



Cardiac Rhabdomyoma in 9 Months Infant: From Echocardiography Perspective

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ABSTRACT

Background: The most prevalent benign cardiac tumor in neonates, cardiac rhabdomyoma, is frequently associated with tuberous sclerosis complex (TSC). Based on their dimensions and positioning, these tumors may result in obstructive cardiac lesions, arrhythmias, or hemodynamic instability. Echocardiography is the primary diagnostic instrument for detecting and monitoring cardiac rhabdomyomas, as well as for differentiating them from other intracardiac malignancies.

Case Report : This case report describes a 9-month-old newborn diagnosed with cardiac rhabdomyoma, focusing on the echocardiographic findings, clinical presentation, and therapy strategy. Echocardiographic imaging identified many hyperechoic masses within the ventricles; further echocardiographic evaluations demonstrated spontaneous regression, thereby corroborating the diagnosis. **Conclusion :** The absence of genetic evidence between MRI and TSC hinders a definitive connection. This case underscores the importance of non-invasive imaging in guiding the diagnosis and treatment of pediatric cardiac cancers.

Keywords: echocardiography, pediatric cardiac tumor, cardiac rhabdomyoma, tuberous sclerosis complex, ventricular mass

INTRODUCTION

Cardiac rhabdomyomas constitute around 60–80% of all primary pediatric cardiac tumors in neonates and children, representing the most frequently occurring benign cardiac tumors. Typically seen in the heart's ventricles, these tumors consist of striated muscle cells^[1]. Despite being histologically benign, the clinical significance of rhabdomyomas is contingent upon their location, size, and number. Occasionally, arrhythmias, valve dysfunction, or obstruction of blood flow result in hemodynamic instability^[2]. Cardiac rhabdomyoma exhibits a strong correlation with tuberous sclerosis complex (TSC), a hereditary multisystem disorder characterized by benign tumors in various organs, including the brain, kidneys, skin, and heart^[3]. As much as eighty percent of neonates diagnosed with cardiac rhabdomyoma also meet the criteria for tuberous sclerosis complex (TSC)^[3]. Consequently, it is essential to assess affected babies for additional TSC symptoms. However, diagnosing TSC in newborns and infants can be challenging, as the characteristic cutaneous and neurological symptoms require time to manifest. Echocardiography is the primary diagnostic modality for identifying cardiac rhabdomyomas due to its comprehensive non-invasive imaging of intracardiac structures. Typically presenting as homogeneous, well-defined, hyperechoic masses embedded within the ventricular myocardium^[4], these cancers Although not consistently accessible, magnetic resonance imaging (MRI) and genetic testing for TSC1 and TSC2 mutations may assist in further evaluation. This case report discusses the echocardiographic findings, clinical presentation, and management of a 9-month-old infant diagnosed with cardiac rhabdomyoma. We emphasize the necessity of a multidisciplinary approach in managing young patients with suspected cardiac malignancies and the role of regular echocardiographic monitoring.

CASE REPORT

A 4-month-old female infant was sent to a pediatric cardiology clinic for A routine assessment for congenital heart disease (CHD). Maternal hypertension resulted in her early birth at 30 weeks of gestation via cesarean surgery. At birth, she weighed 1.3 kilograms, measured 34 cm in length, and had Apgar scores of 8 and 9 at one and five minutes, respectively. The newborn was immediately crying at birth and was sent to the neonatal intensive care unit (NICU) for monitoring. She was stable during the hospitalization and had no significant issues, like feeding difficulties or respiratory distress syndrome. She was discharged in a stable condition after several weeks of supportive care and monitoring. At four months of age, during a regular follow-up, the infant appeared vibrant and well-developed, without signs of cyanosis, respiratory distress, or feeding difficulties. Her parents expressed no concerns regarding her development. Her vital signs were stable: heart rate at 136 bpm, respiratory rate at 28 breaths per minute, temperature at 36.5°C, and oxygen saturation at 98% on room air. Auscultation indicated the absence of cardiac murmurs. Her weight of 1.54 kg and length of 45 cm indicated appropriate weight gain in development parameters. A comprehensive physical examination revealed no abnormalities indicative of syndromic associations such as tuberous sclerosis complex (TSC). Her history of prematurity necessitated a transthoracic echocardiography (TTE) as part of standard cardiac evaluation. Two distinct, homogeneous masses within the myocardium were initially identified in an echocardiography. One tumor, measuring 23.7 mm x 17.7 mm, was located in the right ventricle (RV); another tumor, measuring 14.3 mm x 9.48 mm, was identified in the left ventricle (LV). The ventricular function appeared preserved, with no indications of obstruction in the left ventricular outflow tract (LVOT) or right ventricular outflow tract (RVOT). The patient was scheduled for a standard follow-up echocardiography to evaluate any progression or regression; the masses were presumed to be cardiac rhabdomyomas according to the echocardiographic findings. The newborn remained asymptomatic, exhibiting consistent development and no signs of cardiac dysfunction at a follow-up examination at five months of

