Electroneuromyography was performed, myopathic syndrome was detected. The level of creatine phosphokinasein the blood - 340 IU / L. DNA diagnostics was recommended, deletion of the 7th and 8th exons of the SMN1 gene in the homozygous state was revealed. The child was examined by a neurologist (hypoxic-ischemic encephalopathy, acute period, CNS depression syndrome. Spinal muscular atrophy type I. Differential diagnosis included Pompe disease, galactosemia and intrauterine cytomegalovirus infection. The family received recommendations for further treatment and diagnosis of the child.In Italian clinics, the boy received treatment with Zolgensma, which is FDA-approved, based on a viral vector to replace the defective SMN1 gene with a working copy.

So, timely verification of the clinical diagnosis of Spinal muscular atrophy allowed clinicians to correctly interpret the neurological symptoms in a patient with myopathic syndrome and to conduct timely treatment.

Korotun O.P.

DIAGNOSTIC VALUE OF SOME CLINICAL INDICATORS IN IDENTIFYING THE RISK OF BRONCHIAL REMODELING IN CHILDREN WITH BRONCHIAL ASTHMA

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Respiratory remodeling according to the Inflammatory Theory of the disease is considered to be a multifactorial process that is realized with the participation of many cytokines, chemokines and growth factors. Remodeling is initiated by damage to the epithelium in the process of chronic bronchitis with the subsequent development of such characteristic manifestations as (1) changes in the epithelium and loss of their integrity, (2) thickening of the basement membrane, (3) subepithelial fibrosis, (4) enlargement of the goblet and submucosal glands. (5) hypertrophy and hyperplasia of smooth muscle cells, (6) increased vascularization, especially around the large bronchi.

The study aimed to assess the value of clinical indicators in identifying the risk of bronchial remodeling in children with bronchial asthma. To achieve the goal of the method by simple random sampling, a cohort of 53 children was formed, in which the severe persistent course of asthma was determined. Depending on the characteristics of the rate of acetylation processes, patients were divided into 2 clinical groups: the first (I) group included 25 patients, the second (II) group was formed by 28 school-age children.

The birth-weight of a child less than 2500 g showed high specificity of 97.4 (95% CI 92.0-99.6)% and a moderate predicted value of a positive result of 75.7% (95% CI 41.1-95.7), however, low sensitivity of 8.1% (95% CI 3.5-15.3) showed the probability of bronchial remodeling. The mentioned above birth-weight of a child indicates increased the post-test probability of possible bronchial remodeling by 24.3%, and higher birth weight was associated with a decrease in the probability of this event by 1.5%. The risk ratio with a positive test result was 3.3 (95% CI 0.79-13.7), BP = 1.56 (95% CI 0.4-6.1) with an absolute risk of 0.27.

Indications for passive smoking of children had a moderate specificity 65.8 (95% CI 55.6-75.0)%, but low sensitivity 54.0 (95% CI 43.7-64.1)%, PCPR 61.2 (95% CI 50.2-71.4)%. The posttest probability of this test was + 11.2% and -8.8%. The ratio of the risks of bronchial remodeling with a positive test value reached 2.26 (95% CI 1.3-4.0) with a relative risk value of 1.5 (95% CI 1.1-2.1) and absolute risk - 0.2.

In cases where the source of the harmful effects of tobacco smoke on the child was the mother, the test had a high specificity of 86.8 (95% CI 78.5-92.8)%, but a low sensitivity of 27.0 (95% CI 18.6-36.8)%. The accuracy of this test in detecting bronchial remodeling was 56.9 (95% CI 49.7-69.4)%, relative risk + - 2.05, and probability- - 0.8.probability of positive result increased by 17.2% and decreased by 4.3%. Indications for maternal smoking were associated with a probable risk of formation of structural changes in the bronchi: chances ratio = 2.4 (95% CI 1.17-5.04) with absolute risk = 0.2 and relative risk = 1.5 (95% CI 0.8) -2.7).

Thus, the results of a comprehensive examination of children used as diagnostic tests were mostly reliably specific, but low-sensitive with an unsatisfactory likelihood ratio. The data suggest that none of the proposed tests used to detect a high risk of alteration of bronchial structures had sufficient diagnostic value to detect a high risk of remodeling with a positive result, and, moreover, the exclusion of this risk with a negative test result. Therefore, for this purpose, they should probably be used either in combination (in parallel) or dynamics (sequentially).

Lastivka I.V.

CLINICAL CASE OF TUBEROUS SCLEROSIS

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Tuberous sclerosis is one of the phacomatosis genetically caused by a defect in embryonic development with the formation of tumor-like formations, with damage of all organs and systems, but primarily of the skin and nervous system. Frequency among infants - 1:6000-1:10000. Tuberous sclerosis is inherited by autosomal dominant type. 80%of cases are the result of a de novo mutation. The development of Tuberous sclerosis is determined by two genes: TSC1 (encodes the protein gamartin), and TSC2 (encodes the protein tuberin). Used diagnostic criteria were proposed by Roach E.S. in 1999. Treatment is symptomatic. Prevention and early prenatal diagnosis of the disease are important due to the high degree of disability.

Purpose and objectives of the study were to verify the diagnosis of tuberous sclerosis by molecular genetic diagnosis in a child with epilepsy. Material and methods: targeted high-throughput sequencing of clinically important genes, Sanger sequencing.

Clinical case: the family of a 4-year-old boy, who is under the supervision of neurologists for symptomatic epilepsy, consulted a geneticist. Parents were complaining about the presence of spots on the child's skin (from birth), seizures (from 4 months old), feeding problems. The child was born from the second pregnancy, ran on a background of anemia, the threat of miscarriage in the 1st trimester. Childbirth at 38 weeks of pregnancy finished by cesarean section. The baby from the first pregnancy is healthy, the mother is currently pregnant. Parents' anamnesis is burdened by oncopathology.

Examination of the child: on the skin of the buttocks, torso - multiple dense matte white macules up to 0.6 cm and depigmented spots. MRI of the brain: MRI picture of cortical and subcortical focal changes of the brain, characteristic of tuberous sclerosis. DNA diagnosis: mutation p.1869del (p.Asp624Thrfs * 74) of the TSC2 gene. This mutation was not detected in parents and native siblings. In addition, the patient and his mother were found to carry a pathological mutation p.220C> T (p.Arg74Cys) of the SGSH gene, which is responsible for the development of mucopolysaccharidosis type IIIA. The family was provided with recommendations for further monitoring and planning of subsequent pregnancies. Results: a mutation p.1869del (p.Asp624Thrfs * 74) of the TSC2 gene was detected in a proband usin ghigh-throughput sequencing.

So, the use of modern sequencing methods allowed to identify the pathogenic mutation of Tuberous sclerosis, confirm the clinical diagnosis and conduct medical and genetic counseling in the family.

Lozyuk I.Ya.

FREQUENCY OF HELICOBACTER PYLORI INFECTION IN CHILDREN WITH INFLAMMATORY DISEASES OF THE GASTROINTESTINAL TRACT

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Prior to the discovery of *H. pylori* and its relationship with the development of inflammatory diseases of the upper area of gastrointestinal tract (IDUGIT) the main etiological factor in the development of diseases was considered hyperproduction of hydrochloric acid, so all approaches to treatment were aimed at reducing acid-peptic factor using evolutionary anticholinergics, H2-histamine blockers and histamine blockers. proton pump. However, studies of their effectiveness in