



in this index, which may be due to the short-term use of the “Mamalak” preparation and the anatomical and physiological characteristics of prematurely born infants. At the same time, we have traced a clear tendency to the decrease in the concentration of carbohydrates in feces almost three times against the background of the replacement enzyme therapy. Therefore, the percentage of infants whose concentration of carbohydrates in feces exceeded 0.6% has decreased from 84.5% to 30.7% (Δ 53.8%) in the I group versus the trend from 61.4% to 38.3 % (Δ 23%) in the comparison group. To sum it up, we can state that the obtained results prove the effectiveness of the early use of replacement enzyme therapy in the feeding of pre-term born infants with reduced tolerance to food in the hospital.

Lastivka I.V.

THE ROLE OF MEDICOGENETIC CONSULTATION AT PRADER–WILLI SYNDROME

*Department of Pediatrics and Medical Genetics
Higher State Educational Institution of Ukraine
«Bukovinian State Medical University»*

Prader-Willie's syndrome (PWS) is the most common cause of genetically predisposed high-grade obesity in children over one year old. The syndrome is also characterized by a delay in psycho-verbal and sexual development. Its population frequency is 1:10 000. Prader-Willie's syndrome occurs as a result of disorders in the PWS-AS area function: a) in 70 % of cases – as a result of deletion in the 15 parentage chromosome; b) in 20 % – in the absence of the 15 parentage chromosome in the child's cells or its replacement with a duplicated maternal chromosome (the phenomenon of one-childhood dysosomy); c) in 5 % – due to the deactivation of a structurally normal part of the PWS-AS of the parent chromosome in the fetus as a result of methylation. Similar changes in the allelic region of the 15 chromosome of maternal origin lead to Angelman syndrome, that's why Prader-Willie's syndrome is included in the group of diseases caused by genomic imprinting.

The purpose of the work was to assess the role of medical-genetic counseling in diagnosis of Prader-Willie's syndrome.

Patients with Prader-Willie's syndrome require multidisciplinary, differential, according to the age of the child, monitoring of the state of the main organs and systems, especially nervous and endocrine ones. The diagnosis is verified by a molecular-genetic study of the 15th chromosome pair. For the period of 2000-2016, 15 children (0-18 years old) with a suspicion of Prader-Willie's syndrome presented for medical-genetic counseling: 5 (33,33 %) of them were diagnosed syndromologically (including Lviv MGC), in 3 (20,0 %) of them the diagnosis was confirmed cytogenetically. 7 (46,7 %) families refused to follow up the verification of Prader-Willie's syndrome and as disabled children are observing by psychiatrists and/or neurologists.

Parents of children with Prader-Willie's syndrome are advised to undergo a genetic test before planning the next pregnancy, as there is a risk that the next child may be born with the same syndrome. The probability of a sick child is less than 1 % if it has a gene deletion or unipolar dysemia. If the child has a mutation in a region that is characterized by the phenomenon of imprinting, the probability increases to 50 %. The risk of recurrence, if one of the parents has a balanced translocation, depends on the nature of the translocation, but may reach 25 %; in contrast, all patients with unbalanced translocation, described to date, had a chromosomal reorganization.

Taking into account the foregoing, the medical-genetic counseling of such families is of particular importance, namely the prenatal testing of Prader-Willie's syndrome.

Levytska S.A.

A CASE-CONTROL STUDY OF THE IL-1 β GENE (C-511T) AND IL-4 GENE (C-590T) SINGLE NUCLEOTIDE POLYMORPHISM IN CHILDREN WITH CHRONIC SINUSITIS

*Department of Pediatric
Surgery and Otolaryngology
Higher State Educational Institution of Ukraine
«Bukovinian State Medical University»*

The mutations of the genes coding the components of the immune response may be the basis of the development of chronic inflammatory processes of the upper respiratory airways. The genetically determined balance between cytokines production influences the force and direction of the inflammatory response.

The aim of the study was to evaluate the association between the single nucleotide polymorphism (SGP) of the IL-1 β gene (C-511T) and the IL-4 gene (C-590T) with the development of chronic sinusitis (CS) in children.

We examined 100 children with CS and 35 children of the control group (CG). All children were genotyped for the IL-1 β (C-511T) SGP and the IL-4 (C-590T) SGP by polymerase chain reaction and restriction analysis.

There were no differences between CS and CG in the distribution of C-allele of the IL-1 β SGP. Significantly higher frequency of the T-allele of the IL-4 SGP was revealed in CS-children (43,5% vs. 24,3% in healthy controls, $p < 0,05$). The CC-genotype of the IL-1 β dominated in the CS-children (46% vs. 22,9% in CG, odds ratio – 2,9; CI 1,2-6,9, $p < 0,05$) as well as domination of the CT-genotype (65% vs. 42,9% in CG, odds ratio – 2,5; CI 1,1-5,4, $p < 0,05$) and TT-genotype (11% vs. 2,9% in CG, odds ratio – 4,2; CI 0,5-33,8, $p < 0,05$) of the IL-4 SGP was revealed in CS-patients.

The carriers of the T-allele of the IL-4 (C-590T) SGP and of the CC-genotype of the IL-1 β (C-511T) SGP had increased risk of the development of chronic sinusitis.