



A Word from Our  
Chairman, Michael Heath  
Page 2

Helpline Analysis by  
Victoria Hilton Page 3

Sonalee Laboratory:  
Research Update  
and Summer Students by  
José Aragon-Martin  
Pages 5 & 6

Fundraising Update by  
Gurpreet Madan  
Pages 9 & 10

In Memory Page 11  
Feats of Fundraising  
Pages 12, 13 & 14

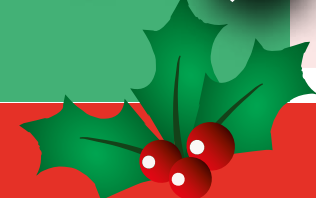


How Tall is Too Tall by  
Dr Anne Child MD FRCP  
Page 4

Psychological Support by  
Dr Zoe Berger  
Page 8

Marfan Information Day  
Page 15

Summer Raffle Winners  
Page 15



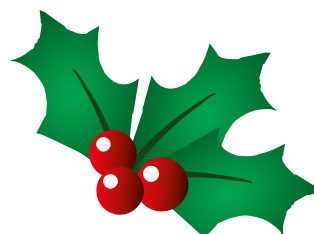
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# Marfan Matters



## Welcome to our Autumn /Winter Newsletter 2021



Dr Nitha Naqvi, Consultant Paediatric Cardiologist  
& Soon-To-Be Marfan Trust Trustee

Autumn has set the trees ablaze and the year is hurtling to its close. While basking in the season's fiery glow, we reflect on 2021. Dominated by a long, dark lockdown and a rapid vaccine rollout, it has proved a uniquely disruptive chunk of time for the world, yet a very productive one for the Marfan Trust. We are busily expanding! As we grow our community of supporters and develop our membership, we are expanding our roster of Trustees.

During the summer, we invited Dr Nitha Naqvi, Consultant Paediatric Cardiologist at Royal Brompton Hospital, into the fold. Winner of Asian Woman of the Year 2019, Dr Naqvi is well known to many of you from her frequent Marfan clinics at Harefield and Brompton Hospitals. An expert in our syndrome, Dr Naqvi spoke informally at our recent Marfan Information Day [page 15] on children, vaccination and myocarditis, clarifying the rampant misinformation, and demystifying recent press reports.

Describing myocarditis as inflammation of the heart muscle, Dr Naqvi confirmed that an

incidence of this condition had been noted in children following the COVID vaccination. However, the incidence is in the order of **1 in 16,700** while the risk of children developing myocarditis after contracting COVID-19 is **five times higher**. Dr Naqvi further warned of COVID continuing into debilitating long COVID, and of the potential to develop a very rare condition known as Paediatric Inflammatory Multi-System Syndrome (PIMS) which can lead to aneurysm. Needless to say that in a practical context, children with COVID miss school, disrupting their education in an already-disrupted school year.

To conclude, Dr Naqvi unambiguously recommends the vaccine for children. No child has died following the COVID vaccine but some have died of COVID. However, given that vaccine-related myocarditis is more common in boys, and more so after second vaccination, she **recommends just one vaccine jab**. There are no specific guidelines for children with **Marfan syndrome** and Dr Naqvi's preference is for **one jab** until the evidence accumulates.

# A Word from Our Chairman

by Michael Heath

It was lovely to see so many of you at our second virtual Information Day which was a resounding success. Once again Dr Child has brought together expert speakers covering the spectrum of Marfan syndrome.

The annual conference is just one of the many examples where Victoria and Gurpreet have created what I believe is some terrific event and social media output, from regular bulletins Marfan Monday and Dr Child's Casebook to ad hoc articles and celebrations of supporters. Our new website is filling with useful and informative content. The team is now busy preparing for 'Marfan Awareness Month' in February 2022.

Even a small charity like ours is an expensive commitment. In short, we cannot continue without your ongoing and generous funding through membership – your fees are a lifeline for us as are your ad hoc generous donations and personal fundraising activities. I cannot thank you enough or express how important it is to a small but vital charity like ours.

In recognising your support, the Trustees have a responsibility to ensure the Trust continues as a going concern. We the Trustees must look forward to secure the best interests of the charity. Therefore, we are reviewing where the Marfan Trust currently sits and how in the future we can best meet your needs and achieve the aims of our Charity.

Please do view our Christmas catalogue in this edition and check out with some fundraising purchases. Finally, may I wish you a very Merry Christmas and best wishes for 2022.



Chairman Michael Heath photographed during a visit to the Sonalee Lab with Dr José Aragon-Martin



A handwritten signature in black ink that reads "Michael Heath".

# Helpline ... A Lifeline

by Victoria Hilton

The sole source of personal support for people with Marfan syndrome in the UK, our niche helpline remains a port in a storm. Readily dispensing comfort, clarity and rarefied medical advice, Dr Child has taken 110 calls since our spring edition newsletter. Of the many assorted enquiries, one theme prevailed and that was the quest for a definitive diagnosis.

There were **25 separate enquires** from people who have been led to suspect that they may have Marfan syndrome, or something similar. This means potentially **25 lives saved**. Opportunities to identify Marfan exist everywhere from opticians and cobblers to GPs and physios. Greater general awareness of the condition is life saving for those living unknowingly with potentially life-threatening Marfan syndrome.

The second most popular line of enquiry was from **supporters seeking general information on the condition**. From the newly diagnosed to long-time sufferers, people often find general advice and answers on Marfan syndrome elusive: “how do I control chronic pain” and “are my gastrointestinal problems a consequence of the condition” are often asked. **Skeletal** (pectus chest, scoliosis, height control) worries are sometimes mentioned, while many contact us to discuss matters of the **heart** – the risk of the aorta expanding during pregnancy and labour for example, the

risk of endocarditis after dental treatment, the risk of myocarditis after the vaccine (see front page article].

