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A Tall Order

With Marfan Syndrome, Detection is Protection

The teenager sitting in your exam room is tall and lanky, with long arms and slender fingers. Sizing up his physique, many people ask him: Do you play basketball? But a more appropriate question may be: Has anyone in your family younger than age 50 ever died suddenly?

That long-limbed stature is a classic feature of Marfan syndrome, a genetic disorder of the connective tissue that can lead to aortic enlargement and rupture. Young athletes with Marfan syndrome may show skills on the court, but high-intensity, competitive sports could endanger their lives.

Being exceptionally tall is only one marker. Marfan syndrome involves a collection of common features. It affects the skeletal system, eyes, lungs, blood vessels and heart, with symptoms emerging as a child develops. An estimated 200,000 Americans have this connective tissue disorder, and in 75 percent of the cases, they share the condition with someone else in their family.[1]

For example, ectopia lentis, is a defining feature. But a combination of other aspects–flat feet, pectus excavatum, mitral valve prolapse, laxity in the joints and severe nearsightedness–may be enough to warrant some questions about a family's history.

In Marfan syndrome, an inherited or spontaneous mutation of the FBN-1 gene causes a defect in the protein fibrillin-1. Fibrillin-1 is an essential part of fibers that provide strength and flexibility to connective tissue throughout the body. Affecting one of the basic building blocks of human anatomy, Marfan syndrome has far-ranging effects.

Marfan syndrome often is discovered when a tall, thin pre-teen seeks an orthopaedic consult for scoliosis. About 60 percent of people with Marfan syndrome have scoliosis, which tends to be rapidly progressive.[2] Scoliosis is typically easy to detect with a physical exam and X-ray.

Patients who are suspected to have Marfan syndrome should be referred to a cardiologist because of the potentially serious effects of the disorder on the aorta. Stress on the aorta will increase with time, so it is important to diagnose the condition as early as possible.

Evaluation begins with family history, patient history and exam. (Yes, it is important to know everyone in the family who died suddenly before the age of 50.) If Marfan syndrome is suspected, an electrocardiogram (EKG) and echocardiogram will reveal if there has been any cardiovascular involvement.

If the findings suggest Marfan syndrome, families should be referred to a clinical geneticist, especially because the syndrome may be yet undetected in other family members. Every child in the family should be screened as well as both parents.

Treatment depends on each child's medical profile. Beta blockers may slow the progression of dilatation of the aorta. Research is determining how effective they are and if other cardiac medications provide similar or better results. In more advanced cases, surgery may be necessary to replace the aorta to prevent a rupture. Surgery also may be necessary to correct scoliosis because children with Marfan syndrome often do not respond well to bracing.

Children with Marfan syndrome can and should maintain active lives. But it is important for them to avoid contact sports or competitive, high-intensity activities, including wrestling, hockey, football, basketball, lacrosse, volleyball, weight-lifting or sprinting. Recreational sports–baseball, tennis, golf, bowling and even long-distance running–pose less risk.

Diagnostic Criteria for Marfan Syndrome

The diagnostic criteria, known as the 2010 revised Ghent nosology, relies on a scoring system that assigns a greater weight to some features than others.[3] In summary, Marfan syndrome is diagnosed if there is:

- Aortic root dilatation or dissection
- Ectopia lentis
- FBN1 mutation (if there is a family history)
- Systemic score of seven or higher for various Marfan syndrome-like features

*Visit the National Marfan Foundation at **www.marfan.org** for more information about the diagnostic criteria and scoring.*

Sometimes a treating physician must be emphatic with parents who want their child to continue to compete in high-level athletics. In rare cases, a call to child protective services might be warranted if a child's life is endangered by intense sports competition. After all, if the aorta dissects, a child may not survive. Undiagnosed or unaware, a child with Marfan syndrome is at risk of sudden death.

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1 National Marfan Foundation. "About Marfan Syndrome." Available at www.marfan.org/marfan/2409/FAQ. Accessed on Sept. 22, 2010. 2 Sponseller PD, Hobbs W, Riley LH 3rd, Pyeritz RE: The thoracolumbar spine in Marfan syndrome. J Bone Joint Surg Am. 1995 , 77(6):867-76. 3 Loeys BL, Dietz HC, Braverman AC, et al. The revised Ghent nosology for the Marfan Syndrome. J Med Genet. 2010;47:476-485.