Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

General Guide to the Charity

FIND OUT MORE TODAY

Helpline: (0)333 011 5256 | www.marfantrust.org | info@marfantrust.org
Marfan Trust, a CIO registered as a charity in England in Wales with charity number 1198847

Registered with FUNDRAISING REGULATOR

What is Marfan Syndrome?

Ashton is 7 and has Marfan syndrome

Marfan syndrome (MFS) is a genetic disorder of the body's connective tissue that affects any gender, race or ethnic group. Connective tissue helps provide structure to the body, binding skin to muscle and muscle to bone. It provides the stretchy strength of tendons and ligaments around joints, and in blood vessel walls. It also supports the internal organs. This tissue is made of fine fibres and 'glue'. One fibre is called fibrillin.

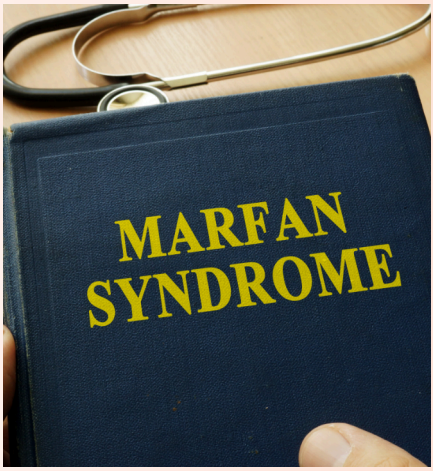
In MFS, a change in the fibrillin-producing gene, fibrillin-1, means that this protein is deficient in connective tissue throughout the body, creating an unusual stretchiness and weakness of tissues. This has far-reaching implications and can affect the eyes, lungs, gut, nervous system, skeleton and, most dangerously, the cardiovascular system. Symptoms can vary widely from person to person with people experiencing mild to severe manifestations.

Lucy has Marfan syndrome

MFS affects roughly 1 in 3,000 people which means that approximately 18,000 people in the UK have MFS. We estimate that half remain undiagnosed. 75% of patients inherit the condition whilst 25% develop it as a result of a spontaneous (new) gene change. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

Diagnosing Marfan Syndrome

Marfan syndrome can be difficult to diagnose because signs of the condition vary greatly from person to person. MFS is multi-faceted and most affected people will not have all the signs and complications of the syndrome.



The most common, visible feature is excessive height with disproportionally long limbs, fingers and toes. This is sometimes accompanied by a protruding or indented chest, curved spine (scoliosis), high-arched palate, crowded teeth, short-sightedness and dislocated eye lenses.

An early, accurate diagnosis is essential, not only for people with Marfan syndrome but also related connective tissue disorders. MFS belongs to a wider family of connective tissue disorders that includes Loeys-Dietz syndrome. Early accurate diagnosis of MFS can be confirmed by genetic testing. The Trust helped to set up Marfan clinics in 24 NHS regional genetic centres in the United Kingdom and here is the link to the list:

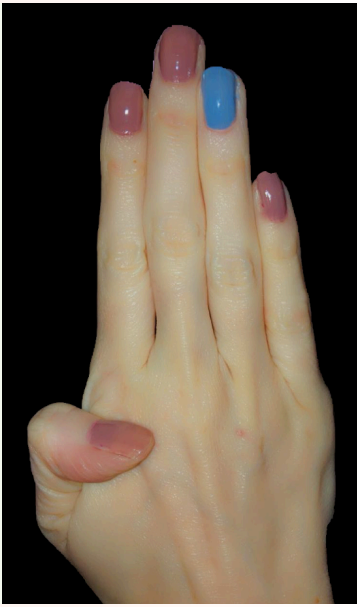
www.marfantrust.org/resources/2-regional-genetics-centres

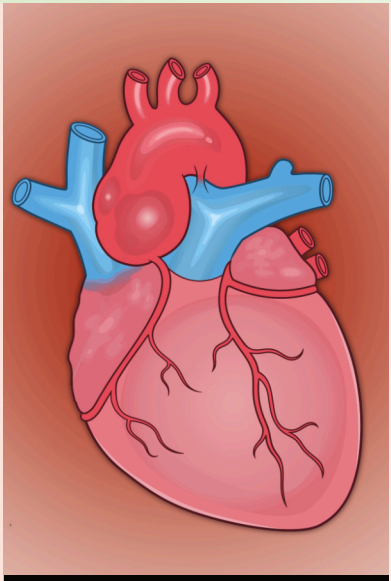
Referral to your nearest centre can be obtained through your GP. Genetic testing involves the analysis of a blood sample for a change in the fibrillin-1 gene. Preimplantation Genetic Diagnosis (PGD) is available, if one parent is affected. This is a technique that can test embryos for the gene change before they are implanted in the womb to ensure the baby is unaffected.