Adapted every Wednesday into **Dr Child’s weekly Casebook**, our Helpline has started a conversation between our supporters. Our **peer-to-peer support** initiative is gathering momentum and we invite you to **join the conversation. It’s good to talk!** If interested, please email Victoria at [info@marfantrust.org](mailto:info@marfantrust.org). Meantime, we are publishing below a special edition of Dr Child’s Casebook with our Chairman’s permission. Michael Heath recently suffered a detached retina and turned to Dr Child for help!



## Dr Child’s Casebook: Detached Focus

by Dr Anne Child & Victoria Hilton

Wilful blindness, a state we sometimes succumb to in matters of medical emergency. Sensing something was awry in his left eye, our Trust Chairman adopted a “blokeish” bravado and ignored the obvious while delaying the inevitable. They say procrastination is the thief of time, but it can also steal your sight, as Dr Child warns.

**Question:** I should have known my left eye was not right! My father had suffered with detached retinas in the mid 1960’s – “it’s the Marfan, stupid”, but did I go straight to an eye hospital? No, I went into typical bloke mode – that is, it will be fine and waited a bit.

Eventually I need to visit an eye clinic on a Friday, receiving a diagnosis of Retinal Detachment and referral to the Vitreoretinal clinic at Moorfields the following day. Thus, very early on a Saturday last March, I am in the main entrance to Moorfields City Road. The seats soon fill up and then it’s join the steady stream to our clinic. I suspect for an eye hospital there is a daily occurrence of detached retinas that need immediate attention. A 61-year-old, I am on the list for surgery for that day and I am lucky there are no children or more serious injuries to go ahead of me. It’s a fascinating procedure completed on me without a full anaesthetic. I have an awareness of what is going on. Once

complete, I recover with tea and a sandwich, head down on my lap and a taxi home that afternoon. Back in the day it would have been a week or more in Hospital.

Once again, a first-class service from the NHS my thanks to all. But I should have presented earlier. If you have any problems you must not delay; the extent of any sight loss is at stake.

At the Marfan conference last Saturday whilst enjoying a fascinating presentation by Mr Robert Henderson, some questions occurred to me for Doctor Child’s Casebook:

1. Why are we with Marfan syndrome so vulnerable?
2. Apart from regular eye examinations with an optician is there anything else we can do to reduce the risk? Finally, any dietary advice other than a daily beer and scotch egg?

**Answer:** Procrastination is NEVER the stance to adopt



when symptoms present! Every second is precious with the eye, as you well know! It is sometimes human instinct to filter into our consciousness what makes us feel good but resist what is damaging to our ego! Please abandon the default stance of blokeish bravado and seek help.

In Marfan syndrome the risk of retinal detachment is significantly increased. It occurs because the light sensitive tissue peels off from the back of the eye. The connective tissue is weakened in Marfan syndrome and the tissue has come away easily.

For our wider public, symptoms that may indicate a retinal detachment are:

- Bright flashes of light, especially in peripheral vision
- Translucent specks of various shapes (floaters) in the eye
- Blurred vision
- Shadow or blindness in a part of the visual field of one eye

To answer your second question, you must undergo comprehensive eye exams with regular ophthalmologic follow up, particularly paying attention to the status of the lens, refractive errors, and retinal health. You will have heard during Mr Henderson's presentation that there is a risk of lens slippage in Marfan syndrome. Known formally as ectopia lentis, the crystalline lens slips out of place because the zonular fibres that hold it in situ are weak. To prevent it happening again avoid sports such as squash involving possible direct blows to the eye.

In terms of your diet – an all inclusive diet, plus a one-a-day multivitamin multimineral tablet will make sure you have all you need to make fibrillin. Other than that, whatever food makes you happy!

**Casebooks are available only to Best and Family Friends.** They are a treasure trove of information so do **join the Trust to read more.**

## How Tall is Too Tall? Treatment for Excessive Stature

by Dr Anne Child MD FRCP

In Marfan syndrome, from birth, children grow on or above the 99th percentile for their age and sex, and are usually one of the tallest in their class at school. In contrast, their average weight is 25th percentile, which gives them their tall, lanky look. When asked, adult males say that 6 foot 8 inches is the tallest they would like to be, since otherwise "they do not fit in this world". Car and aeroplane seats are not comfortable, clothing and shoes are difficult to find, and tall women worry that they may not find a suitable mate.

How should a parent manage this common problem? It is best to keep a growth chart of the child's height measured in bare feet, every year on the child's birthday. When the child reaches 150 cm (59 inches) it is time to be seen by a paediatric endocrinologist, for prediction of final height. Basically, 30 cm is added to the child's height, but to determine final height more accurately, a wrist x-ray for bone age, and pubertal staging through examination for signs such as early breast development, can refine this estimate.

If agreed with the daughter and parents, low dose sex steroid therapy taken orally can be used to induce puberty at an age and height judged to achieve a satisfactory end point. For example, if a girl is not to exceed a final height of 180 cm, she needs to have started breast development (spontaneously or induced with low dose oestrogen) when she has obtained a height of 150 cm. Exactly the same arithmetic applies to boys whose puberty can be induced with injections of low dose testosterone. Expected side effects need to be discussed and acceptable.

With hormone therapy, the long bones cease to grow, as confirmed by the wrist x-ray. This signals the end of the growth spurt.

In recent years females treated with high dose oestrogen have reported unacceptable side effects including cancer, and reduced fertility. Therefore, low doses of sex steroid are now recommended for purposes of safe treatment of tall stature.

