What is it?
Marfan syndrome is a genetic disorder of the protein fibrillin 1 leading to problems in the bones, heart, and eyes. About 75% of cases are inherited from a parent, but 25% of the cases have no other members of the family affected. The incidence is 2-3 per 10,000 individuals and occurs equally among men and women.

Symptoms/Risks
People with this disorder tend to be tall in stature due to long bone overgrowth, but the real risk lies with the tendency for the large blood vessel leaving the heart (aortic root) to dilate and rupture. Without a proper early diagnosis, the rate of sudden death during exercise is greatly increased and can happen during adolescence and young adulthood.
Patients with Marfan syndrome have a variety of symptoms, but can include some or many of the following:

- Tall and slender build
- A breastbone that caves in or out
- Long arms, legs, and fingers
- Abnormal curvature of the spine (scoliosis)
- Heart murmur
- Dilation or rupture of the aorta
- Dislocation of the lens of the eye
- A high-arched palate with crowding of the teeth

Sports Medicine Evaluation & Treatment
A sports medicine physician will perform a comprehensive history and exam of the athlete, paying special attention to any family history of early sudden death. If Marfan syndrome is suspected, the sports medicine physician will involve a geneticist (genetics specialist), ophthalmologist (eye doctor) and cardiologist (heart doctor) to confirm the diagnosis.

Assessment includes:
- A genetic screen
- A slit-lamp examination of the eye to evaluate for lens abnormalities
- An echocardiogram (sonogram of the heart) to evaluate for any abnormalities of the heart valves or aorta
- A thorough musculoskeletal examination must also be performed, including an evaluation of the long bones and an assessment for scoliosis.

Injury Prevention
Annual evaluation of the eyes and heart to identify any new abnormalities or changes from previous evaluations. Athletes with Marfan Syndrome should avoid strenuous activities that could put them at risk for aortic rupture.
AMSSM is a multi-disciplinary organization of sports medicine physicians dedicated to education, research, advocacy and the care of athletes of all ages. The majority of AMSSM members are primary care physicians with fellowship training and added qualification in sports medicine who then combine their practice of sports medicine with their primary specialty. AMSSM includes members who specialize solely in non-surgical sports medicine and serve as team physicians at the youth level, NCAA, NFL, MLB, NBA, WNBA, MLS and NHL, as well as with Olympic teams. By nature of their training and experience, sports medicine physicians are ideally suited to provide comprehensive medical care for athletes, sports teams or active individuals who are simply looking to maintain a healthy lifestyle. Find a sports medicine physician in your area at www.amssm.org.

## MARFAN SYNDROME

### Return to Play

If Marfan syndrome is diagnosed, athletes are disqualified from competitive athletics. If Marfan syndrome is suspected, but not confirmed after genetic testing, competitive athletics are permitted as long as the athlete has a periodic clinical and diagnostic workup.

Recommendations for recreational sports activities for patients with Marfan syndrome include low-intensity activities like golf, bowling, or diving, so long as there is no evidence of aortic root dilation, a heart valve issue called mitral regurgitation, or a family history of aortic rupture or sudden death.

Athletes with Marfan syndrome are not recommended to participate in activities that involve sustained muscle contraction such as weight lifting or rock climbing. High-intensity level activities such as basketball, ice hockey, skiing, baseball, surfing and scuba diving are also not recommended. These activities cause additional strain on the aorta putting it at risk to rupture. Regular aerobic activities will improve bone, heart and psychological health and are recommended in most individuals with Marfan syndrome.

### References