amylase and triglyceride therefore we can't use these parameters to prognosis the efficiency of surgical treatment, even though some studies have shown a prognostic value.

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БИОХИМИЧЕСКИЕ НАРУШЕНИЯ У ПАЦИЕНТОВ С САХАРНЫМ ДИАБЕТОМ С АНГИОПАТИЯМИ НА ЭТАПЕ ХИРУРГИЧЕСКОГО ЛЕЧЕНИЯ

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Проведен анализ биохимических отклонений у больных с диагнозом сахарный диабет с ангиопатией на этапах хирургического лечения для определения его практической значимости.

PRADER-WILLI SYNDROME: A CASE REPORT

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Introduction. Prader-Willi syndrome (ICD 10 Q87.1) is a genetic multisystem disease resulting from lack of expression of paternal imprinting genes of chromosome 15 (q11-q13). This syndrome was first described by Swiss pediatricians A. Prader and H. Willi in 1956. Prader-Willi syndrome occurs with a frequency about 1:15000 to 1:25000 newborns. Around 350000 - 400000 people worldwide suffer from the syndrome [1].

The probability of having a sick child is less than 1% if he has a gene deletion or unipaternal disomy, but if the child has a mutation in the region that is characterized by the phenomenon of imprinting, then this probability increases to 50%, in the case of chromosomal translocations, the risk of developing the disease in the next child is 25% [1]. Prenatal features include decreased fetal activity, fetal growth restriction, breech presentation, and preterm labor. Prenatal testing can be

used for diagnosis. Each age group has its own clinical manifestations. In the neonatal period, usually extreme muscle hypotonia and feeding problems are replaced by hyperphagia in early childhood, leading to obesity. During childhood, short stature is detected, and in adolescence - low levels of growth hormone, decreased growth rates, and hypogonadism can be seen. Growth hormone deficiency may be one of the components of the hypothalamic syndrome in Prader-Willi syndrome, and the pathophysiology of this process is poorly understood. In most cases, Prader-Willi syndrome is sporadic. In practical pediatrics the low incidence of the disease, the polymorphism of clinical manifestations and, often, the reluctance of relatives to admit the presence of deviations in the child's development lead to untimely diagnosis in approximately 75% of cases [2].

Aim of the study. To analyze a rare clinical case of Prader-Willi syndrome, characterized by polymorphism of clinical manifestations and diagnostic complexity.

Materials and methods. When describing this clinical case, a retrospective analysis of the child's medical documentation was carried out, as well as observation of her in the intensive care unit at neonatology department of the State Children's Clinical Hospital.

Research results. The patient is an 18-month-old girl. That was the first pregnancy of the mother which also was a premature birth with 28 weeks of gestation, given by emergency caesarean section. Pregnancy was occurred against the background of isthmic-cervical insufficiency, hydronephrosis on the left, pyeloectasia on the right. Body weight at birth was 1310 g, body length was 37 cm, head circumference was 26 cm, chest circumference was 24 cm. The condition of the child after birth is severe, which is due to respiratory failure and prematurity. Surfactant was administered in the delivery room; the child was on a ventilator. She was transferred to the next stage of nursing with a diagnosis of unspecified congenital infection. Congenital pneumonia. Bronchopulmonary dysplasia, "new form", severe course, respiratory failure of the 3rd degree. Cardiopathy of mixed origin. Minor cardiac anomaly: patent foramen ovale. Circulatory failure 1st degree. Necrotizing enterocolitis grade 1a. Encephalopathy of a newborn of mixed origin of moderate severity. Central nervous system depression syndrome. Intraventricular hemorrhage of the 2nd degree. Ventriculodilation on both sides. Choroid plexus cyst on the right. Submucosal cleft palate. Retinopathy of prematurity stage 2-3, zone 1-2, active phase of both eyes. Severe anemia (condition after correction). Congenital developmental features and prematurity 28 weeks. The child's condition showed positive dynamics. She was extubated at the age of 2 months and was subsequently nursed in the neonatology department. When examining the child, multiple congenital developmental features were noticed; low hair growth, overhanging nape, dysplastic low-set ears, close-set eyes, small mouth, gothic palate, short palpebral fissures. Weak pigmentation of the skin, hair and iris was also noted. In this regard, the child was consulted by a geneticist. A dry spot examination of a drop of blood was performed and a decrease in the concentration of free carnitine was detected.

