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FEATURES OF THE COURSE AND LABORATORY DIAGNOSIS OF SICKLE CELL ANEMIA

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Abstract
Sickle cell anemia is one of the most pressing diseases in the field of hemato-oncology, affecting millions of people worldwide. This hereditary disease, caused by a mutation in the gene responsible for the synthesis of beta-globin, leads to a change in the shape of red blood cells, which in turn causes their abnormal aggregation, impeding blood circulation and reducing the level of oxygen in the tissues. The relevance of the problem lies not only in the high prevalence of the disease, but also in the complexity of its diagnosis and treatment. This hereditary disease, caused by a mutation in the gene responsible for the synthesis of beta-globin, leads to a change in the shape of red blood cells, which in turn causes their abnormal aggregation, impeding blood circulation and reducing the level of oxygen in the tissues. The relevance of the problem lies not only in the high prevalence of the disease, but also in the complexity of its diagnosis and treatment.

Keywords: Sickle cell anemia, clinical symptoms, laboratory diagnostics, treatment.

Introduction

Sickle cell anemia is an inherited blood disorder caused by a mutation in the gene responsible for the synthesis of β-globin. The main cause is the substitution of adenine for thymine in codon 6 of the HBB gene, which leads to the formation of an aberrant hemoglobin known as hemoglobin S**.** When oxygen levels are low, these molecules clump together, taking on a sickle shape, making red blood cells less elastic. These abnormal cells impede blood circulation, leading to congestion and oxygen starvation of tissues.

Other causes of the disease may include heredity, as sickle cell anemia is inherited in an autosomal recessive manner. People with one copy of the mutated gene, known as "sickle cell carriers," may not show symptoms, but in conditions of stress, infection, or high altitude, health risks are significantly increased [1, 8, 17].

The clinical symptoms of this condition include a number of manifestations, primarily anemia, which manifests itself as fatigue, weakness, and shortness of breath. Frequent crises of pain caused

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by the blockage of small vessels by sickle cells become a characteristic feature. These crises can affect various organs, including the chest, abdomen, and joints, causing intense pain [1, 5, 19].

In addition, patients often experience infections, especially pulmonary infections, due to insufficient spleen function. Skin manifestations such as leg ulcers may also result from chronic hypoxia and impaired circulation. Chronic anemia can lead to damage to organs such as the liver and kidneys, requiring close medical monitoring and a comprehensive approach to treatment, including regular transfusions and drugs that stimulate the production of normal red blood cells.

Laboratory diagnostics of sickle cell anemia is based on the detection of specific changes in the blood associated with the presence of abnormal red blood cells. The main method is blood microscopy, which allows visualization of sickle cells, which have a characteristic shape that leads to disruption of microcirculation and tissue oxygenation [3, 12, 13].

A complete blood count (CBC) for sickle cell anemia is important for diagnosis and monitoring of the patient's condition. This disease is characterized by characteristic changes in blood counts. The main symptom is a decrease in hemoglobin levels, which leads to anemia. As a result, a complete blood count may show low red blood cell and hematocrit values, as well as a high reticulocyte level, indicating a compensatory response of the bone marrow [1, 11, 14].

Despite anemia, the formed elements may remain within normal limits, but it is important to identify features such as the presence of sickle-shaped red blood cells, which can be determined using specialized research methods. In addition, the CBC may show signs of hyperleukocytosis and changes in platelets, which indicate possible complications, including infections and thrombosis.

In addition, molecular genetic methods are used to confirm the diagnosis and identify mutations in the HBB gene, which is responsible for the production of beta globin.

Molecular genetic methods play a key role in the study of sickle cell anemia, a hereditary disease caused by a mutation in the HBB gene encoding the beta chains of hemoglobin. Modern technologies such as polymerase chain reaction (PCR) and next-generation sequencing allow for the highly accurate identification of mutations responsible for the disease [2, 10, 15].

These methods not only facilitate early diagnosis, but also open up new horizons for the development of targeted therapies. For example, the use of CRISPR-Cas9-based gene therapy shows promising results in correcting mutations, which can significantly improve the quality of life of patients.

In addition, molecular genetic studies can establish genetic predisposition to the disease, which plays an important role in individualized treatment strategies and preventive measures.

The hemoglobin electrophoresis test for sickle cell anemia is an important diagnostic tool for determining the types of hemoglobin present in the patient's blood. During electrophoresis, the blood is separated into fractions depending on the charge of the hemoglobin molecules, which allows the presence of hemoglobin S, responsible for the pathological condition, to be detected.

This analysis not only confirms the diagnosis, but also helps to assess the severity of anemia, as well as the risk of complications. The clinical significance of electrophoresis is that it can also detect other forms of hemoglobin, such as hemoglobin C and D, which is important for determining a more accurate prognosis and choosing a treatment strategy [3, 9, 16].

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Sickle cell disease causes a variety of complications, including chronic anemia, abdominal pain, organ damage, and an increased risk of infections. The prognosis for people with SCD depends on many factors, including the extent of the disease, access to medical care, and adherence to doctor recommendations.

Modern approaches to treatment and management of the disease have significantly improved the prognosis. Methods such as chemotherapy, stem cell transplantation, and new biological drugs have helped many patients achieve improvement in their condition. However, it is important to note that early diagnosis and regular medical monitoring remain key aspects in the management of SCD [2, 6, 18].

Some patients achieve a normal life expectancy and high quality of life with proper treatment and family support.

Treatment of sickle cell anemia is a multifaceted process aimed at improving the quality of life of patients and reducing the incidence of complications. The main goal of therapy is to prevent crises associated with microthrombosis and maintain normal hematocrit levels. The mainstay of treatment is hydroxyuric acid and transfusion therapy, which help reduce the number of sickle cells and increase the level of normal red blood cells.

Conclusions:

Thus, current research is aimed at studying the mechanisms of the disease, as well as developing new therapeutic approaches, including genetic therapy and innovative methods of symptom management. Raising public and medical awareness of sickle cell disease is an important step towards improving diagnosis and access to needed health care.

Psychosocial support for patients and their families is equally important in helping them adapt to the difficult challenges of living with a chronic illness. Specialized rehabilitation and education programs can significantly improve quality of life and provide hope for better health. Each individual approach to sickle cell disease treatment creates synergies that contribute to advances in this area of medicine.

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