Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Marfan Matters

Autumn/Winter 2023 Edition

Regular Features

Special Features

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Dr Anne Child chairing a session at London Aorta

Autumn heralded fiery hues and copious conferences from which we emerged exhilarated and brimming with new insights. September spelled Aortic Dissection Awareness Month and the body's largest artery was the subject of several big campaigns, and two different conferences spanning three full days, both emphasising the necessity of a multidisciplinary approach to its treatment. See page 10 for soundbites, updates and details.


Assembling for Student Presentations

As summer ebbed away, we grew in knowledge. Three medical students, Elsa Harte, Laxmi Thileepan, and Simon Khatra spent the summer exploring, respectively, Neonatal Marfan Syndrome, the Psychosocial Impact of Marfan Syndrome upon the Family, and Gastrointestinal Symptoms in Marfan Syndrome. Their research, and resulting material, has reaped results, deepening our understanding of the subjects and producing fascinating factsheets for our supporters.

From two recently held high-octane webinars (summary on pages 15) 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.

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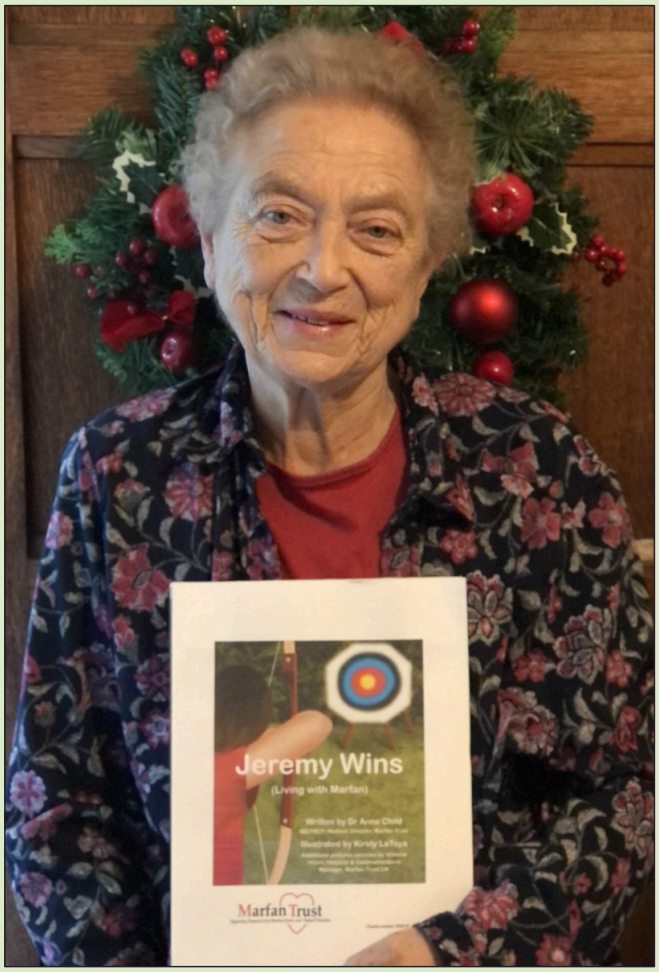
A Word from Our Chair, Dr Anne Child MD FRCP

Hello, Old Friends and New.

At the time of writing we're basking in the afterglow of a wonderful Marfan Information Day. Held just three weeks ago (at the time of writing) we expanded our subjects to include those we felt were under-researched including gastrointestinal symptoms in Marfan syndrome. One of our three Brian Adams Memorial Scholarship students took this on, and his research can be found on page 7.

Financially we are doing well. We have managed to keep most of the legacy of the Marfan Association and have supplemented this with generous donations from our fabulous fundraisers. We are pleased to say that we have a new legacy of £50,000 per year, which we are using half to support our unique laboratory research programme for Marfan syndrome and related disorders, and half to fund our marvellous nurse, Joanne. We hope more of our followers will take up our offer of low-cost will-writing programme (see our website) and include a legacy no matter how small for the Marfan Trust, to ensure the continuity of our important service and research programme reported in this newsletter. This benefits our specialised Marfan community.

We wish you a Very Merry Christmas and a Happy New Year!



Anne H Child

Dr Anne Child MD FRCP

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Helpline Analysis

by Victoria Hilton

The helpline continues apace with questions arriving thick and fast, some that can be answered readily whilst others prompt much research and pleas to our advisory committee. And there are queries for which there is no definitive answer but to give the patient all the available information and leave it to them to decide.

Beta-Blockers for Children

One such question, echoed by several parents, concerns beta-blockers and the wisdom of administering these to Marfan children who, as yet, show no signs of aortic dilation. In each instance, the child was a young boy who'd been prescribed beta-blocker therapy by their paediatric cardiologist. The parents phoned our helpline, seeking our thoughts on the matter, and our response is reproduced in Dr Child's Casebook, "The Path of Marfan", next page.

Hormone Replacement Therapy (HRT) in Menopause

Another question to which there is no definitive answer is use of HRT. Women with Marfan continue to ask if HRT is safe to take. We posed the conundrum whilst at the Marfan Foundation's Patient Symposium in Paris. Dr Juan Bowen of the Mayo Clinic said he would treat Marfan patients as he would his other patients, and recommends they take it for five years. Dr Bart Loeys commented that whilst there's insufficient research on the subject "it is better than the alternative which is osteoporosis". He emphasised that the doses of hormone in replacement therapy are less than those of the natural oestrogen which occurs in high level during pregnancy.

