Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders
Loeys Dietz
CONNECTING THE UNCONNECTED AND STRENGTHENING LIVES

MARFAN MATTERS

Spring/Summer '25 Edition

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SPECIAL FEATURES

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Steeped in a searing heatwave at the time of writing this summer newsletter, we are keeping things cool by quietly preparing for our Birmingham symposium. Hovering on the not-so-distant horizon, our patient conference is a collaboration with Birmingham Women's and Children's Hospital featuring specialists dispensing their expert advice on managing the symptoms of Marfan and Loeys-Dietz syndromes. It will also, just as importantly, offer the opportunity to mix with fellow supporters, ask questions directly of the experts and share stories. A dedicated playroom will be available for children, along with a fun space designed for teenagers. Places are limited for this 6 September conference so sign up today through the Events link on our website!

Summer at the Marfan Trust spells the arrival of medical students who join us for a six-week research project every year. Since 2017, 15 students have deepened our knowledge and explored the underexplored symptoms of Marfan syndrome. We take stock of our alumni in a report on page 5. In the meantime, pictured right is our 2023 research student Elsa Harte who presented her continuing research into neonatal Marfan syndrome at the European Society of Human Genetics in Milan. Congratulations to Dr Harte!

From supporter stories and fundraising feats to medical updates, conference reports and more, this is a newsy newsletter. Happy reading!

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**

Marfan Trust, 24 Oakfield Lane, Keston, Kent, BR2 6BY

A Word from our Chair and Founder, Dr Anne Child

The first half of 2025 has been full of activity as we expand to serve the 3,700 UK families that we know about, whilst raising awareness to reach those with Marfan syndrome, Loeys-Dietz syndrome or new familial aortic aneurysm syndrome who are as yet unidentified.

Aortopathy Network

We have developed great links with aortic clinics across the UK, primarily through the amazing nurses working within these services. We ensure they can signpost to our charity and provide information and leaflets about the support we can provide. You can read more about this ongoing work on page 7.

Future Marfan Care

We are now planning two information days per year - our regular virtual conference in October, and a live, in-person event over the summer. The dates for your diary this year are: Saturday 11 October – a remote day for everyone to attend, and Saturday, 6 September – our live Patient Information day in Birmingham. We are asked frequently about whether gene therapy can be applied to those with Marfan syndrome and hope to hear the answer to this question at our virtual conference. Other topics regarding the future of Marfan syndrome care and research are included in the programme. Don't miss it!

Welfare Benefits

The recent government legislation regarding disability benefits is of great concern to many of our supporters, who fear losing their essential funding. The good news is that those already in receipt of benefits will remain unaffected. New criteria are being applied to new applicants. However, the welfare budget must still be trimmed, and our supporters may be subject to reassessments, in an attempt to reduce payments. Rest assured, the Marfan Trust remains happy to supply essential support letters, to attach to applications for continued support.

Support Groups

Victoria is facilitating the creation of patient support groups in areas with large numbers of Marfan syndrome supporters.

Fatigue

Many of our supporters suffer from fatigue which governs their lives. This is especially difficult to quantify or explain during an assessment. A summer student is doing a postal survey of 1000 supporters to assess the importance of fatigue and pain in our population. The results will be presented at our October 11th meeting, and contributed to a wider European study of fatigue, its causes and treatment. We hope to have new understanding of this symptom by early 2026. If you have received a fatigue questionnaire, please do reply.

Trustee/Treasurer Wanted

We are actively seeking a new trustee/treasurer to join our group of 7 faithful trustees. The volunteer must have certified chartered accountancy training, hopefully with some charity experience. The position will require 2-3 hours per week, and would suit a retired or retiring chartered accountant. May we please hear from any patient, relative or friend who would be willing to volunteer. This is a remote position, and the applicant could work from home, joining quarterly meetings by zoom.

We are also seeking a new fundraiser, because with our plans for expansion, your donations and legacies are extremely helpful, but we do need to actively fundraise ourselves as well.

Legacy

A special thank you to the LIL MacKenzie Trust for their generous three-year legacy which has allowed us to hire Joanne Jessup, Nurse Specialist, to provide valuable patient support in the form of Zoom conversations, and remote clinics.

Please keep your summer donations coming in. We have lots of branded t-shirts and cycling shirts to give away to those participating in walks, runs or cycling events.

Every penny counts!

With warmest wishes,

Anne H Child

Chair and Medical Director, Marfan Trust
Senior Research Associate, Imperial College

Helpline .... a Lifeline


Victoria Hilton, Dr Anne Child, Joanne Jessup

by Victoria Hilton

Eternally busy, our helpline is forever expanding to meet demand, and we have recently introduced online clinics with our cardiac nurse specialist, Joanne Jessup. Running every other Friday afternoon, these structured appointments allow a 20-minute conversation and 'consultation' where the patient's situation can be clarified, and advice dispensed. If you're newly diagnosed, seeking advice on symptoms or simply searching for answers, you can navigate your way to an appointment through the Events link on our website.

Questions to our helpline remain dominated by people suspecting they, or their relative, may have a connective tissue disorder. In response, we discuss their signs and symptoms before advising on their next steps and, when appropriate, helping them on the path to diagnostic certainty. The patients sometimes return to our helpline after echo and ophthalmology tests have revealed all to be reassuringly normal, fearing that with no aortic, or eye involvement, they will be denied a genetic test and left in diagnostic limbo. If there is no family history of a connective tissue disorder or unexplained early death in a close relative, then the geneticist will often not proceed to genetic testing in these cases. When the patient in question is a young child with "marfanoid features" and on the cusp of the pubertal growth spurt, parents naturally worry their aorta will change during this time. We recommend that children are reviewed again by a cardiologist with a repeat echo at the age of 18 to ensure there has been no enlargement. Assuming the aorta is normal, it is highly unlikely the child has MFS, but if there have been changes, then a further referral to genetics would be indicated for a review and possible genetic testing.

