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БУКОВИНСЬКИЙ ДЕРЖАВНИЙ МЕДИЧНИЙ УНІВЕРСИТЕТ»

М А Т Е Р І А Л И

105-ї підсумкової науково-практичної конференції
з міжнародною участю
професорсько-викладацького персоналу
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Матеріали підсумкової 105-ї науково-практичної конференції з міжнародною участю професорсько-викладацького персоналу Буковинського державного медичного університету, присвяченої 80-річчю БДМУ (м. Чернівці, 05, 07, 12 лютого 2024 р.) – Чернівці: Медуніверситет, 2024. – 477 с. іл.

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У збірнику представлені матеріали 105-ї підсумкової науково-практичної конференції з міжнародною участю професорсько-викладацького персоналу Буковинського державного медичного університету, присвяченої 80-річчю БДМУ (м. Чернівці, 05, 07, 12 лютого 2024 р.) із стилістикою та орфографією у авторській редакції. Публікації присвячені актуальним проблемам фундаментальної, теоретичної та клінічної медицини.

Загальна редакція: професор Геруш І.В., професорка Грицюк М.І., професор Безрук В.В.

Наукові рецензенти:
професор Братенко М.К.
професор Булик Р.Є.
професор Гринчук Ф.В.
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професорка Годованець О.І.

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smoothed philtrum, curved raised lip, absence of lip frenulum and incisor papilla, sometimes cleft palate are described. In addition, hypotelorism, anosmia, short stature, delayed psycho-linguistic development. Treatment depends on accompanying abnormalities and must be multidisciplinary and individual in nature, according to the specific needs of the patient.

The aim of the study was to present a clinical case of early diagnosis of maxillary single central incisor syndrome.

Material and methods. Diagnosis was carried out on the basis of phenotypic data of an objective examination of the child, data on the history of life and illness, data on the child's development map, and the results of laboratory and instrumental research methods.

Results. We present the clinical observation of a child with MSCI. A mother with a 2-month-old girl came to see a doctor. Complaints about an increase in the size of the tongue, prolonged jaundice and drowsiness. A child was born from a second full-term pregnancy against the background of anemia, threat of miscarriage, toxicosis, polyhydramnios. Until the 18th week of pregnancy, the woman took Duphaston. It was a first childbirth at 37 weeks of gestation by Cesarean section. Body weight of baby at birth - 2500 g, length - 49 cm. Apgar score - 6/8 points. During the first day after birth, the child was on oxygen support. Heredity on the maternal side is burdened with cardiovascular pathology, on the paternal side – gastrointestinal pathology and diabetes. At the age of 2 months, the child was treated in the neurological department (diagnosis: Hypoxic-ischemic damage of the CNS, fragile child syndrome. Early anemia. Patent foramen oval of a large size with an aneurysm of the atrial septum. On examination, macrosomia of the second degree (25% excess weight), macroglossia, umbilical hernia, delayed stato-kinetic development were seen. Differential diagnosis with congenital hypothyroidism and Beckwith-Wiedemann syndrome was carried out. It was recommended a dynamic monitoring by a geneticist and endocrinologist. When the child was 2 years old, the mother came for a repeat examination. Complaints: delayed language and stato-kinetic development of the child. Examination of the oral cavity revealed one central incisor along the midline of the upper jaw, absence of a bial frenulum and incisor papilla, and an oval palate. Central incisor syndrome is suspected. The diagnosis was confirmed by the method of DNA diagnostics - a heterozygous deletion was detected on 11.18.32-11.23. Deletion in this region is associated with chromosome 18 deletion syndrome (OMIM: 146390) and depending on the size and breakpoints of the deletion on the short arm of chromosome 18, the phenotype can vary widely. The *TGIF1* gene, which is part of the deleted region, is associated with autosomal dominant holoprosencephaly 4 (OMIM: 142946), which may explain the patient's phenotype. Recommended: carrying out the array method to confirm the diagnosis; observation by an endocrinologist and CT scan of the brain.

Conclusion. The case has the clinical significance of early diagnosis of central single incisor syndrome and the need for multidisciplinary (dentist, pediatrician, speech therapist, psychologist, geneticist) observation in the treatment of such patients.

Kovtyuk N.I.

