INCIDENCE, LABORATORY FEATURES AND TREATMENT OF HEMOPHILIA C IN THE REPUBLIC OF UZBEKISTAN

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Hemorrhagic diseases is a generalized name for a group of diseases in the presence of which a person's ability to clot blood is impaired.

Hemorrhagic diseases are considered to be diseases such as hemophilia A, B and C, von Willebrand disease, and other rare diseases that can manifest in severe, moderate and mild forms. With modern treatment, people with hemorrhagic diseases are able to lead a normal and productive life.

At the same time, limited access to treatment can lead to serious consequences in the life of a person with this disease. In fact, bleeding that is not properly treated can be critical and even life-threatening, such as cerebral hemorrhage. All hemorrhagic diseases are considered rare, as they affect less than 5 people out of 10,000.

Despite the rarity of the diseases, diseases such as hemophilia and von Willebrand (the most common of them) are ahead of many other rare diseases, in in the sense that for them there is a diagnosis and various types of treatment.

The difficulty for patients is access to modern treatment, which is often expensive, as well as the availability of specialized medical services, which may be present in only one or two cities in the country.

Hemophilia is a hereditary disease associated with a defect in plasma coagulation factors, characterized by impaired blood clotting. Hemophilia appears due to a change in one gene on the X chromosome. There are three types of hemophilia (A, B and C).

Factor XI deficiency (hemophilia C-Rosenthal factor) is a fairly rare genetic disease (2G4900, 4q35), both dominant and recessive types of inheritance occur. The factor XI gene is located on chromosome 4 (next to the prekallikrein gene).

There are many known mutations that affect the course of the disease, and therefore the frequency and risk of bleeding does not always depend on the degree of factor XI deficiency.

Hemophilia C was first discovered in a young Ashkenazi Jew living in America in the 1950s.

The insufficiency of this factor was discovered in 1953 by Rosenthal. This disease is especially common among Ashkenazi Jews. According to statistics, it affects approximately 8% of this population of both sexes. In other nationalities, the disease occurs in 1% of the population.

It causes spontaneous bleeding and any surgical intervention may cause excessive blood loss, so it is recommended to avoid dangerous situations that may cause bleeding.

The minimum hemostatic level of factor XI activity in the blood for performing operations is 15-25%; with lower activity, the risk of developing postoperative bleeding is extremely high.

The minimum hemostatic level of factor XI activity in the blood to stop bleeding is 5-15%; with lower activity, stopping bleeding without administering factor XI to the patient is impossible. Hemophilia C is a mild form of hemophilia that affects both sexes.

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The pathology is characterized by a decrease in the activity of factor XI, which leads to disruption of the coagulation system. Unlike hemophilia A and B - hereditary deficiency of factors VIII and IX, respectively, spontaneous bleeding from soft tissues and hemarthrosis practically do not occur. More often, bleeding occurs after operations, injuries, and childbirth.

Bleeding from the nasal cavity, oral mucosa and urinary tract, that is, tissues with high fibrinolytic activity, is characteristic. After operations (appendectomy, orthopedic), the duration of bleeding may be longer, but it often stops after treatment. Clinical symptoms associated with factor XI deficiency are rarely related to its activity. Thus, even with a pronounced decrease in the indicator, severe violations are not observed. Women with a decrease in the factor in the blood may have heavy menstruation and prolonged bleeding after termination of pregnancy.

Quite often with hemophilia, bleeding occurs from the gums, nose, kidneys, and gastrointestinal tract. Bleeding can be initiated by any medical procedure (intramuscular injection, tooth extraction, tonsillectomy, etc.). Bleeding from the pharynx and nasopharynx is extremely dangerous for a child with hemophilia, as it can lead to airway obstruction and require emergency tracheostomy. Hemorrhages in the meninges and brain lead to severe damage to the central nervous system or death.

Hematuria in hemophilia can occur spontaneously or as a result of injuries to the lumbar region. In this case, dysuric phenomena are observed, with the formation of blood clots in the urinary tract - attacks of renal colic. In patients with hemophilia, pyelectasia, hydronephrosis, and pyelonephritis are often found.

