Introduction to Human Genetics

Genetics is the branch of biology concerned with the study of inheritance, including the interplay of genes, DNA variation and their interactions with environmental factors.

Genetics is the study of how genes and how traits are passed down from one generation to the next. Human genes carry information that affects health, appearance, and even the personality. So, **Human genetics** is a vast and complex field of study that aims to understand the inheritance of traits in humans. It encompasses various branches that explore different aspects of genetics, from the study of the structure and function of genes to the analysis of how genetic variations contribute to human diseases.

Main basic concepts in genetics that should be clear to anyone studying this field:

Chromosomes are threadlike structures made of protein and DNA that serve to carry the genomic information from cell to cell. Like plants and animals, human chromosomes reside in the nucleus of cells.

Humans have 46 chromosome (23 pairs) , 22 pairs of autosomal chromosomes and one pair of sex chromosomes (XX or XY) . Each pair contains two chromosomes, one coming from each parent, which means that children inherit half of their chromosomes from their mother and half from their father. Chromosomes can be seen through a microscope when the nucleus dissolvesduring cell division.

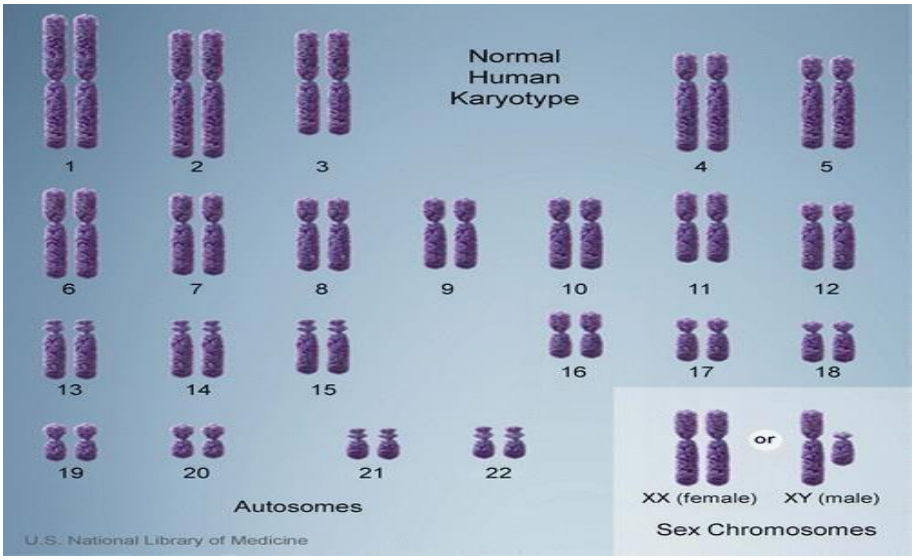

Figure (1): Human Chromosomes.

Chromatid is one of the two identical halves of a chromosome that has been replicated in preparation for cell division. The two "sister" chromatids are joined at a constricted region of the chromosome called the centromere.

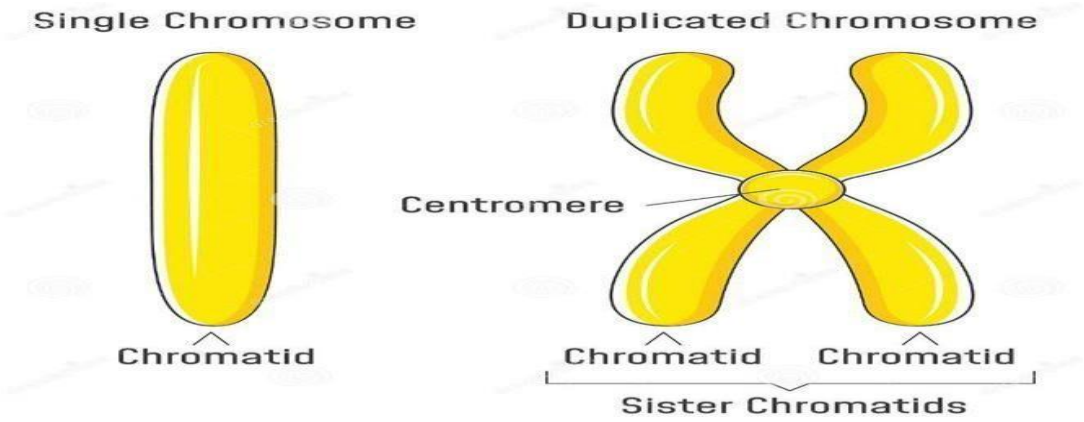

Figure (2) human chromosome chromatid& centrosome

Centromere is the region of a chromosome that links sister chromatids and is essential for their proper separation during cell division. It is the attachment site for spindle fibers, which help pull chromatids apart during mitosis and meiosis.

