

What Is Marfan Syndrome?

Fast Facts: An Easy-to-Read Series of Publications for the Public

Marfan syndrome is a disorder that affects connective tissue. It is usually passed from parent to child through the genes, but may result from a new gene mutation.

What Is Connective Tissue?

Connective tissue supports many parts of your body. You can think of it as a type of “glue” between cells that:

- Helps bring nutrients to the tissues
- Gives tissues form and strength
- Helps some tissues do their work.

If you have Marfan syndrome, many body systems can be affected, such as:

- Skeleton
- Heart and blood vessels
- Eyes
- Skin
- Nervous system
- Lungs.

Who Gets Marfan Syndrome?

Men, women, and children can have Marfan syndrome. It is found in people of all races and ethnic backgrounds.

What Causes Marfan Syndrome?

Connective tissue is made of many kinds of protein. One of these proteins is called fibrillin. Marfan syndrome is caused by a defect in the gene that makes fibrillin.

What Are the Symptoms of Marfan Syndrome?

Marfan syndrome affects people in different ways. Some people have only mild symptoms, and others have severe problems. Most of the time, the symptoms get worse as the person gets older.

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Skeleton

People with Marfan syndrome are often very tall, thin, and loose jointed. They may have:

- Bones (arms, legs, fingers, and toes) that are longer than normal
- A long, narrow face
- Crowded teeth because the roof of the mouth is arched
- A breastbone that sticks out or caves in
- A curved backbone
- Flat feet.

Heart and blood vessels

Most people with Marfan syndrome have problems with the heart and blood vessels, such as:

- A weak part of the aorta (the large artery that carries blood from the heart to the rest of the body). The aorta can tear or rupture.
- Heart valves that leak, causing a "heart murmur." Large leaks may cause shortness of breath, fatigue, and a very fast or uneven heart rate.

Eyes

Some people with Marfan syndrome have problems with the eyes, such as:

- Nearsightedness
- Glaucoma (high pressure within the eye) at a young age
- Cataracts (the eye's lens becomes cloudy)
- A shift in one or both lenses of the eye
- A detached retina in the eye.

Skin

Many people with Marfan syndrome have:

- Stretch marks on the skin. These are not a health problem.
- A hernia (part of an internal organ that pushes through an opening in the organ's wall).

Nervous system

The brain and spinal cord are covered by fluid and a membrane. The membrane is made of connective tissue. When people with Marfan syndrome get older, the membrane may weaken and stretch. This affects the bones in the lower backbone (spine). Symptoms of this problem include:

- Painful abdomen
- Painful, numb, or weak legs.

Lungs

People with Marfan syndrome do not often have problems with their lungs. If symptoms in the lungs do arise, they may include:

- Stiff air sacs in the lungs.
- A collapsed lung if the air sacs become stretched or swollen.
- Snoring or not breathing for short periods (called sleep apnea) while sleeping.

How Is Marfan Syndrome Diagnosed?

There is no single test to diagnose Marfan syndrome. Your doctor may use many tools to make a diagnosis:

- Medical history
- Family history (any family members who have Marfan syndrome or who died at a young age from heart problems)
- A physical exam, including the length of the bones in the arms and legs
- An eye exam, including a “slit lamp” test
- Heart tests such as an echocardiogram (a test that uses ultrasound waves to look at the heart and aorta).

What Types of Doctors Treat Marfan Syndrome?

You may need special kinds of doctors to treat the many symptoms of Marfan syndrome. Your health care team may include:

- A family doctor or pediatrician
- A cardiologist – doctor who treats heart problems
- An orthopaedist – doctor who treats bone problems
- An ophthalmologist – doctor who treats eye problems
- A geneticist – doctor who specializes in genetic diseases

How Is Marfan Syndrome Treated?

There is no cure for Marfan syndrome, but certain activities can help treat and sometimes prevent related problems.

Skeleton

- Getting a yearly exam of the spine and breastbone
- Using a back brace or having surgery for severe problems.

Heart and blood vessels

- Getting regular checkups and echocardiograms
- Seeing a doctor or going to an emergency room for pain in the chest, back, or abdomen

- Wearing a medical alert bracelet
- Taking medicine for heart valve problems
- Having surgery to replace a valve or repair the aorta if the problem is severe.

Eyes

- Getting yearly eye exams
- Wearing eyeglasses or contact lenses
- Having surgery if needed.

Lungs

- Not smoking (because it can hurt your lungs)
- Seeing a doctor if you have any problems with breathing during sleep.

Nervous system

- Taking medicine for pain if the membrane of your spinal cord swells.

Diet

- Eating a balanced diet can help you maintain a healthy lifestyle, even though no vitamin or supplement can slow, cure, or prevent Marfan syndrome.

What Do Pregnant Women with Marfan Syndrome Need to Know?

Women with Marfan syndrome can and do have healthy babies. Because pregnancy can stress the heart, pregnant women should see an obstetrician and other doctors familiar with Marfan syndrome. Your doctor can help prevent problems with your heart while you are pregnant.

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

A genetic disorder can cause social, emotional, and financial stress. It often requires changes in outlook and lifestyle. People with Marfan syndrome may feel many strong emotions, including anger and fear. They may also be concerned about whether their children will have Marfan syndrome.

It helps people with Marfan syndrome to have:

- Proper medical care
- Correct information
- Strong social support.

Genetic counseling may also help you learn about the disease and the risk of passing it on to your children.

What Research Is Being Done on Marfan Syndrome?

Marfan syndrome is the focus of intense research. Scientists are studying:

- Factors that cause heart and blood vessel problems
- The process that leads to skeletal problems
- The role of a chemical messenger called transforming growth factor-beta (TGF- β)
- Genes linked with Marfan syndrome
- Defects in the fibrillin gene
- Families with Marfan syndrome
- How Marfan syndrome affects connective tissue
- Why people with Marfan syndrome have different symptoms
- New treatments, medicines, and surgeries to help people with Marfan syndrome.

For More Information About Marfan Syndrome and Other Related Conditions:

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The information in this publication was summarized in easy-to-read format from information in a more detailed NIAMS publication. To order the Marfan Syndrome Q&A full-text version, please contact NIAMS using the contact information above. To view the complete text or to order online, visit <http://www.niams.nih.gov>.