GENETICS

- Heredity - is the transmission of characteristics from parent to offspring through the gametes.
- Gene - a unit of heredity, a segment of a DNA that contains all the information required for synthesis of polypeptide chain. Each gene has a specific position (locus) on a chromosome.
- Allele- an alternate form of a gene, that can occupy a particular chromosomal locus. In humans and other diploid organism there are two alleles, one on each chromosome of a homologous pair.
- An individual who has two identical alleles for a gene at a particular locus on homologous chromosomes is homozygous for that gene. An individual with two different alleles at a particular locus on homologous chromosomes is heterozygous.

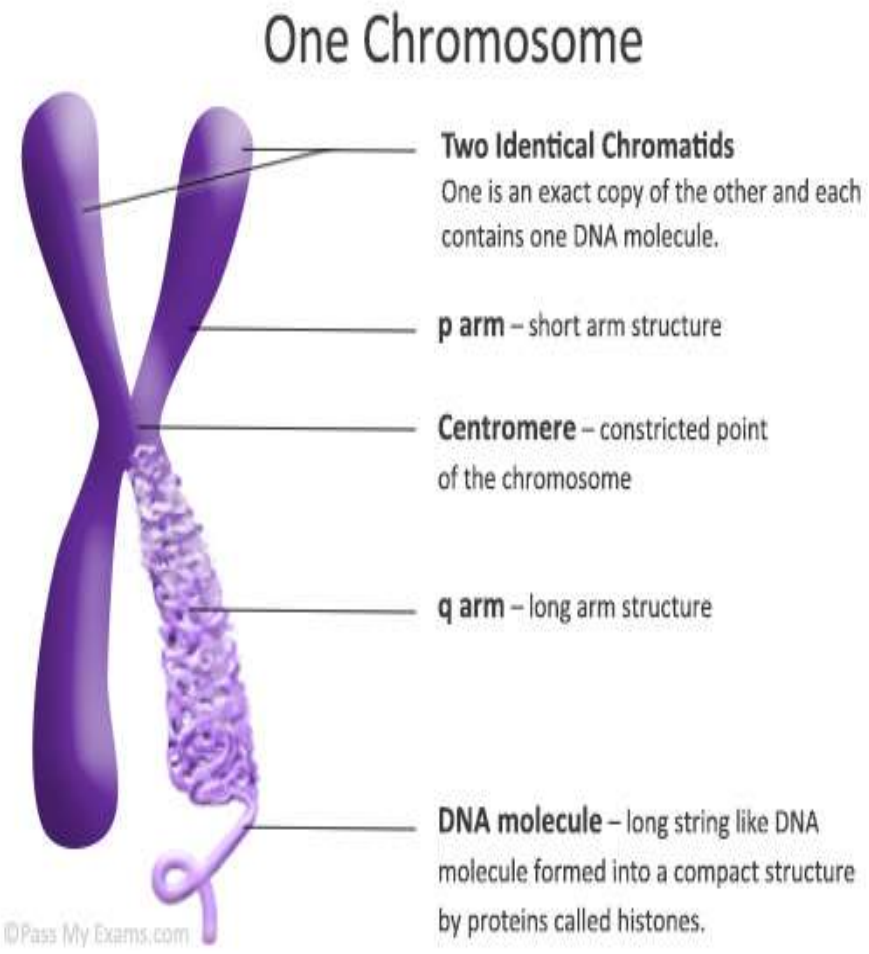
- INHERITANCE - is the way of passing of hereditary information, which depends on the forms of reproduction. During asexual reproduction the main traits are inherited through spores or vegetative cells, thats why the maternal and daughter cells are very similar. During sexual reproduction the main traits are inherited through gametes.
- Genotype- is the genetic constitution of an organism (a diploid set of genes).
- The genotype describes the organisms alleles, whole the phenotype describes the outward expression (physical appearance of an individual) of an allele combination.
- The phenotype of an individual is the observable outward manifestation of the genes that it carries.
- Genome - is a collection of genes of an organism in cells (may be a haploid set of genes).

