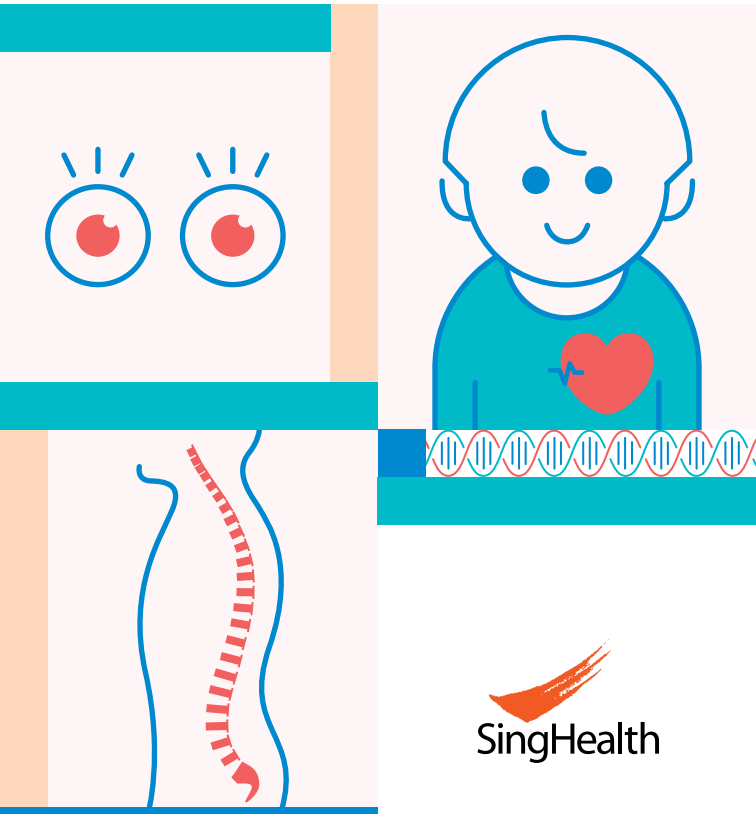
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

Marfan Syndrome



SingHealth

What is Marfan syndrome?

Marfan syndrome (MFS) is a genetic disorder of the connective tissue, which helps to support many parts of the body. MFS particularly affects the heart, blood vessels, skeleton and eyes.

How common is MFS?

It is estimated that **one in 5,000 to 10,000** people are born with MFS. Occurrences of MFS can occur in all races and affect both genders equally.

What are the features of MFS?

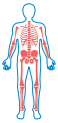
While MFS may be present at birth, some symptoms develop gradually over a number of years. The severity of symptoms varies from person to person, even among affected family members.

The main features include:

Cardiovascular (heart and blood vessels). Weakening and stretching of the blood vessels can occur. This applies in particular to the aorta (the main blood vessel carrying blood away from the heart) and increases the risk of aortic aneurysm, dissection or rupture (bursting).

The heart's valves may be affected, especially the mitral valve. The valve leaflets become floppy and do not close tightly, causing leakage of blood backwards across the valve (mitral valve prolapse). If this progresses, the valve leaks and the condition is called mitral valve regurgitation. If left untreated, this can eventually cause heart failure.

Lungs. Asthma, emphysema, pneumothorax (collapsed lung)



Skeleton. Tall and thin with long arms, legs, fingers and toes. Flat feet, protruding or indented chest bone, loose joints, scoliosis and skin stretch marks.

Eyes. Dislocation of lenses, myopia (short-sightedness), retinal detachment, glaucoma

A **family history** of MFS in a parent, sibling or child

Can MFS be cured?

While it is not possible to cure MFS, much can be done to make sure your child has the best possible outcome.

How is MFS managed?

The multidisciplinary medical team looking after your child will be able to address specific medical concerns and routine MFS-related health issues.

Management measures may include:

- **Regular echocardiograms** to assess the heart and width of the aorta. Medications and/or surgery may be required if the width of the aorta gets too wide.
- **Antibiotic prophylaxis** may be required before dental surgery for children with involvement of the heart valves to prevent infections.

- **Careful monitoring of the skeleton** to detect problems with the spine or chest bone.
- **Regular eye examinations** to monitor for myopia and lens dislocations.

There are also important lifestyle considerations for a child with MFS:

- Ensure a balanced and healthy diet and avoid smoking. These measures are also good for general health.
- Engage in gentle exercise which is good for the heart, blood vessels and joints.
- Discuss with your child's doctor about recommendations for exercise and activities.

With proper management of the cardiovascular manifestations, the life expectancy of someone with MFS approximates that of the general population.

When should your child seek medical attention?

Seek medical attention if there is:

Chest pain or breathlessness

Sudden blurring of vision

Joint pain

What causes MFS?

MFS is caused by changes in the *FBN1* gene located on chromosome 15. Genes are instructions for cells

to make proteins in the body. MFS is the result of a change (mutation) in the *FBN1* gene.

