Ya.A. Munkhalova, S.I. Tumanova, V.M. Argunova, L.I. Semenova, V.B. Egorova, T.E. Burtseva, S.N. Alekseeva, A-U.A. Novikova

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A RARE CLINICAL CASE OF TUBEROUS SCLEROSIS WITH CARDIAC RHABDOMYOMA IN A NEWBORN

This article presents a rare case of tuberous sclerosis with cardiac rhabdomyoma in a newborn. Rhabdomyoma is a benign tumor in the heart cavity, arising from embryonic muscle cells, and is frequently associated with tuberous sclerosis (TS), serving as its diagnostic marker. The disease was not detected prenatally. Postnatally, the infant was found to have a heart murmur; within days depigmented patches appeared all over the body, predominantly on the back, groin area, and legs. The diagnosis was established based on echocardiography. To determine further management tactics, a telemedicine consultation was conducted with the Federal Center for Cardiovascular Surgery in Khabarovsk, and the diagnosis was confirmed. Urgent planned surgical intervention was indicated.

Keywords: cardiac rhabdomyoma, tuberous sclerosis, cardiac tumors, newborn.

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MUNKHALOVA Yana Afanasyevna – PhD, Head of the Department of Pediatrics and Pediatric Surgery, Medical Institute, M.K. Ammosov, North-Eastern Federal University tokmacheva@mail.ru (corresponding author); ORCID: 0000-0002-9657-5612; **TUMANOVA Sakhayana Igorevna** – head of OPNND No. 2, Republican Hospital No. 1 – M.E. Nikolaev National Medical Center miss.tumashkina@mail.ru; **ARGUNOVA Vera Maichna** – Rheumatologist, Republican Hospital No. 1 – M.E. Nikolaev National Medical Center, Chief Freelance Rheumatologist of the Far Eastern Federal District, cardiorevmatologsakha@mail.ru; **SEMENOVA Lyudmila Ivanovna** – neonatologist, OPNND No. 2, Republican Hospital No. 1 – M.E. Nikolaev National Medical Center, semenovali@mail.ru; **EGOROVA Vera Borisovna** – PhD. Associate Professor, Department of Pediatrics and Pediatric Surgery, Medical Institute, M.K. Ammosov North-Eastern Federal University, veraborisovna@yandex.ru; ORCID: 0000-00003-3051-5251; **BURTSEVA Tatyana Egorovna** – MD, DSc – Professor, Department of Pediatrics and Pediatric Surgery, Medical Institute, M..K. Ammosov North-Eastern Federal University, leading researcher, Yakut Scientific Center for Complex Medical Problems, bourtsevat@yandex.ru; ORCID: 0000-0002-5490-2072; **ALEKSEEVA Sargylana Nikolaevna** – PhD, Deputy Director of the Perinatal Center, Republican Hospital No. 1 – M.E. Nikolaev National Medical Center, Associate Professor, Department of Pediatrics and Pediatric Surgery, Medical Institute, M.K. Ammosov North-Eastern Federal University, sargylanao@mail.ru; ORCID: 0000-0002-0550-9397; **NOVIKOVA Anna-Umsuura Alexandrovna** – 2nd year resident in Pediatrics (specialty 31.08.19), Department of Pediatrics and Pediatric Surgery, annaumsuura@mail.ru

Introduction. The most common benign cardiac tumor in children is rhabdomyoma. In 50% of cases, the tumor has an intracavitary localization, in 30% - it affects the myocardium, and it is extremely rare for it to be found on the heart valves [3]. The clinical presentation of rhabdomyoma is variable and depends on the localization, size of the tumor, involvement of the cardiac conduction system, and may manifest as an asymptomatic condition and a result of an incidental finding, or may lead to a fatal outcome in the perinatal period [4]. Rhabdomyoma is frequently (in 50-86% of cases) associated with tuberous sclerosis (TS), and is its diagnostic marker [4]. Tuberous sclerosis (Pringle–Bourneville disease, epiloyia) is an autosomal dominant disorder phenotypically manifesting as benign hamartomas in various organs and systems. Its prevalence is established as ranging from 1:6,000 to 1:10,000 in different populations [5, 6]. The cause of tuberous sclerosis (TS) is mutations in chromosomes 9 and 16 —specifically, in region 34 of the long arm of chromosome 9 (TS type 1—TSC1), and in region 13 of the short arm of chromosome 16 (TS type 2—TSC2) [1, 5, 7, 8].

