



Patrons: Sir Magdi Yacoub FRCS • Lady Maryanna Tavener • Prof. Marjan Jahangiri FRCS (CTh)

Autumn/Winter 2020 ♥

We are reaching the end of a strange and surreal year that has been like no other. Time seems to have stretched and shrunk in weird ways, but it has not stopped for the Marfan Trust which continues apace, as these pages will show. We staged our first ever virtual **Information Day**, and we herald the publication of our first ever, vibrantly visual, **Guide for Young Adults with Marfan Syndrome**.

(Virtual) Marfan Information Day

Members of the medical elite converged virtually on Saturday, 3 October for the Marfan Trust's most highly attended Information Day. Marfan syndrome (MFS) manifests itself in three main systems - eyes, heart and skeleton, and this was reflected in the different disciplines explored. Opening with Professor John Pepper, Consultant Cardiac Surgeon at Royal Brompton Hospital, the day featured seven specialists and 4.5 hours of expert content, exclusive to Marfan syndrome. A further hour was given to Q&As, allowing the 91 attendees, those with, or affected by, Marfan syndrome to engage with the speakers.

These talks are available separately for £5 each from the Marfan Trust. See page 15 for further details.

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'Remote' Insight from our Chairman ♥

As this extraordinary year rumbles on to its conclusion much talk is about family at Christmas. We at the Marfan Trust (the 'Trust') have been thinking a lot about the Marfan family. The team whilst working from home have continued to deliver and develop the Trust; three examples being Dr Child's weekly Casebook, ad hoc articles and our outstanding first ever virtual Information Day - highly attended and well received. It was great to see so many and meet some of you.

How the Trust extends this connection with you and provides the right support is crucial to our future. Our plan of supporter engagement is introduced in a separate article, following this one.

It was my first conference as Chairman of the Trust, and as I said on the day I'm humbled to continue my predecessor Michael Carr's work. Michael saw the Trust through many momentous changes, some of these physical, most significantly our move to Imperial College alongside the Royal Brompton Hospital. The Brompton has generously provided a new home for our research laboratory at the Guy Scadding Building which is slowly filling with cutting-edge new equipment – exciting tools for our resident scientist José Aragon-Martin. While COVID has left the nation in suspended animation, it has not interrupted our work. In fact, it has only served to drive and propel people to our Helpline – now a Lifeline for many of you. With Dr Child at the other end dispensing not only medical expertise but also moral support, we at the Trust are the first port of call, building our community of like-minded Marfan folk. This is reflected in our new and burgeoning social media sites managed by our new members of staff who have created a hub of information delivered in an accessible style which is attracting more and more of you daily. This online presence helps to spread the word, enlightening and educating the wider world of our niche syndrome. And it expands upon your



Michael Heath, Acting Chair

work, you being our busy supporters, you who heighten awareness not just through discussing the syndrome on your forums and blogsites but in your fundraising efforts for us and this is truly indispensable to us as a charity. We are very, very grateful to you.

An unusual condition, Marfan syndrome has an unusually dedicated 'champion' in Dr Anne Child, Medical Director of the Trust. I started on the theme of firsts and will conclude on this very note. It was Dr Child who first organised the international consortium which together located the gene that causes our condition, by tenaciously travelling far and wide collecting blood samples, perhaps from some of you, or your parents if not your grandparents, in her quest to find the cause of Marfan syndrome. That was some 32 ago and her zeal to improve and prolong our lives remains undimmed. From research projects on the boil such as Ectopia Lentis that will help uncover the genes for dislocated lenses, to our life-saving hunt for the missing 15,000 – that is those predicted to be living in this country many unknowingly with Marfan syndrome, the Trust continues, temporarily displaced, but always unstoppable.

The Trust will be working with and for you, as we all get through this winter, the pandemic and look forward to more comfortable times. Be reassured the Marfan Trust continues to: support the Marfan Family, deliver an innovative research programme, and heighten awareness amongst public and medics alike of our unique condition.

Best wishes for 2021 Michael

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Marfan Trust Supporters Survey and Consultation Process ♥

After our very successful October conference we at the Trust have been planning our 'next steps'. We have identified a need for a wide consultation with all of you: the Marfan family, our supporters, stakeholders, friends and other interested persons. We also feel that despite the ongoing upheavals of this year, now is both an opportunity and the right time to do this for the Trust.

The purpose will be to identify 'what people need and want from the Marfan Trust' as the single patient support organisation in the UK. We will then use this information to inform and develop the next Strategy Plan for the Trust.

The intention is to enable you all to participate by way of an online survey and to further gather ideas through a number of subject/group-related focus groups. The outcomes of these can then be linked to our Business Plan.

We are working now on the logistics of this, so watch out for invitations to take part in the survey and possibly participate in a focus group. We are looking to complete this process by the end of the year.

The Trustees feel this is a positive investment for the Charity; it is a key part of our commitment to engaging with our People. It will allow an effective plan to be set to meet the aims and objective of the Marfan Trust in the coming years. It does however mean a slight delay in our roll out of a new membership scheme in order that it is compliant with the outcomes of this consultation.

I would be most grateful for your patience and participation; I confidently look forward to a positive contribution to this process from you all.

