• amino-acids of blood - increased levels of threonine (0.225 mmol / l at a rate of 0,040-0,204) and methionine (0.045 mmol / l at a rate of 0,022-0,043);
• Lactate – 1.84 mmol / L (normal);
• Gas chromatography of urine – identified metabolites of exogenous origin;
• homocysteine of blood – 6.1 mmol / L (normal up to 5);
• Folic acid blood – more than 24 ng / mL (normal, more than 5.38);
• Vitamin B12 of blood – more than 2000 pg / mL (normal, 211-911);
• cortisol, testosterone, 17-OH-progesterone blood – the norm;
• Discovered MTRR A66G polymorphism in the homozygous state;
• Ultrasound of the internal organs - an excess of gall bladder symptoms dizgenezia of biliary, perivascular infiltration in the spleen, kidney - pathology detected, the adrenal glands are not visualized.

In connection with the identified polymorphisms MTRR A66G, the metabolism of cyanocobalamin, homocysteine, the amino acids to probands were given recommendations: a power to exclude meat broths, control blood levels of homocysteine, rehabilitation measures in Hospital № 1, observation of a pediatrician, neurologist, ophthalmologist, infectious disease; medical check-up in HSMGC.

Conclusions. The clinical features thanks to modern genetics can identify key errors of metabolism are associated with congenital CMV infection, which can be influenced.

THE RESULTS OF NTBC TREATMENT OF HEREDITARY TYROSINEMIA TYPE 1 PATIENTIES IN RUSSIA – THE IMPROVEMENT OF LIVER FIBROSES STAGE, RICKETS AND BONE DENSITY

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Background. Effectiveness of nitisinone treatment in hereditary Tyrosinemia type 1 patients (HT1) is well known. We started to treat patients on advanced stages of disease with cirrhotic liver and severe rickets with slender hope for success.

Material/patients The effectiveness of nitisinone was evaluated in 11 children from Russian population.

HT1 was confirmed in 12 children (f/m: 5/7) at the age 37[6; 244] months by elevated succynilacetone level in the urine, amino acid profile and detection of 2 mutations in FAH gene. Cirrhosis was confirmed in most (9 from 12) children, one 10 y/o girl with late diagnosis has been administered to liver transplantation without NTBC treatment. The initial dose of NTBC was 1,5-2 mg/kg in 6 subacute HT1 patients of the age less than 12 months, 0,6-1 mg/kg - with chronic HT1 patients older than 36 mns. The regress of the morphological signs of cirrhosis associated with the AFP normalization was confirmed by various methods of visualization (MRI, CT), radioisotope scanning and fibroelastography. Excellent results of treatment allowed us to avoid needle liver biopsy. The unexpected regression of liver cirrhosis was confirmed by different methods of visualization in 9 pts. Two incompliant patients demonstrated poor results – the same stage of fibrosis.

Two boys with HT1 of 5 and 13 years who started nitisinone treatment in 2009 and 2011 respectively, had a growth deficit of more than 3SD. Both had severe phosphate diabetes, complete Fanconi syndrome (hypophosphatemia, hypocalcaemia, glucosuria, great bicarbonate deficiency, metabolic acidosis). Prior to initiation of therapy both children were immobile. Besides NTBC patients received calcitriol, phosphates, calcium and other adjuvant therapy. Bone mineral density and bone age was determined by densitometry. First child demonstrated normalization of bone mass (from initially BMD =0,56) and bone density (with initial Z-score = -3) in 1.5 years after NTBC treatment and second child showed significant improvement of BMD and Z-score after 2 years of NTBC treatment. Both started to walk without assistance after orthopedic bone reconstructive surgery. Parathyroid hormone levels returned to normal. Patients stopped losing calcium and phosphorus with the urine. The patient's height increased in 4 and 2 years by 24 and 20 cm respectively. Two reconstructive operations on lower extremities have been performed in half year period: wedge resection of saber deformed bones of legs and hips with metal osteosynthesis.