A clinical diagnosis is also possible after careful physical examination by an experienced doctor if there are numerous signs, specifically those of aortic dilatation and dislocated eye lenses, especially if a person has a known family history of Marfan syndrome. However, genetic testing is increasingly used to test patients and has become the most definitive diagnostic tool which can be used in conjunction with other tests and assessments.

Marfan syndrome was first identified in 1896 by French physician Antoine Marfan. The salient signs and symptoms are:

Skeleton: Musculoskeletal problems are common and troublesome in MFS, with patients often growing to excessive height while developing curvature (scoliosis/kyphosis/ lordosis) of the spine. Fingers are often spidery and long, and hammer toes are a frequent feature. Skeletal problems also involve abnormally shaped chest (pectus deformity) , and loose joints which frequently cause pain and occasionally joint dislocation.



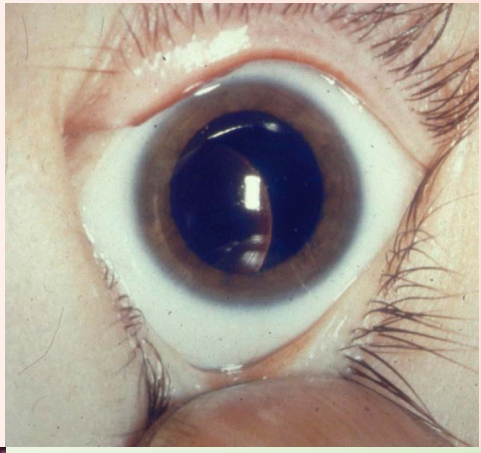


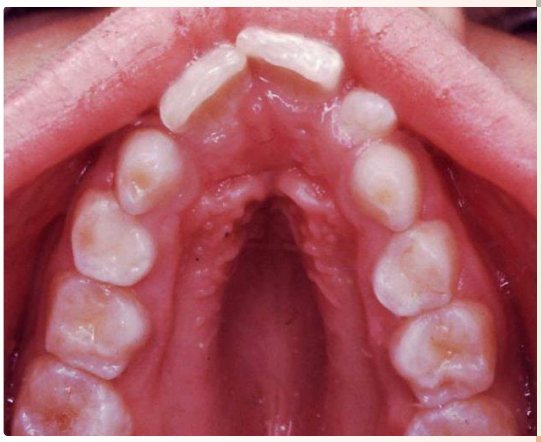
Ascending Aortic Aneurysm

Heart: People can experience ballooning and potentially fatal tearing of the aorta (the body's largest artery, carrying oxygen-rich blood from the heart to the circulatory system) and backward billowing of the heart's valves. Left unmonitored or untreated, these symptoms can be life-threatening.

The key to a long and healthy life with Marfan syndrome is monitoring with regular scans to check the heart function and the size of the aorta. A combination of echocardiograms and MRI or CT scans are usually used. This allows doctors to plan any treatment that may be required.

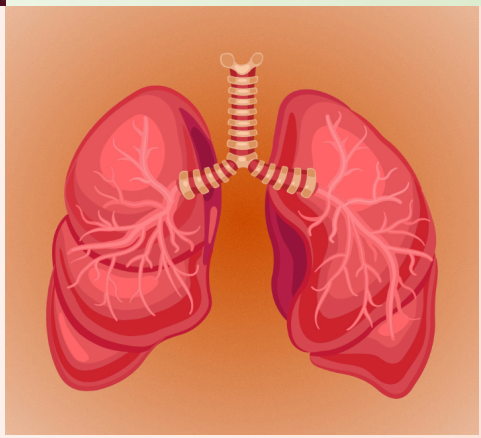
Eyes: People with MFS are often myopic (short-sighted), with some experiencing dislocation of the ocular lens, and retinal detachment. Strabismus (squint) and glaucoma are also ocular signs.





Dental: A high-arched palate and crowding of teeth with a history of tooth extraction for crowding is fairly common amongst Marfan patients.

