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

MARFAN MATTERS

Autumn/Winter '25 Edition

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From left to right: Research Director Dr José Aragon-Martin, Nurse Specialist Joanne Jessup, Marfan Trustee Dr Nitha Naqvi, Marfan Trustee Ed Stilliard, Helpline & Communications Manager Victoria Hilton

Poised on the cusp of Christmas, we have hurtled to the end of a busy and eventful year while looking forward to a new one filled with patient information days, research breakthroughs, inspiring projects, exciting webinars, new collaborations and an ever-busier helpline.

Though taken only three months ago, this sun-kissed photograph above already feels like a distant memory. It was snapped at Austin Court during our Birmingham Patient Information Day where medical experts shared their knowledge, supporters mingled, children enjoyed the playroom and new friendships were forged.

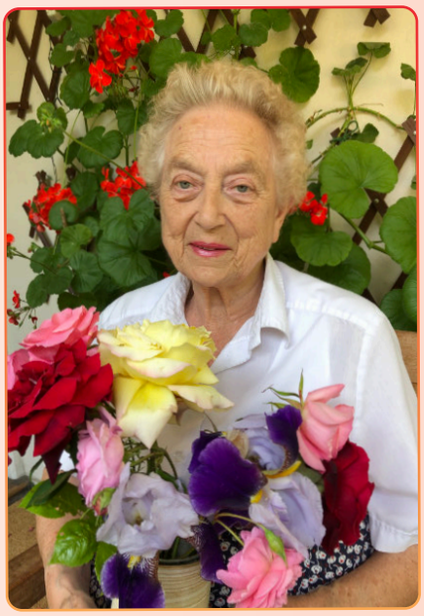
Save the Date: We are resuming our collaboration with the Marfan Foundation and will be hosting another Patient Symposium in Bloomsbury (London) on Saturday, 10 October. Details and a registration page will be released soon. In the meantime, we are also organising a separate patient conference in Glasgow, while continuing our staple remote webinar in spring.

Each page filled with newsworthy news, our festive edition of Marfan Matters is a must-read!

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



Hello, Old Friends, and New.

Festive Greetings! We have somehow reached the end of another eventful year, during which much progress has been made.

As I write this, responses to our nationwide survey are being analysed, and our report will be published very soon. We are incredibly grateful to those who completed the questionnaire; its results will underpin our charity's three-year strategy.

Autumn was very much the season of conferences, opening with our Patient Information Day in Birmingham, Living Well with Marfan & Loeys-Dietz Syndromes. I was sad not to be able to join you all but heard so many good reports of how well it went. The venue Austin Court was conducive to a friendly intimate symposium and the weather cooperated. We are very grateful to Marie O'Donnell and her colleagues from Birmingham Women's and Children's Hospital for collaborating with us on such an important event and making it so special.

Our remote Patient Information Day took place in October and opened with Consultant Thoracic Surgeon Mr Joel Dunning and his barnstorming presentation on Treatment for Chest Abnormalities. I spoke later on during the conference and unveiled the preliminary results of my questionnaire on fatigue. Determined to discover more about fatigue, its intertwined relationship with pain, and its impact in Marfan and Loeys-Dietz syndromes, I distributed a questionnaire over the summer with the assistance of medical student Kannan Maheswaran. Thank you to those who completed it. The full results will be reported soon. In the meantime, here is the link to the videos of our remote conference in October:**https://www.marfantrust.org/articles/marfan-trust-information-day-videos**

Dr José Aragon-Martin's research programme is advancing beautifully as he continues his exploration into the exact genetic causes of Neonatal Marfan syndrome, and new genes causing Hereditary Thoracic Aortic Aneurysm and Dissection, ectopia lentis and glaucoma.

We are expecting to see publication of scientific papers in the New Year from our work.

It's been such a productive year and none of it would have been possible without you and your wonderful fundraising and support! We celebrate a collection of fundraising feats on page 13 of this newsletter.

With best wishes for the festive season,

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

Helpline .... a Lifeline

by Victoria Hilton

A point of clarity in a sometimes disjointed medical landscape, our helpline continues to offer reassurance and guidance to a wide variety of callers, from supporters seeking advice on managing the manifestations of their condition, to patients in search of a particular specialist, to those whose signs and symptoms lead them to suspect they may have Marfan or Loeys-Dietz syndrome. Of the 530 queries taken this year, around 40% of queries fit this latter category. In this instance we offer advice, a free nurse specialist online appointment, and signpost accordingly.

At least a quarter of queries are musculoskeletal in nature and the management of associated pain. Recently held webinars on this subject confirm that anyone with a connective tissue disorder should carefully deliberate before submitting to surgery for chronic joint pain. Specific medications and joint supports should be tried first. An operation may not provide relief, and any joint stabilisation surgery neither reliably resolves the pain nor avoids a very high risk of failure.

Questions to the hotline can sometimes be driven more by curiosity than by immediate concern. One supporter, initially suspected of having diverticulitis, instead learned she had enlarged pelvic veins, a condition known as pelvic venous congestion syndrome, like varicose veins. She wondered whether this finding could be related to Marfan syndrome. Indeed it is! Enlarged veins are a feature of Marfan and Loeys-Dietz syndromes.

Remaining questions concern several areas, most notably the heart. Some parents question the idea of medication for children who show no signs (as yet) of aortic dilatation. This is, unsurprisingly, a knotty issue. Many cardiologists recommend starting treatment immediately rather than waiting for abnormalities to occur. As our paediatric consultant adviser, Dr Nitha Naqvi, puts it: “Do you wait for your washing machine to break and then fix it, or do you care for it, descale it and service it?”