To clarify the complexities of diagnosis, we held a webinar, The Genetics of Marfan and Loeys-Dietz Syndromes, led by Professor Anand Saggar. This is the link to his fantastically comprehensive presentation and subsequent audience discussion: **https://www.youtube.com/watch?v=HH00z93HLnA**

Remaining hotline questions are disparate and manifold in nature. Approximately 25% of calls emerge from patients seeking advice on their heart and aorta, with queries ranging from blood pressure worries and palpitations to the regularity of their aortic scans.

Symptoms of fatigue and pain are closely intertwined and keenly felt by at least 30% supporters who approach us for advice. Loose and lax hypermobile limbs and joints must work much harder than normal, rendering them prone to exhaustion and painful dislocation. We are exploring the correlation between pain, fatigue and Marfan syndrome in a questionnaire which was distributed by post on 1 July.

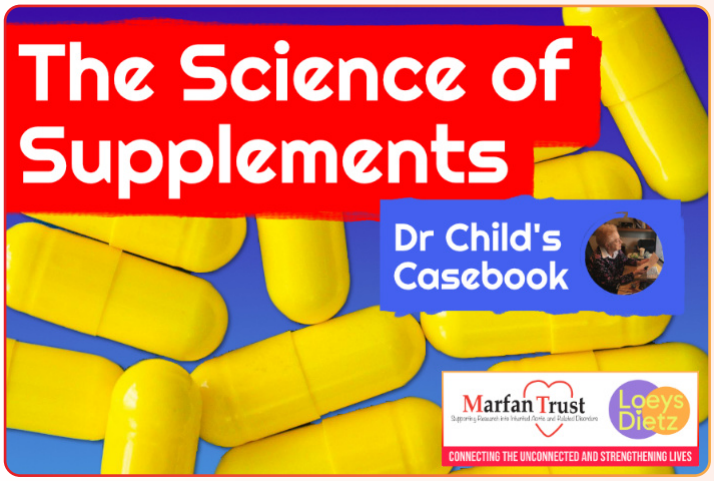
The ocular manifestations of Marfan syndrome trouble many supporters who often ask us if a dislocated lens should be removed or remain? The criterion is whether the ectopic lens is affecting the vision, says Mr Aman Chandra, Consultant Ophthalmic Surgeon. So, if partial and not affecting vision, removal may not be necessary.

Since January of this year, we have taken 300 patient enquires from eight different countries, a mix of medical, financial and emotional pleas for support. In response we often direct people to the Information Leaflet section of our website. An invaluable resource and trove of advice, our library of guides on the many manifestations of Marfan and Loeys-Dietz syndromes is constantly being reviewed, revised, and expanded. We have recently updated our existing pamphlets "Growing Older with MFS & LDS", "Pregnancy: and the "ENT Aspects of MFS & LDS", so download your free copies today: **https://www.marfantrust.org/resources/category/7-information-leaflets**

New Advisor to our Helpline!

The recent appointment of Natasha Woodgate has added an invaluable dimension to our helpline. Natasha is an occupational therapist with Marfan syndrome and a bottomless well of practical advice, guidance, tips and tactics on living with a connective tissue disorder. She recently led a fantastic webinar, Navigating the Education System with Marfan and Loeys-Dietz syndromes, **https://bit.ly/43WQaoK**, and is speaking at our virtual Information Day on Everyday Life and Marfan Syndrome. (see page 15 for details)

Dr Child's Casebook: The Science of Supplements



Hugely hyped, the collagen market is a burgeoning business with its promise of strengthening ageing skin and improving painful joints. But do these supplements actually work? And does increased collagen production compensate for diminished fibrillin in Marfan syndrome?

Q: I have a query about collagen supplements which seem to be quite heavily marketed, especially to women. Are these supplements helpful and is collagen important for Marfan syndrome patients, especially women during the perimenopause? As for symptoms, my joints ache so I'm up for trying any options!

A: I'm so sorry you are having joint pain. It does make you feel tired and limits your lifestyle.

Membership Alert: By joining our Trust for just £3 a month, you will receive exclusive access to Dr Child's Casebook. We have published a sample to whet your appetite.

To elaborate: female hormones are protective so when they start to disappear, you need to find strategies to maintain your bone and joint health despite this. Collagen accounts for 30% of your body's protein and provides structure, support or strength to your skin, muscles, bones and connective tissues. As you age, collagen production correspondingly diminishes.

Osteoporosis means that the bones become gradually weakened and more prone to fracturing. It is sometimes only diagnosed when a fracture occurs. Osteopenia precedes osteoporosis and means that you have lower bone density than other people of your age. Risk factors include: Women losing bone density after menopause due to falling levels of oestrogen.

If you are doing anything stressful such as gardening, standing for a long time, or driving a long distance, joint supports can readily be purchased for wrists, back, or knees, and worn during the activity.

Yoga or Pilates are also exercises that can help with core strength and stability which helps maintain balance and good posture

A healthy diet rich in calcium and vitamin D can be beneficial (The UK recommends that everyone take a vitamin D supplement of at least 10 micrograms a day during autumn and winter as we don't receive sufficient sunlight.) Foods rich in Vitamin D include oily fish.

Take regular weight bearing exercise – this means an exercise that requires you to carry your body weight, like walking, dancing, stair climbing, gardening etc. Swimming is still a great exercise for people with MFS but it is not weight bearing.

