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Marfan Matters

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Spring/Summer 2023 Edition

We have slipped into summer and reached the halfway point of a productive year. It's time to reflect, reminisce and take stock, and we have been doing just that. We have looked afresh at our leaflet collection, dusting off old titles and creating new ones.

With our increasingly busy helpline come helpful new booklets on heart surgery and pregnancy in response to supporter demand, whilst existing titles are being given a modern makeover, including our Guide for Schools & Colleges, written in collaboration with our wonderful members. Visit our website to find out more: **https://bit.ly/3Oui8In**

A time to reflect, summer is also a time to refocus, and we are planning and plotting exciting happenings and events, including our next Information Day (*page 15*). This will take supporters on the full journey of Marfan syndrome from conception to old age, with pitstops in between when the spectrum of the condition will be comprehensively discussed including talks on gastrointestinal problems which has been too little researched, and scleral lenses for the eyes which are too little known.

During the whirlwind blitz of social media posts, stories and articles that was Marfan Awareness Month (*see analysis on page 12*), we quietly introduced a series of monthly drop-ins. Entirely informal and completely private, these hour-long Zoom sessions are held on the last Tuesday of every month at 7pm. Hosted by our clinical nurse specialist, Joanne Jessup, they're a chance to meet fellow supporters and discuss common concerns in a closed, confidential environment. Join in – it's good to talk and we would love to (virtually) meet you! **https://bit.ly/3pFucGq**

Dr Anne Child, Medical Director and Chair of Board of Trustees

2022 was a year devoted to Marfan syndrome children, culminating in a shared Transition Day with the Brompton Inherited Cardiac Condition Nurses, held at Chelsea Football Stadium. This popular event encouraged children with inherited cardiac conditions including Marfan syndrome and their parents to attend to discuss topics of great interest to this age group, 16-24 years (*see report on page 8*).

This year we are concentrating on both ends of life, from neonatal Marfan syndrome to patients over the age of 50 who are writing a new chapter in the life history of patients. All ages are invited to attend our open drop-in on the last Tuesday of each month, organised by Joanne Jessup, our experienced Clinical Nurse Specialist. Victoria Hilton, our Helpline Expert, also participates. Please contact us if you wish to be invited, and bring any questions with you to the webinar.

A new webinar on *Gastrointestinal Disorders* is planned in the near future. This will be the research topic for Simon Khatra, one of our summer research students, and we would like input from our supporters.

Three studentships are funded by the Girdlers' Guild, whose master, Sir Major General Sebastian Roberts, a Marfan sufferer himself, sadly passed away in March (*see In Memory, page 11*).

Another student, Elsa Harte, will advance our knowledge of neonatal Marfan syndrome, by helping Dr Aragon-Martin on this Marfan gene mapping exercise. Can we use a baby's actual gene mutation to predict which systems will be affected in later life?

Laxmi Thileepan, meanwhile, is studying the psychosocial aspects of Marfan syndrome to deepen our understanding of the psychological ramifications of the condition.

We also report an international review of all Losartan/Irbesartan medication trials, and the conclusions and recommendations (*Collaborative Trial page 7*).

Our helpline is busier than ever (*see page 3*), perhaps reflecting the problems in obtaining general practice and specialist care in this year of NHS unrest. Happily, Joanne has joined in by answering many of the questions, and writing new booklets for patients and teachers (*see page 4*).

Dr Anne Child MD FRCP

Our fundraiser, Gurpreet Madan, has launched a will-writing programme, including a plea for legacies (*see page 9*). We are grateful for all fundraising activities, and especially grateful for unexpected legacies which help us to plan special projects.

Our annual Patient Information Day has been arranged and the speakers secured. They will cover topics of interest, such as *Sports Medicine*, *Pregnancy Care*, and *Gastrointestinal Problems*. We hope you will all join us on Saturday 7th October, 9.30am to 4.30pm. You will receive an invitation in the near future, but please put this date in your diary.

We look forward to seeing you (virtually) at the conference.

Anne H Child

Dr Anne Child MD FRCP

Helpline … A Lifeline

by Victoria Hilton


Victoria Hilton

Knowledge is endless and expertise is expanding. As our helpline is increasingly sought after, with 169 calls taken between 1st January and 1st June this year, our network of advisers is correspondingly widening. We have the full spectrum of Marfan syndrome and its many manifestations comprehensively covered but there remain areas, including endocrinology and gastroenterology, that need further research.

Joanne Jessup has added an invaluable dimension to the team, hosting one-to-ones with patients, dispensing advice, offering support and sometimes simply translating our supporters' often impenetrable medical reports. Anything lost in translation over email can be so easily resolved in a simple face-to-face (Zoom) meeting with Joanne.

Arguably a barometer of the times, our helpline has seen a surge in requests for PIP (personal independence payment) applications as the cost of living tightens its grip. In response we can offer letters to reinforce PIP claims and peer help in navigating the forms. The calls to the helpline are otherwise dominated by, amongst other enquiries, the quest for diagnostic certainty. Around 25% of the enquiries are from people hoping to unify their or their child's constellation of seemingly disparate symptoms under an overarching diagnosis, be it Marfan or another connective tissue disorder. The wait for genetic testing is around six months for the appointment and a further three months for the result, which leaves patients feeling naturally impatient and worried. We've clarified the process here (scan the QR code).

