



Marfan Syndrome Facts

WHAT IS MARFAN SYNDROME?

Marfan syndrome (MFS) is a disorder of connective tissue. Connective tissue holds all parts of the body together and helps control how the body grows. Because connective tissue is found throughout the body, MFS features can occur in many different parts of the body.

MFS features are most often found in the heart, blood vessels, bones, joints, and eyes. Sometimes, the lungs and skin are also affected. MFS does not affect intelligence.

WHAT CAUSES MARFAN SYNDROME?

MFS is caused by a defect (mutation) in the gene that tells the body how to make fibrillin-1—a protein that is an important part of connective tissue. This mutation creates different MFS features and causes medical problems for people with MFS.

WHO HAS MARFAN SYNDROME?

About 1 in 5,000 people have MFS. This includes men and women of all races and ethnic groups. People can inherit MFS, meaning that they get the mutation from a parent who has MFS. This happens to about 3 out of 4 people with MFS. Other people have a spontaneous mutation, meaning that they are the first in their family to have MFS. People with MFS have a 1 out of 2 chance of passing the mutation on each time they have a child.

People are born with MFS but may not notice any features until later in life. However, MFS features can appear at any age, including in infants and young children. MFS features and medical problems can get worse as people age.

WHAT ARE MARFAN SYNDROME FEATURES?

MFS features occur in many different parts of the body. Rarely, a person has every feature. Some MFS features are easy to see. Other features, such as heart problems, are hidden and need special tests to find them. It is important that a person with MFS features see a doctor who knows about MFS.

Here are the most common MFS features:

Heart and blood vessels (Cardiovascular system)

- Enlarged or bulging aorta, the main blood vessel that carries blood from the heart (aortic dilation or aneurysm)
- Separation of the layers of the aorta that can cause it to tear (aortic dissection)
- “Floppy” mitral valve (mitral valve prolapse—MVP)



National Marfan Foundation
Education • Research • Support

Serving the needs
of people with
Marfan syndrome
and related disorders

22 Manhasset Avenue
Port Washington, NY 11050

516-883-8712
800-8-MARFAN
516-883-8040 (fax)

www.marfan.org

Bones and Joints (Skeletal system)

- Long arms and legs
- Tall and thin body type
- Curvature of the spine (scoliosis or kyphosis)
- Chest sinks in (pectus excavatum) or sticks out/pigeon breast (pectus carinatum)
- Long, thin fingers
- Flexible joints
- Flat feet
- Teeth that are too crowded

Eyes (Ocular system)

- Severe nearsightedness (myopia)
- Dislocated lens of the eye
- Detached retina
- Early glaucoma or cataracts

Other body systems

- Stretch marks on the skin, not explained by pregnancy or weight gain
- Sudden collapse of the lung (spontaneous pneumothorax)
- Widening or ballooning of the dural sac surrounding the spinal cord (dural ectasia)

WHAT IS LIFE LIKE FOR A PERSON WITH MARFAN SYNDROME?

While there is no cure for MFS, advances in medical care are helping people live longer and enjoy a good quality of life. Research is also finding new ways to treat people with MFS. Most people with MFS can work, go to school, and enjoy active hobbies.

It is very important that people with MFS get treatment and follow medical advice. One reason is that heart problems can cause sudden death if they are not treated. Early diagnosis means helpful treatment can begin early in life. People with MFS should not play active team sports such as football, soccer, or basketball. They should not lift heavy objects when at work, home or the gym.

WHAT SHOULD YOU DO IF YOU SUSPECT MARFAN SYNDROME?

Look for a doctor who knows about MFS so you can be checked for this disorder. Keep in mind that you can have MFS features but not have the disorder. The only way to know for sure is to be checked by a doctor who understands MFS.

HOW CAN YOU LEARN MORE ABOUT MARFAN SYNDROME?

MFS is a complex disorder, with many features that affect different parts of the body. Here are some ways to learn more about MFS:

- Call the National Marfan Foundation (NMF) Resource Center at 1-800-862-7326 ext. 26. When you call, you will speak with a staff member who can answer questions and mail you information. They can also suggest ways to find a doctor who knows about MFS.
- Go to the NMF website at <http://www.marfan.org> You can find more information on the “About Marfan Syndrome” and “Living with Marfan Syndrome” pages.
- Talk with a doctor. Sometimes it helps to take MFS information with you when you visit the doctor. Your doctor can go to the special section for doctors on the NMF website by clicking on “Medical Professionals.”