HEREDITARY DISEASES IN CHILDREN EARLY DETECTION AND PREVENTIVE MEASURES

Nazarova Firuza Ilxomovna
BITI medical biology science teacher
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ABSTRACT

By his decree of December 25, President of Uzbekistan Shavkat Mirziyoyev approved the state program for early detection of congenital and hereditary diseases in children for the period 2018-2022. In 2013-2017, more than 2.1 million pregnant women were examined at the primary health care level, 65 thousand of them had their pregnancies terminated due to the detection of congenital defects in fetal development.

In the 21st century, two major factors exert great pressure on the environment: the first is the rapid growth of the Earth's population, the second is the progress of technology and the impact on nature. A person interacts with the external environment that surrounds him. Water, air, plants, fauna, food, habitat, clothing, noise, vibration, radiation, various medicines, bioprophylactic drugs, modern airliners, various toxic chemicals used against agricultural pests, etc.-this is the environment surrounding humans. On the other hand, these factors directly affect a person's physical and mental health. Social problems in living conditions can also affect people's health. The impact of mutagenic (endogenous, teratogenic) factors, which are increasing in the biosphere due to environmental pollution, causes an increase in hereditary diseases transmitted from generation to generation. Hereditary diseases are very severe compared to other somatic diseases, many of which have not been found effective treatments to date. But the most basic way to prevent hereditary diseases in the current period is medical genetic counseling. Nowadays, our children, who will be born as a result of ecolgias-changes in the external environment, are very likely to be born with mental retardation, disabilities, defects and hereditary diseases. In order to reveal the reasons for this, an in-depth and comprehensive study of the Department of genetics is becoming relevant.

More than 1.1 million pregnant women in the prenatal period were examined in screening centers. Of these, more than 210 thousand women with pathological pregnancy received medical treatment. In general, prenatal examination of pregnant women made it possible to prevent the birth of a child with congenital developmental defects and genetic disorders in more than 21 thousand. More than 1.7 million infants were tested for hypothyroidism and phenylketonuria (congenital and hereditary diseases). Currently, 1,407 patients are under medical supervision, of which 1,117 children with congenital hypothyroidism are constantly provided with free medicines in order to prevent progressive

diseases that can later lead to disability, and 290 children with phenylketonuria are provided with therapeutic nutrition. As a result of the measures carried out, the birth rate of children with congenital anomalies decreased by 1.8 times compared to 2000. At the same time, the current stage of development of medical genetics requires strengthening and qualitative improvement of the material and technical base of screening centers to provide specialized high-tech assistance to expectant mothers and children. The decision provides for:

- improving the material and technical base of screening centers by equipping the balance of high-tech diagnostic medical equipment, components, reagents and zara materials;
- improving measures for the prevention and early diagnosis of congenital and hereditary diseases in the fetus through mass prenatal examination of women in central multidisciplinary district (city) polyclinics in the first three months of pregnancy and the phased introduction of biochemical examination of pregnant women at risk for genetic syndromes;
- the introduction of modern methods of laboratory diagnostics, including cytogenetic and molecular cytogenetic technologies in the diagnosis of chromosomal syndromes in fetuses and young children
- the introduction of modern methods of laboratory diagnostics, including cytogenetic and molecular cytogenetic technologies in the diagnosis of chromosomal syndromes in fetuses and young children
- improving the effectiveness of treatment of hereditary diseases by improving methods of early diagnosis of hereditary diseases in children through mass examination of infants, as well as providing sick children with medicines and medicinal food;
- strengthening the human resources of screening centers and implementing international cooperation with the introduction of modern world experience in practical healthcare. It is planned to allocate 31 million US dollars and 90 million soums for the implementation of the measures provided for by the state program, of which 28 million 460 thousand US dollars are funds from the state budget, 90 million soums and more than 2 million US dollars are funds from the international non-governmental charitable foundation "For a Healthy Generation".

Implementation of the state program:

- the introduction of mass ultrasound examination of pregnant women before childbirth in the central multidisciplinary polyclinics of medical associations of cities and districts;
- step-by-step biochemical blood testing of women in early pregnancy in order to identify a risk group for fetal genetic syndrome (it is planned to examine more than 220 thousand pregnant women);
- 310,000in biochemical screening of pregnant women until childbirth in the second trimester of pregnancy;
- examination of children for confirmation of hereditary exchange diseases by the method of Andem mass spectrometry (more than 18 thousand examinations);
- it makes it possible to reduce the birth rate of children with developmental disabilities by a factor of two.

In addition, the provision of food and medicines that specifically treat children with hereditary diseases who are registered at the dispensary and newly registered will continue.

The implementation of the planned measures will increase the coverage of newborn screening to 95 percent and increase the effectiveness of diagnostic and therapeutic measures for the early detection of children with hereditary diseases leading to childhood disability

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