MODERN MEDICINE AND PRACTICE
https://inovatus.es/index.php/ejmmp
EUROPEAN JOURNAL OF MODERN MEDICINE AND PRACTICE
Vol. 4 No. 9 (Sep - 2024) EJMMP ISSN: 2795-921X

HEREDITARY MICROSPHEROCYTIC HEMOLYTIC ANEMIA (MINKOWSKI-CHAUFFARD DISEASE)

Ruziev Zarif Muhammadovich
Department of Hematology and Clinical Laboratory Diagnostics Bukhara State Medical Institute Bukhara, Uzbekistan

Abstract: Hereditary microspherocytic hemolytic anemia was first described over a century ago by the German physician Oskar Minkowski and the French therapist Anatole Chauffard, who singled out the disease from the group of hemolytic syndromes as a special nosology. This disease has not lost its relevance even today, as it is still a widespread disease of the hereditary type with an incidence rate of 1:5000 live-born children of both sexes, including mild and subclinical forms - 1:2000 live-born children of both sexes. Minkowski - Chauffard hemolytic anemia is encountered quite often in the practical work of a pediatrician, therefore the main methods of diagnosing this disease, providing assistance to children in a state of hemolytic crisis, as well as recommendations during the remission period should be well known to a pediatrician in any area of practical work.

Key words: hereditary microspherocytosis, hemolytic anemia, clinical case.

Congenital microspherocytic hemolytic anemia is a familial disease inherited in an autosomal dominant pattern. The disease is based on a genetic defect in the erythrocyte membrane protein, which increases its permeability to sodium ions, which leads to swelling of the erythrocytes, impaired ability of the erythrocytes to deform, splitting off part of their surface in the spleen, shortening their lifespan and destruction by splenic macrophages. The pathology of the erythrocytes is manifested by a morphological anomaly - microspherocytosis.

The central place in the clinical picture belongs to the hemolytic syndrome, which is manifested by three cardinal signs: jaundice, splenomegaly and anemia. Signs of delayed development, as well as facial skeletal abnormalities in the form of a "tower skull", saddle nose, high palate, abnormal teeth placement, and narrow eye sockets may be observed. The severity of the anemic syndrome varies. A moderate decrease in hemoglobin is often observed. Some patients do not have anemia at all. The most severe anemia is observed during a hemolytic crisis. Microspherocytic hemolytic anemia has a chronic course and is accompanied by periodic crises and remissions. Hemolytic crisis occurs under the influence of provoking factors (infection, hypothermia, fatigue, pregnancy, etc.) and is manifested by a sharp increase in symptoms against the background of continuously ongoing hemolysis. In this case, the temperature rises due to the massive breakdown of red blood cells, the intensity of jaundice increases, hepatomegaly is pronounced , the spleen is dense and smooth, often painful as a result of tension of the capsule during blood filling or perisplenitis . Hemolytic disease is often complicated by attacks of hepatic colic, due to the formation of pigment stones in the gallbladder and bile ducts. Due to attacks of hepatic colic and stagnation of bile in the liver, patients may experience symptoms of angiocholecystitis and parenchymatous hepatitis with the appearance of direct bilirubin in the blood. During an exacerbation of the disease, a tendency to nosebleeds is observed. Despite

the congenital nature, the disease only rarely manifests itself in the first days after birth, usually symptoms appear in childhood, more often at 3-10 years, or adulthood.

The necessary treatment methods are selected individually, determined by the severity of the disease, the patient's age, the presence of developmental anomalies and their expressions

As an example we give a clinical case of hereditary microspherocytic hemolytic anemia , diagnosed at the child is 21 years old.

From the anamnesis of life it is known that the child was born a girl from II pregnancy, 2nd child flowing on background chronic iron deficiency anemia of moderate severity. Weight at birth - 3200 G., length bodies - 44 cm.

The patient's mother said that in 2010, her daughter sought medical attention at her local medical facility due to complaints of jaundice, recurring from time to time on the skin and sclera of the eyes. dizziness, general weakness. She was treated in hospital with a diagnosis of hepatitis, the form of which she does not remember. After treatment, jaundice on the skin and sclera decreased, but periodic signs of jaundice persisted. During 2011 and 2019, she was treated several times with a diagnosis of hepatitis in the infectious diseases department. During the examination, the general condition is moderate, conscious, the mucous membranes of the skin are yellow, peripheral lymph nodes are not palpable, there is no leg edema. Vesicular breathing in the lungs. Muffled heart sounds. The pulse is rhythmic, 88 per 1 minute. Blood pressure is 120/80 mm Hg in the right arm and 120/70 mm Hg in the left arm. The abdomen is soft and painless. The liver is not palpable. The spleen is +2.5 cm. Pasternatsky's symptom is bilaterally negative. Diarrhea and diuresis are regular. Complete blood count: Hemoglobin - 105 g / l, Erythrocytes - 3.9 * 10 12 / l, C / p - 0.81; Reticulocytes - 131 ‰, Platelets - 378 * 10 9 / l, Leukocytes - 9.9 * 10 9 / l, Leukocyte formula: rod nuclear - 1%, segmented nuclear - 69%, lymphocytes - 22%, monocytes - 6%, monocytes - 2%, ESR - 4 mm / hour. Erythrocyte morphology: microspherocytes 40-45%, normocytes 100: 4. Blood biochemistry: ALT - 23 U / L , AST - 42 U / L , Total bilirubin - 65.7 μmol / L, Direct bilirubin - 8.3 μmol / L, Indirect bilirubin - 57.4 μmol / L. Osmotic resistance of red blood cells: beginning 0.5% (normal 0.4%), end 0.4% (normal 0.3% complete hemolysis). Direct Coombs test is negative. Ultrasound conclusion: Diffuse liver changes, cholecystitis.

