DOCUMENT RESUME

ED 443 241 EC 307 935

TITLE	Questions & Answers about...Marfan Syndrome.
INSTITUTION	National Inst. of Arthritis and Musculoskeletal and Skin Diseases (NIH), Bethesda, MD.
SPONS AGENCY	National Institutes of Health (DHHS), Bethesda, MD.
REPORT NO	AR-06-QA
PUB DATE	1999-04-00
NOTE	8p.
AVAILABLE FROM	NIAMS/National Institutes of Health, 1 AMS Circle, Bethesda, MD 20892-3675; Tel: 301-495-4484; Tel: 301-565-2966 (TTY); Fax: 301-718-6366; e-mail: NIAMSInfo@mail.nih.gov; Web site: http://www.gov/niams/.
PUB TYPE	Reports - Descriptive (141)
EDRS PRICE	MF01/PC01 Plus Postage.
DESCRIPTORS	Adults; Children; Clinical Diagnosis; *Etiology; *Individual Characteristics; Intervention; *Outcomes of Treatment; *Physical Disabilities; *Symptoms (Individual Disorders)
IDENTIFIERS	*Marfan Syndrome

ABSTRACT

This fact sheet answers general questions about Marfan syndrome, a heritable condition that affects the connective tissue. It describes the characteristics of the disorder, the diagnostic process, and ways to manage symptoms. Characteristics include: (1) people with Marfan syndrome are typically very tall, slender, and loose jointed; (2) more than half of all people with Marfan syndrome experience dislocation of one or both lenses of the eye; (3) most people with Marfan syndrome have abnormalities associated with the heart and blood vessels; (4) the brain and spinal cord are surrounded by fluid contained by a membrane called the dura, and as people with Marfan syndrome get older, the dura often weakens and stretches, then begins to weigh on the vertebrae in the lower spine and wear away the bone surrounding the spinal cord; and (5) many people with Marfan syndrome develop stretch marks on their skin. Treatment options include orthopedic braces or surgery, eyeglasses or contact lenses, beta-blockers, and medication to minimize any associated pain with dural ectasia. (CR)

QUESTIONS AND ANSWERS ABOUT. . .

MARFAN SYNDROME

National Institute of Arthritis and Musculoskeletal and Skin Diseases Information Clearinghouse

NIAMS/National Institutes of Health
1 AMS Circle
Bethesda, Maryland
20892-3675

Tel. 301.495.4484
TTY 301.565.2966
Fax 301.718.6366
E-mail NIAMSInfo@mail.nih.gov
www.nih.gov/niams/

Public Health Service
Department of Health and Human Services

This fact sheet answers general questions about Marfan syndrome. It describes the characteristics of the disorder, the diagnostic process, and ways to manage symptoms. If you have additional questions after reading this fact sheet, you may wish to discuss them with your doctor.

What Is Marfan Syndrome?

Marfan syndrome is a heritable condition that affects the connective tissue. The primary purpose of connective tissue is to hold the body together and provide a framework for growth and development. In Marfan syndrome, the connective tissue is defective and does not act as it should. Because connective tissue is found throughout the body, Marfan syndrome can affect many body systems, including the skeleton, eyes, heart and blood vessels, nervous system, skin, and lungs.

Marfan syndrome affects men, women, and children, and has been found among people of all races and ethnic backgrounds. It is estimated that at least 25,000 people in the United States have the disease.

What Are the Characteristics of Marfan Syndrome?

Marfan syndrome affects different people in different ways. Some people have only mild symptoms, while others are more severely affected. In most cases, the disease progresses as the person ages. The body systems most often affected by Marfan syndrome are:

- **Skeleton**—People with Marfan syndrome are typically very tall, slender, and loose jointed. Their arms, legs, fingers, and toes may

be disproportionately long in relation to the rest of their body. A person with Marfan syndrome often has a long, narrow face, and the roof of the mouth may be arched, causing the teeth to be crowded. Other skeletal abnormalities include a sternum (breastbone) that is either protruding or indented, curvature of the spine (scoliosis), and flat feet. The joints can also be very loose.

- **Eyes**—More than half of all people with Marfan syndrome experience dislocation of one or both lenses of the eye. The lens may be slightly higher or lower than normal and may be shifted off to one side. The dislocation may be minimal, or it may be pronounced and obvious. Many people with Marfan syndrome are also nearsighted (myopic), and some can develop early glaucoma (high pressure within the eye) or cataracts (the eye's lens loses its clearness).

- **Heart and blood vessels (cardiovascular system)**—Most people with Marfan syndrome have abnormalities associated with the heart and blood vessels. The valve between the left chambers of the heart is defective and may be large and floppy, resulting in an abnormal valve motion when the heart beats. In some cases, the valve may leak, creating a "heart murmur," which a doctor can hear with a stethoscope. Small leaks may not cause any symptoms, but larger ones may cause shortness of breath, fatigue, and palpitations (a very fast or irregular heart rate). Because of faulty connective tissue, the wall of the aorta (the large artery that carries blood from the heart to the rest of the body) may be weakened and stretch, a process called aortic dilation. Aortic dilation increases the risk that the aorta will tear (aortic dissection) or rupture, causing serious heart problems or sometimes even sudden death.

