Sudden death from cardiovascular disease in young athletes: fact or fiction?

S Sharma, G Whyte, W J McKenna

Introduction
The cardiovascular benefits of exercise are well established and epidemiological studies suggest that long term exercise programmes reduce the development of atheroma and the risk of sudden cardiac death (SCD).1-3 It is well recognised that a very small but significant proportion of athletes may die suddenly. The precise incidence of SCD in young athletes is unknown but estimates from the United States suggest that it is very low. Such tragedies are highly publicised, particularly when high profile athletes are involved, causing major concern in the general community who perceive athletes as the most healthy segment of society.4

The sudden and unexpected death of athletes has evoked considerable interest among doctors and pathologists and led to the detection of a wide number of cardiovascular disorders causing sudden death in this population.4-17 Most non-traumatic sudden deaths in young athletes are due to inherited structural and functional cardiovascular abnormalities. Hypertrophic cardiomyopathy (HCM) accounts for 40-50% of all such deaths. Other well recognised causes include arrhythmogenic right ventricular cardiomyopathy, congenital anomalous coronary arteries, premature coronary artery disease, Wolff-Parkinson-White syndrome, long QT syndrome, myocardiitis, and Marfan’s syndrome (table 1). Early identification of affected individuals should prevent sudden death. There is controversy about the extent of the problem, the significance of cardiovascular abnormalities detected at postmortem examination in athletes, and whether or not exercise related sudden death in apparently healthy individuals should be a cause for alarm in the sporting community.18

Definitions
Before proceeding further, it is prudent to define the terms “young”, “competitive athlete”, and “sudden cardiac death”. Sporting individuals aged below 25 years will be classified as young. Although a substantial proportion of the population consider themselves to be athletes, we consider a competitive athlete as an individual who participates in an organised team or individual sport in which regular competition is a component.19 SCD is defined as an event that is non-traumatic, non-violent, unexpected, and resulting from sudden cardiac arrest within six hours of previously witnessed normal health.

SCD in young athletes is thought to be uncommon, but precise data on its exact incidence are lacking. Estimates from the United States suggest an incidence of 1 in 200 000.20-22 This is unlikely to represent the true incidence of SCD for several reasons. When faced with SCD in a young athlete, the coroner’s mandate is to exclude foul play rather than provide the cause of death. An expert pathologist with experience of conditions causing SCD is rarely responsible for carrying out postmortem examination, thus subtle conditions may not be detected. The cause of death is difficult to ascertain in conditions predisposing to fatal cardiac arrhythmias, and SCD in the absence of structural cardiac abnormalities, for example, the long QT syndrome. Furthermore, the evident lack of a systematic national registry for SCD in athletes means that the compilation of statistics on SCD relies heavily upon reports from the media, which usually concentrate on the most elite athletes rather than the general young athletic population, and on voluntary referrals from hospital based pathology registries. Such crude methods of data collection undoubtedly lead to a significant underestimation of exercise related SCDs.

Some 80% of non-traumatic sudden deaths in young competitive athletes are due to inherited/congenital structural or functional cardiovascular abnormalities, most of which provide a pathological substrate for fatal cardiac arrhythmias predisposing to sudden death. A proportion of affected athletes are capable of incredibly high levels of performance, and reports on the sudden deaths of athletes have shown that many had competed at intercollegiate, professional, and national level (table 2).

The majority of deaths occur during or immediately after strenuous physical activity, suggesting that exercise is a strong trigger factor for cardiac arrhythmias in athletes harbouring potentially lethal cardiac conditions. Most deaths occur in men. This may be due to a higher participation rate and competition in
Table 1 Causes of sudden cardiac death in young competitive athletes

<table>
<thead>
<tr>
<th>Common</th>
<th>Uncommon</th>
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<tbody>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>Myocarditis</td>
</tr>
<tr>
<td>Idiopathic concentric left ventricular hypertrophy</td>
<td>Premature coronary artery disease</td>
</tr>
<tr>
<td>Coronary artery anomalies</td>
<td>Long QT syndrome</td>
</tr>
<tr>
<td>Arrhythmogenic right ventricular cardiomyopathy*</td>
<td>Marfan’s syndrome (aortic rupture)</td>
</tr>
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<td>*Commonest cause of death in northern Italy.</td>
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Table 2 Prevalence of inherited or congenital cardiovascular disorders associated with sudden death

