

## Marfan syndrome

Marfan syndrome is caused by a faulty 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, the remainder are due to a spontaneous gene mutation, 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 3–5,000 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 individual 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. These may include:

- Family history of the condition
- Long, narrow face
- Proportionately small lower jaw and narrow, highly arched palate
- Usually, tall lean stature with disproportionate 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
- Striae – stretch marks 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 portion of the spine is affected and causes no symptoms.

### Heart valve problems can be a complication

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.

## Gene abnormality

The child of a person with Marfan syndrome has a one-in-two chance of inheriting the disorder. Around three out of four cases of Marfan syndrome are inherited from one affected parent. The remainder of cases are due to a spontaneous gene mutation at some stage before conception, for reasons unknown. The abnormal gene affects the composition of fibrillin, an important substance that contributes to the integrity of connective tissue.

The gene responsible for Marfan syndrome was identified in 1991. Research into the condition is ongoing, with the hope of devising genetic tests for earlier and more accurate diagnosis. Recently, other genes have been found that might account for a small proportion of cases and cause-related conditions.

## Diagnosis of Marfan syndrome

Gene testing is not readily available, but 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 the 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
- Genetic data.

## Treatment for Marfan syndrome

There is 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
- Yearly echocardiogram tests to monitor heart and aorta size and function
- Yearly 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
- Drugs, such as beta-blocker medication, to reduce blood pressure and relieve the strain on the cardiovascular system. New medications are now being used that better treat the underlying pathology in this condition
- 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.

## Life expectancy has improved

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
- Early diagnosis
- Advances in medical management and medication
- Surgical techniques
- Careful living.

## Lifestyle changes

If you have Marfan syndrome, it is important to be well informed about the condition and how it can be managed. If you are aware of your limitations, it will help you accept the particular lifestyle changes that will reduce the stress on your body. Be advised by your health care provider.

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

- No contact sports
- No physical activities that depend on isometric exercise, such as weight training or moving heavy objects
- Gentle, moderate exercise only
- 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.

## Where to get help

- Your doctor
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Marfan Syndrome Clinic, Alfred Hospital Melbourne Tel. (03) 9076 3263
- Marfan Association Victoria Tel. (03) 5784 1357 or (03) 9740 3651
- Genetic Support Network Victoria Tel. (03) 8341 6315

## Things to remember

- 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.

**This page has been produced in consultation with, and approved by:**

Genetic Health Services Victoria

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