



# friends<sup>®</sup> Together

Vol 7, Issue 1 Spring 2024

## A Night at the Museum

**What a night!** FOP Friends was delighted to have had the opportunity to collaborate with Dr Lucy McKay and Medics 4 Rare Diseases (M4RD) to host an evening reception: 'First do no harm: Expert reflections on a rare bone disease' at the prestigious Hunterian Museum at the Royal College of Surgeons in London.

The event had been imagined by Helen and Lucy, over five years ago. However, other demands, diaries and Covid delayed the project until last year when it was picked up and planned in earnest. Working with Dawn Kemp from The Hunterian Museum and Nadine Jeffries from M4RD, the event aimed to be insightful and thought-provoking, shedding light on the complexities and challenges faced by individuals living with any rare bone disease, not just FOP.

We were delighted to welcome a panel of esteemed speakers, from Professor James Triffitt who has dedicated his life to the discovery of the gene and furthering understanding of FOP, to Oliver, a teenager living with FOP. Professor Richard Keen spoke about clinical research and looking to the future; Chris spoke about being a parent and advocate of someone living with FOP; and Lucy Pratt spoke of living with FOP, the need for healthcare professionals to listen to the patients, and about holding on to hope.

*(continued on p2)*



FOP Friends exists to **help find a treatment and a cure** for the rare genetic condition fibrodysplasia ossificans progressiva (FOP), and to **support the families** affected by it.

Thanks to exceptional collaboration and openness from The Hunterian Museum at The Royal College of Surgeons of England, FOP Friends and Medics4RareDiseases held an event that told the story of so many rare conditions - compassion, perseverance and collaboration

leading to knowledge, community and hope. The speakers who all had different experiences with FOP were exceptional and moved the audience with their stories. They exemplified that even in the most

challenging of situations there is still joy and hope when we unite behind a single cause.

**Lucy McKay CEO of Medics4RareDiseases.**

To learn more about the work of Medics 4 Rare Diseases, and their advocacy for everyone living with a rare condition, please visit [www.m4rd.org](http://www.m4rd.org) or follow them on Instagram and Facebook.



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## A word from Alison



Welcome to our first newsletter of 2024! While I'm personally facing the obstacle of a broken leg, I've had the gift of time to reflect on our past achievements and more importantly to focus on our future as a charity.

As we look forward to spring and hopefully some sunshine, I'm particularly excited about our upcoming Conference. The remarkable lineup of medical experts, researchers, scientists, and pharmaceutical representatives from across the globe is a testament to the tireless dedication of our charity and the profound respect it commands.

As we approach the end of our financial year, part of my responsibility as a trustee is to assess our financial standing. In these challenging times, many small charities are facing fundraising hurdles. This is also true for us. We appreciate many families are finding the cost of living increases a real challenge, but if you are able to contribute in any way, we would truly appreciate it. Take a look at the article on Becoming a Regular Donor, where you'll find an easy scan QR code to get you started. Please also share this with your friends and networks and encourage others to contribute to our cause. Together, we can inch ever closer to the cure we all desperately seek.

I look forward to catching up with old friends and making new ones in Manchester.

Alison, FOP Friends Trustee

## A Night at the Museum cont..

There were guests from all walks of life, from patients, families, and friends, to researchers, healthcare professionals and members of the Royal College of Surgeons, making the event even more meaningful and impactful.

Attendees were able to visit the Hunterian Museum and meet Mr Jeffs, a gentleman from the 1700s who had lived with FOP. Healthcare professionals were also given a rare opportunity to visit the Anatomy and Pathology Museum to see another person who had lived with FOP and selflessly donated their skeleton to science to help future generations to better understand the condition.

Helen, Lucy, Nadine, and Dawn were rightly proud of the success of the evening: it had raised awareness about the challenges associated with living with a rare condition, as well as inspiring hope for a future where compassionate care and scientific innovation converges to improve the lives of those affected. It had succeeded in putting the individual experiences and voices of patients and their families at the forefront of the conversation, allowing all attendees to connect on a deeper level, fostering a sense of community and solidarity in the face of adversity.

The evening was a testament to the power of collaboration, empathy, and advocacy in promoting health equity for all.

FOP Friends would like to say a special thank you to Dawn Kemp for so generously and enthusiastically welcoming us to the Hunterian Museum, to Nadine Jeffries for her hard work behind the scenes in making the event happen, to the incredible speakers, and to everyone else who contributed to make this a truly memorable event.



