GENETICS AND HEALTH

Davlatov Hamidjon Dilshodjon ogli
10th grade student of the 13th general education school of Fergana city.
Scientific supervisor: **Parpieva Odinakhan Rakhmanovna**
Senior lecturer at Central Asian Medical University.
Fergana, Uzbekistan.

Annotation. *Genetics determines many of the characteristics of our body. This information is encrypted in DNA. The DNA sequence is identical in all cells of the body, except the sex cells, and can undergo only minor changes with age (somatic mutations). The DNA sequence is a matrix that determines the structure of all the proteins in our body, as well as how much of these proteins will be produced. In other words, almost all functions and biochemical processes depend on DNA.*

Key words: *heredity, genetics, DNA, chromosomes, genetic disorders, myopia, Down syndrome, Klinefelter's syndrome, hemophelia.*

Heredity is the ability of living organisms to pass on genetic information about certain characteristics from one or both parents to their children. For example, heredity determines some aspects of our appearance, so siblings often resemble each other, and children resemble their parents.

The science that studies heredity is called genetics. The branch of science called human genetics focuses on the study of aspects of heredity and variability in humans.

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Cells are the building blocks for all living things on earth. The human body is made up of trillions of cells. Inside each cell (in the nucleus) there is genetic material. Genetic material is the totality of all the hereditary information of a cell. This genetic information determines, for example, many of a person's external characteristics, such as eye, skin and hair color.

All cells of our body contain a building plan in two copies, each of which in its normal state (norm) is packaged in 23 packages, the so-called “chromosomes”. Chromosomes are passed on to us from our parents: one copy of the blueprint with 23 chromosomes from mom and a second copy with 23 chromosomes from dad. Thus, we have 23 pairs of chromosomes stored in each cell, for a total of 46. Chromosomes are made of a chemical compound called DNA.

Each chromosome consists of a tightly knit strand of genetic material - a double-stranded DNA molecule (DNA - deoxyribonucleic acid). DNA stores the genetic

information of each person's building plan. Some computer scientists even believe that our genetic blueprint is the most powerful computer on earth. In order to organize all genetic information, use it correctly and transmit it further, the cell divided our construction plan into 23 subunits - chromosomes.

Genes are located on thread-like structures called chromosomes. Each of these genes contains the genetic information for protein production. The cells of our body need proteins to function properly. Since each cell in the human body contains two building plans, there are also two instructions for each protein - one from the mother, the other from the father. With these instructions, the cell assembles the available building blocks (amino acids) and thus produces protein. It is important that there are no errors in these instructions.

Heredity is the transfer of genetic information from parents to children. Each person has two blueprints with 46 chromosomes in the nucleus of each cell. The exception is eggs and sperm: each of them contains only one building block, that is, only 23 chromosomes. This is necessary so that after the fusion of the egg and sperm, two building plans with 46 chromosomes are formed in each cell of the child. Thus, each child receives a complete genetic blueprint from both mother and father, and as a result, external similarity to both parents.

Sometimes it can happen that an error creeps into a person's genetic blueprint. In genetics, such errors are called mutations. Each of us carries several of these errors within us - therefore, each of us has genetic plans that deviate slightly from the norm. In most cases, these bugs are found in places where they cause little to no harm. However, if the defect is located at an important site, the result may be a defective protein. Whether such a defective protein will have a further impact on the child's health depends on various factors. The hereditary factor plays a major role in this case. In genetics, there are autosomal dominant, autosomal recessive and X-linked types of inheritance.

Genetic disorders can be divided into three main categories:

- **Autosomal dominant diseases:** disorders of a single gene that occur when a person has one altered copy of that gene and one healthy copy.

Autosomal dominant inheritance:

- One mutated (changed) copy of the gene from one of the parents is enough for the child to develop an autosomal dominant disease.
- The gene that causes the disease is dominant over the "healthy" gene.
- Every individual with a disease usually has one parent who has the same disease.
- Boys and girls get sick with the same frequency.

- **X chromosome disorders:** disorders that reflect the presence of an altered gene on the X chromosome. X disorders are more common in males because they have only one X chromosome. As a result, men only need one copy of the altered gene to experience symptoms. An example is muscular dystrophy.
- **Autosomal recessive diseases:** disorders of a single gene that occur only when a person has two altered versions of the corresponding gene.

Autosomal recessive inheritance:

- The disease develops only in those people who have two mutated copies of the gene (one from each parent).
- Usually an individual with the disease does not have parents with the disease because each person has only one copy of the mutated gene.
- The disease is observed mainly in brothers and sisters (usually within one generation).
- Boys and girls get sick with the same frequency.

There are hereditary diseases that are not found in other family members or human ancestors. The fact is that healthy people can have recessive, that is, hidden, mutations in the genome, and they do not manifest themselves in any way. Two people with the same mutation in DNA or with different mutations, but in the same gene, meet. In this case, the mutation can be passed on to the child if he inherits copies of the mutated gene from both mom and dad. Such diseases include cystic fibrosis, spinal muscular atrophy, hereditary deafness and many others, there are more than 6 thousand of them.

The most common genetic diseases:

- Myopia. The disease makes itself felt in adolescence, and the child has difficulty seeing objects that are far from him. Moreover, if parents have good vision, then the risk of pathologies in children is only 10%.
- Down syndrome. This is a rather dangerous disease in which a person cannot fully be in society and has limited health capabilities. The risk of having a child with Down syndrome increases with the age of the mother.
- Colorblindness. This disease is hereditary in nature, it is expressed in a person's inability to distinguish colors (one or more). This pathology mainly affects representatives of the stronger sex.
- Klinefelter's syndrome. It is characteristic of men and manifests itself mainly during puberty. This disease leads to infertility. Patients have enlarged breasts.
- Hemophelia. Sons can inherit these diseases from their mother. It is characterized by blood clotting disorders and is quite dangerous. A patient with this disease is constantly dependent on medications.
- Cystic fibrosis. It is a hereditary disease in which people suffer from increased sweating and have serious lung problems.
- Migraine. This disease was associated with opium even in ancient papyri. It is accompanied by severe headaches, which are localized in one part of the hemisphere.

Inherited diseases can affect various organs and systems, including the heart, lungs, liver, kidneys, brain and nervous system. The severity of symptoms and the age at which they appear can vary greatly depending on the specific condition and type of genetic disorder. Many inherited diseases have no cure, but some can be managed with medications, lifestyle changes, or other treatments.

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