fop friends®

together

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www.fopfriends.com

Bringing *together* the FOP community

You're reading the very first of our new-look quarterly newsletter – *Together*! Fibrodysplasia ossificans progressiva (FOP) may only affect a very small number of people, but we have a brilliant community and together we are making progress towards a cure. *Together* aims to share what's happening in the FOP community and to connect patients, clinicians, researchers and funders. It will bring you regular updates on research, events and the fantastic fundraising efforts of our supporters, as well as views and personal stories from FOP patients and their families.

We want you to be a part of it, so please share your news and thoughts about FOP with us for future issues, and let us know if you have any feedback or suggestions. If you haven't already, you can sign up on our website to receive your own copy of the newsletter. We look forward to hearing from you!

INSIDE

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.

FOP Friends is a Registered Charity in England and Wales 1147704

A word from Chris

Welcome to the very first issue of our FOP Friends® Together newsletter.

We're delighted to share this edition with you: firstly because we think it looks amazing; secondly, because it showcases the highlights from our 2016 Gathering; but most importantly because it celebrates the amazing things our small but special community are doing in the quest to find a treatment and a cure for FOP.

We're all used to using social media and reading everything on a screen, but we thought we'd go a little 'old school' and get a quarterly newsletter into print. We are therefore thrilled to be fully funded to produce this newsletter for the next two years.

This is your newsletter, whether you're an FOPer, a family member or friend, a medical professional, or simply someone who has chosen to support us on our journey. We hope you'll join in and contribute content or make suggestions for future issues. We want this to be filled with your stories and your news.

Although we'll be doing limited print runs for our community and supporters, a PDF version will be available for you to use at every fair, line dance, garden party, clothes swap and bingo night. With your help it will give others insight into FOP and our dynamic community.

We may be only a small charity, but we're now making a big difference, and this is down to the continued support from everyone who listens to our stories, raises a few pennies (or pounds), and who, most importantly, cares. We thank you all.

Together we are strong; together we have hope; together we will find that treatment.

Chris Bedford-Gay
FOP Friends Chairman

Tough Mudder Madness

WEST LONDON – APRIL/MAY

FOP Friends has some pretty tough and determined supporters who proved they weren't afraid of a bit of mud and pain if it could help make a difference. We had supporters taking part on both days of the West London Tough Mudder weekend, and together they raised over £2,000.

"I thought it would be a great opportunity to raise awareness for FOP itself and raise money for a charity that does such great work very close to home." – Marcus

"It was a great day! We tunnelled, scaled, ran, walked and swam our way through the 11 mile obstacle course in a little over four hours, grinning all the way, spurred on by the knowledge that we were supporting FOP Friends with all their great work and amazing community." – Craig

See what other fundraising feats our supporters have been up to on page 7. And if you fancy being a Tough Mudder yourself, visit www.toughmudder.co.uk.

23 APRIL

#FunFeet4FOP

This year it's 10 years since the discovery of the ACVR1 gene that causes FOP was announced. In April we marked this fact and International FOP Awareness Day with a #FunFeet4FOP campaign on social media, asking people to share pictures of their feet to help us raise awareness. Why feet? Because the first sign of FOP is malformed big toes. This classic sign is seldom recognised because of a lack of awareness of FOP, and this leads to FOP being misdiagnosed in many cases (often as cancer). People from around the world shared pics of their decorated feet or flamboyant footwear. We even had paws and hooves helping with our efforts!

Find us on social media
FOP Friends **@FOPFriends**

NEWSflash

Lack of Oxygen Link to FOP Flare-ups

May

Scientists from the Center for Research in FOP and Related Disorders at the University of Pennsylvania have shown that tissue cells starved of oxygen (a state known as hypoxia) amplify the formation of bone in FOP flare-ups in both human cells and mice.

