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

MARFAN MATTERS

Autumn/Winter '24 Edition

REGULAR FEATURES

SPECIAL FEATURES

KEEP IN CONTACT

www.marfantrust.org
+44 (0)333 011 5256
info@marfantrust.org
Find us on Facebook
Follow us on X
Follow us on Instagram
Watch us on YouTube

Firmly ensconced in the festive season at the time of writing, we have just emerged from behind our Christmas stall at the Royal Brompton Hospital. The first we have held for five years, our tempting table drew a steady stream of customers and started conversations about Marfan and Loeys-Dietz syndromes!


© photograph Simon Burgess

As the year rushes to a close, we reflect on the last few months. Heralded by our International Patient Symposium (right) on 31 August, Autumn was dominated by conferences. September marks Aortic Dissection Awareness Month and the body's largest artery was the subject of several fascinating events. A run of brilliantly educative seminars culminated in our Information Day on 12 October during which we heard from voices old and new.

Our (remote) Information Day also allowed the Marfan Trust's two summer medical students, Mehar Bijral and Navaneethan Adityaraj, to unveil the results of their research. AI met the Aorta in Mehar's project where he analysed over 500 CT scans to predict the measurement at which the aorta may dissect. Meanwhile, Navaneethan Adityaraj spent months investigating the eye in Marfan syndrome, producing a wonderfully comprehensive guide. See page 6 for a summary of their work.

From our international symposium, webinars, fundraising events and festive stall to our casual, low-key monthly drop-ins, we've really enjoyed meeting our supporters over the year and hope you enjoy this festive edition of *Marfan Matters*.

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

Hello, Old Friends and New.

We're finishing the year on a wonderfully festive note, with our Christmas Stall at the Brompton where we have met old friends and made new ones. Ostensibly fundraising, these events are just as important for awareness and we have introduced many passers-by to Marfan and Loeys-Dietz syndromes.

This has been a very good year for awareness, from the 2 million and counting who tuned in to my *Life Scientific* interview to the many events held by supporters around the country for our charity. We are celebrating your fundraising feats on our website and here is the path to the page.

Our research programme is flourishing, so much so we have acquired new equipment to allow Dr José Aragon-Martin and his team of MSc students to more easily identify new genes associated with Hereditary Thoracic Aortic Aneurysm and Dissection. We have a new NGS machine PromethION, Next Generation Sequencing. This is next generation technology that facilitates the sequencing of one entire genome in just one day and will vastly speed up Jose's research.

A definite highlight in the year was our International Patient Symposium. Collaborating with the Marfan Foundation, the Aortic Dissection Charitable Trust and Annabelle's Challenge on the event was a joy. Over 200 patients attended the day-long conference and our report is on pages 8 & 9.

We wish you a wondrous festive season and a fantastic New Year.

Anne H Child

Dr Anne Child MD FRCP

Helpline .... a Lifeline

by Victoria Hilton

In a medical landscape of sometimes disjointed care for Marfan and Loeys-Dietz patients, our helpline is a central source of advice and information. Many patients, especially the newly diagnosed, seek an overarching structure to their or their relative's treatment. They wonder how frequent their cardiac check-ups should be, and where they should turn to address the different symptoms these connective tissue disorders can bring. We aim to be the central point of contact, guiding accordingly.

Prominent amongst the 489 calls received this year are questions over the intervals introduced between cardiology scans. A few years after undergoing successful aortic root replacement surgery, a supporter was told that her echoes would move from being held annually to biannually. Naturally, she was worried. In response, we cited the latest American Heart Association guidelines which recommend that after surgery, a two-year interval can be introduced to monitor the aorta. However, these guidelines also recommend that an MRI or CT be used to check on the distal thoracic aorta, that is the part further away from the heart. This part of the aorta is sometimes more difficult to see with an echo scan alone.

Recommendations for Diagnostic and Surveillance Aortic Imaging in Marfan Syndrome (Continued)

COR	LOE	Recommendations
Imaging After Aortic Root Replacement		
1	C-LD	3. In patients with Marfan syndrome who have undergone aortic root replacement, surveillance imaging of the thoracic aorta by MRI (or CT) is recommended to evaluate for distal TAD, initially annually and then, if normal in diameter and unchanged after 2 years, every other year.[3-6]
2a	C-LD	4. In patients with Marfan syndrome who have undergone aortic root replacement, surveillance imaging every 3 to 5 years for potential AAA is reasonable.[2,6]

Of the nine queries taken every week, around a third are from people wondering if their symptoms signal Marfan syndrome. Many people display the external features of a connective tissue disorder - height, hypermobility etc - leading them to suspect that they may also have the internal, more serious symptoms. Whilst we cannot diagnose from afar, and depending upon the individual situation, we strongly suggest they seek a further review and request an echocardiogram.

Remaining calls to our helpline include questions of the eye which constitute around 15% of queries. Should a dislocated lens 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 it may not be necessary.