Management of symptoms including those relating to the heart are also popularly received and we have printed the latest cutting-edge research on angiotensin receptor blockers (ARBs) on page 7. Some doctors remain reluctant to prescribe ARBs so it may be useful to take along this magazine, with its references, to a potentially resistant cardiologist. Guidance on pain relief is often sought after, as is respite from gastrointestinal problems. The latter is something we are exploring and discussing further in a forthcoming webinar. Finally, the Resources section of our website is a constantly expanding trove of hopefully helpful information, so please do dip in.

From our Helpline emerges Dr Child's (usually) weekly Casebook which is exclusive to our members. We're publishing a special edition below to give non-members a flavour of what to expect from joining the Trust.

Casebook: Marfan Manifestations

by Dr Anne Child & Victoria Hilton

With the (literally) thin skin inherent in Marfan syndrome come many manifestations. You are left vulnerable to bruising and bleeding, but can every mark and blemish be attributed to the syndrome, a supporter wonders.

Q: I have Marfan syndrome and recently fell, leaving a large haematoma on my leg. My nephew, also affected with MFS, came off his bicycle and suffered two haematomas on his legs. Is this a feature of MFS and if so, why?

A: Like other connective tissues disorders (for example Ehlers-Danlos syndrome) the combination of thin skin, lack of subcutaneous fat to cushion blows, and fragile blood vessel walls can lead to prolonged bleeding into soft tissues after a blow. This blood gathers and forms a pool under the skin, called a haematoma. It is not dangerous, but can be unsightly due to subsequent multi-coloured bruising. Arnica cream can be rubbed in to speed healing. Local pressure and ice packs applied at the time may limit the bleeding. Wearing protective pads on knees and legs when cycling may help prevent damage. This is not due to a lack of clotting factors, but simply trauma. Milder sports such as swimming, walking or badminton may be safer. Contact sports such as boxing or karate should definitely be avoided. As humans, we are lucky to have so many sports to choose from.

For yourself, please think why you fell. Was it a mechanical fall (trip over, lost footing) or did you feel faint, dizzy or have any heart palpitations beforehand? Have you fallen more in recent months? If you are concerned, you should discuss this with your doctor. Check footwear, don't carry heavy parcels, ask someone to help with ladder work, and when out walking, carry a stick to provide balance.

I hope this is helpful.

First Impressions *by Joanne Jessup*

Since joining the Marfan Trust in November 2022, I have been busy working with my wonderful new colleagues to continue advocating for our members and responding to their questions and requests for help.

The excellent team of doctors who make up our expert advisory committee mean we have access to up-to-date, evidence-based clinical advice from current practitioners. We have expanded our advisory panel with three further experts in their field: Dr Isma Rafiq, Consultant Cardiologist with specialist interest in Maternal Cardiology; Dr Yaso Emmanuel, Consultant Cardiologist (Congenital Heart Disease and Aortopathy); and Mr Conal Austin (Consultant Cardiothoracic Surgeon with extensive experience of PEARS surgery).

I have been working to produce two leaflets; one is entirely new and deals with the topic of **heart and aortic surgery** with Marfan syndrome. I was delighted to have lots of engagement from our members for the development of this leaflet. People were willing to share their stories and experiences and provide feedback on the final draft to ensure that the information is appropriate and relevant to our community.

Information for teachers

I have also been updating our **Information for Teachers** and again, was glad to have assistance from one of our members with a background in teaching. She also has a grandson with Marfan syndrome so she was able to draw on both her professional and personal life to give great insight into what is important for teachers to know and understand so they can help children and young people reach their full potential. Both these leaflets are going to press as we speak and will be available soon on our website.

Since working on the helpline, I have found that the best way to respond to some queries is a **'face-to-face' conversation**. Most people are now happy to join Zoom or Teams calls so I can offer this as a way of talking through some of the more complex questions. If you would like to talk, please let us know via the helpline and I can get in touch to arrange a convenient time to go through your questions or concerns.

In addition to one-to-one conversations, I have introduced a **Monthly Drop-In** session for anyone who would like to join. These started at the end of Marfan Awareness Month in February. The Drop-In is designed to be a safe space in which you can ask questions, discuss issues and meet other people with Marfan syndrome. Often, there will be people who have been through similar struggles and can offer strategies and ideas that helped them find a resolution.

If you would like to join one of these sessions, they usually take place on the last Tuesday of every month. They will be publicised in the week prior via email and social media and you will be sent a link to join via Zoom. There won't be a presentation or the possibility to answer specific medical questions surrounding diagnosis.

Professional development

It is important for any clinician to continue their professional development and I am no exception! I have attended conferences and talks by other healthcare professionals to ensure that the advice and guidance we are providing always follows the most up-to-date guidelines.

- Teaching Session run by the Network for Inherited Cardiac Conditions in Scotland about Genetic Testing for Children
- Lifetime Management of the Aorta Conference – hosted by UPMC Heart and Vascular Institute Pittsburgh

In addition to hearing from other professionals, I was incredibly grateful to one of our Trustees, Helen Syms, for sharing her story.

Her son Matthew had Marfan syndrome and tragically died at the age of 17 following cardiac surgery. Helen talked openly and honestly about the constant battles she fought to get Matthew the support and care he needed throughout his life and the effect this had on him, her as a parent and on his sister.