Chromosomes

- 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.
- In Eukaryotic cells, genetic material is present in the nucleus in chromosomes, which is made up of highly organized DNA molecules with histone proteins supporting its structure.
- A human cell contains total 23 pair of chromosomes (2n, total 23×2=46), of which 22 are autosomes and 1 sex chromosome.

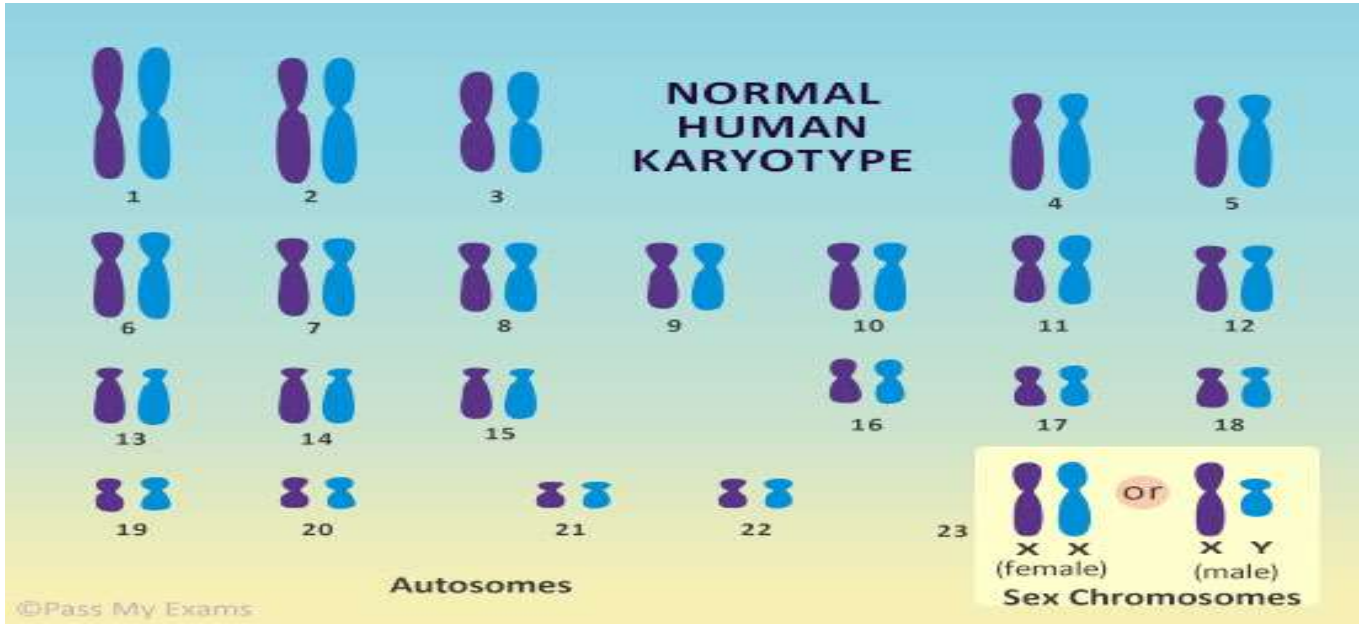
Structure of a chromosome

- The long string like structure that makes up a chromosome is made up of a chemical called **deoxyribonucleic acid (DNA)**. Each chromosome contains one DNA molecule.
- The DNA is coiled tightly around proteins called **histones**. These proteins provide structural support to a chromosome and allow the very long DNA molecule to form a compact shape and fit inside the nucleus of a cell.
- Individual chromosomes cannot be seen very clearly in the periods between cell division.
- This is because during this phase i.e. the interphase the chromosomes exist as very loosely coiled, long, thin threads spread throughout the nucleus.
- Before the nucleus of a cell divides an exact copy of the DNA molecule in a chromosome is made so that at nuclear division the chromosome is a double structure, containing two identical DNA molecules.



