Utility of Real-Time 3-Dimensional Echocardiography and Magnetic Resonance Imaging for Evaluation of Danon Disease

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A 21-year-old man was admitted to the hospital with exertional dyspnea. At the age of 14, he was diagnosed with Danon disease by genetic analysis (a 2–base-pair deletion at positions 288 and 289 in exon 3 was identified in the lysosome-associated membrane protein-2 [LAMP2] gene of the patient, which led to a frameshift and resulted in a premature stop codon). A chest radiograph demonstrated moderate cardiomegaly (cardiothoracic ratio of 58%). The ECG exhibited normal sinus rhythm, wide and bifid P waves with a duration of 170 ms, complete left bundle-branch block with a QRS duration of 200 ms, and a leftward axis (–2°; Figure 1).

Echocardiography revealed left ventricular (LV) dilatation with an end-diastolic internal dimension of 60 mm and diffusely hypokinetic LV wall motion with fractional shortening of 8% (Figure 2; Movie I of the online-only Data Supplement). LV concentric hypertrophy was also found, with a wall thickness of 13 mm, but the LV apical portion was thinner than the other portions (Figure 2). An LV noncompaction, characterized by prominent trabeculations and deep intertrabecular recesses, was observed at the apex and midventricular segment of the LV posterior and lateral walls (small arrows in Figure 2B and 2C). The affected segments had a 2-layer structure, which consisted of a compact epicardial layer and an endocardial layer that consisted of a prominent trabecular meshwork and deep intertrabecular spaces; the ratio of the noncompacted to compacted myocardial layers at the site of maximal wall thickness was 2.3 (Figure 2B and 2C; Movie II of the online-only Data Supplement). In addition, an LV apical mass suggestive of a thrombus was found (Figure 2B). Real-time 3-dimensional echocardiography revealed that the entire trabecular projections and intertrabecular recesses were visualized simultaneously, and the distinctions between the compacted and noncompacted LV myocardium and the thrombus present within the noncompacted LV myocardium were easily demarcated (Figure 3; Movie III of the online-only Data Supplement). Cardiac magnetic resonance imaging 3 weeks after initiation of anticoagulant therapy demonstrated a dilated hypocontractile LV with concentric hypertrophy. Furthermore, first-pass rest perfusion images disclosed a subendocardial perfusion deficit clearly visible within the anterior, lateral, and posterior walls and heavily spongious, “trabecularized” endocardial layers with deep intertrabecular recesses in those regions (Figure 4A; Movie IV of the online-only Data Supplement). Late gadolinium enhancement was found in the subendocardium, especially in the anterior, lateral, and...
posterior LV, and in the midlayer of the interventricular septum (arrows in Figure 4B and 4C). A late gadolinium enhancement was also observed transmurally in the apical portion (Figure 4C, right panel). The patient was discharged receiving anticoagulant therapy, medical treatment for heart failure, and a cardiac resynchronization therapy device with an implantable cardioverter defibrillator, and he has done well with no deterioration of his heart failure during a 6-month follow-up period.

Danon disease is a rare X-linked disorder characterized by cardiomyopathy, skeletal myopathy, and mental retardation. The cardiomyopathy may mimic a dilated phase of hypertrophic cardiomyopathy as in the present case, and prophylactic anticoagulant therapy is recommended in patients with hypokinetic LV wall motion. The recent advances in real-time 3-dimensional echocardiography and magnetic resonance imaging enabled us to assess cardiac function and myocardial tissue characterization precisely and noninvasively. In the present case, real-time 3-dimensional echocardiography was useful for diagnosing and characterizing the apical thrombus and LV noncompaction that are often associated with genetic and neuromuscular disorders. The magnetic resonance imaging revealed that the pattern of the perfusion deficit and late gadolinium enhancement was atypical for hypertrophic cardiomyopathy or myocarditis. Thus, magnetic resonance imaging was useful for differentiating ischemic from nonis-
chemic cardiomyopathies on the basis of the patterns of the enhancement and may be useful in discriminating between different forms of nonischemic cardiomyopathies.3,4

Disclosures
None.

References

Figure 4. Cardiac magnetic resonance images. A, First-pass rest perfusion images in a short-axis orientation. B and C, Late gadolinium enhancement images in a 4-chamber and short-axis orientation. Arrows indicate late gadolinium enhancement in the midlayer of the interventricular septum.
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