Many Thanks
Michael Heath
Chair, Marfan Trust



Helpline ... a Lifeline ♥

By • Dr Anne Child and Victoria Hilton



A symbol of solace in surreal times, the Marfan Trust Helpline became increasingly sought-after during the pandemic's reign. Whilst the world was stilled and modern life suspended, Marfan health problems continued, and worries were nursed. From 'key' workers returning to 'frontline' duty, to housebound folk in chronic pain, the emails to our inbox were disparate and varied. Dispensing clarity and comfort from her virtual consulting room, Dr Child has taken over **100** calls since our Spring Edition newsletter, each one unique yet universal.

In detecting the patterns and trends that have emerged since 1 June, we noted that **16%** of queries were COVID-related. Brought into question were the protective merits of the flu jab, the underlying susceptibility of Marfan folk, and shielding measures. These calls are now ebbing away. Parents of newly diagnosed children left to ponder their next step, and recently diagnosed adults is a continuing theme that constitutes **10%** of calls. This mirrors the converse - those who seek a definitive diagnosis - also **10%**. Pain and its (mis)management (**10%**) is frequently discussed as sufferers of metatarsalgia sought tips such as orthotics, and people feeling disconnected with joint pain sought to reconnect. **7%** of the calls stem from those with symptoms and characteristics that mimic Marfan. **5%** emerge from pregnant couples seeking general advice; **5%** from aortic dissection; **5%** dental; **10%** chest; **7%** gastrointestinal, and **15%** other. The material and advice amassed during these calls has evolved into Dr Child's helpful Weekly Wednesday Casebook to be found on our website.

PS please note we at the Helpline are now returning once a week to the office to collect post and despatch merchandise.

Research Report ♥

As COVID-19 shuttered laboratories across the globe, condemning many scientists to 'dry lab activities', Dr José Aragon-Martin turned lockdown into 'heads down', honing his sequencing skills, analysing his datasets and submitting two manuscripts. *New Gene for Scoliosis*, with Université de Montreal PhD student Hélène Mathieu; and *LMOD1 – A New Gene for Ascending Aortic Aneurysm*, are now being reviewed for publication. And he welcomed the Trust's new summer student, Leshanth Uthayanan, with whom he worked on TAAD, that is identifying the genes involved in Thoracic Aortic Aneurysm and Dissection. See page 6.

Away from the physical lab, the Trust could only set the scene for interrupted and/or future projects. Now returning once a week to the Guy Scadding Building, Drs Aragon-Martin and Child can weave these dreams into reality

Ectopia Lentis (EL) *the dislocation or displacement of the natural crystalline lens.*

The lens of the eye is essential for focusing light rays on the retina. An inherited condition causes the lens to move out of place, becoming dislocated. This is because the small fibres which attach the lens to the eyeball itself are missing a protein component. The first protein to be discovered was Fibrillin-I which causes Marfan syndrome. The second gene involved is known as ADAMTSL4. However, many patients do not have a defect in either of these two genes.

Starting with patients for whom no gene has been found, we will study DNA samples from 28 patients with dislocated lens.

Aims

1. To discover new genes involved in Ectopia Lentis (EL) in patients with no cardiac involvement. This will help save patients' lives by separating Marfan patients (who need cardiac care) from patients who have only EL. It will save money for the NHS,



Dr Anne Child and Dr José Aragon-Martin

because patients who do not carry the Marfan syndrome gene have no cardiac risk, and therefore, follow-up will not be a yearly routine.

2. To create a fish model of the known genes in EL since the fish eye contains the same filaments which attach the lens to the eye. Then, study known mutations in these fish models to better understand how the gene error produces the disease. We can then use the fish model to revert the effect back to normal, using gene therapy, which in future can be used for patients.

Methodology

1. There is enough material in our present database of patients and samples to look for new genes for EL. We are probably the only group in Europe offering screening on a research basis and one of several groups who have published on the topic. A collaborative network of European and UK ophthalmologists has provided a retrospective study group, and will provide further cases with unknown cause. Twenty cases have been analysed to date, and published. Each patient is referred with family tree, and further family samples can be provided when requested to verify gene errors (mutations) discovered.
2. Samples are studied using whole genome sequencing. Gene mutations are verified and family studies performed in the Sonlee Laboratory under the direction of Dr José Aragon-Martin, PhD, Senior Research Associate, Imperial College whose salary and laboratory consumables are funded by the Marfan Trust. Cutting edge strategies will be used to identify disease genes, initially Overlap Strategy, De novo Strategy, and Linkage Strategy. Once an abnormal gene has been found in one of the 28 patients, all the patients' samples will be analysed to study the frequency of this gene in the EL population.

Zebrafish Model

1. The mutated gene will be introduced into a Zebrafish colony, and if dislocated lens is produced, this will confirm that the mutation is a causative

one, and provide a fish model to reverse the damage done to the filaments. This gene therapy could later be translated to the patients.

Resources Required

1. This work will be done in our fully equipped molecular genetic Sonalee Laboratory. Zebrafish models will be studied in the fish facilities, housed in Imperial College.

