The sudden cardiac-related death of an athlete during a sports event is a particularly poignant tragedy. Participation in sports—a wholesome pursuit that provides important physical and psychosocial benefits—is not expected to place young, apparently healthy competitors at risk for cardiovascular collapse. When sudden deaths occur in elite-level athletes who have national media exposure, the concerns become magnified. How safe is sports play? What causes these tragic events? How can they be prevented?

In addressing these questions, particular attention has often been focused on health-care providers, who, it has been assumed, should be able to identify the young athlete at risk and thus prevent sudden cardiac death in sports. It is probably true that unexpected cardiac-related deaths occur only in athletes who have some underlying heart disease or dysfunction (i.e., it is unlikely that a normal heart can be “worked to death”). With their extensive diagnostic tools, it seems that health-care providers should be able to detect athletes who are afflicted by such conditions, restrict their sports competition, and thereby diminish or eliminate the risk of sudden death during sports participation.

In accord with this strategy, national groups have been organized to formulate and publish guidelines for detection and prohibition of athletes at risk from further participation in sports (Maron et al., 1994; 1996b). It is clear, however, that not all underlying disease can be detected by preparticipation screening. Therefore, using current screening methods, it is not possible to prevent all sudden unexpected death in young athletes. Moreover, restriction of the adolescent with certain high-risk cardiac conditions from participating in sports may do little to protect against the danger of sudden death. Still, identifying those conditions that can be detected by history and physical examination, with subsequent restriction from sports participation, is an important means of reducing risks for these youngsters.

Fortunately, the immense tragedy of sudden death in athletes is very rare. Each year, 10-13 such occurrences are reported in the United States (Maron et al., 1996a; Van Camp et al., 1995), probably underestimating the true incidence. If approximately 4 million youngsters are involved in competitive sports, the statistical chance of an apparently healthy adolescent dying from unexpected heart disease during a year of sports participation is no more than 1:250,000. Putting this into perspective with other risks, approximately 14,700 adolescents die each year in motor vehicle accidents, 400 from fires, and 250 as a result of inhaled poisonous fumes (National Safety Council, 1985).

Certain groups of athletes may be at greater risk. Sudden cardiac death appears to be disproportionately more common in males, African Americans, and in
competitors in basketball and football (Maron et al., 1996a). The explanation for these trends is unclear, but they may reflect intensity of play or gender/racial influences on the frequencies of certain cardiovascular abnormalities. Most sudden deaths have occurred during or immediately after training or competition. This time course supports the prohibition of identified at-risk athletes from sports participation.

CAUSES OF SUDDEN DEATH

Many cardiovascular abnormalities can create risk for sudden death, presumably by causing insufficient blood flow or oxygen supply to the heart muscle and/or by causing an increased propensity for fatal arrhythmias of the ventricles of the heart. Fortunately, these diseases are uncommon in the pediatric age group. Autopsy studies of young athletes indicate that four cardiovascular conditions account for approximately 80 percent of unexpected sudden deaths (Table 1). These conditions are 1) diseases of the heart muscle, especially those associated with an enlarged heart (hypertrophic cardiomyopathy); 2) congenital anomalies of the coronary arteries that prevent an adequate supply of blood to the heart muscle; 3) rupturing of the aorta due to hereditary weakness in the vessel wall (aortic aneurysm usually associated with Marfan syndrome); and 4) valvular aortic stenosis (a thickening of the aortic valve that prevents adequate flow of blood out of the left ventricle into the coronary arteries and other major arteries of the body). Unfortunately, it is not possible to specifically define the magnitude of risk of death during sports participation for these abnormalities because such statistics are based on the number of athletes who were allowed to participate and who subsequently died. In other words, the number of athletes with unsuspected cardiovascular defects who play without complications is unknown.

In autopsy reports, no explanation can be found for a small number (2-7%) of sudden deaths in young athletes. In these cases, death has been presumed to be due to noncardiac causes or from arrhythmias and other cardiac conditions that cannot be detected at autopsy.

