A 33-year-old woman was referred for cardiovascular magnetic resonance imaging to further assess a left ventricular mass found after an echocardiography. Her past medical history was remarkable for multiple basal cell carcinomas and surgical resection of odontogenic cysts, and her family history was negative for cardiac tumors. The patient was in her usual state of health until 6 months before presentation, when she noted increasing fatigue, atypical chest pain, and exertional dyspnea. She also complained of intermittent palpitations without syncope. A transthoracic echocardiography demonstrated a 6×3-cm mass in the left ventricular lateral wall, with deformation of the left ventricular cavity. Left ventricular systolic global function was normal, and there was a trace amount of mitral regurgitation.

The patient underwent cardiovascular magnetic resonance examination with a 1.5 T scanner (Signa CV/i, General Electric, Milwaukee, Wis) with an 8-channel cardiac phased-array coil. Bright blood cine images by steady-state free-precession techniques revealed a 5.5×3.8×4.2-cm mass that was hypointense compared with normal myocardium arising within the lateral wall of the left ventricle (Figure 1). T1-weighted, double-inversion recovery fast-spin echo (black-blood technique) images showed the discrete mass was isointense to slightly hypointense relative to the surrounding normal myocardium (Figure 2). First-pass perfusion images by fast gradient recalled-echo echo-planar technique immediately after a bolus injection of 0.075 mmol/kg gadolinium-diethylenetriamine pentaacetic acid (Magnevist, Berlex Pharmaceuticals, Wayne, NJ) demonstrated a lack of first-pass enhancement (Figure 3), suggesting low tumor vascularity. Delayed enhancement images (Figure 4) were obtained using an inversion-recovery segmented gradient echo sequence (to null normal myocardium) 10 minutes after gadolinium administration (cumulative dose, 0.15 mmol/kg). There was intense late gadolinium enhancement reflecting an increased extracellular volume of distribution in the left ventricular myocardium. Collectively, the tumor appearance and tissue characteristics were consistent with a fibroma. A diagnosis of cardiac fibroma in association with Gorlin syndrome was made.

The patient subsequently underwent surgical resection of the myocardial mass, which was confirmed to be a cardiac fibroma on pathological examination (Figures 5 to 7). Postoperatively, she developed significant mitral regurgitation necessitating mechanical prosthetic (St Jude Medical, St Paul, Minn) mitral valve replacement without further complications.

Gorlin syndrome is a rare autosomal dominant disorder with complete penetrance and variable expressivity.1 Its estimated prevalence is 1 in 57 000. It is caused by mutations in the Patched gene, which acts as a cell-cycle regulator. The hallmark of this syndrome is the presence of multiple basal cell carcinomas, which may appear early in infancy. Other associated features may include craniofacial, central nervous system, musculoskeletal, and genitourinary anomalies. Approximately 3% of cases are associated with cardiac fibromas, which may present later during adulthood rather than the typical infancy or childhood period.1

Disclosures

None.

Reference

Figure 1. Steady-state free-precession (bright-blood) short-axis and 4-chamber images showing a slightly hypointense mass in the lateral wall of the left ventricle.

Figure 2. Double-inversion recovery fast spin-echo image in the short-axis plane.

Figure 3. First-pass perfusion image in the short-axis plane showing hypoperfusion of the mass surrounded by the normally perfused myocardium, which is suggestive of low tumor vascularity.
Figure 4. Delayed myocardial enhancement images demonstrating high signal intensity of the mass compared with the nulled normal myocardium.

Figure 5. Gross pathology of the resected left ventricular mass. The cut section showed an off-white whorled surface with foci of calcification. The lesion extended to the inked surgical margin. There was no hemorrhage or necrosis.
At the periphery of the resection, the lesion intermingled with the surrounding myocardium (immunopositive staining with desmin). The tumor cells showed negative staining with desmin, smooth muscle actin, S100, CD34, and C-Kit.

Figure 6. Hematoxylin & eosin-stained section demonstrating proliferation of bland spindle cells, hyalinized collagen, and focal calcifications. No cytological atypia, necrosis, or mitotic activity was identified. Magnification ×4 (top) and ×10. (bottom).

Figure 7. Desmin immunohistochemical stain. Magnification ×2. At the periphery of the resection, the lesion intermingled with the surrounding myocardium (immunopositive staining with desmin). The tumor cells showed negative staining with desmin, smooth muscle actin, S100, CD34, and C-Kit.
Myocardial Fibroma in Gorlin Syndrome by Cardiac Magnetic Resonance Imaging
Andrew T. Yan, Donna M. Coffey, Yi Li, Wing-Sze Chan, Adolphe J. Shayne, Tuan M. Luu, Ronald B. Skorstad, Maung M. Khin, Kenneth A. Brown, Martin J. Lipton and Raymond Y. Kwong

_Circulation_. 2006;114:e376-e379
doi: 10.1161/CIRCULATIONAHA.105.605832

_Circulation_ is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
Copyright © 2006 American Heart Association, Inc. All rights reserved.
Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the World Wide Web at:
http://circ.ahajournals.org/content/114/10/e376

Permissions: Requests for permissions to reproduce figures, tables, or portions of articles originally published in _Circulation_ can be obtained via RightsLink, a service of the Copyright Clearance Center, not the Editorial Office. Once the online version of the published article for which permission is being requested is located, click Request Permissions in the middle column of the Web page under Services. Further information about this process is available in the Permissions and Rights Question and Answer document.

Reprints: Information about reprints can be found online at:
http://www.lww.com/reprints

Subscriptions: Information about subscribing to _Circulation_ is online at:
http://circ.ahajournals.org/subscriptions/