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HEMOLYTIC DEFICIENCIES (MEMBRANOUS DISEASES, THALASSEMIA)

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Abstract: In this article, we will discuss the main aspects, diagnosis, and treatment methods of membrane pathologies and thalassemia diseases. Spherocytosis, Elliptocytosis, Thalassemias discussed, As a result of studies, it was found that the pathogenetic components of thalassemia (hemic hypoxia, hypersideremia) themselves lead to a decrease in cellular and humoral immunity and contribute to an increase in the level of the disease.

Key words: Thalassemias discussed, Hematological deficiencies, hemic hypoxia, Membrane, genetic testing.

Hematological defects are represented by functional and morphological disorders of blood and its components. In most cases, these diseases include hereditary diseases such as membrane pathologies and thalassemia. In this article, we will focus on the main aspects, diagnosis and treatment methods of membrane pathologies and thalassemia diseases. Membrane pathologies are associated with disturbances in the structure and function of the erythrocyte membrane. These diseases are often hereditary and arise as a result of mutations in the membrane of anucleated erythrocytes. Hematological defects, especially membrane pathologies and thalassemia, have a significant impact on human health. Their timely detection and proper treatment are of great importance in preventing the severe consequences of the disease. New medical technologies and genetic tests are increasing the guarantee of success in treating these diseases.

Spherocytosis

Spherocytosis is characterized by the loss of the normal disk shape of erythrocytes and their transformation into a sphere. This increases the sensitivity of erythrocytes and reduces their ability to withstand the effects of drugs. Main symptoms: Anemia (blood deficiency), Jaundice (jaundice), Splenomegaly (enlargement of the spleen)

Elliptocytosis

In elliptocytosis, the red blood cells take on an elliptical or cigar shape. This condition is often asymptomatic, and in some cases leads to anemia.

Thalassemia

Thalassemia is a hereditary disease associated with a defect in hemoglobin synthesis. It occurs as a result of a defect in the synthesis of the alpha or beta chains of hemoglobin.

Alpha-thalassemia

Alpha-thalassemia is associated with a defect in the synthesis of the alpha globin chains. There are varying degrees of this disease:

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• Incompatibility: May be asymptomatic.

• Alpha-thalassemia major: Moderate anemia.

• Hemoglobin H disease: Severe anemia and splenomegaly.

Beta-thalassemia

Beta-thalassemia is caused by a defect in the synthesis of beta-globin chains. There are three main forms of this disease:

• Incompatibility: May be asymptomatic.

• Thalassemia minor: Moderate anemia.

• Thalassemia major (Coulley anemia): The most severe form, requiring frequent blood transfusions and ongoing treatment.

• **Diagnosis**

Diagnosis of hematological disorders is based on blood tests, genetic tests and molecular medicine methods:

• Blood tests: Hemoglobin, hematocrit, shape and size of red blood cells.

• Genetic testing: To detect mutations.

• Molecular medicine: To examine the structure of hemoglobin.

4. Treatment

Treatment may vary depending on the type of disease:

• Blood transfusion: To correct anemia.

• Splenectomy (removal of the spleen): To reduce splenomegaly and rapid destruction of red blood cells.

• Gene therapy: To restore hemoglobin synthesis through new methods

Diagnostics

Diagnosis of hematological defects is carried out on the basis of blood tests, genetic tests and molecular medicine methods:

• Blood tests: Hemoglobin, hematocrit, shape and size of erythrocytes.

• Genetic tests: Identification of mutations.

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• Methods of molecular medicine: Examination of the structure of hemoglobin.

The vast majority of patients - 99 (78.2%) suffered from β-thalassemia (hemoglobinopathy), 25 (19.4%) - fermentopathy and 3 (2.4%) patients - microspherocytic anemia. Thalassemia major was characterized by the following symptoms: regressing anemia with normoblastosis, earthy-icteric coloration of the skin of varying severity, hepatosplenomegaly, hyperbilirubinemia and urobilinuria, osteoporosis, causing peculiar changes in the bones of the face and cranial vault and a sharp lag in sexual and physical development from their peers.

Conclusion

Hematological disorders, especially membrane pathologies and thalassemia, have a significant impact on human health. Their timely detection and proper treatment are essential to prevent the severe consequences of the disease. New medical technologies and genetic testing are increasing the chances of success in treating these diseases.

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