The principal strategy for reducing the incidence of sudden death in sports lies in recognizing would-be athletes with high-risk cardiac disease and restricting them from participation. The following sections address the most common causes of these tragedies and the effectiveness and limitations of the preparticipation medical examination in achieving this goal.

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy is the predominant cause of sudden unexpected cardiac death in athletes, accounting for one-half of cases in most reports. In its full-blown expression, this disease is characterized by unexplained, dramatic ventricular thickening, most prominently in the wall between the two ventricles, with virtual obliteration of the left ventricular cavity. Milder forms have also been observed in autopsy specimens following sudden death.

The mechanism for sudden death in patients with hypertrophic cardiomyopathy is uncertain. However, it is not difficult to predict that inadequate blood flow in the coronary arteries, insufficient filling of the ventricles, and predisposition to fatal disturbances in the normal rhythm of the electrochemical impulses that stimulate ventricular contractions might all play important roles. Significant obstruction to the flow of blood from the left ventricle into the aorta is seen in only a minority of cases and is generally not related to symptoms or risk of death.

Initially, it was thought that the prognosis for patients with hypertrophic cardiomyopathy was grim, with mortality rates as high as 2-4% per year (i.e., 20-40% of patients would not survive 10 years after diagnosis). However, subsequent studies suggested that the natural history of these patients is variable, and individuals with hypertrophic cardiomyopathy have been described who tolerated years of intense participation in athletics without complications (Maron & Klues, 1994).

Participation in physical activity increases the risk of sudden death with hypertrophic cardiomyopathy, but the physical activity is typically not participation in sports. Among 78 adults, 37% died at rest or during sleep, 24% during mild exertion, and 29% with vigorous activity (running, hiking) (Maron et al., 1982). As illustrated in this report, the disproportionately large percentage who died during physical activity has prompted the recommendation that those with hypertrophic cardiomyopathy be restricted from participation in competitive athletics.

<table>
<thead>
<tr>
<th>Cardiovascular Abnormality</th>
<th>Percent of Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>36.0</td>
</tr>
<tr>
<td>Increased cardiac mass (unexplained)</td>
<td>10.0</td>
</tr>
<tr>
<td>Aberrant coronary arteries</td>
<td>13.0</td>
</tr>
<tr>
<td>Other coronary abnormalities</td>
<td>10.0</td>
</tr>
<tr>
<td>Ruptured aortic aneurysm</td>
<td>5.0</td>
</tr>
<tr>
<td>Valvar aortic stenosis</td>
<td>4.0</td>
</tr>
<tr>
<td>Myocarditis</td>
<td>3.0</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>3.0</td>
</tr>
<tr>
<td>Arrhythmogenic right ventricle</td>
<td>3.0</td>
</tr>
<tr>
<td>Idiopathic myocardial scarring</td>
<td>3.0</td>
</tr>
<tr>
<td>Mitral valve prolapse</td>
<td>2.0</td>
</tr>
<tr>
<td>Atherosclerotic coronary disease</td>
<td>2.0</td>
</tr>
<tr>
<td>Other congenital heart disease</td>
<td>1.5</td>
</tr>
<tr>
<td>Long QT syndrome</td>
<td>0.5</td>
</tr>
<tr>
<td>Sarcoidosis</td>
<td>0.5</td>
</tr>
<tr>
<td>Sickle cell trait</td>
<td>0.5</td>
</tr>
<tr>
<td>Unexplained</td>
<td>2.0</td>
</tr>
</tbody>
</table>
The effectiveness of this strategy is limited by the difficulty in detecting athletes with hypertrophic cardiomyopathy during the preparticipation medical examination. Careful recording of the athlete's medical history may provide some clues. Hypertrophic cardiomyopathy is an inherited condition with variable symptoms and signs, and a positive family history can be obtained in about 20% of cases. Approximately one-half of patients may describe symptoms of chest pain, dizziness, fainting, and abnormal breathlessness, particularly with exercise.