Other concerns emerge around headaches. Patients with Marfan syndrome are more vulnerable to migraines, which can sometimes have a postural cause when the patient has scoliosis or kyphosis, for example.

Ocular manifestations affect many of our supporters, and several have questions about the benefits and long-term outcomes of intraocular lens implants. This remains a new and cutting-edge area, and we invited Mr Imran Masood to speak on this topic at our recent Information Day. **https://www.youtube.com/watch?v=iaqHpQW8dbI&t=7s**

Here are some infrequently asked questions:

Do identical twins always have exactly the same DNA, including in the Marfan (FBN1) gene?

We believe identical twins emerge from the same fertilised egg at conception, so they inherit the same DNA sequence. Small genetic and epigenetic differences can emerge over time. Even so, because differences are still possible if only small, it is wise to test each twin's DNA individually to confirm they both carry the same FBN1 variant, i.e. gene change/mutation.

I have been diagnosed with a Variant of Unknown Significance. What does this mean for my family?

A genetic test does not always yield results. You can receive a negative test in which no variant is identified, or you can have a 'Variant of Uncertain Significance' (VUS). Put simply, this means that a genetic change is identified but there may be insufficient information to determine whether or not is it causing the symptoms. In this case, a careful family history needs to be taken and further information about other members of the family collected (e.g. dilated aorta/aortic dissection etc) which will determine whether the family will be advised to undergo monitoring and follow up. Even if no genetic change is identified, it may still be necessary for families to be monitored due to their family history. The family may share a gene change that simply hasn't been identified yet, and this is why continuous research into these conditions is so important.

I am shortly to undergo my 8th or 9th CT scan in just 18 months. I am newly diagnosed with Loeys-Dietz Syndrome, have undergone valve-sparing root surgery and now have inflammation around the site. I understand they are scanning my thymus gland for the source of the inflammation. Are there any important side-effects of serial CT scans that I should be aware of. I am a 30-year-old male with two lovely children.

A CT scan is indispensable for assessing the aorta when urgent or very detailed imaging is needed. It can evaluate dissections or rapid diameter change and provides a clear view when MRI isn't possible for reasons of pacemakers, claustrophobia, poor-quality MRI images. A CT angiogram of the chest typically exposes you to about 5–8 mSv, equivalent to a few years of natural background radiation. I understand your concern about the accumulative risk of exposure to radiation but when scanning your thymus gland it will target the soft tissue not the bone. It is also a long way from the reproductive organs if this is a worry regarding fertility.

Specialist Nurse Clinic on Friday Afternoons

Running every other Friday afternoon, these structured appointments allow a 20-minute conversation and "consultation" with nurse specialist Joanne Jessup where your 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. **https://www.marfantrust.org/pages/83-events**

Parents' Corner

Speaking with people who are similarly situated can be both constructive and comforting. Our Parents' Corner offers an opportunity to chat freely with other parents of children with Marfan or Loeys–Dietz syndrome and discuss common concerns in a warm, friendly (Zoom-based) environment. These low-key gatherings occur on the third Tuesday of every month at 7pm. Join the conversation! **https://www.marfantrust.org/pages/83-events**

Stories from the Helpline

When Dmytro fled the war in Ukraine for refuge in Britain, he turned to the Marfan Trust for help navigating government bureaucracy. Living with Marfan syndrome and a dilating aorta, he later sought our assistance in continuing his medical care and scans once he had settled here, with fantastic support from the local council. We also arranged peer support, ensuring that Dmytro never felt alone.

Dmytro has since returned to Ukraine, where he met the love of his life, and they are to marry later this year. He now has six-monthly scans for his aorta and feels reassured that he is being monitored properly. Despite enduring daily disruptions, including power cuts lasting 12 to 16 hours, he remains hopeful that peace will come soon.

We are here to help you so please do not hesitate to call us on 0333 011 5256, or email info@marfantrust.org

by Dr Jose Aragon-Martin PhD, Sonalee Laboratory Research Director

As gene editing increasingly moves from the research laboratory to clinical practice, where it is radically enhancing lives, we take stock of its promise for people with Marfan syndrome. One of the most significant advances in the field has been the clinical use of gene editing for single-gene disorders, inherited in a similar way to Marfan syndrome.

Medical science is advancing rapidly, and many patients hear exciting headlines about gene editing and "living drugs" in the news. You may have seen stories about new therapies for leukaemia and wondered if they could help Marfan syndrome. Here's what you need to know in simple terms.

1. CAR-T Therapy: A Revolution for Blood Cancers

CAR-T therapy is a new kind of treatment that has made headlines for curing certain blood cancers like leukaemia and lymphoma.

- Scientists take immune cells (called T cells) from a patient or a donor.
- They genetically engineer these cells to hunt down cancer cells.
- The modified cells are put back into the patient's body, where they attack the cancer.
- Some of these therapies, like the BE-CAR7 therapy, even use gene editing (base editing) to make the T cells safer and more effective.

Important for Marfan patients:

- CAR-T therapy does not fix genetic conditions like Marfan syndrome.
- It is designed specifically to target cancer cells, not structural proteins or connective tissues.

2. Prime Editing: Fixing the Genetic Code

Prime editing is a newer technology that works like a "search-and-replace" tool for DNA.

