МЕДИЦИНА, ПЕДАГОГИКА И ТЕХНОЛОГИЯ:
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GENETIC MECHANISMS OF HEREDITARY DISEASES

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Annotation: Hereditary diseases are diseases caused by a violation of genetic information that passes from parent to child; mainly caused by mutations in chromosomes or genes that are passed on to off spring. Mutations can occur as a result of external environmental factors (ionizing Rays, certain biologically active chemical compounds) as well as negative effects on the body and cells. This article provides information on the causes of the origin of hereditary diseases, ways to combat them and their harmful consequences.

Keywords: hereditary diseases, gene, mutation, genetics, metabolism, albinism, phenylketonuria, galactosemia, leukodystrophy, hemochromatosis, enzyme, screening, medical-genetic consultations, Reproductive Center.

Introduction. Hereditary diseases arise as a result of changes (mutations) of the hereditary apparatus of the cell under the influence of radiation, thermal energy, chemical and biological factors. A number of mutations are caused by hereditary recombinations and are caused by errors in the biosynthesis of proteins and nucleic acids. Some hereditary diseases are caused by a mutation in a single gene, while others are caused by the interaction of many genes and environmental factors. Understanding the true causes of hereditary diseases is important in the development of effective treatment and preventive measures. Mutations affect both somatic and germ cells. By reason of origin, hereditary diseases are classified into gene and chromosome diseases.

Mutations in genes can be inherited from one or both parents, or can occur spontaneously during fetal development. Mutations can lead to a wide range of diseases, including monogenic diseases caused by one gene mutation, such as mucovicidosis, sickle cell anemia and Huntington's disease, and complex multifactorial diseases such as cancer, diabetes mellitus, heart disease.

Chromosomal abnormalities can also lead to hereditary diseases. These anomalies can occur during the formation of gametes (eggs and sperm) or during

embryonic development. Examples of chromosomal abnormalities that can lead to hereditary diseases are Down syndrome, Turner syndrome, and Klinefelter syndrome.

In people with gene diseases, protein (enzyme) synthesis is disrupted at the expense of a mutation at the gene level, as a result of which the biochemical reaction carried out in the presence of this enzyme does not proceed normally. This leads to a failure of metabolism.

Main part. Hereditary gene diseases according to which disorders of the metabolism in the body are classified as follows:

1. Diseases caused by impaired amino acid metabolism (phenylketonuria, alkaptonuria, albinism, leucinosis).
2. Diseases caused by impaired carbohydrate metabolism (galactosemia, diabetes mellitus, glycogenosis).
3. Diseases caused by impaired lipid metabolism (leukodystrophy).
4. Diseases caused by disorders of purine and pyrimidine metabolism.
5. Diseases caused by disorders of Mineral Metabolism (hemochromatosis, hepatolenticular degeneration).

Albinism. This is a unique genetic condition caused by mutations or changes in some genes that affect the amount of melanin that our body produces. Melanin is a chemical in the body that controls the pigmentation (color) of the skin, eyes and hair. People with albinism have very pale skin, eyes and hair. People with this disease have visual problems ranging from mild to severe. People with albinism are also sensitive to sun exposure, so they have a higher risk of developing skin cancer. There are two main types of albinism, caused by changes in the genes responsible for melanin production.

General albinism (oculocutaneous albinism - OCA) : the most common type of albinism. People with OCA have very pale hair, skin and eyes.

Ocular albinism is less common than Oca. Eye albinism mainly affects the eyes. This does not affect the skin or hair much. OA usually leads to symptoms such as blurred vision, sensitivity to light. Ocular albinism is associated with changes in the gpr143 gene. Although there is no cure for albinism, people with the disease can take steps to protect their skin and eyes, and to properly care for their eyes and skin.