Chromatin is the complex of DNA and histone proteins that form the chromosomes found in nucleus of human's cells. Chromatin exists in two forms euchromatin and heterochromatin**.** The chromatin condenses to form **chromosomes**.

Histones are small, positively charged proteins that bind to negatively charged DNA, facilitating the formation of a compact structure known as chromatin. They are essential components of chromatin, which helps to package DNA into a more condensed form, allowing it to fit within the nucleus of eukaryotic cells.

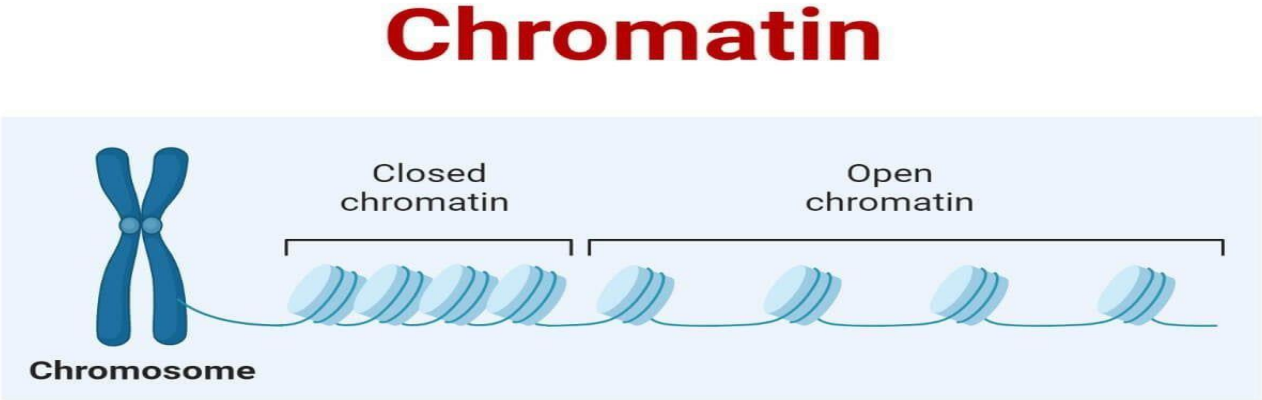

Figure (3) chromatin

Deoxyribonucleic acid (DNA) is the molecule that carries genetic information for the development and functioning of an organism. DNA is made of two linked strands that wind around each other to resemble a twisted ladder — a shape known as a double helix. Each strand has a backbone made of alternating sugar (deoxyribose) and phosphate groups Attached to each sugar is one of four bases: adenine (A), cytosine (C), guanine (G) or thymine (T). The two strands are connected by chemical bonds between the bases: A bonds with T, and C bonds with G. The sequence of thebases along DNA's backbone encodes biological information, such as the instructions for making a protein or RNA molecule.

