



# Marfan Syndrome Facts



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## 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. Always, a person with MFS has at least 3 features in 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 3 or more 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)

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**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
- Early cataracts

**Other body systems**

- Stretch marks on the skin, not explained by pregnancy or weight gain
- Sudden collapse of the lung (spontaneous pneumothorax)

**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.

**WAYS TO LEARN MORE**

- Contact the Marfan Syndrome Support Group Ireland at [info@marfan.ie](mailto:info@marfan.ie).
- Talk to your doctor. Sometimes it helps to use information like this fact sheet when you speak with the doctor.
- Visit the Marfan Syndrome Support Group website at [www.marfan.ie](http://www.marfan.ie).