To resolve this definitively, we approached Consultant Cardiologist Dr Isma Rafiq, at the Royal Brompton Hospital, with two scenarios. The first involved a peri-menopausal woman in her late forties with an ascending aortic aneurysm, and the second a menopausal woman in her fifties with no aortic involvement. Dr Rafiq confirmed that both 'patients' can safely take HRT.

All patients wanting to take HRT should discuss this with their cardiologist so recommendations can be made on an individual basis and referrals to endocrinology or gynaecology specialists can be made if appropriate. However, several doctors remain equivocal about prescribing HRT. Dr Child adds that patients must continue to have their annual echocardiograms while taking HRT in order to pick up any increase in aortic dilatation.

Insomnia

In other news, several supporters suffer from sleeplessness. The catalyst for insomnia varies and it is sometimes stress-induced, but one should consider sleep apnoea which can happen with Marfan syndrome (signs include disturbed sleep during the night, snoring with varied breathing rates and resultant daytime fatigue and drowsiness owing to lack of sound sleep). This is due to weakened connective tissue which affects the mechanical act of breathing. Whilst it's often associated with being overweight in the general population, sleep apnoea can be found in the slim Marfan community. It may be worth discussing this further with your GP, so they can ask about any signs or symptoms of sleep apnoea and refer you to a local respiratory team for sleep studies if appropriate.

Personal Independence Payment (PIP)

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.

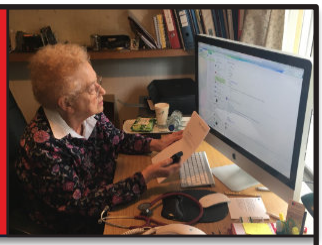
Pain

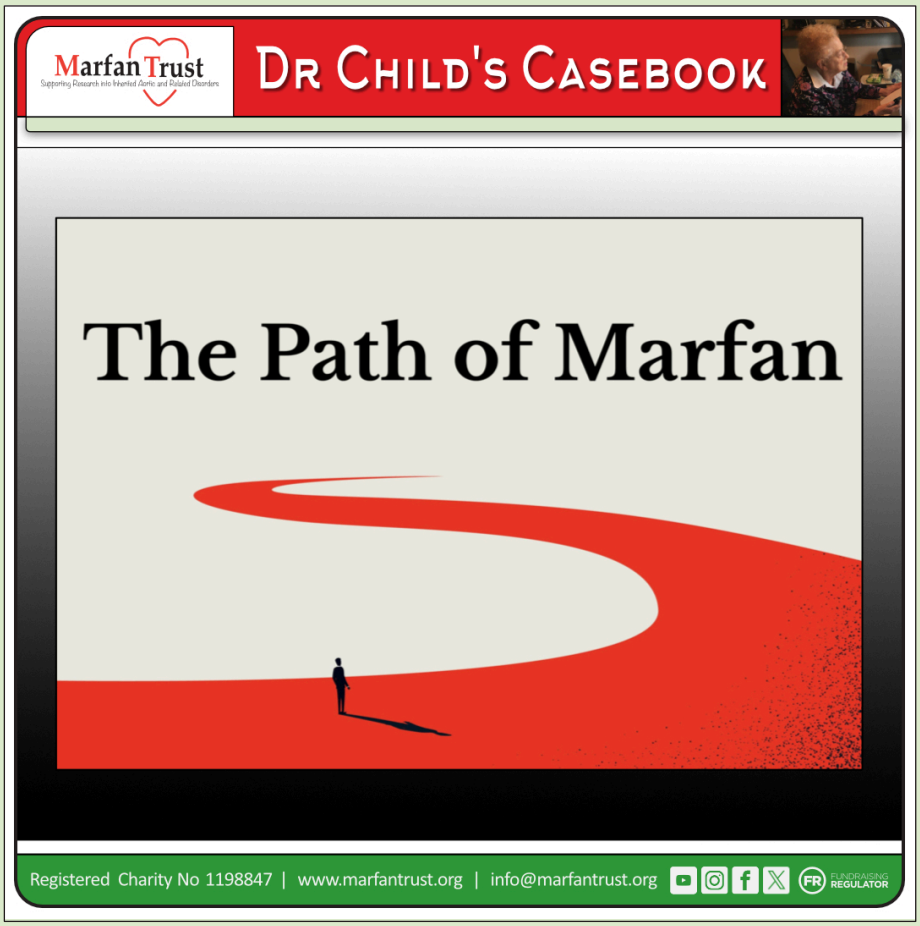
Pain is invisible but inescapable and is commonly and keenly felt in Marfan syndrome. Many approach the helpline hoping for advice on how to alleviate it. Dr Child has been investigating an emerging form of pain relief and her findings are published on page 10.

At the time of writing (20 October 2023) we've received 324 calls for help and advice thus far this year. This translates to **8 queries per week**, compared to 6 last year. Twenty-five per cent of queries are from those wondering if they, or a family member, has the syndrome which means we are continuing to find the undiagnosed.

Helpline Victoria & Joanne

Dr Child's Casebook: The Path of Marfan





Predicting the path of Marfan syndrome is made easier when the condition is inherited. The gene is said to 'breed true' and often follows the familial pattern. Doctors can take parental history and anticipate accordingly with medication. But what if they pre-empt something neither the child nor the parent displays, and prescribe preventative medication with unpleasant side-effects?