- Scientists can use it to correct specific genetic mutations.
- Unlike older gene-editing tools, prime editing is very precise and causes fewer unwanted changes.
- For Marfan syndrome, which is caused by changes in the FBN1 gene, prime editing could theoretically fix the faulty gene at its source but is still in early research stages.

Challenges today:

- **Marfan syndrome** affects many parts of the body (heart, aorta, eyes, skeleton). Reaching every affected tissue with a new therapy is extremely difficult
- Delivery of prime editing safely to the right cells is still experimental.
- Currently, prime editing is not available as a treatment for Marfan syndrome — it's being studied in laboratories.
- 3. What This Means for You
- CAR-T therapy: Exciting and life-saving for some cancers, not suitable for Marfan syndrome.
- Prime editing: Holds promise as a potential future therapy for genetic conditions like Marfan syndrome, but it is still in early research.
- Researchers are learning more every year, and gene-editing technologies may eventually offer new ways to treat or even prevent Marfan syndrome at its genetic root.

3. What This Means for You

- CAR-T therapy: Exciting and life-saving for some cancers, not suitable for Marfan syndrome.
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Gene Editing Tool	How It Works (Simple Explanation)	What It Can Do	Limitations / Notes
CRISPR-Cas9	A molecular “scissors” that can cut DNA at a specific spot.	Can remove, insert, or change a gene. Used in research and some experimental therapies	Can sometimes cut the wrong spot (“off-target” effects). Delivery to all affected cells is challenging
Base Editing	A “letter-by-letter” editor that changes a single DNA base (A, T, C, or G) without cutting the DNA	Can fix small point mutations that cause disease.	Only works for certain types of mutations. Still experimental in humans.
Prime Editing	A “search-and-replace” tool that can rewrite small sections of DNA very precisely	Can correct more types of mutations than base editing, including insertions or deletions.	Delivery to the right tissues is still experimental. Not yet used as a treatment for genetic diseases like Marfan.
Zinc Finger Nucleases (ZFNs)	Custom-made proteins that cut DNA at specific sites.	Can remove or fix genes in research settings.	Complex to design and less flexible than CRISPR.
TALENs	Similar to ZFNs but easier to program to target specific DNA sequences.	Can edit genes in cells for research and experimental therapies.	

Takeaway for Patients:

Gene-editing tools are like precision tools for DNA. Some act like scissors (CRISPR-Cas9), others like a pencil that can correct a single letter (base editing) or rewrite a small section (prime editing).Right now, these tools are being developed in research and experimental stages. For Marfan syndrome, they are not yet available as treatments, but we are studying how they might fix the genetic cause in the future.

Bottom Line:

If you have Marfan syndrome, the current recommended management remains:

- Regular cardiac monitoring (heart and aorta).
- Medications to reduce stress on the aorta.
- surgical interventions when needed (proven safe and effective).
- Lifestyle adjustments and ongoing care with a Marfan specialist.

Photograph features from l to r: Dr Jose Aragon-Martin; PhD students Archana and Chandana at the American Society of Human Genetics in Boston, USA

Gene editing is exciting, but for now, it is not a treatment option, though it may become a reality in the future.

The Sonalee Laboratory: Advancing Genetics Research by Dr José Aragon-Martin

The Sonalee Laboratory continues to explore the genetic basis of connective tissue and ocular (eye) disorders.

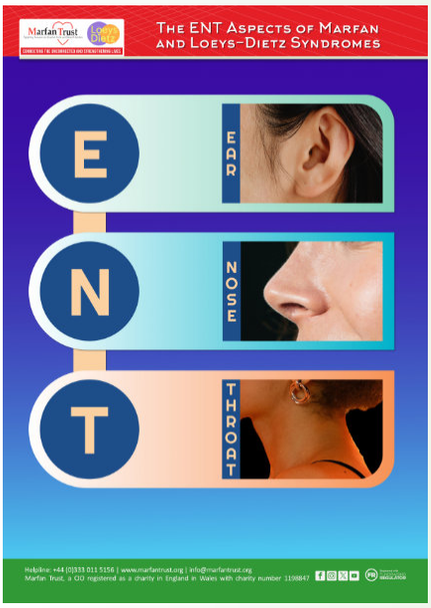
We are investigating the genetic changes that cause neonatal Marfan syndrome using advanced approaches. These include 3D protein modelling, which helps us understand how specific mutations affect protein structure and function, and whole-genome sequencing, which examines the entire genetic code of a patient. By incorporating long-read sequencing, we can read longer stretches of DNA, capturing complex regions and structural changes that traditional short-read methods might miss, giving a more complete picture of the genome. Using computational analyses, we explore how specific genetic variations relate to the traits and symptoms seen in patients and identify additional genes that may influence disease severity.

In parallel, we are studying glaucoma samples to pinpoint genetic variants that may contribute to the condition. Caused by abnormal pressure within the eye, glaucoma is known as "the thief of sight". It affects 5% of Marfan syndrome patients. This work is being done in collaboration with the Institute of Ophthalmology and Moorfields Eye Hospital with the goal of uncovering new insights into the underlying biology and potential targets for intervention. Overall, this research aims to deepen our understanding of these diseases, improve diagnosis, and support more personalised care for patients.

For the first time, the Daniel Finkletaub Memorial Summer Studentship has been awarded twice to the same student, Jessica Alvarez who wishes to continue in the Sonalee Laboratory to complete a PhD. Jessica has been exploring the genetic variants of neonatal Marfan syndrome and the severity of the disease. This year she will be studying gene changes behind familial thoracic aortic aneurysm and dissection.