Women may consider using Hormone Replacement Therapy (HRT) both to prevent menopausal symptoms and to maintain bone density. Not everyone is able to take HRT and this will need to be discussed with your specialist doctors e.g. cardiologist.

Medication can be marvellous if you find the one which works for you. I find Brufen, starting with 200 mg twice daily, then 400 mg twice daily even up to 800 mg twice daily is the most effective for patients. You can purchase the 200 mg tablets over the counter. If you find it helps, then go to your GP and ask for a prescription. Take with, or after food.

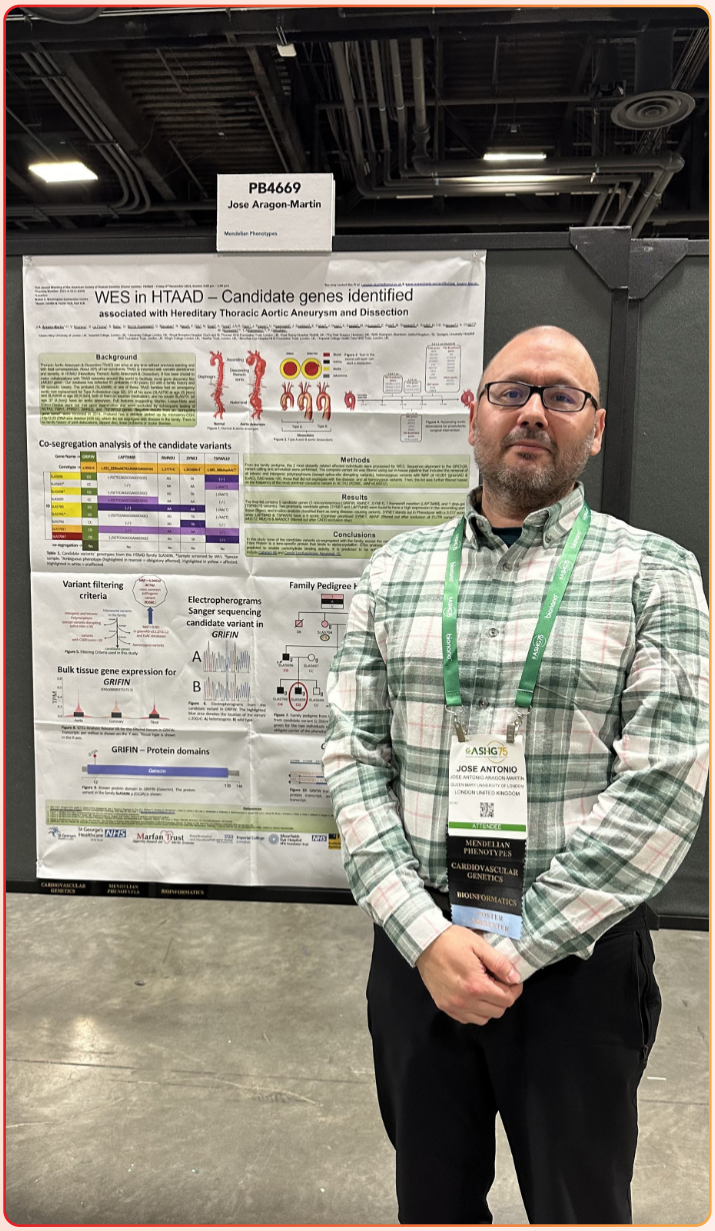
One a day multivitamin/multimineral may help with collagen production - a pathway with many cofactors. Fibrillin and collagen exist in bundles around joints so Increased collagen production may compensate for diminished fibrillin production.

Stretching exercises performed daily seem to help the nerve endings under strain from lack of fibrillin support. One of my patients who can't take medicines because of sensitive stomach, stretches every morning and that keeps her pain at bay.

With every large joint, take it to its extreme limit, put it under firm but gentle stretch and HOLD it there to count of 5. She was given this tip by her physiotherapist.

A physio who offers ultrasound or soft laser local treatment may be helpful. Swimming or just exercising/ stretching in warm water may also help. Keep your weight low, and this will help back, knees, ankles, and feet. Arch support insoles for flat feet are a must. Podiatrists make these. Ankle support boots or trainers help too. Take your inner soles and try them in new shoes before purchasing. Make sure you have room for toes. If knees hurt at night try sleeping with a pillow under your knees.

I hope these tried-and-true remedies are helpful. By all means, try oral collagen but if you already have a balanced diet with good protein, it may not make any difference. After all, it is broken down into amino acids in the digestive system. Then it has to be incorporated into new protein. That's where the genetic defect lies - on the fibrillin pathway. And you get the amino acids in your normal dietary protein (fish, meat, eggs, cheese milk).