In summary, parents can help by measuring the child's height annually and taking these figures to the appointment when the child is referred at height 150 cm by the general practitioner to a paediatric endocrinologist who can supervise pubertal management safely.

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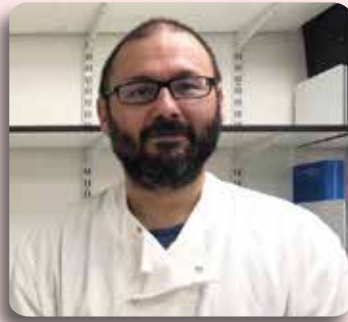
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# Marfan Trust Research Update

Our research programme into the causes and management of Marfan syndrome was created 20 years ago, replete with a specifically equipped laboratory. The Sonalee Lab was opened by the parents of its namesake, Sonalee Wijetunge, a young doctor who died during her ward round, of complications from Marfan syndrome. Over the years it has played host to a number of exciting research projects and every summer welcomes different students into the fold. This summer was no exception, with four young students drawn to the Trust through their interest in clinical genetics.



Dr José Aragon-Martin

## Summer Studentship Report

By *Husnaa Fathima Ali Azamathullah Khan*

I would like to thank Dr Aragon-Martin and the entire Marfan Trust team for giving me the opportunity to work on the project “Genotype – Phenotype associations in Marfan syndrome” this summer.

Since I had this opportunity to work on this project, I had the chance to develop remarkable skills and expand my knowledge on Marfan syndrome. Throughout these eight weeks, I had hands-on learning which fostered my critical thinking and analytical skills to further expand knowledge on this very important medical condition.

As a fourth-year medical student studying in the Queens University of Belfast, we often have our clinical rotations throughout the entire academic year. Having the opportunity to be a part of this research team helped me realise how important research is.

The project I was working on was ‘Genotype – phenotype associations in Marfan syndrome’. In this project, we are not only looking at the position of the amino acid that has been exchanged in Marfan patients but are also looking at the different positions of the amino acid that has been exchanged resulting in the difference in the clinical features that were seen. We focused on the mutation in the FBN1 gene.

I decided to apply for the sponsorship based on whether I can try to start a new society here at Queens that further helps people understand what Marfan syndrome is and the complications it can lead to if not diagnosed at an early stage. I am hoping to make a difference starting first amongst medical students here in Northern Ireland.

## Volunteers in the Sonalee Laboratory

A volunteer medical student from Imperial College, Rohan Bhupar, has spent a productive summer cataloguing the blood and DNA samples in our freezer collection, so that we are ready for HTA inspection (Human Tissue Authority) in our new location, as part of the Brompton Hospital database and biobank. He has also learned to draw pedigrees on the computer, and has updated our Thoracic Aortic Aneurysm and Dissection TAAD pedigrees in preparation for a new grant application to the British Heart Foundation in 2022. Rohan once volunteered for us as a Year 12 student and remains interested in our research.



Rohan Bhupar

## Thank you, Girdlers' Guild!

By *Dr Anne Child, Medical Director, Marfan Trust*  
& *Dr Jose Aragon-Martin, Sonalee Laboratory Director, Marfan Trust*

We remain grateful to the Girdlers' Guild for their continued support of our charity. Their loyalty has allowed us to enhance our knowledge of the causes and management of aortic aneurysms including Marfan syndrome (MFS), and allowed us to update our pamphlet literature for reprinting and distribution.



In proceeding with our research projects, we used their very kind continuation grant of £3,000 donated on the 30th of June 2021 and the additional £1,400 grant for training and education from the Jock French Fund, on the 28th of April.

Imperial College MSc students **Qasim Mansoor Ashraf** and **Lauren Lau** spent the summer studying causative genes in thoracic aortic aneurysm and dissection TAAD. Since they were self-funding, we used the Jock French funding toward the purchase of two updated computers (Apple Mac, total cost £3,731.68). These two young people worked as data scientists, and were handling large amounts of DNA data, requiring external drives for their computers.

**Qasim** was working on the analysis of results from Next Generation Sequencing (NGS) data in Thoracic Aortic Aneurysm and Dissection (TAAD) Cohort 1,

and discovery of novel (TAAD) genes in families 1 and 3. Meanwhile **Lauren** was working on the analysis of results from NGS data in TAAD Cohort 2, and the discovery of novel genes in families 2 and 4.

A third student from Queen Mary's University in Northern Ireland named **Husnaa Khan**, assisted Dr Aragon-Martin in doing a literature review of genotype-phenotype correlation in Marfan syndrome. Upon this basis will be added our own findings to make a very fine publication. Studentship Cost: £1,000.

A further Girdlers' Student, Anna Apará, a science student (pre-medicine) at St Andrew's University in Scotland, took on the updating of our literature pamphlets, ready for publication after reprinting. Some of our very old publications are out of date. These pamphlets pertain to the **Management of Pregnancy, Eye Disease and Psychosocial Problems**, and she has written a new one on **Stroke in Marfan Syndrome – Risk Factors, Types, Incidence, and Prevention** as well as **Management**. These will form the basis for an updated article for general practitioners to increase awareness, probably to be published in the British Medical Journal. Girdlers' Studentship: £1,000.

We are extremely grateful to the Girdlers' Guild for giving these young people a chance to enhance their skills and knowledge in the field of molecular genetics. The total cost of the students and equipment was £5,950.72.

The extra additional funds (£1,550.72) were taken from the Marfan Trust research fund, to supplement the Girdlers' donation. The staff and trustees of the Marfan Trust are extremely grateful for their continued support.