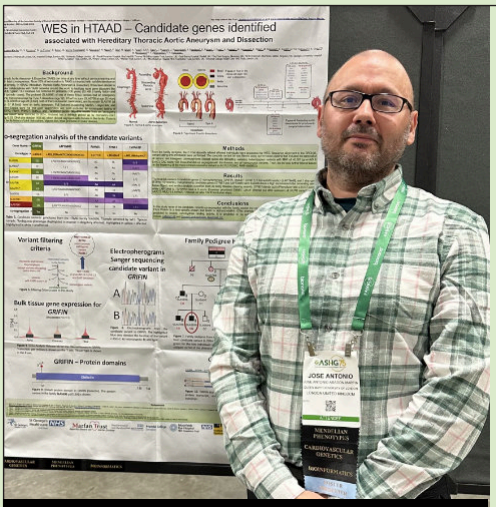
Q: I am a mother who has Marfan syndrome, as does my young four-year-old son. I have no heart involvement, nor does my child. However, he has just been prescribed beta-blocker therapy and I wonder as to the wisdom of this?

A: Some cardiologists recommend starting treatment immediately and not waiting for abnormalities to occur. Do you wait for your washing machine to break and then fix it or do you care for it, descale it and service it? Do you brush your teeth every day and go to the dentist or do you wait until you have toothache? I personally give parents all information concerning benefits and side-effects, and look at family history, but clinicians vary in their approach. I certainly have children with normal aortas taking medication.

If it is tolerated, this treatment buys time, by protecting the aortic wall against enlargement. Common side-effects are drowsiness, dizziness and occasionally nightmares.
Also, it is well documented that in many Marfan families the women are often mildly affected, while the men develop aortic aneurysm requiring surgery. The Marfan gene expresses itself more completely in the male. So, although this mother is mildly affected and does not require medication, it may be wise to start preventative treatment for her son. This will cover him during his rapid growth years, the period when aneurysms can first develop. Along with each annual echocardiogram, his heart involvement and treatment can be reassessed.

If side-effects occur the medication can be reduced, replaced, or stopped. Mother could ask for a further appointment to explain her feelings and hear the doctor's rationale. Beta-blockers usually are very well tolerated in children and are routinely given from birth.

Research Update
by Dr José Aragon-Martin



Jose: ASGH poster exhibition Washington 2023

A despatch from the genetic frontier …

At the time of writing, I am unveiling my new research at the American Society of Human Genetics (ASHG) 2023 Convention in Washington DC.

I have just presented a newly discovered gene causing Heritable Thoracic Aortic Aneurysm & Dissection (HTAAD). This is a momentous finding and was detected using whole exome sequencing (WES). The gene is called GRIFIN and is still in its infancy as further research must be performed on functional studies (how the gene relates to biological factors) and other families with the gene to clarify its nature. However, we currently have a UK HTAAD family that seems to be linking their clinical feature to GRIFIN.

My MSc students, Vatsala and Silvia have passed their exams and are joining me here in Washington DC for poster presentations. Whilst they have left the Sonalee Laboratory for private laboratory work, they are demonstrating their findings from the Marfan Trust at the ASHG. As discussed in the spring edition newsletter, Vatsala, Silvia, and a third student Sanjana (pictured) have been studying four families:

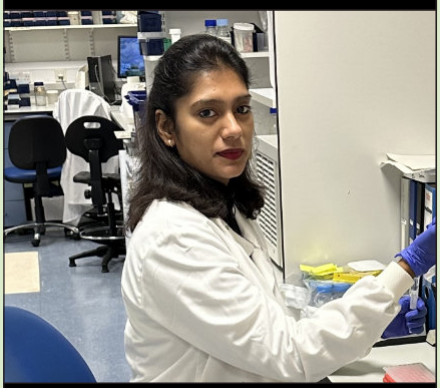
2 x HTAAD (Hereditary Thoracic Aortic Aneurysm & Dissection)

2 x hEDS (hypermobile Ehlers-Danlos syndrome)

Both are overlapping clinical conditions, and perhaps, genetic pathways. This last one must be proved first.

We were working with Sanger sequencing to genotype variants in the family members that approached us to do the research. We thought we had some failures, because we were not able to pick up the variant in Sanger, while it was observed in the NGS (Next Generation Sequencing) screening. We realised that perhaps these special variants are not artefacts (mistakes created by the technique) but either not picked up by the designed primers (therefore, new primers need to be designed), or these variants are from DNA fragments that belong to another region of the genome (translocation). We strongly think it is the latter, but to demonstrate this we will need to sequence family members with a technique that can prove this: either long read sequence (Oxford Nanopore, or PacBio), or Optical Genome Mapping. In an ideal world we would like to use Oxford Nanopore and Optical Genome Mapping at the same time to cover as much as we can when it comes to research.


from left: Vatsala Khurana & Silvia La Penna with Jose at ASHG 2023


Sanjana Babu Working in the Lab on Hypermobile EDS

That's all for now! I wish you a Very Merry Christmas and a Happy New Year!'

Summer Student Research

Summer Student Research

Breathing new life and bringing fresh ideas into the Marfan Trust, three medical students joined our team this summer, deepening our knowledge of the condition's manifestations, and enhancing our approach to our supporters.

Neonatal Marfan Syndrome *by Elsa Harte*


Elsa Harte

I had the opportunity to undertake a Summer Studentship working with Dr Aragon-Martin at the William Harvey Research Institute. Our project was researching Neonatal Marfan syndrome, which is a less prevalent subset of Marfan syndrome. Neonatal Marfan syndrome can present at birth or can be diagnosed in early childhood through a combination of clinical examinations by a doctor, scans, and genetic testing. Unlike classical Marfan syndrome, which is inherited from a parent's genes, Neonatal Marfan syndrome occurs due to a spontaneous (new) mutation and is a much more severe form of the syndrome

Our research aimed to collate the patients identified as having Neonatal Marfan syndrome from the patient case reports and corresponding genetic test results available at the Marfan Trust. We did this to identify the common features and genetic changes in Neonatal Marfan syndrome and how this may differ from classical Marfan syndrome. This is important when aiming to raise awareness about what is considered a rare condition, as it allows doctors to identify patients who may have neonatal Marfan syndrome, allowing for a quicker diagnosis and developing an understanding of what treatment may work best.