Gastrointestinal bleeding in patients with hemophilia may be associated with taking NSAIDs and other drugs, with exacerbation of the latent course of gastric and duodenal ulcers, erosive gastritis, and hemorrhoids. With hemorrhages in the mesentery and omentum, a picture of an acute abdomen develops, requiring differential diagnosis with acute appendicitis, intestinal obstruction, etc.

Scientific novelty

Early detection and improvement of diagnosis and treatment of patients with Hemophilia C in the Republic of Uzbekistan, as well as reduction of disability and mortality among them.

Materials and methods of research. The study is based on a clinical analysis of dynamic observations of 17 patients with hemophilia C undergoing treatment at the

Republican Specialized Clinical Hospital of the Ministry of Health of the Republic of Uzbekistan. Among the examined patients were all males and females aged from 2 to 55 years, with an average age of 25.2 years.

All patients were examined by clinical, biochemical, coagulological and serological methods.

Results and discussion. Laboratory data before treatment:

APTT 81 sec., fibrinogen 4.8 g/l, prothrombin according to Quick 108%, FVII 102%, FII 98%, FV 100%; FVIII 14%, FIX 98%, FX 96%, FXI 5.9%, FW 101%, FXII 103%, platelet aggregation with ristomycin 65%, platelet aggregation with collagen 73%, platelet aggregation with ADP 69%. The absence of an inhibitor to factor XI allowed us to exclude acquired XI deficiency.

In the general blood test, the patients' hemoglobin was 107 g/l, erythrocytes 3.4 x 1012/l, platelets 190 x 109/l; leukocytes 6.8 x 109/l; in the biochemical blood test - total protein 69 g/l, albumin 40 g/l, alanine aminotransferase 42 units/l, aspartate aminotransferase 29 units/l, creatinine 85 μmol/l.

Laboratory data after treatment:

APTT 38 sec., fibrinogen 3.4 g/l, prothrombin according to Quick 115%, FVII 105%, FII 103%, FV 100%; FVIII 124%, FIX 98%, FX 106%, FXI 71.0%, FW 105%, FXII 101%, platelet aggregation with ristomycin 68%, platelet aggregation with collagen 78%, platelet aggregation with ADP 69%. In the general blood test of patients, hemoglobin increased by 112 g/l, erythrocytes 3.8 x 1012/l, platelets 202 x 109/l; leukocytes 7 x 109/l;

Taking into account the anamnestic, clinical and laboratory data, the patients were diagnosed with Hemophilia C. FFP transfusions were continued at a dose of 700 ml/day (8 ml/kg body weight), after stabilizing the patient's condition, the daily dose of FFP was reduced, he was discharged from the Russian Scientific and Practical Center for Hematology after 10 days.

Taking into account the anamnestic, clinical and laboratory data, the patients were diagnosed with Hemophilia S. FFP transfusions were continued at a dose of 700 ml/day (8 ml/kg body weight), after stabilizing the patient's condition, the daily dose of FFP was reduced, he was discharged from the Russian Scientific and Practical Center for Hematology after 10 days.

Thus, treatment is reduced to stopping bleeding that has occurred by transfusion of media containing factor XI (plasma, serum, recombinant blood coagulation factors XI, etc.).

The clinical effect of the transfusion usually lasts for three weeks, although laboratory deficiency of the factor is again detected after a few hours.

A key aspect of improving health status and quality of life in the treatment of Hemophilia C is the prevention of bleeding: life-threatening bleeding and hemorrhage (in the central nervous system (CNS), gastrointestinal tract (GIT), etc.).

In addition to specific hemostatic and replacement therapy, a hematologist may use additional medications in the treatment of patients with Hemophilia C, for example, hormonal therapy in women with recurrent severe uterine bleeding.

Tranexamic acid and other antifibrinolytic agents can be used to relieve menorrhagia and mild mucosal bleeding, with the exception of renal bleeding. Antifibrinolytic drugs can be used alone or in addition to clotting factor concentrates.