### Recent Research in the Sonalee Laboratory

By Dr José Aragon-Martin

In this report for our Autumn 2021 Newsletter I will talk of my Summer Research at the Marfan Trust. I have several projects in motion, remote and within the Sonalee Lab. As I write this, I am currently working with Dr Sanjay Sinha at the Cambridge Stem



José and Lauren at work

Cell Institute, discovering new drugs that will help us to better understand Marfan syndrome. I am also working on genotype-phenotype association in MFS to find out if a particular mutation in the FBN1 gene produces a particular clinical feature and, in this way, try to predict the clinical features in a newly diagnosed Marfan patient. This way we can better access the number of visits per year the patient will make to the clinic for follow-ups. We are also working on the fine mapping of the FBN1 gene to describe better

those variants that are causative and those that are just so-called normal.

We are looking to discover new causative genes in thoracic aortic aneurysm and dissection TAAD, in ectopia lentis (dislocation of the crystalline lens in the eye) and Ehlers-Danlos. When a patient with clinical features comes to our clinic it is our duty to find out the genetic cause in the patient. If the condition is fatal like Marfan syndrome we must rule out FBN1 involvement. If we screen the patient for next generation sequencing (NGS) it gives us the power to find the cause of the condition.

Our Imperial College MSc students Lauren and Qasim worked over the summer on NGS and thoracic aortic aneurysm and dissection TAAD. Both students of genomics, Qasim and Lauren were drawn to the TAAD project through their interest in clinical genetics with TAAD an overlapping feature. Qasim looked at a cohort of 20 patients, all unresolved cases, with the aim of identifying the potentially causative gene. Lauren meanwhile examined cohort 2 at 19 cases, also with the aim of identifying the causing gene.

Qasim and Lauren were able to find variants of great significance in their TAAD cohorts but we have not yet had time to go over their data to analyse these variants and confirm their responsibility to the disorder. We are still waiting for further data analysis and confirmation. We have to check that the variants are significant and important before using DNA from family members to confirm segregation of the variant in the family. Our samples are currently being audited by the Royal Brompton and once confirmed we can research the samples.



# THE DANIEL FINKLETAUB PRIZE 2021

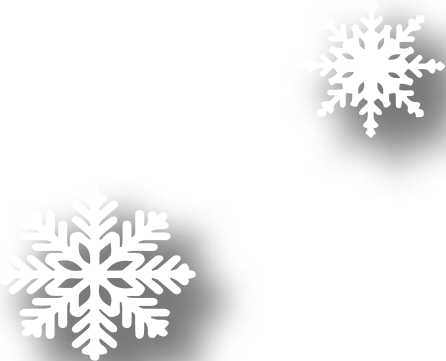
To commemorate their beloved son and brother Daniel, Carole and Elaine Finkletaub created a student prize in his name. Daniel died in 2009 at just 41 of complications from Marfan syndrome. Although his life ended prematurely, it was a life lived fully. He worked hard, completing two degrees and rising 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[s] of the Daniel Finkletaub Prize:**  
Mohamed Abdelrazek and Thomas Whittaker

Final-year medical student at Cardiff University, Mohamed occupies both sides of the doctor's consulting table. The first member of his family to be affected by Marfan syndrome he suffers its symptoms visibly and invisibly. While working in Wales' hospital hotspots on COVID-19 wards during the pandemic's reign he decided, with fellow medical student Tom Whittaker, to investigate the effects of the virus on his own Marfan community. In doing so, he and Tom made an exciting discovery. Reassuringly, Marfan patients do not appear to be affected severely by COVID-19. The clinical pattern differs significantly from the normal population with less fever, but more fatigue and muscle aches. This may provide evidence that MFS is a protective factor due to reduced adipose tissue (fat). On this happy note, we can say that Mohamed and Tom are worthy winners of the prize!



Mohamed and Tom



# Psychological Well-Being: Living and Thriving

by Dr Zoe Berger, Consultant Clinical Psychologist, Great Ormond Street Hospital

## What Do We Know from Literature?

The vast majority of people with a chronic health condition cope and adjust well. Approximately 20% of such children experience psychological difficulties. Research shows no consistent link between severity and outcome. Parental mental health is strongly correlated to the child's mental health. Children often have a parent who has the same health issue.

## Impact on Psychological Well-Being

Feelings of being different from peer group loom large, and give rise to body image concerns and low self-esteem, low mood and anxiety, worries about the future, and about living with uncertainty. The child must adhere to regimes including medication, treatment, hospitalisation, and surgery. Loss and grief if another family member has died can be an important issue.

## Visible Differences

From the age of five, affected children realise that they are different from their peer group. Some are teased and bullied. Handling this requires cooperating with teachers, talking about it, reporting it, and building social skills to help the child manage the problem.

## What Do We Mean by Coping?

This is a psychological process carried out in response to a stressor, which involves problem-focused and emotion-focused approaches. These help a person to deal with something difficult in their life.

## What is Resilience?

Resilience is something that can be developed in the face of challenges, and allows the person to overcome stress or adversity. This may require personal changes allowing survival.

## What Can We Do to Improve Well Being?

- Practise good selfcare e.g. healthy eating, sleeping well
- Do one pleasurable thing per day
- Build in relaxation and time to replenish
- Ask for help
- Talk openly and honestly about the medical condition
- Listen to your child without judgement
- Teach about differences from a young age. No one is perfect.
- Get the school involved
- Help identify strengths to build self-esteem

## Dealing with Questions

Practising example responses in role play with your child will equip them. Most people ask questions because

they are curious. Explain, reassure, and distract them in conversation. Dealing with staring can include looking back, smiling and holding the person's gaze, moving away, or asking them to stop staring. Distract yourself by thinking of a pleasant thought.



