

Marfan Association Queensland

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Marfan Association Queensland

The Marfan Association Queensland was formed in 1994 by people who suffer from Marfan Syndrome, their family members and health care professionals. The objectives of the Association are:

- To share current and accurate information about Marfan Syndrome world wide and facilitate international communication among the medical professions.
- To provide a united body to represent the needs of Marfan Syndrome affected people.
- To support and foster research.
- To expand the knowledge of all health professionals.
- To educate the general public about the Syndrome.
- To provide a forum that facilitates the sharing of knowledge and the provision of support for Marfan affected people and their families by:
 - The production, obtaining and distribution of educational material.
 - The holding and promoting of conferences and seminars.
- To produce a regular newsletter for distribution to members and for sale to interested people.



How is Marfan Syndrome Treated?

People affected by Marfan Syndrome should be treated by a physician familiar with the condition and the ways 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.

Every affected person should work closely with his/her physician(s) to develop a customised treatment plan. However, in general, treatment may include:

- 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 by an ophthalmologist.
- Careful monitoring of the skeletal system, especially during childhood and adolescence.

In addition,

- Beta-blocker medication may be prescribed to lower blood pressure and, consequently, reduce the stress on the aorta and the risk of dissection.
- Antibiotics may be prescribed prior to dental or genitourinary procedures to reduce the risk of infection in people who have mitral valve prolapse.
- Lifestyle adaptations, such as avoidance of strenuous exercise and contact sports, are often necessary to reduce the risk of injury to the aorta, lungs and eyes.

The Marfan Clinic at Prince Charles Hospital

The clinic has been established for diagnosis and management of Marfan Syndrome. It is held every three months. For appointments and information, phone _____ 0417 194 162 or email j.west@uq.edu.au. The Marfan association works closely with clinic staff to ensure that people with Marfan Syndrome and members of their families have access to appropriate diagnosis and care.

Marfan Syndrome



A support group for adults and children affected with Marfan Syndrome, their families and other interested people.

MEMBERSHIP FORM

Name _____

Address _____

Postcode _____

Phone Home _____

Work _____

Email _____

If health care professional, please indicate field of specialty

How many in your household have Marfan Syndrome?

Number _____ Ages _____

Membership (\$22.00 per annum) \$ _____

Donation (optional) \$ _____

Total amount enclosed \$ _____

Cheque Money Order Cash Bank Cheque

Please make all cheques payable to: Marfan Association Queensland

To fulfill one of the aims of the Association, it is important that members are able to make contact with each other for support and sharing information. Are you willing for your details to be made available to other members? Indicate the preferred way to contact you.

Yes: Name Address Phone Email

No Not Yet

Date / / Signature _____

Membership is due 1 July annually.

What is Marfan Syndrome?

Marfan Syndrome is a heritable disorder of the connective tissue that affects many organ systems, including the skeleton, heart and blood vessels, eyes and lungs. The condition affects both men and women of all races and ethnic groups. It may be diagnosed in children, teenagers or adults.

What Medical Problems are associated with Marfan Syndrome?

The Cardiovascular System

- The most serious problems associated with Marfan Syndrome involve the cardiovascular system (heart and blood vessels). The aorta (the main artery carrying blood away from the heart) is generally wider and more fragile in patients with Marfan Syndrome. This widening is progressive and can cause leakage of the aortic valve and/or tears (dissection) in the aorta wall. When the aorta becomes greatly widened or dissects, surgical repair may be necessary.
- In addition, the two flaps of the mitral valve in the heart may billow backwards when the heart contracts (mitral valve prolapse). This can lead to inefficient operation of the valve or irregular heart rhythm.

The Skeleton

- People with Marfan Syndrome often have long arms and legs, usually resulting in tall stature. They may have a hollow or pigeon chest, long fingers and toes, flat feet and curvature of the spine (scoliosis). Often they are loose-jointed and have a high palate and crowded teeth.

The Eyes

- The most significant problem with the eyes is subluxation of the lens, when the lens no longer sits behind the pupil but moves or becomes detached. People with Marfan Syndrome are also often short sighted (myopic) and may have a flat cornea (clear part of the eye).

Other Clinical Problems

- People with Marfan Syndrome are prone to pneumothorax (collapsed lung) and hernias. They may have a congenital abnormality of the spine which can only be diagnosed using MRI or CT scan.

What causes Marfan Syndrome?

Marfan Syndrome is a dominant genetic condition. Most affected people have an affected parent although about one third do not and are thought to have new (or spontaneous) mutation. A person with Marfan Syndrome has a 50:50 risk of passing it on to each offspring. The gene which is mutated in Marfan Syndrome is located on human chromosome 15.

How is Marfan Syndrome diagnosed?

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.

To make a diagnosis of Marfan Syndrome, a complete physical examination which focuses on the systems affected by the disorder is needed.

This includes:

- Echocardiogram, a sound wave picture of the heart and aorta, assessed by a cardiologist.
- Slit lamp examination of the eye with dilated pupil by an ophthalmologist.
- Skeletal examination.
- Discussion of family history.

In 1991, the gene which is abnormal (mutated) in Marfan Syndrome was discovered. It carries the recipe for a component of connective tissue called fibrillin, which is involved in giving tissues elasticity. In some families it may be possible to look at this gene and identify the change which causes the condition. However, this test is only available through one private laboratory in Australia and the family must bear the cost.