

Cardiac Rhabdomyoma

Olivia R. Rubin; Richard B. Towbin, MD; Jeffrey A. Towbin, MD; Jason N. Johnson, MD, MHS; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

Pediatric cardiac rhabdomyoma (CR) is a benign, congenital cardiac tumor that typically forms in utero. The most common presenting symptoms include arrhythmias and blood flow restriction, which can result in a reduction in peripheral pulses, cyanosis, shortness of breath, heart murmurs, and heart failure. CR is associated with tuberous sclerosis in over 90% of patients and presents with multiple benign tumors. It is typically diagnosed with echocardiography and MRI. Most CRs resolve on their own. In more severe cases, cardiac surgery may be necessary.

Keywords: Cardiac, Oncology, Benign

Case Summary

An infant with a history of seizures also has hypomelanotic macules of the skin, a loud, harsh systolic ejection murmur at the left upper sternal border, and subependymal nodules of the brain (Figure 1).

Imaging Findings

The transthoracic echocardiogram (Figure 2) at 1 month of age showed multiple homogeneous hyperechoic masses with a large mass in the right ventricular outflow tract causing moderate obstruction. A repeat echocardiogram (Figure 3) at 6 years of age showed near-complete resolution of the multiple homogeneous hyperechoic masses.

Diagnosis

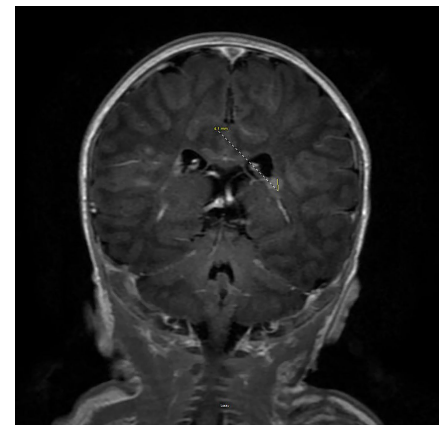
Cardiac rhabdomyoma (CR).

The differential diagnoses include atrial myxoma (AM), cardiac fibroma, and hemangioma.

Discussion

CR is a rare, benign cardiac tumor of striated muscle that occurs in 0.2% of children. However, they are the most common cardiac tumors, making up 45% of pediatric cardiac tumors. CRs are nonspecific to race or ethnicity and occur equally in males and females. A child born with mutations on the TSC1 or TSC2 genes will have tuberous sclerosis (TS) complex, which may also result in rhabdomyomas.¹ CR is typically located in the heart's ventricles and can interfere with the functions of the heart, such as blood flow restrictions and arrhythmias. Over 90% of CRs are associated with TS and almost always present with multiple cardiac tumors. In almost all cases of non-syndromic rhabdomyoma, the tumors are solitary, and their cause

Figure 1. Coronal T1 FLAIR of the brain. There is a subependymal nodule along the lateral margin of the body of the lateral ventricle.



is unknown.² 94% of CRs are multiple and related to TS. 6% of the CR tumors are solitary and typically unrelated to TS.³ Although these children are usually asymptomatic, symptomatic presentation may include arrhythmias, heart murmurs, and, in rare cases, heart failure, which leads to hydrops.¹

CR can be diagnosed in utero using three-dimensional echocardiography and MRI. MRI commonly employs the use of balanced steady-state free precession and single-shot fast spin echo black blood sequences, which differentiate blood (hyperintense) and myocardium (hypointense). MRI is the preferred imaging modality for the evaluation of

Affiliations: Scottsdale Preparatory Academy, Scottsdale, Arizona (Rubin); Department of Radiology, Phoenix Children's Hospital, Phoenix, Arizona (RB Towbin, Schaefer); Cardiology, Le Bonheur Children's Hospital, Memphis, Tennessee (JA Towbin, Johnson); Department of Radiology, Cincinnati Children's Hospital, Cincinnati, Ohio (A Towbin).

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Figure 2. (A) Two-dimensional transthoracic echocardiogram parasternal short axis. There is a large homogeneous hyperechoic mass (*) of the right ventricular outflow tract consistent with a rhabdomyoma. AV, aortic valve; LA, left atrium; MPA, main pulmonary artery; RA, right atrium. (B) Two-dimensional transthoracic echocardiogram four-chamber view at end-diastole. There are multiple homogeneous hyperechoic masses (*) of the right and left ventricular free wall and ventricular septum consistent with rhabdomyomas. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (C) Two-dimensional and color Doppler compare the transthoracic echocardiogram four-chamber view angled anterior at peak systole. There are multiple homogeneous hyperechoic masses (*) of the right ventricular outflow tract, left ventricular free wall, and ventricular septum consistent with rhabdomyomas. There is moderate obstruction of the right ventricular outflow tract (yellow*). MPA, main pulmonary artery; RV, right ventricle.

