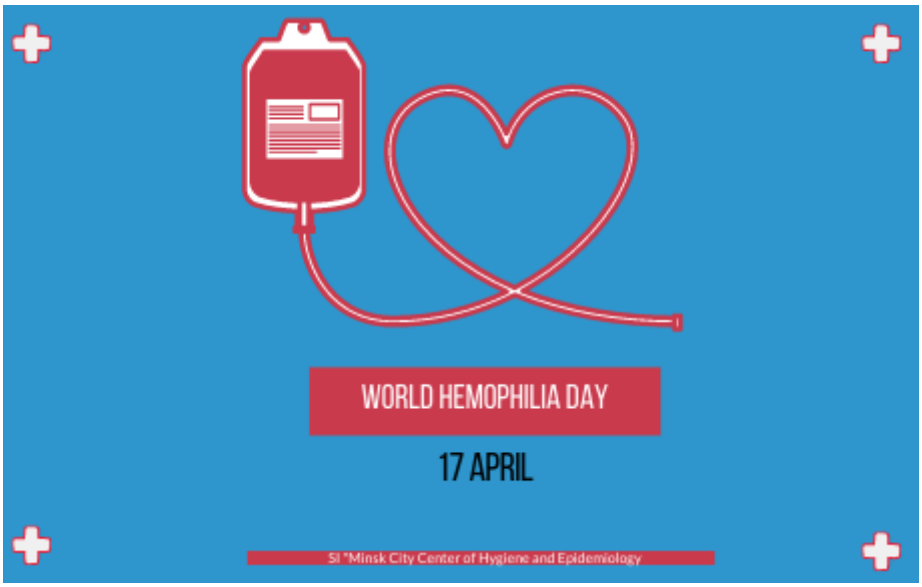
17 APRIL WORLD HEMOPHILIA DAY



Hemophilia is a rare hereditary disease associated with blood clotting disorders. The disease causes hemorrhages in joints, muscles, and internal organs, either spontaneously or as a result of trauma or surgery.

The clotting system ensures the preservation of blood inside the blood vessels when their integrity is compromised (injuries, medical interventions), preventing it from pouring out by closing the damage with special clots (thrombi). Clots are formed as a result of biochemical reactions between molecules present in each person's blood. In the study of the clotting system they are called "clotting factors.

The cause of increased bleeding in hemophilia is a disorder in the synthesis of plasma clotting factor molecules. In this regard, the following main forms of hemophilia are distinguished:

- Hemophilia A – caused by clotting factor VIII

deficiency;

- Hemophilia B – caused by factor IX deficiency;

Hemophilia A accounts for 80% of cases and hemophilia B for 12%. The remaining 8% account for other disorders caused by a defect in factor production or a lack of factors with other numbers.

The hemophilia genes are located in the X chromosome, which is passed from grandfather to grandchild through a healthy daughter, a carrier of the defective gene. That is, men usually suffer from the disease, while women act as carriers of hemophilia and may give birth to sick sons or daughters who are carriers. According to WHO statistics, about one male infant in 5,000 is born with hemophilia A, regardless of nationality or race.

Symptoms of hemophilia

The leading symptoms of hemophilia A and B are increased bleeding from the first months of life; subcutaneous, intermuscular, subfascial, retroperitoneal hematomas due to contusions, cuts, various surgical interventions; profuse post-traumatic bleeding; hemarthrosis of large joints, with secondary inflammatory changes.

In newborn children, signs of hemophilia may include prolonged bleeding from the umbilical wound, subcutaneous hematomas. Bleeding in children in the first year of life can be associated with teething, the sharp edges of baby teeth can cause biting of the tongue, lips, cheeks and bleeding from the mucous membranes of the mouth. However, hemophilia rarely deburs in infancy due to the fact that mother’s milk contains sufficient amounts of an active enzyme, thrombokinase, which can improve clot formation.

The likelihood of post-traumatic bleeding increases significantly when the child with hemophilia begins to get up

and walk. Nasal bleeding, subcutaneous and intermuscular hematomas, and large joint hemorrhages are common in children after one year of age. Exacerbations of hemorrhagic diathesis occur after infections (acute respiratory viral infections, chickenpox, rubella, measles, flu, etc.) due to impaired vascular permeability. In this case, spontaneous small hemorrhages often occur. Because of constant and prolonged bleeding, children with hemophilia develop anemia of varying severity.

A characteristic feature of hemophilia is the delayed nature of bleeding, which usually develops not immediately after injury, but some time later, sometimes 6-12 hours later.

Hemophilia is not necessarily hereditary. Spontaneous mutations in human DNA occur all the time. And so it is possible that hemophilia can occur in a family where no one has ever suffered from this disease – the so-called sporadic hemophilia. It is not so rare – in one third of all cases of the disease.

To diagnose hemophilia the following is used: coagulogram, determination of clotting time, adding plasma samples with the absence of one of the clotting factors.

Treatment of hemophilia

Substitution therapy is the main method of treatment for hemophilia. For this purpose, concentrates of VIII and IX clotting factors are used in individual doses for each patient and type of bleeding.

Prevention of hemophilia

In order to prevent the birth of a child with hemophilia, there is medical genetic counseling and prenatal diagnosis is possible.

Patients are counseled at the hematology departments of regional hospitals, counseling centers in large cities, and

the Republican National Perinatal Center.