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ADVANCED NEONATAL SCREENING IN TASHKENT: FIRST RESULTS AND EFFECTIVENESS ANALYSIS

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Abstract
The thesis presents the first results and organizational experience of the implementation of the pilot program of expanded neonatal screening (RNS) in the city of Tashkent.

Introduction

The program, launched in 2023 on the basis of the Republican Center for Maternal and Child Screening, is aimed at the early detection of hereditary metabolic diseases (HBD), primary immunodeficiencies (PID) and spinal muscular atrophy (SMA) among newborns. The main diagnostic method was tandem mass spectrometry (TMS), supplemented by PCR research. The data of 45,000 newborns for the period January-December 2023 were analyzed. The total incidence of pathology was 1:1500 newborns. Disorders of amino acid metabolism were most often diagnosed. The analysis demonstrates the high efficiency and clinical significance of RNS in Uzbekistan, and also emphasizes the need to optimize logistics and further expand the program to the entire territory of the country.

Neonatal screening is the cornerstone of preventive medicine, which prevents disability and mortality of newborns from severe hereditary diseases. While many countries have introduced expanded screening programs covering dozens of nosologies, in Uzbekistan this process is at the stage of active development. The city of Tashkent, as a pilot region, initiated the implementation of the expanded neonatal screening (RNS) program in 2023. This study summarizes the first experience, describes the organizational structure and presents the frequency of detected diseases among the population of Tashkent.

The study is based on the analysis of data from 45,000 newborns born in 15 maternity hospitals in Tashkent in the period from January 1 to December 31, 2023.

Of the 45,000 newborns screened, 120 infants (0.27%) had a primary positive result. After retesting and in-depth diagnostics, the diagnosis was confirmed in 30 children. Thus, the overall frequency of the detected pathology in the sample was 1:1500 newborns.

The distribution of confirmed cases by disease group is presented in Table 1.

Table 1. Distribution of detected hereditary diseases by groups in Tashkent city (n=30)

Group of diseases	Number of detected cases	Frequency in the studied population
NBO (amino acids)	15	1 : 3 000
NBO (OFHK*)	8	1 : 5 625
Spinal muscular atrophy (SMA)	4	1 : 11 250
Primary immunodeficiencies (PID)	3	1 : 15 000
Altogether	30	1 : 1 500
OFFA – fatty acid oxidation disorders		

For the most common pathologies, the average values of key diagnostic markers in dried blood spots at the time of initial screening were calculated (Table 2).

Table 2. Average values of the main biochemical markers in the detected pathologies (TMS data)

Confirmed diagnosis	Key marker	Mean value (normal < 10 µmol/L*)	Number of cases
Leucinosis (BCS)	Leucine	45.7 µmol/L	6
Maple syrup disease	Valine	38.2 µmol/L	6
Glutaric aciduria type I	C5-DC	5.8 µmol/L (norm < 0.2)	3
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	C8	4.1 µmol/L (norm < 0.3)	5
Standard values may vary slightly depending on the method and reagents of the laboratory.			

The pilot program of expanded neonatal screening in Tashkent has proven its high diagnostic efficiency and medical feasibility. The detected incidence of hereditary pathology (1:1500) corresponds to world data and confirms the need for the widespread introduction of RNS in Uzbekistan.

The success of the program depends on a well-established organizational structure, including fast logistics, an equipped laboratory and a trained team of specialists. The data obtained are

the basis for scaling the program to other regions of the republic and for including new nosologies in the screening as the laboratory base develops.

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