Marfan Trust

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Marfan Syndrome: A Guide for Schools and Colleges

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

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What is it?

Marfan syndrome (MFS) is an inherited genetic disorder of the body's connective tissue that affects both boys and girls of any race or ethnic group. It was first described by a French paediatrician, Dr Antoine Marfan, in 1896.

MFS can affect the cardiovascular system, eyes, lungs, gut, nervous system, and skeleton. Connective tissue, which helps provide structure to the body, binding skin to muscle, muscle to bone, is made of fine fibres and 'glue' called fibrillin. This tissue provides the stretchy strength of tendons and ligaments around joints and in blood vessel walls. It is also important in the eyes, lungs, and gut.

In MFS, fibrillin is deficient in connective tissue throughout the body, accounting for the unusual stretchiness and weakness of tissues.

Symptoms can differ widely from person to person with people experiencing mild to severe disability. Approximately 50% of people with MFS remain undiagnosed.

- In 75% of cases, it is an inherited condition due to a genetic change that is passed on from parent to child; 25% of cases are the result of a spontaneous genetic change (i.e. they are the first person in their family to have MFS).
- Any child of an affected parent has a 50% chance of inheriting MFS.
- It is caused by a change in the gene for fibrillin-1 on chromosome 15.
- MFS is rare, occurring in approximately 1 in 5,000 people worldwide. This would suggest there are around 15,000-20,000 people with MFS in the UK.
- Every year, on average in the UK there are over 200 new cases of Marfan Syndrome diagnosed.

Medical Emergency

The following emergencies are rare, but it is imperative that all adults in the school or college are aware of them before dealing with young people with MFS.

There are five **rare** complications requiring emergency care. If you suspect that a child in your care is experiencing any of these, please call an ambulance. When you speak to the paramedic call handler, it is important to mention that the child suffers from Marfan syndrome and that this puts them at increased risk of these rare, but severe problems.

- **Aortic dissection** – the child may present with severe chest or back pain and may become unconscious. This is **a life-threatening medical emergency** and the child should be transferred to a hospital as soon as possible.

- **Collapse of lung (pneumothorax)** – this can occur spontaneously in MFS and may happen during periods of exertion. The child could present with severe breathlessness, may become blue and complain of chest pain which is worse when they try to breathe in. This condition is not always life-threatening but requires **immediate hospitalisation**.

- **Glaucoma** – a dislocated lens may produce a sudden increase of pressure within the eye, causing acute eye pain. This condition requires **urgent hospitalisation and treatment to avoid blindness**.

- **Detached retina** – this is a separation of the membrane at the back of the eye from its supporting layers and can also cause severe vision loss or blindness if not treated. This requires **urgent medical attention**.

- **Joint dislocations** – loose ligaments do not provide adequate support for joints, and dislocation (knees and shoulders are most frequently affected) may require hospital admission for treatment.

It is important that all staff (teachers/clerical staff/lunchtime supervisors) within the school/college are aware of the procedures to be followed in a medical emergency and that plans are put in place for field trips/school outings as well as routine school days. It is also important to note that these potential complications are rare and children with MFS should be given the opportunity to participate in all suitable school activities without fear. For most children this will include all school and play activities other than contact sport.

Marfan-associated medical issues: their treatment and impact on school/college life

1. Physical features which may be present

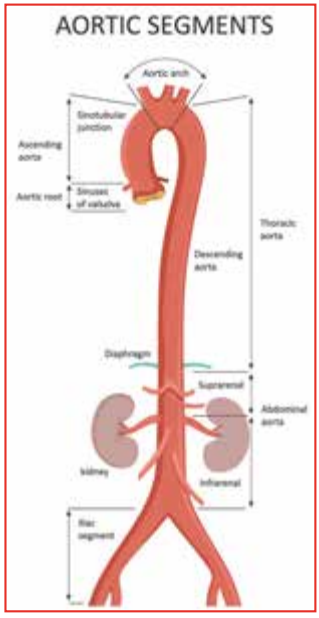
- Long arms, legs, and fingers
- Tall and thin body type
- Curved spine
- Sunken or protruding chest
- Flexible joints
- Flat feet
- Crowded teeth, high arched palate
- Unexplained stretch marks on the skin

There is no cure for MFS, but symptoms can be managed. Everyone with this condition needs careful monitoring tailored to their specific situation. The symptoms of Marfan syndrome vary from person to person.

Some pupils with MFS will be attending regular medical appointments with a whole variety of specialists, often far away, and may miss many days of school as a result.

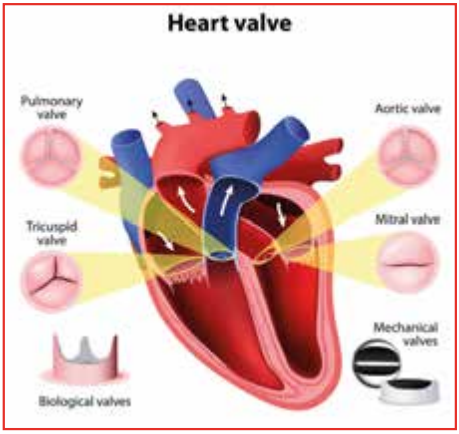
2. Cardiovascular system

The most serious, life-threatening problems associated with MFS involve the cardiovascular system. Within the heart, the two leaflets of the mitral valve may billow backwards when the heart contracts, a condition called "mitral valve prolapse". If the mitral valve prolapse becomes severe it can lead to heart failure and/or rhythm disturbances so sometimes an operation will be needed to fix this. The aorta (the main artery carrying blood away from the heart) is sometimes wider and more fragile in people with MFS. This widening is progressive and may result in leakage of the aortic valve or the development of tears (dissection) in the aorta wall.



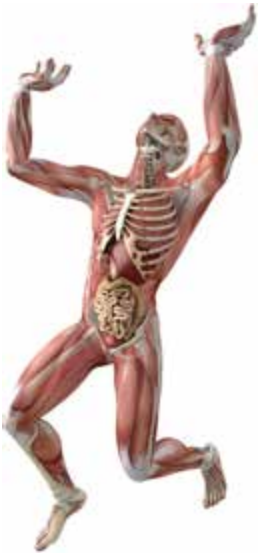
Medications (e.g. beta blockers or angiotensin receptor blockers to slow aortic enlargement) may

be prescribed to reduce strain on the aorta and regulate the heart rhythm. The medications can lead to tiredness and lethargy. Surgical repair may be required if the aorta continues to widen, and the risk of tears therefore increases.



Children and young people will be seen regularly by a Cardiologist for scans of their heart and aorta. These scans will check for any change to the heart function or size of the aorta and are usually done on at least an annual basis or if there are any changes to the child's symptoms (e.g. becoming more short of breath during PE or at play).

3. Skeleton and nervous system



Musculo-skeletal problems are common and troublesome in MFS. Involvement of the skeleton can include curvature of the spine (scoliosis/kyphosis/lordosis), abnormally shaped chest ("pectus" deformity), tall stature and loose joints (joint hypermobility) that can lead to chronic pain and joint dislocations.

Careful monitoring of the skeleton is important, particularly during the rapid periods of growth in childhood and adolescence. In addition to an Orthopaedic doctor, children may need to see other healthcare professionals such as an Orthotist if aids or shoe inserts are needed to assist with posture and walking. Sometimes actions are taken to inhibit growth in an attempt to prevent excessive height. This can be done with surgery or medication and requires input from an Endocrinologist.

Dural ectasia (widening or ballooning of the dural sac which surrounds the brain and spinal cord) can cause back pain, leg pain, abdominal pain, and headache amongst other symptoms.

Crowded teeth might require regular Orthodontist visits. The discomfort or pain from skeletal problems may need several adaptations to a child's routine or the equipment they use.

4. Eyes

People with MFS are generally short-sighted (myopic). In addition, some have dislocation of the ocular lens and/or retinal detachment due to the weakened connective tissues holding the lens in place. This may require surgery. This may affect their ability to see in class or when working.

Regular eye tests will be required; the child will be seen by an Ophthalmologist if there are problems with lens dislocation and if surgery is required. Any difficulty with a young person's ability to see will need to be accommodated.

5. Lungs

Spontaneous collapse of the lung (pneumothorax) with breathlessness and chest pain occurs in about one affected child in 20. This problem requires prompt attention but is usually not life-threatening.

6. Bowel

A proportion of patients with MFS have gastrointestinal disturbances with abdominal pain, bloating, constipation and diarrhoea. In the general population, IBS (irritable bowel syndrome) commonly first develops in young adults and teenagers. They may need to access a toilet frequently or quickly. Bowel issues requiring GP or Gastroenterologist visits, medication and dietary requirements may also need to be accommodated by school or college.

The requirement for appointments with several different specialists means that sometimes absences from school will be unavoidable to ensure the child is getting all the care and input they need to meet their full potential. For some pupils this will mean a significant disruption to their learning at times during their school life.

School/college accessibility and adaptations

Most children and young people with MFS will cope well in the regular school system but they may need adaptations and adjustments to achieve their full potential within the learning environment.

Legislation including the Equality Act 2010 and the Children and Families Act 2014 protects the right to these equal opportunities in law.

Children with MFS demonstrate the same range of abilities as all children. However, the variability of symptoms in MFS requires an Individual Education Plan

for each child and careful dialogue with the child, parents, school, and healthcare professionals will be needed.

Parents may require a legally binding "Individual Education, Health and Care Plan" if the school requires additional resources to make the necessary adaptations. Age-appropriate focus on a child's strengths and their educational requirements are vital.

Of note, children with MFS can be exceptionally tall and this can have a variety of consequences. They can appear much older than their chronological age and therefore often be treated as such. It is important to remember this and expect the same of them as their peers.

The child may outgrow small classroom furniture very quickly and need more appropriate chairs or desks to allow them to work comfortably and effectively; as the child moves into senior school, these adaptations need to be in place in all areas the child will visit. These differences can make the child feel "apart" from their peers and it is important to monitor their integration within the class.

The following are just a few examples of adaptations that might be required: (taken from the Disability Rights UK website, 2022):

- https://www.disabilityrightsuk.org/adjustments-disabled-students-and-apprentices
- https://www.gov.uk/topic/schools-colleges-childrens-services/special-educational-needs-disabilities

Regular dialogue with staff/teachers if the condition is hidden/fluctuating

Specialist or adapted furniture or computer equipment

Tailored examination arrangements such as extra time or supervised rest breaks as detailed in Exam Access Arrangements https://www.jcq.org.uk/exams-office/access-arrangements-and-special-consideration/

Arrangements to ameliorate impact of frequent medical appointments

Provision of work that can be done at home if the child is fatigued or in pain

Physically accessible classrooms, exam rooms, toilets, catering, and leisure facilities

Permission to stand or move around to avoid discomfort

These tables break down some of the issues that a child with MFS may experience, the impacts on their learning, and potential adaptations to minimise this.

Heart issues

Issue	Impact	Possible adaptations
Aortic dissection (rare in school-aged children but a medical emergency if does occur)	Fatigue	Take medical complaints seriously
Aortic root dilatation/ aortic aneurysm	Restrictions on lifting	Easy access to school nurse and place to rest
Mitral valve prolapse	Restriction on physical activities	Adapt schedule or deadlines to allow for rest if necessary
Irregular/erratic heartbeat	Medication regime	Provide additional time to get to classes
		Modify (PE) curriculum or offer alternative

Nervous system

Issue	Impact	Possible adaptations
Dural ectasia	Pain (often chronic) which can affect ability to sit and focus for prolonged periods of time	Easy access to the school nurse
	Difficulty participating in PE	Access to pain medication as required
	Difficulty completing homework/assessments on time	Adjust school timetable to give opportunities for rest/lying down
		Allow extra time for homework/ exams if needed
Gut discomfort	Pain	Arrangement to go home if necessary
	Need for easy toilet access	Allow for rapid access to toilet
	Lack of sleep	More frequent eating arrangements
	Need to eat smaller/ more frequent snacks	

Eyes

Issue	Possible impact	Possible adaptations
Severe short-sightedness	Vision may fluctuate	Provide large print books/texts
Dislocated lens	Difficulty reading for prolonged periods	Provide seating at the front of the class
Risk of detached retina	Difficulty reading small/ light-coloured fonts	Use dark/clear font on school materials
	Difficulty seeing whiteboard	Provide IT equipment with settings to accommodate visual impairment

Bones and joints

Issue	Impact	Possible adaptations
Tall stature with long arms, fingers, and legs	Difficulty sitting in standard desks and chairs	Provide desks/chairs at a suitable height for classrooms, home study and exams
Loose and flexible joints	Difficulty sitting for extended periods of time	Allow student to stand/move around if uncomfortable and provide a bed in a rest area if necessary
Underdeveloped muscles	Difficulty walking long distances	Provide extra time to get to class
Pain (often chronic)	Inability to carry heavy books long distances	Schedule classes in rooms closer together
Pectus excavatum or Pectus carinatum (chest bone that dips inwards or protrudes outwards)	Difficulty writing	Assisted mobility for longer distances
Curved spine (scoliosis)	Prone to joint injuries	Use of a lift if necessary
Flat feet	Muscle fatigue	Allow access to nurse for pain medication if necessary
	Body image issues	Modify PE curriculum or offer alternative
	Some students may require a wheelchair (rare)	Use aids for handwriting and consider scribe/laptop use in lessons and adapted exam conditions such as additional time for tests/exams
	Difficulty sitting on the floor	

Sports and activities

Regular exercise, important for the maintenance of good physical and mental health, is equally important for children with MFS.

MFS affects all individuals differently and it is therefore vital to plan safe access to PE and sports for children, considering their specific needs. **This will require close liaison with parents and advice from the child's medical practitioners so that children can maximise their participation whilst remaining safe.**

Recent research within the Marfan community has demonstrated that regular, moderate physical activity is beneficial in improving an individual's health-related quality of life and in early studies there has been evidence to show slowing of aortic dilatation in groups undertaking tailored exercise plans. This is an area that is constantly evolving.

A useful first step can be to provide parents with information about the PE/ sporting curriculum for the coming academic year so they can obtain specific, tailored advice from the doctors.

The key issues listed previously – heart, bones and joints, eyes, nervous system – are the same things that will need to be considered here. The aim is to help the child develop a healthy lifestyle incorporating physical activity whilst not putting them at increased risk of complications.

As a rule, children with MFS should be encouraged to choose low-impact, low-intensity activities and their expectations need to be managed: participating in sports at an elite or competitive level will not be possible and this can be difficult to hear, particularly if their diagnosis is made at a later age and they are already playing or competing at a high level.

Considerations during physical activities

1. Heart

Children may have a dilated aorta, a heart valve that leaks or may have already undergone heart surgery. They may be taking medications. The dilated aorta requires protection and contact sports should be avoided.

Heavy weightlifting or other isometric exercise should also be avoided due to the spikes in blood pressure which stress the heart and aorta.

Beta blockers will slow the heart rate and reduce the blood pressure; the pulse rate becomes less responsive to exercise, so it is harder for children to make rapid changes to their activity level and pulse rate is a less reliable way to assess their level of exertion.

If a child has had heart surgery, they may also be taking a blood-thinning medication and in these instances the avoidance of contact sports is particularly important as they will be at increased risk of bleeding.

2. Bones and joints

The child may have joint hypermobility or loose joints which put them at increased risk of injury. Their height and long limbs may also affect their hand-eye co-ordination. Lack of muscle bulk can lead to fatigue at an earlier stage than that of their peers. These children will need the option of resting during PE and taking part for short periods of time and to be given extra support to find sports and activities that they enjoy.

3. Eyes

Short-sightedness can affect co-ordination and may increase their risk of collisions. Appropriate glasses that can be worn during exercise will be an important consideration.

There is the risk of retinal detachment and lens dislocation, so appropriate eye protection should be used and contact sports should be avoided. It is important to listen to the child during physical activity and provide ready access to the school nurse if they report new symptoms that need assessment.

4. Guidelines for activity

Each individual is affected differently, and these general recommendations need to be discussed with the child and parents, alongside advice from their own medical professionals.

- Take regular low/moderate recreational aerobic activities.
- Avoid contact sports, high-level competitive sports and static/isometric exercise in which the muscles and blood vessels contract, causing spikes in blood pressure.

Try to maintain an aerobic level of activity (the child should be able to continue to hold a conversation whilst exercising)

Light weights can be used with higher numbers of repetitions rather than heavy weights with fewer repetitions

Wear protective equipment e.g. bike helmet

Incorporate adequate time for warm-up and cool-down

Avoid 'fitness tests' that pit children with MFS against other children; try to focus on personal achievements and achieving a 'personal best'

Activities such as football, basketball, cycling and dance can all be done at high, strenuous levels, but they can also be enjoyed at a more leisurely, low-impact level and children need to be taught that they can enjoy these pastimes but at a lower intensity

Regular physical activity is important and children with MFS should be encouraged to find a sport or activity that they enjoy

The age of diagnosis will impact the child's response to restrictions on their physical activity. If they have a pre-school diagnosis, their activities can be tailored from an early age and other activities can be incorporated, e.g. hobbies such as art, music and photography can foster a community and a social circle that is common within sport. If a child is diagnosed at an older age, they may already be participating in sports at a high level and be a valued member of a team that is an important part of their identity and social circle. This is challenging and can lead to them feeling excluded, angry and depressed if they can no longer be a part of this team. This requires careful management and can be incredibly difficult for a teenager.

Final advice should always come from the child's own doctor.

Absence from school due to ill health

Some children will require surgery or prolonged periods of time off school due to ill health. School can help minimise the disruption to their education by being understanding of the importance of these absences, planning appropriate work for the child to do whilst not at school and liaising with other caregivers, e.g. parents, hospital teachers/play specialists who can support learning in a different environment.

Planning for surgery

Planned surgery for any individual will be anxiety-provoking, but this can be particularly difficult for teenagers to deal with and careful liaison between parents and teachers is vital to smooth the path. Teenagers may have a range of worries and concerns:

- **Loss of control**
- **Fear of the operation itself and pain afterwards**
- **A change in their appearance due to scars**
- **Missing friends and family**
- **Fear of the unknown**
- **Missing a large period of time at school or clubs and wondering what people will think**

Teachers can be an important piece of the puzzle in the build-up to surgery; they can watch out for mood swings and behaviour changes.

In addition, they can modify work assignments and put together a plan for phased return to school so that the young person has a clear plan for the post-op period.

A teacher or pastoral head at school may be the trusted person that a teen wants to speak to about their worries and concerns; it is important that the school are aware of the plans for surgery so they can provide appropriate support.

Psychological wellbeing

Attention needs to be paid to the psychosocial and mental health of a child with MFS. There is limited research in this area, but it suggests that children and adolescents with MFS generally have poorer Health-Related Quality of Life (HRQoL) and mental health than their peers.

They may perceive themselves as 'different', perhaps feeling ostracised or bullied. They may understandably feel left out of exciting things due to absence or illness, missing peer group enjoyments.

Limitations to daily life due to fatigue and physical symptoms or different bodily appearance can increase social stigma and have an impact on psychological wellbeing and quality of life (Gritti *et al*, 2015).

The World Health Organisation defines HRQoL as the perceived (subjective) health-related physical, mental, and social functioning of children and adolescents.

Mental health is defined as a state of wellbeing in which children and adolescents realise their own abilities, can cope with the normal stresses of life, can study/work productively, and are able to contribute to community (World Health Organisation, 2007).

Handisides *et al* (2019) found that children and adolescents with MFS up to the age of 18 reported lower quality of life when considering physical, psychological, emotional, social, and school functioning than their peers.

Interestingly, young adults aged 19-25 in this study still had lower physical scores but their scores for emotional, social, and school/work functioning were higher than those of their peers.

It is possible to speculate that children with a chronic condition such as MFS can develop effective coping strategies, and this should be supported during their childhood years.

Support of a child's mental health requires close liaison between school and home: information needs to be shared between parents, teachers and pastoral staff, medical professionals, and the child in an age-appropriate way.

Bullying

The NSPCC (2022) defines bullying as 'behaviour that hurts someone else. It includes name calling, hitting, pushing, spreading rumours, threatening, or undermining someone. It can happen anywhere – at school, at home or online. It is usually repeated over a prolonged period of time and can hurt a child both physically and emotionally.'

Children with MFS may appear physically different to some of their peers; they may frequently miss school due to medical tests or appointments and this can put them at risk of bullying which needs to be monitored.

Children should be encouraged to talk to a trusted adult if they are experiencing bullying and careful attention should be paid to their behaviour by teachers/ parents and other care givers to look for any changes that may signify bullying. This needs to be dealt with swiftly and effectively with ongoing review.

As a result of being aware of being different, a child with MFS may have fragile self-esteem and can interpret innocuous comments as bullying which may benefit from discussion.