<table>
<thead>
<tr>
<th>Condition</th>
<th>Estimated prevalence</th>
<th>Estimated number affected in the United Kingdom</th>
</tr>
</thead>
<tbody>
<tr>
<td>HCM</td>
<td>1/500</td>
<td>100 000</td>
</tr>
<tr>
<td>ARVC</td>
<td>1/10 000</td>
<td>5000</td>
</tr>
<tr>
<td>Wolff-Parkinson-White syndrome</td>
<td>1.5/1000</td>
<td>75 000</td>
</tr>
<tr>
<td>Long QT syndrome</td>
<td>1/10 000</td>
<td>5000</td>
</tr>
<tr>
<td>CAA</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>Mitral valve prolapse</td>
<td>2/100</td>
<td>1 000 000</td>
</tr>
<tr>
<td>Marfan’s syndrome</td>
<td>1/5000</td>
<td>10 000</td>
</tr>
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</table>

more rigorous sports compared with women. Most athletes are allegedly asymptomatic before death. In a recent review of SCD in 157 young athletes (mean age 17) in the United States between 1985 and 1995, 90% died during or immediately after a training session and only 18% had prodromal symptoms thought to be cardiovascular in origin in the preceding 36 months.31 The lack of prodromal symptoms may be due to under-reporting on the part of the victim through fear of being perceived as “unfit” by sporting bodies, or, more importantly, to false reassurance as the result of the triviality attached to potentially serious symptoms by medical practitioners who perceive athletes to be extremely healthy. An apparently healthy young athlete presenting with chest pain or syncope during exertion is often inappropriately reassured rather than investigated.

The commonest cause of SCD in young athletes is HCM which accounts for between 40 and 50% of deaths.1,1,21 Other important causes include arrhythmogenic right ventricular cardiomyopathy, anomalous coronary arteries, Wolff-Parkinson-White syndrome, long QT syndrome, Marfan’s syndrome, mitral valve prolapse, myocarditis, and premature coronary artery disease. Premature coronary artery disease is the most predominant cause of death in older athletes (see box 1).24,25

**Hypertrophic cardiomyopathy**

HCM is defined as ventricular hypertrophy in the absence of a cardiac or systemic cause.26 It is inherited as an autosomal dominant trait with a high degree of penetrance. Missense mutations within genes encoding four different cardiac sarcomeric contractile proteins are responsible for the disease. These include β myosin heavy chain (30%), cardiac troponin T (15–20%), α troponin (2–5%), and myosin binding protein C (10–15%).27 The most common pattern of hypertrophy is asymmetric septal but almost any pattern is possible.28,29 Left ventricular outflow tract obstruction secondary to systolic anterior motion of the mitral valve apparatus is present in 20%. The histological hallmark of the disease is myocyte and myofibrillar disarray, which contributes to a variety of pathophysiological disturbances and electrical instability.30,31 The prevalence of the disease in young adults is 0.2%, which means there are approximately 100 000 affected individuals in the United Kingdom.32

Chest pain and dyspnoea are the commonest symptoms of HCM. Other symptoms include palpitation, dizziness on exertion, and syncope. The majority of young patients are asymptomatic or have minor symptoms.33,34 Physical examination is helpful in those individuals with left ventricular outflow tract obstruction for which palpation of the carotid artery may show up a jerky pulse, and palpation and auscultation of the precordium may detect a double apical impulse and a harsh ejection systolic murmur respectively (see box 2).

Sudden death is often the first presentation. Data from referral institutions show a death rate of about 2.5% per annum, reaching a maximum of 6% during childhood and adolescence.35 The exact death rate among young athletes is not known but is probably significantly higher than in the non-exercising population. In a study comparing sports related and non-sports related SCD in young athletes, HCM was a much more common cause of death in sports related deaths than in non-sports related deaths: eight of 34 deaths compared with 20 of 656 (P = 0.00019).36 The highly significant increase in exercise related sudden death from HCM in young athletes suggests that the propensity to life threatening supraventricular and ventricular arrhythmias resulting from myocardial disarray may be triggered by the stress of exercise.

