Medical genetics
Senior / First Lab

WHAT ARE MEDICAL GENETICS?

Medical genetics is a field of study that focuses on genetic and techniques to diagnose and manage genetic disorders and diseases in individual. As a result of rapid progress in molecular genetics, DNA-based diagnosis has been accessible for numerous inherited conditions, and gene therapy the insertion of normal genes into patients to correct genetic disease is showing promise for some conditions.

Basic Cell Biology: Structure and Function of Genes and Chromosomes.

All genetic diseases are characterized by cell-level defects. For this reason, one must understand basic cell biology to under- stand genetic disease. Errors can occur in the replication of genetic material and translation of DNA into proteins. Such errors frequently result in single-gene disorders. Errors that occur during cell division can also result in chromosomal disorders.

Chromosome

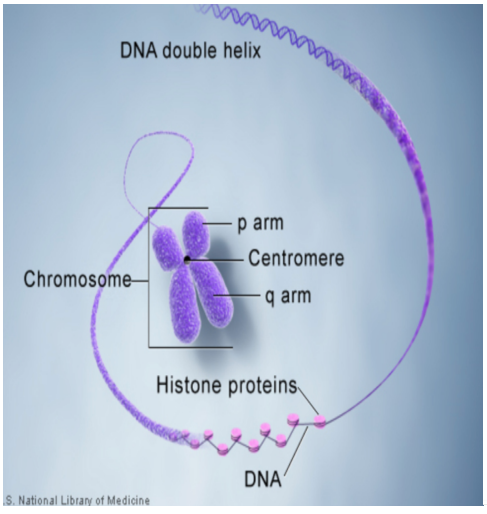
A chromosome is an organized structure rod shaped filamentous bodies present in the nucleus, which become visible during cell division, it's structure of DNA and protein that is found in cell. Contains many genes, regulatory elements, and other nucleotide sequences.

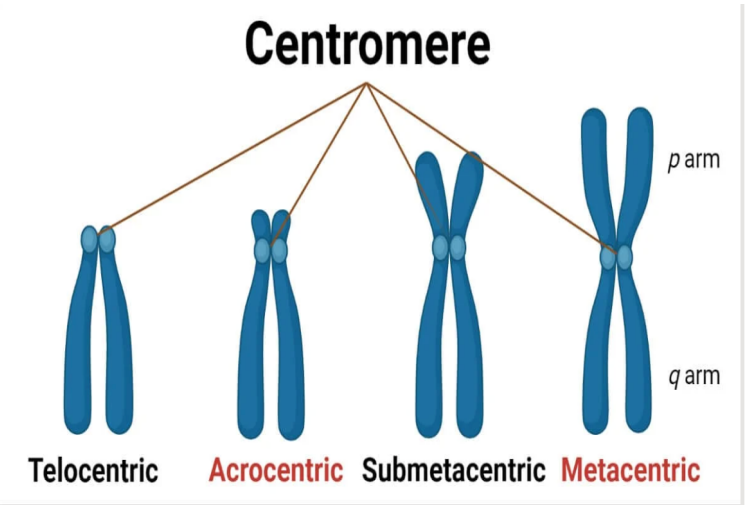
STRUCTURE OF CHROMOSOME

The size of the chromosome is normally measured at mitotic metaphase in the continuous process of cell growth and cell division.

Chromosomes occur in the form of thin, coiled, and contractile, thread-like stainable structures of two symmetrical structures called chromatids that are attached to each other.

Classification of chromosomes morphologically, **chromosomes are classified according to the location of the centromere.**

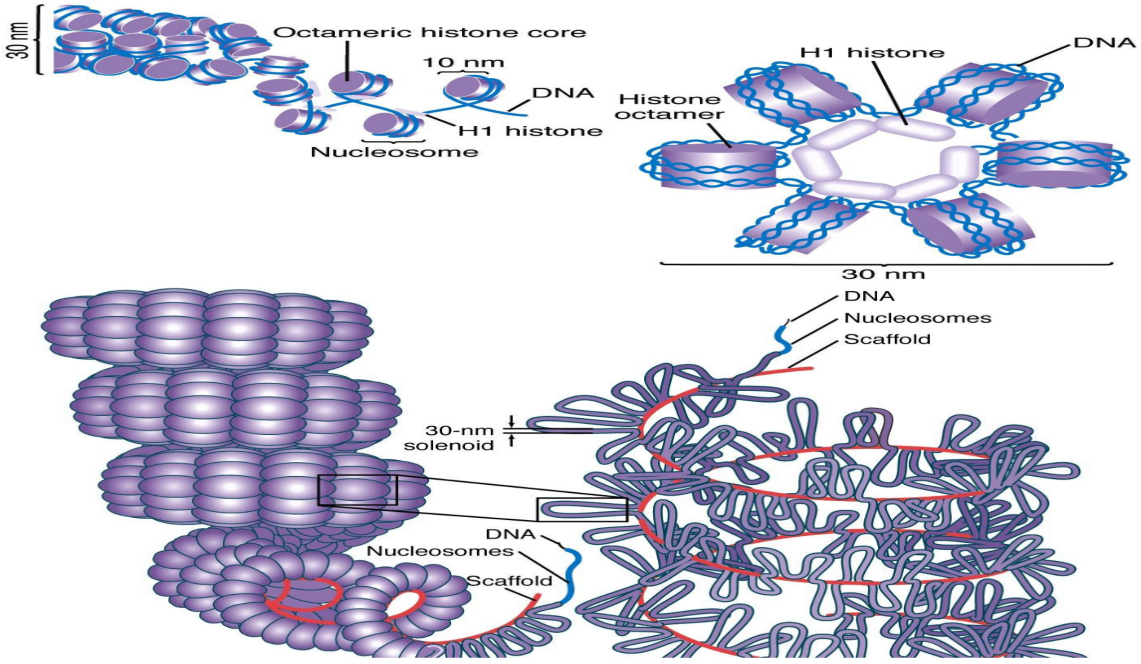




Eukaryotic

- Genomes are composed of multiple chromosomes each containing a linear molecule of DNA. DNA consists of nucleotides while the primary protein component is histones
- Histones are unique features of eukaryotic cells.
- The packaging of DNA is due to histones.

