CASE REPORT

Non-compaction cardiomyopathy in an asymptomatic athlete

Margaret Kapor Manus, Satyajee Roy, Rosemarie Stag, Daniel Hyman

Department of Internal Medicine, Cooper University Hospital, Camden, New Jersey, USA

Correspondence to

Dr Margaret Kapor Manus, manus-margaret@cooperhealth.edu

Accepted 2 August 2016

SUMMARY

Prevention of sudden cardiac death in athletes requires the screening and recognition of pathologies that often remain clinically silent for years until provoked by a physiologic stressor. This can result in the manifestation of disease and even death. Left ventricular non-compaction cardiomyopathy (LVNC), newly classified as a distinct entity arising in the adult population, is a cardiomyopathy that at initial presentation can manifest as a wide spectrum of symptoms from asymptomatic to ventricular arrhythmias, systemic embolism and even sudden cardiac death. We present the case of an asymptomatic athlete found to have LVNC and discuss the implications this finding may have on sports participation.

BACKGROUND

Left ventricular non-compaction cardiomyopathy (LVNC) is the most recent cardiomyopathy to be classified as a distinct entity by the American Heart Association (AHA). Once thought to be a rare diagnosis, seen largely only in the paediatric population, LVNC is now being detected with increased frequency in the adult population.¹ Individuals diagnosed with LVNC have been found to be at an increased risk of ventricular arrhythmias, systemic embolism, progressive heart failure and even sudden cardiac death.²

CASE PRESENTATION

A 20-year-old African-American man presented to the outpatient office for a routine sports physical examination prior to joining a university-level basketball team. On presentation, he offered no symptoms. He stated that he was very active and played basketball year round. On review of symptoms, he denied having chest pain, palpitations, dizziness, syncope or undue shortness of breath with or after physical activity. His medical history was significant for asthma as a child. He took no medications. He had no significant family history including early or sudden death prior to the age of 50 years. At the time of his examination, the patient's temperature was 97.5°F (36.4°C), blood pressure 110/82 mm Hg and heart rate 67 bpm. On physical examination, the patient had a normal first heart sound, a late grade 3/6 systolic murmur best heard at the second intercostal space along the left sternal border and a second heart sound with a fixed split S2. The remainder of the examination was within normal limits.

INVESTIGATIONS

An ECG demonstrated normal sinus rhythm, no ST changes, normal T waves and a normal QTc interval. There were no abnormal ventricular conduction dysrhythmias. An initial two-dimensional echocardiogram (2-D echo) revealed cardiomegaly with an ejection fraction visually estimated to be between 35% and 40%. His left ventricular (LV) apex had moderate trabeculations. His exercise stress test was normal. A cardiac MRI study showed hypertrabeculation along the lateral and apical wall of the left ventricle. The ratio of trabeculated (13 mm) and compacted (5 mm) layers in end diastole was 2.6. The left ventricular ejection fraction (LVEF) calculated on MRI was estimated to be 45% (figure 1). The patient was diagnosed with LVNC.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis includes hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM). In HCM and DCM, it is uncharacteristic for the ratio between trabeculated myocardium and normal myocardium to be >2. Additionally, HCM and DCM demonstrate a diffuse pattern of trabeculations.²

TREATMENT

No additional therapies were started at this time.

OUTCOME AND FOLLOW-UP

The patient was referred to a cardiologist specialising in sports medicine. The patient underwent a tailored exercise stress test that consisted of periods of burst stress, up to 10 miles per hour at a 1% grade, meant to simulate sports participation. This study showed no arrhythmias, excellent exercise tolerance, normal resting LV function and a resting ejection fraction of 55%. These data were suggestive of a low-risk state in the setting of LVNC. The ejection fraction of 45% determined by the cardiac MRI categorised our patient as "borderline or intermediate ejection fraction group" as defined by the 2013 American College of Cardiology Foundation (AACF)/AHA Task Force practice guidelines. According to the AACF/AHA practice guidelines, a LVEF value between 41% and 49% falls into the borderline or intermediate ejection fraction group. The clinical course, outcome and therapeutic intervention of this group appear to be similar to those of patients with preserved ejection fraction (LVEF ≥50%).³ With this information, as well as a normal ECG, the patient was cleared for sports participation with periodic scheduled

CrossMark

To cite: Manus MK, Roy S, Stag R, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-216339

Figure 1 Cardiac MRI showing characteristic thin outer compact layer and thicker inner non-compact layer with deep recesses.

monitoring. The patient has been followed on a 6-month basis for cardiac imaging and will continue to do so for long as he remains asymptomatic. He has been advised to report to the nearest emergency room immediately should he develop symptoms, such as fainting, palpitations, dizziness, chest pain or shortness of breath.