Unfortunately, the results of the physical examination may be deceptively "normal." A heart murmur is typically absent or unimpressive unless the flow of blood from the left ventricle to the aorta is obstructed. The peripheral pulses can be bifid (two parts), and any murmur may intensify with standing or during a Valsalva maneuver (forcefully contracting the abdominal muscles while holding one's breath), but such clues are not always evident.

The electrocardiograms in most patients with hypertrophic cardiomyopathy demonstrate are abnormal, but the most definitive test for hypertrophic cardiomyopathy is the echocardiogram, in which sound waves are passed through the chest to enable the examiner to visualize heart structures while the heart is beating. The echocardiogram for a person with hypertrophic cardiomyopathy typically demonstrates that the thickness of the septum or wall between the two ventricles exceeds 18 mm and that there is a general thickening of the heart muscle. There may also be abnormal movements of the mitral valve, through which blood passes from the left atrium to the left ventricle, along with abnormal filling of the left ventricle during the relaxation (diastolic) phase of the cardiac cycle, and, in some patients, varying degrees of obstruction of blood flow leaving the left ventricle.

While a diagnosis of hypertrophic cardiomyopathy can be established confidently when all these features are present, certain features (e.g., left ventricular enlargement and atypical electrocardiogram) are also observed in the normal response to long-term intensive athletic training ("the athlete's heart"). Therefore, in some athletes, the health-care provider is faced with the dilemma of trying to decide if these findings reflect a highly trained athlete who can be encouraged to continue participating or represent a mild form of hypertrophic cardiomyopathy, a sign that the athlete should stop participating in sports.

Several criteria have been suggested to help make this differential diagnosis. The diagnosis of true hypertrophic cardiomyopathy is suggested by a) a ventricular septal thickness greater than 18 mm, b) normal or small left ventricular end-diastolic cavity dimension, c) failure of myocardial hypertrophy to regress following a period of detraining, d) evidence that ventricular thickening has been inherited, and e) abnormal patterns of left ventricular filling patterns as shown with Doppler echocardiography (Maron et al., 1995).

Coronary Artery Abnormalities

Atherosclerotic coronary artery disease is the most common cause of sudden cardiac death in adults, but this is not a consideration in young athletes except in extremely rare situations (i.e., inherited hypercholesterolemia). However, congenital structural abnormalities of the coronary vessels may create a risk for sudden death in children and adolescents, and such deaths can be precipitated by participation in vigorous physical activity.

Several forms of coronary artery abnormalities have been observed at autopsy following sudden unexpected death in young athletes. These include abnormal origin of the coronary arteries from the aorta, incomplete development of the arteries, coronary arteries buried deep within the heart muscle, and anomalous origin of the left coronary artery from the pulmonary artery instead of the aorta. At autopsy, investigators have also noted thickening of the walls of coronary arteries causing narrowing of critical coronary artery branches (e.g., those supplying the atrioventricular node, which transmits critical electrical impulses from the atria to the ventricles). The specific mechanism underlying death in these conditions is uncertain, but it presumably causes an insufficient flow of blood to the heart muscle when intense physical activity places greater demands on myocardial metabolism.

Congenital abnormalities of the coronary arteries that predispose an individual to sudden death are extraordinarily rare in the general population. That they rank as the second most common cause of sudden cardiac death in young athletes is a reflection of the difficulty in recognizing such abnormalities clinically. While cases of fainting during exercise occasionally lead directly to the diagnosis of coronary artery abnormalities, the most common initial presentation of this condition is sudden collapse and death of a previously apparently healthy athlete.

The risk and occult nature of congenital abnormalities of the coronary arteries was highlighted by the sudden death of the basketball star Pete Maravich. Following a long and highly successful career as one of the best college and professional players ever, Maravich died suddenly following participation in a recreational game when he was 40 years old. Autopsy revealed a complete absence of the left main coronary artery. The right coronary artery served the left coronary arteries, a reflection of the difficulty in recognizing such abnormalities clinically. While cases of fainting during exercise occasionally lead directly to the diagnosis of coronary artery abnormalities, the most common initial presentation of this condition is sudden collapse and death of a previously apparently healthy athlete.