## STOPFOP Update

STOPFOP is the clinical trial FOP Friends has been supporting, from laboratory to the current clinical trial. We caught up with Dr Marelise Eekhoff, Principal Investigator, from VUmc, Amsterdam, to get a quick update. We are also excited to announce that Dr. Eekhoff will be attending our conference in May, with Vincent Verhey, sub-coordinator of STOPFOP.



STOPFOP is the project that is investigating the safety and efficacy of oral saracatinib, an ACVR1 kinase inhibitor. Despite the significant challenges it encountered at inception from Covid and Brexit, the study continues to persevere.

A group of 17 patients are actively participating through the Dutch site (Amsterdam UMC) and the German site (Klinikum Garmisch-Partenkirchen), and while the UK site in London (RNOH) still awaits approval, several UK patients have been included through the Amsterdam site in the interim.

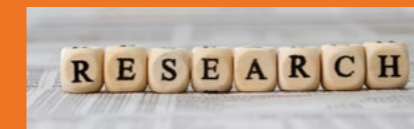
With the addition of these UK patients, the trial now involves participants from eight different European countries. With growing pre-clinical data on the potential of saracatinib, variant mutations could also be included after an amendment to the initial protocol was approved.

The trial's progress is exciting for both researchers and participants. There has been no serious drug related adverse events to date and all participants who finished the trial chose to continue saracatinib through a prolonged extension phase of the trial. The STOPFOP team receives great support from the FOP associations and have also attracted interest from people outside the community.

The team hopes to expand the scope of saracatinib to research in children, should their first data, that is expected later in 2024, be promising.



## Research update: Interesting news from the Patient-R project



In February, the findings from the Resilient Patient Project were published in the Journal of Bone and Mineral Research.

Several FOP specialists were involved in the project, including Professors Fred Kaplan, Robert Pignolo and Eileen Shore. The results from the findings were fascinating but it is important to note that the MMP-9 inhibitors have not been tested in people diagnosed with FOP.



Our newest trustee, Andrew Rankin, summarised the project's findings:

The Resilient Patient Project focused on a healthy 35-year-old man (called patient-R) who had classic FOP. Despite having the typical FOP gene mutation and a toe malformation since birth, patient-R has developed very little extra bone and can move almost normally. The researchers hypothesised that patient-R lacked a sufficient inflammatory trigger for spontaneous bone growth – or heterotopic ossification (HO). Tests on patient-R's blood showed lower levels of a protein called matrix metalloproteinase-9 (MMP-9) compared to other people with FOP and healthy individuals.

The publication details the observations in patient-R with mutations reducing MMP-9 production and activity, and multiple FOP mouse model studies inhibiting MMP-9 support a role for MMP-9 as an inflammatory trigger for Activin A-mediated heterotopic ossification in FOP.

The authors propose that reducing MMP-9 levels in FOP patients pharmaceutically could potentially protect against trauma-induced HO by decreasing the activity of the Activin A pathway.

However, more studies are required to fully understand the connection between MMP-9 and Activin A, and the exact role of MMP-9 in the trauma-induced formation of HO in FOP.

Additionally, there is the potential for off-target activity with an MMP-9 therapeutic due to the high similarity with other MMPs that have different actions and roles in the body.

Nevertheless, MMP-9 inhibition offers a potential novel additional therapeutic target for reducing HO formation in FOP patients beyond those currently in clinical development.

You can read a more detailed explanation and discussion of the report on our website:

[www.fopfriends.com/mmp-9](http://www.fopfriends.com/mmp-9)

## Back to the Board

It hardly seems a year ago since Helen attended her first in-person board meeting for the IFOPA, yet last month she headed for sunnier climes as she flew to Orlando, Florida. Unfortunately, it was only a short visit so there was no time to meet up with a certain famous mouse!

The Board meeting was held over two days and Helen found it inspiring to spend time with like-minded people who were all looking for ways to support those living with FOP whilst also searching for a treatment for FOP.

Two new Board members were welcomed, and FOP mum Rebecca Wallace took up her new role as Chair. The Board heard the IFOPA's team reflect on the successes of the past year, and their vision for the next 12 months. It was interesting to hear Danielle Kerkovich's research updates and it gave much hope to Helen as a parent.

There is so much great work going on in our small-but-mighty FOP world. We are truly lucky to be part of such a strong community, both in the UK and across the globe. We still have a long way to go until we get the treatment we all so desperately need, but we are going in the right direction and more importantly we have HOPE. **#strongertogether**



## Q&A with Dr Alex Chesover

At the end of 2023, the FOP team at the Royal National Orthopaedic Hospital were pleased to welcome Dr Alex Chesover to their team, bringing a specialist paediatrician on board. Dr Chesover has already met several of our FOP families in London, and attended our 'First, Do no harm' event. So, allow us to introduce Dr Alex Chesover...

