HEMOGLOBINOPATHY

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Abstract*. Hemoglobinopathies are a group of severe hereditary blood diseases caused by a violation of the structure of hemoglobin or a decrease in the synthesis of one or more globin chains. The clinical picture is extremely varied. Common symptoms are hemolytic anemia, enlarged spleen, and bone damage. Diagnosis is carried out using peripheral blood smear microscopy, hemoglobin electrophoresis, and genetic studies. For treatment, transfusions of blood components, hydroxyurea preparations, and infusion therapy are used. In severe patients, splenectomy and stem cell allotransplantation are performed.*

Keywords*: **h**aematology, haemoglobin, molecular diagnosis, sickle cell disease, thalassemia, β-thalassemia intermedia.*

INTRODUCTION.

There are two main groups of hemoglobinopathies: abnormal structural variants of hemoglobin, caused by mutations in the genes encoding it, and thalassemias, which are caused by insufficient synthesis of normal hemoglobin molecules. The main structural varieties of hemoglobin are HbS, HbE and HbC. The main types of thalassemia include alpha thalassemia and beta thalassemia.These two pathologies can be combined because some conditions that cause abnormalities in the structures of hemoglobin proteins also affect their synthesis. Some structural variants of hemoglobin do not cause pathology or anemia and therefore are often not classified as hemoglobinopathies. Hemoglobinopathies have similar pathogenetic mechanisms. The altered structure of hemoglobin predisposes to intense hemolysis. Long-term anemia promotes compensatory bone marrow hyperplasia. Deformation of the skull bones and curvature of the spine occurs. Extramedullary foci of hematopoiesis develop, leading to an increase in the size of the liver and spleen (hepatosplenomegaly). As a result of splenomegaly, hypersplenism occurs - increased destruction of red blood cells by the sinusoids of the spleen.

MATERIALS AND METHODS.

Hemoglobinopathies are a series of congenital hemolytic anemias characterized by changes in the amino acid sequence of hemoglobin or suppression of the formation of globin chains. These pathologies often end in death in early childhood. About 50 types of hemoglobinopathies are known. The most common and life-threatening diseases are sickle cell anemia (SCA) and thalassemia. Hemoglobinopathies are autosomal recessive genetic diseases. Qualitative hemoglobinopathies develop due to mutations in genes responsible for the synthesis of certain amino acids in the beta chain of globin. As a result, one amino acid is replaced by another (glutamic acid with valine, lysine, etc.). This leads to the formation of abnormal hemoglobin, which is much less soluble than normal hemoglobin A, giving the red blood cells a different shape (target-shaped, sickle-shaped), which impairs their function and reduces life expectancy. Quantitative hemoglobinopathies are caused by mutations in genes that encode the entire globin chain (usually

alpha and beta). At the same time, the balance between globin chains shifts - with insufficient synthesis of alpha chains, an excess of beta chains occurs, and vice versa. The size of red blood cells decreases, their hemoglobin content decreases, and the membrane becomes more susceptible to various damages.

RESULT AND DISCUSSIONS.

Hemoglobinopathies are common in Central Africa and South Asia and are observed mainly in people of the Negroid race. Thalassemias also occur in Mediterranean countries. About 350,000 children are born each year with hemoglobin defects.Transport of oxygen from the lungs to the tissues: This is due to the special interaction of the globin chains, which allows the molecule to absorb more oxygen where the oxygen content is high and release oxygen when the oxygen concentration is low. In high-quality hemoglobinopathies, under the influence of reduced oxygen content in the blood, molecules of insoluble abnormal hemoglobin stretch the membrane of red blood cells, which leads to a change in their shape. Deformed red blood cells carry oxygen worse and are also able to adhere to the vascular endothelium, thereby clogging small vessels, causing thrombosis, occlusion and infarction.

Common complications of hemoglobinopathies are cholelithiasis and pathological fractures of long tubular bones. Heterozygous forms are rarely accompanied by adverse events, as they have a mild course. With quantitative hemoglobinopathies, heart failure, liver cirrhosis, and type 2 diabetes develop due to the deposition of excess iron in the internal organs.

Qualitative hemoglobin pathologies are characterized by a wide range of adverse consequences. The most dangerous are considered to be pulmonary embolism, myocardial infarction, stroke, which lead to death in approximately 10% of patients. Blockage of the microvessels supplying the bones leads to avascular necrosis of the femoral head (AFH). Due to constant infarctions of the spleen, functional aspleism occurs, which is why bacterial infections (bronchitis, pneumonia) often develop with a severe course, often with a fatal outcome.

Hemoglobinopathies are differentiated from other congenital hemolytic anemias (membranopathy, fermentopathy, microspherocytic anemia of Minkowski-Choffard). Persistent thrombosis must be differentiated from various thrombophilias. Iron overload should be distinguished from hereditary hemochromatosis. Anemia and ossalgia require the exclusion of malignant myeloproliferative diseases.

CONCLUSION

Why we need to learn it -Some hemoglobinopathies (and related diseases such as glucose-6-phosphate dehydrogenase deficiency) appear to have conferred an evolutionary advantage, especially to heterozygous organisms, in areas where malaria is endemic. Malarial plasmodia live inside red blood cells but interfere with their function. In patients predisposed to rapid red blood cell clearance, this may lead to early destruction of cells infected by the parasite and increase the chances of survival of the carrier of this trait. There are factors that provoke severe attacks (crises). These include dehydration, hypothermia, infections accompanied by high fever. In women, exacerbations often develop during pregnancy. But the main pathological stimulus of high-quality hemoglobinopathies is a decrease in the concentration of oxygen in the blood (hypoxia). This can happen, for example, when rising to a high altitude (climbing a mountain, flying on an airplane), where the partial pressure of oxygen in the air is reduced, or with severe diseases of the respiratory system (pneumonia).

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