

Hypertrophic Cardiomyopathy and Exercise Limitation: Time to Reconsider. A Case Report

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Abstract

The health benefits of physical activity are well established. Patients with hypertrophic cardiomyopathy (HCM) are often discouraged from participating in physical activity because of a perceived increased risk of sudden cardiac death (SCD). Although most SCDs are unrelated to HCM, guidelines have traditionally focused on preventing the potential risk by limiting physical activity, without clear recommendations on how to reverse the negative health effects of inactivity. Given the growing evidence that carefully supervised exercise can be both beneficial and safe in patients with HCM, the author describes an original case report that draws the attention of sports medicine and cardiology experts to whether it is time to abandon exercise restrictions in HCM in selected cases.

Keywords: Pre-participation sports screening, Hypertrophic Cardiomyopathy, Physical Activity, Sudden Cardiac Death.

Introduction

HCM has a prevalence of approximately 1:500 and is characterised by either segmental or diffuse LV hypertrophy without cavity dilatation and in the absence of other cardiac or systemic causes. The diagnosis of HCM is made when the LV wall thickness is ≥ 15 mm. In cases where LV hypertrophy is milder, with LV wall thickness between 13-14 mm, the diagnosis can still be made if there is a positive family history, symptoms, ECG abnormalities and/or pathogenic genetic variants.¹ Initially, pathological studies suggested that HCM was a major cause of SCD in athletes, accounting for up to 40%. However, more recent evidence suggests a much lower percentage (6-16%). Studies have also shown that tailored exercise programmes for HCM patients can be safe.² For example, a study with a median follow-up of 7 years in 88 HCM patients found no difference in symptoms and adverse events between those who stopped exercising after diagnosis and those who continued, with no adverse events reported in the 27 who continued to exercise.³ Another study in 53 low-risk HCM athletes, including professional athletes, documented only two events during follow-up.⁴ An international registry of athletes with ICDs found that only one of the 65 athletes with HCM experienced a shock during follow-up.⁵ A recent survey also highlighted a similar incidence of adverse events between sedentary and physically active HCM patients.⁶ These findings suggest that

'low risk' HCM patients are reasonably unlikely to experience adverse events during low to moderate intensity exercise. Therefore, it may be possible to determine eligibility for competitive exercise, taking into account risks and therapy, with assessments best carried out in specialised centres. The purpose of this report is to illustrate an original case of a young adult athlete, in good health and completely asymptomatic, who was excluded from competitive activity following the discovery of non-obstructive hypertrophic cardiomyopathy, thus denying him any possibility of engaging in physical activity, even at a recreational level. The aim of this article is therefore also to dispel the collective notion that a young athlete suffering from hypertrophic cardiomyopathy is a priori excluded from any physical activity.

Case report

In this report, the author describes the original clinical case of a 33-year-old male footballer, apparently in good health, completely asymptomatic and with a negative family history of cardiomyopathy. This athlete was withdrawn from competitive sport several years ago during a pre-participation screening because of resting ECG changes (i.e., biphasic and negative T waves on antero-lateral leads) suggestive of hypertrophic cardiomyopathy (HCM) (Figure 1).