Pictured l - r: A-Level student Gemma was given a tour of the lab by PhD student Jessica Alvarez Salgado

New Information Guides!



Hidden away yet widely experienced, the gastrointestinal manifestations of Marfan and Loeys-Dietz syndromes have long been under-explored. But no more. Our new, free guide is now available to download. It discusses the functional gastrointestinal disorders including IBS and constipation while also touching on rarer complications sometimes associated with connective tissue disorders such as small bowel diverticulosis. Other new leaflets including the **Dental Manifestations of Marfan & Loeys-Dietz Syndromes**, and the **ENT Aspects of Marfan & Loeys-Dietz Syndromes**. Download your free copies today: **https://www.marfantrust.org/resources/information-guides-**

The UK's Pioneering Role in Aortic Root Surgery for Marfan Syndrome

by Darren McDean

Marfan syndrome is a complex genetic disorder affecting the body's connective tissue, and its most life-threatening complication is the progressive weakening and enlargement of the aorta, particularly at the aortic root. Without intervention, this can lead to a catastrophic tear, or dissection, of the aorta, often resulting in sudden death. The history of surgical treatment for this condition is a story of a dramatic shift from high-risk, reactive procedures to proactive, life-extending and valve-preserving operations, with the UK playing a significant role in this evolution.

Early Innovations: The Bentall Procedure

Professor Hugh Bentall

The foundation of modern aortic root surgery was laid in the late 1960s with the development of the "Bentall procedure" by UK surgeons Professor Hugh Bentall and Mr Antony De Bono. This groundbreaking operation addressed both the aneurysmal aortic root and the associated valve problems in one go. The procedure involves replacing the dilated aortic root with a composite graft - a synthetic tube with a pre-sewn mechanical heart valve. The coronary arteries are then re-implanted into the new graft.

For decades, the Bentall procedure was the gold standard for patients with Marfan syndrome and a dilated aortic root. It significantly improved life expectancy and was a major step forward from the grim prognosis of the pre-surgical era. However, a major drawback was the lifelong need for anticoagulation therapy (Warfarin) to prevent blood clots from forming on the mechanical valve, which carried its own risks and lifestyle limitations.

Sir Magdi Yacoub

The Move to Valve Preservation

As surgical techniques and understanding of the disease evolved, surgeons began to question the necessity of replacing a patient's own, often structurally sound, aortic valve. This led to the development of "valve-sparing" procedures.

The two main valve-sparing techniques are the "David procedure" (reimplantation) and the "Yacoub procedure" (remodeling), both named after their pioneering surgeons, Professor Tirone David (Canadian Cardiac Surgeon) and Sir Magdi Yacoub, a prominent UK-based cardiac surgeon. These operations replace the diseased aortic wall with a synthetic graft while preserving and re-suspending the patient's native aortic valve inside the new graft. This avoids the need for a prosthetic valve and, critically, the requirement for lifelong anticoagulation (Warfarin).

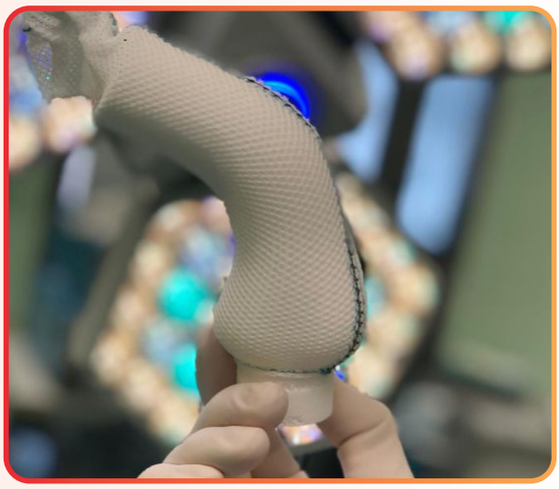
Professor Tirone David

Valve-sparing root replacement has become a preferred option, particularly for younger patients with Marfan syndrome, as it allows for a better quality of life and avoids the long-term implications associated with blood-thinning medication. The UK has been at the forefront of adopting and refining these techniques, with major cardiac centres becoming international referral points for these complex procedures.

A UK-Led Patient-Driven Revolution: The PEARS Procedure


Professor John Pepper & Tal Golesworthy

A truly unique and patient-led innovation in the UK is the Personalised External Aortic Root Support (PEARS) procedure. This story is an inspiring example of collaboration between a patient and a surgeon. Engineer Tal Golesworthy, who has Marfan syndrome, worked with his surgeon, Professor John Pepper at the Royal Brompton Hospital in London, to develop a new approach.

