Objectives: To report a case of a previously asymptomatic patient who was diagnosed with hypotrophic cardiomyopathy (HCM) on echocardiogram, which was later found to represent right ventricular dysplasia (RVD).

Methods: We present a case of a 35-year-old woman who presented with exercise-induced fatigue and chest pain. A transthoracic echocardiogram revealed akinesia of the left ventricle and hypotrophy of the right ventricle. A right heart catheterization was performed which showed normal right heart pressures and tricuspid regurgitation consistent with primary RVD. The patient was referred for endomyocardial biopsy which showed features of both HCM and RVD.

Results: The patient was subsequently referred for a cardiac magnetic resonance imaging (MRI) which confirmed the diagnosis of RVD. The patient was treated with beta-blockers and was asymptomatic at her latest follow-up.

Conclusions: The diagnosis of RVD can be challenging and requires a high index of suspicion. Right heart catheterization and endomyocardial biopsy can help confirm the diagnosis. A comprehensive cardiac MRI is recommended in patients with atypical presentations of RVD.

Keywords: Hypotrophic cardiomyopathy, Right ventricular dysplasia, Cardiac Magnetic Resonance Imaging.
echocardiogram showed a thick to thin compacted layer ratio of 3.2:1, consistent with a diagnosis of LVNC (Zurich and Milwaukee echocardiogram criteria >2:1).\textsuperscript{7} Expert 1’s interpretation of our MRI confirmed the diagnosis of LVNC, showing prominent trabeculations in the left ventricular cavity extending to the apex, and established a ratio of nontrabeculated to compacted myocardium of 3:1 (Petersen MRI criteria ratio >2.3:1).\textsuperscript{8,9} A 24-hour Holter monitor to rule out nonsustained ventricular tachycardia and aspirin, 81 mg/d, for stroke prevention were recommended. An implantable cardiac defibrillator was deferred since the patient was asymptomatic with a preserved ejection fraction. Notably, Expert 1 recommended that the patient continue to refrain from organized sports and avoid “burst exertional” activity, although recreational aerobic activity would be regarded as safe.

Expert 2 performed an exercise treadmill test, which was negative for arrhythmia. A transthoracic echocardiogram again showed an ejection fraction of 60% and left ventricular trabeculations at the apex potentially consistent with LVNC. How-
ever, the noncompacted to compacted segment ratio determined was 1.8, which did not meet the criteria for LVNC. A 24-hour Holter monitor again did not record any ventricular ectopy. Expert 2 concluded that, given normal wall motion as well as a normal enhancement pattern without evidence of strain, this case represented a “normal variant in a black athlete” rather than pathologic LVNC. Furthermore, unlike Expert 1, Expert 2 recommended that the patient be allowed to return to full athletic competition but repeat the 24-hour Holter monitor and cardiac MRI in 2 to 3 months. In addition, it was proposed that all first-degree relatives undergo echocardiographic screening.

Discussion

The discussions with the consultants were reviewed with the patient, family, and the team physicians. We decided that despite conflicting opinions and a likely low yet unquantified risk of sudden death, it would be in the patient’s best interest to return to play. The patient returned to full athletic participation. He has performed very well, has remained asymptomatic, and is reevaluated annually. Family screening echocardiograms have been negative for abnormalities.

Left ventricular noncompaction is characterized by a prominent trabecular meshwork and deep intratrabecular recesses that extend into the left ventricular wall. It is believed to be caused by the arrest of normal intrauterine myocardial morphogenesis. Left ventricular noncompaction is an extremely rare cardiomyopathy with a prevalence of less than 0.3% in the adult population. Hypertrabeculation also can be seen in association with a number of other cardiac abnormalities as well as in the healthy heart. In the largest study of its kind, Gati et al used echocardiography to show that athletes had a higher prevalence of hypertrabeculation compared with nonathletes (18.3% vs 7%). This finding was even more pronounced in African and Afro-Caribbean athletes, but only a small proportion experienced systolic dysfunction and marked repolarization suggestive of cardiomyopathy. Captur et al applied fractal geometry analysis to cardiac MRI to quantify left ventricular trabeculation. They showed increased trabeculation in healthy African Americans compared with healthy whites. Interestingly, a large proportion of patients with sickle cell disease demonstrate increased left ventricular trabeculation that meets diagnostic criteria for LVNC, while it is unlikely they have LVNC. It was hypothesized that hypertrabeculation could be an ethnically determined adaptation to increased preload seen in both sickle cell disease and athletic training.

Typical clinical manifestations of LVNC that occur equally in adults and children include the triad of heart failure, arrhythmia, and embolic events. Electrocardiographic findings often demonstrate left ventricular hypertrophy with increased voltages, inverted T waves, and Wolff-Parkinson-White syndrome. There is no pathognomonic histologic feature to aid in the diagnosis of LVNC, although fibrosis has been reported. Diagnosis is usually made using echocardiography with increasing use of cardiac MRI. Jenni et al defined LVNC as a ratio of the thick noncompact layer to the thin compact layer of more than 2.0, as measured in the short-axis view. Using MRI, Petersen et al defined LVNC as a ratio between noncompact and compact layers of more than 2.3 at end-diastole. Magnetic resonance imaging technology has improved the definition of the anatomy but is not as accessible as echocardiography, especially in the context of preparticipation screening. The patient’s case emphasizes the caveats of preparticipation screening. He was a highly recruited athlete expected to compete at an extreme level, and obtaining an echocardiogram to assess a new murmur was reasonable. The incidental findings suggestive of LVNC, beyond the obvious medical impact, could have curtailed his collegiate career immediately as well as his prospects as a professional athlete. We were faced with a controversial diagnosis that, unlike conditions such as hypertrophic cardiomyopathy and long QT syndrome, has limited data associated with it to guide management. Regardless of guidelines, expert consultation should be considered in challenging cases such as ours to provide the patient and family and his or her health care providers with as much information as possible before deciding on participation or restriction.

Conclusions

To our knowledge, no reported cases of sudden death in athletes have been attributed to LVNC. We expect as imaging modalities continue to improve and as preparticipation screening becomes more prevalent, scenarios such as ours will become increasingly common. Patients with hypertrabeculation but preserved left ventricular function may represent a low-risk group. Close follow-up along with longitudinal registry studies will continue to be important to establishing risk in patients such as ours.

REFERENCES