This article presents a clinical example of a child with cardiac rhabdomyoma in the setting of tuberous sclerosis.

Objective: To describe a rare clinical case of a child with cardiac rhabdomyoma in tuberous sclerosis.

Materials and Methods: A retrospective analysis of the medical records of a patient hospitalized in the Department of Pathology of Newborns and Premature Infants No. 2 (DPNPI No. 2), Republican Hospital No. 1 – National Medical Center named after M.E. Nikolaev (RH No. 1 – NMC), was performed.

Clinical Case. A newborn girl was admitted to State Autonomous Institution RS(Y) "RH No. 1–NMC named after M.E. Nikolaev ", Department of Pathology of Newborns and Premature Infants No. 2 (DPNPI No. 2) on the 6th day of life. According to the mother's obstetric history, she had been under observation at an antenatal clinic since 7–8 weeks of pregnancy. The child was born from the fourth pregnancy. The first half of pregnancy was complicated by threatened miscarriage, marginal placenta previa, and retrochorial hematoma, for which she received inpatient treatment at her place of residence. In the second half of pregnancy, she developed grade 1 anemia, which was treated on an outpatient basis.

Prenatal screening results: First screening (12 weeks): marginal chorionic previa, retrochorial hematoma; Second screening (22 weeks and 4 days): no pathological findings; Third screening: not performed.

The third delivery occurred at 40 weeks of gestation, with vertex presentation. Birth weight was 3896 g, length 57 cm, head circumference 37 cm, chest circumference 37 cm. Apgar score was 8/9 points. The newborn's condition at birth was assessed as relatively satisfactory. A pronounced blowing murmur was detected at all auscultation points of the heart. No resuscitation was required.

At the central district hospital (CDH), a complete blood count and blood bio-

chemistry showed no abnormalities; blood gas analysis was not performed. Abdominal ultrasound and chest X-ray revealed no pathology. Echocardiography (ECHO-KG) was not performed due to the lack of equipment.

Upon admission to DPNPI No. 2: the skin was pink with peeling, and there were single round depigmented patches, up to 1.0 mm in diameter, on the posterior trunk and buttocks (Fig. 1).

The heart sounds were rhythmic but muffled, with a systolic murmur of moderate intensity at all auscultation points. In the subsequent days, the spots spread over the entire body, predominantly on the back, inguinal area, and legs; there were numerous solitary whitish lesions resembling areas of depigmentation, without hyperemia.

The results of the complete blood count, blood biochemistry, blood gas analysis, ELISA, PCR for intrauterine infections, infectious screening, coagulation profile, and urinalysis were unremarkable. Instrumental studies – Including neurosonography (NSG), abdominal ultrasound (US), hip joint ultrasound, EEG, abdominal CT, and brain MRI – revealed no pathology.

During the clinical-instrumental examination, the following findings were identified:

- Chest X-ray: Congenital heart defect. Pulmonary hypertension. Hypervolemia in the pulmonary circulation. Grade 3 thymomegaly.

- ECG: Intraventricular conduction disturbance. Increased potentials of the right ventricle (RV). Disturbed repolarization processes.

- Echocardiography (EchoCG): Additional masses detected in the left ventricular (LV) cavities; the right ventricle shows obstruction of the inflow tract of the left ventricle and of the inflow tract of the right ventricle (presumably rhabdomyomas). Patent foramen ovale (0.34 cm). Grade 1 regurgitation at the tricuspid valve. Right ventricular dilation. Ejection fraction 74.2% (Fig. 2).