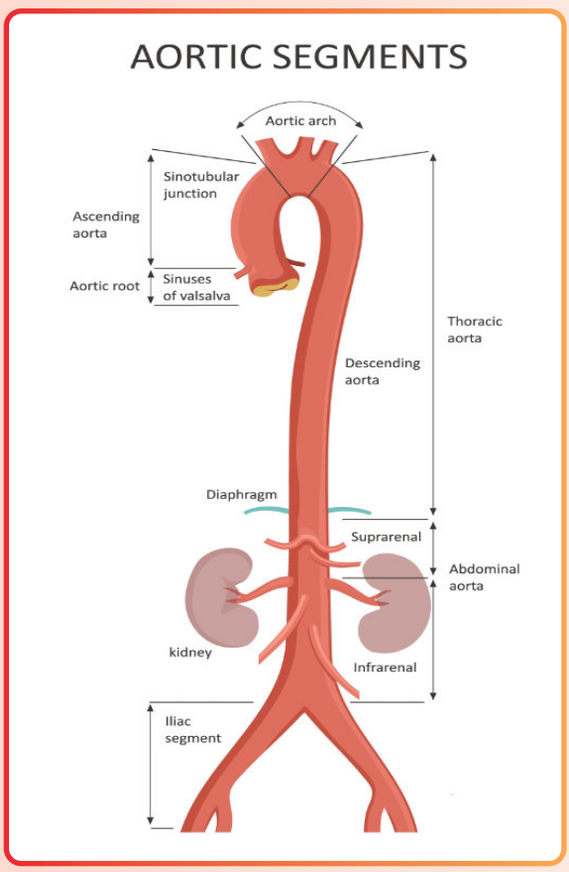


Recognizing that the problem was the weakening of the aortic wall itself, Mr Golesworthy used his engineering expertise to design a custom-made, 3D-printed mesh sleeve. This sleeve is placed around the outside of the existing aortic root, acting as a scaffold to prevent further dilation.The PEARS procedure is a less invasive and less complex option than full root replacement. It does not involve any cutting or stitching of the aorta itself, and it preserves the native aortic tissue and valve completely. It is particularly suitable for patients with a smaller degree of dilation and no aortic valve issues. First performed in 2004 on Mr Golesworthy himself, the PEARS procedure highlights the UK's commitment to pioneering new, less invasive solutions, driven by a patient-centred approach.

The Future of Aortic Surgery in the UK

Today, the UK remains a leader in the surgical management of Marfan syndrome, offering a full spectrum of treatments. Clinical practice is guided by clear criteria for intervention, with surgery typically considered when the aortic root reaches a certain size (often around 4.5-5 cm for Marfan patients, a far lower threshold than for the general population).

The UK’s approach emphasizes a multidisciplinary team model, with cardiologists, geneticists, and cardiac surgeons working together to provide comprehensive care. This collaborative framework ensures that patients are monitored closely and receive the most appropriate and timely intervention, whether it be a Bentall procedure, a valve-sparing operation, or the PEARS procedure. The history of aortic root surgery for Marfan syndrome in the UK is a testament to constant innovation, a commitment to improving patient outcomes, and a willingness to embrace novel, patient-driven solutions.



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Yacoub MH, Ahmed A, Hosny H, et al. A New Technique for Shaping the Aortic Sinuses and Conserving Dynamism in the Remodeling **https://www.sciencedirect.com/science/article/abs/pii/S0003497520320294**

https://www.ctsnet.org/article/yacoub-ii-valve-conserving-operation-aortic-root-remodeling

Connective tissue disorder and high-risk pregnancy: a case series with personalised external aortic root support (PEARS)

As PEARS is increasingly performed, research is steadily accumulating, and the most recent relates to pregnancy. Our Trustee, Consultant Cardiologist Professor Christoph Nienaber, is amongst the authors of a newly published article in Nature Magazine, and our synopsis is below.

This case series reports nine successful pregnancies in women with Marfan or Loeys–Dietz syndromes who had previously undergone the PEARS aortic root support procedure. No aortic dissections or maternal deaths were observed.

Aortic dimensions remained stable throughout pregnancy and postpartum, and no hypertensive pregnancy disorders occurred. Both vaginal delivery and Caesarean section were safely undertaken.

Although small, the study suggests PEARS may be a promising pre-conception option for selected women with aortopathy. Here is a link to Professor Christoph Nienaber's full article. **https://bit.ly/3KuSGwC**

Newly Published Papers on Blood Pressure

A reliable measure of health and happiness, your blood pressure matters! Like water travelling through a garden hose, your blood flows around your arteries, exerting force against the walls. When the pressure exerted is too high, it can be dangerous. But how high is too high? Our advisor, Professor Christoph Nienaber, Royal Brompton Hospital reports on present opinions:

Blood Pressure, Hypertension, and the Risk of Aortic Dissection Incidence and Mortality: Results From the J-SCH Study, the UK Biobank Study, and a Meta-Analysis of Cohort Studies

An inextricable link between hypertension and aortic dissection is borne out through a new analysis of Japanese and UK data. People with hypertension face over a threefold higher risk of aortic dissection. Importantly, the risk does not rise only once hypertension is diagnosed but increases steadily as systolic and diastolic pressure soar, becoming significant above 132/75 mmHg. These findings signal that even so-called "normal" values may mask meaningful risk. Earlier, more pre-emptive blood pressure control could prevent catastrophic aortic events. Furthermore, a healthy diet, exercise, weight control, no smoking, low salt and/or medication may be needed earlier than traditionally thought. For fuller details, click here to read the full article.

Taming Hypertension to Prevent Aortic Dissection: Universal Recognition of a "New Normal" Blood Pressure?

The notion of normal is inherently subjective. What is normal?

A newly published editorial by Professor Christoph Nienaber argues that new evidence calls for rethinking what constitutes "normal" blood pressure, because risk rises even at the upper end of the accepted range. The authors emphasise that keeping pressure as low as is safely possible may substantially reduce the likelihood of dissection, and that this principle appears consistent across populations and is supported by major trials. They call for a shift in practice towards more stringent, lifelong management of blood pressure. In the meantime, healthy everyday habits that lower blood pressure such as exercising, eating well, reducing salt, managing weight, and not smoking unequivocally help to reduce the risk of aortic dissection. Click here to read the full article: **https://bit.ly/49ZmE6k**

We remember those we lost in 2025 …

Suzanne Brown, by her daughter Joanne

In Sue's memory, instead of flowers we asked for donations to the Marfan Trust. Sue suffered from Marfan syndrome along with some of her family members. It meant that she became frail before her time and despite essential surgery her heart just wasn't strong enough to cope with everyday life. Along with other complications with her health, she and Richard, who has devotedly cared for her, were unable to spend their retirement as they'd dreamt. She has supported the Marfan Association since before she was diagnosed and often knitted items to raise funds. It is a little-known syndrome and as such the charity receives very little funding. We raised £2532.50 in memory of my wonderful mother.

