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NIEMANN-PICK DISEASE

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Among the hereditary metabolic diseases lysosomal storage diseases take the specific position. They develop as a result of disturbances of the structure, function, transport of lysosomal enzymes etc. Today we know about 40 lysosomal storage diseases, which are characterized by an early manifestation, rapid development of irreversible damage to organs and organ systems, leading to disability and death. Among lysosomal diseases mucopolysaccharidoses are the most familiar to pediatricians. There are 4 disease types: classic infantile or neurological form (type A), visceral, form without neurological signs (type B), sub-acute or juvenile form (type C) and new Scottish variant (type D).

Etiology of NP disease.The disease is a genetic defect of chromosome 11 (types A and B), particular in the gene of sphingomyelin phosphodiesterase 1 (SMPD1). This gene encodes an acidic sphingomyelinase. It would be useful to mention that the NPD's disease represents an autosomal recessive inheritance pattern, and the specific weight of type A is about 70-80% of all cases. Therefore, in this work, we will focus specifically on this type of disease.

Type A is the most unfavorable form for the life prognosis. Manifestation of the disease appears mainly in the early age of 4 to 6 months. The first thing that attracts attention is baby lethargy, feeding difficulties, unreasonable temperature rise. The stomach increases gradually due to the spleno- and hepatomegaly, ascites and jaundice develop. Extremities look very thin in comparison to the enlarged abdomen. Prominent symptom is a regression of acquired skills, reduction of interest for the outer world, difficulties in holding objects, confined contact with parents. Children stop to hold their heads, sit and turn over themselves. In the terminal stages of the disease bulbar disturbances are observed, lack of hearing and vision, absence of tendon reflexes, spasticity and so on. On examination of the fundus, about half the cases, the specific symptom of "cherry stone" is determined – a dark red spot on the retina. There may be lenticular opacity of cornea and appearance of brown lens color. Light microscopy reveals a large number of cells with lipid inclusions, particularly a large number of them is in the liver and spleen. Obviously histobiochemical studies ascertain the accumulation of sphingomyelin and unesterified cholesterol. In the analysis of blood moderate anemia and thrombocytopenia are observed. We would like to dwell on the main symptoms which indicate exactly Niemann-Pick disease (as many metabolic diseases may have similar symptoms, such as "Hurler-like syndrome", which is also characteristic for the majority of mucopolysaccharidoses): manifestation of the main symptoms of the disease in the first year of life; progression of the clinical course; rough facial features; stiffness of joints; hepato- and splenomegaly; anemia and thrombocytopenia; cherry-red spot; reduction of sphingomyelinase activity ; frothy liver cells.

Thus, at present there is a possibility of biochemical and molecular genetic diagnosis of NPD disease type A in the postnatal and antenatal periods. Particular attention should be paid to the severity of the disease and its early fatal similarity to other diseases of accumulation, the difficulties of differential diagnosis. As already mentioned, unfortunately, there is no effective treatment. The therapy is aimed at symptom control and relief of the patient's condition. Gene therapy attempts are made only to animals and have positive results. The prognosis is unfavourable.

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