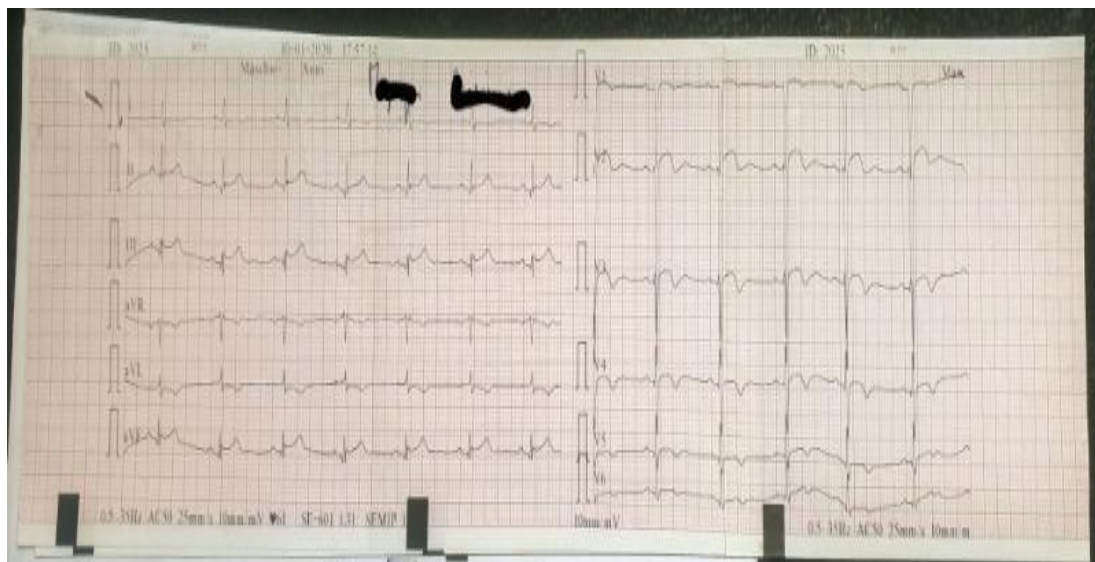


Figure 1 shows an old ECG (2020) with abnormal ventricular repolarisation in the antero-lateral leads, suggesting hypertrophic cardiomyopathy.

HCM was subsequently confirmed by echocardiography and more recently by magnetic resonance imaging (Figure 2, Videos 1-2).

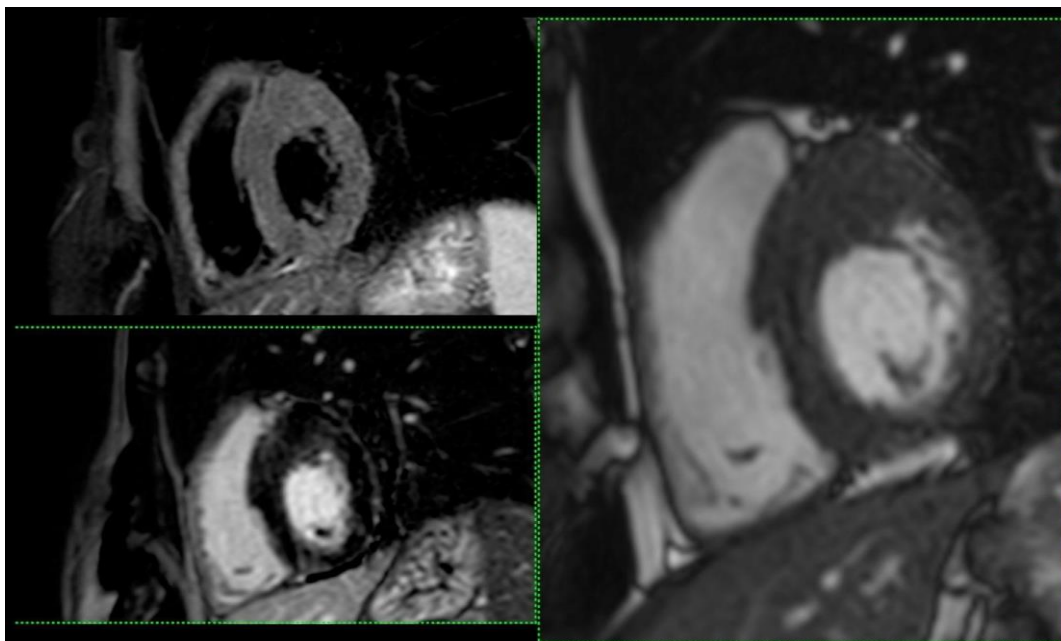


Figure 2 and Videos 1-2 show the cardiac magnetic resonance findings of asymmetric antero-basal septal hypertrophy without scar (LGE) and LVOT tract flow obstruction.



Case report HCM
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However, the genotype analysis was negative. In order to obtain at least a certificate for non-competitive sporting activities, the athlete contacted our centre for the appropriate assessments, which were finalised to obtain eligibility for physical activity. As already described, the family history was negative for cardiomyopathies, heart disease and sudden death. In addition, the athlete reported that he had never experienced symptoms

such as syncope, dyspnoea on exertion, chest pain or palpitations. The resting ECG (Figure 3) showed the presence of small Q waves in the infero-lateral leads with mild ST-segment elevation and deep S waves in the right precordial leads suggestive for septal LVH, while ventricular repolarisation was within normal limits.

