

MARFAN SYNDROME DIAGNOSIS

Marfan syndrome is a serious condition, and some complications are potentially life-threatening. Advances in medical care have made it possible for people with Marfan syndrome to live a normal lifespan if they are diagnosed and treated properly.

Marfan syndrome most often affects the heart, blood vessels, bones, joints, and eyes.



What is Marfan syndrome?

Marfan syndrome 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, Marfan syndrome features can occur in many different parts of the body. Most often the condition affects the heart, blood vessels, bones, joints, and eyes. Sometimes, the lungs and skin are also affected. Marfan syndrome does not affect intelligence.

What causes Marfan syndrome?

Marfan syndrome is caused by a change (mutation) in the FBN1 gene that tells the body how to make fibrillin-1, a protein that is an important part of connective tissue. This change creates Marfan syndrome features and causes medical problems.

How is Marfan syndrome inherited?

- About 75 percent of people with Marfan syndrome inherit it from a parent who has the condition.

- About 25 percent of people with Marfan syndrome do not inherit it from a parent; their condition is a result of a spontaneous mutation and 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.
- Marfan syndrome affects about 1 in 5,000 men and women of all races and ethnic groups.

What are the features of Marfan syndrome?

Marfan syndrome features occur in many different parts of the body. It is rare that a person has every feature. Some Marfan syndrome features are easy to see. Other features, such as heart problems, require special tests to find them. The common features include:

Heart and blood vessels

- Enlarged or bulging aorta, the main blood vessel that carries blood away from the heart (aortic dilation or aneurysm)
- Tear in the inner wall of the aorta that causes blood to flow between the layers of the aorta wall (aortic dissection)
- “Floppy” mitral valve (mitral valve prolapse)

Bones and joints

- Disproportionately 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
- High arched palate
- Teeth that are too crowded

Eyes

- 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/loss
- Sudden collapse of the lung (spontaneous pneumothorax)
- Widening or ballooning of the dural sac surrounding the spinal cord (dural ectasia)

If you have Marfan syndrome, it is something you are born with, although you may not notice any features until later in life. Marfan syndrome features can appear at any age—including in infants, teens, and older adults—and they can get worse as people age.

What should you do if you suspect Marfan syndrome?

If you suspect that you or a family member may have Marfan syndrome, find a doctor who is knowledgeable about the condition. Since Marfan syndrome is rare, not all doctors know about it. Ideally, the diagnostic process should be coordinated by a medical geneticist (a doctor who specializes in genetic conditions). If you cannot see a medical geneticist, visit a cardiologist (heart doctor). Make sure the cardiologist has treated people who have Marfan syndrome.

It is possible to have some Marfan syndrome features, but not enough for a confirmed diagnosis. The only way to know for sure is to be checked by a doctor who understands Marfan syndrome.

To find a doctor:

- Call our help center at 800-862-7326, ext. 126
- Request our directory of medical institutions (through our website, Marfan.org)
- Ask your primary doctor for a referral
- Call the doctor referral service at your local hospital
- Contact your insurance company

Find out about your family's medical history. You can download the family health history kit from our website to compile the details, including:

- Past illnesses, operations, and hospitalizations
- Medications
- Reasons you think you or your family member might have Marfan syndrome
- Family members who have, or might have, Marfan syndrome
- Family members who died of a heart or vascular problem

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. The evaluation includes:

- 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 before doing this test.

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.

Should you consider genetic testing?