Fatigue and Pain

Fatigue and pain also loom large amongst helpline calls. The two are intertwined and very hard to manage. To address the latter, we secured a wonderful speaker for our recent Information Day. Dr Helen Cohen is a Consultant Rheumatologist at the Royal National Orthopaedic Hospital and gave a vivid, comprehensive presentation clarifying the anatomy of pain, its various mechanisms, and how to approach seeking help. Here is the path to the YouTube video. **https://www.marfantrust.org/articles/chronic-pain-and-marfan-syndrome**

Support

A barometer of the times, our helpline has recently seen a surge in requests for help with PIP (personal independence payment) applications as the cost of living tightens its grip. In response we can offer to provide letters to reinforce claims, and peer support in navigating the tricky forms.

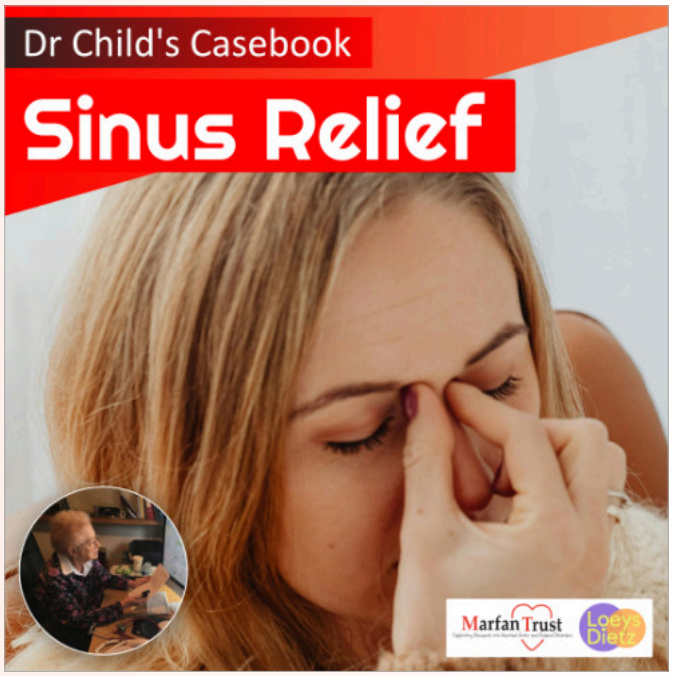
In-Person Support Groups

We are recreating the local support groups that once existed across the country. There is nothing quite like chatting in person to someone similarly situated with whom you can share common concerns and make MarFriends! Email **info@marfantrust.org** if you are interested in joining or leading a regional support bubble. The link to our support page is here: **https://www.marfantrust.org/pages/local-support-groups**

Over the year we have taken calls from the Republic of Ireland, Australia, Canada, Pakistan, South Africa and Norway in addition to the UK!

Join the Trust today and receive access to Dr Child's Casebook. Published below is a sample casebook to whet your appetite.

Casebook Special: Sinus Relief



Tiny but mighty, our sinuses constitute four paired empty spaces, filled with possibility. This network of paranasal cavities is instrumental in helping to modulate your voice, fend off infection, safeguard your skull, and more. But in Marfan syndrome, sinuses can be compromised. Which medication will help?

Q: I have been recently prescribed Fluticasone nasal spray for chronic sinusitis, which I believe is a steroid. What is the recommendation for topical steroid use on the nasal linings in Marfan Syndrome, with respect to tissue fragility/weakness?

A: Thank you so much for approaching us with the interesting question. Your sinuses are surprisingly important. They produce mucus that helps to lubricate your nasal passage and filter away dust and bacteria. They protect your skull and face in the event of a trauma such as a crash. They provide an immunological defence, and help to add warmth to your voice. Their roles are many!

In Marfan syndrome, the sinuses can be much narrower than normal and underdeveloped. The drainage passage is consequently narrower which means that bacteria are not filtered away efficiently, leading to infection.

In regard to your specific question of medication, I suggest that you use Fluticasone sparingly for short acute episodes and read the instructions.

Your sinusitis may need review of your prescribed medication. Is it a side-effect? Can the identified drug be substituted?

Can the steroid spray be substituted with another non-steroidal spray when the acute episode is brought under control, e.g. try Otrivin nasal spray?

However, these medications ALL put your blood pressure up. So, can you find the cause of the sinusitis?