Hearing a first-hand experience of the daily decisions that need to be made, from getting the right glasses to deciding which sports to do as a family, to campaigning for the adaptations Matthew needed at school to fulfil his potential, I have a much deeper understanding of the condition and how it can affect all members of a family, even if only one member has it. Thank you, Helen.

Plans for the next few months

- Create two new alert cards based on patient feedback
  - Fluroquinoline antibiotics
  - Post-heart surgery information and alert card
- Build a network and provide education to nurses working with aortopathy patients across the UK

RESEARCH UPDATE

Not confining himself to Marfan syndrome but extending his research to its overlapping conditions, Dr Aragon-Martin and his Sonalee Laboratory are dividing their time between two prestigious establishments, the William Harvey Heart Research Institute and the Institute of Ophthalmology. Joined by his MSc summer students, José is researching new genes and new gene changes causing a variety of disorders. Read on.

We finally moved our laboratory – the relevant equipment and consumables – to the William Harvey Research Institute at Queen Mary University of London (QMUL). Here, I am overseeing Vatsala Khurana (*pictured right*) who is studying for her MSc in Genomics Medicine at QMUL, continuing on from her medical degree in India.

Vatsala is exploring the genetics and gene changes causing unexplained and **inherited thoracic aortic aneurysm and dissection** – TAAD. Together we are working with a family, previously assessed at the Marfan Unit of St George's Hospital, to discover if they have Marfan syndrome or a similar, related connective tissue disorder causing aortic aneurysm.

I can also be found at the Institute of Ophthalmology (UCL) where again, I am exploring new genes and gene changes, but for those that underlie unexplained, **inherited joint hypermobility disorder**. MSc genomics student Silvia La Penna is running my project, and I have prepared the relevant files and script to run the programs on the Sonalee Lab iMacs.

Silvia is studying for her MSc in Genomics Medicine at QMUL, after earning a BSc from the same university. She has worked in Florence as a lab assistant on cancer research. Her fluency in Italian, German and English has proved invaluable in translating external clinical reports and journals. We are studying a hypermobility family, who have been assessed at the Marfan Unit of St George's Hospital, to discover if they have a connective tissue disorder which overlaps with Marfan syndrome.

Sanjana Babu is the third MSc student from QMUL who also is looking for an answer to inherited joint hypermobility disorder families.

Sanjana Babu

I am continuing to research **ectopia lentis** (dislocated lenses) families, small and large, for a second and third new gene at the Institute of Ophthalmology with Professor Mariya Moosajee. This will help to distinguish these families from Marfan syndrome at the time of diagnosis.

Finally, there are other exciting projects afoot this summer in the Sonalee Laboratory when we will be studying neonatal Marfan syndrome as well as the psychosocial aspects of Marfan syndrome. These were made possible by the generosity of the late Sir Sebastian Roberts and the Girdlers' Guild.

José with MSc genomics student, Silvia La Penna
Below: José with Vatsala

Thank you Derek, Elizabeth, Christine and Phillipa for your very kind donations towards this essential laptop equipment in the Sonalee Lab, to be used daily by Dr José Antonio Aragon-Martin and his MSc and medical students, in the search for new genes which overlap Marfan syndrome, and indeed mutations in the fibrilin-1 gene which causes Marfan syndrome itself.

PECTUS UPDATE *by Joanne Jessup*

After many discussions and much debate, NHS England has agreed a new approach to the treatment of pectus deformities. Dr Child joined a consortium of surgeons and physicians, engaging and lobbying for this important service development. Formerly, and controversially, NHS England has not funded pectus deformity surgery, viewing it not as a medical but a cosmetic problem. This stance has changed, and NHS England will fund treatment for the most severe cases of pectus deformity, but only after a multi-disciplinary process of assessment. Read on…

Pectus deformities are abnormalities in the shape of a person's chest wall. They are a known feature of Marfan syndrome and some other connective tissue disorders but can happen unrelated to these conditions. Pectus deformities can be present from birth, but the problem often becomes more pronounced during adolescence and periods of rapid growth.

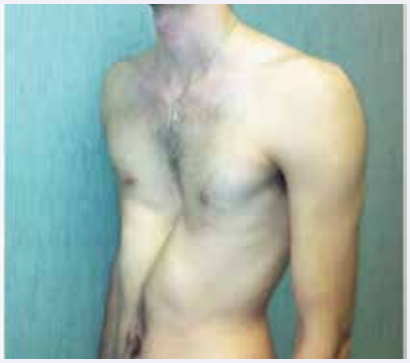
At the Marfan Trust we are often contacted by members who are struggling with symptoms but don't know where to turn. Hopefully this will start to change thanks to some new guidelines and pathways championed by a dedicated group of cardiac and thoracic surgeons who are aware of the need for treatment options.

Most pectus deformities are mild and do not interfere with daily activity. However, a small proportion of deformities are severe and can cause both psychological and physical effects. The psychological effects can include issues with self-esteem, depression, withdrawal from activities that could 'expose' the chest (dancing/swimming) and avoidance of social situations like sleepovers and trips to the beach. This often coincides with puberty and teenage years when formation of strong friendship bonds is so important.

Symptoms

A very small minority of people with a severe pectus deformity can experience physical effects due to the compression of their heart or lungs. These can include breathlessness on exercise or with minimal exertion, difficulties swallowing due to pressure on the oesophagus, heart rhythm disturbances or blackouts due to compression of major blood vessels or the pumping chambers of the heart.