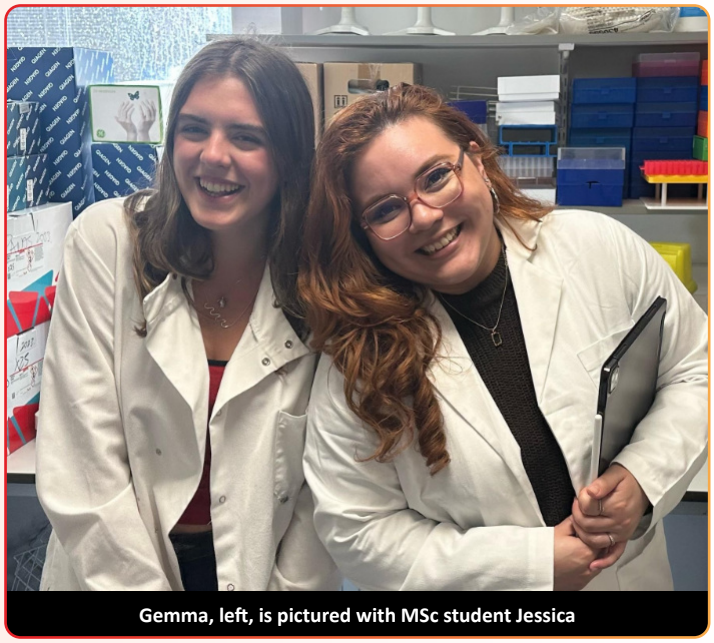
Dr Jose Antonio Aragon-Martin

At the Sonalee Laboratory, our research is focused on uncovering the genetic causes of Marfan syndrome and related connective tissue conditions such as Heritable Thoracic Aneurysm and Dissection HTAAD, hypermobile Ehlers-Danlos syndrome (hEDS), ectopia lentis, and scoliosis.

We are collaborating with Professors Mansoor Sarfarazi (USA) and Mariya Moosajee (University College London) on a project involving primary congenital glaucoma. Samples have now arrived in our lab and will soon be sent to Qatar University for free next-generation sequencing, once the final paperwork is completed.

We're also working closely with Professors Panos Deloukas and Aung Oo on a funding application for a PhD project investigating the molecular causes of aortic aneurysm in families with Marfan syndrome, and other connective tissue disorders who currently lack a genetic diagnosis. Standard tests usually focus on DNA that codes for proteins, but our approach goes further. Using long-read sequencing technology from Oxford Nanopore, we are now able to study non-coding regions and structural variations in the genome, providing new opportunities to uncover disease-causing variants that traditional methods might miss.

As part of this effort, we are also learning how to process whole genome (WGS) and whole exome sequencing (WES) data to detect structural variants—large-scale changes in the genome that may be missed by standard analysis pipelines. We're gaining hands-on experience with tools such as DELLY2, LUMPY, MANTA, VAQUITA, and others to improve variant detection and interpretation.



Gemma, left, is pictured with MSc student Jessica

Our team includes MSc, summer, and work experience students who are actively contributing to pedigree studies, literature reviews, and genetic analysis. Their work has led to four poster submissions for ASHG 2025, covering topics from neonatal Marfan syndrome to genotype-phenotype associations.

Thanks to support from Queen Mary's University of London, we have secured new freezer space that will allow us to store additional samples and continue expanding our research.

We are truly grateful to the Marfan Trust supporters for your continued fundraising; your commitment makes this work possible.

Finally, to say that you can always visit our Lab, as Gemma (pictured above) did recently!

Summer Studentship Programme

by Dr Anne Child

Summer Medical Student of 2023, Laxmi Thileepan

Since 2017, 15 summer students, all medical students, have applied for a six-week funded research post, to pursue a research topic of great interest to our supporters. This has included nine female and six male students. They have applied from medical schools all over the UK, and one from India. Some working remotely and some actively with staff in London. Speciality supervision has been arranged through members of our advisory committee. All projects have been presented remotely to our supporters, in our October annual national information day. Most recently, this day was attended by over 159 supporters.

Funding is provided from supporter donations, and each student is awarded £1.000. In addition, travel funding is provided if the topic is presented at a conference nationally or internationally, reaching a wider audience.

Results

Their results have been used for information pamphlets and videos posted on our website and distributed throughout our Marfan clinic network. They have also inspired webinars. One student is co-author of a chapter on gastrointestinal problems in Marfan syndrome.

Research topics have included diagnosis and management, newborn Marfan syndrome, prenatal diagnosis, eye disease, pain management, DNA mutations, growth management, GI problems, CT scans, and psychological impact on families, which was the project of Laxmi Thileepan, pictured left.

Summer 2025

This summer's students will be tackling fatigue in Marfan and LDS syndromes, update on pain management, and digital pedigree preparation in families with aortic aneurysms, under investigation for new causative genes.

Impact

A survey of the impact of these projects on the students has indicated a 100% enthusiasm, with students enjoying their time, learning research techniques which in one case has led to a career in medical research, and in all cases, leading students to an understanding of Marfan syndrome diagnosis and management.

In one instance a Marfan Trust student, now a medical graduate who was in A&E at 3am admitted a 30-year-old man with unexplained chest pain and shortness of breath. The doctor suspected Marfan syndrome, a chest x-ray revealed pneumothorax, and an echocardiogram revealed a slightly enlarged but intact aorta. The pneumothorax was treated, and two days later the patient left, free of chest pain, and with follow-up genetic and cardiac appointments. His family was also screened for Marfan syndrome.

Annual Aortic Nurses Symposium

By Joanne Jessup (seen here on left)

On Friday 6th June, Birmingham hosted the 2nd annual Aortic Nurses Symposium. Around 80 health care professionals met for a full day of lectures, round-table discussions and networking, with the ultimate aim of improving every aspect of care for people with aortic conditions, like Marfan syndrome. I was able to catch up with colleagues I know, as well as meeting new members of this community, nurses had travelled from all corners (from Plymouth to Liverpool and everywhere in between!).

The morning began with some great talks from leading lights in the area of aortic surgery, Dr Elefteriades (Professor of Surgery at Yale) and Mr Mark Field (Aortic Surgeon at Liverpool Heart and Chest Hospital). They shared useful insights into the clinics they have established at their hospitals (with the help of the aortic nurse specialist) and generated discussion about the key ingredients for a service offering quality care to everyone undergoing aortic surgery.