What did we find?

From the 1,334 case reports and 524 genetic probands (families), five cases of neonatal Marfan syndrome were identified. Furthermore, we had case reports from three patients currently under the care of the Marfan Trust. This is the largest collective of neonatal Marfan syndrome patients to date, which has allowed us to identify their common FBN1 exon mutations. All eight patients had mutations in the 24–32 exon regions. Exons are essentially the coding region of our genes that determine how we present characteristically. These specific exon changes are consistent with what we have seen published internationally in previous cases of neonatal Marfan syndrome and therefore add to the current evidence base.

Furthermore, the patients we identified presented with varying levels of cardiovascular compromise, which included a widened aorta and heart valve regurgitation. Although cardiac failure has previously been considered the most common cause of mortality in neonatal Marfan syndrome patients, our research has importantly identified several effective interventions which have allowed these patients to enjoy a good quality of life for longer. These include medications such as Irbesartan and Atenolol, which aim to reduce blood pressure and alleviate the stress on the heart, as well as surgical interventions such as valve repair/ replacement and aortic root support procedures.

These interventions coincide with the overall care these patients receive from their specialised team of nurses, physiotherapists, orthopaedic surgeons, and ophthalmologists to support any musculoskeletal and ocular conditions they may also experience, similar to the care received for classical Marfan syndrome.

Overall, the growing research into neonatal Marfan syndrome and the increasing evidence for effective interventions demonstrates promise for current and future patients with this condition.

Summer Student Research

The Psychosocial Impact of Marfan Syndrome Upon the Family *by Laxmi Thileepan*


Laxmi Thileepan

During the summer, I embarked on a research project focusing on the psychological impact of Marfan syndrome on families. This endeavour led to the creation of a structured questionnaire, which was subsequently distributed internally to families registered with the Marfan Trust. We received a notable and substantial response, totalling 108 completed surveys. This influx of data yielded meaningful insights into the utilisation of available resources and the emotional experiences accompanying the diagnosis of Marfan syndrome. In addition, I undertook the task of crafting informative pamphlets, distinctively tailored for parents and healthcare providers. These pamphlets direct individuals to a comprehensive array of resources within the Marfan community.

Analysis of the survey responses unveiled a pertinent finding: while resources were indeed accessible, a significant proportion—approximately 58% of families—remained unaware of, or did not fully engage with, support groups. For those who did participate in support groups, their experiences underscored the pivotal role these groups play in achieving and maintaining their mental health, acquiring practical tips, and fostering a sense of community.

The cornerstone of my research was centred on the application of the biopsychosocial model, a comprehensive framework widely used when making clinical decisions as it addresses many aspects of a patient's well-being. This model examines biological, psychological, and social factors. Our findings illuminated the substantial emotional, financial, and physical burden borne by caregivers. Many parents expressed sentiments of guilt and self-blame, emphasising the necessity for psychoeducational interventions to mitigate these emotions. Anxiety stemming from the inherent uncertainty of the future emerged as a recurring theme, underscoring the profound impact of these complex emotions. Coping strategies encompass participation in support groups, engagement in psychotherapy, and a deliberate effort to focus on manageable aspects, such as being able to advocate for your child.

It is noteworthy that the repercussions of grappling with a chronic illness extend beyond the individual patient and encompass the entire family unit. This can manifest as heightened tension within the family's social dynamics as there is added stress such as dealing with medication. To navigate these challenges, it is imperative to harness available support avenues, including charitable organisations, support groups, online forums, and healthcare providers.

A central conclusion arising from this research is the pressing need for heightened awareness concerning readily available support services. Achieving this objective necessitates a multifaceted approach, including the provision of tailored activities for parents to engage their children in, as well as clear signposting of available support resources. This can be accomplished through educating healthcare professionals, parents, and caregivers.

Summer Student Research


Simon Khatra

Gastrointestinal symptoms experienced by people affected by Marfan syndrome *by Simon Khatra*

In this super short summary, I will discuss some of the gastrointestinal symptoms that the Marfan population *may* experience. Gastrointestinal complications in the Marfan population have been poorly researched and evidenced since they are not a 'characteristic' trait of Marfan. Highlighted below is a small collection of scientific studies and anonymised patient interviews that I conducted.

Some of the Marfan population may experience symptoms that belong to a functional gastrointestinal disorder (these are symptoms that continue in the absence of any pathology or structural abnormality). The prevalence of these symptoms often include: diarrhoea, constipation, abdominal pain, bloating etc. Opinions in scientific literature vary when comparing these to the general population. However, after my interviews, all of the following symptoms were often described- including, 'abdominal pains, pain in the bowels, constipation, nausea and vomiting'.

Some methods that worked (**not an exhaustive list**):

- Everyone had found that changing/improving their current diet, changing out unhealthy options for healthier alternatives usually improved their symptoms.
- Moreover, by tackling and changing specific food items, this could be an effective way to track which certain foods may exacerbate/reduce the symptoms that they were experiencing.
- Often, by increasing the amount of fibre in the diet, this led to a reduction in gastrointestinal symptoms.

In addition, there have been several case reports in scientific literature highlighting the structural complications that can arise (and thus explain some of the gastrointestinal symptoms). Anecdotally, from my small sample size of interviewed patients, this seems to correspond with the lower numbers present also in the general population. Structural problems according to a handful of case reports in scientific literature may include: midgut volvulus (twisted bowel), small bowel diverticulosis (infected outpouchings), upper gastrointestinal bleeding and various types of hernia.