Possible Sources of Inflammation:

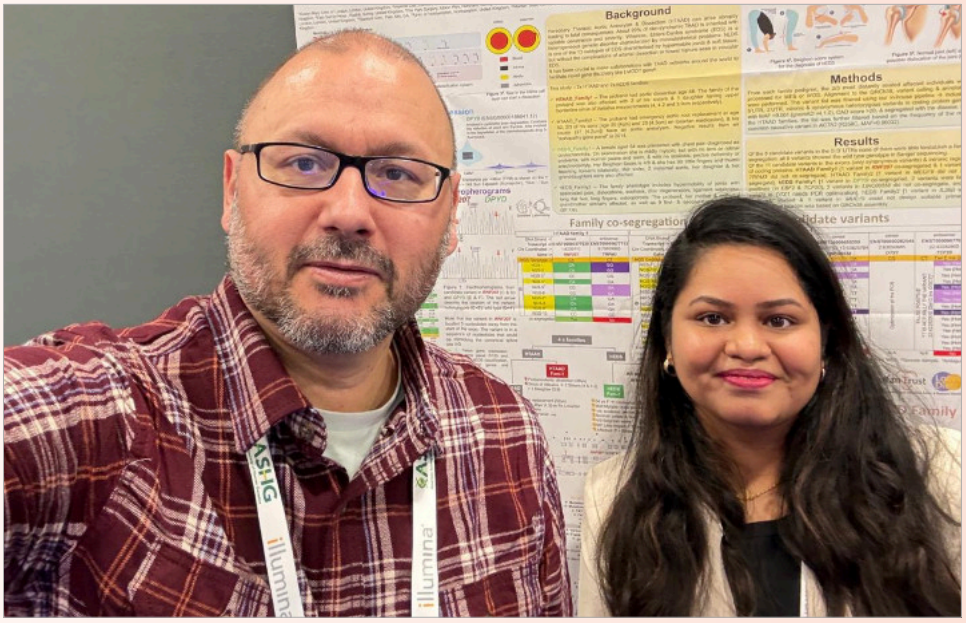
Allergy (dust; pet - if so buy a poodle or poodle-cross, or keep goldfish!)

See an ENT specialist:

Sometimes an X-ray shows small or obstructed sinuses; or indeed a deviated septum (the middle bone in the nose) which can be corrected.

Research Update

Our research programme continues apace, as Dr Aragon-Martin explores new causative genes behind hereditary thoracic aortic aneurysm and dissection (HTAAD), genotype-phenotype correlations in Marfan syndrome, dislocated lenses and more. Here is his update on activities in the Sonalee Laboratory.



by Dr José Aragon-Martin
I'm dividing my time between the William Harvey Research Institute at Queen Mary University of London, and the Institute of Ophthalmology.

William Harvey Research Institute

The Sonalee Laboratory maintains a valuable database of samples from UK families affected by Heritable Thoracic Aortic Aneurysms and Dissections (HTAAD). In several of these families, all the major known genes associated with HTAAD have been ruled out. Exploiting the latest technology, my MSc students and I are searching for novel genes that may play a role in the HTAAD pathway.

Genomic medicine in the UK is constantly evolving and the gene panel, known as R125, used to screen people suspected of having HTAAD (as well as Marfan and Loeys-Dietz syndromes) now tests for well over 30 genes. Our continued work at the William Harvey Research Institute aims to identify further genes that will contribute to this panel, potentially saving lives.

We have recently acquired new equipment that facilitates the sequencing of one entire genome in just one day and will vastly speed up our research. The PromethION P2 Solo allows scientists to sequence entire genomes or transcriptomes rapidly and at a lower cost. This is a fantastic coup for our lab and was made possible only through our supporters' fundraising. Thank you!

With Msc student Chandana Veshala, I unveiled my research at the American Society of Human Genetics symposium in Denver. We showcased the results of our Next Generation Sequencing (NGS) study into the gene changes potentially responsible for HTAAD and Hypermobile Ehlers-Danlos Syndrome (hEDS). We are pictured together, above.

Pictured from left to right:
Msc students: Roshanak, Chandana, Jeyabharathi, Srivarshini and Silvia.

Chandana is one of four MSc students who have been analysing NGS (Next Generation Sequencing) data on HTAAD and hEDS (hypermobile Ehlers-Danlos syndrome) to find genetic biomarkers that might be causing these conditions. Here is a summary of their activity.

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).

We continue to map gene changes causing the condition, onto the international gene map for Marfan syndrome. This helps us predict how a new mutation will affect a baby who has just been diagnosed.

Institute of Ophthalmology – University College London

Over the last academic year (2023-2024), we supervised one UCL PhD student project to analyse NGS data on PCG (Primary Congenital Glaucoma) to find genetic biomarkers that might be causing this condition:

1. Dr Nicola Cronbach studied 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 basis will allow us to better understand MFS.