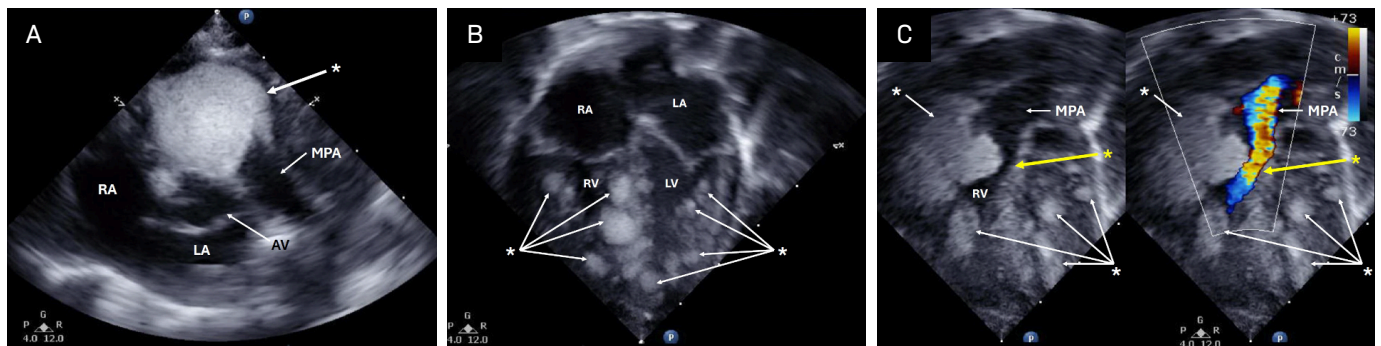
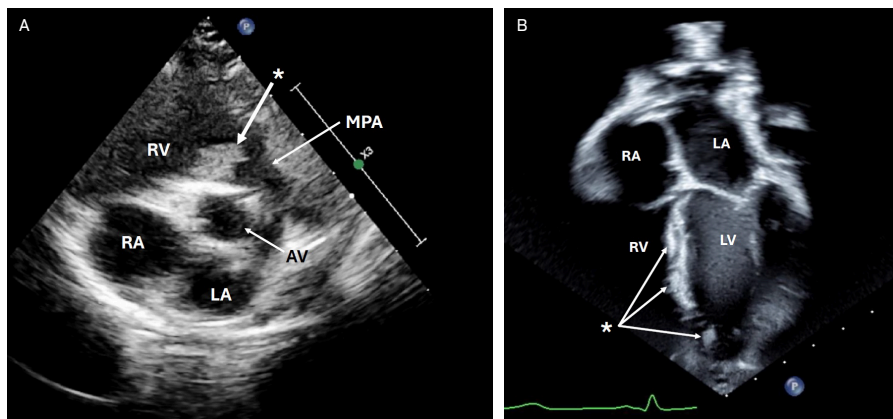


Figure 3. (A) Two-dimensional transthoracic echocardiogram parasternal short axis. There is a small homogeneous hyperechoic mass (*) of the right ventricular outflow tract consistent with a rhabdomyoma. AV, aortic valve; LA, left atrium; MPA, main pulmonary artery; RA, right atrium. (B) Two-dimensional transthoracic echocardiogram four-chamber view at end-diastole. There are three homogeneous hyperechoic masses (*) of the ventricular septum consistent with rhabdomyomas. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.



CR. On MRI, CR typically presents as solid, homogeneous masses with smooth, well-defined borders.⁴ CRs typically are enlarged until approximately 32 weeks of gestational age, after which time they can gradually decrease in size.³ Maternal ultrasound findings of CR include nodular, well-defined, oval-shaped, and hyperechoic lesions. Additional findings include myocardial edema and pericardial effusion.¹ Doppler evaluation of blood flow. A biopsy is needed for a histologic diagnosis of CR. The typical histology features the presence of

“spider cells” that are large, polygonal cells with glycogen. The peripherally displaced nucleus, cytoplasmic vacuoles, and radiating myofibrils.

The differential diagnosis of a cardiac mass includes a cardiac fibroma, the second most common cardiac tumor of childhood, which occurs in 0.03-0.32% of cases.⁵ These benign tumors usually arise from the cardiac septum and may grow into the myocardium.⁶ Cardiac fibroma does not usually resolve spontaneously after birth, as CRs do. While both cardiac fibroma and CR may present

with arrhythmias, cardiac fibroma can have more serious effects, such as heart failure, cyanosis, syncope, chest pain, and sudden death.²

AM is a benign cardiac tumor that primarily appears later in life in females between the ages of 40 and 60 years.⁷ AM can be differentiated from CR based on the number and location of the tumor. AMs arise in the atria and are singular, while CRs are in the ventricles and are usually multiple. In children with a solitary CR, the ventricular location helps distinguish between the two lesions. MRI is the primary means of distinguishing between the two. In contrast to rhabdomyoma, AM does not penetrate the subjacent myocardium.⁵

Cardiac hemangiomas (CHs) have different imaging features on echocardiography than CRs. CHs can occur anywhere in the heart or pericardium and are hyperechoic and show marked enhancement on CT. On MRI, they are heterogeneous with intermediate to high signal on T1-weighted images and diffuse high signal on T2-weighted sequences. After contrast injection, the lesion enhances intensely for a prolonged time. Histologically, a CR is located in the myocardium of the ventricles, is round and uniform in nature, and has small, well-defined myocardial nodules that can protrude into

the ventricle. In comparison, the histology of CH presents as engorged vascularity surrounded by endothelial cells.⁸

While the majority of CRs are asymptomatic and resolve on their own, rare instances of heart failure and hemodynamic compromise can occur.¹ Treatment of a patient with a symptomatic cardiac condition can include inhibitors, digitalis, and diuretics. In more severe cases with larger tumors, surgical removal may be necessary.

Conclusion

Pediatric CR is a benign, congenital cardiac tumor that typically forms in utero. The most common presenting symptoms include arrhythmias and blood flow restriction, which can result in a reduction in peripheral pulses, cyanosis,

shortness of breath, heart murmurs, and heart failure.¹ CR is associated with TS in over 90% of patients and presents with multiple benign tumors.⁹ It is typically diagnosed with echocardiography and MRI. Most CRs resolve on their own. In more severe cases, cardiac surgery may be necessary.

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