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THE JOURNEY OF TUBEROUS SCLEROSIS IN CHILDREN AND THE EXPECTED OUTCOME

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Relevance. Tuberous Sclerosis, also known as tuberous sclerosis complex (TSC), is a rare genetic disorder that affects multiple organs in the body. It is caused by mutations in either the TSC1 or TSC2 genes, which regulate cell growth and division. TSC is characterized by the growth of benign tumors in various organs, including brain, heart, kidneys, and lungs. TSC can cause a range of symptoms, including seizures, developmental delays, and behavioral problems.

Aim: to present a clinical case of tuberous sclerosis in infant and describe diagnostic issues which have been faced.

Materials and methods. This study was conducted on a child diagnosed with TSC in Belarus and included a detailed medical history, physical examination, and laboratory investigations. The imaging studies included computed tomography (CT) scans, magnetic resonance imaging (MRI), neurosonography, and echocardiography. The data collected was analyzed using descriptive statistics.

Results and their discussion. The tumors associated with TSC can cause a variety of microscopic and macroscopic changes in affected organs. In the brain, the tumors can cause abnormal cell growth and disorganization, leading to seizures and other neurological symptoms. These tumors can be composed of a variety of cell types, including astrocytes, oligodendrocytes, and neurons. Our patient (1-year-old) had a small subendimal cyst on the right side of the brain. He was admitted to the ICU with suspected hyperglycemic coma (blood glucose level was 14 mmol/l). From anamnesis: acute respiratory tract infection, weakness, leaning forwards while crawling. Upon administration the child wasn't conscious, blephoraptosis was also present. Even though, 27 reflexes were symmetrical the child still had hypotonic muscle tone. MRI: In basal nuclei on both sides, in white matter of both hemispheres of the brain, asymmetrical multi-sized hyper intensity areas are determined - calcification with approximate sizes from 3 mm to 10 mm. There were no tumors in the heart, stable haemodynamics. On the cardio monitor sinus rhythm is periodic with tachycardia against the background of a decrease in sedation and anxiety, extrasystole units are recorded. Heart chambers are not dilated. the walls are not thickened. Systolic and diastolic functions of both ventricles are not impaired. Regurgitation on MV and PV is minimal, on TV 1 degree according to the Echo CG. Breathing is carried out on both sides, normal, no wheezing. The skin is pale, without cyanosis, there are three ash-colored spots on the skin of the trunk and limbs. The limbs are cold to the touch. Meningeal symptoms absent, no focal neurological symptoms. After receiving the results of blood (decompensated respiratory acidosis (pH-7.03. pCO2-94. p02-180. BE: -7, HCO3: 24)) the child was urgently intubated. He was treated symptomatically, get stable and transferred to neurological department.

Conclusion: the severity of TSC varies widely among affected individuals, and the age of onset of symptoms also varies widely. Our clinical observation demonstrates a patient with manifestation of the disease with vomiting, weakness, lethargy, impaired consciousness (stupor) with suspected coma of unknown origin**.** The disease was estimate by the neurological symptoms, brain MRI, skin spots. It is important for healthcare providers to closely monitor these changes and develop appropriate treatment plans to address them, which may include medication, surgery, or other interventions. Further research is needed to better understand the underlying mechanisms of TSC and to develop more effective treatments for this complex disorder. And as we saw the child did get a little bit better and long term treatment was the best option in this case.