age. The RV mass measured 22.8 mm × 16.5 mm, and the LV mass measured 13.2 mm × 7.1 mm; a subsequent echocardiographic assessment indicated a little reduction in the size of the masses. No hemodynamic impairment or obstruction was seen once again. Ongoing cautious management with periodic echocardiographic assessment was warranted due to the stable nature of the masses and the absence of clinical symptoms. The patient's clinical condition remained stable by the time she reached nine months of age. She exhibited standard growth milestones, adequate weight gain, and no cardiac-related issues, including signs of heart failure or exertional dyspnea. The right ventricular mass measured 1.4 cm x 0.8 cm, whereas the left ventricular mass measured 2.5 cm x 1.8 cm; echocardiographic evaluation revealed additional tumor reduction. Additionally, the absence of hemodynamic impairment and the observed pattern of spontaneous regression corroborated the diagnosis of cardiac rhabdomyoma. The patient had to undergo ongoing monitoring to identify any late complications and ensure sustained tumor decrease.

DISCUSSION

Cardiac rhabdomyomas are typically identified during prenatal or early neonatal stages with routine echocardiography. Their projected prevalence in the general population varies from 0.02% to 0.08%. Although benign, their potential implications include embolization, arrhythmias, and outflow obstruction. Cardiac rhabdomyoma is evidently associated with tuberous sclerosis complex (TSC), as it occurs in up to 80% of instances involving individuals with TSC^[5]. However, in this instance, the absence of genetic testing or other clinical data consistent with TSC renders a definitive diagnosis of TSC difficult to ascertain. We are unable to ascertain whether this case aligns with TSC or represents an isolated rhabdomyoma due to the constraints of our diagnostic methods. The diagnosis of cardiac rhabdomyoma was established based on echocardiographic findings specific to this neoplasm. Characteristic features of rhabdomyoma include a homogeneous, hyperechoic, and well-defined mass^[4]. Furthermore, the lesions were numerous and located in the ventricular myocardium, a distribution typically associated with

rhabdomyomas^[6]. The echocardiographic characteristics of rhabdomyoma are distinct from those of other pediatric cardiac neoplasms. Fibromas are typically solitary and larger than rhabdomyomas. Their internal calcifications^[4], which were not present in our case, are occasionally observable. Typically pedunculated, irregularly shaped, and predominantly located in the atria rather than the ventricles, myxomas represent an alternative diagnosis^[4]. Typically associated with pericardial effusions, absent in this patient, teratomas—originating from pluripotent cells—generally have a combination of solid and cystic components^[4]. Although uncommon, hemangiomas are characterized by increased vascularity on Doppler imaging, a feature not seen in this case. In addition to the echocardiographic characteristics, our diagnosis is corroborated by the normal progression of cardiac rhabdomyoma. The propensity of rhabdomyomas for spontaneous regression is one of their most notable characteristics^[7]. In contrast to fibromas and myxomas, which generally necessitate surgical excision due to continuous growth or obstructive consequences, rhabdomyomas gradually regress over time without intervention^[7]. The phenomenon has been attributed to postnatal hormonal and genetic regulation. Serial echocardiographic evaluations of our patient demonstrated a sustained decrease in tumor growth over several months, indicating a higher likelihood of rhabdomyoma as opposed to an alternative cardiac tumor. Furthermore, the absence of arrhythmias or hemodynamic compromise in this patient aligns with the typically benign nature of rhabdomyomas. The patient remained asymptomatic during follow-up, despite the possibility of conduction problems or ventricular outflow tract block in certain circumstances. The findings indicate that the most appropriate course of action remains ongoing conservative therapy with echocardiographic monitoring.

CONCLUSION

Cardiac rhabdomyoma is a rare but significant heart tumor found in infants. The evaluation of tumor burden and hemodynamic consequences relies on early detection facilitated by echocardiography. Given that the majority of cases resolve spontaneously, non-invasive surveillance is of heightened significance. This case

underscores the necessity of echocardiography in directing diagnosis and treatment to ensure optimal outcomes in young patients with cardiac tumors.

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