The antagonist for HCM being an important cause of SCD may argue that left ventricular hypertrophy is common in athletes participating in regular endurance sport, and there is a possibility that postmortem demonstration of

**Box 1**

SCD in athletes
- SCD in athletes is uncommon.
- 80% of non-traumatic sudden deaths are attributed to inherited or congenital cardiovascular disorders.
- 40–50% of all deaths are from HCM.
- The majority of deaths occur during or immediately after physical activity.
- Prodromal symptoms of cardiovascular disease may be absent.

**Box 2**

Symptoms of cardiovascular disease
- Chest pain on exertion
- Disproportionate breathlessness in relation to the exercise being performed.
- Dizziness on exertion.
- Syncopal episodes during or after exertion.
- Palpitations.
ventricular hypertrophy is often falsely attributed to HCM rather than physiological adaptation to exercise. However, physiological hypertrophy is usually mild, and left ventricular hypertrophy exceeding 13 mm is rare even in the most elite athletes. Major publications documenting HCM as the cause of death have based their findings on the demonstration of gross left ventricular hypertrophy with a heart weight usually exceeding 500 g or a maximal left ventricular wall thickness of 20 mm in the absence of a dilated ventricular cavity, together with histological evidence of extensive myocardial disarray and scarring. To date there is a conspicuous lack of reputable reports on athletes with a ventricular wall thickness exceeding 16 mm or left ventricular mass exceeding 400 g.

Patients with HCM have extensive disarray with about 20% of the myocardium affected. There are reports on SCD in athletes where postmortem examination has shown concentric left ventricular hypertrophy with a wall thickness range of 15–19 mm, a non-dilated left ventricular cavity, and a modest increase in left ventricular mass with heart weights between 400 and 490 g. Histological examination of the myocardium has failed to detect myocardial disarray, and the cause of SCD in this group has been classified under a separate entity termed “idiopathic concentric ventricular hypertrophy” even though the macroscopic findings are compatible with the diagnosis of HCM. Most affected athletes have been black, and it is possible that these changes may be an exaggerated race related response to undetected hypertension during life. Alternatively, the protagonists for HCM causing SCD in athletes may argue that these cases represent mild or atypical forms of HCM. The latter is in keeping with the fact that HCM is a significantly more common cause of SCD in young black athletes.

The genetic heterogeneity of HCM currently makes it impractical to perform genetic testing on all individuals suspected of having HCM. Two dimensional echocardiography and Doppler studies continue to serve as the gold standard test for the diagnosis of HCM. The demonstration of left ventricular hypertrophy with a wall thickness exceeding 15 mm and a small left ventricular cavity is usually diagnostic. Additional features such as systolic anterior motion of the mitral valve, redundant mitral valve leaflets, a gradient across the left ventricular outflow tract, and Doppler evidence of abnormal ventricular filling may also be present.

The autosomal dominant inheritance of HCM makes it prudent that all first degree relatives of patients with HCM are screened for the condition with echocardiography. Thus athletes with affected first degree relatives should be offered screening. The early diagnosis of HCM in competitive athletes should reduce the death rate by preventing affected individuals from participating in vigorous training and further competition and, where appropriate, undergoing treatment to prevent sudden death.

**Differentiation of the normal athlete’s heart from the condition of HCM with non-invasive cardiac tests**

Regular physical training induces structural and functional cardiac changes as part of a normal adaptive physiological process which may simulate cardiac pathology. The structural features of an athlete’s heart include a 10% increase in left ventricular cavity, 10–20% increase in left ventricular wall thickness, and a 45% increase in left ventricular mass. Although cavity dilatation and hypertrophy are common, absolute cardiac dimensions are usually within normal limits irrespective of body surface area. In a few cases, left ventricular hypertrophy is quite pronounced, and may raise the differential diagnosis of HCM. This is highlighted by a recent echocardiographic study involving 947 elite Italian athletes in which about 2% had left ventricular hypertrophy of between 13 and 15 mm. The distinction between the athlete’s heart and HCM in this rare situation has important implications because identification of HCM in an athlete may be the basis of disqualification from competition to minimise the risk of sudden death. An erroneous diagnosis of HCM in an athlete may lead to unnecessary withdrawal from sport with profound consequences to the sportsman in terms of physical, financial, and psychological well being.