- These two part structure of chromosomes are called **chromatids** with each chromatid of the pair containing one of the two identical DNA molecules.
- Just before nuclear division takes place the chromosomes coil up into shorter, thicker more compact structures and the chromatids become recognisable as separate structures.
- It is these structures that are much more visible under a microscope and therefore more commonly used when showing chromosomes.
- The image below shows the structure of a chromosome just before nuclear division made up of two **chromatids**. The chromatids are held together at a point called the **centromere**. The centromere may occur anywhere along the length of the chromosome.
- When the sets of chromosomes from a human are lined up according to size it can be seen that they exist in pairs.
- These are called **homologous pairs** because they are similar in structure.

- An image of such an arrangement of chromosomes is called a **karyogram** and the set of chromosomes is called the **karyotype**.
- Karyotyping is a technique to study the structure of chromosomes present in a species. Chromosomes are isolated, stained and photographed. This technique is useful in finding out any chromosomal abnormalities.
- In humans there are 23 pairs of chromosomes. The reason the chromosomes are in pairs is because one set of chromosomes comes from the female parent via the egg and the other set of chromosomes comes from the male parent by way of the sperm.
- During fertilisation when the sperm cell fuses with the egg cell the resulting cell is called a zygote and contains two sets of chromosomes.



Types of chromosomes

- Human chromosomes are of two types **autosomes** and **sex chromosomes**.
- Genetic traits that are linked to the sex of the person are passed on through the sex chromosomes. The rest of the genetic information is present in the autosomes.
- Humans have 23 pairs of chromosomes in their cells, of which 22 pairs are autosomes and one pair of sex chromosomes, making a total of 46 chromosomes in each cell.

Based on the number of centromeres

- **Monocentric** with one centromere.
- **Dicentric** with two centromeres.
- **Polycentric** with more than two centromeres
- **Acentric** without centromere. Such chromosomes represent freshly broken segments of chromosomes which do not survive for long.
- **Diffused or non-located** with indistinct centromere diffused throughout the length of chromosome.

Based on the location of centromere

- **Telocentric** are rod-shaped chromosomes with centromere occupying the terminal position, so that the chromosome has just one arm.
- **Acrocentric** are also rod-shaped chromosomes with centromere occupying a sub-terminal position. One arm is very long and the other is very short.
- **Sub-metacentric** chromosomes are with centromere slightly away from the mid-point so that the two arms are unequal.
- **Metacentric** are V-shaped chromosomes in which centromere lies in the middle of chromosome so that the two arms are almost equal.

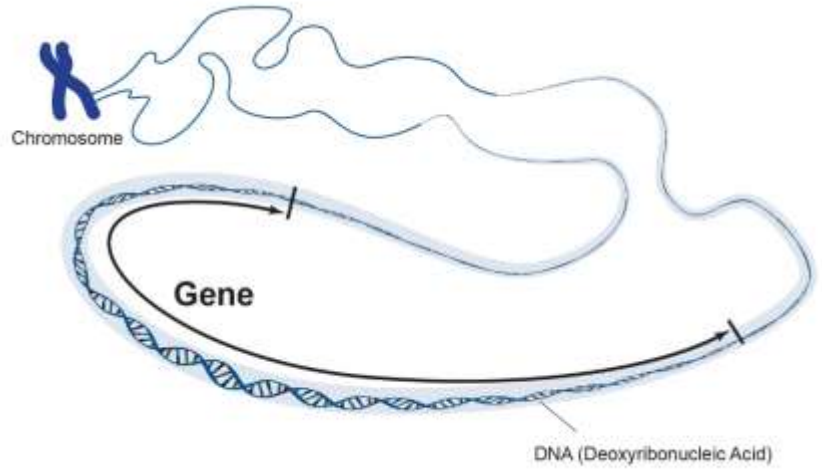
Functions and significance of chromosomes

- The number of the chromosomes is constant for a particular species. Therefore, these are of great importance in the determination of the phylogeny and taxonomy of the species.
- **Genetic Code Storage**: Chromosome contains the genetic material that is required by the organism to develop and grow. DNA molecules are made of chain of units called genes. Genes are those sections of the DNA which code for specific proteins required by the cell for its proper functioning.
- **Sex Determination:** Humans have 23 pairs of chromosomes out of which one pair is the sex chromosome. Females have two X chromosomes and males have one X and one Y chromosome. The sex of the child is determined by the chromosome passed down by the male. If X chromosome is passed out of XY chromosome, the child will be a female and if a Y chromosome is passed, a male child develops.