Histones proteins basic (+ charged lysine and arginine) amino acids that bind DNA, The **DNA** molecule is wrapped twice around a **Histone Octamer** to make a **Nucleosome**. Six Nucleosomes are assembled into a **Solenoid** in association with **H1 histones**. The solenoids are in turn coiled onto a **Scaffold**, which is further coiled to make the **chromosomal matrix**. Fore core histones in nucleosomes Two of each of H2A, H2B, H3A and H4 fifth h, H1 is the linker histone appearance that chromatin fibers have a beaded appearance, with the beads at intervals of approximately 200 base Paris



Number of chromosomes

All the individuals of species have a specific number of chromosomes which distinguishes it from other species. Presence of a whole set of chromosomes is called **euploidy**, Gametes normally contain only one set of chromosomes this number is called **haploid**

Somatic cells usually contain two sets of chromosomes-2n: Diploid 3n- called triploid while 4n-called tetraploid. The conditions in which the chromosomes set are present in a multiple of "n" is called polyploidy

Variation in chromosome number

Aneuploidy, monoploidy, and euploidy are all ways in which the number of chromosomes can

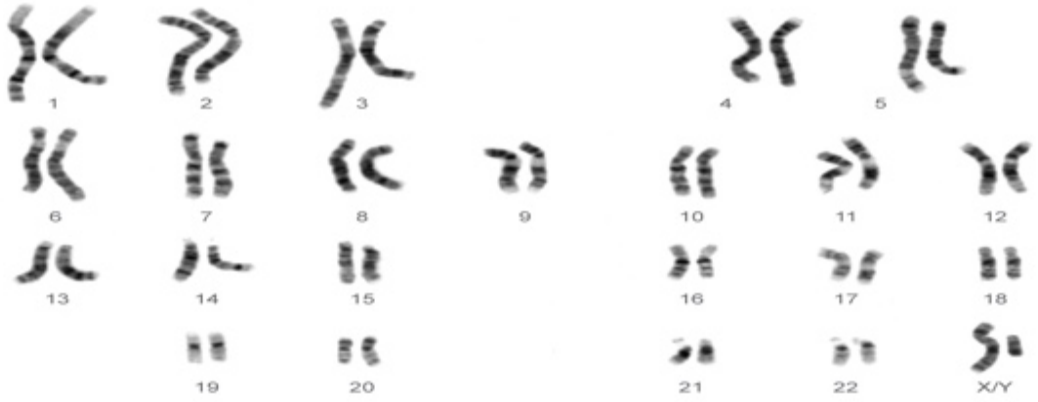
change. Change can occur by the addition of all or part of a chromosome (**aneuploidy**), the loss of an entire set of chromosomes (**monoploidy**) or the gain of one or more complete sets of chromosomes (**euploidy**). Each one of these disorders deviates from the typical diploid number of chromosomes. Each of these conditions is a variation on the normal diploid number of chromosomes can have a significant impact on phenotypic expression.

The different conditions of aneuploidy are:

1. **Nullisomy** - the loss of both pairs of homologous chromosomes; individuals are called nullisomics and their chromosomal composition is (2n-2)
2. Monosomy - the loss of a single chromosome; individuals are called monosomics and their chromosomal composition is (2n-1)
3. **Trisomy** - the gain of an extra copy of a chromosome; individuals are called trisomics and their chromosomal composition is (2n+1)
4. **Tetrasomic** - the gain of an extra pair of homologous chromosomes; individuals are called tetrasomics and their chromosomal composition is (2n+2).

Karyotype analysis

Somatic cells in human have 46 chromosomes, cells studied before division where chromosomes arrange in pairs, the member of each pair have the same size, shape and constriction (location of kinetochore) also had the same banding pattern. This is called **karyotype analysis.**



Regions of chromosomes
There are several different chemical techniques are used to identify certain chromosomal regions by staining them.

There are 2 types of regions in the chromosomes.

Euchromatin
- Light staining regions
- Lightly packed form of chromatin and is often under active transcription

Heterochromatin

- Darkly staining and tightly coiled throughout the cell cycle and that is for the most part, genetically inactive

Constitutive heterochromatin

- It is fixed and irreversible in form and function
- It does not reverse to the euchromatic stage
- Chromosomes 1, 9, 16 and the Y chromosomes contain regions of constitutive heterochromatin

Facultative heterochromatin

- It has the faculty to return to the normal euchromatic state
- Consists of euchromatin that takes on the staining and compactness characteristics of heterochromatin during some phase of development
- The inactive X chromosomes is made up of facultative heterochromatin

Autosomes and allosomes

Chromosomes that are not directly concerned with reproduction and sex determination are called **autosomes.**

Allosomes/heterosome

- These chromosomes are directly associated with reproduction and differ from autosomes in size, form, and behavior.
- Usually there is a single pair of allosomes in mammals termed as “X” and “Y” chromosomes
- In bugs of Heteropteran like locusts, the female has two X chromosomes while the male has one X. The Y chromosomes in absent in these species

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