In conclusion, serious gastrointestinal manifestations are rare; however, this does not mean that the patients who suffer from Marfan syndrome may experience gastrointestinal symptoms which affect them on day-day basis. Due to these symptoms not being a 'classic' trait in Marfan, awareness of the prevalence is also severely reduced. This is evidenced in the fact that **ALL** of my patient interviews stated that they were in fact **NOT** told about the potential gastrointestinal complications that could be linked to Marfan syndrome.

The symptoms may present in a variety of ways in patients, and it is important for any patient experiencing any gastrointestinal symptoms to speak with a doctor as not only may there be a functional/motility disorder underlying the symptoms,but there could also be a structural complication, and to highlight the possible link with Marfan.

I would like to thank the Marfan Trust, my supervisor- Dr Child for her vast knowledge and guidance throughout the project. Victoria Hilton, in particular for helping to arrange the patient interviews and the GI webinar. Lastly, I would also like to thank the patients in the Marfan Trust who agreed to be interviewed, and hopefully their anonymised experiences will help increase the awareness of potential gastrointestinal complications in the Marfan population.

From the students' research have sprung factsheets. Scan the code to download your copy.

Lindsey's Story

by Victoria Hilton


In Conversation with Lindsey

"You're finished!" shouted Lindsey's mother in response to her daughter's diagnosis of Marfan syndrome. Lindsey was anything but, defying her mother's doom-laden words to live a life less ordinary. Whilst navigating through the physical pain of Marfan, she has found love and fulfilment in art, and recently wrote a racy novel about royalists. I visited Lindsey in her beautiful, ephemera-filled home in The Cotswolds to discuss her unusual life and work.

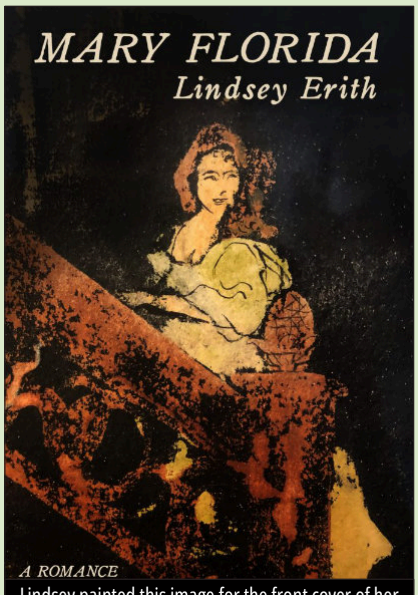
An elegant woman offering a glass of wine was the most welcome sight on an unusually cold spring day following a long coach journey from Victoria. Lindsey greeted me warmly into her gorgeous home, its walls covered with memories and art. Her father was the photographer, John Erith, and snapped the English actor Dennis Price whose portrait hangs amongst others in her decorative home. Over a delicious lunch we chatted, and Lindsey talked about her fascinating life.

Lindsey survived a girls' public school, and a complete and unexpected collapse in health at the age of 16 that left her bedridden. She had grown terribly tall but remained very thin. Trapped in a world of pain for months on end, unable to even sit upright, Lindsey let her imagination roam free and took solace in stories and secret scribbles. Unbeknownst to her, she had been diagnosed with Marfan syndrome (by Arthur Holman in University College Hospital). Her mother would not be seen with her, and refused to have her told. Lindsey had to learn to walk, and, after eventually learning of her medical condition, recalls laughing in her parents' face: "I'll show you". And so, she did.

Lindsey's father facilitated her escape to Art School where she lived in the appropriate 'artist's attic'. Her time there was punctuated by three hospital admissions as she realised that Marfan syndrome – the attendant pain, chronic fatigue and other untold features - was for life. But she nevertheless completed her Graphics diploma, then promptly fell in love and had a youthful romance with an Irish racehorse trainer. This led to her expert niche in equine and human portraiture, consolidating her interest in character and likeness. To the left is Lindsey's portrait of the fine steeplechaser who was the Queen Mother's 100th winner. It hangs in her late husband's 'office', a haven of sheet music and artworks.

Music filled the air as we continued the conversation into Lindsey's living area, a long, beamed room festooned with pictures including an arrestingly beautiful portrait of Lindsey. In the background plays Verdi's Requiem, produced by John Boyden, who was the love of Lindsey's life. Their Valentine's Day marriage was short but so sweet. He sadly died in 2021. A musicologist and record producer, John was the first managing director of the London Symphony Orchestra, and went on to manage the New Queen's Hall Orchestra. He emboldened Lindsey to take the next step in her career – that of novelist. Progressively weak wrists meant Lindsey no longer had the control for delicate artwork, and so she turned to capturing man's likeness in words on the page.

On her typewriter, Lindsey quietly conjured a character, whose adventures inform a racy novel about royalists. Set at the close of the English Civil War, *Mary Florida,* reinvents what it means to be a hero and the siren song for a lost cause. Its titular character, Mary Florida, is the alluring motivation for the protagonist's actions and their romance unfolds lavishly chapter by chapter. Lindsey tentatively gave her husband the manuscript of her historical romance, and upon reading it, he encouraged her to immediately seek a publisher. It was released by Cranthorpe Millner, the first publisher Lindsey (tentatively) approached, in July 2022 and is a true page turner. (It's now available on audible and is read by Clive Heyward, an award-winning British actor, and the lute music was specially recorded for by Sam Brown). She's now working on her second novel, Wanton Troopers.