Sue is much loved and will always be remembered.

Maureen "Mosey" Walker a Eulogy

Mosey was born in Westerham on 5 January 1945, just as the Second World War was drawing to a close. Her mum, Annie and dad Cyril had three older sons, Ted, Mike and Tim. Mosey was their very much loved but much younger daughter as Tim was 15 when she was born. Mike and Ted were in the RAF at the time, fighting for their country.

Mosey's first job worked at Friary Meux in Westerham. There she met her future husband Chris Walker, and they married. She and Chris went on to have two children, Stuart and then Beverley. They parted amicably.

Mosey first met her second husband and love her of life Colin when he installed her Aga and initial their friendship grew into something more, and Colin moved in with Mosey. They then shared the next 36 years together, enjoying the same interests, cooking, gardening and going to National Trust Places and the RHS. They also enjoyed wonderful holidays together. Colin had two children, Natasha and Jason, and Mosey welcomed them in on their weekends and they naturally loved her delicious cooking. Colin and Mosey were soulmates, best friends and loved each other with all their hearts.

As a Grandma Mosey loved it when the grandchildren visited and they would look forward to endless amounts of food which came from the kitchen. Mosey was an amazing cook, cooking Christmas Lunch for all her family.

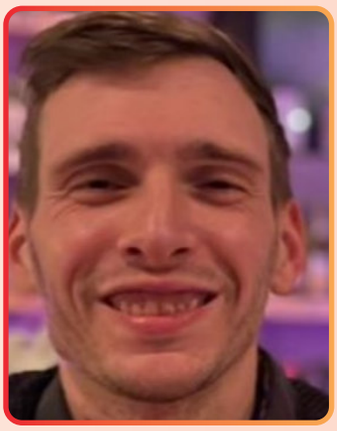
Unfortunately, Mosey had Marfan Syndrome and was a long-standing member of the Marfan Association. This came with a lot of medical issues, and as she got older it began to take a toll on her, with her scoliosis causing mobility problems. In 2018 she was additionally diagnosed with Parkinsons. She had to endure a lot throughout her life, but she was strong willed, and she carried on with life as best as she could. Over time, Colin became her full-time carer, and did so with an open heart and willingness to care for his beloved Mosey. She in turn knew that he was there for her in every way, and their love for each other was as strong as it always was, and always will be.

It was heartbreaking to see Mosey suffer, but she received so much love and care from Colin and Beverley and the family. They would like to thank all of the nurses and doctors and staff at Pondtail Surgery, the first community health and care team and her neurologist Dr Kimber, who went above and beyond to help.

Mosey sadly passed on Sunday 3rd August. Losing her has been dreadful, and a shock for all the family. She was so loved and is greatly missed.

Mosey's family and friends raised nearly £900 in her memory with the donations to be divided between us and Parkinsons UK.

Josh England 1995 - 2025



A tribute to my beautiful boy, by Margaret England

Josh was born on January 4th 1995, a beautiful, healthy baby.

After a happy healthy childhood, Josh grew into a very tall gangly teenager. We joked about his height as I'm only 5ft. A few illnesses included excessive headaches leaving his eyesight very poor were put down to a mild case of meningitis. His painful joints were put down to growing pains! But he grew into a healthy young man, completing a degree in Business Management.

Josh loved to play football and trained our local football team under 11s. The kids loved him and his dry sense of humour. But Josh loved to travel and set off to see Thailand and surrounding countries: yet he always returned home. He moved to the outer Hebrides living on the Isle of Harris and loved every minute of it there. He came home in November 2024 after developing a bad pain in his back. This was the start of our nightmare.

After being admitted to hospital as he couldn't breath, Josh was diagnosised with kidney infection and then pneumonia! He was moved to intensive care where ONE doctor recognised it as Marfan syndrome. We had never heard of it, nor had his two siblings.

After an aorta replacement, he had a stent fitted in an elephant trunk (artery). Then he had an aneurysm at the end of the artery so he then had a Candy Plug fitted, only the second time this operation had been done in the UK. He was still very poorly; his heart was enlarged and he needed a heart transplant but sadly all his arteries had been worn out and a transplant was impossible. He was given three weeks to live!!

In those three weeks we had fun in the hospital as he had friends arriving from all corners of the world. We had food parties as his appetite was still good. The staff were brilliant but into our third week he was sleeping more and more. Finally on Monday, May 26th 2025 at 5.26pm he slipped quietly away. Our six months of hell had come to an end and my beautiful boy had died at the young age of 30 years old with his whole life that should have been before him.

I have put on his resting place

"no greater pain have I felt than when your heart stopped beating and mine carried on"

Thank you for this opportunity to pay tribute to Josh I really appreciate it.

I hope for future sufferers that their Marfan syndrome can be recognised earlier and symptoms can be picked up sooner.

Thank you most sincerely

Fundraising Feats

Without your fantastic feats of fundraising we would not be able to continue our work, running the helpline and conducting cutting-edge research. Here is a small selection of the wonderful campaigns over the last six months.

