Marfan Syndrome
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After completing this article, the reader should be able to:
- Explain the genetic aspect of Marfan syndrome.
- Summarize the history of Marfan syndrome.
- Explain how Marfan syndrome is diagnosed.
- Identify the clinical manifestations of Marfan syndrome.
- Discuss treatment options for different manifestations of Marfan syndrome.
- Describe disorders that are similar to Marfan syndrome.

Television and newspapers bear witness to the effects of Marfan syndrome. When an athlete collapses on the court or field, death sometimes occurs instantaneously. This situation does not happen very often, but when it does it usually makes headlines. The question on everyone’s mind is why an asymptomatic athlete suddenly dies for no apparent reason. Flo Hyman, an Olympic volleyball star, collapsed and died at a game in Japan in 1986. She was only 32 years old. Death occurred as a result of an aortic dissection, and an autopsy revealed that she had Marfan syndrome. Sudden death related to Marfan syndrome is not limited to athletes. Jonathan Larsen, the composer of the acclaimed musical “Rent,” died the morning of the show’s opening night, just days before his 36th birthday. Both Hyman and Larsen, healthy until their deaths, had one thing in common: an aortic dissection caused by undiagnosed, and therefore untreated, Marfan syndrome. Both deaths brought about a greater public awareness of Marfan syndrome; however, the disease remains difficult to diagnose in some patients.1

Marfan syndrome is a heritable disorder of the connective tissue that affects mainly the cardiovascular system, skeletal system and eyes. Marfan syndrome almost always occurs because of a mutation in the gene (FBN1) that encodes the protein fibrillin. Fibrillin is a component of microfibrils, which are present in the connective tissue throughout the body. Microfibrils are uniform minuscule structures that compose the extracellular matrix. They form the framework of elastic fibers in tissues such as the aorta media and the ligaments of the musculoskeletal system. Other microfibrils occur independent of the elastic fibers in tissue, such as the eye filaments that hold the lens in place.

Fibrillin is one of the larger human proteins and is a major component of connective tissue. The primary purpose of connective tissue is to hold the body together and provide structure for the growth and development of the body. Connective tissue contributes to the function of all organs in all stages of development, from childhood to old age. Thus, it is reasonable to assume that a defect in connective tissue production would interfere with normal skeletal and organ development, which is the case in

Marfan syndrome is a heritable disorder of the connective tissue that affects the cardiovascular, skeletal and ocular systems, and often involves the skin, nervous system and lungs. Historically, a person with Marfan syndrome had a poor prognosis because of the cardiovascular effects of this disorder. The life expectancy has improved dramatically over the past 30 years for individuals with Marfan syndrome because of aggressive medical and molecular research and advances in surgical technology. This article discusses the history, genetics, manifestations, diagnosis and treatment of Marfan syndrome.

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Marfan syndrome. People who have Marfan syndrome are usually tall, slender and have loose-jointed arms, legs and fingers. The first noticeable characteristic of many patients with Marfan syndrome is their long arms and legs that do not seem to match their bodies. They may have scoliosis, prominence or concavity of the chest and long, narrow faces with a high palate and crowded teeth. Their digits may be hypermobile, appearing almost as if they were made from elastic. Because connective tissue is found throughout the body, evidence of Marfan syndrome can be found in the eyes, heart, blood vessels, lungs and nervous system.2

Signs and symptoms of Marfan syndrome can vary from person to person. In some cases, the symptoms may be mild and in others, moderate to severe. The most serious effects of the syndrome involve the cardiovascular system. Weaknesses in the connective tissue of the aorta can potentially cause a life-threatening situation, such as an aortic dissection.

Marfan syndrome is not considered a rare disorder, as it is estimated to occur in about 2 to 3 people per 10,000.3 There are more than 200 different heritable disorders of connective tissue, and some have the same characteristics as Marfan syndrome. This syndrome affects men, women and children of all races, ethnic groups and geographic locations. Because it is an inherited condition, the greatest risk factor for Marfan syndrome is having a parent with the disorder.3

History of Marfan Syndrome
Marfan syndrome was named for French pediatrician Antoine Bernard Jean Marfan, who was asked to evaluate a 5-year-old girl named Gabrielle in 1896.4 Her eyes, heart and intellect appeared normal at the time of her visit to Dr. Marfan, but he thought her long, disproportionate body and spider-like extremities were quite unusual. Marfan had never seen such features and was so impressed by the girl’s long body and arms that he documented her clinical signs in a medical journal, calling the condition “dolichostenomelia,” which is Greek for “long, thin extremities.” (See Fig. 1.) He accompanied this description with drawings of her hands and feet, using the term “pattes d’araignée,” or “spider fingers.” Upon further testing, skeletal radiographs of the child demonstrated possible thoracolumbar kyphoscoliosis, an anterior chest deformity, and pulmonary tuberculosis.

Forty years after this first documented report, Marfan reviewed more than 150 similar cases. By that time, other organ systems were known to be involved, as revealed by congenital displacement of the lens (ectopia lentis) and mitral valve disorders. It was apparent to Marfan by then that the disorder was genetic.3 He gained international recognition for his work and was considered a pioneer of pediatric medicine in France. In 1934 Marfan received an honorary fellowship from Britain’s Royal Society of Medicine.4 As time went on, physicians reported similar clinical signs in their patients, and by the early part of the 20th century, Marfan’s name was linked with this condition. The medical term “syndrome” was added because it means that a group of physical signs occurs often enough to create a recognizable pattern.3

Although the disorder bears his name, Marfan was not the first physician to describe these clinical signs. Approximately 20 years before Marfan’s presentation, physician E. Williams described a family with similar characteristics to the American Ophthalmological Society in July 1875. Williams’ report, titled “Rare Cases, With Practical Remarks,” described a father and his 2 children whose body habitus appeared large and loose-jointed and displayed dislocated lenses of the eyes. Unfortunately, Williams did not include photographs of these patients, and no further comments about them were made. Thus, his description went unnoticed, leaving Marfan’s discovery the memorable one. In 1902 the condition was renamed “arachnodactyly.” This term, along with “dolichostenomelia,” is still used today by physicians when diagnosing Marfan syndrome.3 Complications in the aorta were described in 1943, and by 1955 Victor McKusick had documented