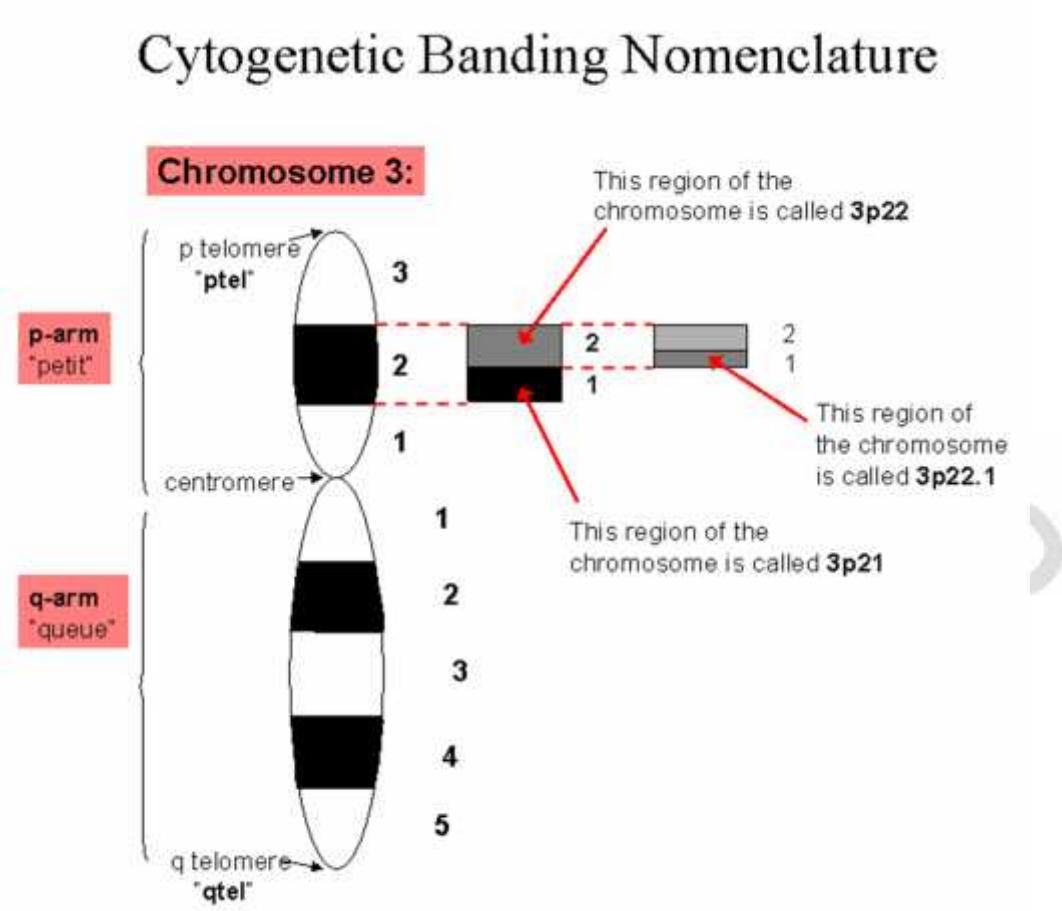




- The location of the centromere on each chromosome gives the chromosome its characteristic shape, and can be used to help describe the location of specific genes.

Each chromosome arm is divided into regions, or cytogenetic bands, that can be seen using a microscope and special stains. The cytogenetic bands are labeled p1, p2, p3, q1, q2, q3, etc., counting from the centromere out toward the telomeres.

At higher resolutions, sub-bands can be seen within the bands. The sub-bands are also numbered from the centromere out toward the telomere.



- For example, the cytogenetic map location of the CFTR gene is 7q31.2, which indicates it is on chromosome 7, q arm, band 3, sub-band 1, and sub-sub-band 2.
- The ends of the chromosomes are labeled ptel and qtel. For example, the notation 7qtel refers to the end of the long arm of chromosome 7.

The commonly used **G-, Q-, and R-banding** techniques show bands distributed along the entire chromosome, whereas the C-, T-, or NOR-banding techniques are used to identify specific chromosome structures that are heritable features

A band is defined as a part of the chromosome that is clearly distinguishable from its adjacent segments based on its staining properties.
As a general rule, a chromosome band contains ~5–10 mega bases (Mb) of DNA.

Giemsa or G-banding is the most common banding method employed in North American cytogenetics laboratories.

Facilitate the identification of structural abnormalities G-dark (positive) bands are AT rich, gene poor, and late replicating.

The early replicating G-light (negative) bands are GC rich, gene rich, and late replicating

C-banding

is particularly useful when identifying the morphologically variable heterochromatin regions of the Y chromosome and chromosomes 1, 9, and 16.

Reverse or R-banding

are useful for analyzing deletion or translocation that involve the telomeres of chromosome.