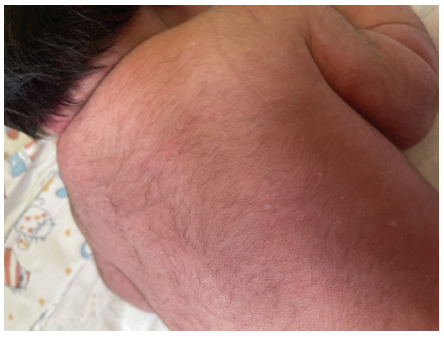

Fig. 1. Areas of skin depigmentation

- Holter ECG: Baseline ventricular repolarization disorders. ECG signs of right ventricular enlargement.

- Cardiac CT with contrast: Additional structures in the outflow tracts of the left and right ventricles. Small areas of ground-glass opacity in the dorsal segments of the lungs, most likely zones of hypoventilation. Grade 3 thymomegaly. Nodule in the right lobe of the thyroid gland.

- Lab studies: Elevated cardiac markers: CK-MB 29.05 U/L, high-sensitivity troponin 35.200 ng/L, and tumor marker AFP 24,590.60 IU/mL.

The child was examined by a cardiologist, dermatovenerologist, geneticist, ophthalmologist, and neurologist. The patient had two major criteria for tuberous sclerosis: skin depigmentation and cardiac rhabdomyoma.

To determine further treatment tactics, a consultation with a federal center was conducted. Following a telemedicine board decision with the Federal Center for Cardiovascular Surgery in Khabarovsk, the patient was indicated for surgical removal of the cardiac tumor-like masses.

On the 8th day of life, considering the increased risk of fatal outcome due to obstruction, the board decided to transfer the child for observation in the ICU. During observation in the ICU, the baby required no respiratory support and was subsequently transferred to the specialized department (DPNPI No. 2) for further preoperative preparation.

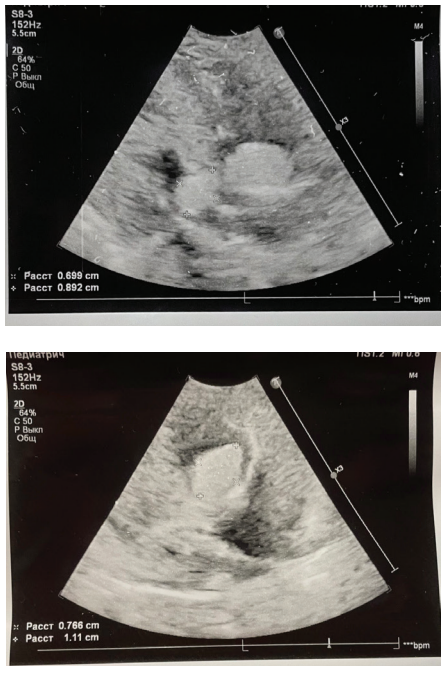

Fig. 2. Masses in the cavities of the left and right ventricles of the heart

During hospitalization, the child was exclusively breastfed and received conservative treatment.

On the 13th day of life, in stable condition, the child was transferred to the Federal Center for Cardiovascular Surgery in Khabarovsk for further surgical treatment with the following diagnosis: Multiple congenital malformations. Benign cardiac neoplasm. Rhabdomyomas (in the left ventricle cavity, 0.7 × 0.9 cm; right ventricle, 0.7 × 0.8 × 1.1 cm and 0.4 × 0.4 cm) with obstruction of the LV outflow tract and right ventricular inlet. Patent foramen ovale, 0.34 cm. Heart failure 0. Tuberous sclerosis. Thyroid gland nodule (right lobe). Unspecified anetoderma.

Conclusion: This case demonstrates the potential for early diagnosis and timely surgical correction of cardiac rhabdomyoma in early childhood. Early diagnosis and surgical intervention improve prognosis. Cardiac rhabdomyoma in newborns requires a multidisciplinary approach (involving cardiologists, cardiac surgeons, and geneticists). The management strategy depends on the tumor size, the presence of symptoms, and its association with tuberous sclerosis.

In recent years, only isolated cases of prenatal diagnosis of cardiac rhabdomyoma have been reported in the literature. For instance, during fetal ultrasound at 21 weeks of gestation, volumetric masses were detected in the region of the right and left ventricles. Histopathological examination revealed the presence of "spider cells." Molecular genetic testing identified a pathogenic variant in the TSC2 gene [2].