This gene controls the production of fibrillin-1, which is a very fine fibre that is found in connective tissue. Fibrillin fibres come together to form an elastic mesh which helps to support certain structures in our body, such as blood vessel walls and the lens of the eye.

Fibrillin-1 also regulates the activation of transforming growth factor beta (TGF-β) binding protein, which takes part in the regulation of many different cell functions and affects connective tissues throughout the body.

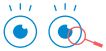
Everyone carries two copies of each gene, one inherited from each parent. MFS follows a **dominant inheritance pattern**, which means the presence of one faulty *FBN1* gene can cause MFS.

How is MFS diagnosed?

MFS is diagnosed based on a full evaluation by a doctor familiar with the disorder. This involves the following:

Thorough physical examination

Complete family history



Full eye examination

Echocardiogram (ultrasound of the heart)

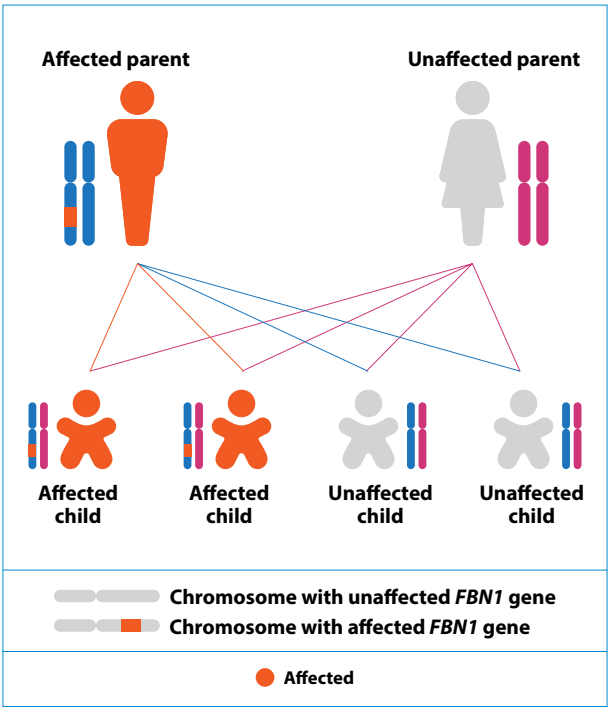
Genetic testing for a causative mutation in the *FBN1* gene can also provide helpful information in some cases

Is MFS inherited?

If either parent has MFS, a child has a 50% chance of inheriting the disease.

Among people with MFS, about 75% of them would have inherited the genetic change from one of their parents. As some people with MFS may have a mild form of the disorder, the parent may not have been previously recognised as having MFS.

The other 25% of people with MFS have it because of a new mutation in the gene.



How likely will I have another child with MFS if neither my spouse nor I have it?

In about 25% of cases, MFS is the result of a spontaneous change (mutation) in the genetic material of the sperm or egg at conception in families with no previous history of the disease.

In such cases, the risk of having another child with MFS is low (< 1%).

MFS is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Groups

Club Rainbow Singapore
Club Rainbow Singapore supports and empowers children with chronic illnesses and their families by providing relevant compassionate services in their journey.
Tel: 6377 1789
Email: contact@clubrainbow.org
www.clubrainbow.org

The Marfan Foundation
www.marfan.org

Acknowledgements
Genetics Service, KK Women's and Children's Hospital

Information contributed by:

SingHealth DukeNUS ACADEMIC MEDICAL CENTRE

Genomic Medicine Centre

KK Women's and Children's Hospital
6294 4050
www.kkh.com.sg

Singapore General Hospital
6321 4377
www.sgh.com.sg

Changi General Hospital
6850 3333
www.cgh.com.sg

Sengkang General Hospital
6930 6000
www.skh.com.sg

National Cancer Centre Singapore
6436 8088
www.nccs.com.sg

National Heart Centre Singapore
6704 2000
www.nhcs.com.sg

National Neuroscience Institute
6321 4377 (SGH Campus)
6330 6363 (TTSH Campus)
www.nni.com.sg

Singapore National Eye Centre
6227 7266
www.snec.com.sg

Find out more about the Centre at:
www.singhealth.com.sg/genomic-medicine-centre

SingHealth
www.singhealth.com.sg

SingHealth Hospitals

Singapore General Hospital | Changi General Hospital | Sengkang General Hospital | KK Women's and Children's Hospital

National Specialty Centres

National Cancer Centre Singapore | National Dental Centre Singapore | National Heart Centre Singapore | National Neuroscience Institute | Singapore National Eye Centre

SingHealth Community and Primary Care

Polyclinics SingHealth | SingHealth Community Hospitals

Reg. No.: 200002698Z
Information correct as at February 2023

Disclaimer: *The information provided does not replace information from your healthcare professional. Please consult your healthcare professional for more information.*

Scan for a digital copy