

# Genetics: Organization and Function of Genes

[See online here](#)

**In the medical field, the topic of Genetics is of significant importance. The topic explains how an organism is able to pass on its characteristics to its offspring. In this article, you will be able to learn about the structure and function of the human gene. Processes such as replication, transcription, and translation are also explained in detail to help you get the required insight on what happens in each process. Furthermore, there are a few questions at the end of the article that will help you to test your understanding. You will find the answers to these questions below the references.**



## Terms and Definitions of Genetics

**Genetics** is the study of heredity or how an organism passes on its characteristics to its offspring. The word is derived from *gene*, a segment of **DNA** (deoxyribonucleic acid, a molecule which is made up of two long twisted strands that contain genetic information). The gene contains the instructions required to produce proteins. It is the basic physical and functional unit of heredity. In humans, genes are found in the nuclei of cells, and they range in size from a few hundred DNA bases to over 2 million bases.

Other terminologies commonly used in the study of genetics include:

**Allele** - Alleles are genes that occupy the same position or locus on each chromosome that forms a pair. They are therefore different forms of the same gene since one is inherited from the father and the other from the mother. Alleles can have the same sequence of DNA bases or can contain slight differences. An occurrence where there is a

pair of identical alleles is known as **homozygosity** while that with non-identical alleles is termed as **heterozygosity**.

**Genome** – A genome is the complete set of genetic material. In other words, a genome contains all the genes of an organism. In humans, the genome contains more than 20,000 genes.

**Karyotype** – Genes are contained in chromosomes, and a karyotype is the full set of chromosomes in an organism's cells.

## Organization of Genes

**Genes** are organized into units known as chromosomes.

Humans have **23 pairs of chromosomes** (46 in total). One chromosome in each pair comes from the person's mother and the other from the father.

The DNA, in human genes, is made up of the following **four types of bases**:

A – Adenine

C – Cytosine

G – Guanine

T – Thymine

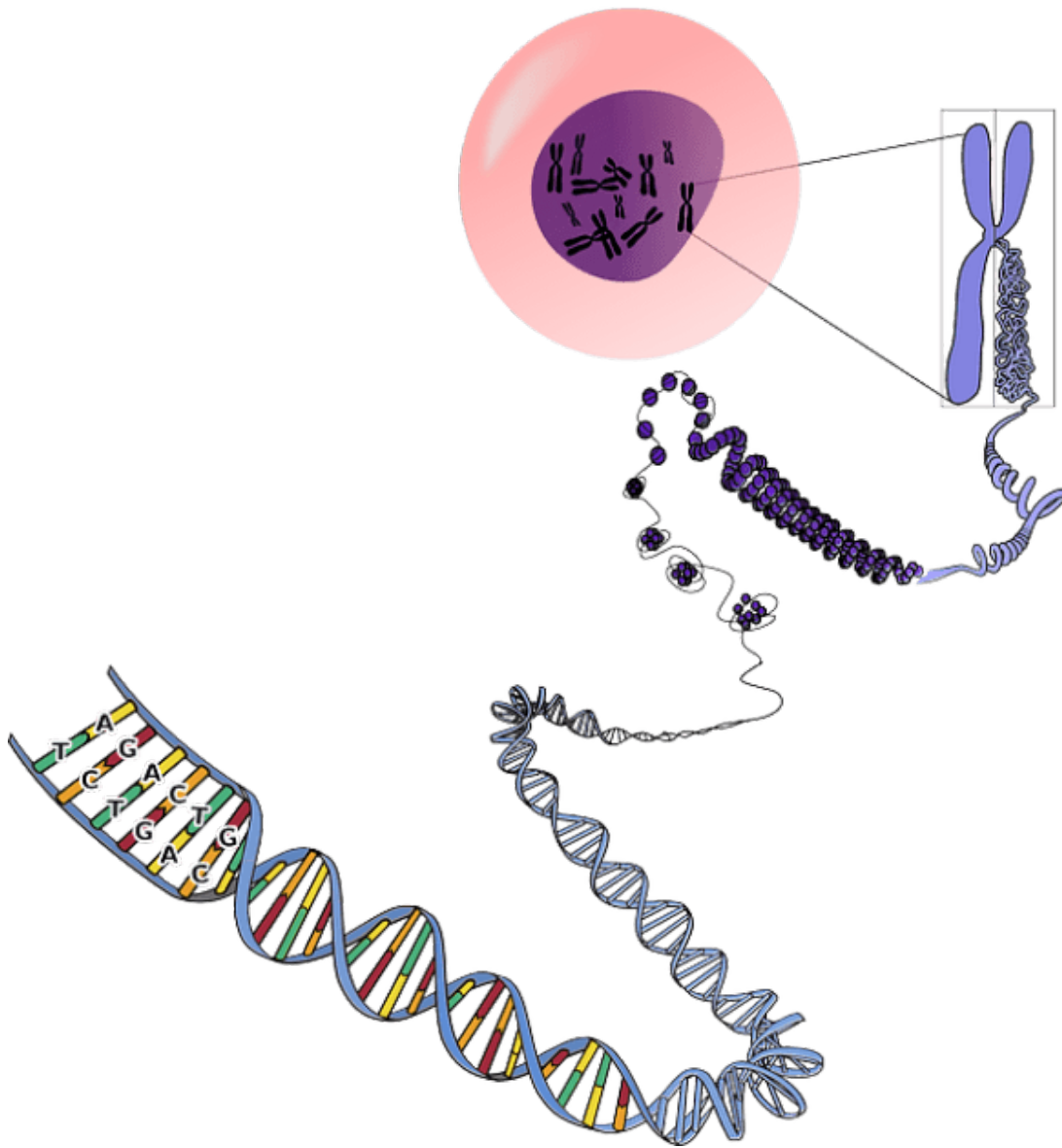
Adenine is a purine base, and it always pairs up with thymine, which is a pyrimidine base. Cytosine is also a purine base, and it always pairs up with guanine which is a pyrimidine base.