The use of genetic testing for the diagnosis of genetic disorders can be very complicated. Input from a geneticist or genetic counselor may be necessary to achieve a full understanding of the capabilities and limitations of genetic testing for Marfan syndrome. A genetic test alone cannot tell you if you do or do not have Marfan syndrome. Here are some situations in which genetic testing may be helpful:

- A child who does not have outward Marfan features, but has a parent with Marfan syndrome, should be monitored on an ongoing basis if the genetic test for the child is positive.
- Individuals who have one of the key features of Marfan syndrome (aortic dilatation or dissection or dislocated lens), but no other obvious signs of the disorder, require additional monitoring if the genetic test is positive.
- When a diagnosis cannot be determined based on clinical evaluation alone, a genetic test may be helpful in considering differential diagnosis (diagnosis of a related condition).
- Families with a history of aortic aneurysm or dissection may choose genetic testing to identify individuals in the family who may be at risk.
- Adults with Marfan syndrome who are considering having children may consider genetic testing to pursue pre-implantation or prenatal diagnosis (options for having a baby without Marfan syndrome).

More information can be found in our resource on genetic testing for Marfan syndrome.

What are the possible outcomes to the diagnostic evaluation?

- Marfan syndrome: If you have enough features of the disorder to meet the diagnostic criteria for Marfan syndrome.
- Non-specific connective tissue disorder: If you don't have enough features of Marfan syndrome. Follow-up echocardiograms may be recommended.

- Potential Marfan syndrome: If you have a confirmed FBN1 mutation (through genetic testing), but your aortic root measurements are too small to meet criteria for a Marfan syndrome diagnosis.
- Another genetic disorder: If you do not meet the diagnostic criteria for Marfan syndrome, but instead have another genetic disorder, such as Ehlers-Danlos syndrome, Loeys-Dietz syndrome, MASS phenotype, familial aortic aneurysm, Sticklers syndrome, ectopia lentis syndrome, or mitral valve prolapse syndrome.
- No specific diagnosis: if you have Marfan features, but do not meet the diagnostic criteria for any known disorder.

Many people with Marfan syndrome features (whether they have a diagnosis or not) need medical treatment and follow-up care. Make sure to talk with your doctor about the care that is right for you.

What are some of the emotional and psychological effects of Marfan syndrome?

People with a genetic disorder sometimes face social and emotional issues. It often requires change in outlook and lifestyle. An adult who receives a Marfan syndrome diagnosis may feel angry or afraid. There may be concerns about passing the disorder to children, as well as worries about genetic implications for siblings.

The parents and siblings 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 child to have the condition.

Some people with Marfan syndrome are advised to restrict their activities. This may require a lifestyle adjustment that can be hard to accept.

It is easier for children and adults to live with Marfan syndrome when they have appropriate medical care, accurate information, and social support. Genetic counseling can increase your understanding of the disorder and its potential impact. You can also contact our help center to learn about our online and local support.

NOTE: If you have been diagnosed with Marfan syndrome and have any of the following features, it is urgent that you talk to your doctor about Loeys-Dietz syndrome, a relatively recently discovered condition that requires more aggressive monitoring and treatment.

- Arteries that twist and wind (arterial tortuosity)
- Aneurysms and dissections in arteries other than the aorta
- Widely-spaced eyes (hypertelorism)
- Wide or split uvula (the tissue that hangs down in the back of the throat)
- Cleft palate (when the roof of the mouth is split at birth)
- Club foot (when the foot is turned inward and upward at birth)
- White of the eye looks blue or gray
- Premature fusion of the bones of the skull (craniosynostosis)

- Heart defects at birth, such as atrial septal defect, patent ductus arteriosus, bicuspid aortic valve
- Features in the skin, such as easy bruising, wide scars, soft skin texture, and translucent skin (when it looks almost see-through)
- Gastrointestinal problems (stomach and intestine problems), such as difficulty absorbing food and chronic (comes and goes but never really goes away) diarrhea, abdominal pain, and/or gastrointestinal bleeding and inflammation
- Food and environmental allergies
- Rupture of the spleen or bowel
- Rupture of the uterus during pregnancy
- Instability or malformation of the spine in the neck (cervical spine instability)
- Poor mineralization of the bones (osteoporosis) that can make the bones more likely to break
- Collection of fluid in the brain (hydrocephalus)
- Part of the brain (cerebellum) with an abnormal shape (Chiari I malformation)

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.