МІНІСТЕРСТВО ОХОРОНИ ЗДОРОВ'Я УКРАЇНИ
БУКОВИНСЬКИЙ ДЕРЖАВНИЙ МЕДИЧНИЙ УНІВЕРСИТЕТ

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106-ї підсумкової науково-практичної конференції
з міжнародною участю
професорсько-викладацького колективу
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Матеріали підсумкової 106-ї науково-практичної конференції з міжнародною участю професорсько-викладацького колективу Буковинського державного медичного університету (м. Чернівці, 03, 05, 10 лютого 2025 р.) – Чернівці: Медуніверситет, 2025. – 450 с. іл.

У збірнику представлені матеріали 106-ї науково-практичної конференції з міжнародною участю професорсько-викладацького колективу Буковинського державного медичного університету (м. Чернівці, 03, 05, 10 лютого 2025 р.) зі стилістикою та орфографією у авторській редакції. Публікації присвячені актуальним проблемам фундаментальної, теоретичної та клінічної медицини.

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POTOCKI-LUPSKI SYNDROME IN A GIRL WITH FEEDING AND BEHAVIOR DISORDERS

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Introduction. Potocki-Lupski syndrome is a congenital chromosomal abnormality that includes muscle weakness, feeding difficulties, delayed physical development, delayed stato-kinetic and linguistic development, autism, and various types of heart abnormalities. The prevalence in the population is 1 per 20,000 people. To date, more than 50 cases of this syndrome have been described in the medical literature.

The aim of the study. The purpose of this study was to demonstrate a clinical case of a rare hereditary syndrome in children with protein-energy deficiency and delay in stato-kinetic development.

Material and methods. Medical and genetic counseling included portrait diagnosis using the Face2Gene Program, NGS study on the Metabolic panel, and genetic testing - karyotype-dependent multiplex ligation probe amplification (MLPA) technique.

Results. The family applied for medical and genetic counseling in order to investigate metabolic disorders in a 4-year-old girl. Complaints about dyspeptic phenomena (flatulence, periodic vomiting) and reduced vision.

The examination revealed dysmorphic facial features (micrognathia, dolichocephaly, bulbous tip of the nose, anti-Mongoloid cut of the eyes), neurological disorders, and delayed physical development.

Medical-genetic counseling included portrait diagnosis using the Face2Gene Program using artificial intelligence, as a result of which a low probability of Potocki-Lupski syndrome was revealed. The result of the NGS study on the Metabolic panel revealed a violation of the number of copies of *RAI1*, *ATPAF2*, and *FLCN* genes on chromosome 17. Because a pathogenic variant was identified, genetic testing was performed, including a karyotype-dependent multiplex ligation (MLPA) probe amplification technique for major microdeletion/microduplication sites (using the SALSA MLPA Probe mix P245 Microdeletion Microduplication Syndromes kit). MLPA test showed duplication of three regions (RAI1, DRC3-6, LLGL1-4RA) of chromosome 17p11.2 corresponding to Potocki-Lupski syndrome.

Conclusions. Children with Potocki-Lupski syndrome are at high risk for neurodevelopmental disorders or autism spectrum disorders, resulting in behavioral or social interaction problems and communication difficulties. In a child with a phenotype indicative of a genetic disease, the primary goal of the pediatrician should be early diagnosis to prevent possible complications. In these cases, a multidisciplinary team is needed. In addition, medical and genetic counseling is extremely important.

Lozyuk I.Ya.

EXTRAGASTRONIC-INTESTINAL (SKIN) MANIFESTATIONS OF FOOD ALLERGY IN CHILDREN WITH INFLAMMATORY DISEASES OF THE UPPER COMPARTMENTS OF THE GASTROINTESTINAL TRACT

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Introduction. Clinical manifestations of food allergy (FA) are diverse and may manifest as oto-pharyngeal, gastrointestinal, respiratory, neurological symptoms, etc., aggravating the course of various pathological processes.