**Note:** In DNA, Adenine always pairs with Thymine. Cytosine always pairs with Guanine. Once the bases pair up, they form **strands**. A DNA molecule is made of up two strands of bases, wound around each other to form a double helix. These strands are held together by bonds that exist between the bases.

In order for long DNA chains to fit into tiny chromosomes within a cell's nucleus, the DNA is wrapped around proteins known as **histones** (H1, H2, H3, and H4), which make it more compact. The resulting DNA-histone complex is known as **chromatin**.

Nucleosomes typically contain 146 base pairs of DNA and eight histone proteins. These nucleosomes then coil around each other to form thicker fibers, which then continue to form coils and loops until they form the double helices.

Chromosomes are made of **chains of nucleosomes**, and this gives them the appearance of a **string of beads** when examined under the electron microscope.

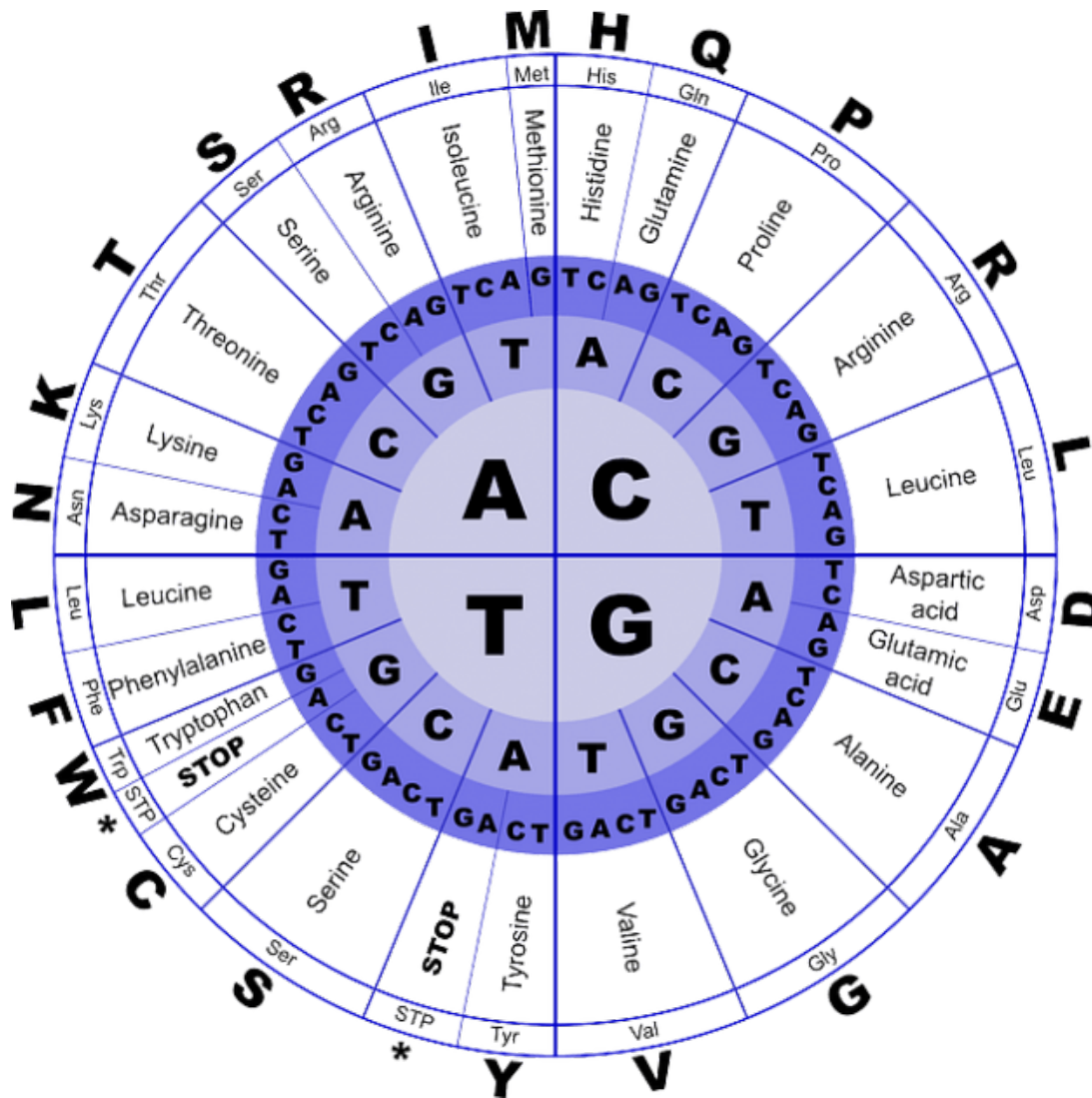


## Function of Genes

The function of genes is to **pass on the instructions** required to produce the proteins that carry an organism's characteristics from one generation to the next. The **genetic code** consists of instructions that are interpreted by the cells to facilitate protein formation. This code consists of **codons**, which are sets made up of three bases that specify the synthesis of the amino acids which form proteins.