Funding for PhD Student

1. This project is perfect for a PhD studentship of three years, and we are seeking funding for the first year. The PhD student will initially be taken on as a technician at salary scale Grade Level 1a £21,570 for the first year. Funding for subsequent years will be sought from other donations.

José's Wish List ♥

It has been a year since we moved to Imperial College. We were welcomed straight away, and they allowed us to share and occupy a laboratory bench as part of a large laboratory group that was already based there. This arrangement was perfect, but then COVID-19 struck. Things moved very quickly, and we were not allowed to go back to work in the lab because of the lockdown. During this period we were granted a new laboratory location where we can now put Sonalee Laboratory back in shape. Sonalee Laboratory managed to move as much equipment as we were allowed but we are in need of crucial pieces of lab equipment:

1- Microwave (£57.00) (Microwave 800W)

2- Gel Visualiser (£7,787.00) (GelDoc Go)

We must use Sanger sequencing in order to validate variants that we found in our research. We can accomplish this by doing PCR, which needs to be run in an agarose gel, and this gel must be visualised with special equipment to assess if it can be used for further analysis or needs to be repeated. The agarose gel is made from heating agarose powder with a liquid mixture in a microwave, and the special equipment to be used to visualise this type of gel is called gel visualiser.

3- A laptop (£5,024.00) is part of the 2020 wish list that will help with Next Generation Sequencing data processing and analysis. The current laptop has been working overtime for the past 7 years and it is time to upgrade to a more helpful one.

Research Strength

1. This work is very important, since as far as we know, no other genetic research group is studying this topic of EL. It will define anatomic components of the lens fibrils, exclude Marfan syndrome, provide a basis for screening children of a patient affected, and a basis for future research to define preventative treatment.

José's Wish List



4- The Sonalee Laboratory needs an under the counter fridge (£400). The one we have right now is not working properly (either too low or too high temperature even though the setting is in the right place). We need the fridge to maintain consumables that need to be between 4C to 8C.

5- DNA quantifier (with fluorescent measurement). (£8,970) DNA needs to be quantified so we can use it in the right amount in research. If we use not enough DNA the project might not work, if we use too much the project might not work and also we will be misusing a precious sample that we might need in future projects. Also, it can quantify more accurately DNA when using fluorescent dyes, which is imperative to use when sending DNA samples for NGS [Next Generation Sequencing]. Then we analyse the results when they return.

6- 3x Bench stands for our bio-bin pipette upright boxes (£20 each, £60 all 3). The disposable tip bins we use are not stable on the lab bench and they can be knocked over spilling their content, making a mess and also making the lab less health and safety for other researchers. We need one for the PCR hood, and two for the laboratory bench.

Leshanth Uthayanan ♥

Marfan Trust Summer Student



Hello! I'm Leshanth, a medical student at St. George's University of London, shortly to intercalate in Cancer Research BSc at Imperial College. Having finished my second year, I wanted to do a summer placement to learn and develop skills which will be important for my academic future. I was looking at many

different research placements available on the internet and getting in contact with different institutions but unfortunately, due to the pandemic, a lot of research and consequently its funding (to my surprise) came to a halt. Many scientists were lost with what to do next as the future was bleak and therefore, I struggled heavily to find a placement. However, owing to the Marfan Trust's previous strong link with St. George's, I discovered the lab via my university and therefore was very fortunate to get in contact with Dr José Antonio Aragon-Martin from the Sonalee Laboratory who consequently became my mentor. Whilst I had strong interests in genomics and data applications, I was only briefly introduced to Marfan syndrome during my studies. I was excited that I managed to find a suitable mentor who could introduce me to the lab's research as well as train me to become a scientist. Therefore, undergoing this project has certainly been very influential in getting me more involved with the Trust.

My summer project mainly involved identifying genes which are involved with Thoracic Aortic Aneurysm and Dissection (TAAD) as well as Ehlers-Danlos Syndrome (EDS), both of which have similar characteristics with Marfan syndrome. The aim was therefore to get a big database of all genes involved which may be involved with the diseases. Due to coronavirus restrictions as well as the relocation of the lab, the task mainly consisted of literature search. I had to read through a mountain load of papers in the first cycle and identify genes that may have some involvement, if any with the diseases mentioned and record them. An example is the ACTA2 gene which is well known for its association with TAAD. I am now currently doing another cycle but now going through genes that are not in the confirmed gene panel and justifying why they should be added. An example is the LTBPI gene which has been in a study proven to be associated with TAAD, but we do not fully understand the pathways (upstream/downstream) involved. Along with

these tasks, I was also helping Dr José Antonio Aragon-Martin with some of his other tasks when needed. One week, I had to help him briefly summarise the current knowledge regarding the genotype-phenotype association in Marfan syndrome. The main aim of my task is to create a gene profile for these diseases. This not only helps us understand the genetic makeup of the disease, but also the consequent proteins involved in the downstream pathways and therefore gives us a better understanding of the disease pathology. Along with that, another main use is that it allows us to find biomarkers that will be very useful in the future just not for diagnosis but also to differentiate between other diseases and to provide optimum treatment to patients once we get a better understanding of the pathology. Once I have completed this, I would re-direct my attention towards understanding about the genotype-phenotype association of Marfan syndrome in greater detail, and if successful, writing that up into a literature review. Along with that, I would also very much enjoy gaining laboratory skills with the help of Dr José Antonio Aragon-Martin.