Stuart Sadler

Defying doctors' bleak predictions, Stuart Sadler is alive, thriving and took part in Run Norwich 2025! Diagnosed at 22 with Marfan syndrome, alongside his mother (it's a family affair), Stu was given a short life expectancy, warned not to have children and never to run again. Now aged 52 he is a proud father of two with a long life ahead, and has endured yet another operation. He raised £2,228 for our Trust in his recent run and here is his story: **https://bit.ly/3I4YpIf**

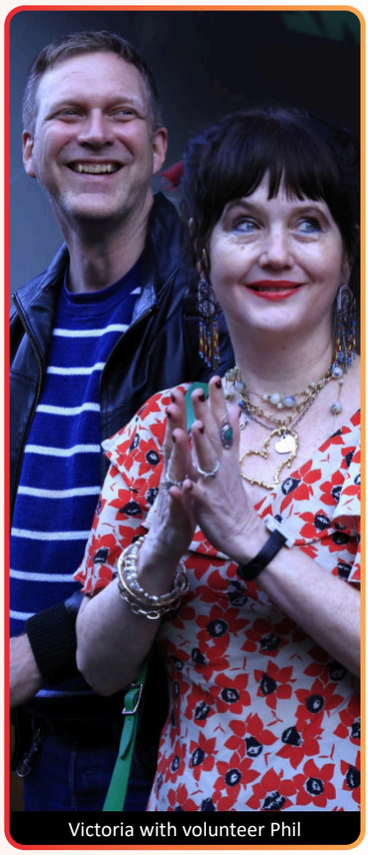
Fundraising Feats

Cannonbawz Run

Kris with his son Lucas

A convoy of elite cars and costumed characters took over Elgin High Street on Saturday 30 August for Kris O'Neill's Cannonbawz Run. Now in its eleventh year, this fundraising phenomenon has steadfastly supported the Marfan Trust, raising both awareness and vital funds.

We stood amongst the crowd as engines roared, heralding the departure of Kris and his crew, as they set off on their rugged road trip along Scotland's scenic NE50. Kris' brother Liam tragically died in 2021 of complications following an operation. He was just six months old when his subluxed lenses alerted a doctor to the possibility of Marfan syndrome. Kris has since regularly raised at least £6,000 a year for our charity. Here is the Cannonbawz Story. **https://bit.ly/3I37zoy**



Victoria with volunteer Phil

David's Challenge

A challenge can be a celebration, and David embraced it as such. He marked his return to health by taking on the Reigate Triathlon in September, raising money for both the Surrey & Sussex Health Charity and the Marfan Trust.

by David

Over the last 12 years, I have suffered three bouts of pericarditis, which is an inflammation of the pericardium, the sac which surrounds the heart. Each time, this has reduced my fitness levels to the bare minimum, and I find it difficult to get through the day or walk up the stairs without being out of breath whilst experiencing some unpleasant chest pains and heartbeat symptoms. However, once the inflammation subsides, it has been swimming, cycling and running that has built my fitness levels back up to be able to play sport and take part in events again. I wish to support two charities close to my heart; the Surrey and Sussex Healthcare Charity (where the money will be sent to the cardiology dpt of East Surry Hospital), and the Marfan Trust for my nephew who has the condition.

David raised over £600 for our Trust!

Honouring Josh

Marking the memory of much-missed brother and friend on his birthday. On 4 January 2026, Josh England would have turned 31, but he died earlier this year following a late diagnosis of Marfan syndrome. To honour their beloved Josh, his brother and best friend will be completing the Three Peaks Challenge. Here is the link to their campaign **https://www.marfantrust.org/articles/in-memory-of-josh** Josh's story is featured above.

Fundraising Feats

Swimming the Bosphorus Straits


Kirsty and Chrissie as students together

In memory of her life-long friend who died in July 2025, Medical Oncologist and Head of Bristol Medical School, Professor Chrissie Thirlwell swam the Bosphorus strait and her story reached the news! (**https://www.bbc.co.uk/news/articles/cy4dwdqdxdpo**). On 24 August, Chrissie joined more than 2,400 competitors from across the globe, swimming 6.5 kilometres from Asia to Europe, battling strong currents to cross one of the world's busiest shipping lanes as part of the Bosphorus Cross-Continental Swim.

Chrissie thought about her remarkable friend Kirsty every stroke of the way. “Kirsty lived a wonderful life and wasn’t at all defined by Marfan,” says Chrissie. “I’m doing this in her memory with her family’s blessing. The fact she would have said I was crazy makes me want to do it even more!”

Kirsty is much missed by her partner Steve, sister Lindsey and many, many friends.

Chrissie raised over £6,000 for the Marfan Trust whilst a further £1,200 was gathered at her funeral.

Conference Titbits

On the sunniest Saturday in September, over 100 patients, 16 children and 13 medical experts gathered at the lovely, light-filled venue Austin Court in Birmingham for our Marfan and Loeys-Dietz Patient Information Day. Living with Marfan and Loeys-Dietz Syndromes was a happy collaboration with Birmingham Women’s and Children’s Hospital, and several wonderful specialists from the hospital were there to deliver presentations and take questions from the audience.

Thoracic Surgeon Mr Ian Hunt whizzed up from St George’s Hospital in London during his on-call weekend to open the conference. Amongst many other things, he discussed the restoration of NHS England funding for severe pectus excavatum, the importance of physiotherapy in the treatment of pectus deformities, and how early diagnosis can prompt earlier and less invasive treatment.