**Note:** Codons are made up of three bases. Specific codons specify specific amino acids. For example, the amino acid methionine is coded for by the codon ATG (Adenine, Thymine, Guanine) codon.

Some amino acids are specified by several codons like isoleucine, whose codons are ATT (Adenine, Thymine, Thymine), ATC (Adenine, Thymine, Cytosine) and ATA (Adenine, Thymine, Adenine).



## DNA Replication and Protein Synthesis

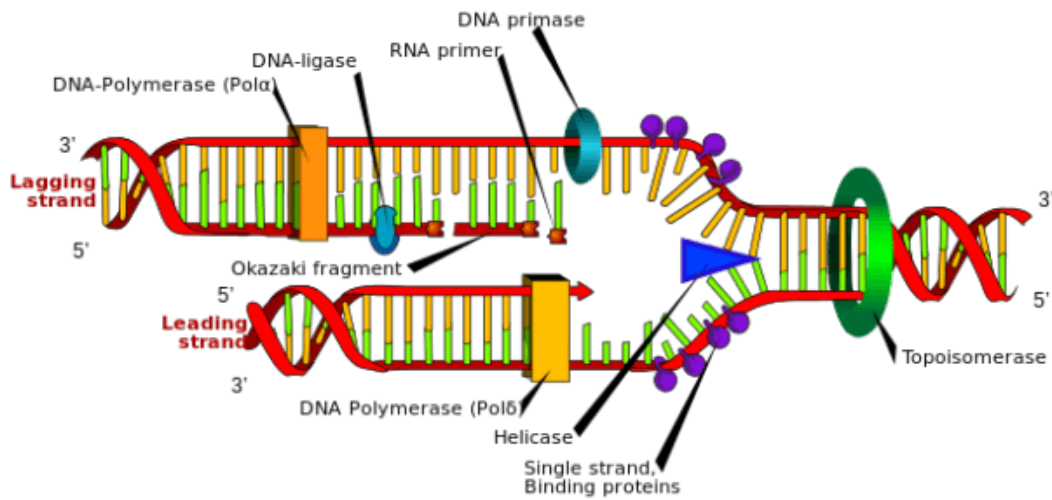
DNA is copied to form RNA (ribonucleic acid) which is then used to make proteins.

The process by which DNA material is copied to form RNA is called **transcription**. The process by which the RNA is used to make proteins is known as **translation**.

**Note:** DNA makes RNA. RNA makes protein.

### DNA Replication

When a cell divides, the double strands that form its DNA split into two single strands of DNA. Every single strand of DNA then acts as a template and forms a new strand of complementary DNA. This results in each new daughter cell with complete double strands of DNA, a process known as DNA replication. This DNA replication process is directed by **DNA polymerase enzymes**.



DNA replication

## DNA Transcription

DNA transcription is the process by which DNA is copied or transcribed into the **mRNA (messenger RNA)**, which carries the information needed to synthesize proteins.

During this process, the DNA unwinds, and the strands separate so that one strand is used as a template to form mRNA. The mRNA has the same base pairs as the original DNA except for thymine, which is replaced by uracil (U).