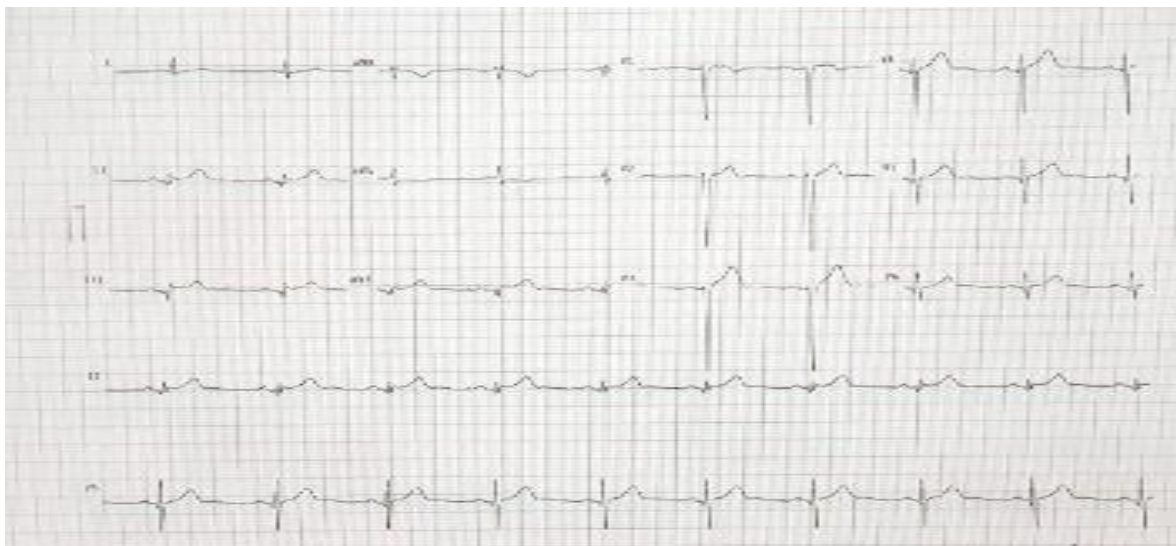


Figure 3 shows a recent ECG (2024) with the presence of small Q waves in the infero-lateral leads and deep S waves in the right precordial leads, suggesting marked septal hypertrophy.

A maximal exercise test was negative for signs of reduced coronary reserve and inducible arrhythmias. A 24-hour Holter ECG recording with exercise session showed no supraventricular or ventricular arrhythmias, and the rhythm remained consistently normal throughout the recording. A post-

exercise 2D TTE echocardiogram showed asymmetric hypertrophy of the left ventricle with marked thickening of the antero-basal septum at approximately 23 mm in the absence of outflow tract obstruction (Fig. 4), but with a slight increase in midventricular flow velocity.

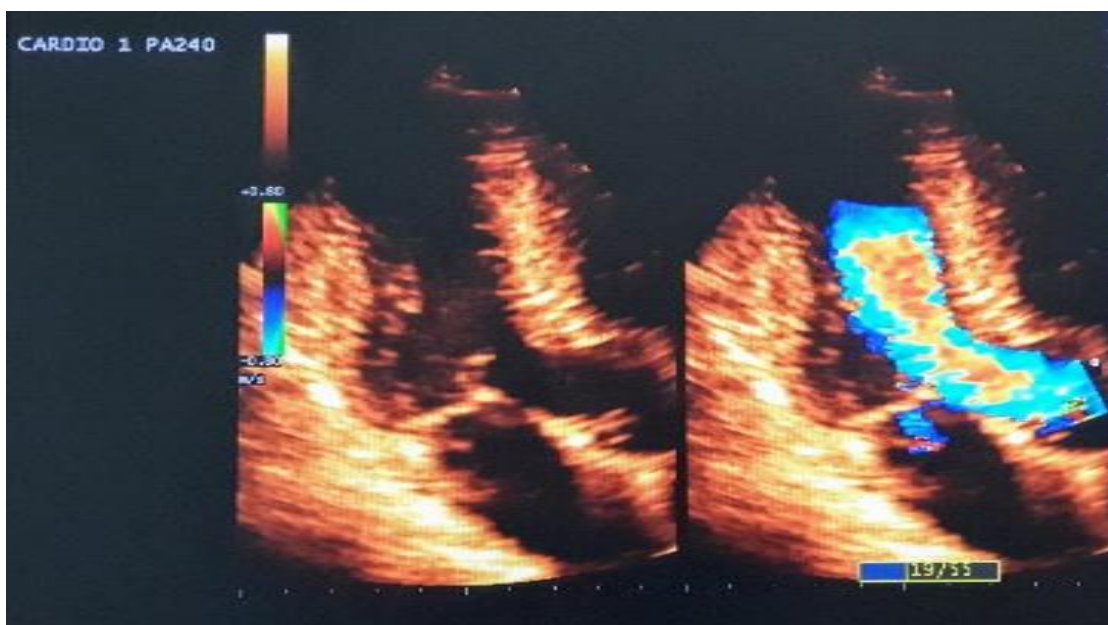


Figure 4 shows 2D TTE features of asymmetric left ventricular septal hypertrophy without LVOT flow obstruction.

