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A Rare Septal Hamartoma of Mature Cardiac Myocytes Manifesting With Cardiac Cachexia in a Teenager

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Hamartomas of mature cardiac myocytes are a rare type of primary cardiac tumor, with only 5 cases of interventricular septal involvement reported to date. They are often challenging to diagnose, and there is currently no standardized approach to surgical resection. Here, we present a rare case of a large 6.0 × 4.4 × 3.8-cm interventricular septal hamartoma that manifested with cardiac cachexia in a teenage patient. Surgical biopsy was used after 4 attempts at endomyocardial biopsy were nondiagnostic, and partial debulking was performed as complete resection was not feasible.

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Primary cardiac tumors, especially those arising from the interventricular septum, are uncommon. Hamartomas of mature cardiac myocytes, an exceptionally rare type of primary cardiac tumor, have been reported to involve the interventricular septum in only 5 cases, of which only 2 were in adults.[1-5] Hamartomas are local malformations involving an abnormal mixture of cells and tissues, typically consisting of cells native to the organ. On histologic evaluation, they can be characterized by myocyte hypertrophy with fiber disorganization. They are also distinguished from other cardiac hamartomas, such as rhabdomyoma, histiocytoid cardiomyopathy, cardiac fibroma, and mesenchymal hamartoma, by myocyte disarray and interstitial fibrosis. Hamartomas can appear to be similar to hypertrophic myopathy, but a key difference is that they contain disorganized mature myocytes characterized by interstitial fibrosis. As cardiac hamartomas may mimic cardiac hypertrophy, the diagnosis may be challenging, and there is currently no standardized approach to guide surgical treatment. Here, we present a rare case of a large interventricular septal hamartoma that manifested with cardiac cachexia in a teenage patient.

A previously healthy 19-year-old woman presented with 30-pound weight loss during 6 months despite adequate nutrition (body mass index, 16.4 kg/m^2). She also reported progressive dyspnea on exertion and weakness. On physical examination, she had a 2/6 systolic ejection murmur and exhibited mild edema to her ankles bilaterally. Laboratory evaluation was unremarkable.

Echocardiography revealed a large interventricular septal mass with concern for left ventricular outflow tract obstruction. There was no evidence of any valvular abnormalities. The mass was further delineated on computed tomography angiography, measuring 6.0 × 4.4 × 3.8 cm (Figure 1). Positron emission tomography scan revealed mild hypermetabolic activity throughout the mass. The patient underwent 4 separate attempts at endomyocardial biopsy for the septal mass, with the first 2 attempts approaching via the right atrium and right ventricle, the next attempt via the aortic valve and left ventricle, and a final attempt via a transseptal approach and through the mitral valve. The biopsy specimens were nondiagnostic, showing myocytes with normal to mild hypertrophy and some irregularity in orientation but otherwise without definite abnormalities. The patient was therefore referred for surgical biopsy and potential intervention. She provided consent for the procedures and for this case report.

In the operating room, transesophageal echocardiography was performed, revealing moderate eccentric aortic insufficiency and severe eccentric mitral regurgitation, neither of which had previously been observed. The decision was made to proceed with surgical biopsy and to address the patient's valvular findings after a definitive diagnosis for the septal mass was established. A median sternotomy was performed and the location of the mass was confirmed with epicardial

