Cardiac Hamartoma: A Diagnostic Challenge

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INTRODUCTION

Cardiac masses have a broad differential diagnosis, including thrombus, cardiac metastasis, primary cardiac neoplasm, nonneoplastic lesion, and infectious process. Cardiac hamartomas are rare and can be detected in pediatric patients as well as adults.

CASE PRESENTATION

A 21-year-old woman was admitted for recent episodes of nonspecific chest pain and shortness of breath. Her medical history was significant for migraine headaches. Physical examination and laboratory workup were unremarkable. Electrocardiography demonstrated normal sinus rhythm with left atrial enlargement and nonspecific ST-T changes. Transthoracic echocardiography was nondiagnostic because of poor image quality, and the patient underwent transesophageal echocardiography instead, which revealed a homogenous echogenic mass in the anterolateral wall of the left ventricle (Figures 1A and 1B and Videos 1A and 1B). The mass was confirmed on subsequent cardiac magnetic resonance imaging (MRI) examination, which demonstrated a neoplasm measuring 1.6 × 1.3 × 1.9 cm along the anterolateral left ventricular wall, eccentrically located toward the subendocardial aspect, partly protruding into the left ventricular cavity (Figure 2). The MRI characteristics of the mass were nonspecific, with the differential diagnosis including both benign and malignant etiologies. Cardiac computed tomography demonstrated that the mass did not contain any calcification and did not encase or directly involve any of the major coronary arterial branches (Figure 3). Fluorodeoxyglucose positron emission tomography was then performed, in an attempt to differentiate benign from malignant etiology. However, the scan revealed mild nonspecific increased fluorodeoxyglucose activity in the cardiac mass, with a standard uptake value of 1.7 and no abnormal fluorodeoxyglucose activity elsewhere in the body. Biopsy of the mass was performed under echocardiographic guidance (Videos 1C and 1D), with detection of unremarkable myocardium on pathology review of the specimen, but no specific tumor diagnosis could be rendered. Because the etiology remained indeterminate and malignancy could not be excluded, the mass was surgically resected, with final pathologic diagnosis of cardiac hamartoma (Figures 4A and 4B).

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DISCUSSION

The clinical presentation of cardiac hamartoma is variable and can range from asymptomatic to arrhythmia, dyspnea, syncope, and sudden death. The left ventricle is the most common location for cardiac hamartomas. Both well-circumscribed and ill-defined morphology has been reported. On pathology analysis cardiac hamartomas are characterized by myocyte hypertrophy and myocyte disarray, with the latter including haphazard, herringbone, pinwheel, and whorled patterns. Other variable features include interstitial fibrosis, interstitial adipose tissue, focal myocyte vacuolization, thick-walled arteries, and dilated venules.[1] Distinguishing hamartoma from other entities, such as focal hypertrophic cardiomyopathy, fibroma, rhabdomyoma, angiosarcoma, and hemangioma, can be challenging.[1-3] Echocardiography, cardiac computed tomography, and MRI can demonstrate the precise location and size of cardiac hamartomas, but rendering a prospective diagnosis is difficult because the imaging appearance is not pathognomonic, and the associated medical imaging literature is composed predominantly of a small number of case reports. On computed tomographic examination, hamartomas demonstrate enhancement and lack of calcification. MRI appearance is variable, ranging from mildly hypointense to hyperintense signal on T2-weighted images, intermediate signal on T1-weighted images, and avid enhancement reported on early- and delayed-phase postcontrast images.[3,4] However, if a focal cardiac mass closely resembles normal myocardium in signal intensity on all MRI pulse sequences, a cardiac hamartoma should be considered in the differential diagnosis, in addition to the focal form of hypertrophic cardiomyopathy. On the other hand, cardiac rhabdomyoma is the most common benign cardiac tumor found in infants and young children. Most of these patients, up to 50%, have signs or family history of tuberous sclerosis.[5,6] These rare neoplasms have solid tan-white homogeneous consistency and are often watery and glistening, with scarce calcification on their cut surface. Microscopically, the presence of large cells with glycogen-containing vacuoles (spider cells) is pathognomonic.[5-7]

Despite the availability of multimodality imaging, in many cases the imaging appearance of cardiac hamartomas is nonspecific, and needle biopsy can be inconclusive given its resemblance to normal myocardium. Thus, if feasible, the only reliable method of establishing the diagnosis of cardiac hamartoma may be surgical resection, followed by microscopic evaluation.[1,2]

CONCLUSIONS

Cardiac hamartoma is a rare clinical entity. Despite advances in multimodality cardiac imaging, surgical resection is the definitive and conclusive diagnostic procedure.

