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.

To understand how traits such as eye color are passed on from one generation to the next, you have to familiarize yourself with the following vocabularies:

**DNA** – deoxyribonucleic acid is a molecule which is made up of two long twisted strands that contain genetic information.

**Gene** – A gene is a segment of DNA that contains the instructions required for the production of proteins. It is the basic physical and functional unit of heredity. In humans, genes are found in the nucleus of cells, and they range in size from a few hundred DNA
bases to over 2 million bases.

**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 their 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 therefore 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 for the development of proteins that carry the characteristics of an organism from one generation to the next.

The **genetic code** is therefore made up of instructions which are interpreted by the cells to facilitate the formation of proteins.

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 splits 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 having its complete double strands of DNA and this process is known as DNA replication. This process of DNA replication is directed by **DNA polymerase enzymes**.
DNA Transcription

DNA transcription is the process by which DNA is copied or transcribed into the mRNA (messenger RNA), which carries the information needed for the synthesis of proteins.

During this process, the DNA unwinds, and the strands separate so that one strand is used as a template for the formation of mRNA. This 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 segments of DNA that carry information needed for regulating 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 type of amino acids to be used for the production of proteins.

**Note:** Exons are DNA segments that carry information about the kind of amino acids to be used in the making of proteins. 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 anticodons of the tRNA (transfer RNA). This tRNA carries an amino acid on one end as well as three nucleotides which make the anticodon on the other end.

The anticodon of the tRNA thus acts as an interpreter by reading the amino acid specified by the codon of the mRNA.
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 for the formation of 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 which have specific codons that code 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 CTT** code for the amino acids histidine, isoleucine, and leucine which will form a different protein if the codons are ordered differently. For example, the order **CTT ATT 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 for the formation of **amino acids** in a specific order. These amino acids are then connected to form protein molecules. The differences in the sequences of the amino acids result in different types of proteins being produced.

### Chromosomes of the Human Genome

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

#### 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** in preparation 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 their father.

22 of these chromosome pairs are known as autosomes 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 in their sex chromosomes while women have two X chromosomes in their sex chromosomes.

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

Review Questions

The answers are below the references.

1. **Which of the following RNA sequences would be transcribed if the DNA strand was ATGATTACATA?**
   - A. AUGAUUAUCAUA
   - B. TACTAATAGTAT
   - C. CUGCUUCUACUC
   - D. GUAGUUGUGUG

2. **In humans, amino acids are required for the synthesis of:**
   - A. Carbohydrates
   - B. Fats
   - C. Proteins
   - D. Lipids

3. **Genetic testing is done on a baby, and they are found to have XXY. What condition do they have?**
   - A. Trisomy 21
   - B. Turner Syndrome
   - C. Triple X Syndrome
   - D. Klinefelter Syndrome