

Asymmetric Hypertrophic Cardiomyopathy in Athletes: Always a Higher Risk for Sudden Death?

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ABSTRACT

Hypertrophic cardiomyopathy (HCM) may be associated with considerable mortality in athletes. However, differentiating myocardial hypertrophy as a physiological adaptation of the heart to exercise can be a clinical challenge. In this context, nuclear magnetic resonance imaging has been shown to be an essential exam for diagnostic elucidation. The case report aimed to depict a young athlete with syncope and an initial investigation suggestive of HCM, which was excluded after deconditioning and serial MRI.

KEYWORDS: Sudden death; Athlete heart; Hypertrophic cardiomyopathy.

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INTRODUCTION

Hypertrophic cardiomyopathy (HCM) is a genetic myocardial disease caused by the contractile protein disarrangement of the sarcomeres, which represents the main reason for sudden death in young people¹. Although the overall prognosis is relatively good, with an annual mortality rate of less than 1%, there is a propensity for potentially lethal ventricular arrhythmias². Myocardial hypertrophy in the absence of heart (e.g. aortic valve stenosis) or systemic diseases (e.g. hypertension and amyloidosis) characterizes HCM, which may be responsible for up to 40% of cardiac death in athletes¹. After excluding secondary causes, the prevalence is as high as 1:500, and should be ruled out in professional or amateur athletes³.

CASE DESCRIPTION

A 31-year-old male patient, with no comorbidities and absence of familiar sudden death or cardiac disease, complained of syncope during soccer practice. He described a sudden loss of consciousness accompanied by muscle spasm and rapid recovery (approximately 1min). He was used to playing soccer three times a week and trained daily, but without previous symptoms. He arrived asymptomatic to the emergency room (ER), with blood pressure of 100/60mmHg, heart rate of 64bpm, and normal cardiac physical examination. The patient was released after excluding acute coronary syndrome through electrocardiogram (ECG) and laboratory investigation, with a recommendation to outpatient follow-up. The ECG carried out at the ER was sinus rhythm with minimal abnormality of the T wave and suspicion of left ventricle (LV) hypertrophy (Fig. 1).