The aim of the study. To assess the severity of skin manifestations depending on the cytotoxicity of the H.pylory strain.

Material and methods. Based on the gastroenterology department, 72 children, aged 7-18 years, with H.pylory-associated IDUGIT and FA were examined, and were divided into two clinical

groups: I group (44 people) - patients with H. pylori Cag A+ IDUGIT, II group (28 people) – patients with H. pylori Cag A- IDUGIT.

Results. Among children of the I and II groups, FA was manifested in the form of atopic dermatitis (AD). The SCORAD scale was used to assess the severity of skin manifestations of AD.

It was established that all children of Group I and 92.9% of patients of Group II had AD in an exacerbation stage, and only 7.1% of people of Group II were in the stage of incomplete remission. It was established that all children of group I and 92.9% of patients II group AD were in the exacerbation stage, and only 7.1% of II group individuals were in the stage of incomplete remission.

A localized form of AD was found in 30.6%, widespread in 47.2%, and diffuse in 22.2%.The diffuse form was more likely to be diagnosed in children with the cytotoxic CagA strain of H. pylori (29.6% vs. 10.7%, $p\varphi < 0.05$). Localized rashes were registered in the group of patients with H. pylori CagA+ 2.3 times less often than in the group of patients with H. pylori CagA-, 20.4% versus 46.4%, $p\varphi < 0.05$.

In all children, rashes were accompanied by itching, the intensity of which is higher in patients of the 1st group (7.3 ± 2.2 points versus 4.5 ± 1.9 points, $p < 0.05$).

In individuals of both I and II groups, the facial area was affected with a clear periorbital and perioral limitation and a rash on the flexural surfaces of the upper and lower limbs. 29.5% of the children of the I group and 28.6% of the children of the III group had signs of xerosis ($p\varphi > 0.05$); cheilitis was diagnosed in 13.6% of patients of the I group and 10.7% of patients of the II group.

Erythematous-squamous skin changes prevailed in both comparison groups (56.8% and 57.1%). In a comparative analysis, erythematous-squamous skin changes with lichenification were diagnosed in children with CagA+ probably more often than in patients with CagA- (27.3% vs. 10.7%, $p\varphi < 0.05$), on the other hand, exudative signs were less common in them (9.1% versus 28.6%, $p\varphi < 0.05$).

Calculation of the SCORAD index revealed fluctuations of the indicator from 7.4 to 88.9 um units. On average, in children with CagA+, it was 54.22 ± 18.11 human units, which is probably higher than in children with CagA(-) – 41.84 ± 16.29 human units, $p < 0.05$.

When comprehensively evaluating the severity of AD in children of the comparison groups as a whole, it should be noted the predominance of moderate AD in both groups. However, the comparative assessment showed a predominance of children of the II group, CagA-, with a mild course of AD (39.3% versus 15.9%, $p\varphi < 0.05$), while in individuals of the I group, CagA+, a severe course of AD (25, 0% versus 7.1%, $p\varphi < 0.05$).

Conclusions. Therefore, the cytotoxicity of the strain causes a more severe course of the disease with pronounced clinical and morphological lesions of the skin of a widespread or diffuse nature, accompanied by persistent intense itching.

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CHARACTERISTICS OF BRONCHIOLITIS AND OBSTRUCTIVE BRONCHITIS PROGRESSION IN CHILDREN UNDER ONE YEAR OF AGE

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Introduction. Respiratory diseases remain a pressing issue today, with a yearly increase in the number of young children diagnosed with bronchial obstruction. During wartime, children's bodies experience general maladaptation, disruption of chronobiological rhythms, autonomic disorders, and impaired mucosal immunity (limited access to fresh air, staying in basements during alerts, relocations, living in compact settlements, older children infecting younger ones in closed groups, large families, separation from home, loss of loved ones, anxiety, and other psychological disorders). These factors contribute to increased vulnerability to respiratory diseases, particularly among children from internally displaced families.