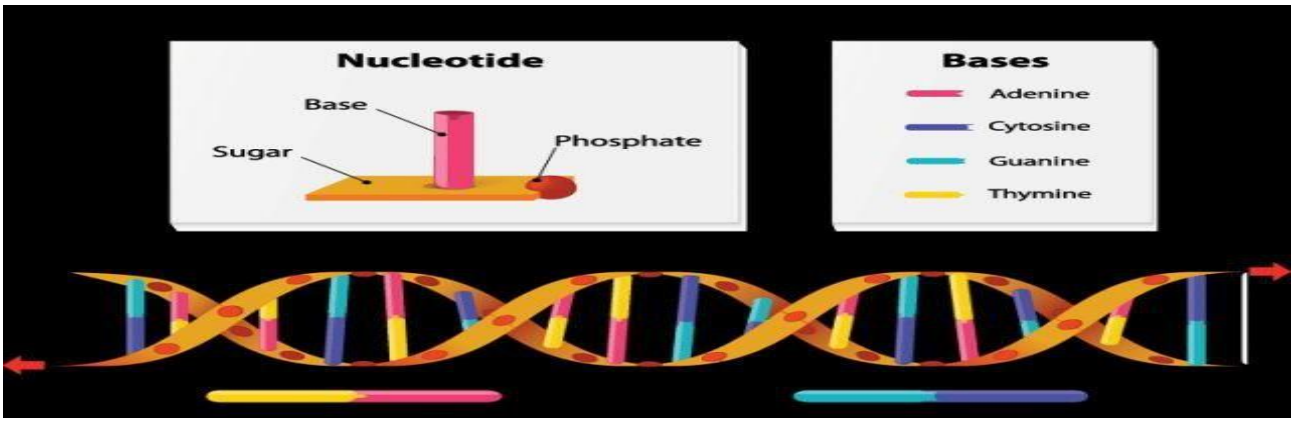

Figure (4) DNA structure

Genes: are considered the basic unit of inheritance, made up of DNA sequences, typically each gene containing from hundreds to thousands of nucleotide bases. Genes passed from parents to offspring and contain the information needed to specify physical and biological traits.

Each gene has regulatory regions (promoters and enhancers) that control its expression and Coding regions (exons) and non-coding regions (introns).

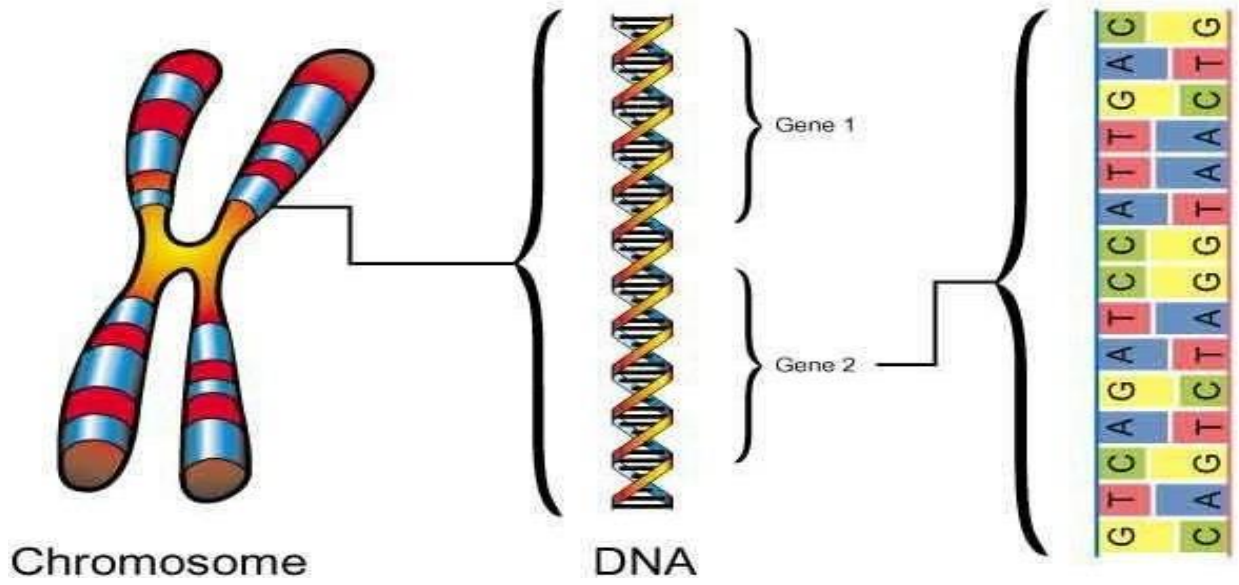

Figure (5): Genes.

Genome is the entire set of DNA instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes located in the cell's nucleus, as well as a small chromosome in the cell's mitochondria. A genome contains all the information needed for an individual to develop and function.

