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Marfan Matters

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

REGULAR FEATURES

A word from our Chair, Dr Anne Child

Helpline Analysis *by Victoria Hilton*

Dr Child's Casebook

Research Update *by Dr José Aragon-Martin*

Fundraising Analysis *by Gurpreet Madan*

In Memory

Feats of Fundraising

Marfan Trust Shop

SPECIAL FEATURES

Introducing our new Clinical Nurse Specialist, Joanne Jessup

Postcards from Paris: Dr José Aragon-Martin & Victoria Hilton

Daniel Finkletaub Award: Loeys-Dietz Syndrome

Dr Michael Carr Scholarship: Growth Management in Marfan Syndrome:

Marfan Information Day

Welcome to our Autumn/ Winter Newsletter 2022

From left to right Diane Rust, Marian Mason (treasurer of the Marfan Association) & Dr Nitha Naqvi

Enveloped in winter's dark embrace and with carols filling the air, we are rushing headlong towards Christmas. The year has whisked by in a busy blur, punctuated by many momentous happenings here at the Trust. Dr José Aragon-Martin and his Sonalee Laboratory have moved from Imperial College to the Institute of Ophthalmology to research the genes behind lens dislocation. Our office has moved from Chelsea to new quarters in Kent. We have hired a cardiac nurse specialist, Joanne Jessup, adding an invaluable dimension to our helpline. We are publishing our helpful and hopeful children's book, "Jeremy Wins", woven throughout with the problems and solutions encountered by Dr Child's many paediatric patients. The world is now largely unlocked after coronavirus and the Marfan Trust continues its unstoppable march. In the meantime, however, we pause with great sadness to report the death of Diane Rust. Irrepressible founder of the Marfan Association, Diane provided support for Marfan folk when there was none and raised awareness of the condition when it was so little known. Her legacy is indelible (see page 10).

A Word from our Chair

Dr Anne Child proudly holding her proof copy of her new book for children, "Jeremy Wins"

2022 is the Year of the Children at the Marfan Trust, as reflected in our Information Day topics, school problems and solutions, and books recommended for children and parents to read together, explaining being different, and building the child's confidence. Using my own experience of advising children how to cope, I have written a book for 5+ year olds, in which our hero Jeremy triumphs over problems to live a happy, healthy life. It is dedicated to Mrs Diane Rust, former Chair of the Marfan Association, who sadly died this year and will be remembered fondly by all who knew her.

"Jeremy Wins" is now available (see back page for shop orders), along with handcrafted items and Christmas cards. All proceeds go towards our programme. There must be many talented readers who are 'crafty'. We would be delighted to receive any donated creations for our shop.

It has been a very productive year, as seen in our features. Staff attended the 'Science in Paris' meeting to hear about international research news regarding Marfan syndrome and other connective tissue disorders.

Membership has increased, largely due to social media articles, and donations are returning to pre-Covid levels.

Every penny is put towards our programme of support, education and research. Thank you all.

We welcome our new staff member, Joanne Jessup, a cardiac nurse, who will help Victoria Hilton, our helpline and communications officer, to answer increasing requests for help and information.

Our research programme, directed by Dr José Aragon-Martin, concentrates on discovery of new genes for thoracic ascending aortic aneurysms, and for dislocated lenses so that these conditions may be separated from Marfan syndrome, and receive correct diagnosis and management.

We thank our volunteers and fundraisers for their most valuable work. We are still a small charity achieving great things, with your donations. Together with you we look forward to further success in 2023.

Merry Christmas and Happy New Year to all!

Anne H Child

Anne H Child MD FRCP
Medical Director and Chair of Trustees
Marfan Trust

Helpline ... A Lifeline

by Victoria Hilton


Victoria Hilton


Dr Anne Child

Continuing the momentum created during lockdown, our helpline is increasingly busy as Marfan syndrome becomes progressively better known. In a cluttered world of competing causes, we strive to bring MFS to the consciousness of the public and professionals alike. This seems to be working, with the prevailing enquiry to our helpline emanating from those wondering if they, a friend, or a family member, has the condition.

The pattern and proportion of our hotline calls was perfectly typified recently during a week in October. Of the ten calls received over seven days, three were from those who suspected themselves (or it had been suggested by a GP or cardiologist) that they or their child had MFS. They came to us seeking diagnostic certainty. A further three called to discuss matters of the heart with one seeking an alternative to their medication, another hoping to move hospitals and a third worried about treatment of arrhythmia. Separately, a supporter rang to discuss long-term debilitating gastric complications for which her medication had been delayed by administrative problems. Another anxiously awaited news on an operation for his pectus excavatum which was painful and inhibited his breathing. A supporter wondered whether to declare Marfan on their job application, and finally, a parent worried for their child's musculoskeletal pains and rapid growth spurt. We have taken 290 calls so far this year and in other weeks there have been many enquiries about eye problems. Several supporters have sought advice on the merits of intraocular lens implants, and on where to go or whom to see for a second opinion or surgery. Our helpline is adapted weekly into Dr Child's Casebook, an edition of which we publish below: On the Spectrum.

In response to the increasing demand on our hotline, we've hired a clinical nurse specialist who has already added a new dimension. Joanne Jessup arrived on 1st November and has hit the ground running!

