

# Marfan Trust

*Supporting Research into Marfan Syndrome*



## Guide to Marfan syndrome

Find out more today



[www.marfantrust.org](http://www.marfantrust.org)

Registered charity number 328070



**M**arfan syndrome is an inherited disorder of the body's connective tissue that affects men and women of any race or ethnic group. Marfan syndrome was identified in 1896 by a French physician Antoine Marfan. The syndrome is an inherited disorder of the body's connective tissue which affects the heart, eyes, lungs and skeleton in handicapping, painful and even mortal ways.

- ◆ 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 Marfan syndrome.
- ◆ It is caused by a mutation in the gene for fibrillin-1 on chromosome 15.
- ◆ It can affect both men and women of any race or ethnic group.
- ◆ Approximately 18,000 of the UK population are affected and 1 in 3,300 worldwide.
- ◆ On average there are over 200 new cases of Marfan syndrome diagnosed every year in the UK.
- ◆ About half of sufferers remain undiagnosed.

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



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## 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), arm span 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:** Dilation of ascending and sometimes descending aorta, incompetence of aortic and mitral valves, aneurysm and dissection of aorta.

**Respiratory:** Pneumothorax (collapse of the lungs), bronchiectasis, fibrosis, 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.



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## 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 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.

## 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.

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 soundwave 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



Photo copyright: Marfan Foundation USA

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.

#### **Genetic Testing:**

Diagnosis can also be confirmed within a family by genetic linkage studies.

Mutations can be found in the fibrillin-1 gene in 97% of patients, assisting with **screening of family members**.

**A Blood test** demonstrating the abnormal gene is available through referral to a clinical geneticist. (See back for list of UK Genetic centres)

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

**Preimplantation genetic diagnosis** through creation of unaffected embryos enables an affected parent to have unaffected children.





## Medical Problems and Treatment

**H**eat: 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 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 between 4.5 and 4.8cms 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, or who have had aortic root surgery.

Lifestyle adaptations, such as the avoidance of strenuous exercise and contact sports, are often necessary to reduce the risk of injury to eyes and 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. Arch supports (orthotics) may help the affected 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 reattachment of retinas.

**Lungs:** Spontaneous pneumothorax (collapse of the lungs) is thought to occur in approximately 10% of patients and requires hospital treatment. Sports involving sudden changes of pressure (parachute jumps, scuba diving) are not recommended.



## Living with Marfan syndrome

### 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 Imaging) CT Scan, lung function tests **prior** to your appointment.

### Fitness

In general it is important for the patient with Marfan syndrome to keep as fit as possible with gentle regular exercise. 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. (Ask for a copy of our Exercise Guide, Marfan syndrome booklet),

### 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 best avoided, but lighter sports such as badminton and cycling on flat ground are suitable.

### Easy Fatigability

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.

## 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.

## 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. If the aortic root measurement is greater than 4.0cm, pregnancy causes at least 10% risk of dissection for affected women. Aortic root replacement prior to pregnancy is an option, ensuring safe pregnancy for both mother and child. Pre-pregnancy genetic counselling should, therefore, be undertaken before starting a family. Genetic testing of the pregnancy is available (see page 4).

## The Marfan Trust Charity

**Registered Charity No. 328070**

Guy Scadding Building,  
Dovehouse Street,  
London  
SW3 6LY

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## Marfan Genetic Clinics

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.

**Genetic Clinics can be found in the following UK locations:-**

### **Birmingham**

West Midlands Clinical Genetics Service,  
Birmingham Women's Hospital,  
Metchley Park Road, Edgbaston,  
Birmingham, B15 2TG

**Tel: 0121 627 2630**

### **Leeds**

Yorkshire Regional Genetics Service  
Department of Clinical Genetics  
Ward 10, 3<sup>rd</sup> Floor Chapel Allerton Hospital,  
Chapeltown Road  
Leeds, LS7 4SA

**Tel: 0113 392 4454**

### **Bristol**

Bristol Clinical Genetics Service  
Bristol Royal Infirmary, NHS Foundation  
Trust, Level 7, Upper Maudlin Street  
Bristol, BS2 8HW

**Tel: 0117 9804070**

### **Leicester**

Leicestershire Clinical Genetics Services,  
Leicester Royal Infirmary  
Leicester, LE1 5WW

**Tel: 0116 258 5736**

### **Cambridge**

East Anglia Regional Genetic Service,  
Department of Clinical Genetics,  
PO Box 134, Addenbrooke's Hospital NHS  
Trust, Cambridge, CB2 0QQ