To date, there has not been a unified approach to treating pectus and NHS England has not always funded the surgery.



Pectus Excavatum

However, determined work by surgeons from the Society for Cardiothoracic Surgery, Royal College of Surgeons of England, Royal College of Surgeons of Edinburgh, British Orthopaedic Association, British Association of Paediatric Surgeons, Royal College of Physicians and Surgeons of Glasgow in addition to input from our Medical Director, Dr Anne Child, has raised the profile of intervention for pectus again and there is work in progress to create a new set of guidelines for treatment.

There will be a new pathway for referrals to ensure equitable access for all patients. Importantly, in individuals with pectus who do not have a diagnosis of a connective tissue disorder, like Marfan syndrome, there will be additional examinations and referral to genetic services if necessary to rule out a genetic cause of the pectus deformity.

Referral pathway

Individuals with a pectus deformity should be referred to their local thoracic surgery service or paediatrician for review and baseline testing which needs to include the following:

- Cardiopulmonary Exercise Test (CPEX)
- Lung function test (to check for any other causes of symptoms)
- Echo scan of the heart (to check for any other causes of symptoms)
- MRI to assess Haller Index (measurement of distance between the front of the chest and the spine)

These tests give doctors quantifiable, objective measures of the pectus deformity and its effect on the body as well as ruling out other possible causes of symptoms. Once these tests have been completed, the patient will be discussed in a national multi-disciplinary meeting (MDT) so a joint decision can be made about treatment.

This MDT will be based at St Bartholomew's Hospital, London, and will consist of specialist cardiac surgeons, thoracic surgeons and cardiologists. Recommendations for surgery/non-invasive treatment or psychological support can be made.

The aim is to collect data from the MDT and from surgeries/non-invasive treatments performed to support further development of the service.

Treatment

The treatments that should be available will include:

- Surgical – Nuss or Ravitch procedures

- Non-surgical – bracing (for pectus carinatum) or vacuum Bell (for pectus excavatum)
- Psychological support

How can I be referred?

If you or a family member have a pectus deformity that is causing troubling symptoms, what do you do?

- See your GP and discuss your symptoms
- Ask for a referral to a specialist in your area who can examine you and if necessary, arrange the tests that are required
- If appropriate, your case will be referred to the MDT for review and a decision about treatment can be made by the experts in this field.

As with all surgery, these operations carry risks and need to be carried out in centres where complications can be dealt with in the event of any problems. For this reason, it may not always be possible to be treated in your local hospital.

The latest NHS England guidelines can be found by scanning this QR code.

COLLABORATIVE TRIALS PROVE LOSARTAN/IRBESARTAN EFFECTIVE IN MARFAN SYNDROME

Tried and tested medications are the prescription of choice amongst doctors, especially when long established. As and when a new (and possibly better) remedy enters the scene, it can take time to take hold and enter prescriptive practice. It's become evident from calls to our helpline that some doctors remain reluctant to administer the 'new medication on the block' – Irbesartan – preferring to stick with the traditional beta-blockers. We're republishing an article that first appeared on our website in September 2022. This is something you can take to doctors should they prove resistant to prescribing Irbesartan.
(Please note that Irbesartan is not in liquid form and therefore not suitable for children under six).

In 2022, a landmark publication in the prestigious medical journal *The Lancet* confirmed that a second class of medications named angiotensin receptor blockers (ARBs) halves the rate of growth of aortic aneurysms, and is safe to use in children and adults with Marfan syndrome. A huge task, this statistical analysis of seven randomised trials involved 1,442 patients from USA and Canada, UK, France, the Netherlands, Belgium, Spain and Taiwan. All patients who volunteered had no prior aortic root surgery.

Fifty-four per cent were female and 75 per cent were already on beta-blockers such as Atenolol. Overall 83 per cent of genotyped individuals had causative fibrillin-1 gene mutations. All patients obeyed the revised Ghent criteria for diagnosis. Patients were divided randomly into treatment or placebo control groups, in double-blind trials in which neither the patient nor the doctor knew which group the patients had been assigned to until the code was broken when the trial was completed. Treatment was well tolerated.

In this international collaborative effort, all detailed results, including those from the **UK AIMS trial** (Aortic Irbesartan Marfan Syndrome) were contributed to the University of Oxford to be analysed by Professor Colin Baigent and Dr Alex Pitcher's clinical trial team.

RESULTS: During a median follow-up of three years in which each patient had an annual echocardiogram, taking an ARB (Losartan or Irbesartan) **halved the rate of enlargement of the aortic root size. In comparing two sub-groups, those on beta-blockers and those on ARBs, the annual change in aortic root diameter was similar in both groups.**

CONCLUSION: The effects of ARBs are similar to those of beta-blockers. This provides evidence of a second class of medicines (ARBs) which are effective in delaying aortic root enlargement.

RECOMMENDATION: Patient tolerance of dosage and side-effects should determine which medication is prescribed. Our analyses revealed that the effects of ARBs showed evidence of consistent effect, whether or not the patient was already on beta-blocker therapy. We expect that the effects of ARBs would be additive to any effects of beta-blockers. **Therefore, the medications would work well together, producing increased effect**.

Treatment commenced early in life is expected to delay the need for future aortic surgery, and should be maintained life-long to protect the whole aorta. For affected children, the **UK AIMS trial** showed greatest effect of **Irbesartan** during the rapid growth years, ages six to 16 years.

