EDITORIAL

Distinguishing hypertrophic cardiomyopathy from athlete's heart: a clinical problem of increasing magnitude and significance

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As hypertrophic cardiomyopathy is a common cause of sudden deaths among athletes, differentiating this condition from the non-pathological "athlete's heart" presents an important challenge

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Sudden unexpected deaths of young trained athletes are highly visible events with substantial impact on the physician and lay communities.¹ Although a focal point first in the USA in the early 1980s,² this issue has now become one of increasing concern in many parts of the industrialised world (particularly Europe).^{3,4} The visibility accorded this medical issue is underscored by the highly publicised sudden deaths of several elite professional athletes, including Hank Gathers and Reggie Lewis in the USA, and more recently several athletes from Europe, South America, and Africa including Marc-Vivian Foe (Cameroon; soccer) who was known to have hypertrophic cardiomyopathy (HCM).⁵

IMPLICATIONS OF DIFFERENTIATING HEART DISEASE FROM ATHLETE'S HEART

Interest in athletic field deaths has also accelerated because of the recognition that these catastrophic events are probably more common than previously thought, occur in young people of both sexes and under-served minorities, and that the responsible but usually unsuspected structural cardiovascular diseases are often clinically identifiable—including by pre-participation screening.⁴ Furthermore, application of the implantable cardioverter-defibrillator to selected high risk patients has made diagnosis of these diseases, and disqualification from certain sports,^{1,6–8} particularly relevant.

The cardiac diseases that predispose athletes to lethal ventricular tachyarrhythmias have been documented in several necropsy based studies.^{1,3,9,10} In the USA, the single most common cause of athletic field deaths is HCM, accounting for about one third.^{1,3,9} Consequently, differential diagnosis between HCM and physiologic non-pathologic left ventricular (LV) hypertrophy associated with systematic training (commonly referred to as "athlete's heart") has become a not uncommon clinical dilemma.¹¹ This diagnostic distinction has important implications since identification of cardiovascular diseases associated with sudden death may be the basis for disqualification from competition to minimise

risk.⁸ Conversely, improper over-diagnosis of cardiac disease may lead to unnecessary withdrawal from athletics, thereby depriving that individual of the varied psychological and sometimes economic benefits of competitive sports. Interest in application of non-invasive markers that may resolve such differential diagnosis has increased with the greater visibility of athletic field deaths^{1–3,5} and focus on pre-participation screening in both the USA and Europe.^{3,5} Furthermore, legal liability incurred by evaluating competitive athletes with cardiovascular disease (presently most substantial in the USA) has raised the general level of anxiety surrounding these clinical situations.

STRATEGIES FOR DIFFERENTIAL DIAGNOSIS

An algorithm for differential diagnosis between HCM (with mild phenotypic expression) and extreme physiologic hypertrophy of athlete's heart has evolved, with particular relevance to trained athletes in whom left ventricular (LV) wall thickness falls into a "grey zone" of overlap between these clinical scenarios (fig 1)—that is, ventricular septal thickness of 13–15 mm present in 2% of highly trained male athletes.^{1,11} Of note, this clinical distinction is relevant only to non-obstructive HCM under resting (basal) conditions, in which mitral valve systolic anterior motion (SAM) is absent, since SAM is inconsistent with athlete's heart and strongly suggests HCM.

Genetic testing

The most definitive resolution of this important differential diagnosis can come from genetic testing. Indeed, a rapid genetic test is now available,¹² analysing by direct DNA sequencing mutations in the eight most common HCM causing genes. While a positive test result in an athlete can resolve the diagnostic ambiguity between athlete's heart and HCM, there is however significant potential for false negative test results in which a HCM diagnosis cannot be excluded.

Cavity dimension

In lieu of genetic testing, other echocardiographic parameters may be useful to distinguish between athlete's heart and HCM, including LV end diastolic cavity enlargement > 55 mm. LV

Abbreviations: HCM, hypertrophic cardiomyopathy; LV, left ventricular; SAM, systolic anterior motion

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IMAGES IN CARDIOLOGY

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Pitfalls in the echocardiographic diagnosis of isolated non-compaction of the ventricular myocardium

A 66 year old man presented with a one year history of atypical chest pain. The physical examination was normal, the ECG showed sinus rhythm and left anterior fascicular block, and the exercise test was normal. Two echocardiograms were performed, demonstrating an apical obliteration, which led to suspicion of endomyocardial fibrosis or apical tumour. Magnetic resonance imaging (MRI) was undertaken, which showed a typical pattern of an apical inner zone of non-compacted myocardium (NCM) distinguished from the thin outer zones of compacted myocardium (panels A, B). Late enhancement with gadolinium was also normal. The echocardiogram with second harmonic imaging was repeated (panel C) with a more detailed evaluation of the left ventricular apex, revealing trabeculations with intertrabecular recesses perfused from the left ventricular cavity (panel D). The left ventricle presented with mild dilation with mild globally depressed systolic function, and mild central mitral regurgitation. The pulsed wave tissue Doppler of septal mitral annulus showed an E' peak velocity of 5.9 cm/s, characterising impaired relaxation, and E/E' of 6.6, demonstrating a normal left ventricular end diastolic pressure. Screening showed one sister with the same echocardiographic pattern and one brother with two parallel false tendons extended between the interventricular septum and lateral wall.

Although echocardiography is the most frequently used first line diagnostic tool for NCM, sometimes the diagnosis is uncertain. Thus, in patients with a ventricular mass, low scale colour flow mapping demonstrates blood flow through deep recesses in continuity with the ventricular cavity. In addition, other imaging modality such as MRI, transoesophageal echocardiography, or contrast enhanced echocardiography may be performed to confirm the diagnosis.

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