As you all probably know, our peer-to peer-support is expanding and we'd love you to join the conversation. As our community of supporters grows, the need for support correspondingly increases and we do need willing mentors to help their fellow Marfan folk. Email info@marfantrust.org

Casebook Special: On the Spectrum?

by Dr Anne Child & Victoria Hilton

Detecting patterns and drawing connections is a natural way of navigating life and making sense of the world. But sometimes these connections, though seemingly absolute, are faulty. Several in our Marfan community have drawn a link between autism and MFS, but does one exist? Dr Child has a considered opinion.

Question: Parents on my Marfan Syndrome UK Facebook site have again brought up the question of the possibility of a link between Marfan and autism and I wondered if my physician's research had helped to link the two or is it too early to say?

Answer: In my experience and in the medical literature there is no reported increased frequency of autism in Marfan syndrome. You must remember autism is very common in the general population. Autism is defined by the National Institute of Mental Health as a condition "affecting how people interact with others, communicate, learn, and behave". Although autism can be diagnosed at any age, it is described as a "developmental disorder" because symptoms generally appear in the first two years of life.

Autism affects up to 1 in 100 people and the incidence of Marfan syndrome is 1 in 3000 people. There will therefore be those within the Marfan community who have both.

However, there is a definite link between autism and Ehlers Danlos (EDS) syndrome- a similar Collagen deficiency (not fibrillin deficiency).

So much so, that if a patient has Marfan syndrome and autism, I would look at the diagnosis carefully to make sure the Marfan diagnosis is not Ehlers Danlos instead. Is there a fibrillin mutation? If not, the diagnosis may well be EDS not Marfan.

Introducing Joanne Jessup

I am excited to be joining the Marfan Trust as a Clinical Nurse Specialist (CNS) and hope that I can add to the excellent work already being done by the charity.

I qualified as a nurse in 1998 and worked at King's College Hospital in London. Most of my work has been in the Cardiac speciality. I spent several years caring for patients following different types of heart surgery, including interventions to the aorta.

I moved from the wards into research and again spent many years helping to coordinate and run clinical trials in the Cardiac Department at King's. Clinical trials and research are so important in allowing us to provide evidence-based care for patients and continually improve the medications and interventions we can offer.

Recently, I have continued my nursing career as a CNS working at the Royal Sussex County Hospital in Brighton. I joined the team there in 2019 and helped to set up the Aortopathy Service as well as looking after patients with Congenital Heart Disease (heart conditions that patients are born with). In the aortopathy clinic I was able to coordinate aortic screening and follow up for patients who required monitoring for all different reasons, including connective tissue disorders like Marfan Syndrome, Loeys-Dietz and Ehlers Danlos.

I am married and have a son aged 15 and a daughter who is 12. We have travelled a great deal due to my husband's job and it's something we all enjoy doing. Both our children were born in South Africa where we spent several years and we also lived in Bahrain, both great experiences. In my spare time I love to walk, read and bake.

Research Report

by Dr José Aragon-Martin, PhD

Laboratories are portable and adaptable, settling quickly into new environments and circumstances. The Sonalee Laboratory has moved to the Institute of Ophthalmology, giving Dr Aragon-Martin a new focus as he explores the genes behind dislocated lenses (ectopia lentis). José "would really like to thank Professor Mariya Moosajee for hosting me in her Molecular Genetic laboratory at the Institute of Ophthalmology, the research arm of Moorfields Eye Hospital".

I am working simultaneously on three projects outlined below.

1. Marfan syndrome

We are creating a neonatal Marfan syndrome UK database along the lines of the French international database for the gene fibrillin-1, which causes Marfan syndrome. This will help us correlate the clinical presentation of neonatal, infantile and classical Marfan syndrome patients who all have mutations in the central exons 24-32 of this gene. Rohan Bhupal, medical student at Imperial College, is helping me with this project.

2. Thoracic Aortic Aneurysm and Dissection

Our discovery of a new gene for TAAD, named LMOD1, has been accepted for publication. We continue to analyse the NGS (gene sequencing) data from 40 families with unexplained inherited aortic aneurysm. We are applying for a British Heart Foundation grant to continue this work in collaboration with Imperial College and Queen Mary's College, University of London.

3. Ectopia Lentis

We continue to analyse Ectopia Lentis families, small and large, for a second and third new gene. This will help us to separate these families from Marfan syndrome families at the time of diagnosis, as they do not need cardiac follow-up.

Thank you

We would like to thank all donors who have specifically requested that their funds be used for research.

Special thanks to the Aortic Dissection Charitable Trust for their generous donation towards my research programme.