At the age of 3 months, the child was discharged from the hospital for outpatient observation by a local pediatrician with a recommendation for repeated

consultation with a geneticist at 6 months. The child began to hold her head well from the age of 4 months, at 6 months she began to independently roll over from his back to her stomach and from her stomach to her back, and at 8 months she began to sit up independently. No psychomotor developmental delay was observed. The child's weight at 6 months was 5600 g (+ 4290 from the moment of birth), height was 59 cm (+ 35 cm from the moment of birth). There were also no delays in physical development. At the age of 6 months, based on clinical manifestations, a geneticist recommended a prometaphase cytogenetic study of the chromosomal region 15q11.2 to determine possible chromosomal abnormalities associated with Prader-Willi syndrome. Molecular genetic diagnostics were carried out using the method of fluorescence in situ hybridization (FISH) and samples for the critical region of Prader-Willi syndromes in the clinical diagnostic genetic laboratory at the Republican Scientific and Practical Center "Mother and Child". One of the advantages of this method is its ability to detect microdeletions that are not detected by classical karyotyping or polymerase chain reaction.

Cytogenetic examination data were obtained: the child's karyotype is 46XX, tandem mass spectrometry - no diagnostically significant disorders were identified, DNA diagnosis of spinal muscular atrophy - deletion of exon 7 of the SMN1 gene was not detected, DNA diagnosis of Prader-Willi syndrome - a deletion of the entire 15q11 region with methylation changes characteristic of Prader-Willi syndrome was detected (genes MKRN3, MAGEL2, NDN, SNRPN, UBEA3A, ATP10A, GARB3, OCA2). Final diagnosis: Prader-Willi syndrome.

Conclusions. Interest in this clinical case is due to the rarity of the disease and the polymorphism of clinical manifestations. Multiple developmental features, such as short hair growth, drooping nape, dysplastic low-set ears, close-set eyes, small mouth, a gothic palate, short palpebral fissures, muscle hypotonia, weak pigmentation of the skin, hair and iris served as grounds to suspect a hereditary disease and refer the child for molecular genetic diagnostics. As a result, a deletion of the entire 15q11 region with changes in methylation was identified, on the basis of which a diagnosis of Prader-Willi syndrome was made [**Ошибка! Источник ссылки не найден.**]. In turn, early diagnosis of the syndrome will make it possible to observe the child in order to identify possible disorders as early as possible and carry out their timely correction.

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СИНДРОМ ПРАДЕРА-ВИЛЛИ: КЛИНИЧЕСКИЙ СЛУЧАЙ

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Синдром Прадера-Вилли (МКБ 10 Q87.1) – генетическое мультисистемное заболевание, возникающее в результате недостаточной экспрессии отцовских импринтинговых генов хромосомы 15 (q11-q13). Интерес к данному клиническому случаю обусловлен редкостью заболевания и полиморфизмом клинических проявлений. Ранняя диагностика синдрома позволит наблюдать ребенка с целью максимально раннего выявления возможных нарушений и проводить их своевременную коррекцию.

PRIMARY PULMONARY HYPERTENSION: A CASE REPORT

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Introduction. Pulmonary hypertension is a rare medical condition characterized by elevated pulmonary artery pressure exceeding 25 mmHg at rest, which can significantly impact the morbidity and mortality rates in children. Recent studies have shown that prevalence is increased by 75% to 79% among young children [1]. In comparison to pulmonary hypertension in adults, pediatric pulmonary hypertension is often attributed to multifactorial and transient factors [2]. If left untreated, this condition can lead to irreversible damage of the pulmonary vasculature, as well as vascular remodeling and dysfunction of the pulmonary artery. Consequently, pulmonary resistance increases, resulting in elevated arterial pressure. The ensuing pressure gradient profoundly affects the right ventricle, leading to heart failure [2].

Research objective. To present a clinical case of primary pulmonary hypertension.

Research materials and methods. The materials of the case report of patient M., 10 years old boy were represented. He was admitted to the cardiological department with tachycardia attacks, palpitation and stabbing pain in the chest area, weakness, sweating and fatigue after physical activities. Past medical history: ARVI, chickenpox, pneumonia, adenoids, congenital diaphragmatic hernia. The patient has received all routine age-appropriate vaccinations.

Results of studies. The patient's chief complaints included intermittent headache, dizziness, and fatigue after physical activities and decreasing exercise tolerance during the last 5 years. His height is 163 cm and weight is 40 kg, body surface area – 1,38, body mass index – 15,1 kg/m^2. Temperature is 36.6 degree