Marfan Syndrome

Marfan syndrome is an inherited disease characterized by laxity of connective tissue. Research has identified defective formation of a specific protein in patients with this syndrome, an expression of one of several chromosomal abnormalities. In the full-blown disorder, individuals have heart disease (dilated aorta and regurgitation of blood through heart valves), abnormalities of the eyes (dislocated lens and myopia), musculoskeletal disorders (spinal curvature and a depressed sternum),
tall stature, abnormally long and slender fingers and toes, and hyperextensible joints. Commonly, however, patients with Marfan syndrome present with varying combinations of these findings.

The cardiovascular abnormalities present the greatest risk for mortality and morbidity in Marfan patients. Progressive widening of the root of the aorta where it leaves the left ventricle is common, with a risk of eventual formation and sudden rupture of an aortic aneurysm. This defect is typically accompanied by abnormalities of the aortic and mitral valves and insufficient blood flow into the aorta. Efforts to prevent rupture of an aortic aneurysm include the use of beta-blocker medication and surgery to narrow the aortic root when it reaches a critical diameter of 55-60 mm.

The risk of ruptured aortic aneurysms from sports play in athletes with Marfan syndrome was demonstrated by the sudden death of the volleyball player Flo Hyman, the 6-foot 5-inch star of the 1984 United States Olympic team, who collapsed and died during a game in Japan in 1985. Autopsy revealed findings typical of Marfan syndrome, with a ruptured aortic aneurysm and accumulation of blood in the pericardial sac surrounding the heart. This tragedy resulted in an awareness that tall athletes with undiagnosed Marfan syndrome might be at risk and led to recommendations for restricting such individuals from participating in certain types of physical activity (i.e., avoidance of resistance training, sports that might involve a blow to the chest, and endurance training) (Braverman, 1998; Pyeritz & McKusick, 1979).

The diagnosis of Marfan syndrome rests with clinical recognition of its typical physical features as described earlier. Cardiac examination of most patients with Marfan syndrome reveals abnormal sounds associated with a dysfunctional mitral valve.

A careful family history may reveal other members who have Marfan syndrome, but having several tall, healthy relatives is more likely to be evidence of familial tall stature. Chest x-rays and electrocardiograms are typically normal in patients with Marfan syndrome. Suspicion of Marfan syndrome on preparticipation screening should prompt referral to a cardiologist for assessment of cardiac manifestations of this disease. The diagnosis is supported by echocardiographic findings of mitral valve prolapse in conjunction with widening of the aortic root. (Prolapse of the mitral valve is excessive backward movement of the valve into the left atrium during contraction of the left ventricle. This allows regurgitation of blood into the atrium, which reduces flow into the aorta.)

Valvar Aortic Stenosis
Among the cardiac diseases that cause sudden death in athletes, congenital narrowing of the aortic valve (valvar aortic stenosis) is the most prevalent in the population at large. Yet this anomaly accounts for only a small percentage of sudden-death reports. The explanation presumably lies in the fact that, as opposed to the other causes that are difficult to detect, patients with aortic stenosis exhibit typical physical findings that are easily observed.

Patients with valvar aortic stenosis have an obstructed flow of blood from the left ventricle into the aorta, and risk of sudden death is presumably created by either inadequate blood flow to the heart muscle during exercise or marked elevation of left ventricular pressure accompanying physical activity that triggers a strong vagal nerve reflex to disrupt the heartbeat rhythm (Rowland, 1995). Risk of sudden death, which appears to be more common during vigorous physical activity, is high only in those with significant obstruction of the aorta as indicated by abnormalities in cardiac pressures, an abnormal electrocardiogram, and/or chest pain. Would-be athletes with valvar aortic stenosis who are at risk of sudden death while participating in sports should be easily recognizable during a cardiac examination by the presence of specific heart murmurs. Such findings should prompt referral to a cardiologist.