Transvalvular gradients were within normal limits and good biventricular systolic and diastolic function was noted without valvular abnormalities. A subsequent stress echocardiogram confirmed the findings of the transthoracic echocardiogram without evidence of an increased LVOT gradient. In conclusion, in view of the clinical and instrumental evidence, and in particular the estimation of a low cardiovascular risk score, it was decided that this amateur athlete should undoubtedly be allowed to participate in recreational physical activity, as suggested by the latest guidelines and the COCIS 2023 recommendations.

Discussion

People with hypertrophic cardiomyopathy (HCM) have traditionally been advised to limit exercise and sports

participation to low-intensity activities because of concerns about sudden cardiac arrest (SCA). However, recent data have shown that SCA is rare in patients with HCM, and new data support the safety of exercise in this patient population. Recent guidelines support exercise in patients with HCM after comprehensive assessment and shared decision making with an expert provider.

The guidelines consider HCM patients to be 'low risk' if the following criteria are not met:

1. age under 30 years, as the disease is not considered stable;
2. history of cardiac arrest or sustained VT;
3. symptoms such as chest pain, near-syncope, syncope or recurrent/exercise-induced palpitations;
4. history of SCD in first-degree relatives before the age of 40 years;

5. moderate to severe LV hypertrophy;
6. Severe left atrial enlargement;
7. LV EF less than 50%;
8. LVOT obstruction (intraventricular gradient ≥ 30 mmHg at baseline, after Valsalva manoeuvre or during exercise testing);
9. Moderate to severe mitral regurgitation;
10. Exercise-induced myocardial ischaemia or abnormal BP response to exercise (less than a 20 mmHg increase at peak exercise);
11. Atrial fibrillation, paroxysmal supraventricular tachycardia, frequent and polymorphic PVBs, nonsustained VT (≥ 3 beats, >120 bpm);
12. QRS duration ≥ 120 ms (except after myomectomy);
13. Late enhancement $>15\%$ of myocardial mass on CMR (to be performed in specialised centres);

The HCM Risk-SCD estimates the risk of sudden cardiac death at 5 years in patients with hypertrophic cardiomyopathy. The risk of SCD at 5 years for an individual HCM patient can be calculated using the following equation Probability of Sudden Cardiac Death at 5 years = $1 - 0.998^{\text{exp (Prognostic Index)}}$ where Prognostic Index = $0.15939858 * \text{Maximum wall thickness (mm)} - 0.00294271 * \text{Maximum wall thickness}^2 \text{ (mm}^2) + 0.0259082 * \text{Left atrial diameter (mm)} + 0.00446131 * \text{Maximum left ventricular outflow tract gradient (mmHg)} + 0.4583082 * \text{Family history of SCD} + 0.82639195 * \text{NSVT} + 0.71650361 * \text{Unexplained syncope} - 0.01799934 * \text{Age at clinical evaluation (years)}$. According to ESC guidelines, an ICD is generally not indicated in patients with a 5-year risk of SCD $<4\%$, may be considered in patients with a risk of 4 to less than 6%, and should be considered in patients with a 5-year risk $\geq 6\%$.⁷⁻⁸ For these reasons, and in light of the growing evidence that carefully supervised exercise can be beneficial and safe in patients with HCM, it was a reasonable decision to allow this young athlete with non-obstructive HCM to engage in recreational exercise, which will certainly encourage open-minded physicians and sports cardiologists in general to consider abandoning exercise restrictions in HCM in selected cases, such as the one described in this clinical case.

Conclusion

The 2023 COCIS (Italian Cardiological Guidelines for Competitive Sport Eligibility in Athletes with Heart Disease: update 2024) recommendations for granting competitive sport eligibility to subjects with hypertrophic cardiomyopathy state that "adult subjects with a definite diagnosis of HCM, in the absence of risk markers, may participate in non-competitive

sports after a careful, strictly individualized assessment of the clinical stage, also taking into account the sport for which eligibility is sought, carried out in expert reference centers".

Conflicts of interest

The author has no competing interests to declare.

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