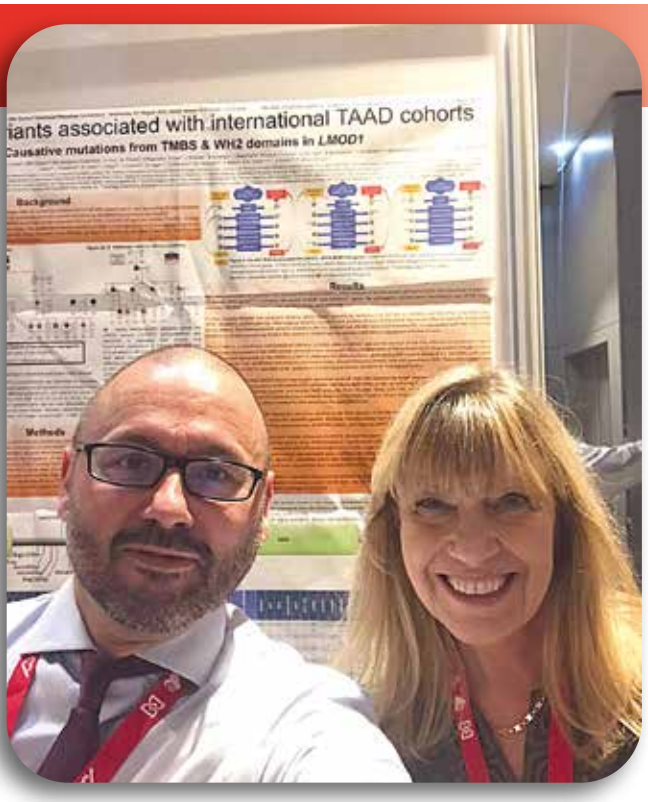
Science in Paris

This is the third year of the pandemic and the restrictions have relaxed, allowing me to attend the Marfan Foundation's conference in Paris.

The 5th scientific meeting on Ehlers-Danlos syndrome and the 11th International Symposium on Marfan syndrome and Related Conditions occurred together in August. I presented my work on a new gene for aortic aneurysm called LMOD1 as a poster presentation. This has been accepted into the aortic aneurysm screening panels by

Professor Dianna Milewicz in Texas, and Professor Bart Loeys from Antwerp, as well as the Brompton Hospital Screening Panel. I was able to meet and talk with collaborators from around the world, I was also able to present a poster for a London colleague who could not attend. The topic was a sub-study of the AIMS trial of Irbesartan in Marfan syndrome.

Rohan Bhupal (one of our summer students working with the Marfan Trust) and myself were given the topic of Neonatal Marfan syndrome. There is currently great interest in the classification, health problems, and gene mutations in this most severe group of patients diagnosed at birth. I am collaborating with new planned international studies, and am also planning a UK collection database of all neonatal cases.



José with Professor Dianna Milewicz

This conference provided increased awareness amongst the international community of the work of the Marfan Trust, and gave me an opportunity to network, and discuss present and future collaborations to speed better treatment and understanding of Marfan syndrome.

Summer Students

by Rohan Bhupal

I joined this summer on my summer studentship at the Marfan Trust and started working on my project set on Neonatal Marfan Syndrome.

This is a devastating variation of Marfan syndrome and has a very poor prognosis. This means children often die at a young age and for this reason it is a rare, uninherited form of the disease. Due to the rare nature of the disease, neonatal Marfan syndrome (MFS) has little research and background literature, I wanted to collect what was available to review what is known and identify further areas for future research. My research involved collecting information about the diagnosis, epidemiology, management and future treatments of this disease, using reports of patients with the condition from the last decade. My research also involved collecting mutations in the FBN1 gene that caused neonatal and classical Marfan Syndrome, specifically in the neonatal region of the gene located (exons 24-32). I then used this information to create a domain map of mutation in this central area of the gene. I have received new information which has meant I will now extend this project to the end of the year to complete a full review of this condition.

Postcard from Paris

by Victoria Hilton
Helpline and Communications Officer

I was propelled onto the genetic frontier during the Marfan Foundation one-day patient conference in Paris. It was a molecular landscape of signalling pathways and vascular interfaces, with the emphasis on science but also exercise. Covering all but the eyes in Marfan syndrome, the symposium was bookended by presentations from geneticists Drs Bart Loeys and Hal Dietz and I watched as several attendees met these namesakes of their connective tissue condition.

From living in a largely virtual zoom-bound world it was a real thrill to meet people in person, including **Eileen Masciale** of the Marfan Foundation USA, and Marfan patient **Gareth Owens**, Chair of the national patient charity Aortic Dissection Awareness UK & Ireland and leader of the global THINK AORTA campaign. My notes are below.

Genetics of Aortic Conditions
Professor Bart Loeys, University of Antwerp
Of thoracic aortic aneurysms, 20% have a family history, and 5% a syndrome. Most are autosomal dominant, with a 50% risk of passing on the condition. There are 38 known TAAD genes, with the most genetic variations having a high degree of non-penetrance (the person carries the gene but it has not expressed itself). Each new variation only explains a small proportion (1%) of TAAD patients. The known genetic variations explain 30% of familial TAAD and there are three pathways. Professor Loeys' research group have discovered four new genes for TAAD, dubbed variously 'an old friend seeking a new relationship', a 'new connective boyfriend', 'connective cousin' and 'a new girlfriend for connective tissues', which interact with the fibrillin pathway.

Heart Issues
Dr Kim Eagle, Consultant Cardiologist, University of Michigan
Dr Eagle discussed what to expect on the medical landscape in the next 5-10 years. Better imaging resolution, more precise genetic risk prediction, better, less invasive intervention for aortic and valve disease, improved understanding of treatment options and preventative strategies.

Knowledge is increasing exponentially through research. Physicians can make precise predictions of aortic behaviour based on knowledge of the gene. In the meantime, Dr Eagle stressed the benefits of exercise.

