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The Need for Prenatal Diagnosis of Hereditary Coagulopathies for Prophylactic Purposes in the Republic of Uzbekistan

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Abstract The article presents a literary review of hereditary coagulopathies, world data and the state on this problem in the Republic of Uzbekistan. Arguments are given for the widespread introduction of prenatal diagnostics. The goal of introducing prenatal diagnostics in all regions of the Republic is to prevent the birth of children with severe hereditary and congenital diseases, to identify and register pregnant women at risk of having children with hereditary defects.

Keywords Hemophilia, Von Willebrand disease, Hereditary coagulopathy, Prenatal diagnosis

In the development of hereditary pathologies, the main role is played by mutation in the structure of the gene or chromosome where the mutation of a certain part of the gene occurs. In the development of such diseases, the main role is played by mutations in the structure of the gene or chromosome. In addition, hereditary pathology also includes diseases with a hereditary predisposition - diseases (hemophilia A and B, cystic fibrosis, adrenogenital syndrome, Duchenne – Becker muscular dystrophy, phenylketonuria and a number of other diseases) [1].

Prenatal diagnosis is a set of highly skilled diagnostic methods that can be used to detect genetic disorders in the development of the fetus and identify chromosomal syndromes and diseases. At the moment, in Uzbekistan, prenatal diagnosis is one of the youngest and most developing areas in medical genetics [8].

Coagulopathy is a complex of processes in which the process of blood clotting is disrupted. The condition can be congenital or acquired. These diseases are dangerous with prolong clotting time, bleeding time which can result in both the development of chronic iron deficiency anemia and life threatening hemorrhage [4]. Hereditary coagulopathies are all genetically determined disorders in the blood coagulation system, which are associated with a deficiency or molecular abnormalities of 13 plasma coagulation factors and the components of the kallikrein-kinin system that take in this process [7].

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The hemostatic system, i.e. blood clotting is a multicomponent and very complex sequential mechanism that plays a critical role in the human body. This mechanism is carried out at the expense of 3 parts of the human body: vascular, platelet and plasma coagulation components [5]. Pathologies that are manifested by the inability of blood coagulation factors to implement and ensure the normal process of thrombotic clot formation constitute the most extensive group of hereditary coagulopathies. This group of diseases is represented by such pathologies as hemophilia A - 68-78% (cases of occurrence), von Willebrand disease - 9-18%, hemophilia B - 8-13%. These forms are called "prevailing", which the doctor should think about first when detecting signs of prolonged bleeding in a child [6].

There are also infrequent forms, which account for about 2-3% of coagulopathies and rare forms (casuistic) occurring in 0.01% of cases [3,9].

The cause of these mutations is believed to be the result of the combined action of environmental factors and a specific set of gene defect, which create conditions conducive to the development of the pathological process [3].

It can be argued that hereditary coagulopathies are quite rare - 1: 100,000 of the population, and are diagnosed even less often, prenatal diagnosis is completely absent for this type of pathology [2]. This is due to the fact that this type of pathology has: low prevalence, combined with a subclinical course of the disease (with mild forms of hemophilia and von Willebrand disease) and a far from perfect system of laboratory diagnostics for hemostatic disorders in a large number of regions of our state, or the absolute absence of this laboratory unit [4]. The number of patients with hemophilia in Uzbekistan is slightly more than 1.8 thousand people, and

with von Willebrand disease about 200. The exact figure has not been established, since according to preliminary calculations, taking into account world statistics, there should be about 3.5 thousand patients in our country, i.e. e. diagnosed just over half. And, such forms of hereditary coagulopathies as hypoproconvertinemia (deficiency of factor VII), hypo- and afibrinogenemia, deficiency of factors XII, XIII, XI, V are extremely rare.

Hemophilia is an inherited disease caused by mutations in the genes of factor 8 or 9 (F8 or F9) and their various types, as a result of which there is a hereditary deficiency of clotting factor VIII (hemophilia A) or IX (hemophilia B). These genes are localized on the long arm of the X chromosome and are inherited in a recessive manner, passing through the mother only to male offspring. In girls, hemophilia is very rare, but they do not live to be 15 years old, due to the development of massive uterine bleeding. In healthy people, the level of factors VIII and IX ranges from 100 ± 50%, but in women who are carriers of hemophilia, it may be lower than normal due to functional inferiority of one of the two genes. The severity of hemophilia is established depending on the activity of the level of factor VIII or IX in the patient's blood [1]. In severe hemophilia, factor VIII (or IX) is absent or remains active (less than 1%). In this case, the disease usually manifests itself from early childhood or even in the neonatal period, when the newborn does not stop bleeding from the umbilical wound.

Hereditary coagulopathies are an incurable pathology, and the recent possibilities of modern substitution therapy with blood coagulation factors, which provide patients with a longer duration and an adequate quality of life, leave much to be desired. These patients in early childhood become disabled with significant damage and defeats of the musculoskeletal system and are supported by the state for life. The quality of life of patients is low, because most of life is associated with the processes of treatment and rehabilitation, there is no opportunity to get a good education and profession, create a full-fledged family and support it. Hemophilia, von Willebrand disease and hypoproconvertinemia belong to the group of social diseases, and without the support of the state, people suffering from hereditary forms of coagulopathies are doomed to disability and vulnerability from any impact of a traumatic factor. Also invaluable is the social, economic, moral damage to parents and loved ones of patients with hemophilia.

The key point in the therapeutic efficacy of hereditary coagulopathies is: early diagnosis, selection of the correct treatment model and full, continuous provision of patients with expensive coagulation factors VIII or IX [1]. If patients do not receive substitution therapy or the amount of the drug is insufficient for treatment, the disease begins to progress steadily and can nullify all previous efforts. Since the majority of these patients are young people, the state suffers enormous economic losses in the form of disability benefits and the loss of valuable workers.

All over the world, these patients are managed in specialized hemophilia centers, in our republic there is also a Hemophilia center on the basis of the Republican Specialized Scientific and Practical Medical Center for Hematology of the Ministry of Health of the Republic of Uzbekistan. The success of this center is the introduction of preventive treatment for children under 5 years of age with hemophilia A and B providing them with expensive preparations of blood coagulation factors, which patients can receive both on an outpatient basis and at home. Also, a medical register of patients with hereditary coagulopathies is maintained, which allows tracking the dynamics of a patient's health over many years and adjusting the dose of drug treatment, which is based on national standards and clinical treatment protocols.

In developed countries, there are various approaches to prenatal diagnosis, today this is an extremely expensive diagnostic method and is practically inaccessible for our citizens. Since prenatal diagnostics of hemophilia in our republic is just beginning to develop, there is no way to obtain reliable data on the molecular genetic characteristics of patients with hemophilia, there is no way to reduce the number of patients with hemophilia.

The goal of introducing prenatal diagnostics in all regions of the Republic is to prevent the birth of children with severe hereditary and congenital diseases, to identify and register pregnant women at risk of having children with hereditary defects. Further research on this hereditary genetic disorder can significantly reduce the number of congenital pathologies and will become a priority for disadvantaged parents of our state. Knowing these issues will enable them to make wise decisions about prenatal diagnosis and decisions about the availability of treatment to prevent birth defects, thereby saving lives and reducing government spending on people with disabilities.

Thus, it is necessary to increase the knowledge of medical personnel about the indications for prenatal diagnosis and its importance for reducing the number of patients in our republic. It is necessary to develop and implement in the republic an algorithm for prenatal - DNA diagnostics of hereditary coagulopathy to prevent the birth of children with hereditary coagulopathies.

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