Lindsey painted this image for the front cover of her novel

Woven throughout Lindsey's life has been much Marfan-induced physical misery - circulatory pain, sinus trouble (thank heaven for Doxycycline), hip problems (eventually resolved by Orthopaedic Surgeon Sarah Muirhead Allwood) amongst many other complications. But the golden thread of music, art and romance has provided Lindsey with personal and professional fulfilment, and as I said goodbye to the exquisitely beautiful figure framed in the doorway I felt giddy with the conversation.

Industry Updates and Conference Gems

An Emerging Treatment for Pain

Invisible but inescapable, chronic pain is a common feature of Marfan syndrome and we are increasingly asked about the use of Cannabinoid products for its treatment. This is an area that is currently the subject of scientific interest and research studies.

There are clinics that can offer trials of cannabinoid medication with robust assessment and follow up: at present this is usually offered on a private basis.

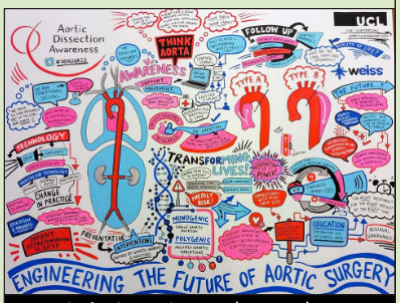
In response to the growing interest in this topic amongst our members, Dr Anne Child has visited one such clinic, the Jorja Emerson Centre, in London and spoken at length to their Medical Director Dr David Tang about the service they offer. “In the three years that I have been prescribing medical cannabis, it is really quite obvious that in the patient with chronic pain, medical cannabis offers a very well tolerated substitute to the opiates, gabapentinoids and tricyclic medications (amitriptyline/nortriptyline). Further to this, substituting out these pharmaceutical medications will not only remove the side effects of these medications suffered by the patient, but also will confer much improved sleep in the vast majority of patients, something we know is a part of the clinical conundrum that is chronic pain”.

More information is available on their website https://www.jorjaemersoncentre.com/, and we will be inviting Dr Tang to participate in a webinar on this topic in the coming months.

Conference Gems–Aortic Dissection Awareness Day & London Aorta

Once dubbed the body’s ‘orphan organ’, the aorta was the focus of many campaigns, discussions and two conferences during September which spells Aortic Dissection Awareness Month. We attended both symposiums and left feeling very knowledgeable and hopeful.

Emphasising the patient experience, **Aortic Dissection Awareness Day** on 19 September at the Royal Papworth Hospital featured an array of compelling speakers from surgeons, cardiologists, anaesthetists and nurses to patients and their families. ‘Humbling’ was the word woven throughout the medical presentations in response to the eloquently told stories from dissection survivors, and relatives of those lost to dissection. Organised by patient charity, Aortic Dissection Awareness UK and Ireland, the conference stressed the themes of early diagnosis and safe transfer, eg ensuring tight blood pressure control during transport. Genetic testing and early rehabilitation to facilitate recovery and a coming-to-terms with dissection were also main topics of discussion during the day.



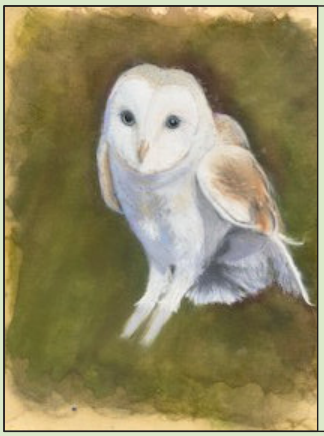
Artist Jenny Leonard created an illustrative montage of the day, with the salient points of the conference vividly rendered.

Across to the other side of the consulting table, **London Aorta** at the Royal College of Physicians, is a two-day conference, held annually, whose aim is to foster a multidisciplinary approach to the treatment of the ‘pulse of life’. No more the ‘forgotten’ organ, the aorta now has many medical champions! This year’s symposium expanded the selection of speakers beyond surgeons and physicians to include nurses and also a psychologist, ie those perhaps more intimately involved in the long-term care of parents.

With a nod to the past and an eye on the future Professor John Pepper ushered in the two-day discussion, focussing on the burgeoning role of AI in cardiovascular medicine. A paradigm full of possibilities but also problems, AI has facilitated imaging which has, in turn, revolutionised early diagnosis and management of aortic disease, leading to personalized risk stratification for individuals at risk of dissection. The future is bright!

Continuing the theme of technology and medicine, Dr Ganan Srithara described a health community app called Health Share in North London which is demonstrating health benefits in various different communities from diabetes to aortic dissection. Aim is to engage patients so they can take a more proactive role in the management of their condition whilst having resources from professionals available and creating a community of patients who can support each other.

In Memory



Annie Anderson

Beloved wife, mother, grandmother, friend, and supporter of the Marfan Association and Trust, Annie Anderson sadly died earlier in the year. Annie was a keen artist and crafter, and very helpful to others in her art group in Dunsford which was held every Friday morning. A close friend of Annie's, Babs Sawyers, sold her paintings and cards at their local church art & craft fayre, sending the proceeds to the Marfan Trust. We reproduce here one of Annie's beautiful works.

Caroline Tudgey

Over £1,500 was raised for the Marfan Trust in memory of Caroline Tudgey, the beloved wife of Graham and mother of Stephanie. The generous donations were from Caroline's close family and friends.

Carole Finkletaub

A warm-hearted woman with spirit and style who turned her private sorrow into a force for good. We are terribly sad to report the sudden death of Carole Finkletaub, who spoke at our Marfan Information Day on 7 October. Carole lost her son Daniel to complications of Marfan syndrome and responded by creating a student award in his memory. In doing so she has facilitated life-saving research into the condition.