- **Control of Cell Division:** Chromosomes check successful division of cells during the process of mitosis. The chromosomes of the parent cells insure that the correct information is passed on to the daughter cells required by the cell to grow and develop correctly.
- **Formation of Proteins and Storage:** The chromosomes direct the sequences of proteins formed in our body and also maintain the order of DNA. The proteins are also stored in the coiled structure of the chromosomes. These proteins bound to the DNA help in proper packaging of the DNA

Genes

The gene is the basic physical unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify traits. Genes are arranged, one after another, on structures called chromosomes. A chromosome contains a single, long DNA molecule, only a portion of which corresponds to a single gene. Humans have approximately 20,000 genes arranged on their chromosomes.



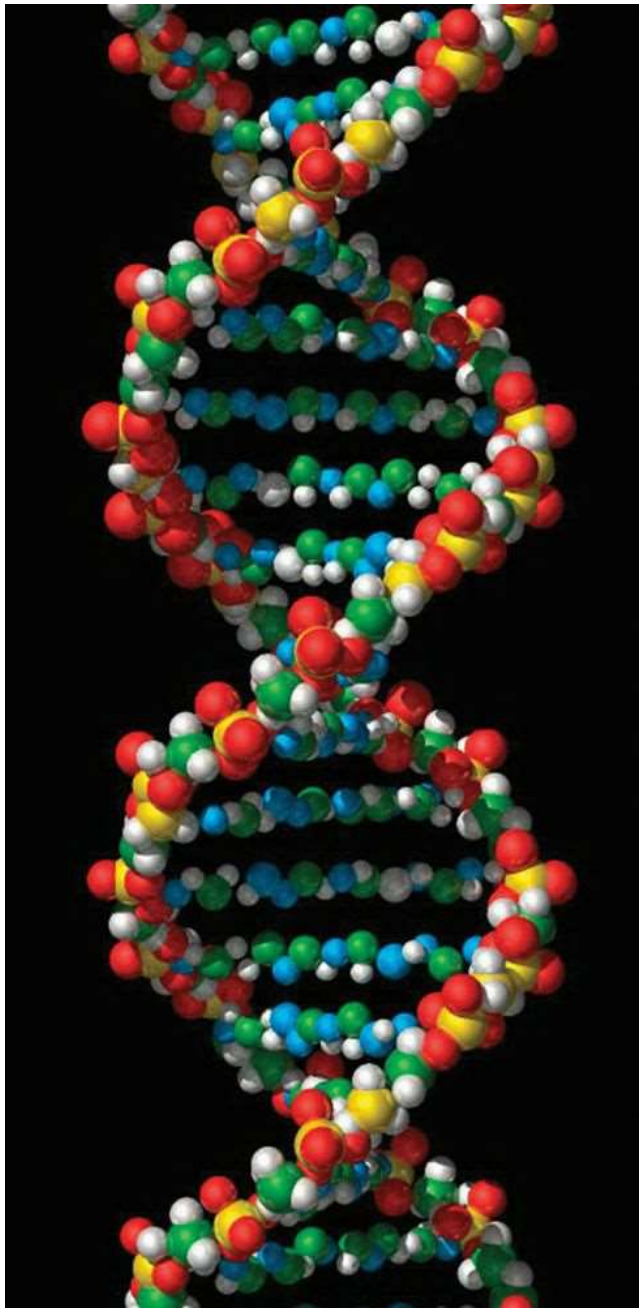
- There are about 30000 genes in each cell of the human body. DNA present in the gene comprises of only 2 percent of the genome.
- Genes come in pairs in the same way like the chromosomes. Each parent of a human being carries two copies of their genes and each parent passes one copy of genes to their child. This is the reason why the child has many characteristics of both the parents like hair colour, same eyes etc.

