A.S. Khandogina (III place)

I.M. Sechenov First Moscow State Medical University, Moscow, Russian Federation

COURSE FEATURES OF PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS OF I AND II TYPES (PFIC I AND PFIC II)

Relevance. Progressive familial intrahepatic cholestasis (PFIC) refers to a group of rare autosomal recessive disorders caused by genetically determined changes of hepatocytes canalicular membranes structure, which violates the excretion of bile. The prevalence varies from 1/50 to 1/100 000 000 infants in the world.

Object: to establish the debut and course features of PFIC I and PFIC II in children with a genetically confirmed disease.

Materials and methods. Retrospective case study of the first hospitalizations of children with PFIC I and PFIC II at the age between 2 and 3 years (age median 1 year 3 months ± 4 months) who were treated at the FSBI "SCCH". The clinical picture, trophological status of children and the most important laboratory parameters: cytolysis (AST, ALT), cholestasis (alkaline phosphatase, direct bilirubin), gamma glutamyl transferase (GGT), and total bilirubin, were evaluated.

Results. Growth (from -3δ to -1δ) and psychomotor development retardation was observed in all children. In 62% of these the disease had debuted in the first days of life in the form of cholestasis, jaundice, itching, hepato- and splenomegaly, in 38% - during the first months. In 7 (54%) children the level of cytolysis was 3-5 AST norms, corresponding to mild hepatitis, in 2 (15%) - >10 norms, corresponding to high activity hepatitis. GGT figures in all the patients did not exceed reference values: Me 24 (18; 25) U/L at a rate of up to 35 U/L in children older than 6 months. In 62% of children, alkaline phosphatase was > 350: Me 375 (216, 1642), U/L. In ll the children, general and direct bilirubin indexes exceeded the reference values: Me 155,5 (63,3; 443,3) and 107.6 (394, 219.9) mcmol/L, respectively.

Conclusions. PFIC is a serious illness, making its debut in the form of cholestasis during neonatal age and infancy. PFIC is characterized by liver lesions - intracellular cholestasis, typical normal levels of gamma-glutamyl transpeptidase, and progressive liver failure with the transformation into cirrhosis, making the patients candidates for a liver transplant.