Marfan Association UK

Registered Charity No. 802727 Established 1984

Rochester House, 5 Aldershot Road, Fleet, Hampshire, GU51 3NG
Tel: +44 (0) 1252 810472, Fax: 810473, Answer phone: 617320
E:mail: marfan@tinyonline.co.uk, www.marfan-association.org.uk

Fact Sheet on Marfan Syndrome

First described by the French doctor Bernard J.A. Marfan in 1896.
Marfan Syndrome (MFS) is a variable disorder of the connective tissue that affects many organ systems including the skeleton, eyes, heart, lungs and blood vessels.

Severity differs in each individual.

In 75% of cases it is an inherited disorder, 25% occurring as a result of a spontaneous (new) mutation.

Each child of an affected parent has a 50% chance of inheriting MFS.

It is caused by a mutation in the gene for fibrillin-1 on chromosome 15.

It can affect both men and women of any ethnic group.

Around 10,000 in the UK have Marfan Syndrome (incidence approx. 1 in 5,000)

SALIENT FEATURES

Skeletal:
Tall thin physique, disproportionately long limbs, fingers and toes, lax ankles, flat feet, spinal curvature, abnormally shaped narrow chest (with pigeon or funnel deformity), armspan usually greater than height, joint hypermobility or contractures and dislocations, striae (stretch marks). Dilatation of the lumbar dural sac occurs in about 75% of patients. Hernias are common.

Cardiovascular:
Dilatation of ascending and sometimes descending aorta, incompetence of aortic and mitral valves, aneurysm and dissection of aorta.

Respiratory:
Pneumothorax (collapse of the lungs), bronchiectasis, emphysema and asthma.

Ocular:
Subluxation or dislocation of lens, myopia (short sight) and unstable refraction, detachment of retina, strabismus (squint), glaucoma.

Dental:
High-arched palate, crowding of teeth.

Genetic:
Even within one family the severity and pattern of disease varies.

***A Syndrome* is a collection of physical features which, when they occur together, enable a physician to recognise a certain condition.**

WHAT CAUSES MARFAN SYNDROME?

A single abnormal (mutant) gene on Chromosome 15 causes the condition. This abnormal gene controls production of fibrillin, a very fine fibre in connective tissue throughout the body (the "glue and scaffolding of the body"). Most of the time this gene is inherited from a parent who is also affected. However, about 25% of the cases occur when the abnormal gene appears in an egg or a sperm (a spontaneous "new" mutation) producing an *affected* child from two *unaffected* parents.

Marfan Syndrome is inherited as an "autosomal dominant" condition. This means that someone with Marfan Syndrome has a 50-50 chance that each offspring will inherit the condition, regardless of sex. On average one in ten affected children are seriously affected.

HOW IS MARFAN SYNDROME DIAGNOSED?

Marfan Syndrome may be difficult to diagnose because signs of the condition vary greatly from one person to the next. Most affected people will not have all the signs and complications of Marfan Syndrome.

Diagnosis can be confirmed within a family by genetic linkage studies. Mutations can be found in the fibrillin-1 gene in 80% of patients, assisting with screening of family members.

The identification of the chromosome, gene in 1990 and the component of connective tissue (fibrillin) in 1991, in which the mutations for the Marfan Syndrome are located, offers great promise as a diagnostic aid. It is hoped that as a better understanding of fibrillin is gained, earlier and more accurate diagnosis of the Marfan Syndrome will be possible.

Blood tests demonstrating the abnormal gene are becoming available for diagnosis through referral to a clinical geneticist.

Prenatal diagnosis is now available for some families with this condition, especially those families where a mutation (change) in the fibrillin gene has been demonstrated.

In general, Marfan Syndrome is diagnosed after careful physical examination, particularly focusing on the main systems involved – Eyes, Skeleton, Heart and Lungs. Certain tests, such as an echocardiogram (a sound-wave picture of the heart) are useful in making the diagnosis.