Lungs: Pulmonary complications occur in 10% of patients, commonly cystic changes in the lung, and occasionally pneumothorax (air trapped outside the lung due to an air leak).



Co-founded in 1988 by Dr Anne Child, the Marfan Trust is the sole charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome.


Dr Anne Child

The Marfan Trust's three main objectives are to:

- Provide personalised support and medical guidance through its helpline.
- Conduct cutting-edge medical research through its designated Sonalee Laboratory, named after a young doctor who tragically died of complications from MFS during her ward round. The results of the lab's internationally recognised research enable doctors and surgeons to provide better treatment for patients, and prolong lifespan.
- Educate the public and professionals alike, through educational literature on- and offline, webinars and annual conference, while raising awareness of the condition so that the symptoms are recognised and more diagnoses are made in good time.

Since its inception in 1988, the Marfan Trust has pioneered and funded many ground-breaking projects. The Trust is responsible for:

- Pioneering the international consortium that discovered the causative gene – fibrillin-1 - for Marfan syndrome in 1991.
- Hosting a research fellow who made a major contribution to the discovery of the fibrillin-1 gene.
- Assisting a researcher who studied pregnancy-associated risks in Marfan syndrome patients.
- Hosting 25 PhD and MSc students who have helped to discover, analyse and identify gene mutations in UK patients.
- Sponsoring 28 medical student summer fellowships to research clinical aspects, and produce a patient pamphlet on each topic.
- Publishing information leaflets covering every aspect and manifestation of Marfan syndrome, empowering patients with the knowledge they need to better manage their condition while providing education for medical practitioners.
- Participating in the Aortic Irbesartan in Marfan Syndrome (AIMS) national drug trial, co-funded by the British Heart Foundation, assessing the positive effects of Irbesartan on preserving the aortic wall and prolonging lifespan.

How you can help

The Marfan Trust relies solely on the goodwill of its unstoppable supporters who tirelessly raise funds and awareness, allowing the charity to continue its good work and lift the shadow from this condition.

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You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month:

www.marfantrust.org/pages/10-membership

JustGiving®

Just Giving – **http://bit.ly/3Scj51w**

PayPal

PayPal Giving – https://bit.ly/45NCuwQ

- BANK: Charities Aid Foundation (CAF) ACCOUNT NAME: Marfan Trust
- SORT CODE: 40-52-40
- ACCOUNT NUMBER: 00017677
- REFERENCE: Your Name (plus campaign name if relevant)

By donating to the Marfan Trust, you are contributing to an ever-growing body of knowledge on the condition, allowing more doctors and medical specialists to deliver the best possible treatment to patients affected by Marfan syndrome.

You can also help to fund a piece of equipment in Dr José Aragon-Martin's Sonalee Laboratory. Email info@marfantrust.org to find out more.

Introducing Our Cover Stars

Ashton is 7 and is at his happiest when he is in the water. He loves swimming, and also sketching and making people laugh. He can often be found researching jokes on his iPad! He takes daily heart medication and wears splints for his hypermobility. Despite all this he achieves Greater Depth in everything he does at primary school. Ashton was diagnosed with Marfan syndrome as a baby, after his grandmother, a nurse, noticed that his fingers were particularly long and thin. He is a very sweet and funny boy who smiles through his chronic pain.

Lucy lives with her mother and pet tortoise, Herbie. She is a crystal-collecting, plant-loving, creative crafter and maintains a widely read blog on her life with Marfan syndrome. She was diagnosed as a child by a locum GP and struggles with the musculoskeletal and gastrointestinal aspects of the condition. Lucy is a great friend of the charity, sharing her thoughts and also holding raffles for us. Her blog can be found here: www.marfantrust.org/articles/category/15-blog.

Raising Awareness

Critical to a healthy life is early diagnosis. Once MFS is diagnosed, a treatment plan is put in place and every aspect of the condition will be managed. But, as a little-known, rare condition, Marfan syndrome is often missed. You can change this. Opportunities exist everywhere in everyday life to recognise the syndrome, from opticians and GP surgeries to rheumatologists and dentists.

Spread the word by distributing our leaflets. Follow us on social media and share our posts.

You can also hold a fundraising event and here's a link to our web page, a trove of helpful ideas. www.marfantrust.org/pages/3-fundraising-ideas

All this will help to lift the shadow of Marfan syndrome from the estimated 18,000 affected people in the UK.

Thank you!

The Hospital Saturday Fund Supported by The Hospital Saturday Fund