### Where were you before you joined RNOH?

I joined Great Ormond Street Hospital for Children (GOSH) in 2021 as a Consultant in Paediatric Endocrinology. Since 2023, my time is equally shared between GOSH and the RNOH, and I have an increased focus on paediatric metabolic bone disorders.

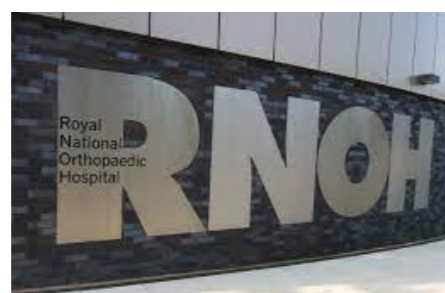
I qualified in medicine from University College London Medical School; completed paediatric speciality training in the East of England; and then a three-year fellowship in Paediatric Endocrinology and Diabetes at The Hospital for Sick Children, Toronto.

### When did you first become aware of FOP?

As part of the multidisciplinary and multiagency care of a newly diagnosed young person and their family. It highlighted the importance of interdisciplinary working, and the wide range of community support and specialist input for families living with FOP.

### How did you get involved in working with patients with FOP?

I lead a monthly postgraduate teaching programme for Paediatric Endocrinology and Diabetes at GOSH. One month, I focussed on FOP and was extremely fortunate to host Prof Keen, Ms Abou-Ameira, and Helen Bedford-Gay, who kindly shared their expertise and perspectives. I was encouraged to join the RNOH by the opportunity to lead the Paediatric FOP Clinic, working alongside Dr Bubbear and Prof Keen.



### What is the best thing about working as part of the team at RNOH?

The team at RNOH care, listen, and support one another. It is a friendly environment, and there is pride in delivering exceptional specialist care, training, and mentorship.

### Do you specialise in any other conditions?

I work in Paediatric Endocrinology at GOSH, and sub-specialise in metabolic bone disorders, which includes disorders of calcium metabolism, hypophosphataemic rickets, and osteogenesis imperfecta. I also have an interest in the care of young people in healthcare transition and have established a new Young Adult Metabolic Bone Clinic at RNOH.

### Are you currently working on any FOP research projects? What is your role in them?

Currently, my focus is establishing myself in the Paediatric FOP Clinic at RNOH and introducing myself to the FOP community. However, there are research opportunities on the horizon in which I will take a lead role at RNOH.

### What is one thing you can't get through the day without?

A smile.

### What is your dream holiday destination and why?

The mountains - wherever in the world - for the culture, the camaraderie, the peace, and the adventure.

Thank you so much for taking the time to let us get to know you Dr Chesover. We look forward to welcoming you to Manchester in May to meet more members of our FOP community.

## Have you booked on our Conference?

As this newsletter goes to print, we are planning the final touches for our fifth Conference at the Manchester Airport Radisson Blu Hotel.



While this is a UK event, we welcome families from around the world...here are already attendees from eight different countries!

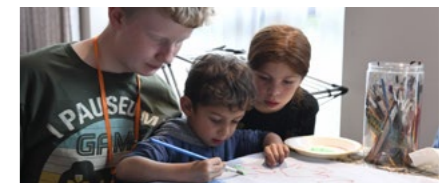
### So, what's on?

There will be presentations by FOP specialists from the UK and around the world. Those living with FOP will be given the opportunity to attend up to three clinical appointments: general, dental, and new for 2024, dermatological. Children and young people will have the benefit of our FriendZone again, and we are welcoming some new furry friends this year - therapy animals. We will hear from Nina Kundi, an emotional wellbeing coach and there will be a presentation 'Caring for the Carer' for those who in an unpaid caring role, with guidance on how to make sure you are receiving the help and support you need. We will be hosting our



Family Dinner with a quiz, with the opportunity to just relax and enjoy the company of those who are on a similar journey to you.

Everyone who has attended our event in the past always tells us that their biggest take-away from the event was the friendship and the realisation that you do not need to face this journey alone. For further information, scan the QR code now. We look forward to welcoming old friends and new to Manchester!



## Great North Run 2024: Places now available

The Great North Run is back again - on Sunday 8th September.