One of the causes of hypoxia is inflammation, which we know from previous research is associated with FOP lesions. When a cell is hypoxic, the HIF-1α protein triggers a molecular alarm. In this study, the researchers found that by inhibiting this HIF-1α protein they could quieten the alarm, and that this resulted in a reduction of bone formation in FOP mice. Most importantly, in human FOP bone cells this approach resulted in signalling of bone growth slowing to what you would see in non-hypoxic cells.

So, what does this all mean? This study has identified the HIF-1α protein as a potential target for treating FOP: “The implications for targeted clinical trials and for compassionate clinical use of HIF-1α inhibitors in the treatment of FOP flare-ups are promising, however we need more data on dosing, duration, timing, rebound, resistance and long-term safety,” said Robert Pignolo, Associate Professor of Medicine at the Hospital of the University of Pennsylvania.

Findings from the study were published in the *Journal of Bone and Mineral Research* in May.

Partnership with SGC Oxford to Continue

April

FOP Friends, FOP France and FOP Switzerland will continue to help fund the Structural Genomics Consortium (SGC) research team at the University of Oxford. The team currently consists of two postdoctoral fellows who are working on developing a treatment for FOP. In 2015/2016 we managed to fund their research to the tune of £125,000, and we hope to be able to at least match this in the coming year.

A Big Microscopic Upgrade

April

The SGC Oxford team received a new microscope in April, following our successful grant application to The Hospital Saturday Fund. The EVOS XL Core Imaging System is used by the researchers daily, and lets them examine and compare the growth of cells containing the FOP mutation to that of unaffected cells.

10th FOP Italia Meeting

14-16 April

Researchers and advocates for FOP families and groups from around the world (including our own Chris Bedford-Gay) gathered in Liverno, Italy in April for FOP Italia’s 10th annual meeting. A series of excellent presentations highlighted the latest research and advances around FOP.

Congratulations FOP Australia!

June

FOP Australia held an official launch event in Brisbane on 4th June 2016. The event also marked one year since the organisation was established through the efforts of Lara Boniface, whose son Jarvis was diagnosed with FOP in 2014. Lara came to the FOP Friends UK conference in 2014, and left determined to set up a similar organisation in Australia.

You can find out more at www.fopaustralia.org.

Have news, views or stories to share? We would love to hear from you. Email us – info@fopfriends.com

"These types of gatherings truly make a difference."

2016 Conference and Family Gathering

On Saturday 21 May, we held our 2016 conference and family gathering in Manchester. As in 2014, it was a chance to learn, share and connect with others who are in one way or another affected by FOP. Thank you to everyone who contributed by being there, asking questions, answering questions, and generously sharing insights and experiences. We are always overwhelmed by the care and dedication that exists in our community when we get together.

BIG LOTTERY FUND
LOTTERY FUNDED

Once again we were privileged to have some of the world's leading researchers and clinical experts in FOP join us at the conference. Families could book clinical appointments from the Thursday through to the Sunday of our event weekend.

The Saturday opened with a talk from Professor Fred Kaplan and Dr Robert Pignolo from the University of Pennsylvania, who tackled *What is FOP? The science for non-scientists*. Delegates had the chance to ask questions, so that we could all start with a solid understanding of what it is we're dealing with. Professor Kaplan and Dr Pignolo were followed by their colleague, Professor Eileen Shore who introduced the session on *How do we fix FOP?* by discussing the foundations for future treatments. This was expanded on by research scientists from Oxford and Harvard and representatives from the pharmaceutical industry. We then had an update on the FOP Natural History Study and heard about progress being made with the FOP Connection Registry. Dr Gehan Abou-Ameira, a paediatric dental consultant from Great Ormond Street hospital rounded off our presentations talking about FOP and dental treatments, speaking to our adult delegates before taking her tooth brushes down to the crèche to talk to the children about looking after their teeth.

Later in the afternoon, we trialled a new approach with three workshops that delegates could attend. The workshop themes were *Genetic Counselling, Neuro Linguistic Programming* and *Meet the Scientists*. A workshop leader facilitated discussions, giving delegates the opportunity to share their experiences, ask the scientists more detailed questions and push them on the future plans for treatment, which led to some heated and informative debates between the different schools of thought.