Carole's son Daniel died in 2009 at the age of 41 of complications from Marfan syndrome. Although his life ended prematurely, it was a life lived fully, adventurously, and studiously! Daniel worked hard, completing two degrees and rising through the ranks of corporate marketing in companies from ABN AMRO to Vocalink. Constantly curious he enjoyed travelling, visiting countries to experience their food and culture. Daniel was diagnosed with Marfan syndrome at the age of 14 and remained healthy until a heart valve replacement when he was 26 years old. Thankfully that operation was successful, allowing him a quality of life until his late thirties.

Daniel became involved with Dr Anne Child and the Marfan Trust by chance when he read an article in the Radio Times in the late 1980s. He later served on the Trust's Board for six years between 1991 and 1996.

To continue the Trust's precious research into Marfan syndrome, Carole and her daughter Elaine, created a Student Prize in memory of their beloved son and brother.

Elaine will continue to fund the prize in Daniel's name, ensuring that her mother and brother's legacy is long and lasting.

Feats Of Fundraising

Unstoppable in their stamina, courage and loyalty, Marfan Trust fundraisers have delved deep into their inner resources and turned their lives around to meet the demands of daunting charity challenges. Here we celebrate just a few of their amazing feats.

Freddie Beard

Freddie Swam the Serpentine

His life saved by the chance intervention of a junior doctor on placement, Freddie went from studying to surgery, and in September swam six miles for the Marfan Trust. In 2013, Freddie was admitted to hospital with a protruding ribcage, recognised by an astute young doctor to be a sign of Marfan syndrome. In 2016 he underwent open-heart surgery, which interrupted his university studies (though he was back after just a month, revising and attending lectures). And this summer he swam six miles in three hours, raising over £1,000.

Freddie has a message for our readers: "I would really like to thank all of those who were generous enough to donate and for giving me all the encouragement and support to get through this swim. Without it, I don't think I could have pushed myself as hard and as fast as I did! Once again, I hope this is a push to those with Marfan, and even those without, that if you put your mind to it you can achieve anything!

All of us with Marfan are superstars, and nothing can hold us back."

Life is a Cabaret by Louise Baskeyfield

Following on from our successful Charity cabaret held in 2021 which raised over £5000 for the Marfan Trust, we held another charity cabaret on the 16th June in a community centre, in the village of Duxford in Cambridgeshire. This time the event was for two charities close to the hearts of the organisers. The Marfan Trust was one of the charities and we were able to raise £2,205.

The Marfan Trust was chosen following the loss of my father who died of an aortic dissection on 23 December 2019. Just two weeks earlier, I suffered a heart attack following a spontaneous coronary artery dissection (SCAD). My father, my son and I had all been under genetics for several years to try and identify a faulty gene.

It was only after these traumatic events that a diagnosis of Loeys-Dietz was confirmed. My children were subsequently tested and confirmed to also have LDS.

The charity event featured West End singers, comedians and a magician and it was a roaring success. We hope the money raised will help the Marfan Trust with the wonderful work they do.

My father was a wonderful man who did much fundraising throughout his life and did a lot for the village community. However, he lived his life unaware of the condition he had and, poignantly, would never have heard of the Marfan Trust.


Louise Baskeyfield with Family & Friends at the Cabaret

Feats Of Fundraising

Cannonbawz Run

Continuing gratitude for the unstoppable support from Kris O'Neill and his Cannonbawz crew. Their exhilarating movie-themed car rallies in the most scenic spots of Scotland have drawn much public awareness to Marfan syndrome and raised huge funds for the Trust. Recent rallies in 2023, including one styled along the lines of their namesake, Smokey and the Bandit, have garnered over £6,000.

An idea that combined his love of cars with a wish to help his brother, Liam, and others with Marfan syndrome, came to Kris some nine years ago. He converted it into the wonderful reality that is the Cannonbawz Run, and a lasting tribute to Liam who tragically died in 2021 of complications following surgery.

Celebrating George Franks-Herbert

We didn't have the pleasure of meeting George Franks-Herbert but felt we got to know him a little when he was celebrated and remembered so vividly at Cardinal Newman R C School in 2022. George died at just 18 of complications from Marfan syndrome whilst in his first year at Bristol University. His former school hosted a memorial, attended by the Marfan Trust, at which funds and awareness were raised by family and school friends. Cardinal Newman also created an annual football match in George's memory, and this summer raised £600 in a contest between staff and students. Apparently, the teachers won!


George's brother and sister enjoying the memorial match


James' Friend, Rich (left), Will Be Running for the Marfan Trust in Tenerife Next Year

James Kenny

His daughter's recent diagnosis of Marfan syndrome prompted James to take part in a 16km run on the South Downs. Here he is in the white vest, exhilarated and energised after his amazing run, having raised £745 for the Trust.

Feats Of Fundraising

Scott Ran for a Better Future

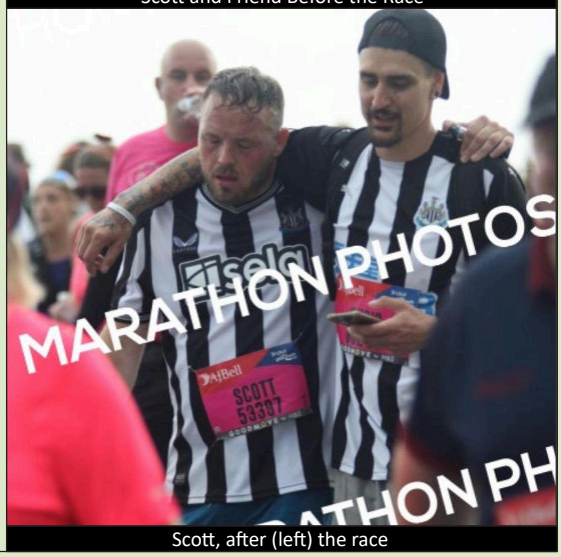
Amongst the many who took part in the 42nd Great North Run this September was Scott Fairbairn-Smith. Running in memory, but also in hope of a better future for Marfan patients, Scott shed weight and quit alcohol in preparation for his daunting challenge.