The GNR is the world's largest half marathon. And if you are lucky enough to take part, you will never forget the sensational atmosphere as you make your way around the course!

Everyone who takes part says it's a run to remember!

Be a part of #TeamFOPFriends and do something amazing!

For more information and to book your place visit: [www.fopfriends.com/gnr/](http://www.fopfriends.com/gnr/)



## Brilliant Batchwood!

In 2023, Will Smeulders chose FOP Friends as his charity when he assumed the role of Captain at Batchwood Golf Club in St. Alban's.

Will knew Lexi Robins and her family through the golf club, and when he learned of Lexi's journey ahead and the challenges those living with FOP face, he wanted to do something to support the research into a treatment for everyone affected by FOP. As Lexi's grandad Graham has been a member of the Golf Club for many years, it was a cause close to many people's hearts. Throughout Will's year of captaincy, a series of fundraising events were held, including quizzes, games, and raffles. Any 'fines' incurred by members also went to his chosen cause. They held golfing events too, including the Lexi Cup.



Last year the club raised £770 which was then fund-matched by local company Cam Wells totalling £1,540. In February of this year, the members held their final event. The grand total raised by Batchwood Golf Club was an amazing £10,329!

### Lexi's mum, Alex said

We are so appreciative of everyone's support, it means so much. I'd like to say a special thank you to my dad who has helped so much this year.

### Will says

It has been an absolute pleasure raising funds for FOP Friends. It's been a great year as Club Captain 2023 and I have been truly amazed at the generosity of the members and others who have joined in with such a worthy cause. I'm proud that our total is the biggest fundraised total ever for the club. Thank you to everyone who has wholeheartedly supported our efforts this year.

# Salford University Project Update

It has been an absolute pleasure working with the students from the University of Salford. It has allowed us to raise awareness of FOP with the next generation of occupational therapists and engineers.

Rachel Winnard (who is living with FOP), Helen, Leo (Oliver's brother) and others visited the team at the University of Salford at the beginning of February to catch up with the progress of their adaptive tool. Rachel had requested something that would allow her to eat simple food such as sandwiches and pizza independently, she also wanted a table that would be the perfect height for her in her chair. The engineering team had some prototypes for her to try out. Rachel gave some suggestions for improvements to the design, and we'll be going back for the final product very soon. We can't wait!

Through our collaboration with the students, Helen was invited to attend the Occupational Therapy Conference at the University. Kathryn, Vicky, and Clare had prepared a presentation about their projects and their key learnings, and they shared this with their peers and other students attending the conference. Their presentation was informative and insightful, with learnings which will be transferable to supporting any person with higher care needs.



## Global Awareness Campaign

**We will be combining forces with the other patient organisations around the world to take part in a Global**

**We will be combining forces with the other patient organisations around the world to take part in a Global FOP Awareness campaign. Look out for our posts and facts on social media and help us to raise awareness beyond our community by sharing the posts on your platforms. It only takes a moment to share but makes a massive difference. #sharingiscaring**

If you want to raise awareness of FOP throughout the month of April and beyond, why not order this year's Global Awareness Day tee here: [www.fopfriends.com/shop](http://www.fopfriends.com/shop)

## FunFeet4FOP

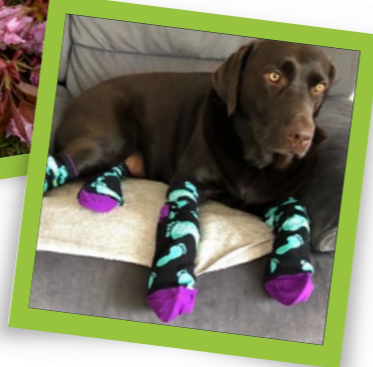
**April is FOP Awareness Month and, with that, we will be hosting our annual #FunFeet4FOP campaign!**

FunFeet4FOP, which we first launched in 2006, is an online campaign that aims to raise awareness of FOP through posting fun pictures of feet! Individuals with FOP usually have 'turned in toes' or 'funny toes'. This classic sign is often not recognised by healthcare professionals, due to the rarity of FOP and the lack of awareness of the common physical FOP symptoms. This can lead to misdiagnosis and a long journey time to a correct diagnosis.

We will continue our active campaigning this year on our social media platforms so keep an eye out on those spaces! Our community has been brilliant in their participation over the years, and we are sure this year will be no different!

So, starting thinking about how you will make your feet fun for FOP Awareness Day on 23rd April!