- **Nervous system**—The brain and spinal cord are surrounded by fluid contained by a membrane called the dura, which is composed of connective tissue. As people with Marfan syndrome get older, the dura often weakens and stretches, then begins to weigh on the vertebrae in the lower spine and wear away the bone surrounding the spinal cord. These changes may cause only mild discomfort or may lead to radiated pain in the abdomen or legs. This is called dural ectasia.

- **Skin**—Many people with Marfan syndrome develop stretch marks on their skin, even without any weight change. These stretch marks can occur at any age and pose no health risk. However, people with Marfan syndrome are also at increased risk for developing an abdominal hernia: a weak part in the abdominal wall that can bulge and contain part of the intestines.

- **Lungs**—Although connective tissue abnormalities make the tiny air sacs within the lungs less elastic, people with Marfan syndrome generally do not experience noticeable problems with their lungs. If, however, these tiny air sacs become stretched or swollen, the risk of lung collapse may increase. Rarely, people with Marfan syndrome may have sleep-related breathing disorders such as snoring or sleep apnea (a sleep disorder characterized by brief periods when breathing stops).

What Causes Marfan Syndrome?

Marfan syndrome is caused by a defect in the gene that determines the structure of fibrillin, a protein that is an important part of connective tissue. A person with Marfan syndrome is born with the disorder, even though it may not be diagnosed until later in life. Although everyone with Marfan syndrome has the same defective gene, not everyone experiences the same symptoms to the same degree. This is called "variable expression," meaning that the defective gene expresses itself in different ways in different people. Scientists do not yet understand why variable expression occurs in people with Marfan syndrome.

The defective gene can be inherited: The child of a person who has Marfan syndrome has a 50-percent chance of inheriting the disease. Sometimes, a new gene defect (mutation) occurs during the formation of sperm or egg cells, but two unaffected parents have only a 1 in 10,000 chance of having a child with Marfan syndrome.

How Is Marfan Syndrome Diagnosed?

There is no specific test to diagnose Marfan syndrome. The doctor and/or geneticist (a doctor with special knowledge about inherited diseases) relies on a complete medical history, including

- information about any family members who may have the disorder,
- a thorough physical examination, including an evaluation of the skeletal frame for the ratio of arm/leg size to trunk size
- an eye examination, including a "split lens" evaluation
- heart tests such as an echocardiogram (a test that uses ultrasound waves to examine the heart and aorta).

The doctor may diagnose Marfan syndrome if the patient has a family history of the disease and specific problems in at least two of the body systems known to be affected. For a patient with no family history of the disease, at least three body systems must be affected before a diagnosis is made. Moreover, two of the systems must show clear signs that are relatively specific for Marfan syndrome. In some cases, a genetic analysis may be useful, but such analyses are often time consuming and may not provide any additional helpful information.

Family members of a person diagnosed with Marfan syndrome should not assume they are not affected if there is no known family history of the disorder. They should also be evaluated for signs of Marfan syndrome.

What Types of Doctors Treat Marfan Syndrome?

Because a number of body systems may be affected, a person with Marfan syndrome may be cared for by several different types of doctors. A general practitioner or pediatrician may oversee routine health care and refer the patient to specialists such as a cardiologist (a doctor who specializes in heart disorders) or an ophthalmologist (a doctor who specializes in eye disorders) as needed. Some people with Marfan syndrome are also treated by a geneticist.

What Treatment Options Are Available?

There is no cure for Marfan syndrome. To develop one, scientists may have to identify and change the specific gene responsible for the disorder before birth. However, a range of treatment options can reduce symptoms. The appropriate specialists will develop an individualized treatment program; the approach the doctor uses depends on which systems have been affected.

- **Skeletal**—Annual evaluations are important to detect any changes in the spine or sternum. This is particularly important in times of rapid growth, such as adolescence. A serious deformity can not only be disfiguring but can also prevent the heart and lungs from functioning properly. In some cases, an orthopedic brace or surgery may be recommended to limit damage and disfigurement.

- **Eyes**—Early, regular eye examinations are key to catching and correcting any vision problems associated with Marfan syndrome. In most cases, eyeglasses or contact lenses can correct the problem, although surgery may be necessary in some cases.

- **Heart and blood vessels**—Regular checkups and echocardiograms help the doctor evaluate the size of the aorta and the way the heart is working. The earlier a potential problem is identified and treated, the lower the risk of life-threatening complications. Some heart valve problems can be managed with drugs such as beta-blockers, which may help decrease stress on the aorta. In other cases, surgery to replace a valve or repair the aorta may be necessary. Surgery should be performed before the aorta reaches a size that puts it at high risk for tear or rupture.

- **Nervous system**—Although there is no way to prevent dural ectasia from developing, medication may help minimize any associated pain.

- **Lungs**—It is especially important that people with Marfan syndrome not smoke, as they are already at increased risk for lung damage. Any problems with breathing during sleep should be assessed by a doctor.