SCREENING FOR RISK
Among the millions of young athletes who undergo preparticipation medical evaluation each year, very few are at risk for sudden cardiac death. The magnitude of these tragedies, however, demands an effective plan to minimize their occurrence. It is important, then, to determine the most accurate yet efficient methods for recognizing these "needles in the haystack."

A substantial reduction in the incidence of sudden death in athletes requires that screening tools be effective in recognizing the four high-risk conditions outlined above. Unfortunately, the medical history and physical examination, mainstays of the preparticipation evaluation, are likely to be effective in detecting only two of these conditions—aortic aneurysms associated with Marfan syndrome and congenital valvar aortic stenosis.

Recognition of hypertrophic cardiomyopathy becomes more problematic. Description in the medical history of chest pain, dizziness, fainting, or abnormal breathlessness during physical activities, or a family history of sudden early death would provide valuable clues. However, these features are not evident in a majority of cases. A harsh murmur during systole detected with the stethoscope placed along the left border of the sternum, particularly if it increases when the patient performs a Valsalva maneuver, should also raise suspicion. Although the effectiveness of history and physical examination in detecting hypertrophic cardiomyopathy during the preparticipation evaluation is not definitively known, the available information is pessimistic. For instance, in a recent compilation of 134 cardiac deaths in athletes, only one of 48 cases of hypertrophic cardiomyopathy had previously been recognized (Maron et al., 1996a). Congenital coronary artery abnormalities will probably escape detection by history and examination, but patients' reports of exercise-induced chest pain, dizziness, or fainting should prompt further evaluation.
In summary, a careful history and physical examination are important in identifying certain individuals who would be at risk for sudden cardiac death during sports participation. However, it is unlikely that such a preparticipation evaluation will reduce the current rate of these tragedies. This conclusion has stimulated efforts to find more accurate means of identifying those at risk.

Echocardiography will effectively identify individuals with hypertrophic cardiomyopathy and has therefore been suggested as a routine screening tool in the preparticipation assessment. A quick, inexpensive cardiac ultrasound study that is both sensitive and specific to hypertrophic cardiomyopathy should reduce the incidence of sudden cardiac death in athletes by one-half. Unfortunately, echocardiography does not satisfy these criteria. Echocardiography is expensive ($500-1,000 for a complete study) and requires prohibitively-costly equipment ($100,000-$200,000). At $500 per athlete, it would cost $2 billion annually to identify 3-5 athletes who would otherwise die of hypertrophic cardiomyopathy. Performing an abbreviated echocardiogram with donated equipment is also not a cost-effective approach that would allow widespread use of echocardiograms for screening millions of athletes.

Another serious drawback of routine screening by echocardiography is the effect of false positive results (Braden & Strong, 1988). While full-blown hypertrophic cardiomyopathy can be fairly easily detected, a large overlap may occur in heart-wall thickness values that represent milder cardiomyopathy, the "athlete's heart," or normal anatomic variant. Considering that mild ventricular thickening would be detected in tens of thousands of normal youngsters, such findings "would generate heavy emotional, financial, and medical burdens for the athlete, family, team, and institution by virtue of the uncertainty created and the requirement for additional testing" (Maron et al., 1996b).

Similar objections can be raised about the use of the electrocardiogram as a screening tool. Consequently, it has been recommended that a medical history and physical examination constitute routine preparticipation screening and that other cardiac studies such as echocardiography, electrocardiography, and exercise stress tests be reserved for secondary evaluation of those with suspected heart disease (Braden & Strong, 1988; Maron et al., 1996b).

A similar situation exists with hypertrophic cardiomyopathy. While certain genetic markers of this disease are known, there is insufficient precision to allow their use in its detection. Still, given the limitations of the current screening approach, the potential ability to detect those at risk by a simple blood test is attractive. Future developments in genetic analysis may create an accurate, low-cost method.