I am very happy to be part of this team at Marfan Trust. It gave me an interesting insight into the complexity of a single disease, and I helped them with some of the lab's logistics. Along with this, whilst I intended to purely volunteer, the Trust has been very kind to assist me with funding donated by the **Girdlers' Guild**. The fund not only was very helpful for me during the pandemic but also will help open up more opportunities for me to explore. This goes to show how impactful and significant your donations are to this Trust as we try to tackle this disease during these times. This, we are grateful for. Dr José Antonio Aragon-Martin and Dr Anne Child have both been very supportive and helpful throughout the project and have played key roles for the development of my skills from programming, data collection, analysis and literature search. They have also introduced me not just to research skills but also knowledge and general skills that will be very important throughout my life. It was refreshing working with the team, where I gained a new perspective about the world of academia. The struggles, politics and regulatory policies that the laboratory had to overcome has been eye-opening; definitely gave me a new insight and something that I will remember in the future.

The main goal of the Marfan Trust studentship programme is to influence and recruit more scientists into researching the disease. I believe that their goal has been successful as I intend to continue my time with what I see as a new chapter for the Sonalee Laboratory. Thank you 😊

Managing the medical manifestations of Marfan syndrome is much emphasised by professionals, less so are the condition's psychological and practical implications. This 'imbalance' is addressed below.

A Guide for Young Adults with Marfan Syndrome ♥

Visually vibrant, our newly published Young Adult Booklet is a vividly practical guide to living life with Marfan syndrome. From navigating education, sex and relationships to shopping for the taller, leaner end of

Psychosocial Aspects of Marfan Syndrome ♥

By • Dr Dieter Benninghoven

“Let Marfan syndrome be the passenger on your bus, not its driver”

With emphasis naturally placed on its medical management, the clinical approach to Marfan syndrome has dwelt far less on the condition's psychosocial ramifications, of which there are many. To represent this 'invisible' aspect of the syndrome, the mental manifestation, we invited leading German psychologist Dr Benninghoven, from the University of Luebeck and the Muehlenberg Rehabilitations Clinic to our recent Information Day.

Opening the afternoon session, Dr Benninghoven spoke immediately of his clinic's long-established – and prescient in retrospect - treatment regime. Since 2014, the Muehlenberg Rehabilitation Clinic with the University of Hamburg has welcomed groups of 10 to 12 Marfan patients for a three-week rehabilitation camp within a multi-disciplinary context. In doing so it has gathered precious empirical data that is otherwise rare. Whilst research into the medical consequences of Marfan syndrome has burgeoned over the years, studies of the syndrome's attendant psychological effects are conversely few.

Empirically speaking, it is known that Marfan syndrome as a chronic illness can impair one's quality of life - socially, physically and psychologically. Curiously this does not correlate to the severity of the syndrome's manifestation in the individual. Depression tends to

Contents

- What is Marfan Syndrome?
- What causes Marfan syndrome?
- Diagnosis & where to obtain it
- Clinical features & modern management
- Relationships & Sex
- Mental health problems
- Psychosocial aspects
- Questions & Answers (careers, travel etc)

the spectrum, it is a compact yet comprehensive trove. Please see back page for order details.



first set in during Marfan adolescence when body image insecurity is acutely felt, and a reluctance to comply with doctors' recommendations is similarly felt. The prospect of a life-limiting condition, the feeling of 'otherness' amongst your peers, all of this is a lot to contend with when navigating teenage hormones. As Marfan folk grow older and form relationships, the possibility of children and the attendant potential of passing on your genes is worrisome. 50% of Marfan patients marry and go on to have children. In the context of careers, Marfan syndrome can usher an earlier-than-normal retirement and limit your physical involvement.

To live fully and happily with Marfan syndrome, Dr Benninghoven advocates an attitude of acceptance, whilst stressing that knowledge is power! Become intimately acquainted with your condition! See below for his top tips. And in the meantime, his three-week camps in Northern Germany stimulate physical and psychological well-being. If only we had more here.

* Accept what you cannot change. * Accept your fears.
* Continue to pursue your personal goals * Engage in active coping style * Ask for information * Connect with other affected people * Practise Mindfulness – is your Mind Full or are you Mindful? * Clarify personal values * Accept reality gaps.

For more information please email info@marfantrust.org

Silent Auction ♥

Let your imagination roam free with our Silent Auction. 2020 may be keeping us largely confined and indoors but all the better to shop and dream from ... your sofa! It's time to restock your jewellery collection, reboot your leisurewear and plan those trips in 2021 ... <https://www.32auctions.com/MarfanTrust2020>

Amongst a trove of offerings are a holiday on the English Riviera, a tour of the iconic Morgan Motor Factory and vouchers to spend freely on Amazon.