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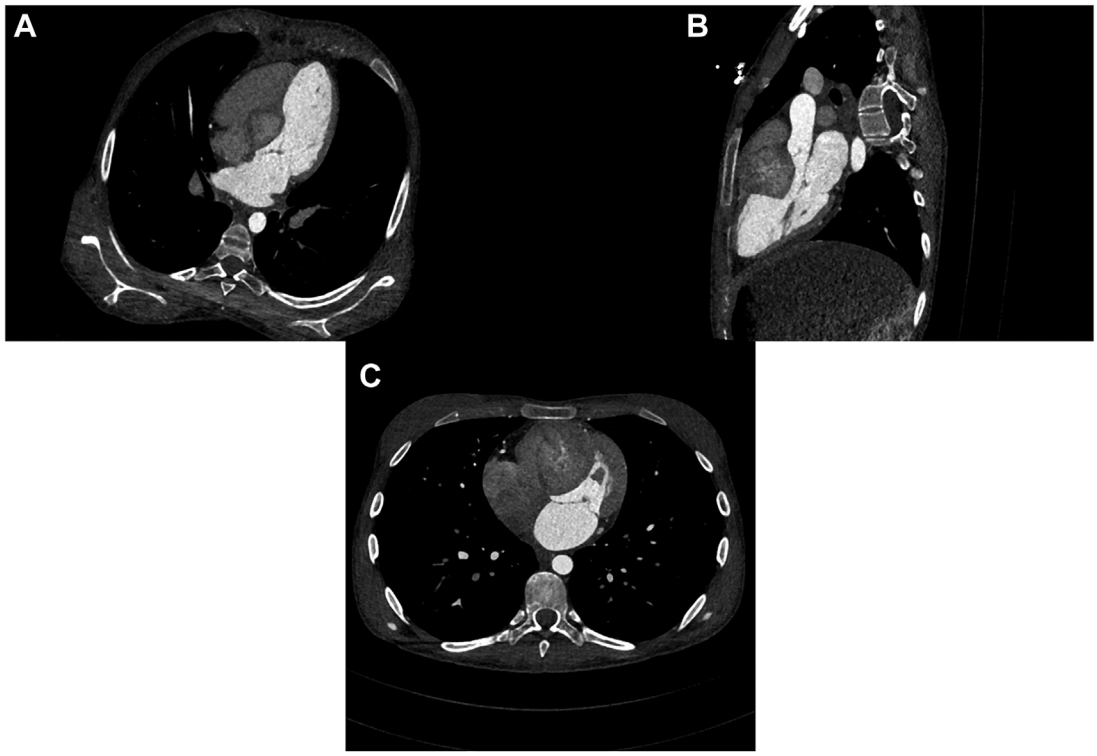

FIGURE 1 Computed tomography images of the septal hamartoma: (A) 4-chamber view; (B) left ventricular long-axis view; and (C) short-axis view.

ultrasound. A polypropylene purse-string suture was placed over the anterior interventricular septum. Multiple passes were made with a Tru-Cut needle to biopsy the mass. The purse-string suture was then tied, ensuring hemostasis, and the chest was closed. The final pathologic diagnosis was hamartoma of mature cardiac myocytes (Figure 2). After a heart team discussion involving the patient and her family, the decision was made to proceed with mass debulking and concomitant aortic and mitral valve surgery.

Two weeks after the surgical biopsy, the patient underwent redo sternotomy. Cardiopulmonary bypass was initiated with central aortic and bicaval venous cannulation. With the heart arrested, a transverse aortotomy was made and the aortic valve was inspected, revealing an irreparable large perforation at the nadir of the right coronary cusp. The aortic valve leaflets were excised, and the septal mass was debulked through the aortic valve until the left ventricular outflow tract was sufficiently wide open. A longitudinal main pulmonary artery arteriotomy was then made, and further debulking of the septal mass was performed through the pulmonary valve until the right ventricular outflow tract was sufficiently wide open. In total, 8 g of tissue was removed. Next, the mitral valve was exposed through Sondergaard's groove. A tear in the A3 leaflet was observed and closed with running polypropylene suture. A 28-mm Physio ring (Edwards Lifesciences) was then implanted onto the mitral annulus. Finally, the aortic valve was replaced with a 23-mm Inspiris valve (Edwards Lifesciences). The heart initially regained a sinus rhythm but subsequently developed complete heart block intraoperatively. The patient had a dual-chamber permanent pacemaker implanted on postoperative day 7. At 4 months after the operation, the patient is gaining weight and has returned to an active lifestyle without dyspnea on exertion.

