## History of cytogenetics

The beginning of human cytogenetics attributed to Walther Flemming an Austrian cytologist and professor of human chromosomes in 1882.

**Flemming** referred to the stain able protein of the nucleus as chromatin and first used the term Mitosis.

**Weldely** introduced the word chromosome (Greek words for "colored body" after discovery of Menedilian inheritance in 1900.

**Sutton** developed chromosome theory of inheritance and combined cytology and genetics when he referred to the study of chromosomes as cytogenetics.

In 1923, **Painter** studied meiotic based on his work on chromosome derived from testicles of men. Reported human diploid chromosome number to be 48 chromosomes then corrected to 46 chromosomes and proposed X and Y chromosome (sex chromosomes).

**Levitsky** formulated the term karyotype refer to the ordered arrangement of chromosomes.

**Cytogenetics:** is a branch of genetics that is concerned with the study of the structure and function of chromosome.

In addition, cytogenetics is a fusion science, joining cytology (study of the cell) with genetics (the study of inherited variation).

**Cytogenetics types:**

1- Physiological Cytogenetics
2- Molecular Cytogenetics
3- Immune Cytogenetics
4- Biochemical Cytogenetics

Other classification depended on the aim of the study

1- Human Cytogenetics
2- Animals Cytogenetics
3- Plants Cytogenetics

**Chromosome** is a thread like structure found in the nuclei of both animal and plant cells. They made protein and one molecule DNA.

**Chromosome structure:**

Chromosome consists of two sister chromatids. Each of which is comprised of compacted double helix of DNA.

- **Centromere:** is a constriction visible on metaphase chromosomes where the two sister chromatids are joined. It is function of centromere is essential to the survival of chromosomes during cell division.

- **Telomeres**: are the physical ends of chromosomes act as protective cap to chromosome ends. Preventing end-to-end fusion of chromosomes and DNA degradation resulting after chromosome breakage. Telomere contain tandem repeates of TTAGGG.

- **Chromatin**: complex DNA and protein found in euokaryotype nucleus that packages chromosomes.

  Chromosomes vary in shape and size and vary in the shape depending position of centromere.

1- **Metacentric**: the centromere in the middle of chromosome and two arm is equal length.
2- **Sub metacentric**: the two arms vary in length.
3- **Acrocentric**: the centromere close to the end.

**DNA: Deoxyribonucleic acid**

DNA is the raw material of inheritance. Watsen and Crick proposed molecular structure of DNA in 1953 using X- ray, and proposed the double helix: a twisted spiral ladder structure consisting of two long chains wound around each other and held together by hydrogen bonds. Each nucleotide consists of deoxyribose sugar, phosphate group and one of four nitrogen bases. (Adenin, Guanine , Cytosine and thymine).

DNA synthesis Occurs in a process requires many enzymes and cofactors, called DNA replication.

Protein synthesis called Transcription, during the transcription of DNA copied into a complementary piece of mRNA modified by removal of introns segment of the next step in protein synthesis, the last called translation. During translation by using the newly transcribed mRNA molecules as template with help of third ribonucleic acid, tRNA.

tRNA contain anticodons that complementary to the codons on mRNA. This type of tRNA transfer the specific amino acids to the synthesizing protein chain. Amino acid joined to this chain by peptide bonds this process is continued until a stop codon is reached the new protein molecule is then released into the cell milieu.

## Cell division

There are two types of cell division

- Mitosis
- Meiosis

**Mitosis**: is the division of somatic cells, whereas meiosis is the special type of division that occurs only in gametes .

**Mitosis:**

1- Prophase
2- Prometaphase
3- Metaphase
4- Anaphase
5- Telophase

1- Prophase: Chromosomes begin to coil, condensed, and become visible to as discrete.
2- Prometaphase: Short period between prophase and metaphase. Spindle fibers begin to appear and chromosomes attach to spindle fibers at their kinetochores.
3- Metaphase: During metaphase, centrioles divide and move to opposite poles then reached their maximum state of contraction during this phase.
4- Anaphase: centromeres divide and chromatids separate during this stage. Sister chromatids migrate to opposite poles.
5- Telophase: final stage, chromosomes uncoil and become indistinguishable again. This stage followed by cytokinesis (cytoplasmic division).

Product of mitosis is two identical daughter cells, poses the same number of chromosomes.

**Meiosis:** is a process takes place only in ovaries and testes. Involving one duplication of DNA and two cell divisions II) and (I. Reduced the number of chromosomes from the diploid number (2n) to haploid number (1n).

**Meiosis I:**

1- Prophase I : complex stage that is further subdivided as follows :

A- Leptotene
B- Zygotene
C- Pachytene
D- Diplotene

Leptotene: 46 chromosomes, each chromosome contain two chromatids. The chromosome begin to condense but are not yet visible by light microscope.

Zygotene: chromosomes appear thread-like structures, pair locus for locus. This pairing called Synapsis. The synaptonemal complex is necessary for the phenomenon crossing-over, which take place later.

Pachytene: synapsis is complete during pachytene, chromsomes continue to condense and now appear thicker threads. The phenomenon of crossing – over takes place during this stage.

Crossing – over: segments of DNA are exchanged between non-sister chromatids of the bivalents. The result of crossing – over is recombination of genetic material and creating new combinations of genes in daughter cells.

Diplotene: chromosomes continue to shorten and thicken then the homologous chromosomes begin to repel each other, this point (held together) called chiasmata.

Metaphase I: formation meiotic spindle, bivalents oriented toward opposite poles.

Anaphase I: centromeres of each bivalents, sparate and migrate to opposite poles.

Telophase I: two haploid sets of chromosomes reach oppsite poles and the cytoplasm divides. The result is two cells containing 23 chromosomes.

**Meiosis II**: the cells move directly from telophase I to metaphase II with no prophase.

Meiosis II proceeds like mitotic cell division except that each cell contains only 23 chromosomes.
23 chromosomes each other contain two chromatids, the chromatids separate and move to opposite poles in anaphase II.

**Cytokinesis occurs in telophase II**

The result of meiosis II is four cells, each cell contains 23 chromosomes, each of chromosome contain single chromatids, and the new cells differs genetically from the original cell.