Research Manuscript Publications and Dissemination of the Results

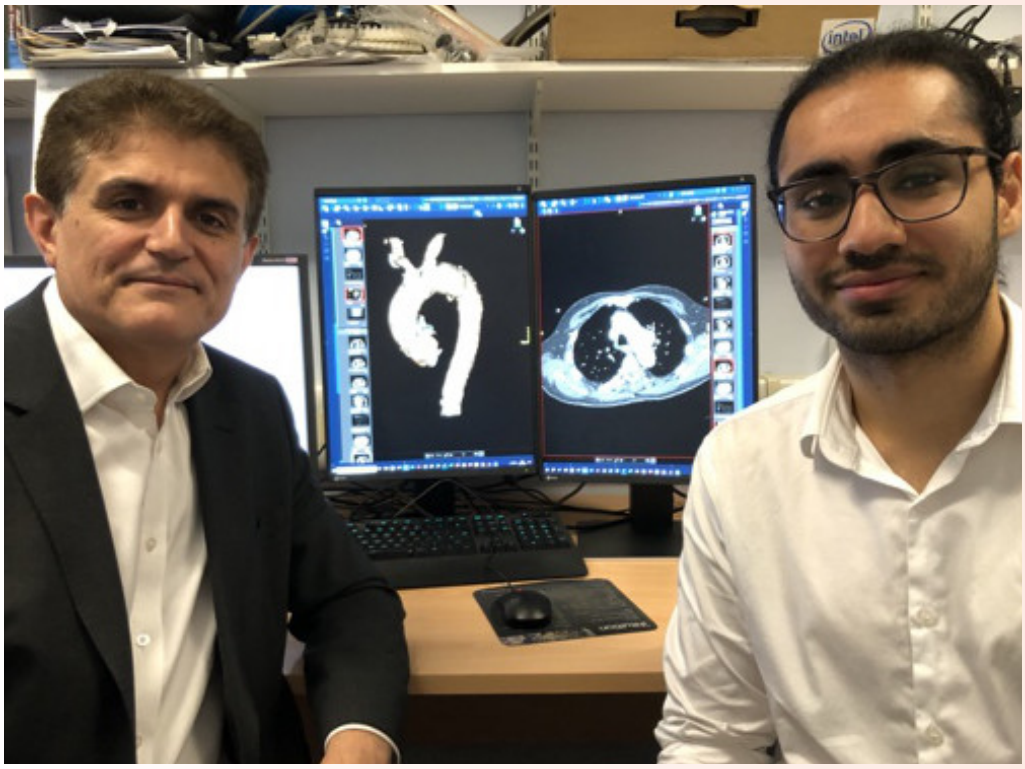
We are one of the authors in a review with one of our Ectopia Lentis (EL) collaborators, Dr Aman Chandra. This is expected to be published imminently. It is going to be an updated review on the genetics of EL.

Link of the published paper:
Article "Zonulopathies as Genetic Disorders of the Extracellular Matrix" has been published in Genes and is available online at the following links:

Website: **https://www.mdpi.com/2073-4425/15/12/1632**

PDF Version: **https://www.mdpi.com/2073-4425/15/12/1632/pdf**

When AI Met the Aorta



Breathing new life and bringing fresh ideas to the Marfan Trust, two medical students joined our team this summer, expanding our booklet selection and deepening our knowledge of aortic imaging.

AI met the Aorta in Mehar's research project, Picture Perfect, where he analysed over 500 CT scans to predict the measurement at which the aorta may dissect. Currently, the main measurement used to risk stratify patients is aortic diameter. The guidelines we use recommend surgery at certain diameters depending on factors such as the specific gene change an individual has. Unfortunately, we still see people who have aortic dissections at lower diameters, and we need to understand why this happens to try and prevent this life-threatening complication. This research looked beyond simply assessing the diameter of the aorta to whether there are other measurements that may help us to more accurately predict aortic dissection in the future, guiding timely intervention. Mehar used AI to look at aortic wall volumes in addition to diameter. He hopes that in the future, AI tools like this will be used to assess the aorta so that more individualised care can be provided. He has produced both a booklet (**https://www.marfantrust.org/resources/information-guides-**) and a presentation (**https://www.youtube.com/watch?v=aqzlvDrUKP0**).

The Eye in Marfan syndrome

Meanwhile, Navaneethan Adityaraj spent months investigating the eye in Marfan syndrome, producing a comprehensive guide which is now available to download. **https://www.marfantrust.org/resources/category/7-**

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**

Thriving with Marfan Syndrome: Embracing Life Beyond the Diagnosis

By Sarah Nuttman

Have you ever taken a moment to appreciate just how incredible it is to navigate life with a chronic condition while juggling everything else life throws at you? If not, you should. You're doing something remarkable every single day.

I'm Sarah. I'm 35 years young and living proof that Marfan syndrome doesn't have to define your entire life. I work full-time as a Civil Servant in Corporate Communications, and I live in Swansea with my long-term partner and our 2-year old cat, Winnie. My spare time is filled with things I love: catching up with friends, losing myself in a good book, writing, exploring new places, "dopamine dressing" (think bold, happy outfits!), and enjoying musical theatre.

My journey hasn't been without challenges. I've had four major aortic surgeries, suffered a stroke, and spent my 27th birthday in an induced coma. But I've also achieved incredible things: earning a bachelor's degree, dancing at Glastonbury, and buying a home with my partner.

I inherited Marfan Syndrome from my father and was diagnosed at birth. Like many of you, my life has involved a constant rhythm of medication, hospital appointments, surgeries, and managing symptoms. But here's the thing: Marfan Syndrome is just one part of who I am. My life is also full of joy, growth, and unforgettable moments.