The Daniel Finkletaub Prize ♥



Natasha Anno-Barnieh



Carole Finkletaub

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

**Winner of the Daniel Finkletaub Prize:
Natasha Anno-Barnieh - Pain Management
in Marfan Syndrome**

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

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

Focusing on the pain commonly felt by Marfan patients, our 2019 summer student Natasha Anno-Barnieh investigated its management in her award-winning thesis. From a widely circulated survey the hip, head, abdomen and back emerged as the most acutely afflicted areas, and the pain therein often undermanaged. 60% of Marfan patients reported experiencing daily pain for periods longer than seven days. Pain medication had been used by 43%, physiotherapy 23%, and joint supports 16%. Associated fatigue was reported as the most challenging problem for working adults.

Head pain typically emanated from migraine, dural ectasia, glaucoma and sinusitis. It was reported that Migralve or Imigran relieved migraine, while lying flat can alleviate dural ectasia-induced pain. Sinusitis may occur in later years because of the small diameter of drainage passages, and

large physical features such as adenoids and tonsils which may have to be removed in order to preserve hearing, and prevent endocarditis from recurrent streptococcus infections. Antibiotics are recommended.

Suggested prevention and/or treatment for joint hypermobility pain – the most ubiquitous - is rest, analgesia, supports, while appropriate bed and seating are important, as is gentle, regular exercise.

Natasha concluded that awareness amongst clinicians and patients of techniques for pain management must be

increased. General practitioners can now refer to regional pain clinics to find management solutions.

Natasha has since graduated from St George's University Hospital in London and joined the Princess Royal University Hospital's A&E department, where she is witnessing an alarming surge in coronavirus admissions. We thank Natasha for her work and wish her good luck in her future career. Her thesis is being adapted into a new pamphlet by the Marfan Trust.



In 2020, the Girdlers' Charity once again supported the Marfan Trust with three generous grants.

A remote working coronavirus grant (£1500) was used to purchase essential office equipment for our staff working remotely as a team. Three summer studentships (£1000 each) helped support three medical students working virtually on projects since the laboratory was temporarily closed during lockdown. Each student is now fully aware of Marfan syndrome, and has contributed to publications in preparation. Lastly a donation of £1400 was granted to help Darren McDean, who is himself affected with Marfan syndrome, to co-write a booklet for patients aged 16 to 24, a guide for young adults with Marfan syndrome. This was funded for printing and distribution, and details are on page 7 and back page.

The history of the Girdlers' Guild is fascinating. They are one of 120 livery companies of the City of London, and founded in the 14th century. Their trade was the making of decorative belts from which both men and women hung their purses, knives and anything else they needed to have about them. This trade died out as trousers with pockets came in during the 16th century. However, the Guild had acquired property, and the funds are now used for charitable benevolence, and active support of selected charities.

We are extremely grateful for their kind support during 2020, a year when decreased donations were inevitable due to cancellation of large fundraising activities such as the London Marathon.

Fundraising Update for Newsletter ♥

As with all charities, the Marfan Trust had to transform, innovate and adapt to the changing times. Embracing the digital age and ensuring our supporters have accurate and up-to-date information and advice has been paramount to us.

We focused on resurrecting our social media and creating educational content for our supporters, which have all been very well received.

"I would like to thank the Marfan Trust for the regular emails, especially during the lockdown. They are very informative and explain a lot of conditions that I and some other members of my immediate family had that were totally missed as being unconnected with Marfan's."

Quote from a supporter

A lot of time and energy went into creating an exceptional conference for all to enjoy. 91 supporters attended and raised

an amazing £995. The income generated is being used to provide support, research and educational literature to those affected by Marfan syndrome. A big thank you to all the attendees for purchasing a ticket and making the conference a success.

"Thank you to all the speakers and organisers who made this day possible. It was most informative and good to hear so many varied experiences and questions from the attendees. Thank you. You all deserve a well-earned rest."

Quote from a supporter



Gurpreet Madan

Although many supporter fundraising events had to be cancelled, we've had some wonderful supporters who planned their own bike rides and runs for the Trust (more details are shared in this newsletter) for which we are truly very grateful. The support, love, feedback, shares that we have received over the last few months is what motivates us to deliver better services and support.

We have events coming up this year and could really do with your support:

- **Act Now, Save A Life Campaign** (running from 29 Oct to 11 Dec)

The Sonalee lab relocation has prompted the Marfan Trust to launch an appeal for £5,000. As the sole UK charity funding its own medical research projects to aid the diagnosis and treatment of Marfan syndrome, we hope you are able to support us [see our social media pages and website for more, and overleaf for].

- **Silent Auction** (launched on 17 Nov and to end on 11 Dec)

Still thinking about those Christmas gifts and wondering how you will purchase them during the lockdown, well fret not! Marfan Trust has got you covered. Please keep a look out for your emails, our website and social media to join in our Silent Auction. A massive Thank You to those that donated Auction Prizes. Your support is crucial in helping us bridge the financial gap that has materialised due to Coronavirus. [see website and social media pages].

Other ways to support us:

- **Keep things interesting and entertaining by setting up a Fundraiser**

Some ideas: a binge of your least favourite TV show, a live stream of you cooking a family recipe, sharing a unique skill that you might have like yoga, dance, arts crafts. Be creative and get in touch for more ideas!

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

- **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

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



Sonalee Laboratory ♥

The story behind the Sonalee Laboratory is one of sadness and hope ...

