Hypertrophic cardiomyopathy and athlete’s heart: a tale of two entities

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Sudden death during sports activities, although unfrequent, is a tragic event with great impact on both the general and medical communities. The two commonest conditions leading to sudden cardiac death in young athletes are hypertrophic cardiomyopathy (HCM), the main cause in the USA, and arrhythmogenic right ventricular cardiomyopathy, which is the leading cause in Europe.

We report the case of a 17-year-old football player with a pathological electrocardiography (ECG) in the pre-participation screening programme, highly suggestive of HCM, in which ECG study showed a septum thickness of 28 mm. Genetic analysis revealed R 495 W mutation in the 18 exon of the MyBPC3 (myosin-binding protein C) and sports activities were contraindicated. Two years later, septum thickness was 19.5 mm. Usefulness of 12-lead ECG, differential diagnosis between athlete’s heart and HCM, and the stratification in patients with HCM are discussed.

KEYWORDS
Hypertrophic cardiomyopathy; Athlete’s heart; Pre-participation screening programme

Introduction

Sudden death during sports activities, although unfrequent, is a dramatic event with great impact on both the general and medical communities. The two commonest conditions leading to sudden cardiac death in young athletes are hypertrophic cardiomyopathy (HCM), the main cause in the USA, and arrhythmogenic right ventricular cardiomyopathy, which is the leading cause in Europe.1,2

Hypertrophic cardiomyopathy is a primary and heterogeneous myocardial disease, with a prevalence in the population of 0.2%; numerous genetic, anatomic, and clinical variations of this cardiomyopathy have been described. It is mainly caused by mutations in genes encoding sarcomeric contractile proteins and is characterized by an autosomal-dominant mode of inheritance. Once diagnosed, it is obligatory to advise against competitive sports activities, but sometimes things are not so easy and there are some open questions and controversies about this topic. We have the international complex debate about the pre-participation screening programme, differences between European and American recommendations are well known, and the usefulness of 12-lead electrocardiography (ECG) is the main point of controversy. On the other hand, differentiating HCM from a non-pathological athlete’s heart is sometimes difficult and can be a diagnostic and personal dilemma. Echocardiography plays an important role in the differential diagnosis: the magnitude and distribution of thickening of the left ventricle wall, the dimension of the left ventricular cavity, and the use of Doppler myocardial imaging in the evaluation on myocardial systolic and diastolic function are important ECG features.3–6

Of course, ECG abnormalities such as prominent Q waves, a marked increase in voltages or deep negative T waves, and, finally, the persistence of hypertrophy after 6 months of detraining can help to resolve the dilemma. In the case we report, although it was clearly an HCM there was also a response to deconditioning.

Case report

A 17-year-old football player with a regular intensive training (14 h/week), asymptomatic and without familial history of sudden death or cardiomyopathy, with a pathological ECG (left ventricle hypertrophy and T wave inversion in precordial leads) highly suggestive of HCM, was submitted for ECG analysis. The ECG was practised as a part of the pre-participation screening programme.

Echocardiogram (July 2005) revealed a septal thickness of 28 mm (Figure 1) without systolic anterior motion of the...
mitral valve or outflow tract obstruction, left atrial and left ventricle diameters were normal. Genetic analysis revealed R 495 W mutation in the 18 exon of the MyBPC3 (myosin-binding protein C) gene (Arg 495 Trp) which was also present in his mother (48 years old) and grandfather (81 years old), this one had a septum thickness on 16 mm in the echocardiogram while mother’s echo was normal. Diagnosis of HCM was made and he was advised against sports activities. Holter monitoring and exercise stress test were normal. During follow-up, ECG studies revealed a progressive decrease in the septal wall thickness, 2 years later (August 2007), septal thickness was of 19.5 mm (Figure 2). There were not ECG changes. He is still under follow-up.

Discussion

This case represents an example of the complex interactions between genetics and environmental factors. It is known that patients with MyBPC3 mutations present a broad range of phenotypes and some of them are characterized by a benign clinical course and a delayed onset of the disease (age-related penetrance). However, in our football player, the physiological adaptations secondary to intensive physical training were magnified by a positive genetic substrate, which lead to a severe and partially reversible hypertrophy at a young age. Detraining for 2 years resulted in regression of left ventricular hypertrophy suggesting that, even in individuals with HCM, the pre-load and after-load stresses associated with exercise are a contributing factor to left ventricular hypertrophy. On the other hand, intense sports activities could be responsible of an earlier development of left ventricular hypertrophy in this young football player carrier of a MyBPC3 mutation. This case may be a reflect of the role of different factors in regulating genotype-phenotype correlations and clinical expressions in each individual patient.

About the recommendation against sports activities, Maron and Klues previously published a group of 14 patients with diagnosis of HCM (left ventricular wall thickness 18–28 mm) who competed actively in various sports and maintained high levels of achievement during years without symptoms, disease progression, or sudden death. However, and although our patient has a considered benign mutation with normal stratification we think that there are not proven risk algorithms that can safely predict low risk in all affected individuals, and recommendations of the ESC and AHA guidelines must be follow-up.

Finally, we firmly think that 12-lead ECG must be a part of the pre-participation screening programme, as it is not only useful for identifying HCM but it also enables detection of other potentially lethal conditions such as WPW, long QT syndrome or Brugada’s disease. About the differential diagnosis...
between HCM and athlete’s heart sometimes it is not easy, but, both ECG and genetic analysis, when possible, are two useful tools for solving the dilemma, and of course, detraining during weeks can resolve the problem.\(^{10,11}\)

References