CONDITION

# Marfan syndrome

Marfan syndrome is an inherited connective tissue condition that can affect the aorta, eyes, bones, and joints.

**WHAT IT IS**
Inherited condition affecting connective tissue throughout the body

**BODY SYSTEM**
Heart and circulation, eyes, bones and joints

**MAIN SYMPTOMS**
Long limbs, flexible joints, chest changes, blurred vision

**WHAT CAUSES IT**
Inherited or new FBN1 gene change

**HOW IT IS DIAGNOSED**
Clinical criteria, heart imaging, eye examination, genetic testing

**MAIN TREATMENT**
Lifelong monitoring, medicines, and preventive aortic surgery

**CAN IT BE CURED?**
No - genetic change remains lifelong

**IS IT AN EMERGENCY?**
Sometimes - sudden severe chest or back pain

**LONG-TERM OUTLOOK**
Often manageable with monitoring and timely treatment

## Overview

Marfan syndrome is a lifelong condition affecting connective tissue. Connective tissue supports and strengthens structures throughout the body, including blood vessels, eyes, bones, joints, skin, and lungs.

The most important effect is possible weakening and enlargement of the aorta. The aorta is the main artery carrying blood away from the heart. Enlargement often causes no symptoms, but it can increase the risk of a tear in the aortic wall. Regular imaging allows changes to be found before they become an emergency.

Marfan syndrome affects people differently. Some people have noticeable skeletal or eye features but little heart involvement. Others need close monitoring or treatment for changes in the aorta. Features may also become clearer as a child grows.

Having long limbs, flexible joints, or short-sightedness does not by itself mean someone has Marfan syndrome. Diagnosis requires a detailed assessment because several other connective tissue conditions can look similar.

## Symptoms

Marfan syndrome can affect several parts of the body. A person may have only some of its features.

Skeletal features can include:

Eye problems may include severe short-sightedness or blurred vision. The lens inside the eye can move out of its normal position, known as lens dislocation or ectopia lentis. This is an important feature of Marfan syndrome. Less commonly, problems such as early cataracts, glaucoma, or retinal detachment can occur.

Heart and blood vessel changes usually do not cause early symptoms. The aortic root, the first part of the aorta, may slowly enlarge. A heart valve may also become loose and leak, sometimes causing a racing heartbeat, tiredness, breathlessness, or awareness of the heartbeat.

Some people develop stretch marks unrelated to major weight change. Others have lung problems, including a higher risk of a collapsed lung. Widening of the tissue around the lower spinal cord, called dural ectasia, can contribute to lower back pain, leg pain, or headaches in some people.

- Long arms, legs, fingers, or toes compared with the rest of the body
- A breastbone that pushes inward or outward
- Curvature of the spine, called scoliosis
- Flat feet, flexible joints, or reduced elbow movement
- A high, narrow roof of the mouth and crowded teeth

## Causes and risk factors

Marfan syndrome is usually caused by a harmful change in the FBN1 gene. This gene provides instructions for making fibrillin-1, a protein that helps connective tissue stay strong and flexible.

The condition follows an autosomal dominant inheritance pattern. This means a person with

Marfan syndrome has a 1 in 2 chance of passing the gene change to each child. The outcome is separate for every pregnancy.

Some people are the first in their family to have Marfan syndrome because the gene change occurred for the first time in them. This is called a new or de novo genetic change.

Lifestyle, parenting, diet, or something that happened during pregnancy does not cause Marfan syndrome. People with the same gene change, including members of one family, may have different features and levels of aortic risk.

## Diagnosis

Diagnosis is based on a combination of medical history, physical findings, heart imaging, an eye examination, and sometimes genetic testing. Clinicians use an agreed framework called the revised Ghent criteria, which gives particular importance to aortic enlargement, lens dislocation, family history, and an FBN1 gene change.

An echocardiogram uses sound waves to examine the heart, valves, and aortic root. CT or MRI scans may be used to view more of the aorta or obtain detailed measurements.

An eye specialist can use a slit-lamp examination to look for lens dislocation and other eye problems. Measurements and examination may also identify skeletal features. These can be combined into a systemic score, but that score is only one part of the assessment.

A blood or saliva test may find a disease-causing FBN1 change. Genetic testing can support or confirm the diagnosis in the right clinical setting and can help test relatives. However, results require careful interpretation. Not every uncertain gene finding causes Marfan syndrome, and a negative result does not always settle the diagnosis.

Assessment may take time, especially in children whose features are still developing. Other inherited aortic and connective tissue conditions may need to be considered.

## Treatment and management

There is no treatment that removes the underlying genetic change. Management aims to protect the aorta, treat eye or skeletal problems, and support everyday activity.

Regular imaging tracks the width of the aorta and how quickly it is changing. The timing of follow-up is based on age, previous measurements, growth rate, family history, and any past surgery. Blood pressure is also monitored because greater pressure puts more strain on the aortic wall.