COMMENT

More than 100 cases of cardiac hamartomas have been reported in the literature, but only 5 have involved the interventricular septum. As space-occupying lesions, hamartomas are often mistaken for cardiac hypertrophy. The best way to distinguish hamartomas from hypertrophic cardiomyopathy is with imaging such as echocardiography, cardiac computed tomography, and magnetic resonance imaging. These allow the identification of the exact location and size of cardiac

hamartomas, although imaging appearance may not be pathognomonic.[3,6]

Most cardiac hamartomas affect the ventricular free wall, which often allows complete resection. In this case, however, the patient's interventricular septal mass was in a central location in the heart and unresectable. Instead, her treatment options included conservative medical management (not feasible because of the patient's progressive symptoms), listing for heart transplantation (not ideal because of the need for lifelong immunosuppressants), and surgical debulking. We opted for debulking in this case, recognizing that hamartomas are benign tumors that generally have a slow growth rate, portending a favorable long-term prognosis.[3,4]

This case is also unusual because of damage to the mitral and aortic valves, most likely iatrogenic in origin. Whereas endomyocardial biopsy may be the least invasive approach to diagnosing primary cardiac tumors including hamartomas,[7] it is likely that the observed valvular damage occurred secondary to multiple attempted endomyocardial biopsies that were nondiagnostic. Open surgical biopsy may be considered if early attempts at endomyocardial biopsy are unsuccessful, especially because hamartomas can appear similar to normal myocytes histologically and may therefore require a higher threshold for the amount of tissue needed for diagnosis.

Complete heart block developed postoperatively and the patient required a pacemaker. A nationwide study reported that the incidence of complete heart block requiring pacemaker implantation in patients who underwent septal myectomy is 9%.[8] As such, surgeons will need to be mindful of the cardiac conduction system when considering debulking surgery for interventricular septal masses.

Overall, we describe the rare case of a young woman presenting with a large interventricular septal hamartoma that was treated by surgical debulking with significant symptom improvement. Whereas complete resection may not be feasible for large septal masses, partial debulking can be considered in selected cases for benign tumors including cardiac hamartomas.

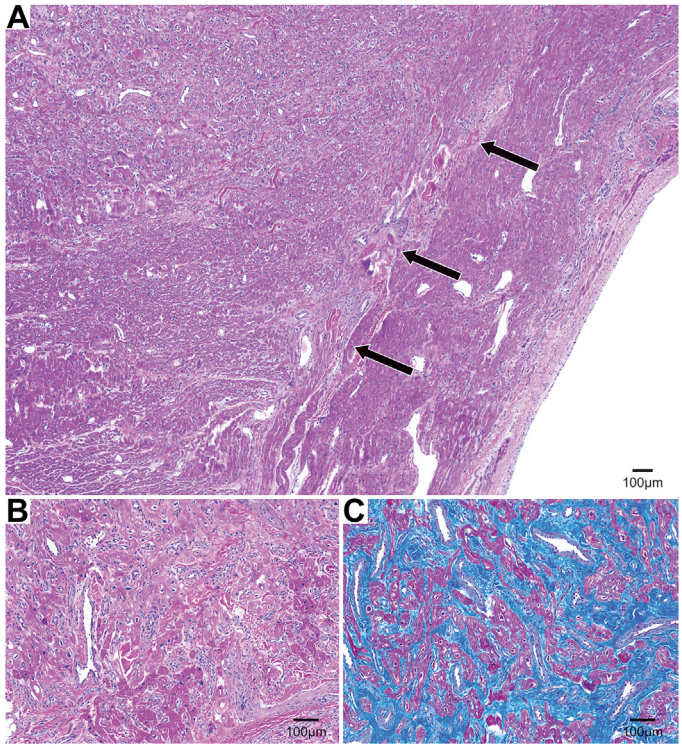

FIGURE 2 Histopathologic features of hamartoma of mature cardiac myocytes. (A) Low-power magnification showing sharp demarcation of the lesion (arrows) from the adjacent subendocardial myocardium and endocardial layer (hematoxylin and eosin, magnification ×40). (B) High-power magnification of hamartomatous lesion composed of enlarged, irregularly distributed myocytes. Myocyte disorganization is accentuated by the widened, fibrotic interstitium containing dilated venules (hematoxylin and eosin, magnification ×100). (C) Masson trichrome stain highlights the interstitial expansion by mature collagen, occasional myocyte vacuolization, and interstitial vasculature (magnification ×100).

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The authors have no conflicts of interest to disclose.

PATIENT CONSENT

Obtained.

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