Scott's brother-in-law Fred was one of triplets. It was later discovered that he, his triplet sister (Scott's partner), and younger brother had inherited Marfan syndrome from their mother who had developed the condition spontaneously. Tragically, Fred died at just 30 following surgery to repair an aortic dissection, leaving behind a 9-year-old daughter.

In response, Scott wanted to raise funds but also awareness of this condition. Both his sons have inherited Marfan. Upon deciding to take on the Great North Run, Scott began seriously training, losing 3.5 stone in six months and giving up drinking. He completely turned around his physical and mental health.

On a very hot Sunday in September (10th) Scott and his friend ran 13.1 miles, raising £1,400 - and much awareness - for the Marfan Trust. It was no mean feat and the run took its toll as you can see from the before and after pictures. Thank you so much, Scott.

Scott and Friend Before the Race



Scott, after (left) the race

Lisa Newsome demonstrating her mobility scooter

Lisa's New Scooter

Lisa shown modelling her new Monarch Air mobility scooter, which is the lightest scooter available, and folds down into a very compact form. It comes with various optional extras, for which Lisa bought a travelling bag.

Obtaining it was made possible due to financial contributions of £500 each from the charities Independence at Home and The Hospital Saturday Fund. Lisa applied enclosing a letter of support from the Marfan Trust.

Huge thanks to the above, and Dr Anne Child & all at the Marfan Trust for making this possible.

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.lusnews.uk/2023/10/a-brief-update.html**

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 Day

Marfan Information Day

A seamless success, our Information Day whisked supporters on the full journey of Marfan syndrome, from conception to old age, concentrating as much on lifestyle as on medicine. 'Thoroughly captivating' was the response from one attendee. We thank our wonderful speakers and supporters, and we are excited to announce that next year's conference will be an in-person event in London! We are collaborating with the Marfan Foundation of the United States in organising this patient symposium, and will keep you posted on developments as and when they happen. In the meantime, we ask that you:
SAVE THE DATE - 31 August 2024

Gastrointestinal Symptoms in Marfan Syndrome

The correlation between Marfan syndrome and gastrointestinal symptoms is little known and under-researched. On Friday 15 September, Professor Aziz comprehensively demonstrated the association between joint hypermobility disorders such as Ehlers-Danlos (EDS) and gastrointestinal symptoms in functional GI disorders including IBS & IBD. He further focussed on GI symptoms in Marfan and the overlap with EDS. Here's the link to his talk:**https://www.youtube.com/watch?v=e-U7qZ2C-Tk**


Professor Qasim Aziz

New Booklet Alert - Pregnancy Leaflet

Echoing the words of Consultant Cardiologist Dr Isma Rafiq from the Royal Brompton Hospital who spoke at our Information Day, there can be risk during pregnancy with Marfan syndrome. "If there is significant dilatation of the aortic root, that is high-risk during pregnancy and childbirth … the risk, and therefore the delivery options, vary depending on the extent of aortic dilatation. For women with little enlargement of the aorta when assessed, the risks of pregnancy and childbirth should be low. For those with moderate to severe dilatation, Caesarean section may be the best course". Our new booklet, Pregnancy with Marfan Syndrome, discusses this and more. It can be downloaded here: **www.marfantrust.org**

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). Please retain the barcode. These stamps can be of any description from any country. Please send to:**Mrs Pauline Moses The Waves, Coast Drive, St Mary's Bay, Romney Marsh, Kent TN29 0HN**

New Exciting Additions to our Shop:

Completing today's luxury look is the humble baseball cap, briefly doomed to the fringes of fashion but now enjoying a runway resurgence. We have a new line at the Marfan Trust that make a subtle statement. We know a diagnosis doesn't define you, but we're united on a mission to make the world more Marfan Aware & your head piece could become a conversation piece. Buy your cap today! (See page 16)

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**

The Marfan Trust Online Shop

Jeremy Wins

Tote Bag


Marfan Pen

Zest for Life


Heart Keyring

Marfan Mug


Willow

Emergency


Wristband

Pin Badge


T-shirt


Scrunchie


All Occasions


Robin Card


Three Kings


Angels


South Bank

Marfan Cap

Welcome to our shop, a trove of lovely things our shop is newly expanded with caps and mugs.

Link to website 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 Cost
Willow, the Marf Giraffe	£5.50		
Jeremy Wins Book	£8.00		
Marfan Trust Tote Bag	£3.50		
Marfan Trust Pen	£2.00		
A Zest for Life Cookbook	£6.00		
Hand-crocheted Red Heart Keyring	£3.50		
Marfan Trust Emergency Card	£2.00		
Marfan Trust Wristband	£2.00		
Hand-crocheted Scrunchie	£3.00		
Marfan Cap	£10.00		
Marfan Trust Pin Badge	£2.00		
Marfan Mug	£9.00		
Marfan Trust T-shirt	£5.50 (each)	Small Medium Large XLarge	
Marfan Trust All Occasions Card (10 cards)	£4.00		
Robin on a Snowy Branch Card (10 cards)	£4.00		
Three Kings of Orient Card (10 cards)	£4.00		
Song of the Angels Card (10 cards)	£4.00		
Snow on the South Bank Card (10 cards)	£4.00		
Postage cost	£3.00		
SUM TOTAL (including postage)			