Genetics

There is a 50% chance of each offspring of a Marfan Patient inheriting the Marfan Gene. The severity and pattern of this condition can be variable even within the same family. Most of the time this Gene is inherited from a parent who is affected. However, about 25% of cases occur as the result of a spontaneous mutation. In 1990 the Marfan Syndrome Gene was located on chromosome 15q. An error in this gene results in diminished fibrillin production. Fibrillin is a very fine fibre in connective tissue throughout the body.

How can you help?

If you, or any member of your family are aware that you have Marfan Syndrome, you should have your GP arrange the necessary investigations, as careful management of Marfans can greatly improve the longevity and quality of life.

How can you support?

If you wish to support us, donations can be made to:

Marfan Syndrome Support Group Irl

Bank of Ireland, The Parade, Kilkenny.

Sort Code: 90 60 64 Acc No:69208838

Marfan Syndrome Support Group

Founded by a group of people who are directly affected by Marfan Syndrome, to provide physical and emotional support to Marfan Sufferers and their families.

The Support Group was also set up to provide information and create awareness for Patients, Relatives, the Medical Profession and the wider community.

To Obtain Further Information Please Contact any of the Following: -

CORK	Johanna & Petronella	021 4630936
DUBLIN	Martin & Mary	01 6239563
KILKENNY	Margaret	056 7767894
LIMERICK	Daphne	061 377276
MEATH	Brian & Jo	046 9071232
MAYO	Caroline & Pat	097 9370899
CLARE	Peggy	065 7084337

Web site: www.marfan.ie
Email: marfan@eircom.net

Aided by :

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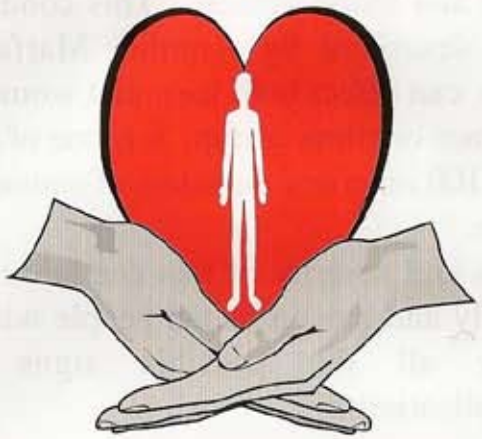
INFORMATION LEAFLET

MARFAN SYNDROME



SUPPORT GROUP

Ireland Ltd

Patrons

Prof. Andrew Green, Geneticist

Prof. Mark Redmond, Cardio Thoracic Surgeon

Dr. Gary Treacy, Ophthalmologist

Dr. John Kenny, Cardiologist

Kate O'Flaherty, Sunday Tribune

Registered Charity No: CHY 13401

Registered No: 307549

What is Marfan Syndrome?

Marfan Syndrome is classified as a genetic/heritable, variable disorder of connective tissue, which may affect many organs including the skeleton, lungs, eyes, heart and blood vessels. This condition, first described by Antoine Marfan in 1896, can affect both men and women of any race or ethnic group. It is one of more than 100 inherited disorders of connective tissue.

Signs and severity of this condition vary greatly and most affected people will not show all the possible signs and complications.

Skeleton Features

Affected people are often tall, slender and double-jointed (joint hypermobility). The arms, legs and fingers may be disproportionately long when compared with the trunk. Spinal curvature is common and may become quite severe without treatment. Easy fatigability is a frequent complaint and migraine headaches, cold hands and feet can be signs of poor circulation.

Cardiovascular

The most serious problems associated with Marfan Syndrome involve the cardiovascular system.

Dilation of the ascending and sometimes the descending aorta, incompetence of the aortic and mitral valves, aneurysm and dissection of aorta. The aorta is usually wider and more fragile. Surgical repair is necessary when the aorta becomes greatly widened (greater than 5cms). Each person suspected of having Marfan Syndrome should have an **Echocardiogram** and regular check-ups so that this function of the heart and aorta can be assessed before serious problems arise. (An Electrocardiogram ECG or chest X-ray is not adequate screening) For someone with no serious cardiovascular problems an annual evaluation is adequate.

Serious heart murmurs or aortic enlargement may dictate more frequent medical check-ups. Aortic enlargement may begin quite early in life, although in some cases it may not occur until much later and in some instances it may never occur at all. People with potential or slight enlargement have been shown to benefit from Beta Blocker Drugs Treatment.

Ocular Manifestation

The eyes require careful attention from early in childhood. The lens of the eyes is off-centre or dislocated in about 75% of persons with Marfan Syndrome. Dislocation of the lens occurs in relatively few other conditions and is therefore, an important hallmark of the Marfan Syndrome, when present. Lens Dislocation usually occurs in early childhood. The most common effect on the eye is near sightedness (Myopia). Holes or tears on the inner lining of the eye (Retinal Detachments) may occur.

Lungs

In some cases Pneumothorax (collapse of the lungs) may occur requiring hospital treatment. Recurrent Pneumothorax may be treated to prevent further episodes.

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