PHENYLKETONURIA IN CHILDREN IN THE REPUBLIC OF BELARUS

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In Belarus, as in other countries with high level of medical care and low neonatal and perinatal mortality, congenital and hereditary pathology occupies one of the leading places in the stage of childhood morbidity, disability and mortality. Most congenital defects are revealed in early childhood.

Keywords: phenylketonuria, phenylalanine, screening.

Phenylketonuria (PKU) is an inherited disease that causes the accumulation of phenylalanine in the amino acid pattern. The pathogenesis of these phenomena is based on a hereditary metabolic disorder associated with a deficiency of the enzyme phenylalanine hydroxylase and an increased content of phenylalanine [1].

Delayed diagnosis and lack of treatment for PKU leads to mental retardation, seizures, behavioral problems and mental disorders. A child with this pathology has disturbances in embryonic development in the form of microcephaly, causing a delay in the intrauterine development of the cardiovascular system [2].

Correction of PKU is carried out using drug therapy, diet therapy with the prescription of specialized products based on mixed amino acids without phenylalanine, and the prescription of nutritional supplements using amino acids if they are deficient in diet therapy with a low phenylalanine content.

For early detection, appropriate treatment, prevention of the development of diseases and disability, as well as reduction of child mortality, neonatal screening of this pathology is carried out. This is a medical diagnostic technology for a continuous, sample-free laboratory examination of all newborns for metabolic diseases, the purpose of which is to ensure timely detection and initiation of treatment for sick children in order to prevent their disability. Every year, these screening programs help keep children alive and healthy.

In the Republic of Belarus, widespread neonatal screening of newborns is carried out for a number of hereditary diseases, including PKU. Mass screening of newborns is a cyclic component of preventive and personalized drug therapy aimed at the prevention and early diagnosis of diseases, the development of which is a priority in the activities of the healthcare system at the present stage [2].

In Belarus, more than 1000 people participate in PKU, 300 of them are under the age of 18. Every year, 15–20 children are born with an established diagnosis of PKU. For the period 2012-2021 more than 4,000,000 newborns were examined and more than 600 patients with PKU were identified, who, thanks to timely therapy, have normal intellectual development.

Antenatal and neonatal screening programs can reduce the cost to society of lifelong care for disabled children by preventing long-term disability.

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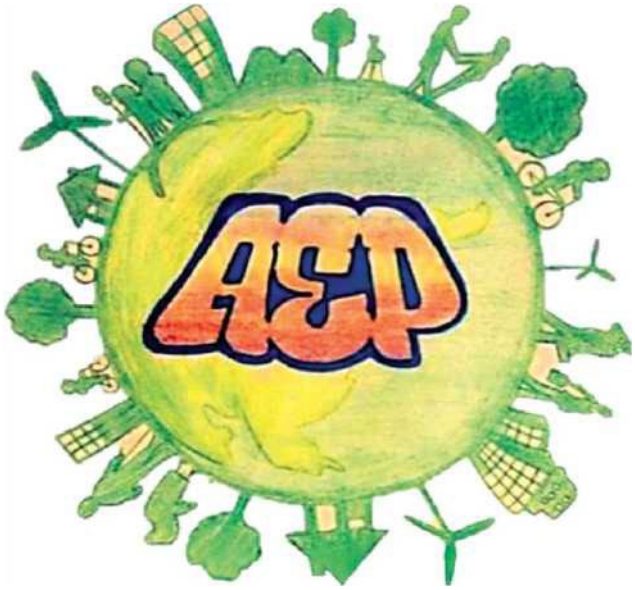

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