As Marfan patients, we're also human beings with dreams, responsibilities, and passions just like everyone else. Living with a chronic condition adds an extra layer of complexity to our lives, one that others may never fully understand. Yet, we continue to thrive, adapt, and build meaningful, fulfilling lives.

Navigating Work and Health: Finding Balance

Since graduating from university, I've been working full-time, taking three extended periods of sick leave for surgeries, each lasting about six months. Some days are harder than others. Pain or fatigue might force me to slow down or wrap up my day early. I've been fortunate to have an understanding employer, but I've learned that success in the workplace with a chronic condition comes down to two key things: **honest communication and self-awareness.**

Understanding your limits and advocating for the support you need is crucial. Whether it's consulting with your GP, seeking advice from Occupational Health, or simply being upfront with your manager, clear communication makes a world of difference.

Embrace Your Strength

Living with Marfan Syndrome isn't easy, but it doesn't have to define your life or your happiness. Each of us has unique experiences and challenges, but we also have incredible resilience and strength. Celebrate your achievements, no matter how big or small. Seek out joy and connection in your daily life.

You're not just living with a chronic condition—you're thriving in spite of it. And that's something truly extraordinary.

International Patient Symposium

On a sunny Saturday in August, 220 patients, 13 medical experts, five patient advocates and four like-minded charities assembled in Bloomsbury, London, to discuss Living Better with Marfan, Loeys-Dietz and Vascular Ehlers-Danlos syndromes.

A year in the making, the structure of our symposium evolved through careful planning with the Marfan Foundation, the Aortic Dissection Charitable Trust and Annabelle's Challenge. To ensure it was accessible to all, the Marfan Trust offered travel bursaries to low-income households and provided a playroom for children. This would not have been possible without generous sponsorship from The National Lottery, The Worshipful Company of Basketmakers and Mazars.

Expert Information

Our Medical Director, **Dr Anne Child**, opened the conference, welcoming patients and their families to the Radisson Blu Hotel. With seven formal expert presentations and a choice of 12 breakout sessions, it promised to be a day crowded with niche knowledge and fantastic content.

Dr Child was also one of the expert speakers, imparting her expansive knowledge as she led a session on 'Ageing Well with a Connective Tissue Condition'. She presented the results of a survey of 50 patients with Marfan syndrome over the age of 50. Thirty years of research means an added 30 years of life, and the average life expectancy has risen from 32 years to 68 years. Medical and surgical care have improved. Surgery in later life might include hernia repair, varicose veins, joint surgery. Easy fatigability is an increasing problem. Specialist travel insurance may be necessary. Fitness should be maintained with gentle, regular exercise. Patients should not smoke. Be the best person you can be, and a model to the family.

Staying Healthy with an Aortic or Vascular Condition

Physical activity is a hot topic amongst people with Marfan and Loeys-Dietz syndromes. Medical emphasis has shifted from a negative to a positive approach, as exercise is increasingly proven to enhance the mood, mind and health of people with aortic conditions.

Dr Siddarth Prakesh from the University of Texas gave valuable advice including appropriate choices both pre- and post-aortic surgery. It is important to have a complete physical check before starting an exercise program and avoid isometric exercises where you contract muscles without moving joints. Light to moderate aerobic exercise daily, about 2 ½ hours per week total is recommended. If you take a beta blocker, heart rate may not be an accurate indicator of your exercise intensity. You should not exert yourself to the point of exhaustion; start low, go slow. A personal risk for exercise should be calculated with your doctor depending on your medical history, family history, blood pressure monitoring and echocardiogram, as well as a treadmill exercise test.

Options in Aortic Surgery

Professor Christoph Nienaber and **Mr Andreas Hoschtitzky**, Consultant Cardiologist and Cardiothoracic Surgeon respectively and both from Royal Brompton Hospital, shared insights and options for aortic surgery. The question of prediction for Type A aortic dissection has been elucidated by a new non-invasive test. Analysis of the length of the entire aorta shows that age-dependent lengthening is increased in connective tissue disorders. This provides a new predictive sign, helping surgeons decide when to operate to prevent dissection. Surgical techniques, including personalised external aortic root support (PEARS) were discussed giving hope for the continued development of new treatment options.

Vascular Surgery

Dr Sherene Shalhub, a vascular surgeon from Oregon explained that in vascular surgery, the choice of timing and type of surgery is dependent on the mutation and gene involved (genotype – surgical phenotype correlation). This is because the tissue strength is different in different connective tissue disorders.

Recovering from Heart Surgery

Marfan Trust Clinical Nurse Specialist, **Joanne Jessup**, led a session for those patients anticipating heart surgery. As a cardiac nurse she is uniquely qualified to dispense practical and psychological tactics and tips on what to expect and how to recover as well as possible.

