
Marfan syndrome

Summary

- Marfan syndrome is a genetic disorder of the body's connective tissue, which may affect the heart, eyes, skeleton and lungs.
 - The most serious effects include those on the cardiovascular system, particularly the heart valves and aorta.
 - There is no cure, but the syndrome can be managed with careful monitoring, medication, surgery and lifestyle changes, including the avoidance of heavy physical activity and contact sports.
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Marfan syndrome is caused by a change in a gene that affects connective tissue. Connective tissue offers support to many structures, including bones, tendons, ligaments, cartilage, heart valves and blood vessels.

While most cases of Marfan syndrome are inherited, some are due to a spontaneous change in a gene, with no family history. Marfan syndrome can be mild to severe, and may become worse with age, depending on which area is affected and to what degree.

In Marfan syndrome, the heart is often affected. The aorta, the major artery of the body, may be more dilated (widened) than average. This can cause the aorta to tear or bulge under pressure – for example, during strenuous exercise or childbirth. This can cause bleeding into the various layers of aorta or increase the risk of an aneurysm.

If the heart valves are affected, blood may seep backwards and place extra strain on the heart. The heart may then enlarge to cope with the extra workload.

Other common problems include spinal curvature (scoliosis) and nearsightedness. Recent estimates suggest one in 3,000–5,000 people have Marfan syndrome. Men, women and different ethnic groups all have a similar risk. There is no cure, but the potential complications can be managed, so early and accurate diagnosis is essential.

Symptoms of Marfan syndrome

The symptoms of Marfan syndrome differ from one person to the next, depending on which body part is affected and to what degree. Some people may not even realise they have the condition, because their features are either mild or not obvious.

Symptoms may include:

- family history of the condition
 - long, narrow face
 - proportionately small lower jaw and narrow, highly arched palate
 - usually, tall lean stature with disproportionately long arms and legs
 - thin, long fingers and toes (arachnodactyly)
 - sunken or protruding chest
 - loose and flexible joints, often prone to dislocation
 - scoliosis (sideways curve in the spine)
 - visual problems, such as nearsightedness (myopia) or dislocation of the ocular lens
 - enlarged aorta, mitral valve prolapse
 - stretch marks (striae) –often appear at an early age, but they are not related to weight fluctuation
 - pneumothorax – spontaneous collapse of the lung
 - dural ectasia – enlargement of the outer membrane around the brain and spinal cord. Usually, the lower
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portion of the spine is affected and causes no symptoms.

Complications of Marfan syndrome

A heart valve (such as the mitral valve) affected by Marfan syndrome is not as stiff and resilient as it should be, which allows it to collapse backwards under pressure. This is called mitral valve prolapse. To cope with the backflow, the heart has to work much harder to pump the blood around the body.

People whose Marfan syndrome affects their heart valves are at increased risk of **endocarditis**, which is an infection of the heart wall or valves. This can have life-threatening consequences. Before various types of surgery are performed, a preventative course of antibiotics to prevent bacterial infection is recommended to minimise this risk.

Causes of Marfan syndrome

Marfan syndrome is caused by a change in the *FBN1* gene. A child of a person with Marfan syndrome has a one-in-two chance of inheriting the condition. Around three out of four cases of Marfan syndrome are inherited from one affected parent.

Around one in four cases of Marfan syndrome are due to a spontaneous change in the *FBN1* gene at some stage before conception, for reasons unknown. The changed gene affects the composition of fibrillin, an important substance that contributes to the integrity of connective tissue.

Research into the condition is ongoing, with the hope of devising more specific genetic tests for earlier and more accurate diagnosis. With technological advances in genetic testing, other genes have recently been found that might account for a small proportion of cases and other condition-related symptoms. Research is ongoing to understand this better.

Diagnosis of Marfan syndrome

Gene testing can be organised for some families who meet the diagnostic criteria. A specific collection of major characteristic features must be present to establish the syndrome. A confirmed diagnosis requires coordinated evaluation by a cardiologist, ophthalmologist, orthopaedic surgeon and geneticist, all experienced with Marfan syndrome.

The condition is diagnosed using a number of tests, including:

- physical examination
- family history
- echocardiogram (picture of the heart using sound waves)
- eye examination, using a slit lamp to detect dislocated lenses
- skeletal tests such as x-rays
- comparison of results with a genetic database.

Over time, it has become apparent that a small proportion of people previously thought to have Marfan syndrome actually have a related but distinct condition called Loeys-Dietz syndrome. Loeys-Dietz syndrome has many features in common with Marfan syndrome; but people with Loeys-Dietz syndrome have more aggressive blood vessel disease and do not develop the eye complications seen in Marfan syndrome.

Treatment for Marfan syndrome

There is currently no cure for Marfan syndrome. Depending on which body parts are affected and to what degree, regular monitoring and treatment options may include:

- lifestyle changes
- regular echocardiogram tests to monitor heart and aorta size and function
- regular eye examinations and treatment
- regular check-ups to monitor the skeleton and offer corrective treatment if necessary. This is particularly important during the growth spurt of puberty

- medication, such as beta-blockers, to reduce blood pressure and relieve the strain on the cardiovascular system
- measures such as a course of antibiotics to prevent infection prior to any type of surgery involving the teeth, genitals or urinary system to reduce the risk of heart valve problems, including infection (endocarditis)
- heart surgery, such as surgical repair of valves and aorta.

Living with Marfan syndrome

The expected lifespan of a person with Marfan syndrome used to be around the mid-40s age group, but now extends into the 70s – which is comparable to the general population.

This improved lifespan is mainly due to:

- improved awareness of Marfan syndrome across health professionals
- early diagnosis
- advances in medical management and medication
- improved and more effective surgical techniques
- appropriate lifestyle changes.

Lifestyle changes

If you have Marfan syndrome, it is important to be well informed about the condition and how it can be managed. General wellbeing and quality of life for individuals with Marfan syndrome can be improved by making lifestyle choices that reduce the stress on their body. A healthcare professional can advise you on suitable lifestyle suggestions.

Depending on which body parts are affected and to what degree, such changes may include:

- avoiding contact sports
- avoiding physical activities that depend on isometric exercise, such as weight training or moving heavy objects
- doing only gentle, moderate exercise
- having specialised counselling. A woman who has a **dilated** aorta requires specialised counselling about the higher risks involved when considering a pregnancy, due to the associated strain on this major artery and the risk of rupture, particularly during childbirth. Cardiac assessment pre-pregnancy and repeatedly throughout pregnancy is recommended.

Genetic counselling and Marfan syndrome

If you or a family member have been diagnosed with Marfan syndrome, or if it runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Marfan syndrome and what causes it, how it is inherited, and what a diagnosis means for you or your child's health and development. Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Marfan syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by MCKD.

Where to get help

- Your **GP (doctor)**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**
- **Genetic Support Network Victoria** Tel. **(03) 8341 6315**

- **Inherited Diseases Clinic**, Alfred Hospital Melbourne Tel. **(03) 9076 3263**

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