Functions of Genes

- Genes control the functions of DNA and RNA.
- Proteins are the most important materials in the human body which not only help by being the building blocks for muscles, connecting tissue and skin but also takes care of the enzymes production.
- These enzymes play an important role in conducting various chemical processes and reactions within the body. Therefore, protein synthesis is responsible for all activities carried on by the body and are mainly controlled by the genes.
- Genes consist of a particular set of instructions or specific functions. For example, globin gene was instructed to produce haemoglobin. Haemoglobin is a protein that helps to carry oxygen in the blood.

DNA

Deoxyribonucleic acid, or DNA, is a molecule that contains the instructions an organism needs to develop, live and reproduce. These instructions are found inside every cell, and are passed down from parents to their children. Its one long molecule that contains our genetic ‘code’.

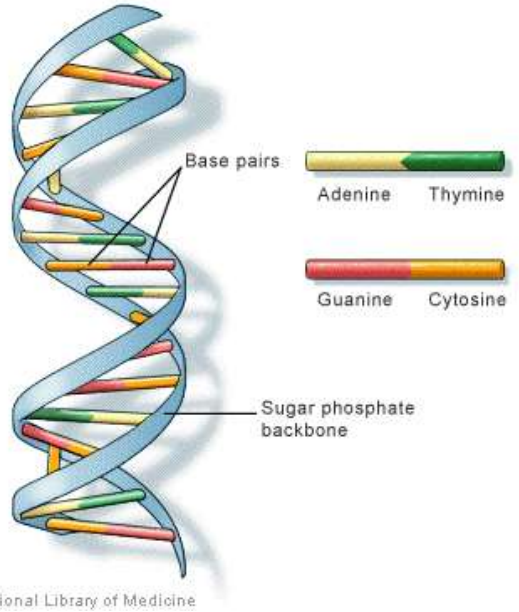


Structure of DNA

- DNA is a double-stranded helix. That is each DNA molecule is comprised of two biopolymer strands coiling around each other to form a double helix structure.
- These two DNA strands are called polynucleotides, as they are made of simpler monomer units called nucleotides.
- Each strand has a 5’end (with a phosphate group) and a 3’end (with a hydroxyl group).
- The strands are antiparallel, meaning that one strand runs in a 5’to 3’direction, while the other strand runs in a 3’ to 5’ direction.

- The two strands are held together by hydrogen bonds and are complimentary to each other.
- Basically, the DNA is composed of deoxyribonucleotides.
- The deoxyribonucleotides are linked together by 3′ – 5′phosphodiester bonds.
- Each nucleotide contains a sugar and phosphate group as well as nitrogen bases.
- These nitrogen bases are further broken down into four types,

  including Adenine (A) Cytosine (C) Guanine (G) Thymine (T)