How to be a Rockstar in a Crisis

The parent of an affected child, Carolin Oder joined us from Austria to stress how very important a positive outlook was in coping with her son's diagnosis and surgery. Best case scenario should always be the focus, which allows a parent or a patient to be optimistic in a health crisis. She recommends singing along to Elton John's 'I'm Still Standing' to give yourself courage and here we all are, still standing.

Creating Connections

During the coffee and lunch breaks, connections were made and friendships forged amongst the participants over tea … Here is Gareth Owens, Chair of the Aortic Dissection Awareness UK & Ireland talking to a fellow patient in a wonderful t-shirt.

and whilst enjoying the playroom:

Lottie and Georgie (left) both have Loeys-Dietz syndrome. They met for the first time at our Symposium where they discovered that they are navigating the same medical journey at the same hospital! It's the beginning of a new, mutually supportive partnership!

© Photography by Simon Burgess

The Marfan Trust table was constantly busy!

Whilst facilitating new friendships, the playroom also kept children creatively occupied as Isabella, pictured right, demonstrates. They emerged brandishing handmade tote bags, badges and keyrings! Isabella has created a character, the 'Marfasaurus' who will shortly appear in a children's book, one that will help youngsters to navigate the syndrome.

With grateful thanks to The National Lottery, The Worshipful Company of Basketmakers and Mazars for their support.

Further gratitude to Simon Burgess for his fantastic photography during the conference.

We are grateful to our fundraiser Amanda Corcoran for raising sponsorship so rapidly.

Marfan Europe Network – M.E.N.

United behind the motto Together We Can, the Marfan Europe Network (M.E.N.) is a coalition of 18 wonderful patient organisations. In September we attended their four-day meeting in Vienna at which we toasted M.E.N.'s new legal status, discussed future collaborations, listened to educational lectures and, just as importantly, socialised.

Teamwork

During the four-day congress we approved M.E.N.'s new bylaws, such that the organisation is now legally empowered to better represent the member states of Europe. A new website will shortly follow, as will a closer collaboration between each of the member states.

On the first full day we discussed our aspirations and vision for the M.E.N. and how to achieve these. Some teamwork building exercises ensued and here we are, creating marshmallow towers.

Young Adult Network

Day Three opened with a fascinating lecture from Dr Christine Pees of the Children's Heart Centre in Vienna. Dr Pees discussed the discovery and treatment of Loeys-Dietz syndrome and how it differs from Marfan syndrome. In 2005, Marfan Type II became formally known as Loeys-Dietz. It is

characterised by the 'triad' of:

- Hypertelorism (widely spaced eyes)
- Cleft Palate / Bifid Uvula
- Arterial Tortuosity, Aneurysm & The congress involved representatives from: Austria, Belgium, Denmark, Finland, France, Luxembourg, The Netherlands, Norway, Spain and the United Kingdom, with special guest star Eileen Novins from the Marfan Foundation in the USA.
- There was also a small coalition of Young Adults who've transcended their country borders to create a pan-European network.

If you are interested in joining this friendly group of people aged roughly between 18 and 30, please do email us. They're keen to meet fellow Marfan/Loeys-Dietz patients from the UK.

We invited Dr Pees to speak at our recent Information Day and you can watch her full talk by following this link: **https://www.youtube.com/watch?v=7ETgvVGoO4s**

Following Dr Pees' lecture came **Romain Alderweireldt** whose nine-year-old son has a severe form of MFS. inspired by the maverick spirit of PEARS-pioneer Tal Golesworthy, Romain and his partner Ludivine Verboogen, created 101 Genomes, a research project exploring the modifier genes that counter the cardiovascular implications of Marfan syndrome. The catalyst for this particular type of research was the discovery of apparently healthy individuals with pathogenic variants on the *FBN1* gene. Their healthy status suggests that they may be genetically protected from even the most severe forms of Marfan syndrome by the action of a so-called protective (epistatic) gene.

Romain opened our Marfan Information Day on Saturday, 12 October and here is the link to his full presentation: **https://www.youtube.com/watch?v=wOaTi-df7fY**

After a morning filled with fascinating lectures we left our cosy hotel bubble for a nostalgic tram ride through the centre of Vienna and Storm Boris!


Victoria with Ludivine Verboogen on the 'nostalgie tram'

A huge thank you to the M.E.N. and the Austrian hosts - Margit and Dagmar - for organising such a friendly and fantastic conference. We are looking forward to a close working collaboration and raising Europe-wide awareness of Marfan syndrome.

In Memory

Michael Edwards

We were so saddened and shocked to hear of the sudden death of Michael Edwards in November of this year. Michael was in frequent contact with the Marfan Trust, offering helpful health suggestions and giving us insights into his life with Marfan syndrome. He was a talented viola player who brightened the dark days of lockdown with his rendition of the Ashokan Farewell. We have shared this on YouTube and here is the link: **https://www.youtube.com/@marfantrust3841** We will write a fuller tribute to Michael in our next edition of Marfan Matters. In the meantime, we extend our condolences to all who knew him.