Exercise: What Can I Do?
Dr Alan Braverman, Washington University in St Louis School of Medicine
Precautions vary depending upon the individual's condition and systems affected. It has become increasingly important to exercise, even if this only includes movement. Marfan mice who are kept without exercise show aortic walls which are fragmented, while the aortic wall structure of mice which take dynamic exercise is improved and the wall strengthened. This is with dynamic isotonic exercise which lengthens muscles, as opposed to isometric exercise which contracts the muscles. A pilot study of the effects of moderate intensity exercise on children and young adults with Marfan syndrome deemed it safe.

Because higher blood pressure which occurs with exercise leads to greater aortic wall stress, it is recommended to limit activities which require extreme or maximal exertion at peak capacity. Patients can freely jog, run short distances, swim, play badminton, clean, mow lawns, play doubles tennis or table tennis, and golf, or pursue yoga, and water aerobics. They should favour non-competitive dynamic exercise and avoid contact sports and straining or pushing.

Developing a health physical lifestyle

- Discuss with your doctor about specific health concerns that may impact exercise prescription.
- Perform activities that you enjoy and that do not lead to pain.
- Make a plan to perform low to moderate aerobic exercise for 30 minutes 5 days a week.
- If you can talk in a conversational voice during the activity it is generally considered safe.
- Don't worry about a target heart rate during exercise.

Hormone Replacement Therapy
Dr Juan Bowen, The Mayo Clinic
The question of whether women with Marfan syndrome should be offered HRT for menopausal symptoms is unresolved. Dr Bowan said he would treat Marfan patients as he would his other patients, and that they can take it for 5 years. Dr Bart Loeys commented that 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 levels during pregnancy. The patient should always discuss taking this medication with their cardiologist, since some patients taking anticoagulants should not take HRT apparently. Dr Child adds that patients must continue to have their annual echocardiograms whilst taking HRT, in order to pick up any increase in aortic dilatation due to this new medication.

The Daniel Finkletaub Prize 2022

Recipient: Amy O'Reilly

To commemorate their beloved son and brother, Carole and Elaine Finkletaub created a student prize in his name. Daniel died in 2009 at just 41 of complications from Marfan syndrome. Although his life ended prematurely, it was a life lived fully. He worked hard, completing two degrees and rising through the ranks of corporate marketing, but he also 'played hard', travelling extensively in his spare time. Daniel met Dr Child in the late 1980s and joined the Trust's Board in 1991. He made an indelible contribution to our Charity and continues to do so through his legacy.

Winner: Amy O'Reilly

As our Trust expands we want to make new connections - parallel connections. A member of the connective tissue disorder family, Loeys-Dietz syndrome (LDS) converges with (and diverges from) Marfan syndrome in its clinical features. Some of our existing supporters were misdiagnosed with MFS and subsequently diagnosed with LDS. Amy O'Reilly worked solidly over the summer setting the foundation and framework for a parallel support group within the Marfan Trust for LDS patients. A medical student at Bristol, Amy wrote a soon-to-be published leaflet on LDS and has created an LDS database of supporters within the UK. She has set the scene for this exciting project and is a very worthy winner of the Daniel Finkletaub Award.

Dr Michael Carr Memorial Scholarship 2022

Winner: Dr Sam Sussmes, Foundation Doctor

Our former Chair, Dr Michael Carr, died in 2020. Instrumental in seeing the Trust smoothly through some critical changes, Dr Carr was much admired and liked. His specialist training in academic inspections (Ofsted) made him uniquely qualified for leading the Trust's expansion into a new research programme and during his tenure, six PhD and 15 BSc/MSc students graduated successfully in projects that focused on Marfan syndrome and related disorders. Suffice to say Dr Carr himself had MFS. To memorialise his legacy, Dr Carr's widow Jenny has created an annual scholarship in his name. Dr Sam Sussmes is the award's first recipient and, in a nice coincidence, he graduated from Oxford where Dr Carr lived and worked. Over the summer, Dr Sussmes explored one of the Trust's most oft-asked questions, how to manage the growth of Marfan children.

Growth Management in Marfan Syndrome

by Dr Sam Sussmes

Concerns

Characteristically Marfan patients are tall and thin, with long limbs. Excessive height can bring many day-to-day problems, such as shopping for suitably sized clothes or shoes, struggling to fit into public transport and airline seating. Clinically, there is specific concern regarding the most serious complications of Marfan syndrome affecting the heart and major blood vessels. There is a worry that the extended period of growth in childhood associated with being tall may further weaken the aortic wall, increasing the risk of aneurysm.

Can anything be done?

Currently there is no formal guidance, recommendations or consensus within the medical profession. Two strategies are suggested, namely treatment with sex hormones to shorten the pubertal growth spurt, or surgery.

Side effects of hormone treatment include headaches, weight gain, aggression, and in females recent reports of reduced fertility in later life. The alternative treatment option is called epiphysiodesis. This is an orthopaedic surgery technique in which the knee growth plate is operated on to stop growth. The technique is minimally invasive, without the need for open surgery. The greatest results occur when this operation is performed early in the pubertal period.

In later life, a short length of bone can be removed from the leg, which also corrects the disproportion between leg length and trunk length.

What does this mean practically?

The child must be referred by a GP or paediatrician to a paediatric endocrinologist when the child is 150cm in height, if there are concerns about height or rate of growth or predicted final height. After a thorough discussion between child, parents and clinician, and with the endocrinologist's prediction of final height based on bone age x-ray, a decision can be made. If the parents have kept annual height measurements at home, child standing against the wall without shoes, at annual intervals for example on the child's birthday, this will greatly help the endocrinologist to predict final height.