Comments and questions to Dr Anne Child:
info@marfantrust.org

ROYAL BROMPTON TRANSITION DAY

by Victoria Hilton

Navigating the transition between paediatric and adult care can be tricky. It's a crucible moment and a rite of passage for many patients. The Royal Brompton Hospital makes it as seamless as possible as we experienced in February at their Inherited Cardiac Condition (ICC) Transition Day held at Chelsea FC where there was lots of curiosity and conversation around Marfan syndrome.

A dedicated ICC transition clinic is held monthly at the Royal Brompton. Run by paediatric and adult trained nurses, it facilitates a smooth passage from child to adult care by empowering teenagers, enabling them to make informed decisions about the treatment and monitoring they need. Attended by many teenagers, some with their parents, the ICC Transition Day conference opened at 9.30am with a presentation on *The Normal Heart*. Unsurprisingly the young audience had a good underlying knowledge. It continued until 4.30pm with a series of talks covering lifestyle from diet and exercise to the perils of addiction.

The latter presentation unveiled many interesting factoids: apparently there is more caffeine in a Costa cappuccino than an energy drink for which you need ID (identification), and too much caffeine isn't always good for the heart. The dietician similarly gave helpful titbits: eggs contain good, not bad, cholesterol, and you can eat several per week. Dr Sabiha Gati spoke on *Exercise* – sports to pursue and exercises to avoid when you have a heart condition. She held the audience captive and we seized the moment to invite her to speak at our Information Day on 7th October. She agreed (*page 15*)!

Dr Child arriving at Chelsea

Thank you to Bethan Cowley and the ICC Nurse Transition Team at the Royal Brompton for such a splendid day which culminated in a tour of Chelsea Football Club! And thank you to the two young Marfan Trust volunteers, Sophia and Reggie (*pictured*), for so cheerfully manning our stall and supporting our small team.

This was the first in an annual series of ICC Transition Days and we strongly urge our supporters – those aged between 14 and 19 – to attend. The day offers not only practical advice and guidance but also a chance to meet your peers and there was a lovely huddle of mutually supportive Marfan teenagers present.

For more information on addiction and the ways in which to release yourself from its grip, please visit: **https://www.hopeuk.org/**


Victoria speaking to a supporter

Volunteer Reggie

Fundraising in 2023

by Gurpreet Madan

In the current economic climate, where people's purchasing power is not what it used to be, it is truly heart-warming to see so many supporters continue to donate, fundraise and sign up to be members of the Marfan Trust. We are so appreciative of your support and would genuinely like to express a heartfelt thank you to each and every one of you.

Your support, not just in terms of monetary value but your help in creating awareness, has helped us to connect with more individuals who are either looking for a confirmed diagnosis, think they have Marfan syndrome, or suspect a friend/family member of having it. Together we have been able to make a massive difference to those affected and their families.

Some of the ways you can support us are:

■ Attend our events

Lend your support and partake in Marfan events – whether that's attending our seminars, conference or virtual events.

■ Become a Best Friend

As a paying member, you will receive our fabulous content (weekly case studies, articles, blog posts), bi-annual newsletters, four PDF versions of Marfan Trust's advice pamphlets, and discounts on our events. You will get all of this and more for a small monthly donation of £3.

■ Endorsement

You can support us by tapping into your network and recommending 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 tins

Holding an event or know of a business who will keep our collection tins in-store? Please email **info@marfantrust.org**

■ Ad space

Ask businesses to take out an ad space in our *Marfan Matters* magazine and website. For more information, email **info@marfantrust.org**

■ Wills

We've signed a contract with **The Goodwill Partnership**, a will-writing service that will visit your home to help you draft a will at a low cost. They provide a unique free home visit appointment service to all postcodes in England and Wales, which will allow people to leave a gift to the Marfan Trust should they wish to. Our new collaboration with The Goodwill Partnership offers a new way of raising funds that will ensure the Marfan Trust can continue to meet the increasing demands for our unique support services over a longer period of time. For any queries, please email Gurpreet Madan at **info@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**

■ Easy fundraising

Transform your daily shopping into everyday magic. With the abrupt closure of AmazonSmile we turn to Easy Fundraising for some feel-good shopping, where over 7,000 brands reside in one handy place.

Each brand will donate part of what you spend to a cause of your choice, and in a world of competing causes, we'd love you to choose the Marfan Trust, so sign up today! From booking a holiday to buying groceries, every little helps, as Tesco once famously said: **http://bit.ly/3ZLRKph**

> **"Alone we can do so little, together we can do so much." Helen Keller**

If anyone in our community has more ideas or suggestions, please don't hesitate to email Gurpreet Madan at **info@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

Mr David Gilhooly *by his son, Brian*

David "Dave" Gilhooly died peacefully at home on 30th March 2023 at age 75. He will be greatly missed by his family and friends. A genuine, kind and funny man.

I would like to mention that Dave's death wasn't caused by Marfan syndrome – I take some comfort in knowing that the prognosis for people with Marfan is improving. I have vivid memories of reading the Marfan newsletter as a teen and getting very upset hearing about the lives cut short by Marfan syndrome. My dad lived a full and wonderful life. I know we won't all be as lucky, but this condition isn't the death sentence it once was.