We also had enlightening talks about 'Genetic Screening' and 'Managing Hypertension'. Dr Osio, a Consultant in Clinical Genetics discussed the plans for improved access to genetic testing. While this is unfortunately not a reality for many of our supporters at the moment, the NHS Genomic Medicine Service are working to improve efficiency and deliver genetic tests in a timely manner. Dr Kapil, a specialist in hypertension at the Barts Heart Centre, was accompanied by one of his patients who survived an aortic dissection. They had an honest discussion about why your healthcare team are constantly asking about your blood pressure and the ways in which we can better support you in managing your blood pressure; from listening to your experiences of side effects, to giving you the skills and knowledge you need to confidently measure and monitor your blood pressure at home.

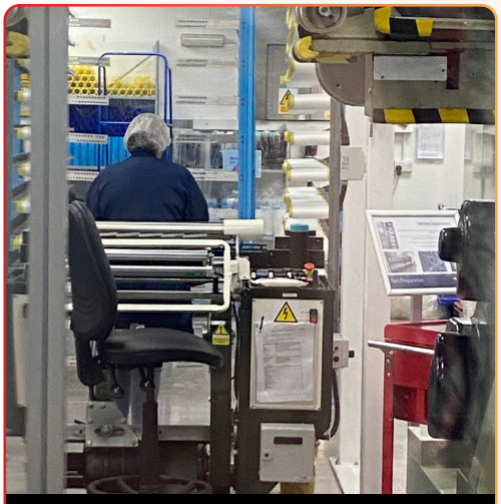
As always, some of the most powerful talks we had were from people who have been personally touched by aortic dissection or a genetic diagnosis that has impacted the whole family. These volunteers bravely shared their stories and talked honestly about their experiences of care and recovery. These insights are invaluable in helping health care professionals reflect on the care they are providing and the services they are building to ensure that they always place patients and their relatives at the centre.

In addition to the annual conference, the Aortic Dissection Charitable Trust (TADCT) are helping us as nurses to build a 'Community of Practice' (CoP) for health care professionals working in this specialist area. I am part of the Working Group to build and develop our CoP, and able to champion the Marfan/LDS community, bringing a different perspective to the group from those working exclusively in the NHS.

Please remember that you can always contact us via the helpline **info@marfantrust.org** if you are struggling with any aspect of your care. We are here to listen, support and offer practical advice.

Visit to Terumo Aortic in Glasgow

Victoria Hilton & Eileen Novins (Marfan Foundation)



The Textiles Room

Fun Fact: Knitted material allows for pliability and porosity. It means the graft can mimic the natural properties of the blood vessels and integrate with the body.

Woven material are used for vascular grafts. These grafts need to have controlled porosity and mechanical strength, to function effectively as replacements for natural blood vessels.

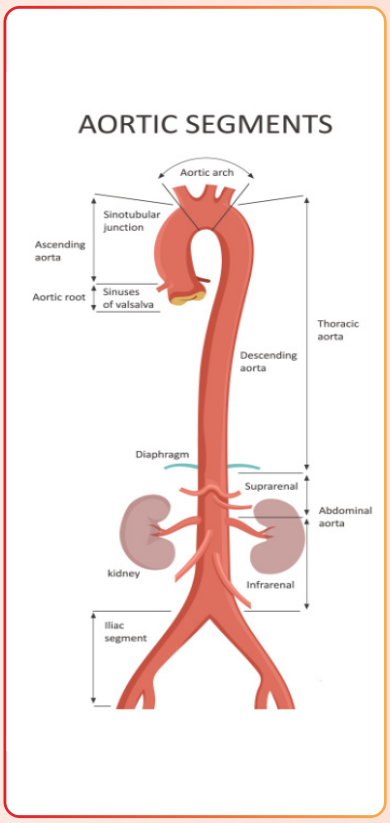
The Surgical Grafts Cleanroom

by Victoria Hilton

A factory with a difference, Terumo Aortic handcrafts the “tubes of life” which sustain 5,600 000+ (and counting) patients across the world. Originally conceived in 1921 as a thermometer manufacturer, Terumo Corporation has evolved to become a global leader in the creation of medical devices, including those for people with aortic and peripheral vascular disease. Eileen Novins from the Marfan Foundation invited me to join her on a tour of this fascinating factory where patients’ faces adorn the walls, reminding employees that every working day, they are saving someone’s life.

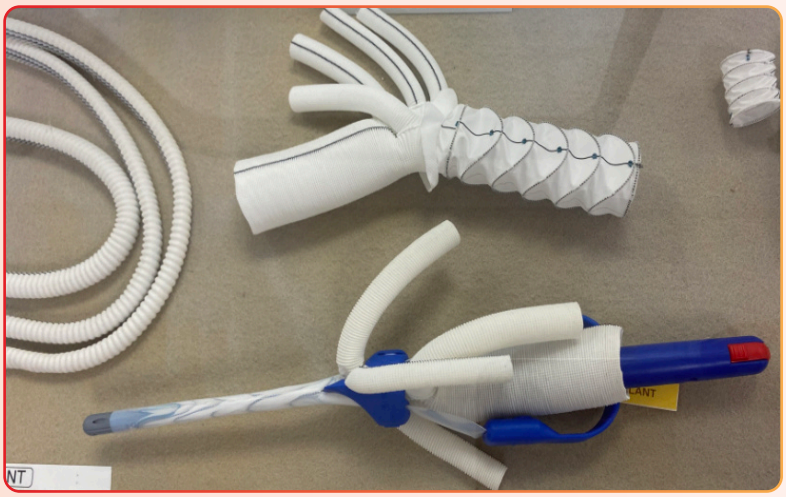
The tour was a revelation! Chief Commercial Officer Tarik Pacuka greeted us and whisked us through the decades, as Terumo expanded its products and locations, from making thermometers in Japan to creating disposable syringes and blood bags, before launching the world’s first microporous hollow-fibre membrane oxygenator (a device used during heart bypass surgery to exchange gases, oxygenating the blood and removing carbon dioxide) in 1982, marking a significant development for the new cardiovascular arm of the business. This recently formed cardiovascular enterprise is now split into three divisions, of which Terumo Aortic is the fastest growing. Underpinned by a relentless drive for quality and a continuity of dedicated staff, Terumo Aortic produces an endlessly evolving, comprehensive range of endovascular, hybrid, aortic root replacement and custom devices to treat both ascending and descending aneurysms, amongst other aortic conditions. And the “consumer” is the clinician.