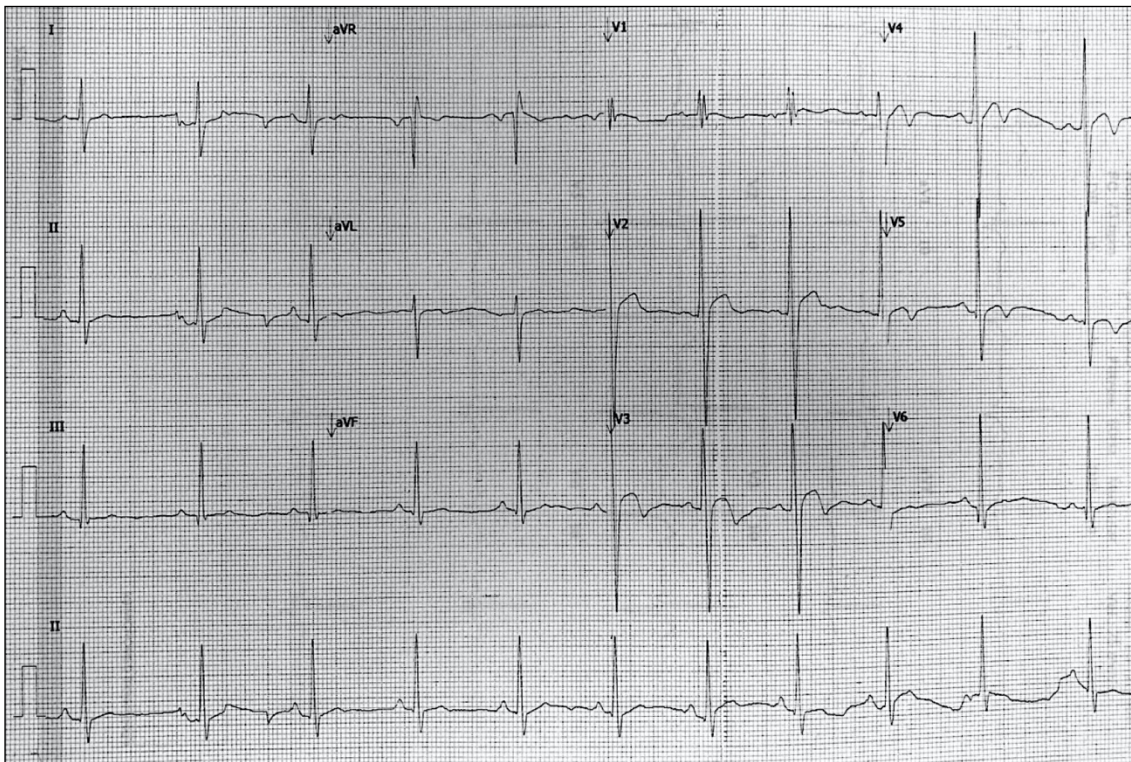


Figure 1. Initial electrocardiogram showing abnormal repolarization.

The patient remained asymptomatic with normal 24h-Holter monitoring and maximal exercise stress test. A transthoracic echocardiogram (TTE) was performed, which revealed normal LV systolic function with an ejection fraction (EF) of 68%, LV end-diastolic diameter (LVEDD) of 45mm, and an asymmetric increase in interventricular septum thickness (IVS, 15mm vs. posterior wall, 8mm) (Fig. 2).

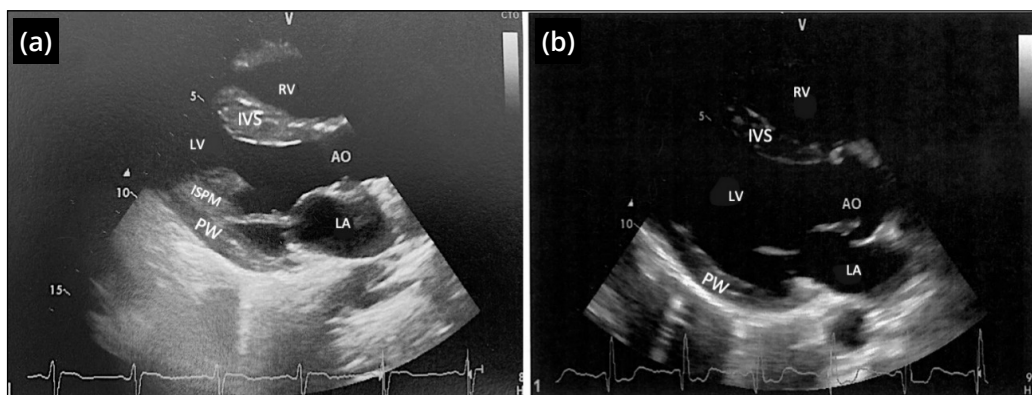


Figure 2. Transthoracic echocardiogram before (a) and after 4 months (b) of deconditioning. LA=left atrium; LV=left ventricle; RV=right ventricle; IVS=interventricular septum; PW=posterior wall; Ao=aorta; ISPM=inferoseptal papillary muscle.

Further evaluation for HCM included cardiac magnetic resonance imaging (MRI) with gadolinium enhancement (Fig. 3). This showed LV hypertrophy exclusively of the IVS with main thickness in the infero-septal segment (14mm). The LVEDD was 54mm and LVEF was 62%. There was a gadolinium enhancement of approximately 8.7% of the LV total mass (9.5g). Heterogeneous delayed enhanced compatibility with fibrosis was detected in the middle myocardium of the inferior and superior segments of the septal wall.

