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PECULIARITIES OF MUCOVISCIDOSIS DIAGNOSIS VERIFICATION IN CHILDREN OF THE FIRST MONTHS OF LIFE

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Mucoviscidosis (MB) is a genetic disease with autosomal recessive mode of inheritance which is the lesion of all excretory glands of human body. MV is a rather widespread disease occurring in 1: 2000 cases among newborn babies. According to the estimation of the World Health Organization, 45-50 thousands children with MV are born in the world every year, and the number of heterozygote disease carriers is tens of millions.

A baby P. (boy), 7 months, was admitted to the Gastric Center of the Regional Children's Clinical Hospital with complaints at hidrosis, undigested fatty stool and insufficient weight gain.

It is known from the anamnesis that the child was born from the 1st pregnancy which was taking a course against the background of chronic toxoplasmosis (the mother was treated with macrolides), placental dysfunction, thrush, 1st stage anemia. During prenatal US screening on the 20th week of pregnancy, high intestine echogenicity was revealed in the fetus. Term birth, 37 weeks of gestational term, weight at birth: 2200 g. On the second day of life, the child underwent surgery – small intestine resection. The result of small intestine biopsy: data indicating at meconium ileus. Post-operative diagnosis: fetal peritonitis, small intestine atresia. Mucoviscidosis is supposed. NaCl content in secretion of perspiratory glands is 40.0 mmol/l (the child's birth is 2100 g). On the 8th day, skin jaundice with olive shade appeared, acholic stool, dark urine, hyperbilirubinemia due to direct fraction appeared. Liver biopsy was performed: manifestations of intracellular cholestasis, moderate portal fibrosis. The following diagnosis was established: congenital defect

of gastrointestinal system development - small intestine atresia, post-operative condition. Cholestatic hepatitis. Post-natal hypotrophy of 1st degree. Deficit anemia. ursodeoxycholic acid and iron supplements, enzymes were prescribed. To 2.5 months, jaundice regression was observed.

At birth, attention was paid in RCCH to static and kinetic development delay, 25% body weight deficit, phenotypic manifestations of rachitis, muscle hypotonia, abdomen size increase. Stool to 6 times a day, light yellow, undigested, fatty.

At examination: 1st type stearrhea, ALT, AST increase to 2 norms, hypocalcemia.

At abdominal US with Doppler sonography – increase of the liver right side linear size by 1.4 cm. Increase of liver parenchyma acoustic solidity, adequate visualization of intrahepatic bile ducts is impossible. Hyperplasia of lymph nodes in porta hepatis. Flaked suspension in the gall bladder. Movable liver fibroelastography: METAVIRF3 score – numerous septa without cirrhosis. At MRI with cholangiography: depletion of intrahepatic small ducts, the left and the right hepatic ducts are visualized well. Pilocarpine test (twice with the 7 day interval) was conducted: 1-a – 77.2 mEq/l, 2-a – 88.2 mEq/l. At molecular and genetic study, mutation of the gene CFTR-F 508 del – heterozygote was revealed. Based on the aforementioned, the following diagnosis was established: mucoviscidosis with pancreatic insufficiency. Meconium ileus, post-operative condition. Hepatic fibrosis, under METAVIR F3, with cholestasis syndrome. 2nd degree hypotrophy, pre- and postnatal, of mixed genesis. Subacute rachitis, eruptive phase.

Thus, the delayed final diagnosing is, most possibly, connected with erroneously negative pilocarpine test results. The children's weight (less than 3 kg) should have been taken into account, together with administration of antibiotics (macrolides) by the mother during pregnancy. It should be kept in mind that these factors influence on the reduction of sodium and chlorine ions in children's perspiratory glands.