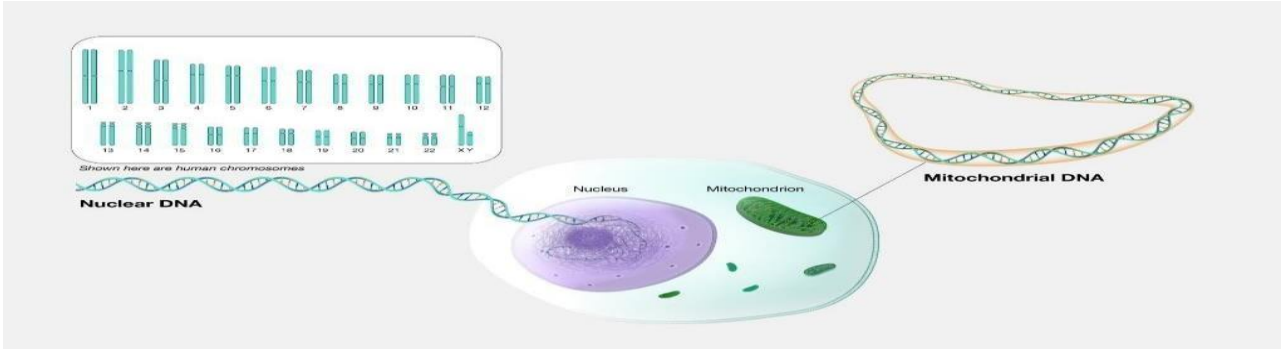

Figure (6): Genome.

Genetic variation refers to DNA sequence differences among individuals or populations. Genetic variation is what makes us all unique, whether in terms of hair color, skin color or even the shape of our faces. Some variants influence biological function (such as a mutation that causes a genetic disease), while others have no biological effects.

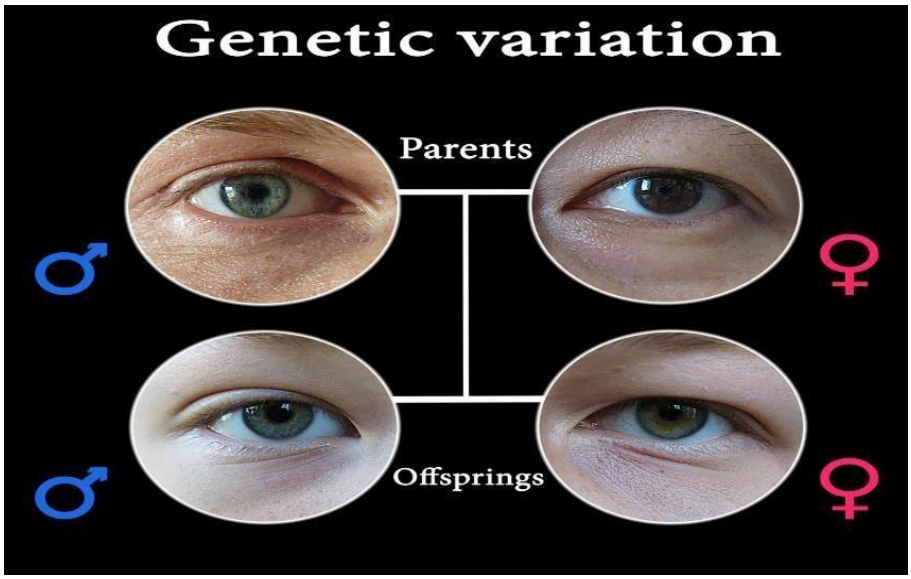

Figure (7): Genetic variation in eyes.

Environmental factors as related to genetics, refers to exposures to substances (such as pesticides or industrial waste) where we live or work, behaviors (such as smoking or poor diet) or stressful situations that can increase an individual's risk of disease. Genetic studies often take environmental factors into consideration, as these exposures can increase an individual's risk of genetic damage or disease.

Allele is different form of a gene which produce variations in a genetically inherited trait. Different alleles produce different hair colors—brown, blond, red, black, etc. An individual inherits two alleles, one from each biological parent. If the two alleles are the same, the individual is homozygous for that allele. If the alleles are different, the individual is heterozygous.

Dominant allele means that in the case of a dominant trait, only one copy of the dominant allele is required to express the trait. The effect of the other allele (the recessive allele) is masked by the dominant allele. Typically, an individual who carries two copies of a dominant allele exhibits the same trait as those who carry only one copy. This contrasts to a recessive trait, which requires that both alleles be present to express the trait.

Recessive allele means that in the case of a recessive trait, the alleles of the trait-causing gene are the same, and both (recessive) alleles must be present to express the trait. A recessive allele does not produce a trait at all when only one copy is present.

