RESEARCH METHODS FOR HEREDITARY ANEMIES

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Hereditary anemias are a heterogeneous group of diseases caused by genetic defects affecting hemoglobin synthesis, erythrocyte structure, enzyme systems, or erythropoiesis processes. Modern diagnosis of these conditions is based on a comprehensive approach, including hematological, biochemical, molecular genetic, and instrumental research methods [2,4,8,9].

Keywords: *general blood count, erythrocytes, hemoglobinopathy, membranopathy, reticulocyte index;*

The primary stage of diagnosis is a general blood test with an assessment of erythrocyte indices (MCV, MCH, MCHC, RDW) that allow for the assumption of anemia type (microcytic, normocytic, macrocytic). Morphological examination of a peripheral blood smear is important for identifying characteristic changes in erythrocytes, such as spherocytes, ovalocytes, schizocytes, and target cells, which allows for suspicion of membranopathy or hemoglobinopathy [1,3,7,9].

Among specialized methods, hemoglobin electrophoresis and high-performance liquid chromatography (HPLC) play an important role in diagnosing hemoglobinopathies, including thalassemia and structural

hemoglobin variants. These methods allow for the quantitative and qualitative assessment of hemoglobin fractions (HbA, HbA2, HbF, and pathological forms) [4,5,6,8,10,11].

To diagnose hereditary membranopathies (e.g., hereditary spherocytosis), tests for erythrocyte osmotic resistance are used, as well as more modern methods such as flow cytometry using eozine-5-maleimide binding (EMA test), which possesses high sensitivity and specificity.

The study of erythrocyte enzyme systems is crucial when enzyme pathology is suspected, such as glucose-6-phosphate dehydrogenase (G6PD) or pyruvate kinase deficiency. Determining the activity of the relevant enzymes allows for the confirmation of the diagnosis and the assessment of the severity of metabolic disorders.

Molecular genetic methods occupy a central place in the modern diagnosis of hereditary anemias. Polymerase chain reaction (PCR), DNA sequencing (including next-generation methods, NGS), and mutation analysis allow for the precise identification of genetic defects underlying the disease. These methods are particularly important for confirming the diagnosis, conducting prenatal diagnostics, and providing genetic counseling [16,17,18,19].

Additionally, methods for assessing hemolysis are used, including determining bilirubin levels, lactate dehydrogenase (LDH), haptoglobin, and the reticulocyte index, which reflects bone marrow activity. Direct and indirect Kumbs tests allow for the differentiation of hereditary anemias from autoimmune hemolytic states.

Instrumental methods, such as ultrasound examination of abdominal organs, are used to evaluate splenomegaly and complications associated with chronic hemolysis [12,13,14,15].

Thus, modern research methods for hereditary anemias are characterized by high informativeness and are aimed at a comprehensive assessment of the morphological, biochemical, and genetic characteristics of the disease. The integration of various diagnostic approaches ensures accurate diagnosis verification, determines treatment tactics, and allows for effective medical and genetic counseling [20,21,22,23,24].

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