More than 100 of you joined us for the dinner on Saturday evening, where the karaoke music quiz let some real talents shine through!

"I am not sure how else to put it, but what you put together was simply terrific... I hope that it could serve as a model for other organisations that deal with rare diseases and have a patient-centric mission and focus. Again, many thanks for inviting me. I felt very honoured and lucky to have been part of this!"

"The one thing I've gotten out of the conference is how much they value us as a family, and how much they value the patients and us contributing. You don't feel like it's 'the doctors' and 'us'."

"It's really motivating. When I go back to the lab...I have faces to put to the condition that I'm working for."

"I absolutely loved seeing my FOP fam! I never have to explain, I never have to be judged by anyone. Everyone understands everyone and I love it!"

"Feel like we are part of a great community."

"[FOP Friends] have pulled off the seemingly impossible task of uniting an army of people with a bunch of world class doctors/researchers and given us hope for a brighter future."

IN NUMBERS

104 conference delegates
71 people whose families have been touched by FOP
31 clinicians and researchers
25 children who built **6** crazy robots in the crèche
11 countries represented
12 speakers from the UK and USA
3 discussion workshops
2 Q&A sessions with the experts
1 mad music quiz
0 new millionaires*

*Dr Richard Keen's million pound question was: What is the chemical formula for Palovarotene? And the answer: $C_{27}H_{30}N_2O_2$

Q&A

with Dr Richard Keen

Dr Richard Keen is a Consultant Rheumatologist at the Royal National Orthopaedic Hospital NHS Trust in London. He currently works with about a dozen FOP patients in the UK.

When did you first become aware of FOP?

I've been in my current job for about 17 years, and one of the very first patients I saw was a teenager with FOP. It was a very steep learning curve for me. At that stage there was still so little information that my focus was on supportive treatment and managing flare-ups.

I became aware of FOP Friends two years ago when I attended their family gathering. One of my patients told me about it. It made me aware of the research and the trials and made me realise there was now a lot more I could offer my patients. I was really motivated by the progress that had already been made.

Are you a part of any clinical networks to support FOP patients?

I'm part of an EU FOP Consortium that includes doctors in the UK, Italy, the Netherlands and France. There's also currently a call out from the EU to establish a reference network for rare bone diseases, and there's been quite a lot of interest in that. Having one would give more FOP patients access to the expertise they need, and it should also drive improvements in all the individual countries involved.

How can the FOP community help support clinicians, and what do you suggest that GPs and other local health professionals do when one of their patients is diagnosed with FOP?

I think the first thing is for the patient to make sure the consultant they're seeing has the most up-to-date knowledge on FOP. Don't be afraid as a patient or family member to question them and make suggestions about who they could contact. The IFOPA and FOP Friends make the latest information available.

I know that patients and their families can often feel isolated and afraid. They can always contact us and we will happily contact their local consultant and GP. We will work with them so that there is a management plan ready for when flare ups do occur. You need to have a plan before they happen, and everyone involved in the patient's care needs to know about them.

I would also really encourage patients to tell us anything that they're experiencing. It may not be relevant, but it could open up new avenues to explore for treatment. There are things that only the patients can tell us and that we have no way of knowing.

Are you directly involved in any FOP research projects?

Yes, we are the only UK centre involved in the Clementia drug trial with palovarotene. We're also involved in the Clementia natural history study. And we may be involved in more trials in the future.

What do you think is the biggest challenge facing FOP research right now?

In the last 10 years, since the gene was discovered, the rate of discoveries of drugs that could work has been rapid. It's incredible how things have advanced in a decade, and it's exciting that there are so many potential drug targets out there now. Regeneron's findings in animal studies look positive, but putting any drug into trial is challenging, and it is even more challenging to take a drug from animal testing to clinical trials when there is such a small patient group. There are many ethical issues: should you trial them on children, where the need is greater but so are the risks, or in adults? Could you do trials in people after they've had surgery? If you could operate to free things up and then give people a drug afterwards that would be great.