Conclusion

The results of growth restriction through hormone therapy or orthopaedic surgery are most striking when intervention occurs early and the process is explained thoroughly to parents and child. Referral to a paediatric endocrinologist is key to success.

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Fundraising Update | *by Gurpreet Madan*

2022 has been a fruitful and exciting year for the Trust. New energetic Trustees have joined our board, we've hired a nurse to support those with Marfan syndrome and held a very successful conference and 2 webinars.

Some Key Highlights since our last Newsletter:
We held our most popularly attended Marfan Information Day and our first ever Eye in Marfan syndrome webinar, details on page 15.

As you are aware, we receive no government funding and in order to continue the work we do, we rely on your goodwill and support.

Can you give us a hand?

- **Attend our Events**
  Lend your support and partake in Marfan events – whether that's attending our seminars, conference or virtual events.
- **Donate, donate and donate**
  Donate to the Marfan Trust– this can be a one-off donation or a monthly standing order.
- **Endorsement**
  You can support us by tapping into your network and recommend us to your Company as their Charity of the Year partner; or why not speak to your Child's school about Fundraising for us – we have packs ready!
- **Collection Boxes**
  Holding an event, or know of a business who will keep our collection boxes in their store? Please email info@marfantrust.org to request a box.
- **Ad space**
  Ask businesses to take out an ad space in our Marfan Matters magazine and website. For more information, email Gmadan@marfantrust.org.
- **Help us spread the word**
  Post on your social media accounts with the hashtag #MarfanTrust or alternatively send stories/videos of how Marfan Trust has helped you to info@marfantrust.org.

With your support, we can make a massive difference to the lives of those affected by Marfan syndrome and their families.

As 2022 starts to appear in our rear-view mirror, we must start planning for 2023. Our future plans include:

- Drafting more trust and grant applications for funding
- Speaking to businesses about advertising in our Newsletter
- Applying for charity of the year partnerships
- Membership review
- Planning for conference & webinars
- Advertising our new will-writing programme

If anyone in our community has more ideas or suggestions, please don't hesitate to email Gurpreet Madan at Gmadan@marfantrust.org. We are always keen to improve our level of support to those affected by Marfan syndrome. Thank you for supporting The Marfan Trust.

In Memory

Diane in her first Marfan Association office

Diane Rust Remembered

With heavy heart, the Trust announced the death of Diane Rust on Friday, 14 October. Diane was the vibrant founder of the Marfan Association which became the lifeline for many Marfan folk. She and her team provided support when there was none, and raised awareness of our condition when it was so little known. Diane's legacy is long and lasting. Tributes poured in when her death was announced. We pay our own tribute here.

by Dr Anne Child & Val Greatorex

Diane Rust was a remarkable woman, who achieved a tremendous amount. We all owe her a debt of gratitude. Working with her as a Trustee for the Marfan Association since the early days, her drive and determination to make Marfan Syndrome (MFS) better known to the outside world, including the medical profession, was infectious. She encouraged us to give our all, yet we could never match the effort and enthusiasm she personally gave towards it.

Diane worked tirelessly in supporting those with the condition and their families, as well as conveying the importance of early diagnosis and treatment by the medical profession. A network of support was set up around the country and telephone help was always available. Research into MFS was a high priority and fundraising enabled many aspects to be explored.

Our gratitude to Diane is immeasurable and her legacy will be that the words MARFAN SYNDROME are now in capital letters in the consultant's Bible.

'TOGETHER WE CAN' was Diane's slogan, and she was right; the proof is that her work is being admirably continued by THE MARFAN TRUST

by Dr Nitha Naqvi

Diane was a truly special inspiring dynamic woman. She was a magnificent advocate, totally committed to making lives of those affected by Marfan syndrome better. Each time I met Diane, I was always struck by her wish to do whatever she could to support healthcare professionals to work with Marfan syndrome. Diane gave enormous support to families for so many years. Her knowledge of the medical aspects of Marfan Syndrome was impressive and she enjoyed interacting with expert speakers and the audience at medical lectures on the condition. We loved having Diane with us at our education days in Chelsea. She taught doctors and nurses about the patient's perspective. Her warm smile was infectious, and Diane always lit up the room with kindness and warmth. She will be missed by so many. Rest in peace Diane.

In Memory

Sue Beer | *by her sister, Margaret Turnbull*

Sue was the wife of Mr Don Beer, a past chairman of the Marfan Trust. Sue's sister, Margaret, pays tribute to her here.

Sue, born on March 24th 1949, was 3 years older than me. She was the clever, sensible big sister who took this responsibility, indeed all responsibility very seriously. A memory that she has never allowed me to forget was when I first started school, her school, Rice Lane, in Liverpool: Sue had just moved up to the Juniors and to my dismay I found that my classroom was in a separate building, in the Infants Department, and that even at playtime her playground was separated from mine… and by big iron railings! I'm told that I would stand there forlornly every morning playtime and cry until (reluctantly) she would abandon her friends to come and… hold my hand through the bars… In truth, she has been holding my hand ever since!

As children, we sisters Sue, Pam and I, had a very sociable childhood. We really enjoyed being part of a close, extended family of kind, caring grandparents, aunts, great aunts, uncles and cousins.