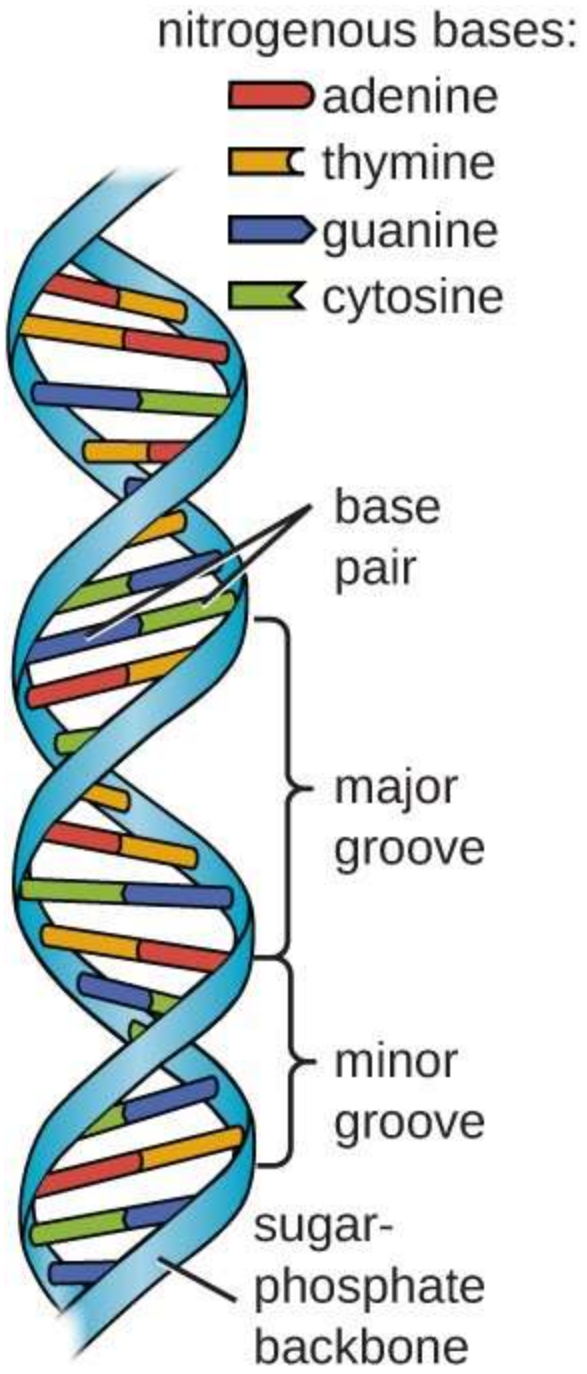


•The nitrogen bases have a specific pairing pattern.

•This pairing pattern occurs because the amount of adenine equals the amount of thymine; the amount of guanine equals the amount of cytosine. The pairs are held together by hydrogen bonds.

•As a result of the double helical nature of DNA, the molecule has two asymmetric grooves. One groove is smaller than the other.

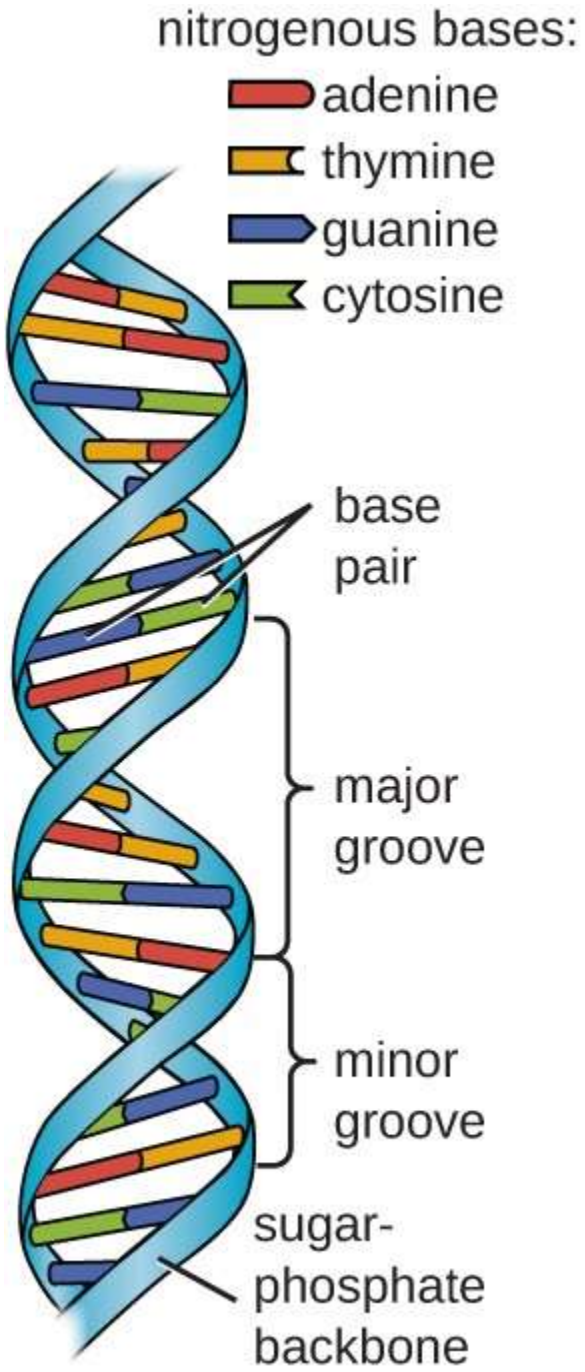
•This asymmetry is a result of the geometrical configuration of the bonds between the phosphate, sugar, and base groups that forces the base groups to attach at 120 degree angles instead of 180 degree.



