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

the treatment of Helicobacter pylori-associated pathology have shown a direct correlation between the duration of remission and the course of the drug.

Given the leading place in the structure of digestive diseases in children IDUGIT, one of the determining predictors of the development of which is *H. pylori*, in order to determine its regional frequency retrospectively analyzed 368 "Medical cards of inpatients" children aged 7-18 years who were hospitalized in the gastroenterology department of the regional children's clinical hospital in Chernivtsi during 2010-2014.

The analysis showed that 216 children were infected with *H. pylori*, which is 58.7%. The dynamics of the frequency of *H. pylori* (+) IDUGIT for a five-year period showed fluctuations: a gradual decrease from 2010 to 2012 (from 59.1% to 56.7%) with a further increase in 2014 (60.6%). There was a difference in the frequency of *H. pylori* infection among children with IDUGIT depending on the place of residence. Despite the preservation of the general trend of the dynamics of the prevalence of *H. pylori* among children in Chernivtsi and districts of the region, significantly higher rates were found among people in the regional center (68.5% vs. 31.5%, $p < 0.05$).

The prevalence of Helicobacter pylori was analyzed for infection in children depending on age. It was found that among children aged 7-11 years the frequency of infection is slightly higher than among persons aged 12-18 years (68% and 57.2%, $p > 0.05$). However, the analysis of the dynamics of the indicator established an inverse correlation of weak strength between the age of the child and the frequency of H. pylori infection ($\varphi = 0.107$).

The structure of H. pylori infection in children with IDUGIT is represented by such nosological forms as chronic gastritis (CG), chronic gastroduodenitis (CGD), chronic duodenitis (CD), peptic ulcer (PUD) and duodenal ulcer. The highest frequency of Helicobacter pylori infection was found in people with HGD, in the second place - with VH, the third - with HG.

According to the results of retrospective analysis, it was found that the frequency of H. pylori infection among children of Chernivtsi region with IDUGIT is 58.7% with probably higher rates among patients with CGD and a tendency to decrease in frequency with age.

Marusyk U.I.

MARKERS OF ATOPIC REACTIVITY IN THE PUPILS, WITH SEVERE BRONCHIAL ASTHMA

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One of the most pressing problems of modern pediatric is allergic disease in children, including bronchial asthma (BA). In different regions of Ukraine this figure ranges from 5 to 9% of child population. Imposing the controlling asthma therapy in children should be considered a feature of the phenotype, defined heterogeneous mechanisms of development, however, is almost identical clinical symptoms. Based on the above, taking into account the performance of atopic reactivity in children that reflect the specific pathogenic mechanisms of realization of asthma, in a comprehensive study of patients will personalize the treatment of asthma and thereby improve treatment of the disease.

Considering this fact the objective of our scientific study was to increase the effectiveness of treatment phenotype of severe asthma in school-age children, taking into account the diagnostic value of indicators atopic reactivity. 60 school-age children with asthma in the remission period were comprehensively examined in the Pulmonology Department of Chernivtsi Regional Children's Clinical Hospital.

Over the course of the disease the patients were divided into two clinical groups. The first (I) clinical group consisted of 30 patients who had been registered severe asthma. The second (II) clinical group formed 30 patients, which was defined moderately severe asthma. For the main clinical features group were not differ. All children performed immunological blood test II - III levels. The contents of serum total immunoglobulin E (IgE), interleukin-4 (IL-4) and interleukin-5 (IL-5) was determined by enzyme-linked immunosorbent assay (ELISA).