David was a lovely loyal member of the Marfan Association and Trust. He contributed to our Facebook challenges, once posting a beautiful poem in response to our Flower Power competition. We proudly publish it here.

Is It Spring Yet?

Through dimmed eyes nature's beauty I see
The season's still cold, naked trees say to me
A vanguard of shoots defy winds and frost
As daylight fights to retrieve hours lost.

The first herald of spring is not sap rising
But hopes and plans and fantasising
Muscles and joints still stiff and sore
Ease with the prospect of living some more

Buds and green growth add to my joy
Foxes so nosey just stare and look coy
Cats from afar sniff out their prey
Pigeons on branches cooing away

I was born in sunshine my mother said
A time for the living not for the dead
If I wither before rites of spring
I ask the bluebells to ring, ring, ring

Dave Gilhooly
January 2020

Tom Reveler

Unusually generous and kind, Tom was a gentle giant with a passion for sport and a love for his job. It's taken Tom's mother another 22 years, the length of her son's short lifetime, to bring herself to read about Marfan syndrome and share his story.

by Lorraine George

My son Tom was nicknamed the gentle giant. He was so kind and thoughtful and tall. He was always positive in everything he did. If there was anything he could not do he used to say, "Never mind, I will do this instead."

Tom worked at Morrisons supermarket and loved his job. He played for the store's football team and was mad about all sports. When we first found out about Tom's condition, that he had Marfan syndrome, we could not believe it. He had never been ill, and he loved life. We had never heard of Marfan.

It came as such a shock because we did not know Tom had a heart condition. We only found out when one day he started getting out of breath. We went to the doctors and they said it was probably asthma. Then, when it happened again, we went to hospital and that's when we found out it was Marfan syndrome. It is so hard to understand why it was not diagnosed before. Looking back, there were signs and symptoms and we feel that it should surely have been picked up.

Tom was so popular and so loved. It's so hard to accept that he is gone. I loved Tom so much. Tom had such a kind, generous manner and everyone loved him. He would do anything for anybody. He died at just 22 years old on 22nd August 2001, on the night before he was to have his heart operation.

I felt so angry that Tom was not diagnosed earlier, and I could not even bring myself to read up about Marfan syndrome. Now I feel I would like to help with a donation to help other families going through it.

Sir Sebastian Roberts

"He was such a wonderful man and so much that was brilliant about him was also bound up with Marfan – his very presence!"

Ebullient, eloquent and elegant, Major General Sir Sebastian Roberts was a "soldier of unusual intellect", an accomplished cartoonist, and a wonderful friend to the Marfan Trust. We are terribly sad to report his death on 9th March.

As Master of **The Girdlers' Company** from 2020 to 2022, Sir Sebastian encouraged and facilitated important pieces of research in our Sonalee Laboratory, by persuading the Girdlers Guild to fund three studentships every summer. From these studentships have emerged studies of newly discovered genes causing thoracic aortic aneurysms and a wealth of information on psychosocial effects and height restriction in Marfan syndrome.

This summer sees our students undertake important research on improved treatment for gastrointestinal disorders, and neonatal Marfan syndrome. None of this would be possible without Sir Sebastian's helpful interventions on our behalf. During the dark days of lockdown when our office was forcibly disbanded, Sir Sebastian ensured our homes had the necessary equipment to continue our roles seamlessly.

Sir Sebastian was never afraid to speak about his Marfan syndrome and used his knowledge and experience to cheer on others who had the same diagnosis. An advocate for our cause and a mentor to many, he gave clarity and comfort to parents and patients anticipating aortic surgery. Only recently a supporter reported feeling as though he had "a new lease of life" after speaking to Sir Sebastian.

"But for [his own] heart problem which prevented him taking on more front-line roles in his later career, he might have gone on to the most senior level in the army" (*The Daily Telegraph*).

Nevertheless, Sir Sebastian was Colonel of the Irish Guards until 2011 when he handed over to HRH the Duke of Cambridge, and the Colonel responsible for military doctrine when he wrote the British Army's doctrine of the Moral Component *Soldiering: The Military Covenant*.

He was also a broadcaster in demand during the late Queen's funeral. These are just three of his many professional achievements.

Diagnosed with Marfan syndrome as a child, Sir Sebastian was under surgeon Professor Marjan Jahangiri and Dr Anne Child, and spoke at the latter's retirement gathering. A revered raconteur, his oratory was "sought from far and wide" (according to his colleagues at The Girdlers) and facts gathered during his many adventures often peppered his anecdotes.

Sir Sebastian's many, inestimable achievements filled us with admiration at the Marfan Trust, and his positive philosophy and charming manner cheered us all. We will miss him hugely and send deepest condolences to his family and many, many friends.

Thoughts on Marfan Awareness Month

By Victoria Hilton

Resuming our Search for the Missing 15,000, we created and maintained a busy flow of disparate social media posts, articles and interviews, eliciting much conversation and creative engagement during this year's Awareness Campaign. Approaches to our helpline tangibly increased, signalling that the word has successfully spread.

In a cluttered world of competing causes, awareness can be diluted, but we emphatically returned the spotlight to our **Quest to Find the Undiagnosed Marfan People in the UK**. We are looking for the 15,000 Marfan folk unknown to the Marfan Trust. Of these, 8,000 remain dangerously undiagnosed. We launched our first official Find the Missing 15,000 in 2021 to great success and resumed the theme this year.