Geneticist Dr Osio then spoke on the diagnosis of connective tissue disorders, followed by Dr Hanadi Kazkaz on musculoskeletal aspects, Drs Chikermane and Clift on cardiology, and Dr Duale on the psychological aspects of living with a rare condition. The afternoon was more relaxed, with break-out sessions and many conversations. With much gratitude to Genetic Counsellor Marie O’Donnell and volunteers from Birmingham Women’s and Children’s Hospital for making it such a special day.

Scenes from the Symposium

Lottie and Georgie met at last year’s Patient Symposium in London and became instant friends. They both have Loeys-Dietz syndrome and are treated at the same hospital, even taking the same medication. Here they are in Birmingham!

Romain Alderweireldt and Ludivine Verboogen (pictured second from left and far right) met their inspiration, Tal Golesworthy during the event. Romain and Ludivine’s son has Marfan syndrome and this prompted their 101 Genomes which researches protective modifier genes that counter the cardiovascular implications of MFS. They were maverick spirit of PEARS pioneer, Tal Golesworthy. Here is the link to their website: **https://www.101gems.be/site** Completing the picture is Consultant Rheumatologist and indispensable advisor to the Marfan Trust, Dr Hanadi Kazkaz.

Thank you to Mary Boag from Terumo Aortic for her wonderful photography!

Remote Marfan and Loeys–Dietz Information Day: Saturday 11 October

Through the lens of expertise, rare disorders Marfan and Loeys-Dietz syndromes, came into focus. Our remote all-day conference, featured a wide array of specialist speakers covering the many manifestations of these conditions. With a tantalising glimpse into the future and a lens on today's treatments and imaging, Professor Christoph Nienaber explored the evolving management of the aorta, the body's 27th organ. From open to non-invasive surgeries and the emerging role of stress mapping, you can discover how innovation is reshaping our increasingly personalised care. . From practical tips on managing fatigue to the latest clinical updates on treatment for the eyes, aorta, chest, and more, this event is a one-stop resource for patients and families alike. Here are the videos from the day **https://www.marfantrust.org/articles/marfan-trust-information-day-videos**

Scottish Adult Congenital Cardiac Services Annual Conference

Rare diseases united in Glasgow at the Scottish Adult Congenital Cardiac Service annual conference on Friday, 28 November! The Marfan Trust stood proudly alongside the Turner Syndrome Support Society and Max Appeal at an event designed for healthcare professionals. We each manned our own table displaying our charity's pamphlets and paraphernalia.

Victoria with Arlene, founder and chief executive of Turner Syndrome Support Society

During the tea and lunch break we took questions from paramedics, nurses, cardiologists, medical students and surgeons about our Trust and what we offer. The medics were quite taken with our emergency cards and left with several of these, along with our tote bags and leaflets.

The afternoon sessions focused on aortopathies, with particular emphasis on the genetic conditions Turner, Marfan and Loeys–Dietz syndromes. Of interest, the threshold for aortic surgical intervention is much lower in Turner syndrome than in Marfan. The doctors then went on to discuss surveillance, the merits of indexing, aortic diameters and other aspects of surgical management. The cardiology team in Glasgow is working to expand its aortopathy service, and we will happily help them.

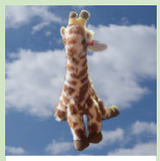
Aortic Dissection Awareness Day

The annual conference was another wonderful opportunity to meet with experts in the field of aortic dissection, from those with lived experience to those working on the frontline. This year, the theme was 'Think Aorta, Think Family'.

In response to this, an exciting piece of research is underway, led by the University of Leicester and supported by ADAUK. The DECIDE-TAD study is developing a Decision Support Tool (DST) that can help individuals and their families decide whether they want to undergo genetic testing, screening and monitoring if they are found to be at risk of Thoracic Aortic Disease (TAD), e.g. if they have a family member with TAD. During the afternoon we spent time in break out groups reviewing the tool and suggesting ways in which it might be developed to make it even better. It is great to see the research team using this group's experiences to build the best DST to support families with these complex decisions. My group taught me so much about some of the challenges they had faced.

As always, my favourite part of the day was meeting people and it was lovely to catch up with our wonderful supporter Rachel, pictured.

The Marfan Trust Online Shop


Willow

Tote Bag


Marfan Pen

Marfan Cap

Heart Keyring


Marfan Mug

Cycle Tops

Jeremy Wins

Water Bottle


Wristband

Pin Badge


Marfan T-shirt

Zest for Life

Marfan/LDS Band

Beanie

Green Tote


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 NEW	Small		Medium		Large		XLarge			£43.00 (each)		
Marfan/LDS Wristband NEW										£2.50		
LDS T-shirt NEW	Small		Medium		Large		XLarge			£11.00		
Marfan Trust T-shirt	Small		Medium		Large		XLarge			£11.00 (each)		
Willow, the Marf Giraffe										£9.00		
Jeremy Wins Book										£8.00		
Marfan Trust Tote Bag										£6.00		
Marfan Trust Pen										£2.00		
A Zest for Life Cookbook										£6.00		
Heart Keyring NEW										£3.00		
Marfan Mug										£9.00		
Marfan Trust Wristband										£2.50		
Marfan Trust Water Bottle NEW										£15.00		
Marfan Cap NEW										£10.00		
Marfan Trust Pin Badge										£2.00		
Watermill Card (10 cards)										£4.00		
Beanie Hat NEW					Black		Grey			£8.00		
Green Willow Tote Bag NEW										£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										£3.00		
SUM TOTAL (including postage)												