Phenylketonuria (PKU) is a hereditary disease caused by a deficiency of the enzyme phenylalanine hydroxylase. Classical PKU is an autosomal recessive disorder caused by mutations in both alleles of the phenylalanine hydroxylase (PAH) gene

located in chromasome 12. In the body, phenylalanine hydroxylase converts the amino acid phenylalanine into tyrosine. Mutations in both copies of the PAH gene mean that the enzyme is inactive, and the concentration of phenylalanine in the body can increase to toxic levels. PKU is inherited from a person's parents. For a child to develop PKU, both parents must contribute to a mutated version of the PAH gene. If both parents have PKU, their children will also have PKU. If only one parent has a mutated gene, the child will not develop PKU. Even if both parents have a mutated PAH Gene, their children may not develop PKU. This is because each of the child's parents has two versions of the PAH gene, only one of which passes during conception.

Galactosemia is a disease in which the body reacts to the processing of simple sugar, called galactose. Signs and symptoms of galactosemia are caused by the inability to use galactose to produce energy. Researchers have identified several types of galactosemia. Each of these conditions is caused by mutations in a particular gene and affects different enzymes involved in the breakdown of galactose.

Mutations in the GALT, GALK1, and GALE genes cause galactosemia. Mutations in the GALT gene cause classical galactosemia (Type I). Most of these genetic changes virtually eliminate the enzymatic activity generated by the GALT gene, leading to life-threatening signs and symptoms of the disease. Type II galactosemia is caused by mutations produced from the GALK1 gene, while mutations in the GALE Gene are based on Type III galactosemia. Enzymes formed from these genes play an important role in processing galactose. The lack of any of them allows galactose and related compounds to rise to toxic levels in the body. The accumulation of these substances damages tissues and organs, which leads to the characteristic features of galactosemia. This condition is inherited in an autosomal recessive pattern, meaning that both copies of the gene in each cell are mutated, but they usually show no signs and symptoms of the disease.

Leukodystrophy is a rare and very rare hereditary disease, in which the myelin sheath of connecting conductive axons between neurons and functional cells of the central nervous system is destroyed. A decrease in the concentration of myelin, which acts as a natural electrical insulator, leads to disturbances in the most complex electrochemical relationships that provide high nervous activity and leads to its gradual general degradation, death. The basis of progressive myelin deficiency is chronic deficiency of lysosomal enzymes in the body, which in turn results from hereditary mutation of specific genes. If both parents have such a false Gene, their child has a

25% chance of developing leukodystrophy. Currently, the only effective way to treat leukodystrophies is to transplant blood from the bone marrow or umbilical cord. This treatment allows, to some extent, to compensate for myelin deficiency and at least slow down the rate of progression of the disease. However, today leukodystrophy remains a serious disability disease with a poor prognosis.

The disease, caused by impaired purine and pyrimidine metabolism, arises from a deficiency of the enzyme hypoxanthine-phosphoribosyl-transferase (GKFBT) in the body. This enzyme promotes the conversion of guanine and hypoxanthine, Free-State purine compounds, into nucleotides. In the absence of an enzyme, the amount of uric acid in the body increases. The disease is diagnosed in a biochemical (screening and complex enzymatic analysis) manner.

Hemochromatosis. Genetic disease hemochromatosis is a rare pathology in which iron absorption in the small intestine is observed and its accumulation in the liver and other organs. The main factor leading to hemochromatosis is the presence of mutations in a number of genes, especially the HFE gene. It encodes a protein involved in regulating iron levels in the body and controls its absorption from food. Hemochromatosis proceeds autosomal recessive from relative to relative. This means that for the disease to manifest, there must be two defective copies of the gene, one from each parent. Both parents manifest themselves if the body contains mutated genes and accidentally developed under conditions in which they entered the child's genome. Therefore, the main way to confirm the presence of the disease is DNA analysis.

Conclusion. It should be noted that many hereditary diseases are complex diseases caused by a combination of hereditary and environmental factors (multifactorial diseases). One of the main tasks facing our medicine at the moment is the various hereditary diseases that arise from marriages between close relatives and their harmful consequences. Under the influence of such harmful consequences, we can see how many families give birth to their children in a disabled way. Understanding the true causes of hereditary diseases is important in the development of effective treatment and preventive measures. To prevent diseases of this type, medical and genetic consultations, a Reproductive Center, screening centers are operating.

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