There are challenges, but I think all these things are solvable and possible with the people that are involved.

What did you gain from the 2016 FOP Family Gathering?

The gathering is primarily for patients and the families, but it's also great for clinicians. In the clinical sessions patients have the opportunity to be reviewed by Professor Kaplan, who is the leading expert, and other clinicians get to learn from him. It's a great opportunity for the dissemination of knowledge to other clinicians.

You and your daughter ran the Great Manchester 10K the day after the conference. Why did you choose to run for FOP Friends?

My daughter Ellie is a teacher in Manchester and has run this race once before. When I realised that the FOP Friends conference was the day before the event it all just seemed to come together. And it made sense that I run for FOP Friends. And no, I'm not a runner! I set myself this challenge at Christmas and it's been hard! I was happy with my time though, just breaking the one hour mark and finishing in 00:59:56!

Together Dr Keen and Ellie raised £1400 for us.

Our Fantastic Supporters

We are continually amazed by what you do to raise money and awareness for FOP. Thank you one and all. Every pound you raise means so much and goes directly to funding research and supporting families affected by FOP.

A Hilly Hastings Half

HASTINGS, SUSSEX – MARCH

Emma Spooner ran the hills of Hastings for FOP Friends in March, raising over £400, despite a raging cold.

"The reason I put myself through 13.1 miles of pain with a stinking cold and really bad cough is because I see my husband fight this horrendous disease. Paul doesn't feel sorry for himself. He works hard for his family and is a brilliant dad. FOP robs him of so much. He can't do things like play golf or go for walks – simple things I take for granted." – Emma

An Azerbaijani Channel Swim

BAKU, AZERBAIJAN – FEBRUARY

In February, five groups of swimmers from the Baku Sharks aged 11 to 16 set out to swim the equivalent of the English Channel in two weeks. Each group collectively swam 32km – that's over 1,200 lengths of a 25m pool! Friends and family pledged money for their hard work and together they raised £360 for FOP.

"It was a great experience for me to manage and organize a fundraiser, and I think it was successful enough for my first shot at it. I hope that next year we can pull out all the stops and do even better than this year." – Anoushka, 16

Awesome Foursome

We were honoured to be chosen as one of the charities for Simon Warner's impressive fundraising feat of running four races for four charities in a week! The London and Milton Keynes marathons sandwiched two 5km races, bringing the total race miles Simon ran that week to almost 60, for which he earned a total of £3,300 in sponsorship.

Student Finance

MANCHESTER, UK – APRIL

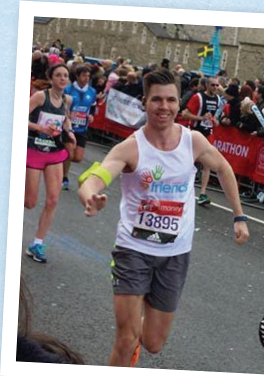
Five students from the University of Manchester chose to undertake a fundraising project as part of a Careers and Project Management Skills module. As well as running the Manchester Marathon Relay, Katie, Olivia, Sophie, Albert and Polly also pooled their skills to host a raffle night. Up for grabs were prizes ranging from a delectable food hamper to tickets to Lords. Together their running and raffling efforts raised over £1,500.

A Swap 4 FOP

Friends of Isla organised a Swap 4 FOP in Milton Keynes that brought in £175 and made sure unused items found some happy new homes.

Running Feats

- Sara and Martyn Cassidy finished the Manchester Marathon in glorious sunshine in April. Together with the University of Manchester Relay team, they raised over £2,500.
- Much further north, Calum Jackson raised £675 by taking on the ANGUS HaM, a half marathon near Dundee with its fair share of hills.
- While in the south, Nick Coogan completed the London Marathon and was still smiling at the end having raised £1,450.