Although Sonalee's Marfan syndrome was not recognised until later on in life, she felt its impact from an early age. As a child, she suffered with scoliosis. For several years, she wore back braces - the Boston brace in the early years and the Milwaukee brace in the later years - to try to correct this. However, when this was unsuccessful, she underwent two spinal fusion operations, at different stages, for this issue. Despite these problems, she excelled at school and went on to qualify as a doctor from St George's Hospital Medical School in June 1998.

Sonalee had a stubborn streak; this inner drive helped her to make the most of the short life she had. She would not allow anything to hold her back. She loved horse riding from childhood, a skill she continued to enjoy into her adult life. She enjoyed travelling, despite the reservations her parents had about some of her trips, (a solo backpacking trip to New Zealand involving white water rafting, glacial walking and hiking in the mountains comes to mind!) Closer to home, she enjoyed spending time with friends, listening to



music, going to the cinema and the theatre, looking after her pet cats and maintaining her extensive collection of cuddly toys. All in all, she enjoyed her life.

Whilst on duty as a junior doctor on 28 June 1999, Sonalee suffered an aortic dissection. Although she was transferred to St George's Hospital for immediate surgery, she passed away after suffering a cardiac arrest on the morning of 6 July 1999.

Having experienced first-hand the impact that Marfan syndrome can have on a loved one, we know how important it is that those with this condition, have it recognised promptly and are able to access treatments that will prolong and improve the quality of their life. Sonalee's father was a Consultant in Emergency Medicine, so as a family we had the advantage of having a ready source of medical insight into Marfan syndrome; not all families of those with the

condition will have this though, so it is vital that they have access to information and support about the condition. Over the past 20 years, The Sonalee Laboratory have been doing sterling research work into Marfan syndrome and many families are benefiting as a result. That is why we are so pleased that the Marfan Trust continue their work in these areas.

(The meaning of "Sonalee" is "Pot of Gold")

Marfan EU: Together We Can ♥

We are not an island ... We have left the EU, but not the Marfan EU.

We remain members of the Marfan Europe Network, a coalition of organisations supporting people affected by Marfan syndrome or related disorders. Founded in 1991, the motto of Marfan Europe Network, "**TOGETHER WE CAN!**", encapsulates its underlying ethos which is to facilitate a free-flowing exchange of information between member organisations.

Members' meetings are organised every second year. These meetings include medical lectures as well as discussions and

exchange of experience between the different associations. A separate **youth group** meeting gives young people affected by Marfan syndrome the opportunity to exchange their personal experiences and tips and tricks for everyday life. An annual summer camp for young people is held in a different EU country every year.

Besides the biannual meetings, Marfan Europe Network keeps in touch with its member associations by **regular newsletters** and emails sharing all information that might be interesting for their members.

<https://www.marfan.eu/>



In Memory ♥

Don Beer ♥ James Bond ♥ Graham Culver
♥ Kenneth Edwards ♥ Jake Fearnside ♥
Timothy Fyffe ♥ Dr Christine Morris
♥ Paul Taylor

Jake Fearnside - by his sister, Jemma
Jake Fearnside passed away on 31st October. He was a computer games designer from West London, who worked for the last eight years for a subsidiary of Sega Games. Jake's Marfan diagnosis led to other family members being tested, and two other relatives are now under the world-beating care of the cardiovascular team at Barts Hospital. Jake travelled extensively, with regular trips to the US with friends and family, but Jake's main passion was Chelsea FC. He was a regular in the Matthew Harding stand at Stamford Bridge and spent many hours following his beloved team through thick and thin, all the way to the Champions League Trophy. Through numerous surgeries over the years, Jake was supported by his mother, sister, nephew and a vast network of amazing, lifelong friends. Jake was always the "go to" person if anyone was



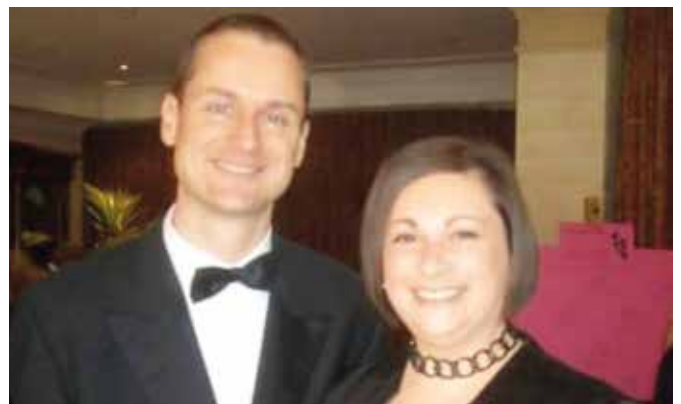
having difficulties in their life, and he counselled us all through cancer, bereavement, divorces, redundancies and similarly life changing events. He is irreplaceable and will be sorely missed by everyone who knew him.

Feats of Fundraising ♥

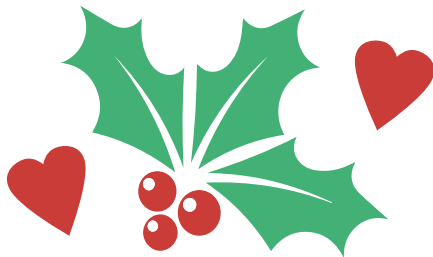
Defying nature's unyielding elements and surmounting lockdown's limitless limitations, our intrepid fundraisers climbed mountains, walked miles, cycled even further and built aircraft to raise funds for the Marfan Trust. At a challenging time for charities, we feel especially indebted to each and every one of you!