**Note:** In RNA, Adenine always pairs with Uracil. Cytosine always pairs with Guanine. Once the pre-messenger RNA is formed, this single strand of RNA is edited to produce the desired mRNA through a process known as **RNA splicing**.

This splicing involves the removal of the mRNA segments transcribed from **introns**, which are the DNA segments that carry information needed to regulate the speed of protein synthesis. After splicing, the mRNA only contains segments transcribed from **exons**, which are the DNA segments that carry information on the types of amino acids to be used for protein production.

**Note:** Exons are DNA segments that carry information about the kinds of amino acids to be used in protein production. Introns are DNA segments that carry information on regulating the speed of protein synthesis.

## DNA Translation

After the mRNA is formed, it is transported out of the cell's nucleus into the ribosome which is located within the cytoplasm. The ribosome is the cell's protein factory, and it is made up of one large subunit and a smaller subunit.

As the mRNA travels through the ribosome, its codons, which are made of three nucleotide bases, engage with the tRNA (transfer RNA) anticodons. This tRNA carries an amino acid on one end as well as three nucleotides which make the **anticodon** on the other end.

The tRNA anticodons thus act as an interpreter by reading the amino acid specified by the mRNA codon.

The specified amino acid is then incorporated into a chain of amino acids according to the sequence determined by the mRNA before it is ejected from the ribosome.

As the amino acid chain is assembled, it folds upon itself and creates the complex 3-dimensional structure of the protein.

**Note:** DNA replication is followed by DNA transcription, which is then followed by DNA translation.

## How Genes Act as Instructions

The DNA information within each gene contains instructions required to form a single protein. All protein-coding regions of the DNA begin with the **“start protein synthesis” codon ATG** (Adenine, Thymine, Guanine) which also codes for methionine.

This is followed by segments of DNA that have specific codons for specific amino acids. The arrangement of the codons on the DNA results in different amino acid sequences, consequently leading to the production of different proteins.

For example, the codons CAT, ATT, and CTT code for the amino acids histidine, isoleucine, and leucine, respectively, which will form a different protein if the codons are ordered differently. For example, the order CTT, ATT, and CAT will form leucine, isoleucine, and histidine.

There are several “stop protein synthesis” codons like TAG and TAA, which mark the end of the protein-coding region. Therefore, the DNA, through the use of mRNAs, gives instructions required to form **amino acids** in a specific order. These amino acids are then connected to form protein molecules. The differences in the amino acid sequences result in different types of proteins.

## Chromosomes of the Human Genome

Genes are usually contained in chromosomes, which are located in the cell nucleus.

### Chromosomes in germ cells

Germ cells, like the **female egg** and the **male sperm** in humans, contain 23 chromosomes. This is the **haploid number of chromosomes** or half the chromosomes in somatic cells. This is because they undergo a process known as **meiosis** to prepare for fertilization when the 23 chromosomes from the egg will be joined with the 23 from the sperm to make a complete set of 46 chromosomes in the fetus.

### Chromosomes in somatic cells

Somatic or non-germ cells in humans have 23 pairs of chromosomes or a total of 46 chromosomes. This is the **diploid number of chromosomes**. Each pair is made up of one chromosome from the person’s mother and another from the father. Autosomes are 22 of these chromosome pairs and are similar for both men and women. These autosomes are numbered according to size in humans.

The 23rd pair is known as the **sex chromosome** since it determines a person’s gender. It is, therefore, different in males and females. Men have one X chromosome and one Y chromosome, while women have two X chromosomes.

The Y chromosome contains the genes that are responsible for male sex differentiation.  
The X chromosome carries genes that are responsible for inherited traits.

## Chromosomal abnormalities

### **Trisomy 21**

Trisomy 21, which is also known as [Down Syndrome](#), is caused by having three copies of chromosome 21 instead of two. It is characterized by having a small head with upward slanting eyes and a small nose.

### **Turner Syndrome**

[Turner Syndrome](#) is caused by having one copy of the X chromosome in women instead of two. It is characterized by short stature and heart defects.

### **Triple X Syndrome**

Triple X Syndrome is caused by having three copies of the X chromosome in women instead of two. It is characterized by mental retardation and sterility.

### **Klinefelter's Syndrome**

Klinefelter's Syndrome is caused by having one Y and two X chromosomes in men instead of one Y and one X. It is characterized by tall stature and impaired fertility.

These conditions can be diagnosed by genetic testing.

## References

[Genetics Home Reference](#) via ghr.nlm.nih.gov

[Merck Manual](#) via merckmanuals.com

[Proteins and Gene Expression](#) via nature.com

**Legal Note:** Unless otherwise stated, all rights reserved by Lecturio GmbH. For further legal regulations see our [legal information page](#).