While eating a balanced diet is important to maintaining a healthy lifestyle, no vitamin or dietary supplement has been shown to help slow, cure, or prevent Marfan syndrome.

What Are Some of the Emotional and Psychological Effects of Marfan Syndrome?

Being diagnosed and learning to live with a genetic disorder can cause social, emotional, and financial stress. It often requires a great deal of adjustment in outlook and lifestyle. A person who is an adult when Marfan syndrome is diagnosed may feel angry or afraid. There may also be concerns about passing the disorder to future generations or about its physical, emotional, and financial implications.

The parents of a child diagnosed with Marfan syndrome may feel sadness, anger, and guilt. It is important for parents to know that nothing that they did caused the fibrillin gene to mutate. Parents may be concerned about the genetic implications for siblings or have questions about the risk to future children. Some children with Marfan syndrome are advised to restrict their activities. This may require a lifestyle adjustment that may be hard for a child to understand or accept.

For both children and adults, appropriate medical care, accurate information, and social support are key to living with the disease. Genetic counseling may also be helpful in understanding the disease and its potential impact on future generations.

What Is the Outlook for a Person With Marfan Syndrome?

While Marfan syndrome is a lifelong disorder, the outlook has improved in recent years. Early diagnosis and advances in medical technology have improved the quality of life for people with Marfan syndrome and lengthened their lifespan. In addition, early identification of risk factors (such as aortic dilation) allows doctors to intervene and prevent or delay complications. Advances being made by researchers provide hope for the future. With early diagnosis and appropriate management, the life expectancy for someone with Marfan syndrome is similar to that of the average person.

What Research Is Being Conducted To Help People With Marfan Syndrome?

Scientists are approaching research on Marfan syndrome from a variety of perspectives. One approach is to better understand what happens once the genetic defect or mutation occurs. How does it change the way connective tissue develops and functions in the body? Why are people with Marfan syndrome affected differently? Scientists are searching for the answers to these questions both by studying the genes themselves and by studying large family groups affected by the disease. A newly developed mouse model that carries a mutation in the fibrillin gene may help scientists better understand the disease.

Other scientists are focusing on ways to treat some of the complications that arise in people with Marfan syndrome. Clinical studies are being conducted to evaluate the usefulness of certain medications in preventing problems with the aorta. Researchers are also working to develop new surgical procedures to help improve the cardiac health of people with Marfan syndrome.

Where Can People Find Additional Information About Marfan Syndrome?

- National Marfan Foundation
  382 Main Street
  Port Washington, NY 11050
  800/8–MARFAN (800/862–7326)
  World Wide Web address: http://www.marfan.org/
  E-mail: staff@marfan.org

This organization helps people who have Marfan syndrome and related connective tissue disorders. It provides information and materials about the disorder and about how to seek appropriate medical care.

- National Institute of Arthritis and Musculoskeletal and Skin Diseases
  Information Clearinghouse
  National Institutes of Health
  1 AMS Circle
  Bethesda, MD 20892–3675
  301/495–4484
  Fax: 301/718–6366
  TTY: 301/565–2966
  NIAMS Fast Facts—For health information that is available 24 hours a day by fax, call 301/881–2731 from a fax machine telephone.
  World Wide Web address: http://www.nih.gov/niams/

This clearinghouse, a public service sponsored by the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), provides information on arthritis and musculoskeletal and skin diseases. The clearinghouse distributes patient and professional education materials and also refers people to other sources of information.

Acknowledgments

The NIAMS gratefully acknowledges the assistance of Bernadette Tyree, Ph.D., of the NIAMS; Harry C. Dietz, M.D., of The Johns Hopkins University School of Medicine in Baltimore, MD; Joel Rosenbloom, M.D., Ph.D., University of Pennsylvania, Philadelphia, Priscilla Ciccariello, M.L.S., National Marfan Foundation and Coalition for Heritable Disorders of Connective Tissue, Port Washington, NY; and the National Marfan Foundation, Port Washington, NY in developing and reviewing this fact sheet.

The National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), a part of the National Institutes of Health (NIH), leads the Federal medical research effort in arthritis and musculoskeletal and skin diseases. The NIAMS supports research and research training throughout the United States, as well as on the NIH campus in Bethesda, MD, and disseminates health and research information. The National Institute of Arthritis and Musculoskeletal and Skin Diseases Information Clearinghouse is a public service sponsored by the NIAMS that provides health information and information sources. Additional information can be found on the NIAMS Web site at http://www.nih.gov/niams/.

DN 3/99
AR–06 QA

U.S. Department of Education
Office of Educational Research and Improvement (OERI)
National Library of Education (NLE)
Educational Resources Information Center (ERIC)

NOTICE

REPRODUCTION BASIS

☐ This document is covered by a signed "Reproduction Release (Blanket) form (on file within the ERIC system), encompassing all or classes of documents from its source organization and, therefore, does not require a "Specific Document" Release form.

☑ This document is Federally-funded, or carries its own permission to reproduce, or is otherwise in the public domain and, therefore, may be reproduced by ERIC without a signed Reproduction Release form (either "Specific Document" or "Blanket").