•The larger groove is called the major groove, occurs when the backbones are far apart; while the smaller one is called the minor groove, occurs when they are close together.

•Since the major and minor grooves expose the edges of the bases, the grooves can be used to tell the base sequence of a specific DNA molecule.

•The possibility for such recognition is critical, since proteins must be able to recognize specific DNA sequences on which to bind in order for the proper functions of the body and cell to be carried out.



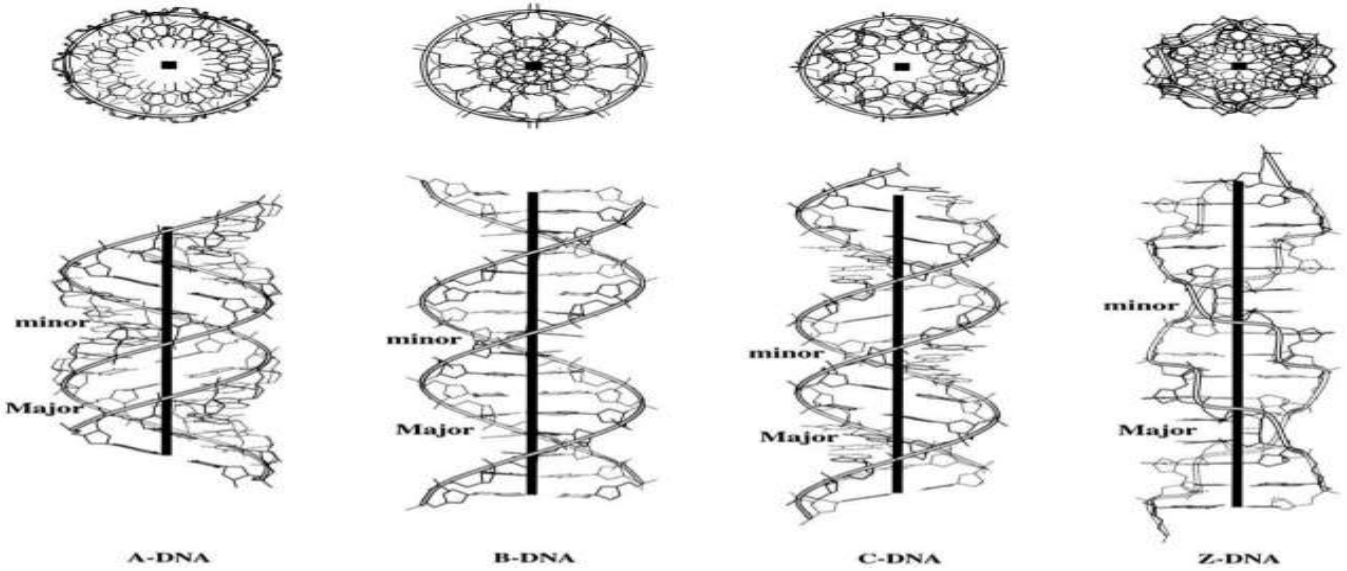
Types of DNA

•Most of the DNA is in the classic Watson-Crick model simply called as B-DNA or B-form DNA.