Dr Zoe Berger

## Acceptance

Accept what you can't change and commit to valued action directed at leading a fulfilled and thriving life.

To get further support contact your GP for referral to local psychological services. School counselling may be available, and the local paediatrician may be able to help. Adult mental health services are also available through the NHS, but often with delay.

Below is a list of organisations that can be helpful on a more urgent basis.

1. **Shout** offers confidential 24/7 crisis support. Text "SHOUT" to 85258.
2. **Childline** is a confidential telephone counselling service for any child with a problem. Call 0800 1111 anytime, or arrange an online chat with a counsellor.
3. **The Mix** provides free confidential support for young people under 25. Call 0808 808 4994 or email
4. **Samaritans** 24/7 365 all year call 116 123 or email [jo@samaritans.org](mailto:jo@samaritans.org)
5. **Crisis Tools** helps professional support young people in crisis, short accessible video guides are available.
6. **Papyrus** provides confidential support for young people or those worried about a young person. Call 0800 068 41 41 or text 07680 039967.
7. **Good Thinking** is London's digital service and provides a range of resources for young people to improve mental wellbeing.
8. **Kooth** a free anonymous online mental wellbeing community including live chat with the team.
9. **Beat** provides support to help young people with an eating disorder, call the Youthline 0808 801 0711 or Studentline 0808 801 0811.



# It's A Wrap....

by Gurpreet Madan

As the year draws to a close, it's important we reflect and gain some perspective on what the Marfan Trust has achieved and why we need your support, while outlining upcoming plans for 2022.

## Marfan Trust's Impact

- We answered over 260 calls this year on the Helpline;
- We held our second virtual **Marfan Trust Information Day**, raising £845;
- We created a campaign during **Marfan Awareness Month** to find the **Missing 15,000**. Working with the amazing Freuds on a pro bono basis we devised a campaign that targeted radio stations. Some statistics to put this into perspective: the number of radio stations reached: 221; Our audience reach: 24,725,780. Similarly, our in-house awareness campaign created an awareness momentum that was accelerated over a series of strategically planned communications. There was a surge in our helpline calls which doubled as the month of February wound on and these continue to rise. According to Facebook, we reached 46,396 people over the month of February. There was also an increase in the number of literature downloads during and after the campaign.
- We launched our **membership scheme** in June 2021, Over 120 people have signed up thus far. To make sure you enjoy the perks of being a member: Sign up here - <https://www.marfantrust.org/pages/10-membership> We hope to welcome you as a Best Friend.
- We held our **first webinar**, on Pregnancy and Preimplantation Genetic Diagnosis and are working on a few more next year stay tuned!
- We launched our **new website** which makes it easier for supporters to navigate and has the functionality to support those who want to make donations or set up their membership directly on the website.
- We introduced a **new design for our newsletter**. Through a poll conducted on social media, we asked you to help us name our new and improved newsletter and you've spoken: Marfan Matters! We hope you enjoy it as much as we enjoyed creating it.

## Why We Need Your Support

- Sole Marfan Charity in the UK  
Since the merger with MA, we are the sole Marfan charity in the UK. We are here to support and advise you on all things Marfan syndrome.

## • Helpline enquiries are rising

With the calls received by the helpline rising, Dr Child and Victoria have been working tirelessly behind the scenes to make sure everyone's queries are answered. If Dr Child is unsure or feels that a Marfan Trust advisor is able to better answer your query, she gets in touch with them to give you a comprehensive answer/explanation.



Gurpreet, Trust Fundraising Manager

## • Research needs to continue

José, Research Director has made an invaluable contribution to the understanding and treatment of Marfan syndrome. Due to lockdown restrictions and the fact that we moved to our new premises and that our new laboratory was only fully equipped by October 2020, Jose spent his time analysing the data we already have. Now that things have returned to normal, José will be focusing on undertaking medical and analytical research so that more is known about Marfan syndrome and its management. Such as searching for new genes for Ectopia Lentis.

## • Awareness

Awareness is very important. Based on Dr Child's calculations, we know that 18,000 people in the UK have Marfan syndrome. However, we only know of 3,000. To find the remaining 15,000, it's important that we raise awareness, that we talk to people about Marfan syndrome and that we spread the word. Through our small efforts during February Awareness Month, we noticed a resulting surge in people contacting us so let's keep the momentum going!

## Upcoming Plans for 2022

### • Fundraising Committee

If you have any experience in fundraising, are willing to share your contacts or have some brilliant ideas, please get in touch and join our fundraising committee. We are planning on our having our first introductory call at the end of the month.

### • Working on strategy and direction of the Trust

With the merger of Marfan Association and lots going on over the last two years, the Trustees will now be making it a priority to discuss the strategy and direction of the Trust. They will be holding their meeting at the end of the year and we hope to share the findings with you early next year.

Continued on page 10

- **Marfan Awareness Month**

Planning for Marfan Awareness Month February 2022 will commence soon and this time around the focus will be around educating medical professionals. Watch this space!

- **Webinars**

We plan on holding more webinars next year. This will be based around helpline queries analysis, supporter feedback and social media. However, if you have any suggestions, please do let us know!

- **Trustee Recruitment**

A couple of Trustees have stepped down and we are in need of new ones. A skill audit was conducted earlier

this year and we now realise we need a Fundraising Trustee and a Treasurer. Could that be you? Or maybe you know someone who fits the bill? If so, please let us know?

If you would like to support the Trust through fundraising or volunteering, please email Gurpreet Madan at [Gmadan@marfantrust.org](mailto:Gmadan@marfantrust.org) for ideas and a chat. Similarly, if you have any suggestions, would like to find out more about what the Trust has been up to or about our becoming a Best Friend, then please do drop me a line. Thank you for being amazing supporters, we wouldn't be around if not for you!