It starts with a thread! From a single thread grows a portfolio of life-saving devices. Our tour took us down long corridors off which lie vast rooms, each filled with a different and distinct area of expertise. The product begins life in the Textiles Room, a cavernous space occupied by enormous beams of yarn, the strands of which are finer than a human hair. Painstakingly fed by hand into various machines, the yarn is wound, twisted, and warped, before being moved on to the knitting or weaving machines, creating a flat fabric to be turned into a tubular graft.



From Textiles to Polyester, the product has reached stage 2 of its creation where the flat fabric is washed, crimped, heated, and handsewn into a graft. (Crimping ensures the graft expands once it reaches its target site; heat setting stabilises the shape and structure of the material). To facilitate X-rays, metal rings are sewn into the fabric.

Its next step depends on its ultimate purpose – a straight graft for a thoracic aortic aneurysm for example, or something more intricate and complicated such as a Gelweave Valsalva for aortic root replacement procedures, in which case it is sent to Sewing for “Extras”. The Valsalva device wears a "skirt"!



Sporting blue spacesuits and stripped of make-up and jewellery (scary), Eileen and I were ushered into the Hybrid Thoraflex™ Sewing Room where magic happens. First approved in 2012, the Thoraflex™ Hybrid device is designed for the open surgical repair of aneurysms and/or dissections in the aortic arch and descending aorta. Pictured left, the four arch branches which facilitate the reconstruction of the major aortic branch vessels, emerge from manually made fenestrations and the tubes are then handsewn into place. The dexterity of the craftsmen was something to behold as they mimic mother nature with their creations.

Continuing the graft's journey, we visited the delivery system department and watched as these blue contraptions, used by surgeons to release the implant into the patient's body, were hand assembled. This is the penultimate step before gel-impregnation, sterilisation and packaging. Boxes are piled one on top of the other and distributed, breathing life across the world.

Thank you to Val, Laura, Tarik, Erik, Kevin, Colin, Sharon, Ian and Robert for an unforgettable journey!

Mind the Aorta!

More Fun facts:

- Terumo was founded in Japan by a group of scientists led by Dr Shibasaburo Kitasato and named Sekisen Ken-onki Corporation which means Red Line Thermometer.
- Terumo Aortic creates 1,000 products a day and 150,000 grafts a year.
- From inception to completion, the medical devices take on average three weeks to make.
- Every product goes through 300 touchpoints!
- Up to 3 million metres of thread is twisted every day.
- Terumo Aortic has plants in America, Vietnam and Japan, in addition to Glasgow.
- Fusing traditional open surgical techniques with minimally invasive endovascular approaches Terumo's hybrid devices are designed to "outlive the patient". The most recent addition to their range is the Thoracoflo. "Current techniques for thoraco-abdominal aortic repair are associated with serious complications and the problem of spinal cord ischaemia remains unsolved. The Thoracoflo device uses the frozen elephant trunk principle. By avoiding a thoracotomy and extracorporeal circulation, it is associated with reduced trauma to the patient".

Pictured from l to r: Erik Pomp (Chief Executive of Terumo Aortic), Victoria Hilton (Marfan Trust, Eileen Novins (Marfan Foundation) and Tarik Pacuka (Chief Commercial Officer, Terumo Aortic]

Marfan Awareness Month

by Victoria Hilton

See the Signs, Save a Life was the rallying cry behind this year's awareness campaign. We posted prolifically on social media, sharing something new every day. Supporters generously allowed us to tell their tales of diagnosis and it was revelatory. Alongside the more standard diagnostic paths were many chance discoveries – a patient's long fingers placed flat on a GP's desk that sparked a suspicion in the clinician's mind, for example. Rich wrote of how, in 1985, he visited his GP with backache and happened to place his hand on the consulting table. The GP had just read about a rare condition called Marfan syndrome, and immediately spotted Rich's slender fingers. After peering into Rich's mouth to find a high-arched palate, she sent him to Southampton Hospital where he was definitively diagnosed. These stories sparked much engagement and started many conversations on Facebook. We interspersed them during the month-long campaign with medical titbits, infographics, research pieces and more. It was a momentous month!

- We enjoyed our widest audience yet on Facebook with a 16-fold increase in the number of accounts reached, and a much higher-than-average (for non-profit organisations) level of engagement.
- We surpassed 1,000 followers on Instagram.
- We secured an article in Optometry Today which has a Facebook following of 26k. **https://www.aop.org.uk/ot/news/2025/02/26/optometrys-life-saving-role-in-identifying-the-signs-of-marfan-syndrome**
- We held a webinar, The Genetics of Marfan and Loeys-Dietz Syndromes with Professor Anand Saggar.
- Our amazing supporters ran fundraising campaigns throughout the month and really spread the word!