DISCUSSION

LVNC is a myocardial disorder characterised by prominent trabeculae, intratrabecular recesses and a LV myocardium with two distinct layers, compacted myocardium and non-compacted myocardium.¹ During normal embryologic development, the heart consists of a network of loose muscle fibres and trabeculations that gradually become compacted during the 12th and 18th weeks of gestation. In LVNC, it is thought that this process of compaction fails to occur, leading to the development of a thickened non-compacted endomyocardial layer and a thin compacted epicardial layer.⁴

The prevalence in the general population of LVNC is unknown. Studies have shown significant variability. One study looking at all individuals referred for echocardiography demonstrated a prevalence of 0.05–0.26%.² In a study conducted on individuals with an LVEF of <45%, the prevalence of LVNC was as high as 3.7%.¹

Patients may present with a variety of symptoms, ranging from asymptomatic to end-stage heart failure symptoms. The most commonly reported complications of LVNC include heart failure, ventricular arrhythmias and systemic embolism.⁵ Consequently, it is associated with significant morbidity and mortality. One of the earliest long-term studies followed individuals diagnosed with LVNC for 44 months, during which time 35.3% of patients died, half of which were due to sudden death, and 11.8% required cardiac transplant.⁶ In a more recent study that followed patients for 46 months, 10.8% died, 42.9% were due to sudden death, and 13.8% required cardiac transplant.⁷ In both studies, no major cardiac events were noted in patients who were asymptomatic at the time of diagnosis.

Echocardiogram and now more frequently cardiac MRI are used to make the diagnosis of LVNC. In our patient, measurement of LVEF differed between the 2-D echo and the cardiac MRI. Studies have demonstrated an observer bias often affects the measurement of LVEF by 2-D echo significantly.⁸ It has been shown that 2-D echo underestimates LV volumes compared to other imaging modalities, such as cardiac MRI. Some studies conclude that the discrepancies are large, with a lack of

agreement of 21–25% between conventional methods.⁹ The 3-D approach of cardiac MRI is considered the preferred technique for LV volume and ejection fraction estimation especially in non-symmetric ventricles due to its superior image quality.¹⁰

Uniform diagnostic criteria have not been established for LVNC. The most commonly employed definition was proposed by Jenni *et al*: (1) the absence of coexisting cardiac anomalies; (2) the presence of multiple, prominent trabeculations; (3) multiple deep intertrabecular recesses filled with blood from ventricular cavity, as demonstrated by colour Doppler imaging; and (4) the maximal end-systolic ratio of non-compacted endocardial layer (NC) to the compacted myocardium (C) of NC to C ratio >2.⁴ Our patient met all four criteria.

There are no specific therapies or follow-up guidelines for LVNC at this time. However, management often focuses on three of the major clinical manifestations: heart failure, arrhythmias and systemic embolic events.² Standard heart failure management often includes the use of ACE-inhibitors, β blockers and diuretics in individuals with a reduced ejection fraction and LVNC. In terms of monitoring for arrhythmias, often individuals with LVNC are periodically monitored with ambulatory ECG. The decision for placement of an ICD is typically reserved for patients with syncope, symptomatic ventricular arrhythmias or severely impaired LV systolic function. The use of anticoagulation has been greatly debated for the purpose of systemic embolism prevention. It has been proposed that individuals with high-risk features such as atrial fibrillation, prior history of systemic embolism, severely impaired systolic function and LV thrombus may benefit from long-term anticoagulation. Other studies have suggested that all patients with LVNC should be placed on long-term anticoagulation.¹¹ In patients with severe heart failure who have failed medical therapy, transplant may be required.⁵