Thus, early diagnosis of cardiac rhabdomyoma is possible in the perinatal period using the tools of prenatal ultrasound and molecular genetic studies.

The authors declare no conflicts of interest.

References

1. Gamirova R.G., Knyazeva O.V., Gadieva A.R., et al. Bolezn' Burnevillya — Pringla: klinicheskij sluchaj iz praktiki detskogo nevrologa [Bourneville — Pringle disease: a clinical case report from pediatric neurologist practice]. Prakticheskaya medicina [Practical Medicine, 2017. Vol. 1. No. 1 P.157-161 (In Russ.).]

2. Dorofeeva M.Yu., Belousova E.D., Pivovarova A.M. Rekomendacii po diagnostike i lecheniyu tuberoznogo skleroza [Recommendations for the diagnosis and treatment of tuberous sclerosisyu]. Zhurnal nevrologii i psihiatrii im. S.S. Korsakova [S.S. Korsakov Journal of Neurology and Psychiatry, 2014. P.58-74 (In Russ.).]

3. Pozgaleva N.V., Borisova A.A., Bykova E.V., et al. Mnozhestvennye rabdomiomy serdca u novorozhdennogo (klinicheskij sluchaj) [Multiple rhabdomyomas of the heart in a newborn (a clinical case)]. Byulleten' medicinskih Internet-kon-

ferencij [Bulletin of medical Internet conferences, 2016. Vol.6. No. 6. P. 1226-1228 (In Russ.).]

4. Naumenko E.I., Anufrieva V.G., Grishutkina I.A. Opuhol' serdca u novorozhdennogo kak marker tuberoznogo skleroza: klinicheskij sluchaj [Cardiac Tumor in Newborn as the Marker of Tuberous Sclerosis: Clinical Case]. Pediatricheskaya farmakologiya [Pediatric Pharmacology]. 2020; 17 (2): 148-151 (In Russ.).] doi.org/10.15690/pf.v17i2.2101.

5. Novikova I.V., Venchikova N.A., Gusina A.A. Sluchaj rabdomiomy serdca u ploda, obuslovlennyj mutaciej v gene TSC2 [Fetal cardiac rhabdomyomas associated with mutation in the TSC2 gene: presentation of a case]. Covremennye perinatal'nye medicinskie tekhnologii v reshenii problem demograficheskoj bezopasnosti [Modern Perinatal Medical Technologies in Solving Demographic Security Problems. 2022, No. 15. P. 564-568 (In Russ.).]

6. Shilyaev R.R., Kharitonova E.V., Kopilova E.B. Tuberoznyj skleroz. Osobennosti klinicheskogo techeniya u detej rannego vozrasta [Tuberous sclerosis: peculiarities of clinical course in children of early age]. Vestnik Ivanovskoj medicinskoj akademii [Bulletin of the Ivanovo Medical Academy. 2010. Vol. 15, No. 1, P. 51-57 (In Russ.).]

7. Dorofeeva M.Y., Belousova E.D., Pivovarova A.M.. Federal'nye klinicheskie rekomendacii (protokoly) po diagnostike i lecheniyu tuberoznogo skleroza u detej [Federal clinical guidelines (protocols) for the diagnosis and treatment of tuberous sclerosis in children]. St. Petersburg, 04.06.2013.Updated international tuberous sclerosis complex diagnostic criteria and surveillance and management recommendations. SPb, 04.06.2013 (data obrashcheniya: 10.10.2025) (In Russ.).]

