11. CLINICAL AND GENETIC ASPECTS OF MELAS SYNDROME (MITOCHONDRIAL ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES).

Author: Rotaru Andreea

Scientific adviser: Mariana Sprincean, PhD, Associate Professor, Department of Molecular Biology and Human Genetics, *Nicolae Testemitanu* State University of Medicine and Pharmacy of the Republic of Moldova.

Introduction. MELAS syndrome (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes) is a rare condition with early childhood outset that affects the nervous and muscular systems. The condition is caused by mutations in the mitochondrial DNA, mutations that may be inherited through the maternal lineage may be spontaneous. Clinical manifestations include seizures, recurrent headaches, stroke-like episodes with hemiplegia, hearing loss, myopathy, cardiomyopathy.

Case presentation. The patient, a 2-year-old and 7-month-old girl, presented to the hospital with the following accusations: superior paraplegia, balance disorders, aphasia. From the history of the disease: at one year and 8 months on the background of pneumonia, the first convulsive crisis appears. He begins to receive anticonvulsant medication. MRI shows ischemic stroke with postischemic encephalomalacia, left cerebrovascular malformation, clinically manifested by right hemiparesis and motor dysphasia. The echocardiographic examination shows restrictive heart disease, ASD, severe atriomegaly, RV diastolic dysfunction, fluid pericarditis and pulmonary hypertension, Heart failure NYHA gr II-III. After about a year, based on the results and the multisystemic effect, the diagnosis of mitochondrial encephalopathy, MELAS syndrome with multisystemic effect is confirmed based on the clinical manifestations as well as the genetic test. In the process of following the patient, other diagnoses were evaluated and excluded.

Discussion. MELAS is a progressive mitochondrial disease whose key element is stroke-like episodes, but it remains heterogeneous and symptomatically variable. Patients with unexplained stroke-like symptoms should be investigated, especially children, for whom early onset of symptoms could indicate a more aggressive phenotype. The presence of cortical and subcortical lesions on MRI and MRS increase the likelihood of mitochondrial pathology. Increased lactate in CSF during acute episodes and skeletal muscle biopsy are specific tests for this syndrome. Confirmation requires genetic testing for mutations in the MT-TL1 gene (up to 80% of cases).

Conclusions: The occurrence of stroke symptoms in pediatric patients requires a multidisciplinary approach and complex investigations. The polymorphism of the manifestations in MELAS syndrome can create difficulties in the diagnosis of the disease, but at the same time its importance lies in the genetic consultation that the family and the patient's treatment will need.