Margaret Evans

Margaret Evans was a beloved headmistress as well as a talented singer and conductor. She died at the age of 84 in August and the Oxshott Choral Society's Christmas Carol Concert was held in her memory, raising £1,079.73. Dr Child attended this magical event at St John's School Chapel in Leatherhead and spoke at the end, introducing the audience to Marfan syndrome. This photograph, taken after the bows, depicts soprano Jane Searle (in burgundy), mezzo-soprano Elizabeth Searle (gold glittery dress) and organist/pianist, Matthew Rickard.

For as long as the present conductor Valerie Beynon has conducted it – 50 years! Margaret was diagnosed with Marfan syndrome quite late in life and nominated our charity to benefit from her Choir's concert activity in 2015.

Fundraising Feats

Fuelled by the goodwill of our fundraisers, the Marfan Trust is very grateful to each and every one of you for your loyalty and support. Without your amazing feats of charity, we would not be able to continue our good work, improving and saving the lives of people with Marfan and Loeys-Dietz syndromes. Here is a small selection of your amazing feats:

Cannonbawz Run

Ten years ago, Kris O'Neill turned a long-cherished ambition into the most exciting reality. He dreamt of a fundraising car rally that would help Marfan patients like his late brother, Liam. Earlier this year, Kris and his 'bawzers celebrated a decade of the Cannonbawz Run, a fundraising phenomenon that celebrates the cinema of Burt Reynolds while taking in the stunning scenery of Scotland's North Coast. This year alone Kris raised £5,000 while navigating the 560-mile journey around the NC 500! Just as importantly he drew public awareness to Marfan syndrome with an interview in The Northern Scot. Thank you so much, Kris and your crew, for your continued support. **https://bit.ly/3v9LNJv**

Downham Young Farmers

In memory of Jamie Morton, the Downham Young Farmers appointed the Marfan Trust as their charity of choice 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. Not long before his passing, Jamie was diagnosed with Marfan syndrome.

The Downham Young Farmers staged a 98-strong tractor pageant in January and have subsequently held other events for the Marfan Trust including clay pigeon shooting. Overall, they have raised £3,000. Thank you so much.

Wooden Wonders

By Ruth Cullingford

My husband David has been creating and selling his lathe-turned wooden items for years. During the year he had a stall at two events where he sold his creations. One was at the Melplash Show in August and the other at a Makers Market in November.

We displayed the Marfan Trust leaflets on the stall as it encourages people to ask questions about the condition, and is informing the public about Marfan.

David's creations raised £600 for the Trust. Thank you!!!!

Fundraising Feats

Michael Farrow

By Michael

I recently put on an event to celebrate one year of Northern Delinquency, an independent music label I run based in Preston. The event was hosted at Chain House Tap Room.

The artists who played the event all received fees from Chain House which we have donated to charity. Chain House have been extremely generous and we would like to therefore make a donation to the Marfan Trust of £200 from the evening.

This event is in memory of my mum, Liz Farrow, who was a volunteer for the Marfan Association with Beryl Henshaw for many years and did tireless fundraising and awareness during her short life regarding Marfan syndrome awareness and research.

And it's a small way of thanking the Marfan Trust for their work and support.

I have attached a selection of photos from the Northern Delinquency First Year Bash here taken by Kyle Ashton. I wish to thank Ryan from Chain House Tap Room for his invaluable help.

Beryl Henshaw was a very close friend of both my mum and the family, she is sorely missed. Before she sadly passed she took time to visit me after my aortic aneurysm. This event is for Liz Farrow, from myself, dad, Bob, and sister, Catherine.

Keep up the good work and thanks again.

7Formation

In December 2023, we were approached by Corby-based construction company 7formation with some wonderful news. We were one of three charities who would benefit from their fantastic fundraising in 2024. Prioritising charity has long been a core value for 7formation and in 2024 they established 7foundation, their charity arm. This wonderful engaged team staged three events during the year: a football match, a long walk, and a golf tournament, raising over £3,000 for the Marfan Trust. Thank you, Alastair, Jak, Kitty and your fantastic team

Trustee Treasurer Needed

Are you a Chartered Accountant? The Trustees of the Marfan Trust CIO are seeking to add a Trustee Treasurer who lives in or near London. The Trust is expanding. Duties would encompass supervising the Finance Officer who helps provide an annual budget and cashflow forecast supplemented by management accounts for each quarterly remote trustee meeting. Yearly audits are prepared by our auditors' with the information provided. When required, an explanation of the finances to the remaining trustees would be valuable so they can discharge their duties properly. This post would be unpaid and would suit a retired or retiring Chartered Accountant. For further information please contact Dr Child via email annechildgenetics@outlook.com or telephone Victoria Hilton on our Information Line 033301152

Lucie's Christmas Stall

Sewing with a smile, Lucie Nicholson created a collection of gorgeous soft toys, following designs by Jo Carter, for her local Christmas Fair ...

by Lucie

I was very lucky to be able to team up with the local Rotary Club for a Christmas Fair at the Bowes Museum, Barnard Castle. They allowed me to share their stand and we attracted customers for each other. As a result, we helped each other to raise funds for various causes, mine being the Marfan Trust!