Guidelines regarding sports participation for patients with LVNC are limited. In the 36th Bethesda Conference Guidelines, published by the American College of Cardiology (ACC), 2005, it was recommended that individuals with LVNC be excluded from competitive sports with the possible exception of those sports of low intensity.¹² The AHA, as of December 2015, released an updated statement to the Bethesda 36 report in regard to eligibility and disqualification criteria for competitive athletes with LVNC. It was their recommendation that participation in competitive sports may be permissible for asymptomatic patients with a diagnosis of LVNC so long as they have a normal systolic function, without ventricular tachyarrhythmias on ambulatory monitoring or exercise testing, and no prior history of unexplained syncope. As for athletes with impaired systolic function, atrial or ventricular tachyarrhythmias on ambulatory monitoring or exercise stress testing, or with a history of syncope, it is the AHA/ACC recommendation that athletes should not participate in competitive sports.¹³

Learning points

- ▶ Left ventricular non-compaction cardiomyopathy (LVNC) carries a high morbidity and mortality.
- ▶ LVNC should be included in the differential diagnosis of an abnormal cardiac examination.
- ▶ Sports participation in those athletes diagnosed with LVNC will have to be considered on an individual basis as participation carries increased risk for sudden death.

Contributors DH, MKM and RS participated in the conception and design of the case report. MKM drafted the article. DH and SR revised the article critically for important intellectual content. All authors participated in the final approval of the version published. MKM, SR, DH and RS all agree to be accountable for the article and to ensure that all questions regarding the accuracy or integrity of the article have been investigated and resolved.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Towbin JA, Lorts A, Jefferies JL. Left ventricular non-compaction cardiomyopathy. *Lancet* 2015;386:813–25.
- 2 Carrilho-Ferreira P, Almeida AG, Pinto FJ. Non-compaction Cardiomyopathy: prevalence, prognosis, pathoetiology, genetics, and risk of cardioembolism. *Curr Heart Fail Rep* 2014;11:393–403.
- 3 Yancy CW, Jessup M, Bozkurt B, *et al.* 2013 ACCF/AHA guideline for the management of heart failure: executive summary: a report of the American College of Cardiology Foundation/American Heart Association Task Force on practice guidelines. *Circulation* 2013;128:1810–52.
- 4 Ikeda U, Minamisawa M, Koyama J. Isolated left ventricular non-compaction cardiomyopathy in adults. *J Cardiol* 2015;65:91–7.
- 5 Hussein A, Karimianpour A, Collier P, *et al.* Isolated noncompaction of the left ventricle in adults. *J Am Coll Cardiol* 2015;66:578–85.
- 6 Oechslin EN, Jost CH, Rojas JR, *et al.* Long-term follow-up of 34 adults with isolated left ventricular noncompaction: a distinct cardiomyopathy with poor prognosis. *J Am Coll Cardiol* 2000;36:493–500.
- 7 Lofiego C, Biagini E, Pasquale F, *et al.* Wide spectrum of presentation and variable outcomes of isolated left ventricular non-compaction. *Heart* 2007;93:65–71.
- 8 Malm S, Frigstad S, Sagberg E, *et al.* Accurate and reproducible measurement of left ventricular volume and ejection fraction by contrast echocardiography—a comparison with magnetic resonance imaging. *J Am Coll Cardiol* 2004;44:1030–5.
- 9 Bernard Y, Meneveau N, Boucher S. Lack of agreement between left ventricular volumes and ejection fraction determined by two-dimensional echocardiography and contrast cineangiography in postinfarction patients. *Echocardiography* 2001;18:113–22.
- 10 Bellenger NG, Burgess MI, Ray SG. Comparison of left ventricular ejection fraction and volumes in heart failure by echocardiography, radionuclide ventriculography and cardiovascular magnetic resonance. Are they interchangeable? *Eur Heart J* 2000;21:1387–96.
- 11 Aras D, Tufekcioglu O, Ergun K, *et al.* Clinical features of isolated ventricular noncompaction in adults long-term clinical course, echocardiographic properties, and predictors of left ventricular failure. *J Card Fail* 2006;12:726–33.
- 12 Maron BJ, Zipes DP. Introduction: eligibility recommendations for competitive athletes with cardiovascular abnormalities—general considerations. *J Am Coll Cardiol* 2005;45:1318–21.
- 13 Maron BJ, Udelson JE, Bonow RO, *et al.* Eligibility and disqualification recommendations for competitive athletes with cardiovascular abnormalities: Task Force 3: hypertrophic cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy and other cardiomyopathies, and myocarditis. *J Am Coll Cardiol* 2015;66:2362–71.

Copyright 2016 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow