Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders
Loeys Dietz

MARFAN MATTERS

Spring/ Summer '24 Edition

Patrons: Sir Magdi Yacoub FRCS | Dr Lady Maryanna Tavener MA, MBBS | Prof Marjan Jahangiri FRCS (CTh) | Prof John Pepper OBE, MA, MChir, FRCS, FESC | Mr Ulrich Rosendahl MD, FETCS

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Poised at the half-way point of another productive year, we look back on a six-month whirlwind of webinars, collaborations and connections, and forward to our Patient Symposium, Information Day and more. All along we have been continuing to renew our resources and expand our advisory panel, while we deepen our knowledge through exciting new projects.

New Patrons


Professor John Pepper OBE


Mr Ulrich Rosendahl

Patrons make things possible, and their influence is far-reaching. For some time now, we have privately dubbed Professor John Pepper OBE our patron saint in response to his invaluable advice and helpful interventions. We have now made it official and are proud to say that in February of this year, Professor Pepper accepted our invitation to become a patron of the Marfan Trust. And it doesn't stop here! His fellow cardiac surgeon, Mr Ulrich Rosendahl of the Royal Brompton Hospital, has also happily agreed to make the leap from Advisor to Patron. We are delighted!

Dr Anne Child on BBC Radio 4's The Life Scientific

Mixing the personal with the professional, Dr Child took to the nation's airwaves on Tuesday, 2 July at 9am to discuss her storied life and career.


Dr Child with Professor Jim Al-Khalili, recording her interview

For 'The Life Scientific', Professor Jim Al-Khalili focuses on an important field of research and interviews the scientist who made it happen, probing their inspiration, and exploring their journey. During his conversation with Dr Child he discovered all about Marfan syndrome and the lightning bolt of love at first sight. It's essential listening! **https://www.bbc.co.uk/programmes/m0020qjj**

YouTube

Our YouTube channel is filling with fascinating content, from high-octane webinars and supporter stories to medical professionals reflecting on Marfan syndrome. Check it out: ***www.youtube.com/@marfantrust3841***

You can help to secure our future by visiting our shop, making a donation, or becoming a member today for just £3 per month. **www.marfantrust.org**

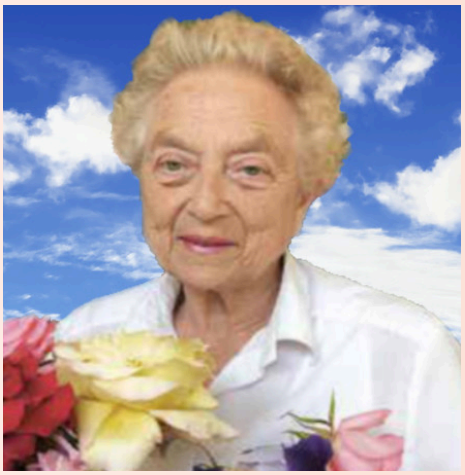
A Word from our Chair and Founder, Dr Anne Child

Dear Friends,

2024 is an exciting year for the Marfan Trust. Our membership is increasing, partly due to the social media posts written by our indefatigable Helpline and Communications officer, Victoria Hilton. Her imaginative journalistic ability has been a great help in raising awareness and solving members' problems, with the assistance of our Advisory Panel. She also hosts a monthly parent drop-in which helps facilitate conversation between parents of children with Marfan syndrome. We have promoted her to become a director and I hope you agree this is a good move. It rewards the many extra hours she devotes to guiding and supporting the aims of the Marfan Trust – support, research and education.

We also have good news regarding Dr Jose Aragon-Martin, Director of the Sonalee Laboratory, who has been awarded a permanent part-time Teaching Fellow position at Queen Mary's College, University of London. He will continue to be funded half-time by the Marfan Trust, and he and his students will analyse DNA from families with Marfan syndrome and related disorders (ascending aortic aneurysm, joint hypermobility, glaucoma, dislocated lens) in his two laboratories. One lab is at QMUL and the other at the Institute of Ophthalmology, where he supervises the programme of Glaucoma Research. Glaucoma affects 5% of all Marfan syndrome patients. He is also searching for new genes for dislocated lens, which is a feature of Marfan syndrome but does occur independently. Congratulations to Jose.

Our Clinical Nurse Specialist, Joanne Jessup, is known to many of you for holding online drop-in clinics for discussion of any and all problems. In addition, she is currently working with other specialist nurses across the UK to create an Aortopathy Nurse Network. The aim is to provide a network for sharing best practice, offering advice and support and working to increase the number of Aortopathy Nurses working across the NHS. Feedback from patients consistently highlights the importance of having a specialist nurse they can contact for support.



The Marfan Trust has now become a Charitable Incorporated Organisation No 1198847, entitled Marfan Trust (no 'The'). The charity itself remains unchanged, as do our goals. This is merely a change in legal structure. We are setting up a new bank account, but donations to the former The Marfan Trust will automatically be transferred to our new bank account.

Lastly, with no government funding, we are entirely reliant on the goodwill and donations of our members, who are most inventive with regard to fundraising. We have also set up a Goodwill legacy writing programme, and we hope you will all consider adding a sentence to your Will, leaving a small donation to the Marfan Trust.

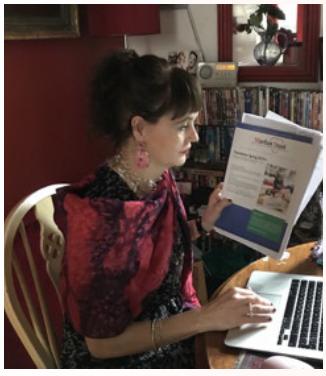
We are thriving, thanks to all our supporters, and plan to continue raising awareness in order to ensure early diagnosis and correct management for the estimated 18,000 affected UK residents, many of whom are as yet unknown to the medical system.

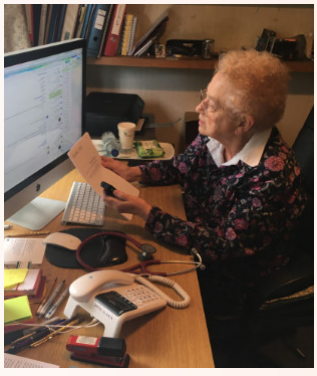
With warmest wishes,

Anne H Child

Dr Anne Child MD FRCP

Helpline Analysis By Victoria Hilton





Gathering momentum since Dr Child's appearance on 'The Life Scientific', the helpline is taking queries from far and wide. Of the 235 calls received this year, 35% stem from people hoping to make sense of a constellation of symptoms, wondering if they can be explained by a connective tissue disorder. How to approach and navigate a diagnosis is the burning question at the forefront of their minds. Approximately 15% of patients sought clarity on heart symptoms including aortic regurgitation whilst 7% approached the helpline for advice on their eyes – the wisdom of using heated eye masks to alleviate a gland dysfunction, for example. Prominent amongst remaining calls are concerns such as fatigue for which we provide coping strategies, and pain, an area in which we sometimes feel impotent to help. We are addressing this at our virtual conference on Saturday, 12 October when a pain specialist will speak on managing this debilitating symptom.

Alert Cards

In response to the potential risk faced by patients anticipating dental treatment and endoscopies/ colonoscopies, we've created an endocarditis alert card. Infective endocarditis is a very real threat to patients with some specific heart problems undergoing these minor procedures, but it's one that can be pre-empted with a simple prescription for prophylactic antibiotics. Order yours today.

We've also expanded our collection with a fluoroquinolone alert card. That which is supposed to help you can also harm you and fluoroquinolone antibiotics should be avoided by Marfan, Loeys-Dietz, and vascular Ehlers-Danlos patients as research shows a small increased risk of aortic aneurysm and dissection.

Peer Support

From making 'Marfriends' to meeting a mentor, our peer support service can offer a helpful conversation between Marfan supporters, or something more specific. We've expanded our network with lots of 'niche knowledge' so if you're seeking advice on securing an education, health and care plan for your child, or navigating the PIP system, just email us and we can pair you with the relevant mentor. If, however, you're simply seeking a cathartic conversation with a like-minded Marfan, we can arrange this too.

Chancey and Isabella

Friday Fact

Our social media presence continues apace with a new series, Friday Fact. These distilled, one-stop pieces on Marfan and its many manifestations give a comprehensive overview of such subjects as joint pain and ectopia lentis, and clarify the difference between seemingly similar conditions like kyphosis and scoliosis. At the time of writing we've completed 27 facts! **https://www.marfantrust.org/articles/category/157-friday-fact**. In the meantime, if you've been confronted with some confusing terminology that needs clarity, or a diagnosis that requires disentangling, then please get in touch and nurse specialist Joanne can help you: **info@marfantrust.org**

Booklets

Victoria and Joanne.

We've augmented our existing collection of booklets with Guides on Heart Surgery and Loeys-Dietz syndrome, and revised and updated existing titles including Dural Ectasia, Physical Activity and a new-look Introduction to our Charity. Download your free copies today. **https://www.marfantrust.org/pages/94-information-leaflets**

Casebook Special: Energy Deficit

From the helpline emerges our Dr Child's Casebook series which is exclusive to members of the Trust. Join the charity today and you will have unlimited access to these fascinating Q&As. Here is a sample to whet your appetite.

Casebook Special: Energy Deficit

Emerging from a long deep sleep only to feel just as fatigued as when your head hit the pillow is dispiriting. Many potential causes could underlie this phenomenon, from bad nutrition and stress to thyroid problems. Is a connective tissue disorder also to blame?

Q: I have Marfan syndrome and feel increasingly tired and lacklustre. Sleep doesn't seem to restore my energy which I severely lack. Even my bones feel tired. I'm only in my early thirties and this lack of motivation is affecting my job. What is the cause and what can I do?

A: It sounds as though you have fatigue as distinct from tiredness. Fatigue is felt in the muscles and joints. It is different from tiredness. Tiredness can be relieved by sleep and rest. Fatigue cannot, although sleep is obviously important.

Fatigue in Marfan syndrome can be a lifelong feature, partly since fibrillin is an important component of muscles, tendons and ligaments. If this is deficient, then these structures are stretchier and weaker than normal.

Defining fatigue is quite difficult as it is fairly intangible. One definition is “an overwhelming sense of tiredness, lack of energy and feeling of exhaustion, mental, physical or both” (Maher et al, 2015).

Velvin et al (2023) interviewed people with Marfan syndrome in a focus group and they used words to describe fatigue that included:

- Tiredness
- Exhaustion
- Lack of vitality
- Foggy feeling
- Parts of the body feeling heavy or paralyzed
- Difficulty concentrating, speaking and listening

Possible Causes of Fatigue

As I mentioned, fatigue can be symptom of Marfan syndrome partly due to the fibrillin deficiency. Listed below are the features of a connective tissue disorder that may well contribute to fatigue, but further work is needed to explore causal relationships and look for other possible contributors to fatigue:

- Chronic pain
- Joint hypermobility
- Reduced physical activity
- Pharmacotherapy
- Living with uncertainty

Other medical conditions also contribute to fatigue:

- Sleep apnoea (which is more common in MFS than in the general population)
- Insomnia
- Underactive thyroid gland
- Postural Orthostatic Tachycardia Syndrome (POTS)
- Anaemia
- Depression
- Medication side effects

It’s important that you and your doctor think about whether any of these might be affecting you as treatment may be effective in relieving the symptoms.

How Does Fatigue Affect Life?

There are numerous studies exploring fatigue in MFS and they highlight the significant impact that it has on all aspects of life.

One study interviewed parents of children with MFS and described how the combination of pain, fatigue and skeletal issues caused negative effects on their child’s physical activity, psychosocial development, education and quality of life. Children with MFS can struggle to keep up with their peers, difficulty with simple tasks like walking, running, throwing and kicking a ball may mean they are left behind at playtime.

Fatigue might mean that older children struggle to keep up with their friends if they have planned a busy day out and must make the decision to stay at home (Warninck et al, 2024).

Fatigue has an effect not just on the child, but on the whole family. Changes to family plans might need to be made, adaptations to holiday plans, days out or activities. This can be frustrating for siblings who might not understand, and difficult for parents to manage.

Adults report similar impacts on their day-to-day life. One study looked at ‘satisfaction with life’ (SWL) in people with Marfan syndrome. Respondents reported that one of the main things that reduced their SWL was severe fatigue, (aortic dissection reported as another important factor limiting SWL).

Coping strategies

It’s important that individuals struggling with fatigue find strategies to try and cope with it while continuing to find satisfaction within their lives. Some of these suggestions might help with the symptoms of fatigue:

- Keep a Fatigue diary for a few weeks, you may begin to see trends or triggers for your fatigue that you can address
- Plan your time wisely and avoid ‘boom and bust’ behaviour e.g. trying to clean the whole house when you are feeling good only to suffer the consequences for the next 2 days
- Take regular exercise
- Eat a healthy, balanced diet
- Reduce your caffeine intake
- Stay well hydrated
- Improve the quality of your sleep (you may be sleeping too little or too much, you can look up advice for good sleep hygiene)
- Deal with anxiety or depression – speak to your GP or refer yourself to Talking Therapies (https://www.nhs.uk/mental-health/talking-therapies-medicine-treatments/talking-therapies-and-counselling/nhs-talking-therapies/)
- Plan and prioritise – think carefully about what you really want/need to do and factor these into your day or week before adding in other activities
- Work
- Talk to your employer if you are struggling as there may be options for flexible working, working from home or other adaptations that could help you

School

- Talk to teachers and obtain support for children and young adults who may need adaptations or additional time or aids to reach their full potential at school

Physical Activity

Physical activity is a hot topic, frequently discussed by individuals with Marfan syndrome. Until recently, there had been a strong focus within healthcare settings on telling people with Marfan syndrome what they CANNOT do rather than what they CAN do. This can lead to people doing little or no physical activity as they deem it to be risky.

However, this needs to change, the physical and psychological benefits of exercise are well established, and these apply to people with Marfan syndrome as well as the general population. It's vital that appropriate regular exercise is built into a daily routine and can help combat fatigue.

Instrumental in this is health care professionals offering tailored, specific advice about the types of activity people can safely do. This will obviously differ from person to person depending on how Marfan syndrome affects them. You can read more about physical activity guidelines **https://www.marfantrust.org/pages/94-information-leaflets**

Along with this advice, there must be a greater focus on rehabilitation and physiotherapy to help people safely become more active. (Velvin et al, 2016). Rehab is available after events such as cardiac surgery.

There has been work done recently to look at the positive effect of physical activity upon the aorta. Research suggests that regular aerobic exercise can reduce the rate of change in the aortic root Z score and increase the elasticity of the aortic wall, which has great implications (Tierney et al, 2024).

[the full version of this casebook can be found in the Resources section of our website: **https://www.marfantrust.org/pages/84-resources]**.

Research Update by Dr José Aragon-Martin

Dividing his time between two prestigious establishments, the William Harvey Research Institute at Queen Mary University of London and the Institute of Ophthalmology, Dr Aragon-Martin continues to deepen his knowledge of connective tissue disorders and their genetic make-up.

Queen Mary University of London (QMUL)

This academic year 2023-2024 I am supervising five Queen Mary University of London MSc students' projects (4x April to September) and (1x June to December) to analyse NGS (Next Generation Sequencing) data on HTAAD (Hereditary Thoracic Aortic Aneurysm & Dissection), and hEDS (hypermobile Ehlers-Danlos syndrome) to find genetic biomarkers that might be causing these conditions; and also, a short project, one student with a literature review on EDS:

All lab-based projects are on 2x HTAAD families and 2x hEDS families:

1. Mrs Jeyabharathi Jeyaraj Kathirvelan is studying the DNA pieces that do not belong to the production of the protein, also known as introns.

2. Ms Roshanak Fekri Yazdi is studying the DNA pieces that come before and after the genes and between genes.

3. Ms Srivarshini Govinda Srinivasan is studying large DNA pieces that can be seen as a problem when there is less or more of this type of DNA than expected, also known as CNV (Copy Number Variations).

4. Ms Chandana Veshala is studying large DNA pieces that can be seen as deletions, duplications, insertions, translocations, inversions, also known as SV (Structural Variants).

All non-lab-based projects are done directly from the laboratory computers:

1. Ms Hsin-Yi Yang is trying to find the latest updates and news on the genetics of EDS by doing a literature search. (Ehlers-Danlos syndrome).

These two types of connective tissue condition share clinical features with Marfan syndrome (MFS) and finding more about its genetic markers will allow us to understand better MFS.

We have been supervising Ms Silvia La Penna for one of the summer studentships 2024: she was able to help us in the lab creating genetic family pedigrees, doing PCRs to finish last year MSc projects, and supervising MSc projects from this academic year.

Institute of Ophthalmology – University College London (UCL)

This academic year 2023-2024 we are supervising one UCL PhD student's project to analyse NGS data on PCG (Primary Congenital Glaucoma) to find genetic biomarkers that might be causing this condition:

1. Mrs Nicola Cronbach is studying the DNA pieces that change the production of proteins, also known as indels and point mutations.

This connective tissue condition shares clinical features with Marfan syndrome (MFS) and isolated Ectopia Lentis, therefore, finding more about its genetic markers will allow us to understand better MFS.

Research Manuscript Publications and Dissemination of the results

We are one of the authors in a review with one of our Ectopia Lentis (EL) collaborators, Dr Aman Chandra. This is expected to be published this year 2024. It is going to be an updated review on the genetics of EL.

We are going to be presenting at the ASHG (American Society of Human Genetics) 2024 in November our research findings from 2 of the MSc students. We are not allowed to discuss any of these findings until further notice, therefore, watch this space.

Research Grant Applications

Although we have been fully funded by Marfan Trust for many years since I have been the director of Sonalee Laboratory, we always continue applying for outside funding to try to help ease the burden cost of the lab. We applied to the British Heart Foundation back in January 2023 for a grant project on HTAAD for £350,000 for 3 years and resubmitted with review comments back in January 2024 for £260,000 for 2 years, however it has come back recently with an unsuccessful proposal, and now we can not apply for the same grant project again on the same topic, the HTAAD screening analysis. We are not completely sad with the outcome since we have already discussed this with other collaborators from our network, and they are experiencing the same outcome.

Our next step will be to apply for the same study but this time as a PhD project for 3 years. Professor Panos de Loukas , my QMUL Head of Department, is very influential and is helping us rewrite the project so it is suitable material for a PhD student. Fingers crossed!

THANK YOU

I just want to take this opportunity to say thank you, no, really, thank you very much. The support by donations to Marfan Trust has helped us to educate students, volunteers, and ourselves in these conditions that are yielding up their secrets as to their causative genes.

Summer Students

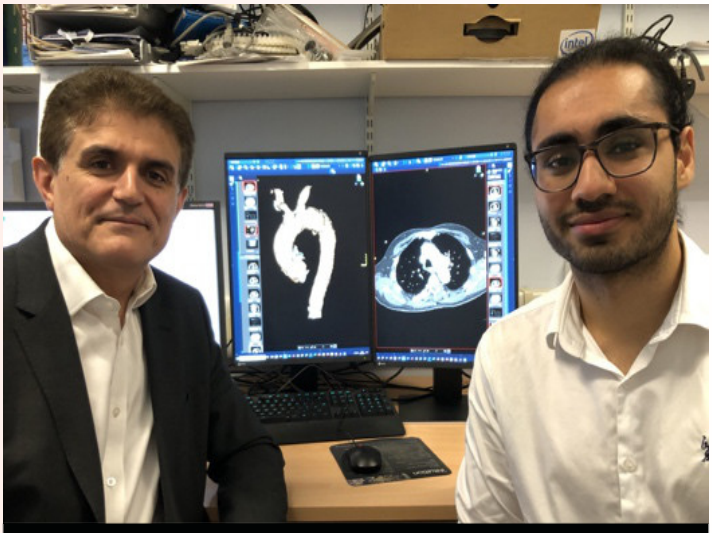
Whether exploring the previously under-explored, or enhancing our existing knowledge, summer students breathe fresh life and bring new ideas to the Marfan Trust. Described below are two current projects, the full results of which will be published in our Autumn/Winter edition newsletter.

The Eye in Marfan Syndrome

Fourth-year medical student Navaneethan Adityaraj has been filling an important gap in our literature. For several months he has been studying the potential manifestations for the eye in Marfan syndrome and has completed a comprehensively brilliant guide which is being reviewed by our ophthalmology advisor, Mr Aman Chandra. It will be published very soon.

Study of CT Scans and Aortic Measurements

Mehar Bijral, a fifth-year medical student from UCL, has been chosen for a £1000 studentship. For six weeks commencing July 11th, he will work closely with Dr Saeed Mirsadraee, Consultant Cardiothoracic Radiologist, Royal Brompton Hospital. Mehar's project involves analysing CT Scans from hundreds of Marfan syndrome patients. Measurements of the aortic wall are held on a database awaiting analysis. There are three groups; patients with no aneurysms (control group); patients with enlarged aortas (in both width and length), who have not dissected; and Marfan patients who have dissected and been operated on to strengthen the aortic wall.



Dr Saeed Mirsadraee (on the left), Consultant Cardiothoracic Radiologist, Royal Brompton Hospital, and Medical Student Mehar Bijral, studying a Marfan syndrome patient's Aortic CT Scan

This study should help answer the question;

"Are there any aortic measurements which predict aortic dissection?" Obviously, such a report would help the surgeons operate just before the aorta dissects, thus saving lives.

We wish Mehar and Saeed well with this important project. Medical student Mehar will report his findings at our Annual Patient Information Day online on Saturday October 5th (see announcement on Pg 15).

The Anatomy of the Aorta

Newly elevated to the status of 'organ in its own right' by the European Association for Cardio-Thoracic Surgery (EACTS), the aorta now sits on a par with the heart and brain. But what does this mean in clinical practice for something once viewed as "just a tube"? In the following piece we translate the EACTS's new guidelines into the context of Marfan syndrome. We also discuss the Marfan aorta.

Background

The aorta is responsible for carrying oxygen-rich blood from the heart to the rest of the body. However, it has become increasingly evident that it also plays an instrumental role in regulating blood pressure and blood flow velocity. In 2022, the EACTS council commissioned the review and revision of the guidelines treating acute and chronic syndromes of the aorta. Until now, aortic diseases had usually been treated either in cardiac surgery or in vascular surgery, depending on their type and location.

The Aorta in Marfan Syndrome

Marfan syndrome is characterised by a lack of connective tissue elasticity and this can compromise the wall of the aorta, causing it to dilate, bulge and tear (dissect).

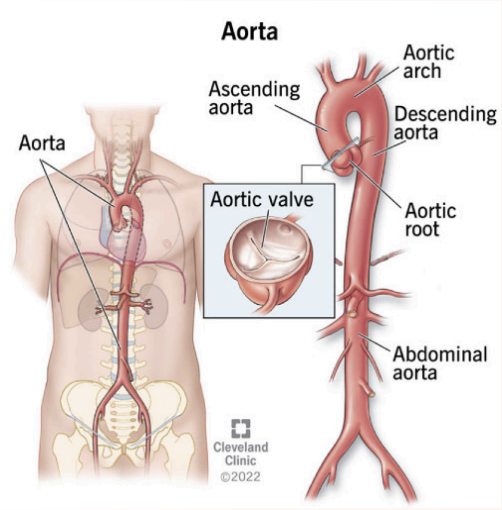
Our body's largest artery, the aorta arises directly from the heart's left ventricle and all blood pumped from the heart must first pass into this artery. After leaving the heart, the aorta branches into large vessels that carry oxygen-rich blood to the entire body. A valve, the aortic valve, lies between the left ventricle and the aorta and functions to keep blood, once pumped out to the aorta from leaking back to the heart.

The wall of the aorta consists of three parts:

1. A thin, inner layer (intima) is the 'tube' through which the blood passes. It contains smooth muscle tissue, connective tissue and endothelial cells. These special cells enable blood to transport oxygen and nutrients without getting absorbed until it reaches the right spot.

2. A thick, elastic middle layer (media). This layer is made of smooth muscle tissue, elastin and collagen (proteins). These substances enable the aorta to meet your body's changing blood flow needs. When more blood is necessary, the aorta widens. If less blood is needed, it narrows.

3. An outer layer (adventitia). The outer layer anchors the aorta in place. It also connects to nearby nerves and tissue.



In Marfan syndrome, the middle layer, the media is weaker than normal. Year after year, as the aortic wall faces the constantly changing stress generated by the beating heart, the wall, particularly in the aortic root which is the first two or four inches above the aortic valve

(sinuses of Valsalva) can gradually stretch. In this region, the diameter of the aorta, usually about one and a half inches (3.7cm) in an adult, widens (dilates). This dilated region is known as an aortic aneurysm. When the enlargement reaches 4.5cm, surgery is considered. If untreated, and the aortic root is left to widen, it may develop a tear – dissection - in the wall. This tear may proceed downward through the middle layer of the aorta through which blood rushes causing the inner layer to also tear which constitutes a dissection. This is the most serious complication someone with Marfan syndrome can suffer**. It is a time-critical medical emergency that when diagnosed and treated quickly has a better than 80% survival rate.**

The beginning portion of the aorta – the root - dilates to some degree in nearly everyone with Marfan syndrome. The echocardiogram is the simplest method for detecting and measuring the aorta in this region. This enlargement usually begins quite early in life and is detectable using echocardiography in most infants with the condition. It can be managed medically with beta-blockers and angiotensin receptor blockers if or until elective surgery is required.

New Guidelines in the Context of Marfan Syndrome

In recent years the aorta has been increasingly viewed by the medical profession as “more than just a tube”. Doctors began taking a holistic approach some time ago to diseases of the aorta (aortopathies), treating them in a multi-disciplinary context. This new attitude has steadily evolved, becoming cemented in the EACTS Guidelines which were published simultaneously with the aorta’s new classification as the ‘body’s 24th organ’. Leading the development of these new guidelines was Professor Martin Czerny from the University of Freiburg.

As Professor Czerny says: *“The new guidelines clearly recommend bundling the treatment of the aorta in a separate speciality, in close coordination with other specialities. We have been practising this integrative approach at the Medical Center - University of Freiburg for a long time and I am delighted that our work is now also being recognised internationally,”*

This is already starting to be practised in the United Kingdom with the creation of aortic teams within many hospitals . These new guidelines will support their development and help collaborative working and the creation of a streamlined approach to patients who need lifelong care and surveillance, like the Marfan community.

The health professionals needed to work together in a team providing care (the list is not exhaustive).

- Cardiothoracic surgeons
- Vascular surgeons
- Anaesthetists
- Radiologists
- Cardiology
- Rheumatology
- Endocrinology
- Genetics
- Specialist nurses
- Psychologists

Professor Czerny went on to say:
“These new guidelines [also] describe the procedure for diagnosing and treating diseases of the aorta such as aortic aneurysm. If the aorta dissects, emergency surgery is required. "These diseases require complex surgical interventions, which we can better understand, research and perform thanks to the holistic view of the aorta," says Czerny. "Very good follow-up care is also very important, as those affected often show dangerous changes in the aorta again later in life".

We have sought views from our Advisory Panel on this exciting development in the journey of the aorta.

Professor John Pepper opines:

“The aorta is a complex organ and not just a simple tube to enable the passage of blood from the heart to the rest of the aorta. The first part of the aorta is also a shock absorber which serves to reduce the power of the pulse so that by the time the blood reaches our organs; brain, gut, kidneys etc. the flow is almost continuous.

The wall of the aorta is made up of elastic tissue, muscle and a specialised type of jelly called the ExtraCellular Matrix or ECM for short. There is a lot of research going on into the ECM as it seems likely that small changes in this material in the Marfan aorta leads to enlargement of the aorta and eventually to the break-up of the elastic fibres. If these early changes can be detected by a simple blood test we would be able to anticipate such disasters as dissection or rupture, weeks or months before the event, and this would be much simpler and cheaper than performing frequent CT or MR scans. This lies in the future, but the future is not far away.”

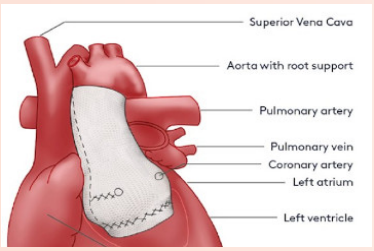
PEARS

The picture was taken at the Royal Brompton Hospital and is Dr Warren Thornton (CAD Engineer), Prof John Pepper(PEARS surgeon), Tal Golesworthy (patient and Project Engineer), Dr Peter Gibson (Exstent CEO) and Prof Tom Treasure (Surgeon)

A source of celebration, anniversaries mark significant events, none more so than the first PEARS procedure performed 20 years ago. On 24 May, 2024, we toasted the 1024 lives that are much better than they might otherwise have been.

The PEARS procedure was conceived by Tal Golesworthy. Unhappy with the existing surgical solution to his expanding aorta, Tal created an alternative, and became the first beneficiary of his elegant invention. As Tal says: "May 24th, 2024 marks the 20th anniversary of the first PEARS surgery carried out by John Pepper on my Marfanoid aorta."

PEARS (Personalised External Aortic Root Support)



A surgical procedure using a bespoke mesh support that wraps around the dilated portion of the aorta to prevent further dilatation and dissection (usually the aortic root and ascending aorta as shown in the image).

PEARS is an alternative to the traditional treatment for aortic dilatation which would be an aortic root/ ascending aorta replacement with or without replacement of the aortic valve.

History

The first PEARS procedure took place in 2004 and much of the surgery has been in people with Marfan syndrome, 73.5% of the first 200 patients undergoing PEARS had MFS (Van Hoof et al, 2016). While MFS was a majority disease type early on, it is no longer the majority of Aortic PEARS cases (as opposed to Ross-PEARS cases): 385/888 = 43%.

Each patient having PEARS requires a specialist CT scan which is used to create the personalised 3D engineered external support for their aorta which is then placed during surgery.

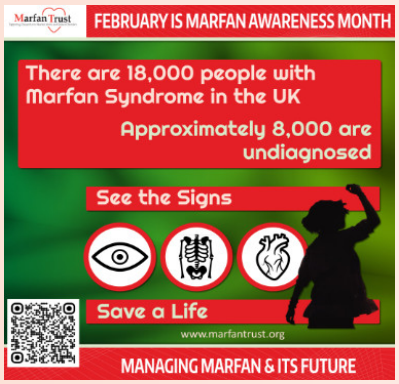
Potential advantages:

- usually, the patient does not need to be put on cardiopulmonary bypass
- the operation time is shorter
- the native aortic valve remains in place which means no anticoagulation medication is needed for a mechanical valve replacement.

Important

- This is still a relatively new procedure which requires a specialist surgeon
- PEARS is not offered at all centres in the UK, but you can ask for a referral for a second opinion
- PEARS will not always be an option and your surgeon will be able to explain the reasons

Marfan Awareness Month



Managing Marfan and its many manifestations was the theme of our awareness campaign this year. Throughout February we ran a series of articles, features, interviews, patient stories and videos on everything from navigating height and chest surgery to physical activity and the benefits system. We were very grateful to supporters who shared their personal experiences. The campaign culminated in a piece on the future for Marfan syndrome which looks increasingly bright and personalised.

Our virtual campaign reached over 30,000!

See the Signs and Save a Life! This became our call to arms during February and made its way onto a sticker which you can order from our shop: **https://www.marfantrust.org/pages/32-shop**

https://www.marfantrust.org/articles/category/74-marfan-awareness-month

.

Meanwhile, we took Marfan syndrome to the fields of Norfolk and the streets of Corby. We've selected some snapshots of our activities during February:

In memory of Jamie Morton, Downham Market Young Farmers have staged a series of fundraising events in 2024. Jamie was a young father and a happy farmer. During lockdown he was designated a 'key worker' and allowed to continue doing the job he loved. One day he fell ill and after many trips to hospital, it was discovered he had suffered a tear in his aorta. Despite two open-heart surgeries, Jamie tragically died. Read his story here: **https://bit.ly/3WTRzYX**

The first of the Farmers' fundraising and awareness events was a spectacular 98-strong tractor pageant which wound its way through the streets of Norwich on a very cold Sunday, before congregating in a field for food and a treat-filled raffle. Victoria met Jamie's lovely family and here they are, pictured left.

Marfan Matters in Corby! We are one of three lucky charities chosen by Corby-based construction company 7formation to benefit from their fantastic fundraising in 2024. Victoria met the lovely team in February and introduced them to Marfan syndrome!

Our website cover star, Amiyah Nason, ran her own awareness campaign during February, alerting everyone at school to the syndrome she suffers from. Amiyah won a star worker award!

In Memory

Len Jacobs

Len Jacobs was one of the longest-serving group support leaders for the Marfan Association/Trust, in the Bristol area. He recently died aged 89, and is sadly missed. He grew up in Ireland and the Clapham area of London, a very keen and skillful football player who later supported Tottenham Hotspur.

His work as a laboratory technician led him to become senior scientific officer serving the genetic unit at Guy's Hospital in London, where he was a patient of Dr Anne Child's. Mildly affected in the eyes, he subsequently went on with his loving wife Angela to have four children. Helen, Mark and Sarah were diagnosed with Marfan syndrome. Their fourth child Karen is unaffected. The family then moved to Bristol, and Len was always a good father to his children, telling them stories from his younger years and others completely fictional. Alongside Angela, he co-created a loving family home for everyone. After retirement he became a bereavement counsellor, mentor to students with additional needs, and prison visitor. The family are very appreciative that Len was cared for at home towards the end.

The Marfan Trust will sadly miss Len, who gave so much, and hope that the family continue to use our support.

Len Jacobs

Remembering John Hirst

By Karen, his wife

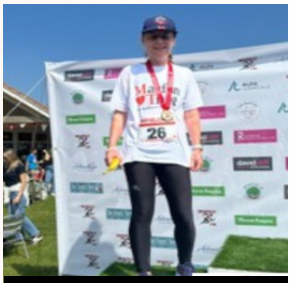
John was a very quiet and calm person, not a worrier (unlike me!) and was very well thought of in his job as a Programming Director for a large pharmaceutical company, working in clinical trials. Many, many people commented on how he was a "great boss": "if ever there should be a wiki page on how to be the perfect boss, John's name should be at the top"; how he was "a lovely gentle man" "such a warm and kind person ". (John would hate all this by the way, never one to 'blow his own trumpet'.)

Again, thank you for reading, I still find it therapeutic to talk/write about John, He was such a lovely person whom I miss every day.

This photo of John was taken at Wembley where he was supporting his lifelong passion – Bolton-Wanderers.

John died in September 2023 of an aortic rupture.

Karen has always participated in her local Binfield Run, and poignantly did so this year. It was the first without her quiet and calm husband greeting her at the finishing line. Karen raised over £1,000 in John's memory.



Karen

Feats of Fundraising

Without our fundraisers, we would not be able to continue our important work, improving and saving the lives of people with Marfan syndrome. Here we celebrate a few of their amazing feats.

Amelia's Story

With her quiet determination and wizard culinary skills, 11-year-old Amelia shrugged off recent knee surgery to complete a triathlon and bake for an entire village. What's more, she enlisted all her school friends in her fundraising mission for the Marfan Trust!

By Jj, Amelia's mother

Amelia Bidwell (11) took part in a Dorset schools triathlon relay with some of her school friends. They each swam 50m, cycled 1,000m and ran 750m on a scorching hot day. A few tears of nervousness were shed before the cycle, but Amelia persevered and completed the course, including running over the finish line with her 3 team mates.

Fundraising for the Trust had included baking cakes, soup and bread which she sold to the village and raised a total of £300 personally but then encouraged all the other girls in her school (Hanford in Dorset) to donate to this cause too.

Amelia has Marfan and had surgery on both knees in December and was also born with skewfoot. To her parents' surprise, Amelia chose to enter the triathlon, and occasionally doubted herself, but as a quietly determined character kept going at her own pace, and, encouraged by her friends, completed the task. She never ceases to amaze her family with her ability to succeed in everything she does - mostly with a smile on her face!

Brooke's Story

Uncle Jason (J-bird) Missed by everyone who knew him. Lived life to the absolute the full x

by Brooke

I got my legs into gear in February, ready for the Brighton Half Marathon.

This time, I ran for the Marfan Trust, in memory of my Uncle Jay / J-Bird / Ladybird but also as my Auntie Leanne is also currently in recovery from a major operation from also having Marfan.

Marfan is something that many are unaware of but something our family knows too much about.

It was an honour to run for this Charity & I was aiming to beat last year's time. This is something that is truly close to my heart.

Brooke raised £1,240 for the Marfan Trust. Thank you so much!

Cannonbawz Run

Many years ago, Kris O'Neill turned a long-held dream into a wonderful reality. He dreamt of a car rally that would help Marfan patients like his brother, Liam. This year marks ten years of the amazing Cannonbawz Run, an event that combines Kris' love of cars and Burt Reynolds with his urge to fundraise for the Marfan Trust.

Liam tragically died three years ago but his legacy is long and lasting, not least in the movie-themed Runs organised by Kris that started in 2014 with just seven cars and now number over 50! Last year alone, Kris and his 'bawzers raised £6,000 and this August will see over 50 cars embark on a 560-mile journey around the North Coast 500 with an overnight stop in Ullapool for a charity auction. Thank you, Kris!

Charlotte Grey

Running Matters for Charlotte! Exhilarated in the wake of completing the Edinburgh Half-Marathon, Charlotte has raised nearly £1,500 for the Marfan Trust. As a busy, newly qualified doctor with Marfan syndrome, this was no mean feat! Read Charlotte's story:

Charlotte approached our charity, offering to volunteer her time and expertise. As a doctor with Marfan syndrome (MFS), she is uniquely qualified to help us spread the word and raise awareness of the condition.

Charlotte wasn't formally diagnosed with MFS until the age of 17. Suspicions that she had Marfan, or something similar, emerged when she was only 19 months old and not walking. A paediatrician examined the little girl and noticed a constellation of signs that suggested a connective tissue disorder. But Charlotte didn't present a classic picture and the geneticist felt she perhaps had only marfanoid habitus (a condition in which a person has the skeletal features of Marfan syndrome but doesn't have any aortic or eye features). Therefore, she remained untested.

Nonetheless, Charlotte was monitored in Oxford with regular echos, as though she had Marfan syndrome. And all the while, she continued to grow taller and taller. At 13, she had her growth plates removed from her knees, which limited her final height to 6 foot 2. Then, at 16, Charlotte developed hip problems and was diagnosed with protrusio acetabuli on both sides, with osteoarthritis. She had a hip replacement at 19 but just prior to that, was genetically tested and found to definitively have Marfan syndrome.

Derek's Do

Emerging from the shadows to take centre stage at the Priory Lodge of Acton 1996's fundraising weekend in Bournemouth, rare diseases Marfan & Williams syndromes stole the spotlight in an evening of copious toasts, talks and dancing.

Derek Goodger lives with Marfan syndrome and is an unstoppable supporter of our charity. He has enlisted his masonic lodge into his fundraising and we have been the lucky beneficiary of their annual charitable Ladies Nights for some years. Over a sumptuous May weekend at the Connaught Hotel in Bournemouth, Marfan and Williams Syndromes were celebrated as Worshipful Master Alf and his son Jake introduced a large audience to these rare conditions. With a very generous supplemental donation from Derek, the evening raised £1,400.

Not only does Derek constantly raise funds but he also heightens awareness of Marfan syndrome, regularly attending PACES Ahead sessions - pre-exam practice and training sessions for doctors aiming to become Registrars and members of the Royal College of Physicians. Thank you, Derek.

7Formation

The sun shone on 7formation's fundraising football tournament in May as barbecue aromas filled the air. We manned a Marfan stand and watched the six-a-side game unfold.

We are one of three lucky charities chosen by Corby-based construction company 7formation to benefit from their fantastic fundraising in 2024. A staff member has a family connection to Marfan syndrome hence we were selected. Many local firms and suppliers took part in May's charity match, raising over £4,534! We are grateful for such generosity. Thank you so much, 7formation for everything you do! www.7foundation.co.uk

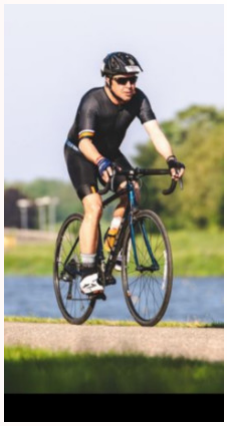
James Fountain

James Fountain

Commemorating a friend who blazed brightly in his relatively short time on earth, James swam, ran and cycled in memory of adventurous David whose life was cut suddenly short by complications of Marfan syndrome. James raised £1,200!

By James

My friend David Graham was a very funny, intelligent man who always made everyone laugh with his wit and was always at the centre of any party. He was a dedicated teacher in the Middle East and before that worked for many years in Interpol. He told me of the problem of his Marfan Syndrome inherited from his family and that it was rare and incurable. He just tried to enjoy his life to the full and as best he could. We will all miss him.



I raised funds with this Nottingham Outlaw Half Ironman, and after 6 hours and 53 minutes of running, swimming and cycling, I completed the 70.3 mile challenge raising £1,200. I want to help people like David who weren't able to have long lives. David tragically passed away in April 2023 aged only 47.

Danny Lovell

From dedicated smoker to committed runner, Danny took on the Birmingham Half-Marathon in May! His step-son, Sam, has Marfan syndrome and Danny was raising funds and awareness for a happy & healthy Marfan future.

"I ran my first ever half-marathon to raise money for Marfan Trust, a charity that's very close to us as a family. There are 18,000 people in the UK with Marfan Syndrome including our Sam. It's a condition that affects people in so many cruel ways and if me running half a marathon helps in any way, I'm glad to do it. I'm running this for Sam, as he's had so much to put up with his throughout his life, but just gets on with things with a strength and determination that I could never do.

Incidentally, Sam was the second beneficiary of the Thoracoflo™ aortic graft in the United Kingdom and is recovering well from the operation. Danny raised over £2,000!

James Stanhope - 80th Birthday Congratulations

Seen left to right, Sarah (grandson's partner), Jane (Jim's daughter), Josie (granddaughter), Jean (Jim's wife of 57 years), Jim himself, great-grandson Jagger, and grandson Josh.

For his 80th birthday celebration, Jim Stanhope invited 50 friends to donate to the Marfan Trust. He is seen here with his family, some of whom came from Australia for the celebration.

Thank you for thinking of the Marfan Trust, and for all the donations.

Stamp for Marfan

Every perforated picture tells a story. Stamps have celebrated and commemorated people, fashion, and events for decades. They have kept us connected in a disconnected world, ensuring the safe passage of letters between lovers, friends and families. And they make a tidy sum for the Marfan Trust. Pauline Moses and her late husband, Raymond, have been collecting stamps, old and new, turning them into donations for our Charity.

Ever tempted to rescue a used pretty stamp from an opened envelope doomed to the recycling bin? For many years, Pauline has been doing just that - collecting used stamps cut from envelopes for the Marfan Association and subsequently, the Trust. These stamps, together with any new unneeded ones, are sold to dealers for cash, raising money for our Charity and thereby transforming lives.

Raymond and Pauline's son Peter tragically died from complications of Marfan syndrome and Pauline herself lives with the condition. As Pauline says of her stamp-collecting enterprise:

Please simply cut, or 'carefully' rip off, the postage stamp from the envelope (leaving 1cm of envelope bordering each stamp) and being careful that you don't damage the stamp itself. Please retain the barcode. These can be stamps of any description and from any country. Please send to:

Mrs Pauline Moses, The Waves, Coast Drive, St Mary's Bay, Romney Marsh, Kent TN29 0HN

Collaborations

The Aortic Nurse Specialist

Amplifying the voice of the nurse specialist, The Aortic Dissection Charitable Trust held a Nurse Conference in June, which proved to be a wonderful day of learning, networking and collaboration to further develop and champion the vital role of Aortic Nurses for our community of patients.

Separately, Marfan nurse specialist Joanne Jessup is working with an ever-growing group of aortopathy nurses who meet quarterly to discuss how they can share best practice and further develop the role of aortopathy nurses across the UK. Nurses work as advocates for their patients and are a great link person for patients and doctors alike. We want to ensure that these important roles are rolled out to more and more centres across the UK.

Heads, Hearts & Minds

Exploring the previously under-explored psychological dimension of an aortovascular diagnosis in Marfan syndrome, Barts ICU Matron Rosalie Magboo discussed the fascinating results of her PhD at our joint webinar held with Aortic Dissection Awareness UK & Ireland.

Rosalie Magboo is an award-winning cardiovascular nurse at St Bartholomew's Hospital. She received a Barts Charity Healthcare Professional Clinical Research Training Fellowship in 2021 to study the under-researched health-related quality of life and psychosocial aspects of living with Marfan syndrome, before and after heart surgery.

Many people with Marfan syndrome develop heart abnormalities, requiring lifelong monitoring and, usually, at least one major surgery. Rosalie's research will help design ways for patients with this condition to improve their mental health and quality of life, before and after heart surgery.

Rosalie recruited around 120 patients to join her study. The webinar is available on our YouTube channel. Meanwhile, Rosalie's studies continue!

b+tlc CHARITY
Funding extraordinary healthcare
Queen Mary University of London
NHS Barts Health NHS Trust

NOTICE TO THE PUBLIC

Marfan Syndrome Mortality and Morbidity Outcomes Study
Re: Use of patient data for research purposes only

Description and Purpose:
Barts Heart Centre will be conducting a research study, generously funded by the Barts Charity and sponsored by Barts Health NHS, to identify the UK incidence and outcomes of aortovascular surgery in Marfan Syndrome (MFS) patients.

This study will help us to further understand the burdens that aortovascular problems place on Marfan Syndrome patients. It will also allow us to evaluate the effectiveness of the care we provide to this patient group.

Data needed for the study
We will be using routinely collected national data from NHS England (Civil Registration Mortality and Hospital Episode Statistics data) and National Institute for Cardiovascular Outcomes Research (NICOR). Data obtained from these three sources will be linked together using patients' identifiable information, such as NHS number, date of birth and date of surgery. Immediately after the data is linked, all patient's identifiable information will be removed, so that data analysis and results presentation will be based on pseudonymised data.

NHS England
Office for National Statistics
Hospital Episode Statistics (HES)
NICOR

Who will be included in the study:
All Marfan Syndrome patients who had aortovascular surgery in England between 1 January 2010 – 31 December 2019.
If you are one of these patients and you prefer that your data will not be included in the analysis for this study, please contact the Principal Investigator, Mrs Rosalie Magboo via email: rosalie.renamagboo@nhs.net
or simply scan the QR code:

Data Controller and contact details:
The Data Controller for this study is Barts Health NHS Trust and can be reached via email: c.g.colechin@qmul.ac.uk. The Data Protection Officer can be reached via: dpo.bartshealth@nhs.net.

Your right to complain:
We work to high standards when it comes to processing your personal information. However, if you have a concern about how our organisation handles your personal information, you have the right to complain to the Information Commissioner's Office: https://ico.org.uk/make-a-complaint/

In collaboration with:
Aortic Dissection Awareness
Liverpool Heart and Chest Hospital NHS Foundation Trust

Medical Management of Marfan Syndrome

A complex condition, Marfan is a lifelong challenge. Medically managing the manifestations of this disorder long-term can be difficult and Consultant Cardiologist Dr Alex Pitcher discussed this very issue on 5 February at a popularly attended webinar organised in collaboration with the Marfan Foundation. Dr Pitcher clearly demonstrated that ARBs have been shown to reduce the rate of aortic growth by half (studies are in patients with no previous aortic surgery). This seems to be an effect IN ADDITION to the effect of beta blockers. His current practice is to aim for his patients with Marfan syndrome to be taking both medications. The dosages can be built up over time and these medications can be taken by children and adults.

Pectus Matters

Not just a cosmetic concern but often a medical one, pectus excavatum can be a sign of Marfan. After much debate and many discussions, surgery for this sometimes life-impairing problem has recently been restored to NHS England. Advocates for the return of NHS funding included members of a wonderful new charity, Pectus Matters.

We collaborated with Pectus Matters on a lunchtime webinar in May and the recording is available to anyone who missed it or would like to refresh their memories. Tune in to hear the patient experience and how to navigate the current landscape for care.

These webinars can be found on our YouTube channel and here's the link: https://www.youtube.com/channel/UC8lxjjN_6xxfTo9E19NDHZQ

Conferences on the Horizon...!

London Patient Symposium – In collaboration with the Marfan Foundation

Rare diseases Marfan, Loeys-Dietz & VEDS syndromes will stand tall in the spotlight on 31 August at our International Patient Day in London. Marking our first collaboration with the Marfan Foundation, TADCT & Annabelle's Challenge, the symposium will assemble medical experts and patient advocates from across the globe to empower patients & their families with the information they need to live their best lives. It's a rare opportunity for rare diseases, giving supporters the chance to hear directly from the expert and create connections with fellow patients

With talks from Dr Anne Child on Living Well with a Connective Tissue disorder, Joanne Jessup on Preparing for Heart Surgery, and Trust advisors Mr Robert Henderson and Mr Hilali Noordeen . **https://www.marfantrust.org/pages/83-events**

Marfan Information Day: Saturday, 12 October

Marfan and its manifestations including pain and dural ectasia will be discussed at our virtual conference on Saturday, 12 October. Speakers include Mr Aman Chandra and Dr Yaso Emmanuel. The results of our summer student projects will also be unveiled.

Book your free tickets today!

https://www.marfantrust.org/pages/83-events

The Marfan Trust Online Shop


Willow

Tote Bag

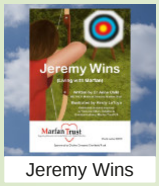

Marfan Pen

Marfan Cap


Xmas Keyring

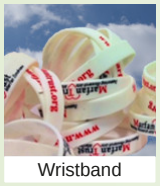

Marfan Mug

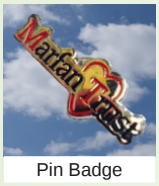
Cycle Tops



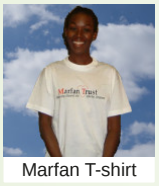
Jeremy Wins

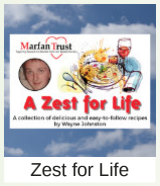

Scrunchie


Wristband



Pin Badge


Marfan T-shirt



Zest for Life

Marfan/LDS Band


All Occasions


Robin Card


Three Kings


Angels


South Bank

Watermill

LDS T-shirt

Welcome to a shop, a trove of lovely things, newly expanded with the Watermill Christmas card, LDS merchandise and more.

Visit Our online shop for a larger selection of merchandise.
www.marfantrust.org/pages/32-shop

☐ I am eligible for Gift Aid: (please tick if relevant and leave name and address below)

Signed ..... Date .....

Please return the response slip to: Marfan Trust, 24 Oakfield Lane, Keston, Kent, BR2 6BY. Please make cheques payable to the Marfan Trust. Thank you!

Name .....

Address .....

Email .....

Please include cost of postage (£3) in sum total. All cards are in packs of 10.

Item									Price	Quantity	Total
Cycle Top NEW	Small		Medium		Large		XLarge		£43.00 (each)		
Marfan/LDS Wristband NEW									£2.50		
LDS T-shirt NEW	Small		Medium		Large		XLarge		£6.00		
Marfan Trust T-shirt	Small		Medium		Large		XLarge		£6.00 (each)		
Willow, the Marf Giraffe									£5.50		
Jeremy Wins Book									£8.00		
Marfan Trust Tote Bag									£3.50		
Marfan Trust Pen									£2.00		
A Zest for Life Cookbook									£6.00		
Hand-crocheted Xmas Sparkle Red Heart Keyring									£3.50		
Marfan Mug									£9.00		
Marfan Trust Wristband									£2.50		
Hand-crocheted Scrunchie									£3.00		
Marfan Cap									£10.00		
Marfan Trust Pin Badge									£2.00		
Watermill Card (10 cards) NEW									£4.00		
Marfan Trust All Occasions Card (10 cards)									£4.00		
Robin on a Snowy Branch Card (10 cards)									£4.00		
Three Kings of Orient Card (10 cards)									£4.00		
Song of the Angels Card (10 cards)									£4.00		
Snow on the South Bank Card (10 cards)									£4.00		
Postage cost									£3.00		
SUM TOTAL (including postage)											