The fostering of an inclusive school ethos, valuing diversity, with an emphasis on kindness is crucial for all children's emotional development and wellbeing. Some children and parents will be keen to share the diagnosis of MFS. This should be encouraged, and the school can assist by giving them a platform to educate and inform teachers and peers. Others may wish to maintain their privacy and confidentiality, and this should also be respected as long as the child understands that some information needs to be shared with their teachers and other caregivers in order to keep them safe at school.

- https://www.youngminds.org.uk/young-person/coping-with-life/bullying/
- https://www.kidscape.org.uk/
- https://www.nspcc.org.uk/what-is-child-abuse/types-of-abuse/bullying-and-cyberbullying/

Clothing and shoes

Excessive height, curved back (scoliosis), protrusion of the chest, long thin arms, legs, hands, and feet cause great difficulty in finding clothes and shoes (including school uniform) which fit. This often results in a poor self-image and the school may need to be flexible with minor non-compliance.

Families may find it challenging to find all pieces of the school uniform that fit children and children may need to wear specially adapted shoes, for example to support lax ankles, flat feet, and unequal foot sizes.

Many online retailers now have ranges that cater for very tall individuals so appropriate clothes should be available, but costs of extra/different items may be difficult for families to meet if they are having to buy adult-sized shoes and clothes for their child.

School trips

School trips of long or short duration are an important part of school life, both socially and academically. Children with MFS should be included as far as possible, and this may require some advance planning to ensure that necessary adaptations can be put in place.

PLEASE ENSURE ALL ADULTS INVOLVED IN A TRIP ARE AWARE OF THE RARE POSSIBILITY OF MEDICAL EMERGENCIES DESCRIBED ON PAGE 2 OF THIS DOCUMENT.

Liaison with parents and students to discuss the trip and the activities on offer is important. As MFS has such a range of symptoms and severity, this will need to be done on an individual basis to meet the specific needs of each child. Further information or advice might be needed from the child's healthcare providers and the school nurse is a useful resource who will be able to help with this.

NOTE: parents will need to consent to any medical information being shared. Once a child reaches their teenage years, they will naturally begin to take more responsibility for their own health and should be involved in decisions. It is vitally important to include teenagers in these discussions so they feel empowered and start learning to weigh up the risks and benefits of different actions.

Career guidance and special interests

As with sport and physical activity, there may be restrictions on career pathways for a child with MFS. Physical issues such as poor eyesight, joint hypermobility, heart murmur or aortic dilatation can mean that pursuing careers that are physically demanding, e.g. armed forces, police service, building, nursing, elite sport may not be an option. Again, this will all depend on the child's individual needs and symptoms of MFS, and guidance from their medical practitioners is key.

Teenagers reaching the point of exam options and career choices need careful, tailored guidance. Suitable work experience placements need to be found along with tailored advice about career pathways. Transition from childhood to adulthood is a challenging time in any teenager's life, but with the added complication of a chronic condition this can be particularly difficult.

Conclusion

With support and understanding, children with Marfan syndrome can all lead a successful and fulfilled educational life. Young people with Marfan syndrome will experience this success and fulfilment more readily if the accommodations and understanding described in this document are in place. Such understanding and arrangements should greatly ease the path to adulthood and an appropriate future career. For further specific advice, please contact the Marfan Trust who should be able to direct you to the appropriate information.

■ With special thanks to the Pothecary family for their help and assistance with the creation of this information.

About the Marfan Trust

Co-founded in 1988, the Marfan Trust is the sole charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome. It is estimated that approximately 18,000 people are living with Marfan syndrome in the United Kingdom and around half of these remain dangerously undiagnosed.

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 self-funded Sonalee Laboratory, named after a young doctor who tragically died of complications from MFS during her ward round;**
- **Continue to provide educational information and raise awareness of the condition.**

How you can help

You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month: https://bit.ly/3GXoSVq

You can also donate to our charity:
BANK: Charities Aid Foundation (CAF)
ACCOUNT NAME: The Marfan Trust
SORT CODE: 40-52-40
ACCOUNT NUMBER: 00017677
REFERENCE: Your Name (plus campaign name if relevant)

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You can also contribute via:

- **Just Giving – http://bit.ly/3Scj51w**
- **PayPal Giving – http://bit.ly/3Z1TLxB**

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 visit our webshop: https://bit.ly/3Vj16qB

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

Registered charity number 1198847