A large number of studies on elite athletes have led to proposed echocardiographic criteria for the differentiation of physiological hypertrophy from HCM (table 3). However, there are cases where echocardiography cannot reliably differentiate physiological from pathological hypertrophy, and, if undue

<table>
<thead>
<tr>
<th>Athlete’s heart</th>
<th>HCM</th>
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<tbody>
<tr>
<td>Left atrial dimension</td>
<td>Common</td>
</tr>
<tr>
<td>LVH (mm)</td>
<td>&lt; 16</td>
</tr>
<tr>
<td>Patter of LVH</td>
<td>Concentric</td>
</tr>
<tr>
<td>LV cavity size in diastole (mm)</td>
<td>&gt; 55</td>
</tr>
<tr>
<td>Indices of diastolic function</td>
<td>Enhanced</td>
</tr>
<tr>
<td>ECG patterns</td>
<td>Large complexes. Voltage criteria LVH</td>
</tr>
<tr>
<td></td>
<td>(Sokolow) in the absence of deep T wave</td>
</tr>
<tr>
<td></td>
<td>inversion in left precordial leads</td>
</tr>
<tr>
<td></td>
<td>ST elevation or ST elevation. Q waves</td>
</tr>
<tr>
<td></td>
<td>Right axis deviation</td>
</tr>
<tr>
<td></td>
<td>RV conduction delay</td>
</tr>
<tr>
<td></td>
<td>Unusual</td>
</tr>
<tr>
<td></td>
<td>&gt; 16</td>
</tr>
<tr>
<td></td>
<td>Heterogeneous distribution</td>
</tr>
<tr>
<td></td>
<td>&lt; 45</td>
</tr>
<tr>
<td></td>
<td>Reduced</td>
</tr>
<tr>
<td></td>
<td>Large complexes. Voltage criteria LVH and deep T wave inversion in left precordial leads</td>
</tr>
<tr>
<td></td>
<td>Leftward axis</td>
</tr>
<tr>
<td></td>
<td>Partial LBBB</td>
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LV, left ventricular; LVH, left ventricular hypertrophy; LBBB, left bundle branch block. Sokolow criterion for LVH is present when the sum of RV1 and SV6 > 35mV.
reliance is placed on this investigation alone, then there is a risk of generating false positives and false negatives. Additional information from family history and the 12 lead electrocardiogram may be helpful in these circumstances. The demonstration of HCM in a first degree relative of an athlete with left ventricular hypertrophy will increase the possibility of the diagnosis of HCM. Voltage criteria of left ventricular hypertrophy and ST segment repolarisation changes on the electrocardiogram (ECG) are common in both conditions; however, the presence of prominent Q waves, left bundle branch block, and deep T wave inversion favours the diagnosis of HCM.

Despite the ECG criteria, there are cases where differentiation of the normal athlete’s heart from HCM remains difficult. A definitive method of resolving the diagnosis is demonstration of regression of ventricular hypertrophy after a three month period of detraining, a feature characteristic of physiological but not pathological hypertrophy. This is usually unacceptable to the athlete striving to achieve honours in competitive sport because it hinders fitness and team selection, but may be feasible during injury or in the off season. Future developments in molecular genetics should allow DNA testing in all individuals with suspected HCM. In the meantime, the development of more specific and sensitive cardiac tests for differentiating between physiological and pathological hypertrophy may obviate the need to detrain.

Arrhythmogenic right ventricular cardiomyopathy (ARVC)
ARVC is a heart muscle disorder characterised by patchy fibrofatty replacement of right ventricular myocardium. The replacement of myocardium by fatty tissue predisposes to inhomogeneous and fatal re-entrant ventricular arrhythmias. It is the commonest cause of exercise related SCD in young athletes in the Veneto region of northern Italy. The strong association with strenuous physical activity incriminates exercise as a trigger factor for arrhythmias. The exact incidence and prevalence are unknown, but the prevalence is estimated to be 1 in 10 000, which means there are potentially 5000 affected individuals in the UK. In about 30–50% of cases the condition is familial, the commonest pattern of inheritance being autosomal dominant although an autosomal recessive pattern has also been described. The main clinical features are palpitation, dyspnoea, presyncope, syncope, and sudden death. The results of physical examination are usually normal. The definitive diagnosis of ARVC relies on histological demonstration of transmural fibrofatty replacement of ventricular myocardium. This is difficult in most living patients and not without risk of perforation. Diagnosis based on endomyocardial biopsy may generate false negatives because of the segmental nature of the disease. In addition, the potential of errors in histological interpretation means that an expert pathologist with ample experience in ARVC histology is generally necessary. The limitations of histological diagnosis means that ante mortem diagnosis usually relies on the combination of cardiovascular symptoms, positive family history of ARVC, electrocardiographic evidence of T wave inversion in the right precordial leads, multiple ventricular tachycardias or ventricular tachycardia of left bundle branch block morphology, delayed potentials on the signal averaged ECG, and demonstration of abnormalities of the right ventricle using cardiac imaging techniques.

