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

Celsius. Physical examination: The skin is pale, pink. Subcutaneous fat is poorly expressed. The skin elasticity and turgor are normal. He has asthenic body type. Oropharinx is clear. There is no lymphadenopathy. The lungs are clear to auscultation. Pulse oximetry is 99% on room air. Breathing rate is 17 per a minute. S1 is decreased at the cardiac apex. Mild systolic murmur most prominent at the cardiac apex. S2 is split and accentuated on the pulmonary artery. Split S1 and systolic murmur at the 5^{th} point. Heart rate is regular, 78 per minute, blood pressure 110/60 mmHg. Pulsation on the femoral arteries is preserved. The abdomen is soft and without distension. The liver and spleen are impalpable. Stool and urination are normal. Laboratory and instrumental diagnostics were carried out.

Laboratory studies revealed a red blood cell count of $5.22x10^{12}$ per liter (normal range, 4.0 to 5.1), hemoglobin 150 grams per liter (normal range, 120 to 150), hematocrit 44.4% (normal range, 32,5-41,5). It is not surprising to observe a secondary erythrocytosis in patients with pronounced PH. A compensatory mechanism of increased red cell production is being launched by hypoxia in case of heart failure.

Instrumental research:

1 – Chest X-ray examination: pulmonary fields are transparent. Lung pattern is increased in the roots area and depleted in the other area. The heart borders are normal. The pulmonary trunk is expanded.

2 – ECG: sinus rhythm, heart rate 60 b/min. The right axis deviation. There are signs of right ventricular hypertrophy and diffuse changes in myocardium of the left ventricle.

3 – Echocardiography: Enlargement of heart right chambers, pulmonary trunk, pulmonary arteries. Hypertrophy of left ventricle. Tricuspid valve regurgitation 2-3 degree, pulmonary valve regurgitation the 1 degree. Significant pulmonary hypertension (85-90 mmhg). The contractile function of left ventricle myocardium is normal. The ejection fraction is 76%.

4 – Transthoracic echocardiography: No congenital heart disease data. The significant dilatation of the right heart chambers. Tricuspid valve regurgitation 2-3 degree. The systolic pressure gradient is 110 mm Hg. The dilatation of the trunk and pulmonary arteries, pulmonary valve regurgitation the 1 degree. The diastolic pressure gradient is 40mm Hg. It's a high pulmonary hypertension.

5 – Endovascular manipulation (oxygen test): Before endovascular oxygen test: Kw value of 35.79, Pp/Ps ratio of 106%, and a TPR/TSR ratio of 112%. The mean pulmonary arterial pressure (PAP) was 90 mmHg, and the pulmonary venous wedge pressure (PVWP) measured 18 mmhg. After using oxygen: Kw value of 29 Pp/Ps ratio of 116%, and a TPR/TSR ratio of 162%. The mean pulmonary arterial pressure (PAP) was 91 mmHg, and the PVWP measured 19 mmhg.

An oxygen test confirmed the severity of primary pulmonary hypertension.

Diagnosis: Primary pulmonary hypertension of a high degree. Tricuspid valve insufficiency 2-3 degree. Heart failure class 2. Displaced nasal septum. Allergic rhinitis.

Initially, the patient was prescribed sildenafil 20mg three times a day than the patient's medication regimen was shifted to bosentan, with a specific dosage 31.5mg twice a day for 4 weeks, followed by an adjustment to 62.5mg twice a day.
As a result of the treatment, the significant clinical improvement was achieved in the patient, including reduced severity of symptoms, enhanced tolerance to physical exercises, reduced functional class of heart failure and improved prognosis.

Conclusions. However, it is crucial to diagnose primary pulmonary hypertension on the first stages. Early diagnosis of this disease is vitally important for initiating modern timely therapy in order to improve the quality and duration of life in such patients.

Literature

1. Global Prevalence of Hypertension in Children / P. Peige Song [et al.] // JAMA Pediatrics. – 2019.

2. Imanina1, S. N. Characteristics of Pulmonary Arterial Hypertension in Children / S. N. Imanina1 // International journal of research publications. – 2022.

ПЕРВИЧНАЯ ЛЕГОЧНАЯ ГИПЕРТЕНЗИЯ: КЛИНИЧЕСКИЙ СЛУЧАЙ

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Легочная гипертензия- редкое заболевание, характеризующееся повышением среднего давления в легочной артерии больше 25 мм рт.ст. в состоянии покоя, которое может значительно повлиять на показатели заболеваемости и смертности в детском возрасте. Недавние исследования показали, что распространенность данного заболевания среди детей младшего возраста увеличилась на 75-79%. По сравнению со взрослыми, легочная гипертензия у детей является гетерогенным многофакторным состоянием.