WHAT IS MARFAN SYNDROME?
Marfan syndrome is an inherited or genetic disorder of connective tissues that can affect the heart, blood vessels, lungs, eyes, bones and ligaments. Connective tissue is the material that holds the tissues of the body together. Marfan syndrome is one of the most common connective tissue disorders, affecting approximately one in 5,000 Americans.

WHAT CAUSES MARFAN SYNDROME?
Marfan syndrome is caused by a single abnormal gene. Marfan syndrome is an autosomal dominant (aw-toe-SO-mal DOM-ih-nant) disorder, meaning that you only need one gene to have the disease and that each child of an affected individual has a 50 percent chance of having the condition. Approximately 25 percent of Marfan syndrome cases occur spontaneously with no previous family history of anyone having the disorder.

HOW IS MARFAN SYNDROME DIAGNOSED?
It is important that you find a doctor who is knowledgeable about Marfan syndrome. You might consider meeting with a clinical geneticist (jin-NEH-ti-sist), a doctor who specializes in inherited diseases, or a rheumatologist (ROO-ma-tall-o-jist), a doctor who specializes in arthritis and related diseases. The National Marfan Foundation or the March of Dimes can refer you to a medical geneticist.

Diagnosis of Marfan syndrome is based on a complete physical examination that focuses on the heart, eyes and musculoskeletal system.

WHAT ARE THE SYMPTOMS AND TREATMENTS OF MARFAN SYNDROME?
Marfan syndrome can cause one or more of the following symptoms:
Heart Problems

In people with Marfan syndrome, the aorta (the main blood vessel that carries blood away from the heart) can become enlarged and weakened. This is called an aortic aneurysm (a-OR-tik an-yeer-IZ-em). The lining of the aorta can split, and in rare cases blood can leak into the chest. In addition, the weakened aorta can lead to problems with the valves of the heart called aortic insufficiency or mitral valve prolapse.

Heart problems may not produce any obvious symptoms, so people with Marfan syndrome should have regular checkups to detect possible complications. Regular echocardiograms (a special examination of the heart made by sound waves) can help detect heart or aortic problems.

Medications called beta blockers are prescribed to reduce strain on the aorta and heart valves. Beta blockers lower high blood pressure and decrease the force of heartbeats. Some people may require surgery if the aorta quickly increases in size or reaches a dangerous size.

Occasionally the retina (light-sensitive tissue at the back of the eye) may become detached, causing sudden vision loss that requires surgery. You should have regular eye exams by an eye specialist called an ophthalmologist (opf-THAL-mall-o-jist).

Spine Problems

Children with Marfan syndrome should be checked regularly for scoliosis (sko-lee-O-sis). Scoliosis is a curvature of the spine. Children with scoliosis should see a doctor who specializes in bone disorders, called an orthopaedist (or-THO-PEE-dist). They may prescribe a removable brace to stabilize the spine. Physical therapy also may be recommended. Surgery may be necessary if curvature increases.

Breast Bone Problems

In some people, the chest may become indented and put pressure on the heart and lungs. In rare cases, surgery may be necessary to relieve the pressure and prevent organ damage.

Joint Pain

Some people with Marfan syndrome experience joint pain, which is frequently associated with loose-jointedness (joints that bend more than normal). Joint pain can be treated with medications, such as nonsteroidal anti-inflammatory drugs (NSAIDs) and exercise, such as walking and water exercise. Examples of NSAIDs include aspirin, ibuprofen and naproxen.

Eye Problems

Nearsightedness is common in people with Marfan syndrome. It can be corrected with prescription glasses or contact lenses. People with Marfan syndrome also may have problems with the lens of their eye (behind the pupil of the eye). The lens can become torn from supporting tissues, resulting in vision problems.
since its inception in 1948. Additionally, the Arthritis Foundation supports key public policy and advocacy efforts at a local and national level in order to make a difference on behalf of 70 million people living with arthritis.

As your partner in taking greater control of arthritis, the Arthritis Foundation also offers a large number of programs and services nationwide to make life with arthritis easier and less painful and to help you become an active partner in your own health care.

Contact us at (800) 283-7800 or visit us on the Web at www.arthritis.org to become an Arthritis Advocate or to find out how you can become involved.

The Arthritis Foundation acknowledges with appreciation Carlo Mainardi, MD, MBA, Newark Beth Israel Medical Center, Newark, NJ; and Laura Robbins, DSW, Hospital for Special Surgery, New York, for their assistance with this booklet.

For more information: The Arthritis Foundation offers a wide variety of books, brochures and videos about different forms of arthritis, treatment and self-management techniques to help you take control of your arthritis. To order any of these products, become an Arthritis Foundation member or to subscribe to the Arthritis Foundation's award-winning consumer health magazine, Arthritis Today, call (800) 283-7800. Call or visit our Web site (www.arthritis.org) to find out how you can take control of your arthritis and start living better today!