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