

## WHAT IS MARFAN SYNDROME?

The Marfan Syndrome is a heritable disorder of the connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels.

Connective tissue "connects", provides structural support and determines the elasticity of the body's organs, bones and ligaments. In Marfan Syndrome, the connective tissue in the heart, lungs, eyes and skeletal systems can stretch and weaken.

The condition, first described by Dr Antoine Marfan in 1896, can affect both men and women of any race or ethnic origin. The incidence of this disease is very rare (There is one person affected in every 5000 person in the population).

## WHAT CAUSES MARFAN SYNDROME?

It is now known that an abnormal gene located on chromosome 15 and containing the coding for a connective tissue protein, is responsible for the syndrome. Most of the time this gene is inherited from a parent who is affected. About 30% of cases occur when the abnormal gene arises in an egg or a sperm of an unaffected parent. Each child of an affected parent has a 50% chance of inheriting the syndrome.

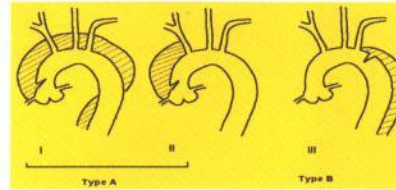
Since genes carry the master plan for all human characteristics, an abnormality of one gene, such as the Marfan gene, results in a specific pattern of bodily changes and medical problems.

## WHAT PROBLEMS ARE ASSOCIATED WITH THE MARFAN SYNDROME?

The following are some of the most common medical characteristics of Marfan Syndrome:

### The Cardiovascular System

- The aorta (the main artery carrying blood away from the heart) can stretch to the point of rupture.



- Leakage of the mitral valve or irregular heart rhythm.

### The Skeleton

- Curvature of the spine (scoliosis).
- Abnormally shaped chest. (pectus deformity).
- Loose jointed-ness.
- Tall stature.
- High arched palate.
- Disproportionately long limbs but not always.
- Long fingers and toes.
- Flat foot.



Scoliosis



Wrist Sign



Thumbs Sign



Flat foot

### The Eyes

- Near-sighted (myopic).
- About 50 percent have dislocation of the ocular lens.
- Retinal detachment.



## HOW IS MARFAN SYNDROME DIAGNOSED?

The Marfan Syndrome is difficult to diagnose because there is no specific laboratory test for the condition. In addition, characteristics of the disorder vary greatly among affected individuals. Most affected people do not have all of the possible signs and complications of the syndrome.

An accurate diagnosis of the Marfan Syndrome can be assessed after a complete physical examination that focuses on the systems affected by the disorder.

### This includes:

- Echocardiogram, a sound wave picture of the heart by a cardiologist.
- Slit-lamp eye examination by an ophthalmologist.
- CT scan or MRI of the spine.
- Complete family history.

## HOW IS MARFAN SYNDROME TREATED?

People affected by the Marfan Syndrome should be treated by a physician familiar with the condition and how it affects all body systems. There is no cure for the disorder yet but careful medical management can greatly improve the prognosis and lengthen the life span.

Treatment for cardiovascular problems is critical and the aorta must be monitored for weakness. Drugs can help to reduce the stress experienced by the aorta but surgery may be needed eventually to replace the vessel.

Every affected person should work closely with his/her physician(s) for their customised treatment plan. However, in general, treatment includes the following:

- Annual echocardiogram to monitor the size and function of the heart and aorta.
- Initial eye examination with a slit-lamp to detect lens dislocation, with periodic follow-up with an ophthalmologist.
- Careful monitoring of the skeletal system, especially during childhood and adolescence.
- Beta-blocker medications may be prescribed to lower blood pressure and consequently, reduce stress on the aorta.

Antibiotics may be prescribed prior to dental or genitourinary procedures to reduce the risk of infection in people who experience mitral valve prolapse or who have artificial heart valves. People who have had aortic surgery must take blood thinning medication. They also need to take antibiotics by injection before dental work and in other situations in which bacteria could enter the blood stream. (Source: *Guidelines for Endocarditis Prophylaxis for People with the Marfan Syndrome Who Have Had Cardiac Surgery*, NMF Professional Advisory Board, June 1991).

Lifestyle adaptations, such as the avoidance of strenuous exercise and contact sports, are often necessary to reduce the risk of injury to the aorta.

## LIVING OPTIMALLY WITH MARFAN SYNDROME

### Be well informed

Know yourself, your body, all available facts about Marfan Syndrome, current medical and surgical treatments, and genetic implications. You should understand your condition and accept it so that you can become an advocate for your own health management.

### Regular assessment

Careful medical management can greatly improve prognosis and lengthen life. Complications can be prevented if patients are seen on a regular basis by their various specialists.

### Emotional support

Marfan patients and their families may seek out information and support to strengthen their coping strategies which are crucial towards living a healthy and productive life.

### Useful websites:

[www.marfan.ca](http://www.marfan.ca)  
[www.marfan.org](http://www.marfan.org)

### DISCLAIMER

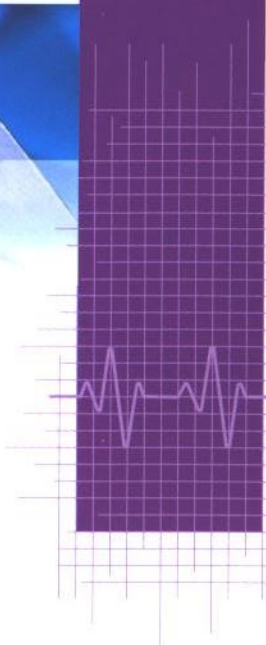
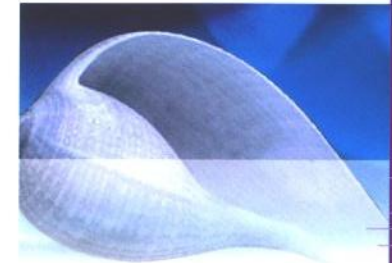
This information is given as a guide only and does not replace medical advice from your doctor. You should seek the advice of your doctor before starting any treatment or if you have any questions related to your health, physical fitness or medical condition.

*The information in this brochure is adapted from:*

*The Marfan Canadian Marfan Foundation and  
 The National Marfan Foundation (USA)*

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## WHAT IS MARFAN SYNDROME? ADULT CONGENITAL HEART DISEASE PROGRAMME



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