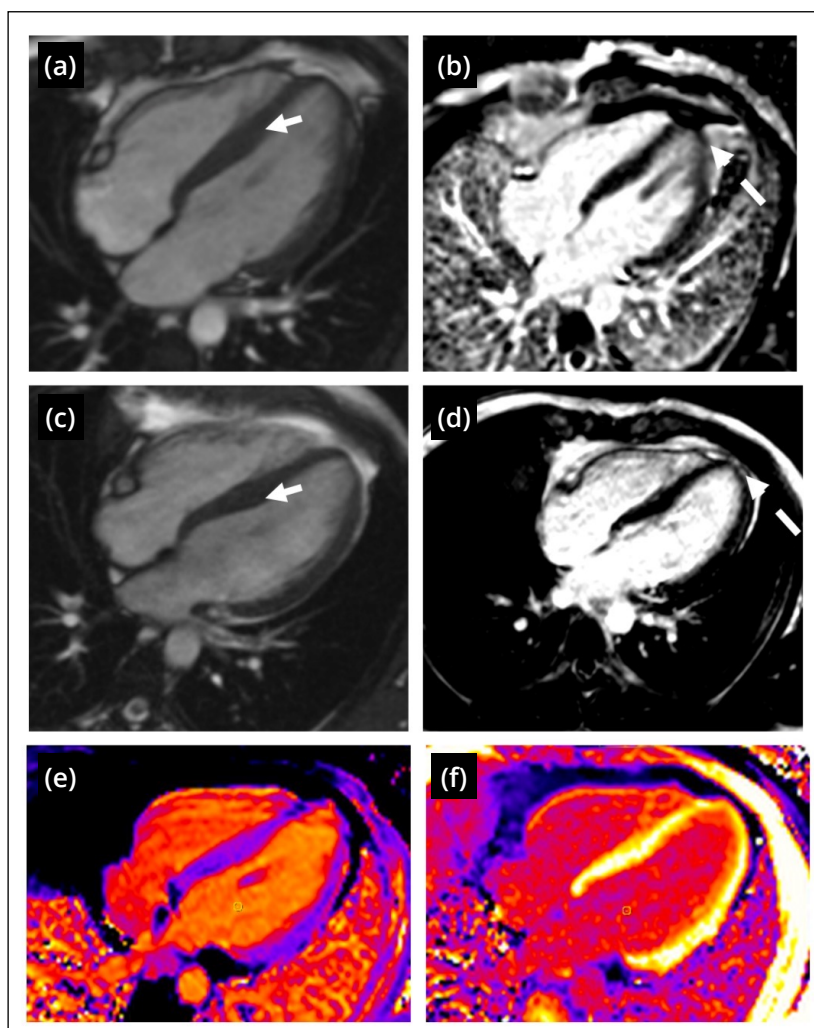


Figure 3. Magnetic resonance imaging (MRI) before and after 5 months of deconditioning. The 4-chamber diastolic (full arrow) shows septal thickening (a). Delayed enhancement suggests middle myocardium fibrosis – dashed arrow (b). After 5 months, MRI shows reuction of septal thickening (c) and absence of fibrosis (d). Native T1 (e) and after gadolinium T1 maps (f) were completely normal.

The risk of sudden death was 2.68% using the HCM Risk-SCD calculator, which did not recommended ICD implantation^{2,4}. He was recommended to completely stop physical exercise. During follow-up, serial ECG showed progressive changes of the ventricular repolarization from abnormally inverted to normal T waves (Fig. 4).

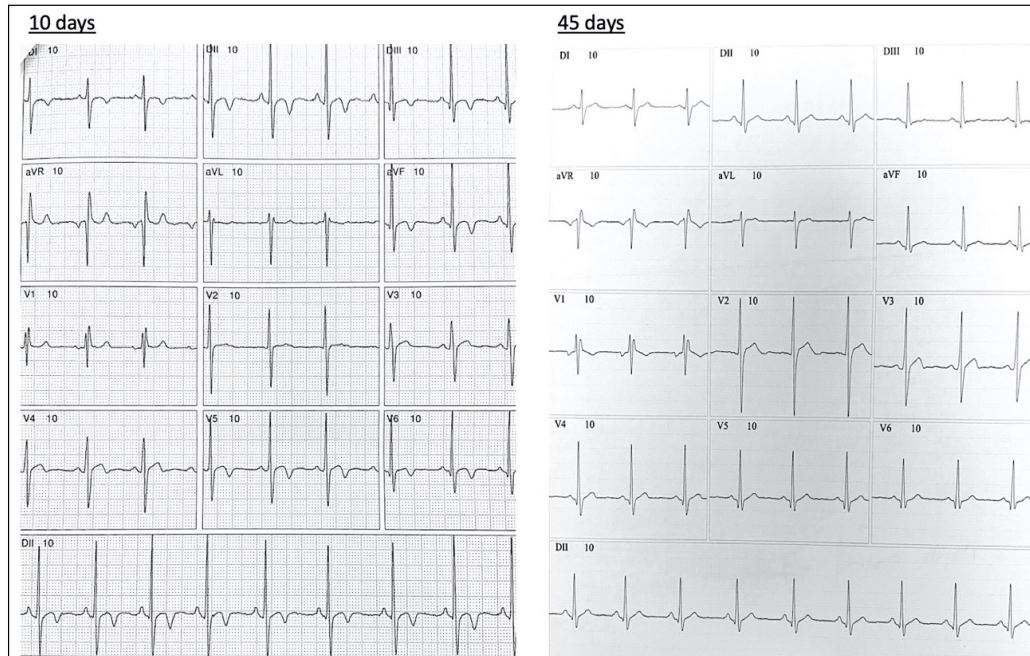


Figure 4. Electrocardiogram after 10 and 45 days of deconditioning.