During February, I visited Preston to discuss the creation of a local support group and travelled to Cirencester where artist, novelist, and Marfan patient Lindsey Erith introduced me to her publication, "Wanton Troopers". Set during the English Civil War, this historical romance follows defeated royalist Hugh Malahide who is "armed by force of character and a charm-the-birds smile as he sets about seduction, burglary and defying Fate". **https://bit.ly/4nHOUifHu https://bit.ly/4nHOUifgh**

Thank you to our supporters for sharing their incredible stories and spreading the word on Marfan and Loeys-Dietz syndromes!

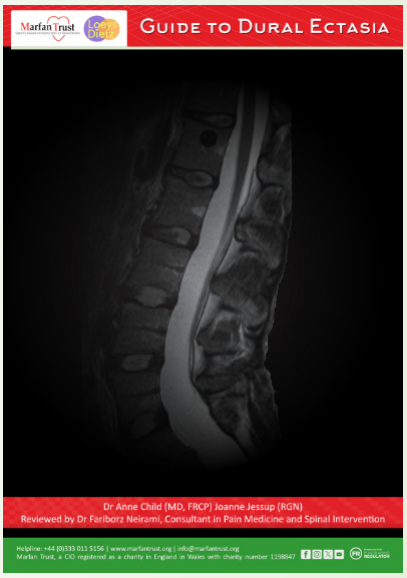
pictured from l to r: Victoria Hilton, author Lindsey Erith, Literary Agent Martha Halford-Fumagalli

PEARS: 25th Anniversary

Twenty-five years ago, in March 2000,engineer Tal Golesworthy discovered that his aorta was reaching the threshold for surgical intervention. Unhappy with the existing solutions on offer, he conceived an elegant, less intrusive alternative. His revolutionary invention was realised four years later, in 2004. Tune in to this fascinating interview in which Tal discusses the genesis and evolution of PEARS (Personalised External Aortic Root Support): **https://bit.ly/4iPeXRX**

Download Our Publications

https://www.marfantrust.org/resources/information-guides-



Stamp for Marfan

Every perforated picture tells a story. Stamps have celebrated and commemorated people, fashion, and events, keeping us connected in a disconnected world. And they make a tidy sum for the Marfan Trust. Pauline and her late husband Raymond Moses have been collecting stamps, old and new, turning them into donations for our Charity, in memory of their beloved son Peter who was lost to complications from Marfan syndrome. 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). Retain the barcode if there is one but if no barcode leave quarter of an inch. These stamps can be 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

Drawing Competition

Life can be a split screen of “Instagram versus Reality” and Rebecca Limb depicted this brilliantly in her wonderful drawing on the theme of Living with Marfan syndrome. She was runner-up in our recent competition “A Picture is a Thousand Words: Convey Life with a Complex Health Condition” and here is her fantastic piece. We will publish the joint winners of this competition in our next newsletter.

by Rebecca

This is my drawing, 'Insta Vs Reality'. The Instagram side of the drawing depicts how I'm often perceived by others and the life I try to lead - one full of joy and optimism. The background is made from words that describe this part of my life. I'm a lawyer and academic, I'm a daughter, sister and friend. I'm sporty, enthusiastic, and have a zest for life. I drew myself in my wheelchair because my chair such a positive part of my life. I celebrate my chair for enabling me to take opportunities.

The reality side shows the medical aspects of my Marfan that I rarely show to others. Here I depict myself attached to my TPN (Total Parenteral Nutrition) which I am connected to for 15 hours a day. I have TPN due to intestinal failure caused by my Marfan. The words in the background describe some of my Marfan symptoms including chronic pain, intestinal failure (and the need for nutrition via TPN), dislocations and hypermobility, eye conditions, cardiac conditions, skeletal symptoms, dural ectasia and fatigue.

In Memory

Joan Fearnside

We were very sad to hear that Marfan Trust supporter Joan Fearnside died earlier this year at the age of 99. Joan has frequently helped our charity since her son James Fearnside, tragically passed away at just 42 in 2002. James had been diagnosed with Marfan syndrome. James’ partner, Annette, wrote to the Trust to say that they had raised £130 in lieu of flowers at the funeral for our charity.

Fundraising Feats

Unstoppable in their support, the Marfan Trust fundraisers make a big difference to our small charity. We receive no government help but rely solely on the goodwill of our community. We feel eternally grateful for their amazing feats of charity and celebrate a small selection of fundraising highlights from the year.

Cannonbawz Run

Taking a rugged road trip through the contrasting scenery of Scotland while shining a light on Marfan syndrome, the Cannonbawz Crew were in action this May! They drove the gorgeous North East 250 route, passing whisky distilleries, mountain ranges and famous castles, while raising funds and awareness for our Trust. Helmed by Kris O'Neill, the Cannonbawz Run is in its eleventh year and has loyally supported not only our charity but many others. Thank you, Kris and your amazing Crew. To read Kris' story, follow this link: **https://bit.ly/4evolqw**

Amy Fletcher

Before: Amy with Wayne

After: Amy with Victoria

A cathartic anniversary and a bold buzzcut! To mark a full year since her fiancé's release from hospital after aortic surgery, hairdresser Amy celebrated in dramatic style, by shaving her head. In doing so, she raised £6,000 for the Marfan Trust and emerged looking even more stunningly beautiful. Victoria from the Trust visited Amy's hair salon in New Milton to witness the act and meet her family and friends.