On your Bikes

In May, a team of 20 colleagues from Savills took on the Surrey Hills in the inaugural Savills 100km Charity Cycle. Together their combined pedal power brought in over £4,900 which was shared between FOP Friends and Dreams Come True. Not content with just completing 100km, one team member, Bob Shattock then cycled from Calais to Amsterdam in three days in mid-May, raising a further £2,675 for the two charities.

Have you been fundraising? Firstly, THANK YOU! And secondly, please let us know how it went and share your experiences with us via email or on Twitter @FOPFriends and Facebook FOP friends

How you can help

FOP is a rare disease, which means that most research is funded through the efforts of families affected by FOP and their families, friends and colleagues. There are so many ways you can help us get closer to a treatment and cure, whether it's raising money, raising awareness or volunteering your time, skills or connections.
We need your help.

JOIN our community

The more of us there are, the greater our influence and our potential to succeed in finding a cure for FOP. Like us on Facebook and follow us on Twitter. Share what we're doing with your friends and colleagues, and encourage them to spread the word.

DONATE in cash or kind

All donations are very gratefully received. You can make a one-off payment or set up a monthly donation online at uk.virginmoneygiving.com/charities/fopfriends. Even the smallest donations make a difference. We also have fundraising events throughout the year and are always looking for raffle and tombola prizes. If you have anything that you could donate we would appreciate the support.

ORGANISE an event

Host a dinner, bake a cake, organise a pub quiz night, a Swap 4 FOP or a charity golf day. You can be as creative as you like. We will help you publicise it and get coverage for any sponsors you have. Mari, our fundraising officer, is on hand to support and advise you. Email mari@fopfriends.com.

CHALLENGE yourself

Run, swim, climb, cycle, wade, walk, dance, knit, sing or Zumba! Do whatever you love (or hate) and get sponsorship from your friends and family. And please let us know what you've been up to.

SHOP through easyfundraising

If you (or your business) shop online, this is an easy and secure way to support us. Simply register with www.easyfundraising.org.uk, select FOP Friends as your charity and start shopping! So long as you go through easyfundraising to visit your online store, we get a percentage of everything you spend, without costing you a penny extra. There are over 3,000 stores signed up to the scheme, and to date we have raised £678.41.

SELL using eBay for Charity

Did you know that when you sell on eBay, you can choose to donate anything from 10% to 100% of the final sale price to your chosen charity? You can also Gift Aid your donation. Just use the 'Advanced Sell' form when you're listing a new item and you'll see the option to donate. Visit charity.ebay.co.uk to find out more.

LEAVE a gift in your will

When people think of leaving a legacy in their will, they often think of wealthy philanthropists. But even a small amount will benefit a small charity like ours enormously. When you're making or revising your will, please consider leaving a gift to help us find a cure for FOP.

SHARE time, skills or contacts

We're always on the lookout for people with new skills, contacts and enthusiasm. If you feel you have something to give to FOP Friends, please get in touch. We'd love to hear from you!

MAKE something special

Are you crafty? Love to bake, make or build? Why not raffle, auction or sell the result of your efforts in aid of FOP Friends. Or if you'd rather not do it yourself, donate it to us and we'll do the raffling or auctioning for you.

CHOOSE FOP Friends

Nominate us to be your company's charity of the year, or suggest that we are one of the beneficiaries of your community's charity fundraisers.

About FOP

Fibrodysplasia ossificans progressiva, or FOP, is one of the rarest and most disabling genetic conditions known to medicine. In people with FOP, bridges of bone develop across the joints, progressively forming a second skeleton that imprisons the body in bone. Currently, there is no known treatment or cure for FOP.

A cure for the disease could also benefit people living with osteoporosis, arthritis and heart disease, as well as those affected by heterotopic ossification (a major complication in hip replacement surgeries and in sports and military injuries), and other more common bone and muscle disorders.

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The Sobell Foundation

Give with confidence
FOP Friends is registered with the Fundraising Standards Board, an independent self-regulatory body for UK fundraising that encourages commitment and compliance with best practice in fundraising.

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