трипсинемія) можуть зустрічатися у новонароджених з низькими показниками за шкалою APGAR, а також у здорових носіїв мутацій (в 3 рази частіше, ніж у популяції), які потребують подальшого вивчення. Необхідно враховувати, що помилково негативні результати скринінгу на МВ можуть зустрічатися при меконієвої непрохідності у новонароджених з МВ; тому всіх дітей з меконієвою непрохідністю необхідно направляти на дослідження хлоридів поту, незалежно від результатів скринінгу, а також при підозрі на ураження легень — ізольованій легеневій формі МВ.

MUCOPOLYSACCARIDOSES

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BACKGROUND: The mucopolysaccharidosis are a group of inherited metabolic diseases in which a defective or missing enzyme causes large amounts of complex sugar molecules to accumulate in harmful amounts in the body's cells and tissues. This accumulation causes permanent, progressive cellular damage that affects appearance, physical abilities, organ and system functioning, and, in most cases, mental development.

PURPOSE: The aim of this work is to provide an overview of mucopolysaccharidosis.

Material and methods:Enzymatic diagnosis of mucopolysaccaridosis.

RESULTS: Seven different types of mucopolysaccaridosis have been diagnosed in a large number of individuals.The prevalence ranged from 1 per 100,000 newborns for mucopolysaccharidoses type 1.Approximately 1 in 100,000 to 1 in 170,000 males for mucopolysaccharidosis type 2 (hunters syndrome). With Sanfilippo syndrome being the most common with an occurrence in 1 per 70,000 neonates.

CONCLUSIONS: Mucopolysaccharidosis is an autosomal recessive disorder , meaning that only individuals inheriting the defective gene from both parents are affected. However,they are relatively prevalent and represent a vital health setback in our society today.their early diagnosis and possible remedies are of utmost importance even though researches are still being conducted on some of them.

CONGENITAL CYTOMEGALOVIRUS INFECTION

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Actuality: Congenital cytomegalovirus (CMV) - a viral disease, which manifests itself polymorphic clinical symptoms with the defeat of the salivary glands, visceral organs, central nervous system and the formation of giant cells with typical intranuclear and cytoplasmic inclusions. Causes of genes mutations.

Objective: Early diagnosis of congenital CMV infection.

Materials and Methods: Patient N. born in 2012, was observated in HSMC in connection with the diagnosis of multiple stigma of disembriogenezis, congenital CMV infection, delayed physical and motor development.

The results of the study:

• study of polymorphic variants of the genes of folate cycle - MTRR A66G polymorphism is found in the homozygous state;

• blood chemistry - alkaline phosphatase 1378.2 U / L (normal up to 1107), total cholesterol 2.77 mmol / L (normal 2,90-5,18), glucose 4.96 mmol / L (normal), AST 35.51 U / L (normal), ALT 22.03 U / L (normal), triglycerides 1.72 mmol / L (normal 0,4-1,24), urea 3.51 mmol / L (normal) Uric acid is 2.32 mg% (normal), calcium 2.59 mmol / L (normal), phosphorus 2.16 mmol / L (normal), creatinine 39.69 m / l (normal), CPK 244.42 U / l (normal), LDH 369.02 U / L (normal), total bilirubin 2.39 mmol / L (normal), GGT 12.53 U / L (normal), total protein, 63.28 g / l (normal) , albumin, 45.02 g / l (normal);

• 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.