8. Northrup H., Aronow ME, Bebin EM, et al. Pediatr Neurol 123:50-66, 2021. doi: 10.1016/j.pediatrneurol.2021.07.011.

M.S. Savvina, V.M. Argunova, M.F. Afonskaya,
M.P. Slobodchikova, Y.A. Munkhalova

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VENOUS THROMBOSIS OF THE LOWER EXTREMITY IN A 13-YEAR-OLD PATIENT

A clinical case of the venous thrombosis of the lower extremity in a 13-year-old patient is represented in the article, the thrombosis occurred in the course of post-COVID syndrome (enzyme-linked immunosorbent assay for SARS-CoV2 by 18.09.2024 detected 50000 BAU/mL. Onset of the disease acute fever up to 38C, abdominal pain. Diagnostic laparoscopy performed in the central district hospital. Diagnosis: right ovarian apoplexy, cystic ovaries, adhesions, anemiya (hemoglobin - 75 g/l, dizziness, weakness). Prior to that, she was examined in Yakutsk, autoimmune thyroiditis was diagnosed. On the 6th day after laparoscopy, fever up to 39,2C, pain in the popliteal pits and lower legs appeared. Transferred to the childrens department of the central district hospital, from there sent to the admission and diagnostic department of the pediatric center RH1. Electroneuromyography revealed signs of progressive muscle damage. Transferred to the cardiorheumatology department for further examination and treatment. Laboratory data showed an increase in all inflammatory markers (CRP, D-dimer, ferritin). Ultrasound imaging revealed occlusive thrombosis of the sural veins of both lower extremities. Received low molecular weight heparin, antucosgulants, aspirin, diclofenac as treatment. The adequate therapy resulted in positive dynamics.

Keywords: thrombosis, thromboembolism, coronavirus, children, vessels, veins

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Introduction. Thrombosis is the formation of the blood clots in the blood vessels both in the arteries and the veins. Venous thromboembolism is most common and results in fatalities in the adult population. However, it is uncommon and rare among children and adolescents. Its estimated prevalence varies from 0.5 to 1.9 per 10,000 children population. Those children diagnosed with blood clotting disorders are at the highest risk [2,8].

Deep vein thrombosis can be complicated with thromboembolism and post-thrombotic syndrome. Most of the thromboses are associated with central veinous catheterization. The other factors are traumas, surgical interventions, post-traumatic disorders, hereditary and acquired thrombophilic conditions, such as the presence of the factor V Leiden mutation, protein C and protein S deficiencies, disorders of the intestines, kidney, circulatory system, infections, rheumatic diseases, antiphospholipid syndrome and many others. The other causes of thrombosis are the inherited abnormalities (inferior vena cava agenesis, atresia, occlusion or uncomplete substitution) [3,6].

As a rule, the onset of deep vein thrombosis in children is asymptomatic, though there can be swollen upper and lower extremities, elevation of body temperature, weakness, erythema, positive Homan's sign (pain response in the dorsiflexion of the foot). The diagnosis of venous thrombosis is confirmed by Duplex ultrasonography with color Doppler imaging; contrast venography, computed tomography, angiography and MRI are less common [2,4,8,9].

Despite all the mentioned causes post-COVID and multisystemic inflammatory system should be added, as they manifest by vasculopathy and hemostasis. The pathogenesis of SARS-coV-2 affects the vascular endothelium, disturbs hemostasis that result in risk of clot formation [10]. The patients after COVID-19 show high indices of D-dimmer and fibrinogen which increase clot formation [3,9].

The clinical case. The female patient K., aged 13, was admitted to the department of cardio-rheumatology of the Pediatric Center of the Republican hospital #1, M.E. Nikolaev National health center. She complained of weakness, pains in the lower extremities when walking, nausea and dizziness.

SAVVINA Maya Semyonovna – Ph.D., senior staff scientist of the laboratory of pediatric health level monitoring, Yakutsk Scientific Center of Complex Medical Problems, maya_savvina@mail.ru; **ARGUNOVA Vera Maichna** – rheumatologist of the Department of Cardiorheumotology, M.E. Nikolaev Pediatric center, Republican hospital No.1, senior non-staff rheumatologist of the Far-Eastern federal district. cardiorevmatologsakha@mail.ru; **AFONSKAYA Marina Viktorovna** – a rheumatologist of the department of cardiorheumotology M.E. Nikolaev Pediatric center, Republican hospital No.1, senior non-staff rheumatologist of the Republic of Sakha (Yakutia); **SLOBODCHIKOVA Maya Pavlovna** – a senior lecturer of the department of foreign languages with the course of Latin language, St. Petersburg state pediatric medical university; **MUNKHALOVA Yana Afanasyevna** – Ph.D., head of the Department of Pediatrics and Pediatric Surgery, Medical Institute M.K. Ammosov Northeastern Federal University