PEOPLE WITH MARFAN SYNDROME SHOULD HAVE AN INITIAL DIAGNOSTIC ECHOCARDIOGRAM WHICH IS REPEATED AT REGULAR INTERVALS.
An Electrocardiogram (ECG) is not adequate screening.
Skeletal X-rays (mainly chest and back) may be necessary and a careful eye examination, using a slit lamp to detect lens dislocation, is recommended.
Family history needs to be taken into account.

MEDICAL PROBLEMS AND TREATMENT

PEOPLE WITH MARFAN SYNDROME SHOULD BE TREATED BY A PHYSICIAN FAMILIAR WITH THE CONDITION, CONVERSANT WITH ITS EFFECTS ON ALL BODY SYSTEMS AND ABLE TO ADVISE ON SCREENING OF THE FAMILY. GENETIC COUNSELLING SHOULD BE GIVEN. THERE IS NO CURE FOR THIS CONDITION YET, BUT CAREFUL MEDICAL AND SURGICAL MANAGEMENT, TOGETHER WITH AN APPROPRIATE LIFESTYLE, CAN GREATLY IMPROVE PROGNOSIS AND LENGTHEN LIFE SPAN. A list of Genetic Centres is available at the Marfan Office.

HEART

The most serious life-threatening problems associated with Marfan Syndrome involve the cardiovascular system. The two leaflets of the mitral valve may billow backwards when the heart contracts, a condition called "mitral valve prolapse". This feature may lead to heart failure or be associated with irregularities of the heart rhythm.

The aorta (the main artery carrying blood away from the heart) is generally wider and more fragile in people with the Marfan Syndrome. This widening is progressive and may result in leakage of the aortic valve or in the development of tears (dissection) in the wall of the aorta. When the aorta becomes widened, medications (e.g. beta blockers, to lower blood pressure) may be prescribed, thereby reducing strain on the aorta and regulating heart rhythm. This may be followed by surgical repair preferably when the aortic root widens to 5 cms diameter, and before it becomes torn.

Patients with Marfan Syndrome are recognised to have a slightly increased morbidity and mortality risk associated with general anaesthesia. Preoperative assessment should include a thorough medical examination with a chest x-ray, electrocardiogram and echocardiogram. Any treatment must be carried out in conjunction with the patient's cardiologist.

Antibiotics may be prescribed prior to dental, genito-urinary or other minor surgical procedures, to reduce the risk of infection (endocarditis) in people who experience mitral valve prolapse.

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

Beta blockers have been shown to slow the dilation of the aortic root and their use should be considered in all patients.

Regular Echocardiograms are important to monitor size and function of the heart and aorta.

SKELETON

Musculo-skeletal problems are common and troublesome in Marfan Syndrome. Indeed, recognition by a rheumatologist could be the first vital step towards diagnosis of the underlying condition.

Involvement of the skeleton includes curvature of the spine (scoliosis/kyphosis/lordosis), abnormally shaped chest ("pectus" deformity), tall stature, and loose jointedness (often causing joint pain and dislocation).

Physiotherapy, pain clinics and bracing may be helpful. In certain instances, surgery is indicated. Careful monitoring is needed, especially during childhood and adolescence. Shoe inserts may help your child. Weak ankles may require lace-up shoes with ankle support.

EYES

People with Marfan Syndrome are generally near-sighted (myopic). In addition, some have dislocation of the ocular lens and retinal detachment. Glasses and/or contact lenses may be prescribed to correct visual defects. Surgery is now available, if required, for removal and replacement of lens(es) and retachment of retinas.

LUNGS

Spontaneous pneumothorax (collapse of the lungs) is thought to occur in approximately 10% of patients and requires hospital treatment.

LIVING WITH MARFAN SYNDROME

Be well informed

As a member of the Marfan Association UK you will be made aware of current medical and surgical treatments, genetic implications and latest research results, enabling you to become an advocate for your own health management.