Going the Distance

On the cusp of performing his first marathon in 2009, experienced charity runner **Robert Austin** died suddenly at 39 of an aortic dissection. His widow, **Danielle**, has since realised her late husband's ambition, completing the Tel Aviv marathon in his memory, on the 10th anniversary of his death, raising over £3,000 for the Trust. Thank you so much, Danielle, our loyal fundraiser; for your enduring support!



Danielle and Robert Austin



Flying High

Touching 6 foot 11 inches tall, avid aviation fan **Stuart Baker** ambitiously hoped to build a model aircraft with a wingspan greater than his own. Tragically he did not live to realise his dream. An undiagnosed heart problem forestalled his life in 2013 when he was only 46. Marfan syndrome was posthumously diagnosed. But his dream lived on. His uncle Brian Fuller has withstood illness in the intervening years to patiently piece together a remote-control Mustang aircraft left unfinished by Stuart. Fittingly, it now surpasses his nephew's preternatural wingspan and was unveiled at a Wings Gathering on 26 October, the 80th anniversary of the Mustang's first flight. Making a splash in their local newspaper, the Farnham Herald, Stuart's sister Lynne talked of the family's hope to sell this big flying machine to a museum or enthusiast with proceeds going to the Marfan Trust. Watch this space!



Stuart Baker

Fabulous Fundraisers ♥

Scaling the Summit for a Friend

Long loyal to the Trust since the death of their friend, Nick Joyner, from Marfan syndrome, the **Brockworth Rugby Club** in Gloucestershire has repeatedly raised thousands for our precious research. In March this year the Club lost another dear friend and member, Graham Turney, to a sudden accident. Graham leaves behind his wife Sally, and young daughters Olivia and Lucy. In the wake of Graham's unexpected death and with restrictions imminent, Brockworth hastily convened a hill-climbing expedition in his memory, scaling the summits of Gloucester. To feel a sense of Graham during their expedition, they wore t-shirts emblazoned with his arm tattoo. Their climb and the subsequent sale of the t-shirts, along with individual



Brockworth Rugby Club

donations, have amounted to nearly £3000. Graham's widow Sally very generously decided this should be donated to the Marfan Trust. We are eternally grateful.



Graham Turney

Ahead of the Game

By presiding over an evening of food, fun and friends, **Natalie Lyall** has raised £180 for the Trust. Marfan syndrome is something she has felt intimately since her husband Mark was diagnosed, and to help him, and others like him, she hosted a sit-down meal complete with guessing games, roulette and a raffle. A festive way in which to raise money. Thank you!



Natalie Lyall and friends

Milestone Upon Milestone

Attributing his success to a "high boredom threshold", **Mark Rainsley** recently completed a 50-mile run on the eve of his 50th birthday! Surpassing his original target of £200 by some £1,500, Mark ran solidly for 11.5 hours, navigating the vagaries of the south-west's hilly coastal path, supported in short stretches by friends but running largely alone. Not only was Mark forced to transcend tricky terrain and weary bones but he also had to negotiate the shifting rules of lockdown and the burning down of his originally chosen turf, Wareham Forest. Streaming his feat live on Facebook, Mark delved into his innermost resources when he greeted first light, knowing his personal marathon would be followed by all!



Mark Rainsley



During his life Mark has undergone thoracic heart surgery and sight-saving eye surgery. In the context of this his run is all the more amazing.

Fabulous Fundraisers♥

The Roads Less Travelled ... Daniel Walker and Guy Padmakumara Potter

Disheartened by the cancellation of this year's London to Brighton Bike Ride, **Daniel Walker** and **Guy Padmakumara Potter** remained undefeated!

With his partner Kate Renoldson, **Daniel** had carefully planned a fundraising campaign, raising close to £3,000, a magnificent sum boosted by a generous £1000 injection from his generous employer. When disappointment struck, and the much-anticipated Bike Ride was deferred, Daniel and Kate plotted their own route. Without the closed roads of the public ride they followed a longer, quieter path from London to Brighton, taking 5.5 hours to cover the 94 kilometres, wearing their Marfan Trust t-shirts most stylishly, as you can see.

In honour of his late step-father and our former Trust Chairman Michael Carr, **Guy** meticulously devised his London to Brighton Bike Ride fundraising campaign, encouraging others to do the same! His hopes and aspirations were captured in our Spring newsletter. As it was, he and his sister Claire took the unofficial route! In Guy's words: "after setting off from Maidenhead train station at about 10am, Clare and I arrived at the family in Abingdon at about 8.30pm. It was dark by that time but our moods were bright, powered by the support we've received from friends and family. It was a beautiful day, ideal for cycling, and it saw us take in some of the lovely countryside and villages of the Thames Valley." Guy and Clare racked up the amazing sum £2,000 for which we are massively grateful!