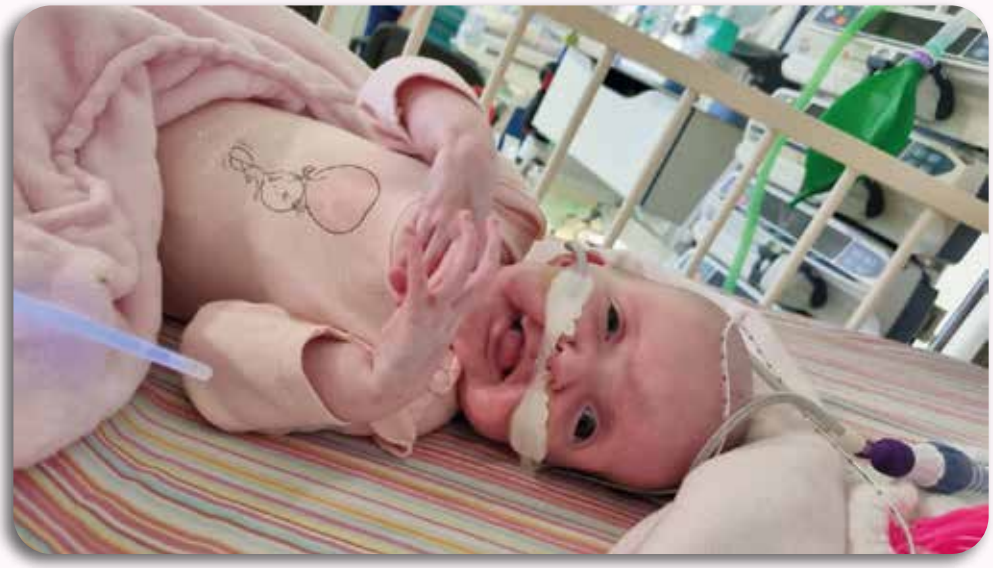
Sue was a very bright child. Despite the lenses on her eyes being dislocated, therefore making focussing really difficult, she had actually learned to read independently at the age of 3, beginning at that early age what was to become her lifelong love of reading. Sue was a very quick learner (yet a VERY impatient teacher!). She was conscientious and really enjoyed learning and academic work. Sue chose not to go to university, instead, after A levels, joining NatWest Bank in Liverpool and eventually moving to work at the bank's head office in London, where she also bought her first home. John Bealer, who had worked with her in London and knew her well, said that Sue was highly thought of and well respected by all of her colleagues, many of whom became lifelong friends.

Marriage, motherhood, new family and Marfan. Sue met Don in London and readily embraced his family. Sue and Don married in Liverpool, and in 1983 produced their much-loved, only child, Nicholas. It was around this time that Sue and Don became very involved with the Marfan Association. Supported by geneticist Anne Child, Sue researched the condition thoroughly. She determined to keep herself informed of any new developments with regard to Marfan and to seek the very best care for Nick as he grew up…and as time went on, also for her grandson, Ronnie. Sue volunteered her help in many ways for the Marfan Association (now merged with the Marfan Trust). Always appreciating the importance of medical research, she willingly participated in the Marfan Trust's continuing research programme.

Sue was always willing to share her experience and understanding of Marfan and readily joined the Trust's support days for patients. Her friend Val, who also has this syndrome said, "I have never known anyone like Sue and I doubt I'll meet anyone like her again." She said that Sue was so experienced in managing and coping with her own symptoms and was so knowledgeable about Marfan generally, that she was the ideal person to speak to anyone newly diagnosed with the syndrome. Although Sue's own health had been in decline for some years, this never broke her amazing spirit or her sheer determination to get well as quickly as possible and to lead a normal life. In hospital, even in ITU and sometimes barely conscious following lengthy operations, she was hugely courageous, always polite, always obliging, keen to never seek attention, to never make a fuss. And afterwards when returning home, she would play down ongoing, (frequently severe) pain and debilitating issues, greeting visitors or people who phoned with genuine delight and interest in their lives, in their health.

I rather think that in time she will not be remembered by anyone for her suffering but for the pleasure and helpful support she continually gave to all. Caring wife, mother, grandmother, in-law, godmother, cousin, aunt, great aunt, niece, sister, friend, Sue took keen interest in us all. Her greatest joy that I recall was when Nicholas and her much-loved daughter-in-law Rose produced Ronnie, her grandson. Sue so loved this little boy and delighted in spending time with him, Fortunately, now aged 5, it's very likely that Ronnie's happy memories of her are now permanently fixed, and will never be forgotten. As her lovely, treasured friend Zandra once whispered at Sue's bedside in hospital, her life again hanging by a thread, "You simply must get well, Sue……Whatever would we all do without you?" She will be so missed.

In Memory



CALI MOWBRAY

Cali was born 23 November 2021. At just a few weeks ago, Cali was diagnosed with neonatal Marfan syndrome and remained very poorly throughout her tragically short life. Before her daughter's birth, Jay had encountered Marfan syndrome before but never knew about neonatal Marfan syndrome. She approached our Charity over Christmas to discuss the syndrome and its implications with Dr Child. Thereafter Jay fought tirelessly for Cali, often coming up against a frightening lack of medical awareness of Marfan. Sadly, Cali died in July 2022. She is survived by her parents and siblings who wear Marfan Trust t-shirts to school hoping to make the syndrome better known.