Recognizing the inability of preparticipation screening to identify all those at risk, the availability of automatic external defibrillators at sports events has been proposed (Cantwell, 1998). This proposal is based on 1) the assumption that the mode of death in sports is often ventricular fibrillation, and 2) the development of relatively-inexpensive defibrillators that can be effectively used by non-medical personnel. The availability of such defibrillators, in fact, has been increasingly common at college and professional sports events as well as non-sports settings to prevent sudden death before emergency personnel can respond. Concerns regarding safety, cost-effectiveness, efficacy, and legal issues still need to be addressed, but these devices provide hope that the incidence of sudden death in athletes might be decreased.

SUMMARY
Each year in the United States, only about one dozen apparently healthy young athletes die suddenly from undiagnosed diseases of the heart and blood vessels. Still, everything possible should be done to minimize these sudden deaths, which often occur during training or competition in sports. Most of these deaths can be attributed to abnormalities in the structure and function of the heart muscle itself, the coronary arteries that supply blood to the heart, the walls of the aorta, or the heart valve that leads to the aorta.

Some of the cardiovascular abnormalities that may lead to sudden death can be detected by routine physical examination during the preparticipation evaluation, but others may not be found without more sophisticated testing. Unfortunately, additional diagnostic procedures such as echocardiography and electrocardiography are not recommended for routine screening of athletes because the techniques are not cost-effective and have a high rate of false positive findings. Future developments, including genetic screening and the availability of defibrillators at the sport site, may decrease the incidence of sudden death. The current limitation in our ability to identify athletes at risk necessitates acceptance that participation in sports carries some risk.

REFERENCES


IDENTIFYING ATHLETES AT RISK FOR SUDDEN DEATH

A careful medical history and cardiac physical examination are the best tools available for detecting heart disease that might pose a risk for participating in sports. Unfortunately, some cardiac abnormalities that can cause death during sports training and competition are not likely to be detected during routine examinations. Nonetheless, the health-care provider performing such evaluations can reduce the risk of sudden death by paying attention to several key aspects of the history and physical examination.

Medical History

* A history of chest pain, dizziness, fainting, or abnormal breathlessness and fatigue during exercise is a "red flag" for possible risk from sports participation. In fact, such symptoms may be the only obvious clues to the presence of high-risk cardiovascular disease.

* Unexpected sudden death of a family member at a young age should raise suspicion of possible inherited cardiac disease. A family history of unusually tall individuals with heart and/or eye problems, particularly if tall stature is not evident in other family members, suggests Marfan syndrome, a serious risk factor for sudden death.

* A history of heart palpitations or heart murmur may signal a need for cardiac consultation.

Because children and adolescents often have insufficient knowledge to respond appropriately to medical-history questions, parents should complete preparticipation medical history forms.

Physical Examination

* Physical signs of Marfan syndrome should be assessed: tall stature, long arms and fingers, sunken sternum, abnormal spinal curvature, flat feet, and high arched palate. The diagnosis of Marfan syndrome is supported by a history of near-sightedness (or dislocated lens of the eye) and certain abnormal heart sounds.

* Athletes with significant and harsh heart murmurs should be referred to a cardiologist. This is particularly true if the murmur is heard with the stethoscope placed on the back, if it becomes louder while the athlete is sitting up or while he or she contracts the abdominal muscles while holding the breath, or if it is accompanied by other abnormal heart sounds.

* Blood pressure should be obtained, and femoral pulses should be evaluated to rule out narrowing of the aorta. A biphasic arterial pulse suggests the possibility of a pathologically enlarged heart.

A focused cardiac history and careful physical examination with attention to the points above can be performed rapidly and should be a routine part of preparticipation screening. Any questions raised in this assessment should be brought to the attention of a cardiologist before the athlete is allowed to participate in strenuous activity.