The diagnosis of ARVC in a competitive athlete is an indication for disqualification from competitive sport to prevent sudden death. Individuals with persistence of life threatening ventricular arrhythmias are managed with anti-arrhythmic medication or by implantation of an automatic cardioverter defibrillator.

Differentiation of the normal athlete’s heart from the condition of ARVC with non-invasive cardiac tests
Some highly trained athletes may show right ventricular enlargement and a variety of repolarisation ECG changes in the right precordial leads, raising the differential diagnosis of ARVC. In this situation the demonstration of right ventricular aneurysms, hypokinetic segments or gross dilatation would support the diagnosis of ARVC, whereas the presence of mild global enlargement together with a slightly dilated hypertrophied left ventricle with good systolic function would be more in keeping with the athlete’s heart.

Congenital coronary artery anomalies (CAAs)
Sudden death can complicate anomalous insertion of the coronary arteries, particularly when the left coronary artery originates from the anterior sinus of Valsalva. In a review of 51 cases of CAAs there were nine sudden deaths. All deaths occurred either during or just after physical activity. In all nine cases the left coronary artery arose from the anterior sinus of Valsalva. The acute leftward passage of the coronary artery along the aortic wall is thought to cause the entrance into the left coronary system to be slit-like. During exercise there is stretching of the left coronary artery and a flap-like closure of the orifice of the left coronary artery causing myocardial ischaemia. There were no deaths when both coronaries arose from the left coronary sinus of Valsalva in this study, but there have been reports of SCD in this type of CAA. Before these reports, CAAs were thought to be minor and associated with a normal life expectancy. CAA is now recognised as the second most common cause of SCD in young athletes in the USA. Most affected patients are asymptomatic, but some may present with exercise induced chest pain, fatigue, and collapse. Exercise testing rarely shows up inducible myocardial ischaemia. The visualisation of the left main stem and right coronary artery is important in the evaluation of all symptoms of athletes suggestive of a cardiovascular abnormality. This is achieved by non-invasive means on the short axis views of the aortic root on two dimensional
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Premature coronary artery disease

Coronary atherosclerosis is a rare cause of SCD in young athletes. Almost all individuals have a family history of premature coronary artery disease from hypercholesterolemia. SCD is almost always the first presentation. Prodromal symptoms of myocardial ischaemia are rare and most individuals are free from established risk factors for coronary artery disease. This is in contrast with older athletes in whom prodromal symptoms are common, and risk factors are identified in approximately 50%. ECG and exercise testing performed on young athletes with premature coronary artery disease before death have often failed to show evidence of myocardial ischaemia.

Death is usually from single vessel disease, predominantly affecting the left anterior descending artery. The macroscopic findings show obstructive atheromatous lesions with acute thrombosis superimposed on a ruptured atheromatous plaque.

Wolf-Parkinson-White syndrome

Wolf-Parkinson-White syndrome is characterised by an accessory conduction pathway with a predilection to re-entrant supraventricular tachyarrhythmias which may precipitate ventricular fibrillation. It is reported as a rare cause of sudden death in athletes. This may be due to failure to identify or appreciate abnormalities of cardiac conduction tissue disease at postmortem examination. Affected patients may present with rapid palpitation, presyncope, or syncope. Wolff-Parkinson-White syndrome can be identified by the presence of delta waves and a short PR interval on the ECG. Exercise testing is useful for predicting the risk of fatal arrhythmias. High risk pathways should be investigated further by electrophysiological studies. Radiofrequency ablation of the accessory is the therapeutic technique most widely used and is particularly attractive for athletes in whom antiarrhythmic agents prove ineffective because of the high sympathetic drive.

