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);