I had 10 toys to sell as my main item and half of them were bought by Rotary members!!! We also had some small items that were added to the Rotary Christmas Quiz; some people have been doing them for years and come back for more.

Whilst at the stall, I met a lady whose brother has Marfan syndrome! He has had a valve replacement; and his son is being tested for the condition. There was a lady who has Ehlers-Danlos syndrome so has heard of MFS and one lady who knew someone from her village with MFS. Then we had an ophthalmologist, who is testing diabetics and has never heard about MFS and a radiographer, who has seen MFS patients in her career.

My son is preparing for his aortic operation early next year and I am glad that I was able to raise funds and awareness for the Trust.

Mr and Mrs Peter and Lisa Newsome are on a tight budget, and have discovered a way to have their own electric car for their carers to drive, to appointments, shopping and outings.

They are using part of their PIP to rent a Dacia Spring Extreme 4 seater car. They are also applying for a driveway in which to park it, and for a home electric charging point to be installed.

Otherwise they must fund mileage to appointments, some of which are far away. They recommend others to google their nearest dealer and try the car. Special discounts are available until January.

Aortic Dissection Awareness Day: 19 September

A momentous date in the diary, Aortic Dissection Awareness Day is marked by copious conferences, many discussions and lots of social media activities. We attended two seminars during which patients' stories were shared and new research projects unveiled.

By Joanne Jessup

To mark Aortic Dissection Awareness Day, ADAUK staged another fascinating conference, the theme of which was 'Aortic Dissection Surgery Around the World'. Brave patient representatives and their surgeons from the US, Canada, Malaysia, Brazil, UK, Ireland and Europe shared moving stories about their personal experiences and hopeful information about the vast amounts of work going on across the globe to improve diagnosis, treatment and outcomes for everyone affected by aortic dissection. Highlights for me included hearing how seven 'lucky breaks' (including a COVID lockdown!) helped contribute to the rapid diagnosis and treatment of one dissection survivor while another described his experience of aortic dissection in Nepal after an epic hiking expedition and the invaluable support of his friends and family.

Next year the conference is titled 'Think Aorta, Think Family' and will focus on the importance of genetic screening and diagnostics for the wider family. This is a topic we are passionate about at the Marfan Trust and we cannot wait to join the team again in 2025.

Meanwhile, the Aortic Dissection Charitable Trust unveiled their three research grant winners for 2025. These three projects will investigate the outcomes and health-related quality of life after an aortic dissection; spotlight AI-driven pathology as a digital tool for aortic dissection, and research the development of a diagnostic blood test for acute aortic dissection. Just as importantly, ADCT recently announced they are funding NHS Trusts to appoint Aortic Specialist Nurses and Advanced Practitioners. These roles will transform aortic dissection care by providing life-saving follow-up, holistic support and genetic screening to protect families.

London Aorta

Several overarching themes threaded their way through this two-day conference. The genetics behind aortic disease were discussed, as was the concept of the aorta as an organ. Much time was spent describing the progress made in treating the aortic arch. Joanne Jessup spoke on the growing prevalence of aortopathy clinics in hospitals and the increasingly recognised importance of aortic nurse specialist. Dr Kurz from the University Medical Hospital of Berlin took a global approach when talking about the time-sensitive nature of Acute Type A Dissection. Discussing country by country he presented data on how fast transportation to high volume medical centres and early diagnosis of Acute Type A Dissection would save lives.

Marfan Information Day

Our virtual conference opened with news of an exciting research project and closed with a compelling talk on how to advocate for yourself from Gareth Owens, Chair of ADAUK. Head to our YouTube channel to find out more.

Lindsey Erith's Wanton Troopers

Marfan patient Lindsey Erith is a portrait artist and a recently published author. Her second book Wanton Troopers is a tale of forbidden love, seduction, betrayal and burglary set during the First English Civil War. It's a page-turner!

Blog Corner

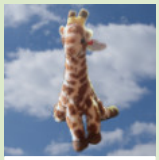
Lucy lives with her mother and pet tortoise. She is a crystal-collecting, plant-loving creative crafter with Marfan syndrome. We often share Lucy's candid blogs on social media and here's the path to her personal site. **https://www.marfantrust.org/articles/category/15-blog**

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

The Marfan Trust Online Shop


Willow

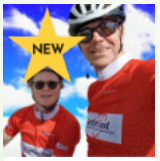
Tote Bag


Marfan Pen

Marfan Cap


Heart Keyring


Marfan Mug



Cycle Tops

Jeremy Wins


Scrunchie


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 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 ★	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		
Hand-crocheted Scrunchie								£3.00		
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								£3.00		
SUM TOTAL (including postage)										