Feats Of Fundraising

Testing the limits of strength, stamina and soul, the Marfan Trust fundraisers have run international marathons, walked many steps, stepped into the muddy unknown, braved the shave, cooked up a storm and then some. Here we celebrate several of their intrepid feats:

Lucy Atkinson Continues to Crochet

Left in limbo while awaiting a diagnosis as her symptoms worsened and her anxiety mounted. Lucy spent months if not years trying to establish the cause of her Marfan-induced gastric symptoms and withstood many attempts to dismiss her problem. She finally has a diagnosis and will get the medication she needs. During her long wait for an answer, Lucy has continued unstintingly to raise funds and awareness for the Marfan Trust, creating lovely items for her regular treat-filled raffles including this cute blanket, pictured. Thank you, Lucy!

Jacqueline Borg Stepped Forward

During April many supporters Stood Tall with the Trust and collaborated on our in-house fundraising campaign. Revolving around the number 15 to represent the Missing 15,000 (those 15,000 in the United Kingdom with Marfan syndrome who are unknown to the Marfan Trust) our campaign was interpreted in different ways with some supporters running 15,000 steps and others like Jacqueline walking 15,000 steps per week. Jacqueline raised over £300 for the Trust. *Thank you*

Feats Of Fundraising

Cannonbawz Run: Fast and Furrybawz

Cruising around Scotland's magnificent North Coast, the Cannonbawz Run and their exhilarating film-themed car rallies have contributed so much to the Marfan Trust. Created in 2015 by Kris O'Neill, the charity rally held particular resonance this summer. August 2022 marked the first anniversary of Liam O'Neill's death from complications of Marfan syndrome. Kris and his brother Liam were very close, bonding over films including "Star Wars", "Smokey and the Bandit" and anything with Arnold Schwarzenegger. Liam was the inspiration for the Cannonbawz and in their August escapade, The Fast and Furrybawz, Kris and his team raised over £3,000, in his memory. *Thank you.*

Tim Canham: Running for a Friend

Woven through life like a beautiful thread, a firm friendship surmounts bad dress sense and stubborn behaviour. Tim ran the London Marathon in memory of his best friend Martin Briggs, an original thinker, a bad dresser, and an argumentative sort who would be turning 60 this year. "Martin and I are part of a group of friends who trace their friendship back to being aged 11 at St Ignatius, a North London comprehensive run by the Jesuits. We all grew up together, went to school, shared holidays, adventures. Got married, had kids. And amazingly stayed in touch. But more than in touch, we remain a very close group. During this time, I became more aware of Marfan syndrome, how it affected my best friend Martin, and how it had taken Martin's dad and sister. Martin was possibly the brightest of our school group (which included some really clever friends that made Oxbridge) and he started his career as a doctor. Tragically, Martin died at just 42 after a heart transplant that had given him a new lease of life. My "plod" around London was about offering something to mark his life, make a contribution to the ongoing battle with the syndrome. And raise some money to assist the research and support." Tim's 43.1km "plod" around London took just 4 hours and 13 minutes, and raised over £2,500.

George Spreads the Word

George has Marfan syndrome and regularly speaks publicly in his home town on the condition, spreading the word. He spoke recently to a large audience over the summer and we are very grateful for him.

Darren Kempson Ran for the Marfan Trust

"In June, I ran 50km to support the Marfan Trust. Following the death of my father in law, my wife was diagnosed with Marfan syndrome and I feel that not enough people are aware of the condition. I work for National Grid and they very generously matched my fundraising. It was an exhilarating run and here I am with my children, proudly wearing my medal."

Feats Of Fundraising

Running for Jamie | *by Sharon Lister*

Jamie and family

"I am running the marathon in memory of Jamie Morton and to help raise awareness of Marfan Syndrome. I met Jamie's wife, Christie, back in 2005 when she was my hairdresser.

As with all hairdresser relationships you share lots of personal details about your lives and we soon became well acquainted with each other and I would look forward to each hair appointment to catch up on daily life. I remember sitting with foils in my hair one day when Christie told me all about her new boyfriend Jamie and her face lit up. It wasn't long before wedding bells were ringing followed by two beautiful children called Archie and Eva. I would often see Jamie when I was at their home having my hair done when he would get home from working on his farm and the children would be so excited to see him come through the door. Everything seemed to be picture perfect until the year of 2020 when the pandemic hit and Jamie started to suffer chest pains. After many visits to hospital it transpired that Jamie had suffered an aortic dissection. Unfortunately, after open-heart surgery and seeming recovery, Jamie died. Just before he died, tests were taken to determine what had caused this problem in a seemingly healthy man. The results of the tests introduced Jamie and his family to Marfan syndrome. I am running the Manchester Marathon to raise awareness of the syndrome".