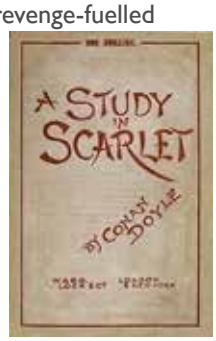
Through interviews with Professor Graham Stuart, supporter stories such as Julie's and Jonathan's, bespoke Marfan pamphlets and information videos, we experienced a 200% surge in audience reach and engagement on Facebook. Our campaign was viewed by over 14,000 on Twitter and we acquired lots of new followers on Instagram. Many approached our helpline wondering if they had the syndrome. We now take an average of **eight calls per week to our hotline**, compared to 6.5 last year, and have sustained the momentum.

Throughout February, our supporters mobilised their own mini Marfan armies, running hugely successful campaigns. Charlotte Nason, her son Kye and daughter Amiyah, engaged their local community with a Marfan-themed street stall and car-wash, raising over £500 and invaluable awareness.

Elsewhere, advocates big and small spoke variously at their school assemblies, doctor's surgery and places of work. Many of our supporters attended as patients the doctors-in-training sessions including Daniel Walker who wrote a fascinating article on his uplifting experience: **https://bit.ly/417rhUZ**

What inspired our Sherlock Holmes-themed lead campaign image?

In his first literary outing, Sherlock Holmes deployed his theory of deduction to unravel a mystery that had crossed continents. *A Study in Scarlet* introduced Holmes to the world, pitting his forensic reasoning against the revenge-fuelled murderous antics of sympathetic baddie Jefferson Hope. Sir Arthur Conan Doyle was a physician, and famously clinical in his descriptions of his characters, creating **in Jefferson Hope** a **physically vivid villain** of **tall stature**, ruddy complexion with susceptibility to epistaxis. **Hope** also suffered a **thoracic aortic aneurysm**. **Did Jefferson Hope have Marfan Syndrome, we and several physicians wonder?**



Meanwhile, our search for the 15,000 continues.

FEATS OF FUNDRAISING

Making a big difference to our small charity, the Marfan Trust fundraisers are our lifeblood and we are grateful for their unending generosity, and awestruck by their courageous campaigns.

SMILING AT THE FINISHING LINE

Picking up many blisters along the way, Sharon Lister ran the Manchester Marathon in April, finding it brutal but finishing it with a smile. Thank you so much to Sharon who raised not only £1,750 for our charity but also much-needed awareness for our condition. Sharon ran in memory of a beloved friend, Jamie, a happy healthy farmer who had undiagnosed Marfan syndrome. Cheering Sharon on every second of the way were Jamie's wife Christie and her children, Archie and Evie. You can read the full story of Jamie on our website: **https://bit.ly/3WTRzYX**

DEREK'S DO!

Forever fundraising, Marfan warrior Derek and his Priory Lodge of Acton 1996 raised £850 in an evening of sumptuous food, amazing prizes and wonderful generosity. Refreshingly, after an endless social desert we (Joanne and Victoria) were able to attend this magnificent fundraising event in person! Seated at the head table with Master of the Evening, John, his wife Aleksandra and their son Olly, we were treated to fabulous company and conversation.

The meal concluded in a grand auction starring a human-sized teddy bear. We introduced the 150 or so guests to Marfan syndrome and little Olly 'flew the flag' for our cause (pictured below on his mother's lap).

We thought to share **Derek's story of Life with Marfan**.

"I was born to profoundly deaf parents. No one in my family had heard of Marfan back then, but I knew something was wrong with me. I had scoliosis as a child that really worsened and eventually led to my spinal fusion in 1970 at 16, when I lost several inches in height. I had spent a year and a half in hospital during this time, in virtual isolation, in a halo frame. I saw no one but a nurse who visited with food. Inevitably I fell in unrequited love with her.

"After the surgery was completed one of my friends said he may have heard the doctors mentioning something called Marfan syndrome. However, I was not given this diagnosis. I did my 2365 electrical 3-year course with LEB in conjunction with Swindon College 232 electrician engineering so started

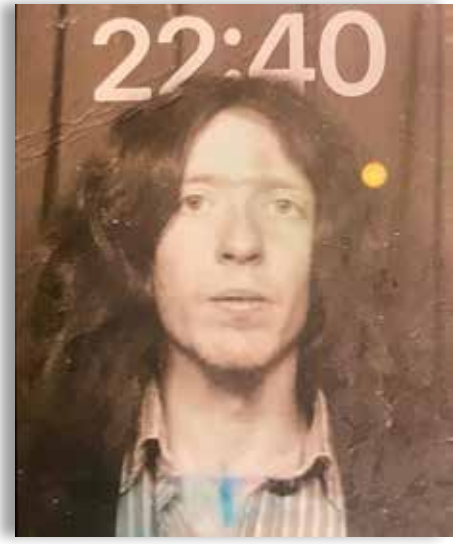
Aleksandra and Olly

CANNONBAWZ

Long-standing, loyal supporters of the Marfan Trust, the Cannonbawz Run are unstoppable in their fundraising feats. Kris and his 'bawzers traversed the north-east of Scotland in their latest car rally in memory of his beloved brother Liam.

my career in underground cabling then moving on to the metering section before coming a Field Service Manager Health & Safety checks on metering installations. I became a jointer, which is quite physical.