Understanding enables you to "come to terms" with the disorder and to concentrate on your life as a person who happens to have Marfan Syndrome rather than as "a Marfan patient". Marfan Syndrome is a variable disorder, affecting everyone differently. The objective should be to achieve a clear understanding of your needs, in partnership with your doctor.

Regular Assessment

Complications can be prevented if patients are seen on a regular basis by their various specialists. Those affected should work closely with physicians for individual care and management. Often your Consultant requires the test results such as X-rays, ECG (Electrocardiogram) Echocardiogram, MRI (Magnetic Resonance Imagining) CT Scan, lung function tests prior to your appointment. Check that this is in hand! The Association would be happy to send updated Marfan information direct to your GP and Specialists. This is often useful prior to your appointment!

Emotional Support

The Marfan Association will be pleased to hear from people with Marfan Syndrome, and their families. Within the organisation we have a National Marfan Support Network offering local support. In addition we have "Circles" e.g. 'Heart to Heart', 'Eye to Eye', 'Bone to Bone' and others. Through these patient groupings, many people have been helped by talking to someone else in similar circumstances and often they themselves have then felt able to help others.

Fitness

In general it is important for the patient with Marfan Syndrome to keep as fit as possible. This improves muscle tone and is good for overall function of the heart and blood vessels, but any exercise should be appropriate to each individual's physical condition. Some activities are best avoided, such as long distance running and heavy lifting.

Activity

The person with Marfan Syndrome should in general be able to take part in appropriate non-competitive sporting activities but should be allowed to stop whenever tired. Contact sports such as basketball and rugby are probably best avoided. Enjoy yourself within your own safe individual limits!

Easy Fatiguability

Fatigue, due to Marfan Syndrome, can be a problem, especially when long periods of concentration are required. Learn to "pace yourself", working within your own comfortable time scales!

Diet

A balanced healthy diet, rich in vitamins and minerals, encourages the production of connective tissue. It makes good sense to choose your foods wisely!

Smoking

Smoking destroys elastin, which is the very protein which is already deficient in anyone who has Marfan Syndrome. It also causes complications in surgery and the recovery period. It is therefore best avoided. Passive smoking again creates a problem, so – if a smoke free zone exists – it makes sense to use it!

Starting a Family

Having children is a very personal decision that should be made solely by prospective parents, but only after acknowledging and understanding the potential risks, especially if the female partner is affected. Pre-pregnancy and genetic counselling should, therefore, be undertaken before starting a family. Remember that, although there is a 50% chance that a child may inherit the Marfan gene, there is a 50% chance that they may not!

The Marfan Association UK was officially founded in 1984 to offer:
"Support for Today with Tomorrow in Mind."
Our three main aims are SUPPORT, EDUCATION AND RESEARCH.
We are a U.K. registered charity with International Outreach

We support **those with Marfan Syndrome and their families…….We are aware that the unaffected family members may also need help to "come to terms with MFS".**
A National Marfan Support Network is available.
We educate **by distributing updated Marfan text to lay and professional sectors, holding and participating in School and Hospital meetings around the country.**
Early diagnosis is vital - we believe this is achieved by spreading information widely.
A Publications List is available covering the many aspects of MFS.
We research **by undertaking, sponsoring, and participating in, national research projects.**
Already the gene has been discovered.

Much can be learned from those with Marfan Syndrome and their families.
Your family history and photographs could greatly help us all to learn more!

We work closely with GPs, Ophthalmologists, Cardiologists, Orthopaedic Specialists, Pulmonary Specialists, Rheumatologists, Geneticists, Surgeons, Anaesthetists, Midwives, Visual Impairment Officers, Social Workers, Teachers, to name but a few, and special information is available for the many different sectors involved in the care of someone with Marfan Syndrome.

Please join the Marfan Association UK today. YOU NEED US AND WE NEED YOU!!
"TOGETHER WE CAN"

"Support for Today With Tomorrow in Mind"
Printed by Printfine Ltd Liverpool
January 2008 Fact Sheet
Registered Charity No. 802727