A Tale of Two Sisters | *by Lana Predmadasa*

At the beginning of 2022, I decided I wanted to run a marathon. I knew, if I was going to be running 26 miles anyway, I wanted to raise money for a good cause. The Marfan Trust is a cause close to my heart. My sister was the first in our family to be diagnosed with Marfan. Prior to her diagnosis, I had never heard of Marfan (although maybe not so surprising as I was a teenager). My mother had only heard about it because a colleague's daughter had some symptoms and a doctor had mentioned it as a possibility to her. After her diagnosis, we found out that neither of my parents had the gene. The genetic testing of first my sister and then both my parents is a process that took several years. For me, running a marathon is something I have always wanted to do, but I never thought was possible. At the beginning of 2021, I was obese, I hadn't run since I was a teenager… and here I am in 2022, running a marathon crossing the finishing line in Amsterdam! " Lana raised a fortune for the Marfan Trust." *Thank you!*

Helen's Muddy Marfan Madness

They say charity begins at home and so it does for our Trustee, Helen Syms, who faced her fears and stepped into the muddy unknown. Helen bravely ventured from her comfort zone and into a world of mud and obstacles, taking on the Nuts Challenge, a five-mile obstacle race that was cold and wet, all for the Marfan Trust! Upon completing the challenge (which was not for the faint-hearted), Helen and her Marfan Trust t-shirt were completely unrecognisable. Whilst Helen was able to clean up afterwards her t-shirt stubbornly refused to be restored after four washes and stayed a permanent mud colour. Helen is our wonderful Trustee whose son Matthew had MFS: "My amazing son Matt had Marfan syndrome and passed away in 2017 after catastrophic complications from a planned corrective heart surgery. I am now a trustee of the charity to contribute to the efforts, supporting families, spreading awareness and education, and conducting life-saving medical research." Helen raised over £1,300 in this latest adventure, an amount doubled by her employee matching scheme making almost £3000 in total.

Marfan Information Day

Members of the medical elite once again converged virtually for our annual Marfan Information Day on 8 October. We cut straight to the heart of the matter with Tal Golesworthy discussing his pioneering invention, PEARS, (personalised external aortic root support) which is spreading across the world as we speak and was recently used to treat a 3-year-old patient in Australia. Tal took the audience through his invention's genesis, its application, and recent statistics. The conference then returned to the beginning of the Marfan journey with genetic insight from Dr Fleur van Dijk who discussed the who, how and when of testing. Thereafter the full Marfan journey was comprehensively explored as vitreoretinal surgeon Mr Ben Clarke led the audience through surgical solutions to ectopia lentis and detached retinas while Dr Fariborz Neirami talked on The Science of Pain and how to manage it. Educationalist Patricia Pothecary advised on improving the school experience for Marfan children, Dr José Aragon-Martin provided insights on his cutting edge research and Dr Anne Child gave tangible tips on combating feet and ankle pain and dural ectasia. The Information Day was recorded in its entirety and the individual videos are available for £5 each.

The Eye in Marfan Syndrome - Webinar

Our window on the world, the eye is the body's smallest organ with the fastest-moving muscles, opening up a Technicolor vista of one million different colours. In Marfan syndrome the eye can be weakened and at risk.

Consultant Ophthalmologist and Vitreoretinal surgeon Mr Aman Chandra led a cosy lunchtime webinar about the Eye and Marfan on Sunday 13 November when he took many questions and resolved many issues. Thank you, Mr Chandra! We recorded the webinar and it is available free for members and £5 for non-members.

Jeremy Wins by *Dr Anne Child*

A young boy with Marfan syndrome overcomes his problems with the help of understanding family and teachers, and triumphs by uncovering his hidden talents, leading to a happy and healthy life.

Written by *Dr Anne Child* who has woven in the real problems and solutions of her many Marfan patients, it is a heartening " how to" story for parents and children age 5 and upwards to read and discuss together.

Its aim is to encourage families to accept that, with regular care, all aspects are treatable, and compatible with a fulfilling life.

To purchase a copy of Jeremy Wins or any of the afore-mentioned webinars, scan the QR code to our shop:

CHRISTMAS SHOP | www.marfantrust.org

With Christmas comes the excitement of gift buying for which you need look no further than the Marfan Trust shop, a trove of lovely things. Newly expanded with "Jeremy Wins", a heartening and helpful book about a Marfan boy who triumphs at school and life (for children of 5+); and crocheted creations, our shop has gifts galore all of which can be handily parcelled into our new Marfan Trust Tote Bag!

Jeremy Wins

Marfan Trust Tote Bag


Marfan Trust Pen

A Zest for Life Cookbook


Red Heart Keyring


Tinsel Heart Keyring

Marfan Trust Emergency Card


Marfan Trust Wristband

Marfan Trust Pin Badge


Marfan Trust T-shirts


Marfan Trust
All Occasions Card


Christmas Card
Robin on a Snowy Branch


Christmas Card
Three Kings of Orient


Christmas Card
Song of the Angels


Christmas Card
Snow on the South Bank

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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!

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Please include cost of postage (£3) in sum total. Christmas cards - Robin on a Snowy Branch, Three Kings of Orient, Song of the Angels, Snow on the Southbank and All Occasions - are in packs of 10.

Item	Price	Quantity	Total Cost
Jeremy Wins	£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		
Tinsel Heart Keyring	£3.50		
Marfan Trust Emergency Card	£2.00		
Marfan Trust Wristband	£2.00		
Marfan Trust Pin Badge	£2.00		
Marfan Trust T-shirt	£5.50 (each)	S..... M..... L..... XL.....	
Marfan Trust All Occasions Card	£4.00		
Robin on a Snowy Branch Card	£4.00		
Three Kings of Orient Card	£4.00		
Song of the Angels Card	£4.00		
Snow on the South Bank Card	£4.00		
Postage cost	£3.00		
SUM TOTAL (including postage)			

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And don't forget feel-good shopping! Shop at smile.amazon.co.uk and donate to the Marfan Trust at no cost to you. Get started!

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