

MARFAN SYNDROME: THE BASIC FACTS

Marfan syndrome is a life-threatening genetic disorder of the body's connective tissue. Knowing the signs of Marfan syndrome, getting a proper diagnosis, and receiving the necessary treatment can enable people with Marfan syndrome to live a long and full life.

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Our community of experts estimates that nearly half of the people who have Marfan syndrome don't know it. Without proper diagnosis and treatment, they are at high risk for an early sudden death.

Marfan syndrome affects our connective tissue, which helps to hold the body's cells and tissues together. It also regulates how our bodies grow.

There are also several disorders related to Marfan syndrome that cause people to struggle with the same or similar physical problems, and anyone affected by these disorders also needs an early and accurate diagnosis.

What are the features of Marfan Syndrome?

Some features of Marfan syndrome are easier to see than others. These include:

- Long arms, legs, and fingers
- Tall and thin body type
- Curved spine
- Sunken or protruding chest
- Flexible joints

- Flat feet
- Crowded teeth
- Unexplained stretch marks on the skin

Harder-to-detect signs include:

- Heart problems, especially related to the aorta, the large blood vessel that carries blood away from the heart

Other signs include:

- Sudden collapse of a lung
- Eye problems, including severe nearsightedness, dislocated lens, detached retina, early glaucoma, and early cataracts.

What causes Marfan syndrome?

Marfan syndrome is caused by a change (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 Marfan syndrome features and causes medical problems.

Who has Marfan syndrome?

- About 1 in 5,000 people have Marfan syndrome. This includes men and women of all races and ethnic groups.
- People can inherit Marfan syndrome; that is, they get the mutation from a parent who has Marfan syndrome. This happens in about 3 out of 4 people with Marfan syndrome. Other people have a spontaneous mutation, meaning that they are the first in their family to have Marfan syndrome.
- People with Marfan syndrome have a 50 percent chance of passing the mutation on each time they have a child.
- People are born with Marfan syndrome, but they may not notice any features until later in life. However, Marfan syndrome features can appear at any age, including in infants and young children. Marfan syndrome features and medical problems can get worse as people age.

What is life like for someone with Marfan syndrome?

Advances in medical care help people live longer and enjoy a good quality of life if they are diagnosed and treated. Most people with Marfan syndrome can work, go to school, and enjoy active hobbies. It is very important that people with Marfan syndrome get treatment and follow medical advice; otherwise, heart problems can cause sudden death. With an early diagnosis, helpful medical treatment can begin early in life. People with Marfan syndrome also need to adapt their physical activity to stay safe. In general, they should not play active team sports such as football, soccer, or basketball. In addition, they should not lift heavy objects when at work, home, or the gym.

What should you do if you suspect Marfan syndrome?

If you suspect that you or a loved one have Marfan syndrome, look for a doctor who knows about Marfan syndrome and make an appointment to be evaluated. Keep in mind that you can have Marfan syndrome features, but not meet the requirements for a firm diagnosis. Regardless of the diagnosis, it is important to get treatment for the features you have and follow-up with your doctor as he/she recommends.

How is Marfan syndrome diagnosed?

A Marfan syndrome diagnosis can often be made after exams of several parts of the body by doctors experienced with connective tissue disorders, including:

- A detailed medical and family history, including information about any family member who may have the disorder or who had an early, unexplained, heart-related death.
- A complete physical examination.

You should also have tests to identify Marfan features that are not visible during the physical exam, including:

- Echocardiogram. This test looks at your heart, its valves, and the aorta (blood vessel that carries blood from the heart to the rest of the body).
- An eye examination, including a “slit lamp” evaluation to see if the lenses in your eyes are out of place. It is important that the doctor fully dilates the pupils.

Genetic testing can provide helpful information in some cases.

- For individuals with a family history of Marfan syndrome, genetic testing can help confirm or rule out the diagnosis of Marfan syndrome in family members who may be at risk.
- Some of the features of Marfan syndrome can be found in disorders related to Marfan syndrome; therefore, genetic testing may be helpful when a diagnosis cannot be determined through an exam by doctors.

It is possible for you to have one or more features of Marfan syndrome, but not enough for you to have a Marfan syndrome diagnosis. You may need additional exams by other doctors and additional genetic testing to see if you have a disorder that is related to Marfan syndrome.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.