## Dear Marfan Family,

First and foremost, thank you for being wonderful supporters of the Marfan Trust, we are thrilled to have you on board.

Unlike many other charities, the Marfan Trust receives no government statutory funding and relies solely on supporter donations. With the pandemic and the additional cost burden this has placed upon us, we need all the help that we can get.

Like the Marfan Association before us, we have launched a membership scheme to help us secure our future and ensure we continue to be around to provide Marfan specific support and research services.

A lot of time and effort has gone into conceptualizing our communication strategy. We hope you've enjoyed and will continue to find articles like Dr Child's Casebook, blog posts, research articles and even our latest addition – Anecdotes and Antidotes useful. We have made it a point to always listen to the needs of our supporters and to adopt these findings into our communications. Therefore, if you have any ideas that will help us be better, please do let us know?

If you can find it in your hearts to give £3 a month (the price of a coffee) it will go a long way to ensuring we stay around for years to come. £3 will help us to continue to provide high-level, interesting content for you and your family. Simply subscribe online to receive all our communications on Marfan syndrome and how to manage the condition.

Your donation of £36 per year is vital in enabling us to respond to two additional calls received by the helpline. Because of you, we are able to ensure that those affected by Marfan syndrome receive the care and support they deserve.



The Marfan Trust Team

On behalf of all of us here at The Marfan Trust, we thank you for being a fabulous supporter of the Trust! If you have any questions, please email Gurpreet Madan at [Gmadan@marfantrust.org](mailto:Gmadan@marfantrust.org)

Yours Sincerely,  
Gurpreet Madan

Gurpreet Madan Fundraising Manager Marfan Trust

# In Memory

**Edit Kovacs • Sheila Smith**

## **Liam O'Neill**

The Marfan Trust was very sad to hear that Liam O'Neill unexpectedly died this August after heart surgery. Liam lived uncomplainingly with Marfan syndrome and was very much loved by his family and friends. Over the years Liam's brother Kris, an air ambulance paramedic, has raised a fortune for the Marfan Association and Trust through his Cannonbawz Run, a car rally in Scotland.

Liam's mother Ingrid wrote to the Trust saying: "we have lived with Marfan syndrome for 37 years and saw how it affected Liam. He had been lucky to receive the best of care from amazing consultants and surgeons and lovely nurses. Liam never complained about his illness and just got on with his life, I think because of this we underestimated the seriousness of the condition. Liam had been through open-heart surgery many times and always came out the other side with a smile on his face.

It has been a huge shock to us all that the effects of Marfan Syndrome ended Liam's life at such a young age, he is sadly missed by so many people."

## **Liam**, by his brother, Kris

"My brother Liam lost his fight this year after a long time in ICU after his aortic arch replacement surgery.

I loved my brother so much: he was my bro', my rock and my hero. We are all so very lost without him, he was my inspiration in life and for starting the Cannonbawz Run, he was an honours degree graduate in networking, a karate teacher to young kids, and was three belts away from black belt, which he was awarded at his celebration of life. To date we have raised over 25k for the Marfan charities and will continue to do so."



Ingrid and Kris raised over £900 for the Trust at Liam's funeral at which the pallbearers were dressed as Stormtroopers and the Star Wars main theme was played.

# Feats of Fundraising

As lockdown gave way to liberation our fabulous fundraisers picked up where they left off, running marathons, swimming lakes, cycling long distances and walking even longer distances. It's been a wonderful summer of fundraising feats. Thank you so much!

## Lucy's Treat-Filled Raffle



Lucy and her Raffle Prizes

One half of the @Marfriends\_ teamworkers Instagram duo, Lucy Atkinson spreads the much-needed digital word on Marfan syndrome through her innovative

posts and candid blogs while raising money for our charity through her treat-filled raffles. Lucy's most recent raffle was a summery sensory feast of candles, prosecco and a vibrant yellow cushion. This lovely confection reaped over £70 for the Marfan Trust. Thank you so much, Lucy!!!

## Life's a Cabaret ..



Louise speaking at her father's memorial cabaret



Performers at the Cabaret

Buoyantly staged on 2 October in memory of her father who died in December 2019, **Louise Baskeyfield's** fundraising cabaret was a rip-roaring success. Over one hundred attendees soaked in an evening of conjuring tricks, amusing antics and sonorous voices produced by a stellar line-up of magicians, panto-dames and professional singers. In doing so they managed to collect over £5,000 for the Marfan Trust. Louise's father had Loews-Dietz Syndrome, and Louise and her children have all inherited this condition. Louise, her family and friends wanted to create an evening that both memorialised her father and helped a

charity working to improve and save the lives of those with a connective tissue disorder. We feel very lucky that they chose the Marfan Trust.

## Sibling Double-Act: Freddie and Alexandra Beard

By Freddie Beard

"Hello to everyone reading this! We are a Brother and Sister Team who followed in our dad's footsteps in conquering the 2-mile swim of Hyde Park's Serpentine this September, all in aid of the Marfan Trust. Having been diagnosed with the condition as a 16-year-old, I (Freddie) am conquering my condition, and my fear of the open water to help out with the great causes and research on Marfan to help any who will be, or are diagnosed with the condition.



Freddie and Alexandra Beard

The swim was a toughie! But I managed to pull through and swim a time of 1 hour 1 minute and 52 seconds which I was personally very happy to achieve. Knowing that I had everyone's support kept me going, along with the encouragement of my amazing girlfriend, my family and my friends.