Long QT syndrome

The long QT syndrome is a rare familial disorder characterised by prolongation of the QT interval (>440 milliseconds; corrected) on the ECG, and the propensity to syncope and fatal ventricular arrhythmias. The prevalence of the disorder is between 1/10 000 and 1/15 000. Both autosomal dominant and autosomal recessive modes of inheritance are recognised. In 10% of cases it is a result of de novo genetic mutations. Mutations on at least four gene loci located on chromosomes 3, 4, 7, and 11 have been identified. The mutations result in defective sodium or potassium channels within cardiac myocytes and are the basis of fatal cardiac rhythm disturbances. The clinical course is variable, ranging from malignant ventricular arrhythmias with recurrent syncope and sudden death in some patients to an asymptomatic course throughout life. Syncope is usually due to polymorphic ventricular tachycardia, which, in most patients, is initiated by acute autonomic change precipitated by fear, sudden auditory stimuli, intense emotion, and physical exertion. Patients at high risk are those with a history of syncope and a strong family history of syncope and premature sudden death. Cardiac morphology is normal, and therefore postmortem identification is not usually possible in the absence of ante mortem ECG information or a positive family history of the condition.

Affected individuals can be identified by ECG. Twenty four hour ambulatory ECG recordings and exercise testing may be helpful in evaluating the seriousness of the disorder. Recent advances in the molecular genetics of the long QT syndrome should prove helpful in the preclinical diagnosis of young patients at risk and in the diagnosis of equivocal or sporadic cases. Avoidance of vigorous physical activity and therapy with β blockers may prevent sudden death in athletes who are identified early.

Myocarditis

Myocarditis is a rare cause of exercise related SCD in young athletes. Viruses and occasionally drugs have been implicated as the cause. Most deaths are related to strenuous physical activity. Myocardial inflammation with lymphocytic infiltration and focal necrosis shown at postmortem examination may be the substrate for exercise induced arrhythmias. Symptoms are non-specific, with coryzal symptoms or a mild febrile illness preceding the clinical picture. Specific signs include tachycardia, evidence of cardiomegaly, and rarely cardiac failure. The ECG information is non-specific but may show cardiac arrhythmias. In severe cases, there may be evidence of ventricular dilatation and impairment at echocardiography, but there are cases where the ECG is entirely normal and therefore a high index of suspicion is required if deaths caused by myocarditis are to be prevented. All athletes with coryzal symptoms or a febrile illness should be rested until completely asymptomatic.

Marfan’s syndrome

Marfan’s syndrome is an inherited connective tissue disorder characterised by skeletal, ocular, and cardiac abnormalities. The gene is located in the long arm of chromosome 15 and encodes the glycoprotein fibrillin (chromosome 15). The prevalence based on clinical criteria is 1 in 5000. Affected patients are excessively tall, have chest wall abnormalities, kyphoscoliosis, arachnodactyly, and high arched palates. Lens dislocation is common. Cardiac involvement is almost always characterised by mitral valve prolapse with or without mitral regurgitation. Aortic root dilatation is a potentially serious cardiac manifestation, and sudden death is usually from aortic dissection or rupture. There may be a family history of premature sudden death. Physical examination should raise suspicion of Marfan’s syndrome. Patients should be referred for assessment of...
the aortic root by echocardiography. Prophylactic surgery is indicated once the aortic root diameter exceeds 6 cm. Abstinence from competitive sport and treatment with β blockers may retard aortic dilatation and prevent death from aortic rupture.

**Mitral valve prolapse**

Mitral valve prolapse is extremely common in the general population. It is thought to affect up to 5% of the population. The condition is generally benign, but there are 60 cases of SCD in the medical literature thought to be associated with mitral valve prolapse. Only four deaths occurred in young individuals, and in only three of the 60 cases was death related to exercise. Symptoms include atrial systole, infra-mammary chest pain, fatigue, dizziness, and occasionally syncope. Mitral valve prolapse can be clinically detected by auscultation and confirmed by echocardiography. Controversy remains about its association with SCD. The detection of mitral valve prolapse after sudden death may be coincidental rather than causal, or it may be due to its association with Wolff-Parkinson-White syndrome or long QT syndrome. The approach to mitral valve prolapse in relation to participation in competitive sport should be pragmatic. In the absence of symptoms and signs and a positive family history of sudden death, full participation is allowed. It would be reasonable to disqualify athletes with a history of syncope, disabling chest pain, complex ventricular arrhythmias, significant mitral regurgitation, prolonged QT interval, Marfan’s syndrome, and a family history of sudden death.

