MARFAN SYNDROME was identified by the American Heart Association (AHA) in 1996 as a cause of sudden death in competitive athletes. Marfan syndrome (MFS) is a connective tissue disorder that primarily affects the ocular, cardiovascular, and musculoskeletal systems. Although MFS affects several systems of the body, cardiovascular complications account for the majority of deaths among individuals with MFS. In 1972, the mean age of death from MFS was 32 years ± 16 years, whereas the mean age of death from MFS in 1993 was 41 years ± 18 years. The increased life expectancy from 1972 to 1993 may be attributed to increases in life expectancy in the general population, earlier identification of MFS through screening, and earlier medical intervention.

Marfan syndrome is a rare condition. Although precise data are unavailable, its prevalence is believed to be greater among athletes than the general population. Although the physical characteristics of individuals with MFS are beneficial in sports such as basketball and volleyball (i.e., increased height and arm-span), the recommendations for physical activity for individuals with MFS limit participation to low-intensity activities with minimal physical contact. Screening for MFS should be incorporated into preparticipation physical examinations (PPE). The purposes of this report are to increase awareness of MFS and to suggest a role for athletic trainers and therapists who assist in the administration of PPEs.

Screening for MFS During the PPE

An important purpose for conducting a PPE is to screen for predisposing conditions that can affect an athlete’s health. The American College of Cardiology suggests that a major objective of the PPE is to identify cardiovascular irregularities that can cause sudden cardiac death. The American Heart Association and the American Academy of Family Physicians have supported screening for symptoms of Marfan syndrome as a component of the PPE.

Evaluation by a subspecialist is recommended for a person who has a family history of MFS and/or is a male taller than 6 ft, or a female taller than 5 ft 10 in, who has two or more clinical symptoms of Marfan syndrome. Clinical manifestations of MFS affect several bodily systems, including the skeletal system. Skeletal manifestations of MFS that are recommended for the PPE screening correspond to the Ghent diagnostic criteria, which include arm-span to height ratio, wrist sign, thumb sign, pes planus, chest deformity, and high-arched palate. The criterion for the arm-span to height ratio is greater than 1.05 (Figure 1). This ratio provides an indication of excessive long bone growth of the arms and legs, which is often present in individuals with MFS. The wrist sign (Figure 2) and thumb sign (Figure 3) are grouped together because both must be positive to classify the findings as a manifestation of MFS. These two tests can identify arachnodactyly and hypermobility of joints. The wrist sign is assessed by wrapping the fifth phalanx and the thumb around the distal radio-ulnar joint. A positive test involves the thumb overlapping or touching the distal phalanx of the fifth digit. The thumb sign is assessed by placing the thumb into the palm and covering the thumb in an enclosed fist. A positive thumb sign is noted when the entire nail bed of the thumb projects beyond the ulnar border of the clenched hand.
Pes planus is frequently present in individuals with MFS as a result of joint laxity and hypermobility. No specific pes criteria have been specified for MFS, but indicators such as the Feiss line could be utilized (Figure 4). The Feiss line quantifies pes planus as the vertical distance from the navicular tubercle to the floor. A positive result is noted when the navicular tubercle drops more than one-third of its vertical distance from the floor when a weightbearing measurement is compared to a non-weightbearing measurement. Chest deformity in an individual with MFS is caused by an overgrowth of the ribs, which pushes the sternum anteriorly (pectus carinatum) or posteriorly (pectus excavatum). Pectus carinatum is discerned by observable protrusion of the chest wall over the sternum and pectus excavatum is indicated by a caved-in or sunken chest appearance. A high arched palate, which is a craniofacial manifestation of MFS, causes the upper teeth to be positioned inside the lower teeth when biting down.

**The PPE Role of Athletic Trainers**

Lack of a sufficient number of available physicians to assess all aspects of every athlete’s status during the PPE necessitates assistance from non-physician health care personnel, which can include performance of the screening tests recommended for identification of MFS. The American Heart Association (AHA) has suggested that a certification course should be developed to ensure proficiency in the performance of the screening tests.
of cardiovascular examinations. Although athletic trainers generally lack extensive knowledge for assessment of the cardiovascular system, the majority of tests recommended for MFS screening relate to skeletal system abnormalities. Athletic trainers clearly possess sufficient knowledge to assist in implementation of the AHA guidelines for cardiovascular screening.*

Assessment of MFS by Athletic Trainers

To evaluate the ability of athletic trainers to identify skeletal manifestations of MFS in a high school athlete, we calculated the intertester reliability of observations made by 12 certified athletic trainers (mean experience = 13.5 years) for the six screening tests previously described in this report. Each of the athletic trainers performed each of the six screening tests to assess the status of a 15-year-old male athlete. The trainers participated in a single 30-min training session, during which the performance of each physical screening procedure for MFS was demonstrated. A printed guide that included descriptions and photographs/drawings of each screening test was also distributed.

The 15-year-old model did not possess any of the physical manifestations of MFS. All of the athletic trainers correctly classified the model as negative for the arm-span to height ratio, wrist sign, thumb sign, chest deformity, and pes planus, 100% agreement among the testers (r = 1.0). However, there was disagreement on the presence of a high-arched palate (3 reporting the existence of a high-arched palate and 9 reporting a negative finding). The intertester correlation among the athletic trainers for the high-arched palate test was r = .24.

The perfect agreement among the athletic trainers for the arm-span to height ratio, the Feiss line, chest deformity test, and the wrist and thumb sign makes them valuable additions to the PPE. Screening female athletes may raise privacy issues associated with chest deformity. The poor level of agreement for the existence of a high-arched palate may be attributed to the subjective nature of the test. The test criteria are vague and the athletic trainers relied on descriptions and pictures to make a determination of whether or not the test was positive. Concerning crowding of the teeth, the evaluators considered feedback from the model about corrective dental procedures performed in the past. Development of more objective criteria for determination that an individual possesses a high-arched palate would almost certainly improve intertester agreement.

Recommendations

The AHA published a statement recommending screening for Marfan syndrome prior to athletic participation. Screening athletes for MFS may lead to earlier diagnosis of the condition and potentially life-saving treatment. Our findings suggest that athletic trainers can reliably interpret the arm-span to height ratio, Feiss line measurement, wrist sign, thumb sign, and chest deformity assessment to identify manifestations of MFS.

References


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