PECULIARITIES OF SALT TASTE SENSITIVITIES IN SHOOLAGE CHILDREN

Department of pediatrics, neonatology and perinatal medicines
Bucovinian State Medical University

Introduction. Taste is the type of sensitivities that help human organism to identify and consume nutrients while avoiding indigestible materials. Taste buds, the end-organs for gustation, detect and respond to a variety of macronutrient and aversive compounds to generate taste perception. It means possibilities to recognize and distinguish at least the “basic” tastes: sweet, umami, sour, salty and bitter. Each of these tastes is believed to represent different nutritional or physiological requirements or pose potential dietary hazards. The capacity to perceive taste sensations significantly influences food choice and food preferences which contribute in a cumulative manner to health status and quality of life. Salty taste governs intake of sodium and other salts, essential for maintaining the body’s water balance and blood circulation. Overconsumption of sodium, primarily in the form of salt (sodium chloride), is associated with an

increased incidence of hypertension, which rises the risk of cardiovascular disease and stroke. Epidemiological studies show that hypertension onset is strongly associated with salt consumption: there is a close relationship between average sodium salt intake and the incidence of hypertension, and restriction of sodium intake substantially decreases blood pressure. The link between dietary sodium and hypertension is well established and dietary modification is a primary step in hypertension risk reduction. Standard clinical advice for the prevention and treatment of hypertension includes limitation of salt intake. Yet, in spite of extensive public health education campaigns, sodium consumption exceeds recommendations. Because sodium is believed to be the ligand for salty taste, a more comprehensive understanding of the factors that drive salt consumption is needed to help develop effective and successful strategies to reduce sodium intake.

The aim of the study was examination of school-age children salty taste oral sensation in association with resting blood pressure.

Material and methods. In total 155 healthy children (ages 10-17 years, mean age - 13.8 years) were examined in schools during screening for elevated blood pressure. The study included assessment of resting blood pressure, salt taste sensitivities, structure and quality of nutrition, food preferences and children quality of life. To obtain the resting blood pressure three measurements were recorded at approximately 3 minute intervals, data from average were used.

Results. Hypertension was defined as a systolic blood pressure over 95 gender/age/height dependent percentile. Salt taste sensitivity to minimal concentrations (0.04%-0.08% of NaCl) was registered in 114 children (73.5%). The group of children with low sensitivity consisted of 41 persons. In some children was registered deviation in taste (disgeusia). Difference between males and females in salt sensitivities level was not established. The association between level of systolic blood pressure and the salt taste sensitivities was shown in regression model with inclusion of some anthropometric data and salt sensitivity.

Conclusions. The taste deviation poses a significant challenge for the health care of children population. Compared with the group of children with normal salt sensitivity persons with low salt sensitivity have higher risk of salt overconsumption and hypertension, they also reported greater liking of food with higher salt levels comparable to regular-sodium products.

Lastivka I.V.

MEDICAL-GENETIC COUNSELING IN NEPHROLOGY

Department of Pediatrics and Medical Genetics
Bukovinian State Medical University

Introduction. Monogenic diseases account for 70% and 10-15% of the prevalence of terminal stage renal failure in children and adults, respectively. Early identification of a monogenic cause of chronic kidney diseases may have important implications for patients and their families regarding treatment, prognosis, genetic counseling, and screening for risk groups among family members. Despite evidence of the diagnostic utility of massively parallel sequencing (MPS) methods, genetic testing is rarely used as a diagnostic tool in routine clinical practice, due to genetic illiteracy, lack of predictable benefit, difficulty in determining the best diagnostic test for a particular patient, difficulty in interpreting results, material costs and the need for post-test counseling.

The aim of the study. The purpose of this study is to acquaint nephrologists with modern methods of genetic research and encourage them to implement genetic testing in their daily practice.

Material and methods. Today, there are numerous methods of genetic studies of patients with kidney diseases: Sanger sequencing, comparative genomic hybridization (CGH), single nucleotide polymorphism (SNP) arrays, multiplex ligation-dependent probe amplification (MLPA), massively parallel sequencing, targeted sequencing, targeted gene panel sequencing, exome sequencing (ES), genome sequencing (GS). Today, most laboratories prefer to use panels of genes associated with the phenotype, which are based on exomes. In cases of unexplained renal failure, the preferred first-tier test is a targeted ES with a second-tier option of whole-exome analysis. CGH, SNP, GS or MLPA are used for copy number (CNV) diagnosis.