**Tel: 0122 3216 446**

### **Liverpool**

Cheshire and Merseyside Clinical Genetics  
Service, Department of Clinical Genetics,  
Liverpool Women's NHS Foundation Trust,  
Service Base, Royal Children's Hospital,  
Alder Hey, Eaton Road, Liverpool, L12 2AP

**Tel: 0151 802 5001/5002**

### **Exeter**

Peninsula Clinical Genetics Department,  
Royal Devon & Exeter Hospital  
(Heavitree), Gladstone Road,  
Exeter, EX1 2ED

**Tel: 0139 2405 726**

### **London North East Thames**

Clinical Genetics Department, Great  
Ormond Street Hospital for Children NHS  
Trust, Great Ormond Street  
London, WC1N 3JH

**Tel: 0207 762 6831/6856/6786/6845**

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## London North West Thames

Regional Genetics Service, Kennedy Galton Centre, Level 8V Northwick Park & St. Marks NHS Trust, Watford Road, Harrow, Middlesex, HA1 3UJ  
Tel: 020 8869 2795

## London South East Thames

The Genetics Department, 7th Floor, New Guy's House, Guy's Hospital, London, SE1 9RT  
Tel: 0207 188 1364

## London South West Thames

Regional Genetics Centre, St. George's University of London, Cranmer Terrace London, SW17 0RE  
Tel: 0208 725 2038

## Manchester

Manchester Regional Genetics Service, Genetic Medicine, 6th Floor, St. Mary's Hospital, CMFT, Oxford Road, Manchester, M13 9WL  
Tel: 0161 276 6506

## Norfolk and Norwich

Norfolk and Norwich University Hospital NHS Foundation Trust, Colney Lane Norwich, NR4 7UY  
Tel: 01603 287068

## Newcastle

Northern Genetics Service, The Newcastle upon Tyne Hospitals NHS Foundation Trust, Institute of Genetic Medicine International Centre for Life, Central Parkway Newcastle upon Tyne, NE1 3BZ  
Tel: 0191 241 8600

## Nottingham

Nottingham Department of Clinical Genetics, The Gables, Nottingham City Hospital NHS Trust, Hucknall Road, Nottingham, NG5 1PB  
Tel: 0115 962 7728

## Oxford

Oxford Regional Genetics Service, The Churchill Hospital, Old Road, Headington, Oxford, OX3 7LJ  
Tel: 0186 5226 009 or 0186 5226 028

## Sheffield

Sheffield Clinical Genetics Service, Department of Clinical Genetics, OPD2, Northern General Hospital, Herries Road, Sheffield, S5 7AU  
Tel: 0114 2717 034

## Southampton

Wessex Clinical Genetics Service Princess Anne Hospital, Coxford Road, Southampton SO16 5YA  
Tel: 0238 1206 170

## WALES: Cardiff

All Wales Medical Genetics Service, Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff, CF14 4XW  
Tel: 029 2074 2577

## Northern Ireland

Northern Ireland Regional Genetics Service, Belfast City Hospital, Lisburn Road, Belfast BT9 7AB  
Tel: 028 9504 8022

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## Scotland Marfan Genetic Clinics:

### Aberdeen

North Scotland Clinical Genetics Service  
Ashgrove House, Foresterhill  
Aberdeen AB25 2ZA  
Tel: 0122 4552 120

### Dundee

East Scotland Human Genetics, Level 6,  
Ninewells Hospital and Medical School,  
Dundee, DD1 9SY  
Tel: 0138 2632 035

### Edinburgh

South East Scotland Regional Genetics  
Service, Western General Hospital  
Crewe Road South  
Edinburgh EH4 2XU  
Tel: 0131 537 1116

### Glasgow

West Scotland Regional Genetics Service  
Level 2, Laboratory Medicine  
Southern General Hospital  
1345 Govan Road, Glasgow G51 4TF  
Tel: 0141 354 9200/9201

There are **many ways you can help** towards the work we do here at the Marfan Trust; please visit our website to find out more:

- ◆ Monthly Donations
- ◆ One off donation
- ◆ Sonalee Wish List purchase
- ◆ Marfan Trust Shop
- ◆ Fundraise for us
- ◆ In memory donation
- ◆ Legacy donation
- ◆ Volunteer

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**W:** [www.marfantrust.org](http://www.marfantrust.org)

**E:** [info@marfantrust.org](mailto:info@marfantrust.org)

**T:** 020 7594 1605

## Make a donation to our work:

All of our leaflets are distributed for free, in order to continue to provide this much needed information we rely on donations, please consider making a donation to The Marfan Trust charity.