Clare and Guy



Kate and Daniel

The Young Mountaineer

Driven to help others like him, 10-year-old Marfan patient Cobie enlisted his parents Jason and Paige in a charity climbing expedition to aid the Marfan Trust. Withstanding lashing rain, biting wind and extreme cold, the trio scaled Mam Tor in Derbyshire, raising £530. Cobie has now acquired a taste for hill-climbing and wants to repeat his adventure year after year.

These are just a few of the many magnificent fundraisers supporting our Trust. Please email your fundraising story to info@marfantrust.org



Jason, Paige and Cobie

Mohamed Abdelrazek ♥

A fourth-year medical student at Cardiff University, Mohamed occupies both sides of the doctor's consulting table. He is the first member of his family to be affected by Marfan syndrome and suffers its symptoms – visibly and invisibly - whilst working long shifts in South Wales's hospital hotspots. Wales felt the virus reawaken with full force in September as infections proliferated alarmingly. In the meantime, Mohamed has been operating quietly behind the fear-inducing headlines of 'broken' doctors and nurses that emerge daily in the newspapers, as he experiences first-hand the rapidly filling intensive care wards.

But he is really interested in COVID's manifestation amongst his own niche community of Marfan folk and would love to hear from you. If and when you have a rare, spare mo-



ment, please request a copy of Mohamed's COVID-19 survey from info@marfantrust.org. Your material will be treated and analysed anonymously.

Marfan Information Day ♥

Experts in the cardiac, skeletal and ocular manifestations of Marfan syndrome each spoke on 3 October, resulting in our most informative, successful conference. The talks are available separately for £5.00 each.

Please email info@marfantrust.org to request details.

PROFESSOR JOHN PEPPER

Consultant Cardiac Surgeon, Royal Brompton and Harefield Hospital NHS Foundation Trust

WHAT'S NEW IN AORTIC and MITRAL VALVE SURGERY

PROFESSOR GRAHAM STUART

Consultant Paediatric Cardiologist, Spire Bristol Hospital

AIMS TRIAL RESULTS (Aortic Irbesartan in Marfan Syndrome). IS IT BETTER, AND HOW DO WE OBTAIN IT?

MR AMAN CHANDRA

Consultant Ophthalmologist and Vitreoretinal Surgeon Southend University Hospital Foundation Trust

THE EYE IN MARFAN SYNDROME (DISLOCATED LENSES: CAUSES AND MANAGEMENT)

DR JOSE ARAGON-MARTIN

Research Director Sonalee Laboratory – Marfan Trust

SONALEE LABORATORY RESEARCH PROGRAMME (Ectopia Lentis, Familial Thoracic Aortic Aneurysms)

“It was a first-class line up with great use of the technology and excellent time management”

DR DIETER BENNINGHOVEN

Clinic of Psychosomatic Medicine and Psychotherapy, University of Luebeck Germany

PSYCHOSOCIAL ASPECTS OF MARFAN SYNDROME

PROFESSOR CHRISTOPH NIENABER

Consultant Cardiologist, Royal Brompton and Harefield Hospitals NHS Foundation Trust

NAVIGATING THE TRANSITION BETWEEN PAEDIATRIC AND ADULT CARE

MR IAN HUNT

Consultant Thoracic Surgeon and Honorary Senior Lecturer St George's Hospital and St George's University of London

PECTUS CHEST PROBLEMS: NEW TREATMENT

DR NATASHA ANNO-BARNIEH

Princess Royal University Hospital, Orpington, Kent

MANAGEMENT OF PAIN IN MARFAN SYNDROME

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!

www.marfantrust.org



Robin on a Snowy Branch



Three Kings of Orient



Nativity Scene



Christmas Wreath



Snow on the South Bank



All Occasion card



Under Guide for Young Adults with Marfan Syndrome

Response Slip (this can be photocopied)

Please return the response slip to the Marfan Trust by post to: Marfan Trust, Guy Scadding Building, Dovehouse Street, London, SW3 6LY. Please make cheques payable to the Marfan Trust; thank you for your support.

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

..... Postcode.....

Email.....

Please insert number of packs required in the relevant box
(Christmas cards are £4.00 per pack of 10 and an additional £3.00 for p&p).

Robin on a Snowy Branch with inside message reading
Season's Greetings and Best Wishes for the New Year
Dimensions: 13.5cm x 13.5cm

Christmas Wreath with inside message reading
Season's Greetings
Dimensions: 13.5cm x 13.5cm

Three Kings of Orient with inside message reading
Season's Greetings and Best Wishes for the New Year
Dimensions: 13.5cm x 13.5cm

Snow on the Southbank with inside message reading
Season's Greetings
Dimensions: 10.5cm x 14.5cm

Nativity Scene with inside message reading
Season's Greetings
Dimensions: 13.5cm x 13.5cm

A Guide for Young Adults with Marfan Syndrome
£7 (including p&p in the UK) **£12** (including p&p international)

Marfan Trust Emergency Card: £2.00 per card: No

Marfan Trust wristbands: £2.00 per wristband No

Marfan Trust T shirts: £5.50 each: Small..... Medium..... Large..... XL.....

Marfan Trust All Occasions Card: £4.00 per pack of 10 No

I am eligible for Gift Aid. Please give your name & address above.

Signed..... Date.....

Registered Charity Number:328070

Registered with