Turning to Google for health advice isn't always wise but in Amy's case it saved a life. Her partner Wayne had struggled with a series of disparate symptoms for many years which were worsening over time. Amy was convinced something lay behind them, while doctors were treating each symptom in isolation. Armed with an A4 sheet scrawled with concerns, Amy joined the dots and Wayne has been diagnosed with Loeys-Dietz syndrome. Read their story by following this link: **https://tinyurl.com/7ww9nmwm**

Fundraising Feats

Donna Fullerton

Clad in tartan and Marfan Trust t-shirts, Donna Fullerton and her husband completed April's 19,000-strong Glasgow Kiltwalk and flew the flag for Marfan syndrome. Donna, her late sister, her mother and children all have the condition. The walk was especially resonant and emotional for Donna when they by chance played her late sister's favourite song at one of the checkpoints. Congratulations to Donna for raising almost £1,400 for our Charity. She was a little sore afterwards but also exhilarated! Here is her story! **https://bit.ly/3Y1rWY3**

Natasha Gillings

Natasha ran the Paris Marathon in memory of her son Micah was only 19 when he died in July, 2020. He was destined for university. While practising his cooking skills one evening in preparation for leaving home for the first time, he collapsed onto the floor. It was later discovered he had suffered a fatal aortic dissection, a result of undiagnosed Marfan syndrome.

This is the third marathon Natasha has completed in Micah's memory and she raised over £1,800.

Tom and Tabitha

Tom ran for a better future during the Brighton Marathon on 6 April, completing the race in just three hours and 37 minutes, and surpassing his fundraising target by over £600. Tom's partner Tabitha survived an aortic dissection in 2021 only to find out she has Marfan syndrome. "Tabs" has since made a full recovery but not every story ends this happily: 50% of dissection patients die before reaching specialist care. Early diagnosis of an aortic condition is key and to draw attention to this, Tom joined the Brighton run. He raised over £1,600 and you can read the full story here: **https://tinyurl.com/yzyk62y5**

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**

Marfan Information Days!

Birmingham Patient Symposium: Saturday, 6 September
IEC Birmingham, Austin Court, 80 Cambridge St, Birmingham B1 2NP

A day filled with expert information, peer support and so much more, our Birmingham Conference is filling up fast. We are collaborating with Birmingham Women's and Children's Hospital on this exciting, free event and many of their specialists will be speaking at the symposium.

Featured Speakers:

- Dr Deborah Osio, Consultant Geneticist: The Genetics of Marfan & Loeys-Dietz Syndromes
- Dr Chikermane, Consultant Paediatric Cardiologist: Paediatric Aortopathies
- Dr Paul Clift, Consultant Cardiologist: The Heart & Marfan/ Loeys-Dietz Syndromes & Ageing Well with a Connective Tissue Disorder
- Dr Dawn Adamson, Consultant Cardiologist: Pregnancy & Marfan/Loeys-Dietz Syndrome
- Mr Ian Hunt, Consultant Thoracic Surgeon: Chest Wall Surgery
- Dr Suad Duale, Consultant Psychologist: Mental Health & Marfan Syndrome
- Ellie Musgrave, Patient Advocate

further speakers to be announced nearer the time.

Food will be provided throughout the day and there will opportunities aplenty to mix and mingle with fellow supporters.

The day will be evenly mixed between formal presentations and break-out sessions. There will be many opportunities to ask questions directly of the experts.

We are offering travel bursaries for low-income households. Email **info@marfantrust.org** to find out more.

Victoria visited the venue, a Georgian warehouse, newly converted into a conference space. It's beautiful and sits peacefully on the canal-side.

Marfan Information Day – Virtual Conference – Saturday, 11 October: 9.15am - 4.30pm Zoom

An annual staple, our remote Information Day this year will feature talks on a wide array of subjects including:

- Gene therapy
- The Vascular implications and treatment of Marfan & Loeys-Dietz syndromes
- Chest Wall Surgery
- Dural Ectasia
- Occupational therapist Natasha Woodgate will discuss everyday life with these connective tissue disorders, offering an abundance of practical tips and advice.
- Fatigue & Marfan Syndrome
- Science in the Sonalee Laboratory

(the full programme will be announced soon).

Sign up today! **https://www.marfantrust.org/pages/83-events**

Newsletter assembled & edited by Victoria Hilton
Marfan Trust, 24 Oakfield Lane, Keston, Kent, BR2 6BY

The Marfan Trust Online Shop


Willow

Tote Bag


Marfan Pen

Marfan Cap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Marfan Mug

Cycle Tops

Jeremy Wins

Water Bottle


Wristband

Pin Badge


Marfan T-shirt

Zest for Life

Marfan/LDS Band

Woolen Beanie

Green Willow Bag


Three Kings


Angels


South Bank

Watermill

LDS T-shirt

Welcome to our shop, a trove of lovely things, newly expanded with our branded water bottle 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	Small	☐	Medium	☐	Large	☐	XLarge ☐	£43.00 (each)		
Marfan/LDS Wristband		☐		☐		☐	☐	£2.50		
LDS T-shirt	Small	☐	Medium	☐	Large	☐	XLarge ☐	£11.00		
Marfan Trust T-shirt	Small	☐	Medium	☐	Large	☐	XLarge ☐	£11.00 (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		
Red Heart Keyring								£3.50		
Marfan Mug								£9.00		
Marfan Trust Wristband								£2.50		
Marfan Trust Water Bottle								£15.00		
Marfan Cap								£10.00		
Marfan Trust Pin Badge								£2.00		
Watermill Card (10 cards)								£4.00		
Beanie Hat					Black	☐	Grey ☐	£8.00		
Green Willow Tote Bag								£5.00		
Three Kings of Orient Card (10 cards)								£4.00		
Angels Card (10 cards)								£4.00		
South Bank Card (10 cards)								£4.00		
Postage cost	Fixed charge no matter how large the order							£3.00		
SUM TOTAL (including postage)										