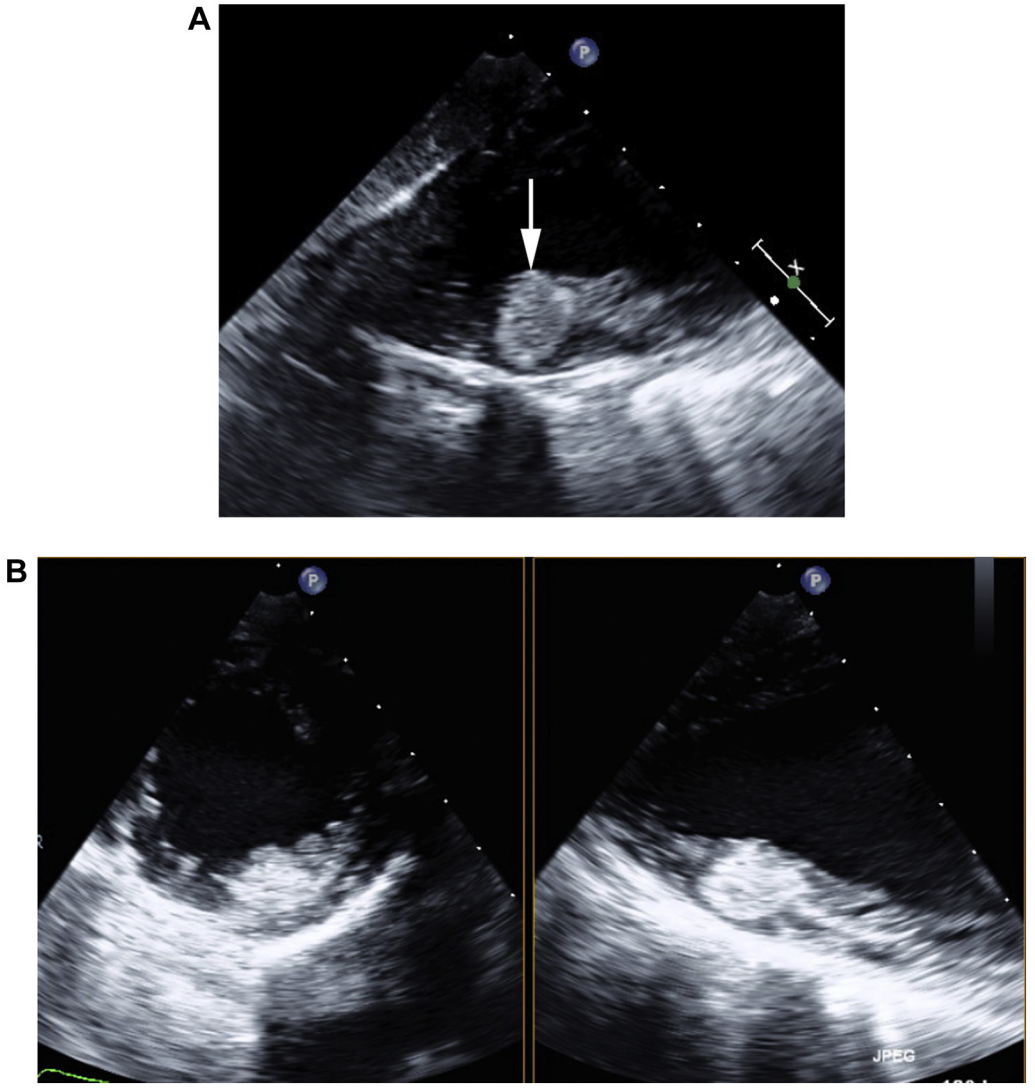

Figure 1 **(A)** Transesophageal echocardiography, transgastric view, angle 92° demonstrates echogenic mass (*arrow*) in the anterolateral wall of the left ventricle partially protruding into the cavity. **(B)** Transesophageal echocardiography, transgastric three-dimensional x-plane with left ventricular short-axis and long-axis views demonstrates the hyperintense mass in the left ventricular wall.