**Screening and prevention of sudden death**

The incidence of cardiovascular conditions predisposing to SCD is low. The ultimate question remains whether all cases of SCD could be prevented if every competitive athlete is screened for cardiovascular disease. The Italian experience is of interest in this respect because all young athletes require an annual certificate of fitness based on a normal physical examination, ECG, and a limited exercise test. The systematic evaluation of SCD victims in northern Italy shows that HCM is a very rare cause of SCD in young athletes. This contrasts with published reports from other countries showing that HCM is by far the commonest cause of SCD. One interpretation is that individuals with HCM are identified early and excluded from sporting activities by the requirements of the Italian fitness certificate. Similarly the potential of detecting Wolff-Parkinson-White syndrome and long QT syndrome on ECG during such fitness screens may reduce the incidence of sudden death from these conditions.

The financial constraints of public health services mean that large scale screening programmes are not possible; a substantial number of athletes would need to be screened to detect a small proportion at risk of SCD. This should not deter major independent financially endowed sporting bodies from protecting their athletes by providing funds for cardiovascular screening because a significant proportion of deaths occur in the absence of symptoms. Where cost is a major issue, raising public awareness of the risk of SCD in athletes with emphasis on the warning features may be more practical. Many athletes at risk may not present with warning features, but many will, and greater sensitivity and awareness within the athletic community should improve identification of those athletes who are at risk of sudden death. Athletes with a family history of premature sudden death or symptoms of palpitation, chest pain, disproportionate breathlessness in relation to the intensity of the exercise being performed, dizziness, or syncope should undergo evaluation in a specialist cardiology centre to exclude underlying cardiovascular disease. Initial investigation with ECG will detect Wolff-Parkinson-White syndrome and long QT syndrome.

Echocardiography with particular reference to left ventricular wall thickness, right ventricular architecture and function, aortic root dimension and demonstration of the origins of

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**Box 3: Cardiovascular assessment of an athlete**

- Screening for cardiovascular disease is warranted in athletes with symptoms of cardiovascular origin and a family history of premature (<40 years) non-traumatic sudden death.
- Initial screening should involve thorough cardiovascular examination with particular reference to blood pressure and cardiac murmurs, 12 lead ECG and two dimensional echocardiography.
- Physical examination will detect forms of HCM with obstruction (20–25%), valvular disorders, and Marfan’s syndrome with or without cardiovascular involvement.
- The ECG is abnormal but not diagnostic in almost all patients with HCM and 90% of patients with ARVC.
- The presence of a long QT interval or accessory pathway on the 12 lead ECG should raise the suspicion of long QT syndrome and Wolff-Parkinson-White syndrome respectively and should prompt referral to a specialist cardiology unit for further evaluation with exercise testing and electrophysiological studies.
- Echocardiography with particular reference to left ventricular thickness, right ventricular architecture and function, aortic root dimension, and demonstration of the origins of the coronary arteries will detect all HCM, Marfan’s syndrome, mitral valve prolapse and a large proportion of ARVC and CAA.
- Further invasive diagnostic investigations may be warranted in some individuals.
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the coronary arteries will detect all HCM, Marfan’s syndrome, mitral valve prolapse and a large proportion of ARVC and CAA. Further invasive diagnostic investigations may be warranted in some individuals (see box 3).

Conclusion

SCD in athletes is rare, but the true incidence of the problem is not known. Most deaths are due to HCM. SCD from HCM is more common in athletes than in sedentary individuals. Exercise is a recognised trigger of fatal arrhythmias culminating in sudden death. There have been claims that ventricular hypertrophy from physiological adaptation to exercise may be misinterpreted as HCM when detected at postmortem examination; however, the diagnosis of HCM in athletes that die suddenly has been based on the demonstration of gross ventricular hypertrophy, massive left ventricular mass, and histological evidence of widespread myocardial disarray, fibrosis, and scarring, none of which are characteristic of physiological hypertrophy. In addition to HCM, there are several other recognised cardiovascular abnormalities that predispose to sudden death in athletes. Early identification of all causes of SCD in young athletes should help to prevent death by allowing the recommendation of abstention from vigorous exercise and, if appropriate, initiation of medical therapy. Individuals with symptoms suggestive of cardiovascular disease or those with a family history of premature sudden death should undergo thorough cardiovascular evaluation at a specialist centre.

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