Based on the examination, the diagnosis was: D – 58 Hereditary microspherocytic hemolytic anemia.

In recent years, hereditary microspherocytic hemolytic anemia has been the subject of intensive research and therefore has recommendations such as:

1. Patients with newly diagnosed NMHA with a family history, typical clinical manifestations (anemia, jaundice and splenomegaly) and laboratory data (spherocytes in a peripheral blood smear, increased MCHC, increased reticulocyte count) do not require any additional examinations.
2. If the diagnosis is ambiguous, for example, in cases where the peripheral blood smear contains a small number of spherocytes and there are no other laboratory, clinical or family data, laboratory tests with high information content can be used: cryohemolysis , EMA test. The high prognostic value of both tests for the diagnosis of NS can be improved in combination with clinical data and erythrocyte indices.
3. Erythrocyte membrane protein electrophoresis can be used solely as a supplementary laboratory test. 4. Confirmation of the diagnosis may be necessary when laboratory test results are equivocal or borderline. Red blood cell membrane protein electrophoresis is the method of choice. This method is informative for revealing the degree of membrane protein deficiency in a patient. The main disadvantage of this method is low sensitivity in mild and asymptomatic forms. Red blood cell membrane protein electrophoresis is indicated in the following cases: 10 - when the clinical phenotype is more severe than that expected based on red blood cell morphology; - when red blood cell morphology is more severe

than that expected based on the parent's blood test, if the parent has NS; - if the diagnosis is unclear before splenectomy and the patient may have a monovalent cation permeability abnormality. If the morphology is typical, there should be no doubt. In more questionable cases (when MCV>100 fL), clarification may be required. 5. The diagnosis of NS does not require further molecular genetic testing to identify gene mutations.

Treatment of NMHA. Surgical tactics splenectomy is a very effective method when it is necessary to reduce hemolysis and increase the lifespan of red blood cells. Clinical manifestations and the risk of complications (gallstones) are significantly reduced in severe forms of NS and are completely stopped in milder forms, but the risk of life-threatening sepsis from encapsulated microorganisms, especially Streptococcus, increases. pneumoniae . Recent data demonstrate that splenectomy in children with NS is quite safe (in the short term, no fatal outcomes, rare complications have been noted).

Indications for splenectomy: Severe form at the age of not earlier than 3 years; Moderate form at the age of 6-12 years; Mild form - in the presence of stones in the gallbladder during simultaneous splenectomy and cholecystectomy at any age over 6 years; with high bilirubinemia and reticulocytosis with normal Hb at the age of over 6 years (to prevent the development of cholelithiasis). The choice of technique for performing splenectomy (endoscopic or laparotomic) is made by the surgeon. Preference is given to the endoscopic method due to a decrease in pain syndrome, a decrease in the length of the patient's stay in hospital and a good cosmetic effect. Partial resection of the spleen and endovascular occlusion of the spleen are not recommended due to the high risk of complications (postoperative splenosis in the first case, severe adhesive disease in the other case) and the short-term effect.

LITERATURE.

1. Okorokov A.N. Diagnostics of diseases of internal organs: Guide // Moscow: Medical literature, 2001. - V. 4. - P. 112-153.
2. Handbook of Hematology edited by Vorobyov A.I. Moscow VA : Medicine. – 1985. – T. 2. – P. 46-93.
3. Pogorelov V.M., Kozinets G.I., Kovaleva L.G. Laboratory and clinical diagnostics of anemia. - Moscow. Medical Information Agency, 2004. - P. 136-137.
4. Kokolina V.F., Rumyantsev A.G. Practical guide to childhood diseases. Hematology/Oncology of childhood. - Volume 4. - M.: Medpraktika -M, 2004. - P. 169-172. Vorobyov A.I. Handbook of hematology. - Volume 2. - M.: Meditsina, 1985. - P. 46-54.
5. Kishkun A.A. Guide to laboratory diagnostic methods . M.: GEOTAR-Media, 2007. - pp. 468-680.
6. Kassirsky I.A., Alekseev G.A. Clinical hematology. - M.: Medicine, 1970. - P. 235-242.
7. Lugovskaya S.A., Pochtar M.E. Hematological atlas. - M.: Triada, 2004. - 227 p.
8. Kolokolov G.R., Gerasina E.V., Ananyev O.L., et al. Analyses. Complete reference book. — M.: Eksmo , 2008. — P. 497-499.
9. Maksimov V.A., Damidovich K.K., Fedorchuk A.N. et al. Rare diseases, clinical syndromes and symptoms of diseases of the digestive organs. - M.: AdamantЪ , 2007. - P. 128-129.
10. Rukavitsin O.A., Skvortsov S.V., Zenina M.N. Hematology. Atlas-directory. St. Petersburg: Detstvo-Press, 2009. - pp. 219-220.
11. Pogorelov V.M., Kozinets G.I., Dyagileva O.A. et al. Color atlas of blood system cells. - M.: Practical Medicine, 2007. - P. 98-99.