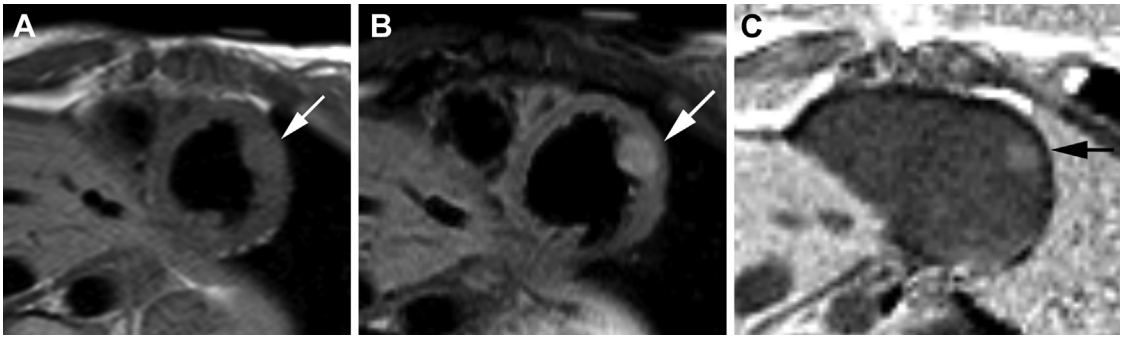

Figure 2 Magnetic resonance images. **(A)** Short-axis T1-weighted image demonstrates intermediate signal intensity of the mass (*arrow*). **(B)** Short-axis T2-weighted image demonstrates intermediate to mildly hyperintense signal of the myocardial mass in the anterolateral wall of the left ventricle (*arrow*). **(C)** Delayed postintravenous gadolinium phase-sensitive inversion recovery image demonstrates avid enhancement of the mass (*arrow*).

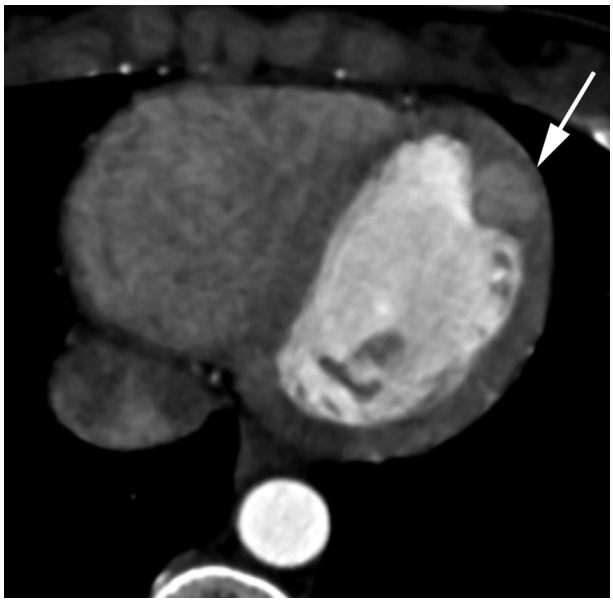

Figure 3 Computed tomographic image demonstrates well-circumscribed enhancing myocardial mass in the left ventricle with no calcification (*arrow*).

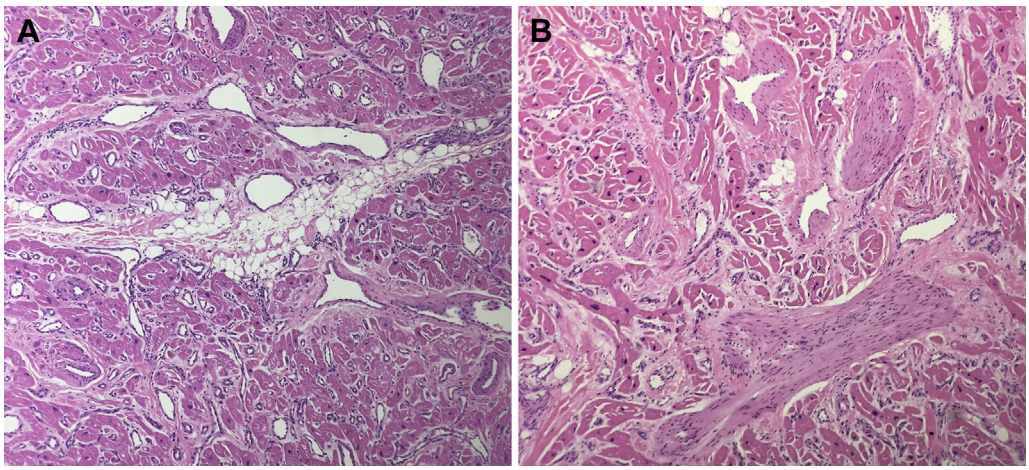

Figure 4 **(A,B)** Histologic sections demonstrate that the resected mass contains hypertrophic myocytes within a disorganized proliferative pattern. The myocytes are present in a background of thick-walled arteries, dilated venules, fibrocollagenous tissue, and mature adipose tissue. The histologic features are consistent with cardiac hamartoma.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.case.2017.01.014.

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