*Here are some of our favourite snaps from past years...*



## Engraver Gamer Gifts

While many of us know him for his streaming skills, Engraver Gamer took on a more magical role as Santa this Christmas to treat the children in our community with deliciously wonderful gifts!

He worked closely with Helen to compile an assortment of gifts for all the children, including hand-decorated gingerbread biscuits from The Biscuiteers, a hot chocolate mug gift set, and an Amazon gift voucher! A Raid Train fundraising event, hosted by Engraver, raised an amazing £1,062.19, with contributions from all over the world.

All the children were over the moon to receive their presents – just look at their big smiles in the photos!

Thank you again to Engraver Gamer for his incredibly generous and thoughtful efforts – his support to FOP Friends has been overwhelming and we cannot thank him enough!



## Walkden Gardens' Winter Wonderland

In the wintry embrace of December, the intrepid Bedford-Gay family ventured forth into the crisp air, undeterred by the chill, to grace the local Walkden Gardens Christmas Fair with an FOP Friends stall.

Amidst the festivities orchestrated by the Friends of Walkden Gardens, where carols danced around the Dovecote and the aroma of mince pies and mulled wine enchanted the senses, the community gathered in harmonious celebration.

It was amidst this backdrop of communal joy that Leo, with his boundless creativity, fashioned a charming 'boy' to showcase our splendid merchandise, a testament to his ingenuity and the spirit of the season. And as if touched by a sprinkle of winter's magic, a gentle dusting of snow adorned the landscape, casting a spell of enchantment upon all who partook in the merriment, heralding the arrival of the Christmas season in all its splendour.



## The Willow's Women's Institute, Sale

Many of our friends and followers will know of Janet Plumb, one of our longest-serving supporters. Janet was Oliver's childminder when he was just two years old and has been by our side since.

Janet has been a longtime member of The Willow's Women's Institute group in Sale, Cheshire, and nominated FOP Friends to be their charity of the year. At the beginning of the charity year, Janet gave a presentation to the members about FOP and how it affects people. Through their weekly raffle, the group has raised a fabulous £501.50 for Oliver and all his friends! Helen was honoured to be presented with the cheque last week by the chair Sarah Turner-Wilson and Janet. Oliver and all his friends are so thankful to have been your charity of choice for the year.



# Apoiando uma criança com FOP: um guia prático para sua jornada de aprendizagem

For those of you who don't speak Portuguese, this translates to 'Supporting a child with FOP: a practical guide to their learning journey' and is first of several translations of the original schoolbook.



This newsletter has been made possible thanks to a grant from **The Zochonis Charitable Trust**.



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We hope you enjoy receiving our newsletter. However, if you no longer wish to be on our mailing list, please email us at [info@fopfriends.com](mailto:info@fopfriends.com)

Kryscia Napoli, Dr Patricia Delai and Helen have been working together on the Brazilian adaptation of the school guide, ensuring it is up to date and relates to the Brazilian school system. There are also updated photos showcasing more of our FOP superstars and celebrating their achievements. Helen says,

It's been a fabulous project to be involved with. I love being able to speak with other people around the world, sharing ideas. It's been really rewarding to be part of such a motivated and enthusiastic team and see the project come to life.

They are working on the final touches in the guide, with the official release on the horizon. Excitingly, this is the first of several adaptations planned, with other languages and countries in the planning. **We would like to thank Regeneron for a generous grant which has made this project happen.**

## A bit of fun...

Did you spot our version of an "Easter egg" hidden in this edition of our newsletter?

Like most people, we have been playing with AI text so thought it would be amusing to use AI to re-write one of our features in the style of a favourite tv character of the trustees.

**Did you find it?**

**Can you guess the character?**



## Become a Regular Donor!

**Have you considered becoming a regular donor to FOP Friends?** We appreciate that things are challenging for many of us right now, with costs seemingly increasing week on week. There really does seem to be such a lot of month left at the end of the money!



However, maybe you grab a take-out coffee or two throughout the week and could consider turning one into a regular donation to FOP Friends instead? We are extremely grateful for any and all donations - big or small. If all our supporters were able to set up a regular donation for as little as £2 a month, it would soon build up into a big change. Every contribution we receive allows us to continue to support the research into FOP as well as support the FOP community and their families. With your continued support, we can reach even greater achievements in our endeavours to discover a treatment and ultimately a cure for FOP.

If you want to find out how your donation will make a difference, please get in touch

**Scan the QR code to visit our website** and find out how to set up your regular donation. Sharing across your networks and social media platforms is another great way to help your charity and can be done easily in just a few clicks.