Medicines that reduce blood pressure and stress on the aorta are commonly used. These may include beta blockers or angiotensin receptor blockers. The choice and monitoring plan are individual decisions, particularly during pregnancy or when other health conditions are present.

Planned surgery can replace an enlarged section of the aortic root before it tears. The decision is based on more than one measurement. Aortic size, recent growth, body size, family history, valve function, pregnancy plans, and the person's overall situation may all matter. People who have surgery still need lifelong imaging because other parts of the aorta can change.

Glasses or contact lenses may correct short-sightedness. Lens or retinal problems sometimes require surgery. Scoliosis, chest-wall differences, foot problems, and joint pain may be managed with physiotherapy, supports, pain treatment, or surgery when symptoms are significant.

Exercise is usually encouraged, but activities that cause intense straining, large blood-pressure surges, or forceful collisions may be unsuitable for some people. An appropriate activity plan is based on aortic measurements, heart valve function, treatment, and previous surgery.

## Complications

The main serious complication is progressive enlargement of the aorta. This can lead to an aneurysm, meaning a weakened and enlarged section, or an aortic dissection, in which blood enters a tear between layers of the artery wall. A rupture is less common but life-threatening.

Other possible complications include leaking heart valves, abnormal heart rhythms, heart failure, retinal detachment, glaucoma, cataracts, collapsed lung, worsening scoliosis, and persistent pain related to joints or dural ectasia.

These complications are not inevitable. Routine monitoring is intended to find treatable changes early, including problems that have not caused symptoms.

## Prevention

Marfan syndrome cannot be prevented because it results from a genetic change. Nothing a parent did caused it.

Complications can sometimes be prevented or reduced through regular aortic imaging, blood-pressure management, suitable physical activity, eye care, and planned surgery when needed.

People considering pregnancy may choose genetic counselling. This can explain inheritance,

testing options, and reproductive choices without directing a person toward any one decision.

## Living with it and what to expect

Marfan syndrome remains present throughout life, but its effects may change. Skeletal features often become more noticeable during growth. Eye problems may remain stable or need treatment. The aorta may stay at a similar size for years or may gradually enlarge without causing symptoms.

Most people need lifelong follow-up with clinicians experienced in inherited aortic conditions. Children may need coordinated heart, eye, and skeletal care as they grow. Adults continue aortic monitoring even if they feel well or have had successful surgery.

Many people attend school, work, travel, exercise, and have families. Some need adjustments because of pain, fatigue, eyesight, physical restrictions, or repeated appointments. Emotional support can also help if uncertainty about the aorta or family inheritance becomes difficult.

Pregnancy places extra strain on the heart and aorta. Someone with Marfan syndrome who is pregnant or planning pregnancy usually needs assessment before conception and coordinated care during pregnancy and after birth. Risk is strongly influenced by the condition of the aorta and any previous surgery.

Outcomes have improved with regular monitoring, blood-pressure treatment, and preventive surgery. An individual course cannot be predicted from appearance alone.

## When to get medical help

Arrange routine follow-up if you have Marfan syndrome and develop new breathlessness, palpitations, reduced exercise tolerance, persistent back or joint pain, or changes in vision. These symptoms do not always indicate a serious complication, but they may need assessment.

Seek urgent same-day eye care for sudden flashes of light, many new floaters, or a dark curtain or shadow across the vision. These can be signs of retinal detachment.

Call emergency services for:

Do not travel to hospital without emergency help if an aortic emergency is possible.

- Sudden, severe chest, upper back, or abdominal pain, especially if tearing or spreading
- Sudden breathlessness, collapse, fainting, or cold sweating

- Sudden weakness, facial drooping, speech difficulty, or loss of coordination
- Sudden chest pain with difficulty breathing, which may indicate a collapsed lung

## Common questions

Can Marfan syndrome be cured?

No. The genetic change remains throughout life. Treatment can reduce stress on the aorta, correct some eye or skeletal problems, and lower the risk of serious complications.

Does being tall with long fingers mean I have Marfan syndrome?

No. These features are common in people without Marfan syndrome. Diagnosis considers the aorta, eyes, skeleton, family history, and genetic findings together.

Can I exercise if I have Marfan syndrome?

Most people can remain active, but the safest type and intensity are individual. Moderate activities may be suitable, while heavy straining, intense competition, or collision sports may place too much stress on the aorta for some people.

Can Marfan syndrome skip a generation?

A person who does not inherit the family's disease-causing FBN1 change does not usually pass that change to their children. However, mild or unrecognised features can sometimes make it appear that the condition skipped a generation.

Can someone with Marfan syndrome have a baby?

Yes, but pregnancy may increase aortic risk. Assessment before pregnancy helps review the aorta, medicines, inheritance, and monitoring needs. Each child of an affected parent has a 1 in 2 chance of inheriting the gene change.

Will everyone with Marfan syndrome need heart surgery?

No. Surgery is considered when aortic enlargement or another heart problem reaches a level where planned treatment is safer than continued observation. Some people never reach that point, while others need one or more operations.

## Sources

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