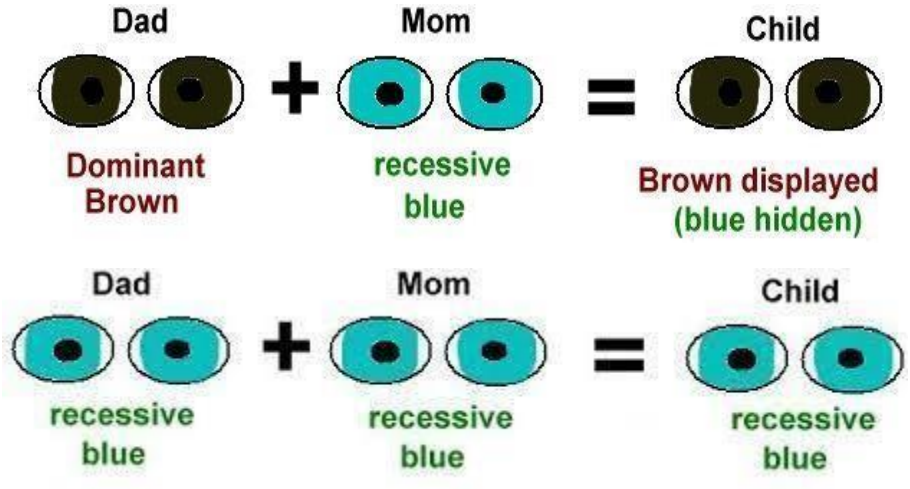

Figure (8): Dominants and recessive alleles.

Homozygous means when organism possess two similar sets of alleles at the same locus for a particular trait, is defined as **Homozygous organism**.

Heterozygous means when organism possess two different sets of alleles at the same locus for a particular trait, is defined as **Heterozygous organism.**

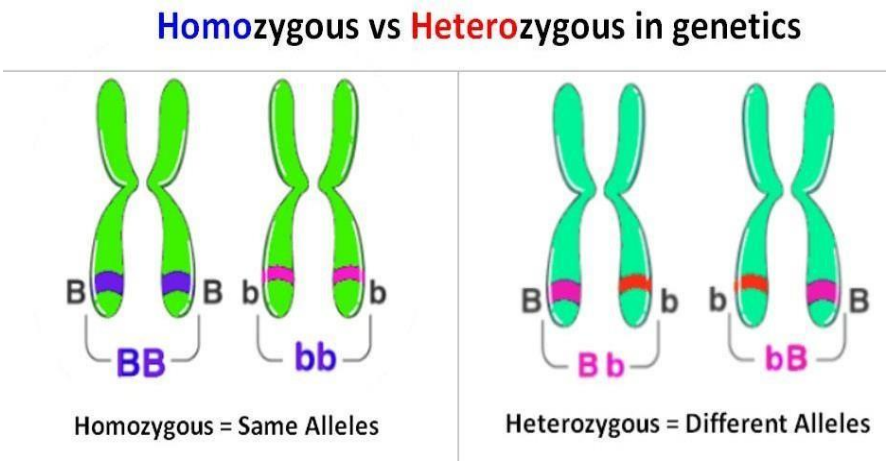

Figure (9): Homozygous and heterozygous alleles.

Genotype is internal heredity information that contain genetic code, in other word"genotype" is genetic makeup of organism.

Phenotype refers to an individual's observable traits, such as height, eye color and blood type. A person's phenotype is determined by both their genomic makeup (genotype) and environmental factors.

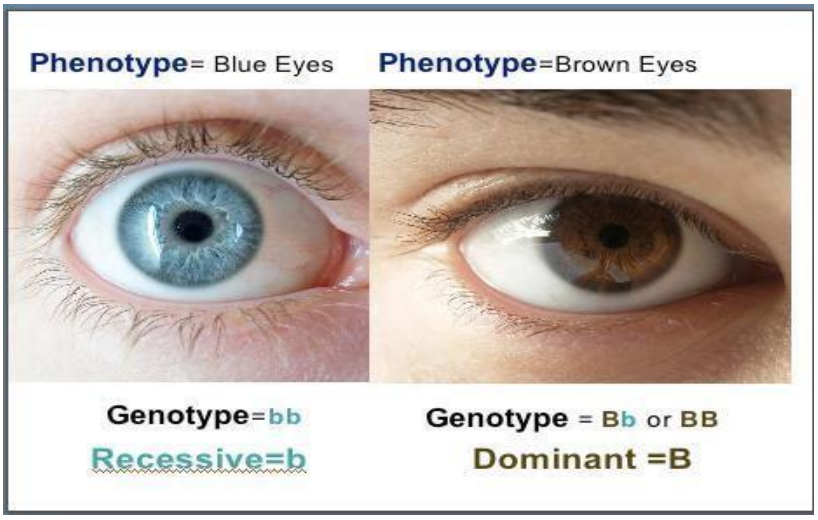

Figure (10): Human Genotype and Phenotype.