Being able to achieve this with my sister, who herself overcame many hurdles to conquer the course with a great time of 1 hour 30 mins was so amazing. We pushed each other through our training and we were there for one another before and after the race. I couldn't be prouder of her, so well done sis, here's to the next physical challenge!

It was a massive personal challenge for me too, but it was one I was delighted to overcome to support the work towards Marfan syndrome. I also hope that it helps anyone else who is, or may become diagnosed, to know that it doesn't hold you back from achieving your goals. To quote Back To The Future's Doc Brown **"if you put your mind to it you can achieve anything!"** and I truly believe that applies to all of us with Marfan syndrome.

**So let's celebrate now, and here's to all those with Marfan! You are not alone and there is nothing that can stop us!"**

Freddie and Alex raised £2,223 for the Marfan Trust!

# Feats of Fundraising

## In Memory of Micah



Cambridge Cats

Micah Gillings died suddenly and unexpectedly in July 2020 at just 19 years of age, leaving his family, his girlfriend and his many, many friends devastated. Micah also left behind innumerable happy memories and his legacy will be long and lasting, not least at his basketball club, the Cambridge Cats. A talented player, Micah was very popular at the Club and they recently staged a tournament in his memory. Organised by coach Terry Reaves, it was a very emotional day as Natasha, Micah's mother describes: "Team Micah" came third, which was amazing as it was made up of his friends, family & past team mates, some of whom didn't even know each other let alone play as a team. They were playing against five other proper teams. It will become a yearly pre-season tournament now which is just amazing, he would be so happy 😊. I've just heard that £890 was raised, which is amazing!"

## The Roads Less Travelled: Derby to London



Peter Maurachea



Pete's Props

A diagnosis of Marfan syndrome and subsequent heart surgery in 2017 left Pete Maurachea feeling pessimistic. The world suddenly seemed a bleak place. But with time, patience and the support of family, friends and his wife Sam, he eventually made a

full recovery and is now unstoppable! Lockdown unlocked a newfound thirst for cycling which he took up in November 2020 and he hasn't stopped since. It was whilst cycling during winter of last year and feeling the cold more acutely than ever before that an idea formed in his head: wouldn't it be fun to cycle from his home town of Derby to London. And cycle he did, raising £2000 for the Marfan Trust in the process. Cruising past such significant personal signposts as the hospital that saved his life (Glenfield) and with props including Brewdog and Warfarin, Pete reached London, tired but triumphant.

Pete wishes to especially thank the manager of Rutland Cycling Pitsford Water: "I had a bike malfunction and the manager came out over 13 miles away and fixed my bike, I told him what I was doing and he did the service free of charge and pledged a donation to the charity. This restores my faith in people."

## A Walk of Two Halves



Stephen Rudge Walking for the Marfan Trust

Two years ago, Stephen Rudge set off on foot from John O'Groats, destined for Land's End. Seven days later a fractured shin intervened, curtailing his ambitious expedition and leaving him in agonising pain. Once he recovered, lockdown then intervened and with no end in sight Stephen decided to complete his walk in the areas around his home and beyond. He covered a distance of 1,064 miles in some six weeks and two days. This is fantastic! In doing so Stephen raised £2,700 for the Marfan Trust, to help his niece Louise, whose husband and son have the syndrome. Thank you so much, Stephen!

# Feats of Fundraising

## At the Finishing Line



Stuart Did It!

Defying doctors' predictions and surmounting weak ankles, **Stuart Sadler** is enjoying a full and 'marvellous' family life while running miles and miles for the Marfan Trust. Diagnosed at 22 with Marfan syndrome, alongside his mother, he was given a short life expectancy, warned not to have children and never to run again. Now aged 49 he is a proud father of two with a long life ahead, and has been running non-stop for the last four years for the Marfan Trust. He's raised over £8,000 and recently completed the London Landmark Half-Marathon in 2 hours, 25 mins, 38 seconds and the Great North Run. Thank you very much!!!

## Leap of Faith

By Hannah Shepard and Caroline McCarthy



Hannah and Caroline with Jonny's mother, Elizabeth Jonny Edwards

"Two years ago, we lost **Jonathan Edwards**, someone who was very close to our hearts and someone who was loved by many. Jonny died from an aneurysm. Prior to this he was fit and healthy.

Had we heard of Marfan syndrome we may have been able to join the dots. Jonny would have been tested and probably undergone surgery. In retrospect, the signs

were all there. He was tall and thin, his rib cage stuck out (pigeon chest), his toes curled, he had stretch marks on his back and he was able to touch his wrist with this thumb. His wing span was longer than his height.

We wanted to raise awareness and money for this condition and on Saturday, 16 October completed a sky dive, making £1,865 for the Marfan Trust."

## Daniel and Kate Cycle from London to Brighton



Daniel and Kate at the Starting Line

Last year's London to Brighton Cycle Ride was tragically cancelled, leaving **Daniel Walker** and his partner **Kate Renoldson** to take the roads less travelled. Without the closed roads of

the public ride they plotted their own route, covering 94 kilometres in 5.5 hours, and making a fortune for the Trust. This year the London to Brighton Cycle Ride was emphatically back in business. Daniel and Kate joined the throngs of cyclists, heroically covering 88.34km in 4 hours and 40 cycling minutes, at a maximum speed of 73 km/h (though Daniel admits some of the downhill were pretty steep). Daniel has Marfan syndrome and he wishes to thank all who contributed to his campaign which "has gone amazingly well, currently standing at £2,341.19 so just short of our target of £2,500. However, the Gift Aid will add in a further £452.50. Thanks all!".

