HEREDITARY CHOLESTATIC DISEASES IN EARLY AGES: ALAGILLE SYNDROME, CAROLI DISEASE, PFIC 1 AND 2 AND OTHER IN COMPARISON WITH BILIARY ATHRESIA

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High cytolysis (>5 norm), jaundice with conjugated hyperbilirubinemia, hypo- and discolored stool are the sings of extrinsic and intrinsic biliar obstruction incombination with hepatocellular injury and sinusoid/ duct cholestasis. Yearly diagnosis of BA -biliary athresia (best before 2 months of age) and surgical correction (portoenteroanastomosis by Kasai) is determined to save a liver preventing of cholestatic hepatitis and cirrhosis. Differential diagnosis includes other cholestatic disease of non-infectious origin.

The similar clinical manifestation of various cholestatic diseases sometimes needs a complex of biochemistry, visual and functional investigations, isotope scanning, and gene sequencing. We want to present our experience in management of 12 ptnts with Alagille syndrome, 4 - with Caroli syndrome and disease, 8 children with PFIC 1and 2 (confirmed with gene sequence), non-syndromic duct hypoplasia (n=6), BA (n= 30).

Alagille syndrome diagnosis is based on the confirmation of three of the five major phenotypic traits, which confirm abnormal tissues formation during embryogenesis.

Liver transplantation is indicated not only for children with decompensated cirrhosis, but also with the severe itching, even if patient demonstrates normal protein- synthetic function of the liver and the absence of coagulopathy. Symptomatic treatment as AS and other forms of intrahepatic hypoplasia is based on UDCA (10-40 mg / kg / day in 1-2 doses), fat-soluble vitamins A, E, D and K, correction of the bone metabolism (calcitriol , calcium supplements and phosphorus). Diet was to offer special blends rich medium chain triglycerides.

We can conclude, that the isotope scanning is the most informative method for liver and biliary tract investigation and does not need a special preparation or anesthesia, and has a small radiation exposure. The most important are the half-life of isotope activity over the liver and the emergence of activity on the area of the intestine. Isotope activity in the liver persists more than 24 hours in case of BA and does not appear in the intestine. Constructing curves from different areas (liver segments, portal tract, choledochus zone, gall-bladder and duodenum) allow to imagine the pathophysiology of biliary excretion and anatomical characteristics of the child - the level of blockade, intra- /extra hepatic cholestasis.