LABORATORY DIAGNOSIS OF THALASSEMIA

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ЛАБОРАТОРНАЯ ДИАГНОСТИКА ТАЛАССЕМИИ

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Annotation. Laboratory diagnosis of thalassemia occupies an important place in the modern approach to the treatment and prevention of this hereditary disease**.** Thalassemia, a group of disorders involving hemoglobin synthesis, can cause serious complications including anemia, cardiovascular problems and growth retardation. Early diagnosis is critical to selecting optimal treatment methods and monitoring the patient's condition.

Key words: thalassemia, laboratory diagnostics, modern research methods, pathogenesis.

Аннотация. Лабораторная диагностика талассемии занимает важное место в современном подходе к лечению и профилактике этого наследственного заболевания. Талассемия, представляющая собой группу нарушений, связанных с синтезом гемоглобина, может вызывать серьезные осложнения, включая анемию, проблемы с сердечно-сосудистой системой и задержку роста. Ранняя диагностика имеет решающее значение для выбора оптимальных методов лечения и мониторинга состояния пациента.

Ключевые слова: талассемия, лабораторная диагностика, современные методы исследования, патогенез.

Introduction. Laboratory diagnosis of thalassemia plays a key role in identifying and managing this inherited disease. Thalassemia is a group of inherited diseases resulting from mutations in the genes responsible for the synthesis of globins, which are the main components of hemoglobin. These mutations can lead to changes in the structure or quantity of globins, which in turn causes disruption of hemoglobin formation and leads to anemia. Depending on the genes affected, thalassemia is divided into alpha and beta thalassemia, each of which has its own subtypes, differing in clinical manifestations and severity of the disease.

The etiology of thalassemia is associated with genetic predisposition passed from parents to children. In most cases, the disease occurs as part of an autosomal recessive inheritance mechanism. This means that for a person to exhibit symptoms, they must inherit the mutated genes from both parents. A high prevalence of thalassemia is observed in regions where malaria was endemic, as carriage of the mutation provides some protection against this disease [1, 4, 10].

The pathogenesis of thalassemia is associated with impaired synthesis of globin chains, which leads to an abnormal form of hemoglobin and, accordingly, anemia. The main mechanism is mutations affecting the genes encoding alpha and beta globins. As a result, the balance between alpha and beta chains is disrupted, which leads to the formation of unstable forms of hemoglobin and their destruction in the bone marrow and peripheral blood.

There are two main forms of thalassemia: alpha and beta thalassemia, each with its own specific genetic causes and clinical manifestations. Alpha thalassemia is characterized by a deficiency of alpha globin chains, while beta thalassemia is associated with a deficiency of beta chains. The severity of the disease depends on the number of mutations and the level of expression of the corresponding genes, which can vary from asymptomatic carriage to severe anemia requiring regular blood transfusions [2, 9, 15].

In addition, disturbances in the pathogenesis of thalassemia may manifest themselves in the form of changes in erythrocyte morphology and additional activation of interactive mechanisms, such as hyperactivity of the cell working to clear the body of abnormal cells. These aspects lead to various complications, including changes in the function of the liver, spleen and other organs, which can lead to serious long-term consequences for the health of patients [1, 16, 17].

Laboratory diagnosis of thalassemia plays a key role in identifying and managing this inherited disease. Essential elements of diagnosis include clinical examination, laboratory tests, and genetic counseling. Initially, general blood tests are examined,

which may reveal anemia, decreased hemoglobin levels, and anisocytosis. For a more in-depth analysis, hemoglobin electrophoresis is performed, which allows one to determine the levels of different types of hemoglobin and confirm the diagnosis of thalassemia. There is also molecular genetic testing that allows one to identify specific mutations in the genes responsible for hemoglobin synthesis [2, 5, 11].

Modern laboratory diagnosis of thalassemia is an important aspect of medicine aimed at early detection and management of this hereditary disease. One of the key methods is DNA diagnostics, which makes it possible to identify mutations in genes responsible for the synthesis of alpha and beta globins [3, 8, 14]. This approach not only confirms the diagnosis, but also helps in prenatal diagnosis, which is especially important for families at risk of transmitting the disease. The development of new technologies, such as next generation sequencing, significantly increases the accuracy and speed of diagnosis.

In addition, a comprehensive approach to diagnosis includes analysis of inflammatory markers, iron and antioxidant status, which allows identifying associated conditions and tailoring therapeutic strategies. Thus, modern laboratory diagnosis of thalassemia is undergoing significant changes, which opens new horizons for effective control of this disease.

Treatment of thalassemia requires a comprehensive approach and depends on the type of disease: alpha or beta thalassemia. The main goal of therapy is to maintain normal hemoglobin levels and prevent complications. One of the key treatments is regular blood transfusions, which help replenish the lack of red blood cells and hemoglobin. However, long-term use of this method can lead to excessive accumulation of iron in the body, which requires the additional use of chelating agents [1, 7, 13]. These drugs help remove excess iron and help reduce the risk of serious complications such as liver damage and cardiovascular disease.

Gene therapy and stem cells are becoming increasingly popular for the treatment of thalassemia. They have the potential not only to improve the quality of life of patients, but also to eliminate the very cause of the disease. It is important that patients and their families are involved in the decision-making process regarding treatment, as each treatment has its own risks and benefits [3, 18, 19].

Previously, severe forms of thalassemia (such as beta thalassemia major) usually resulted in death in early childhood. The situation has changed in recent decades. Now, periodic blood transfusions in combination with therapy aimed at removing excess iron often allow patients to live into middle age and even old age. However, it should be remembered that this therapy does not lead to a complete cure and should be lifelong [3, 6, 12].

Bone marrow transplantation, if successful, leads to normalization of hematopoiesis, but its implementation is associated with certain risks, especially if the patient does not have a compatible related donor.

Conclusions. Thus, laboratory diagnostics not only serves as the basis for determining the presence of the disease, but also contributes to more effective treatment and improvement of the quality of life of patients. Modern laboratory diagnostic methods, such as molecular genetic studies, make it possible not only to confirm the diagnosis, but also to identify carriers of the thalassemia gene in an asymptomatic state. This is especially important for populations where the incidence is high, as it makes it possible to promptly inform patients and conduct genetic counseling to prevent the birth of children with severe forms of the disease.

In addition, regular screening and monitoring of the health status of people with thalassemia helps to identify the development of complications and improve the quality of life of patients. The inclusion of innovative technologies in laboratory diagnostics also contributes to a more accurate and rapid determination of the necessary treatment, which makes it a relevant and integral part of modern medicine.

It is important to consider family history and genetic testing since thalassemia runs in families. Psychosocial support for patients and their families is also an integral part of treatment, as living with thalassemia requires constant attention and care.

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