"Later in life I went to a refraction clinic at Moorfield's and they asked me who I was under for my heart. I was under no one! Clearly something was wrong with my heart and in 1979 I was given a mechanical valve at St George's under Dr Pumphrey. I was then referred to a follow-up cardiologist and ended up going to St Bart's for two years. My heart valve was replaced in 1999 at St George's. Both eyes were operated on at Moorfields, and I have had two pacemakers over the years. I now have an implantable cardioverter defibrillator – ICD – implant after a cardiac arrest circa 2019. I read an article in the *Daily Mail* by Dr Anne Child about Marfan syndrome and I recognised all the traits in me. I got in touch with Dr Child, got a firm diagnosis, and the rest is history."



Derek as a young man

FEATS OF FUNDRAISING

INTREPID ISABELLA

After much pedalling and a few tears, intrepid Isabella completed her first ever fundraising bike ride in two hours.

Defying a bitterly cold April day, she cycled 10 miles along the pretty paths of Stevenage accompanied by her brother and cheered on by her family. Isabella raised an incredible £1,500.

Diagnosed at two and discussing it at eight, Isabella is the only member of her family to have Marfan syndrome. As Isabella says: "I found out I had Marfan at two years old when my parents reported my eyes to be tested as I couldn't see my books or read them properly.

They soon found out my lens was dislocated. After this the doctors then tested my family for the faulty gene and I was diagnosed with Marfan.

Unfortunately, my heart and spine are affected but I have a great team of doctors looking after me. Everyone who has Marfan is doing really well. Keep it up!!!

I also spoke at my school assembly during February to help others understand my condition. My school has been incredibly generous and supportive, raising money over Christmas for the Marfan Trust."

The William Ransom School has donated over £700 to the Marfan Trust – thank you!

UNSTOPPABLE JORJA

by her mother, Jo

My daughter, Jorja, had a PEARS (personalised external aortic root support) and mitral valve repair in 2021. She emerged from the surgery unstoppable and is now in her second year at university. I never thought this was possible. With the Laurencekirk we held a Gala in aid of the Marfan Trust and raised £400.

Thank you, Jo and Jorja!!!

STAMP FOR MARFAN

Every perforated picture tells a story. Stamps have celebrated and commemorated people, fashion, and events for decades. They have kept us connected in a disconnected world, ensuring the safe passage of letters between lovers, friends and families. And they make a tidy sum for the Marfan Trust. Pauline Moses and her late husband, Raymond, have been collecting stamps, old and new, turning them into donations for our charity.

Ever tempted to rescue a used pretty stamp from an opened envelope doomed to the recycling bin? For many years, Pauline has been doing just that – collecting used stamps cut from envelopes for the Marfan Association and subsequently, the Trust. These stamps, together with any new unneeded ones, are sold to dealers for cash, raising money for our charity and thereby transforming lives.

Raymond and Pauline's son Peter tragically died from complications of Marfan syndrome and Pauline herself lives with the condition. As Pauline says of her stamp-collecting enterprise: **"I think with the price of stamps and general hardship, people need to know just a few from 100 people could weigh a kilogram!"**

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 can be stamps of any description and from any country. Please send to:

Mrs Pauline Moses
The Waves, Coast Drive, St Mary's Bay,
Romney Marsh, Kent TN29 0HN

SAVE THE DATE
MARFAN INFORMATION DAY: SATURDAY 7th OCTOBER

Our next Information Day takes supporters on the full journey of Marfan syndrome from conception to old age, with pitstops in between where the spectrum of the condition will be comprehensively discussed including:

- Pregnancy & Marfan syndrome **Dr Isma Rafiq**
- Information & Treatment for Descending Aortic Dissection **Professor Christoph Nienaber**
- Contact & Scleral Lenses in Marfan Syndrome **Dr Yetunde Obadeyi**
- Gastrointestinal Problems & Marfan Syndrome **Professor Anton Emmanuel**
- Exercise & Marfan Syndrome **Dr Sabiha Gati**

INTRODUCING WILLOW

Elegantly feasting from treetops, the giraffe carries its lofty stature with a swaying walk and gentle pride. This graceful creature reaches the leaves others cannot and is the emblem of Marfan syndrome. We have introduced our mighty mascot into our shop and named it Willow after a tightly-contested poll.

WESTMINSTER HEALTH FORUM NEED YOUR HELP

Critical to improved health and quality of life is early diagnosis, something arguably hard to achieve with rarely seen illnesses. Joanne Jessup recently attended the Westminster Health Forum which hopes to change this but needs your help in accomplishing its four main priorities for faster diagnosis, better co-ordinated care, increased awareness amongst medical professionals and improved access to specialist care. Her article on the event is online and she asks that you please do click on these links to participate and help secure a healthier and happier future for others with a connective tissue disorder: bit.ly/3le1N0i

THE MARFAN WEB SHOP

Welcome to our shop, a trove of lovely things and newly expanded with Willow, the Marfan Giraffe.

Jeremy Wins

Marfan Trust Tote Bag


Marfan Trust Pen

A Zest for Life Cookbook


Red Heart Keyring


Willow, the Marfan Giraffe

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

Response Slip (this can be photocopied)

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**.....

..... **Postcode**.....

Email.....

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
Willow, the Marf Giraffe	£5.50		
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		
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)			

☐ I am eligible for Gift Aid:
(please tick if relevant and leave name and address above) **Signed**..... **Date**.....