

If couples are interested in prenatal diagnosis they should be seen at their local genetics centre as testing may be possible in some cases.

For more information

If you need more advice about any aspect of Marfan syndrome, you are welcome to contact:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

The Marfan Association UK

Rochester House

5 Aldershot Road

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01252 617320 (answerphone)

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Seen in clinic by

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Marfan Syndrome



Patient Information Leaflet

What is Marfan syndrome?

Marfan syndrome is an inherited disorder which can affect many parts of the body including the skeleton, lungs, eyes, heart and blood vessels. The condition is caused by a change ('mutation') in a gene which produces fibrillin. Fibrillin is a fine fibre which acts like scaffolding within 'connective tissue' throughout the body.

It is estimated that approximately 1 in 5000 people in Britain have Marfan syndrome.

How is Marfan syndrome diagnosed?

The diagnosis of Marfan syndrome is made on the basis of family history and physical examination, focusing particularly on the eyes, skeleton and heart. Tests such as an echocardiogram (a sound-wave picture of the heart) and eye examination (using a special piece of equipment called a slit lamp) are often helpful in making a diagnosis. Although it is possible to do a genetic test for Marfan syndrome, in practice this is still difficult and is not routinely done at present.

What medical problems are associated with Marfan syndrome?

Heart and blood vessels

The most serious problems associated with Marfan syndrome involve the heart and blood vessels. The mitral valve may billow backwards when the heart contracts (mitral valve prolapse). Rarely this may lead to problems such as an irregular heart rhythm. The aorta (the main artery carrying blood away from the heart) is often wider and more fragile in patients with Marfan syndrome. This widening can increase over time and result in leakage if a tear develops in the wall of the aorta, a condition called aortic dissection. If the aorta becomes greatly widened or torn, surgery may be needed.

For this reason, it is very important that everyone suspected of having Marfan syndrome should have an echocardiogram. People who are affected with Marfan syndrome should have an echocardiogram on a regular basis.

Eyes

Shortsightedness is common. Dislocation (displacement) of the lens, glaucoma or detachment of the retina (the lining at the back of the eye) may also occur. Any changes in vision should prompt urgent referral to an eye specialist.

Skeleton

Patients with Marfan syndrome are typically tall with long fingers and toes. Joints can be loose and sometimes painful. Curvature of the spine (scoliosis) and an unusually shaped breast bone are also associated with this condition.

Lungs

Sudden lung collapse (pneumothorax) can occur

Dental

A high-arched palate (roof of the mouth) and/or crowding of the teeth are common.

It is important to remember that it is very rare for one person to have all of these problems.

Many people with Marfan syndrome have only mild problems. Others are more severely affected. Even within the same family there may be considerable variability between affected individuals in the severity of the condition.

How is Marfan syndrome treated?

There is no cure for this condition yet but careful medical management can reduce the risk of serious complications.

- Affected individuals should have regular echocardiograms to check the size and function of the heart and aorta.

- Medications such as beta blockers may be used to lower blood pressure and slow the widening of the aorta.

- Antibiotics may be prescribed prior to dental/ surgical procedures in patients with mitral valve prolapse.

- Avoidance of strenuous exercise and contact sport may be necessary to reduce the risk of injury to the aorta and/or the eyes. A leaflet with 'Sports Guidelines' is available from the Marfan Association (see reverse).

- Regular eye checks are recommended.

- During childhood and adolescence, regular physical examination to check for curvature of the spine and good dental care are recommended.

How is Marfan syndrome inherited?

Each child of a person with Marfan syndrome has a 50:50 (1 in 2) chance of inheriting the condition.

Most of the time the gene change is inherited from a parent who is also affected. However, in around 25% of people with Marfan syndrome, no signs of the condition are found in either of their parents, making it most likely that the gene change has occurred for the first time in them. In this situation, the unaffected parents have a very low chance of having another child with Marfan syndrome.