## Helen Jacobs' musical Memorial Mass raised £320 for the Marfan Trust



Helen Jacobs

# Marfan Information Day 2021

Members of the medical elite converged virtually on Saturday 9 October for our second Zoom-held Information Day. Covering the Marfan spectrum of heart, eyes and skeleton, Consultant Cardiac Surgeon Mr Ulrich Rosendahl, Consultant Ophthalmic Surgeon Mr Robert Henderson and Consultant Orthopaedic Surgeon Mr Hilali Noordeen each spoke on their specialty and took questions from the large audience. They were joined by Consultant Child Psychologist Dr Zoe Berger from Great Ormond Street Hospital, and Dr Anna-Maria Choy from Dundee University making for a very impressive line-up. Over 60 people attended our conference and the talks are now on sale for £5 each.

## The Luck of the Draw: Summer Raffle

It was on a rain-soaked stormy day that we held our Summer Raffle Draw! Picking the winning tickets from a large box of stubs were, from left to right: Victoria Hilton, Helpline; Sandra, our fantastic new finance volunteer; Qasim our brilliant young Imperial College MSc student and José who needs no introduction.

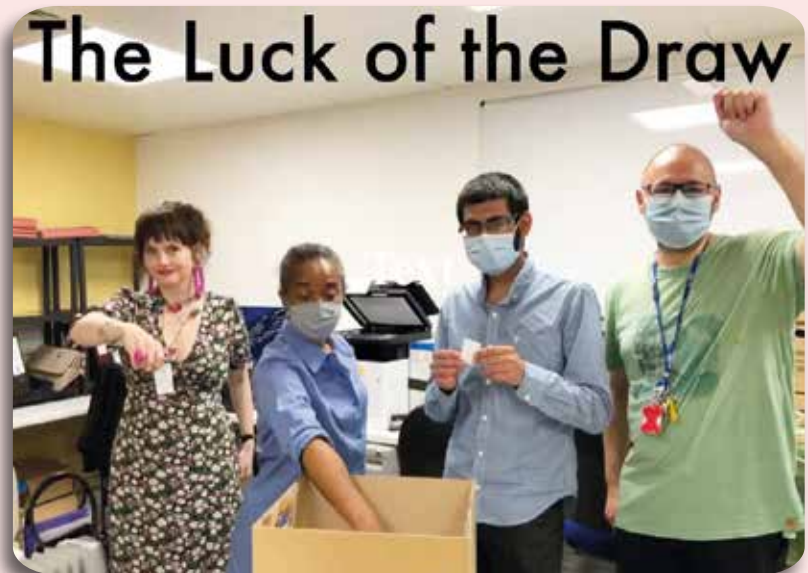
**First Prize:** Val Greatorex won tickets to Classical Spectacular at the Royal Albert Hall

**Second Prize:** Riikka Sikanen won a £50 Amazon voucher

**Third Prize:** Mr and Mrs Kinnard won a National Garden Voucher

**Fourth Prize:** Xan Randall enjoyed the company of goats at Buttercup Sanctuary

**Joint Runners-up:** Jo Gore & Mr Price enjoyed decadent treats from Hotel Chocolat



## Turn Old Postage Stamps Into New Money

Pauline Moses continues to sell old postage stamps to dealers, in doing so raising money for the Marfan Trust. She and her husband, the late Raymond Moses, have made a veritable fortune for the Marfan Association and Trust, through their stamp initiative. Please send any of your old stamps to: Mrs Pauline Moses, The Waves, Coastal Drive, St Mary's Bay, Romney Marsh, Kent, TN29 0HN



# CHRISTMAS SHOP

Spread some festive cheer this year with our quintessential Christmas scenes. In doing so you will help the Marfan Trust! With five different designs, each emblematic of Yuletide's celestial magic, our cards are a celebratory must. And at £4.00 for a pack of 10 (excluding p&p) they are a seasonal steal! What's more, we proudly unveil Zest for Life, a treasure trove of cookable recipes. This is published in memory of chef Wayne Johnston, beloved son of Pam and Ivan, brother of Lyn and father of Lewis.

Welcome to  
Our Shop

[www.marfantrust.org](http://www.marfantrust.org)



Robin on a Snowy Branch



Christmas Wreath



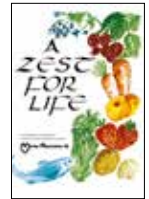
Three Kings of Orient



Snow on the South Bank



\*NEW\* Song of the Angels



\*NEW\* Zest for Life

## Response Slip (this can be photocopied)

Please return the response slip to the Marfan Trust, Guy Scadding Building, Imperial College, Dovehouse Street, London, SW3 6LY. Please make cheque payable to the Marfan Trust. Thank you for your support!

Name..... Address.....

Postcode.....

Email.....

Please insert number of packs required in the relevant box and include the cost of postage as appropriate. (Cost of Postage £3.00)

Card Design	Price per pack	Your Order quantity Enter No. of packs	Enter the Total cost in £s (No. of packs x cost)
Robin on a Snowy Branch	£4.00		
Christmas Wreath	£4.00		
Three Kings of Orient	£4.00		
Snow on the South Bank	£4.00		
*NEW* Song of the Angels	£4.00		
*NEW* Zest for Life Cookbook	£6.00		
Marfan Trust All Occasions Card	£4.00		
Marfan Trust Emergency Card	£2.00		
Marfan Trust Wristbands	£2.00		
Marfan Trust T-shirts	£5.50 (each)	Small ..... Medium ..... Large ..... XL .....	
Postage cost			£3.00
<b>Enter the Total cost of all items ordered plus postage</b>			

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

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



Registered Charity Number:328070

Marfan Trust, Guy Scadding Building, Imperial College, Dovehouse Street, London SW3 6LY



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