ASPECTS OF THE LABORATORY DIAGNOSTICS OF CYSTIC FIBROSIS

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Introdution. Cystic Fibrosis(CF) – is the most common hereditary striking many organs pathology, which characterized by a pronounced genetic and clinical polymorphism. It is a monogenne disease caused by a mutation of a gene and is characterized by lesions of the hemadens of vital organs and is usually a heavy current and forecast. CF is an important medical and social problem. Previously, this disease is attributed to the category of "fatal" or "lethal" because life expectancy does not exceed 5 years. In our days, thanks to the deepening of knowledges about the CF and the elaboration of effective methods of therapy, the disease is diagnosed much earlier, and the average life expectancy of patients has increasedis.

Aim. The object of work is the determination of the optimal methods of diagnostics of this hereditary disease.

Materials and methods. Our research carried out on the basis of Kharkiv specialized medical-genetic Center and included molecular genetic study, neonatalstudy, pilokarpin test, neonatal, screening of new-born, which includes diagnosis of immunoreactive tripsin.

Results and discussion. Investigated levels of immunoreactive tripsin (IRT) in dry blood spots 191 newborn that has been done by means of fluorescent immune assay using multifunction Analyzer test and VICTOR DELFIA Neonatal IRT kit. Screening on the CF allows timely and appropriate treatment and rehabilitation measures that have a positive impact both on the condition of the patients and the average duration of their lives. Early detection of patients with cystic fibrosis, creates the possibility of adequate medical and genetic counseling based on DNA diagnosis and selection of rare forms forms of CF. Screening will determine the frequency of the CF in different regions of the country and/or ethnic groups, which is important for planning the amount of medical and preventive care for this category of patients.According to preliminary data falsely positive results (transient gipertripsinemia) may occur in infants with low APGAR scale, indicators, as well as healthy carriers of mutations (3 times more frequently than in a population) that require further study. The value of the chlorides of sweat has fluctuated from 50 mmol/l to 120 mmol/l among the patients with a positive test was 94.3% of the patients, in others-boundary numbers.

Conclusion. Main in diagnostics and research of CF is a sweat test that detects a larger (2-5 times compared to normal) concentration in sweat chlorides. Research of sweat in terms of elektroforez stimulation is considered the single most credible test in diagnosis of cystic fibrosis. The test gives positive results during first 3-5 weeks of life most affected children. Sweat chlorides increased in CF (in 98% of patients).