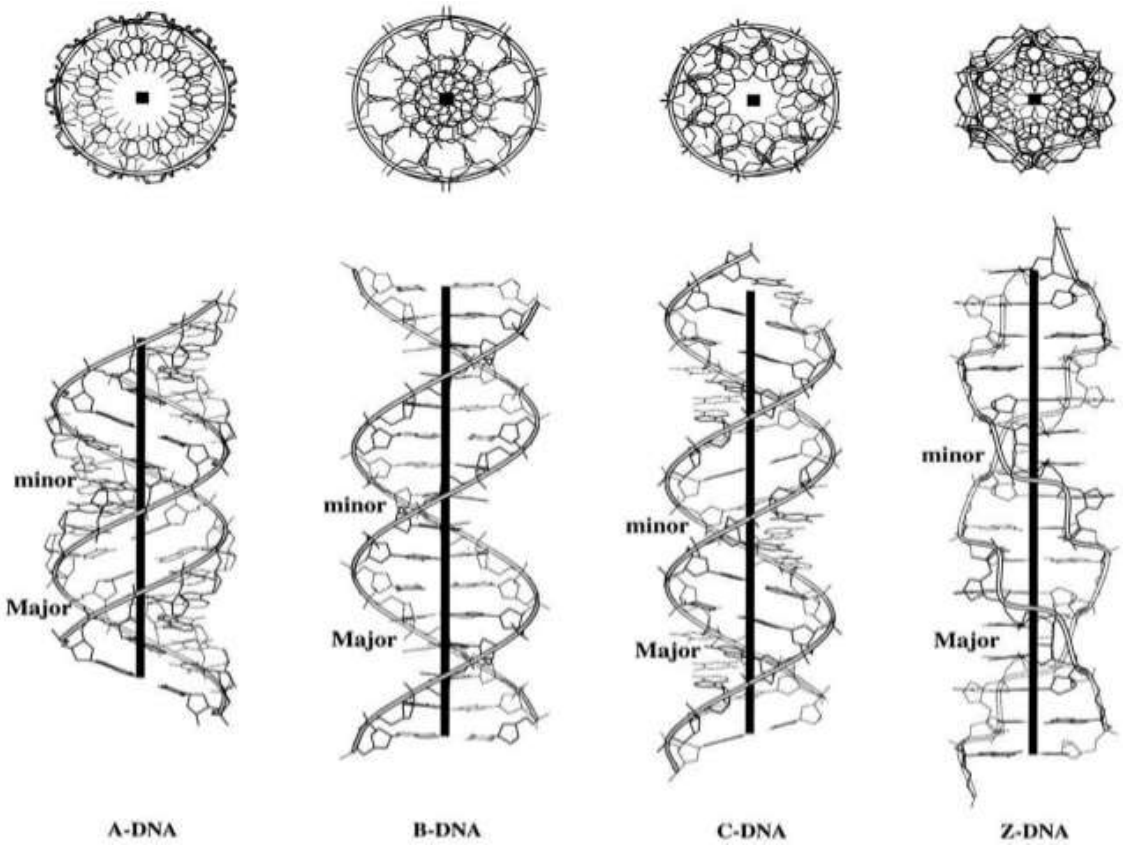
•In certain condition, different forms of DNAs are found to be appeared like A-DNA,Z-DNA,C- DNA,D-DNA,E-DNA.

•This deviation in forms are based on their structural diversity.

- **B-DNA**-Most common, originally deduced from X-ray diffraction of sodium salt of DNA fibres at 92% relative humidity.



- **A-DNA**-Originally identified by X-ray diffraction of analysis of DNA fibres at 75% relative humidity.
- **Z-DNA**-Left handed double helical structure winds to the left in a zig- zag pattern.
- **C-DNA**-Formed at 66% relative humidity and in presence of Li+ and Mg2+ ions.
- **D-DNA**-Rare variant with 8 base pairs per helical turn, form in structure devoid of guanine .
- **E- DNA**-Extended or eccentric DNA.



Functions of DNA

DNA has a crucial role as genetic material in most living organisms. It carries genetic information from cell to cell and from generation to generation.

Thus its major functions include:

- Storing genetic information
- Directing protein synthesis
- Determining genetic coding
- Directly responsible for metabolic activities, evolution, heredity, and differentiation.

It is a stable molecule and holds more complex information for longer periods of time.