The patient had no more syncope during 4 months of follow-up. TTE was compatible with normality (Fig. 2). Another MRI was performed, which revealed complete regression and normalization of the septal thickness (9mm). Delayed enhanced and T1 mapping showed absence of myocardial fibrosis (Fig. 3). A head-up tilt table (HUT) testing was then performed with a positive response that reproduced the previous syncopal event. Asymmetrical HCM was ruled out and the patient was then released for sports activities, being diagnosed for Athlete's Heart (AH).

DISCUSSION

Hypertrophic cardiomyopathy is a genetic cause of sudden death, whose incidence in the general population is not negligible. This pathology, being the main cause of death in athletes, should be ruled out in people who take part in competitive sports. The distinction between pathological abnormalities of HCM and AH is a real diagnostic challenge, with several implications for the patient, ranging from exposure to the risk of sudden death to the unnecessary withdrawal from physical activities.

Some strategies, as genetic and imaging evaluation, can be adopted in this scenario to help clarify the cause of hypertrophy. Genetic testing in this context is very enlightening if positive, though a negative test does not exclude the possibility of HCM⁵. Another useful test is the TTE, showing a LV diastolic diameter greater than 55mm, which is common in trained athletes but rarely present in patients with HCM, wherein the cavity diameter is commonly less than 45mm. However, diameters above 55mm may be present in HCM, but only in advanced stages⁶. Diastolic disturbances seen in tissue and pulsatile doppler, regardless of outflow tract obstruction, are common in HCM, but absent in AH⁶.

Magnetic resonance imaging has a fundamental role in the detailed assessment of the myocardium. This diagnostic tool allows the assessment of ejection fraction and ventricular volumes as well as an accurate measurement of thickness and myocardial mass. Through assisting techniques such as delayed enhancement and T1/T2 maps, it is possible to assess the myocardial structure by identifying myocardial edema, alteration of extracellular volume and myocardial fibrosis. Adaptive physiological modifications in the myocardium of trained athletes and overlapping conditions may be assessed by MRI⁷.

Typical MRI findings in trained athletes are mild dilation, more pronounced in the LV and possibly also in the right ventricle (RV), with preserved biventricular EF. Myocardial thickness may be slightly increased (usually less than 15mm) and with homogeneous or symmetrical cardiac involvement. Myocardial fibrosis is not evident, and there are no signs of myocardial infiltration such as edema or increased extracellular volume (Tab. 1). The case report shows an asymmetric septal hypertrophy with delayed enhancement at baseline MRI, which were normalized in the image after deconditioning:

- 1) myocardial mass was reduced;
- 2) septal thickness was decreased; and
- 3) there was remission of late enhancement.

Table 1. Differential diagnosis by cardiac resonance imaging.

Findings	Athlete	Hypertrophic cardiomyopathy
LV hypertrophy	<15mm	>13mm
LV morphology	symmetric	asymmetric
Systolic LV function	normal	supranormal
Delayed enhancement	absent	present
T1/T2 mapping	normal	abnormal
deconditioning	normalize	unchanged

LV=left ventricle.

These findings were corroborated by the absence of myocardial infiltration in the native and post-gadolinium T1 and T2 maps.

Physical deconditioning in elite athletes with LV hypertrophy promote a 2-5mm reduction in wall thickness over a 4-month period⁸. This decrease in LV wall thickness associated with deconditioning confirms that LV hypertrophy is a physiological consequence of training. Such changes in wall thickness are inconsistent with pathological HCM hypertrophy. However, deconditioning requires athletes compliance and serial TTE or MRI.

We reported a young patient with neuromediated syncope, as demonstrated by HUT testing, and initial anatomical findings suggesting asymmetrical HCM reversed by physical deconditioning. This case shows the importance of physical deconditioning and serial assessment of trained patients to avoid misdiagnosis and ensure correct management.

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