



NEWSLETTER

Genetic Overgrowth PI3K Support

www.gopi3ks.com

December 2020

Welcome to the newsletter for GoPI3Ks – Genetic Overgrowth PI3K Support. Here we will keep you informed of the current research, fundraising and stories from overgrowth patients.




*Thank you to Dr Ralitsa Madsen for her research update & her continued support for the PROS community.
(UCL Cancer Institute, London, UK)*

Dear GoPI3K community,
When Mandy Sellars approached me about a contribution to this year's GoPI3K newsletter, I could not help asking myself the same old question:
“Is this year ALREADY over?”

As usual, this question triggered substantial reflections. It is hard to believe that it has been a bit over a year already since I moved to Bart Vanhaesebroeck's group at University College London's Cancer Institute, with plans to study the complexity of “[the PI3K radio](#)” (covered in last year's Newsletter). This research will hopefully form the basis for improved disease understanding and potentially also for better treatment solutions, not only in the context of *PIK3CA*-related overgrowth syndrome (PROS; includes CLOVES) but also for other diseases such as cancer. So to recap, among the key questions that I am interested in studying, and the ones that I think will resonate with many of you, are:

- Do different cells or tissues in the body respond differently to the same *PIK3CA* mutation? (for example, some of you may share the same *PIK3CA* mutation, e.g. H1047R, yet suffer from strikingly different symptoms due to differences in the affected cells/tissues)
- What cell-based mechanisms explain differences in disease severity with different *PIK3CA* mutations? (for example, two individuals may both suffer from vascular malformations but due to differences in the type of *PIK3CA* mutation, one case may be milder than the other – why is that?)
- Not all cells in an affected tissue carry a *PIK3CA* mutation, so what is the contribution of the ‘normal bystanders’ to the overall disease?
- Finally, can the above knowledge be used to identify pharmacological therapies that are more suitable for different types of PROS?



I always try to be mindful of the fact that not everyone will be familiar with some of the above academic jargon, so let me include a short explainer when it comes to some key terms. **PIK3CA** is the name of a stretch of DNA which provides our cells with the code for producing an enzyme that is critical for growth and survival. PROS is characterised by the presence of changes, or mutations, in this stretch of DNA, thereby resulting in a more active enzyme. The higher activity of the mutant enzyme makes it hard for an affected cell to turn 'growth' programmes off. Due to differences in the timing during development when a mutation is acquired, the exact cells that are affected and the type of **PIK3CA** mutation, people with PROS can suffer from vastly different symptoms.

Some good news from 2020

The end of a year is always a good time to take stock – of what has happened and what is to happen. Although 2020 was not exactly a year full of good news, I am pleased to let you know that I was successful in my application for a research fellowship from the Wellcome Trust. This means that my salary and research funding for PROS/cancer-related work is secured for another 4 years. Among others, I am incredibly grateful for the support of my previous (Prof Robert Semple) and current (Prof Bart Vanhaesebroeck) mentors, without whom this achievement would not have been possible.

I also received a substantial research contribution from the CLOVES Community – thank you for that! I have invested the money in making a novel cell model that is of relevance to understanding PROS, and one that will be critical for tackling the key questions listed above. More specifically, I have tasked some of the leading gene engineering companies, Synthego and GeneWiz, with the generation of this cell model (a research tool) and additional components for its downstream use. These are by no means trivial tasks, so my wish this Christmas is to hear back from both Synthego and GeneWiz with successful news regarding this project!

There is also good news when it comes to immediate treatment prospects for PROS – as some of you may know, a worldwide clinical trial of Alpelisib in PROS is in the pipeline. This will allow an accurate picture to be obtained of which parts of PROS Alpelisib is most beneficial for, and of which ages of patients are best to treat, and, critically, how significant side effects are. Novartis, the company who makes Alpelisib, is in advanced stages of finalising such a trial, spread across several countries, and projected to last up to 5 years. The US arm of the study is already registered (<https://clinicaltrials.gov/ct2/show/NCT04589650>), and other countries are expected to follow soon.

Upwards and onwards into 2021

The year 2020 has certainly been a tough one for many, and I am sincerely hoping that you are all well and safe. Despite its bleak nature, the above goes to show that there are always glimmers of hope for all of us. For me, research comes with many ups and many more downs. In fact, it is almost universally true that as a researcher, you spend most of your time figuring out why an experiment is not working or how it can be improved. The more technically complex the experiment (most of them are nowadays!), the higher the risk of failure. Yet, on days when I feel particularly defeated by yet another hurdle, I keep reminding myself that I am not just doing this for me – I am doing it because it may one day have a positive impact on your lives, and that keeps me going. So thank you all for your continued support and faith in my work. It is hugely appreciated.

Merry Christmas and a Happy New Year to all of you!
(Dr Ralitsa Madsen (UCL Cancer Institute, London, UK))

OUR 2020 FUNDRAISING FOR GOPI3KS & AWARENESS OF PROS.

This year has been a difficult one for us all due to Covid-19.

Sadly many charities have struggled to raise much needed funds.

However, I feel very humbled as I write this to say a big **THANK YOU** to everyone who has helped, whether this be by running fundraisers, donating & raising awareness of GoPI3Ks & PROS.

Your continued help & support really makes a difference.

Thank You

FEBURARY 2020

I ran an online birthday fundraiser to raise money for GoPI3Ks.

Every person that donated was placed in to a raffle to win one of the following items:

This year we raised **OVER £1,000**



AUGUST 2020

One of our supporters
Jet was very kind to hold a birthday fundraiser
Online raising the fab total of: £111



SEPTEMBER 2020

My lovely friend Kerry decided for her birthday to raise money for GoPI3Ks. She raised a fabulous: £90



PAYPAL JUST GIVING FUND

Thank You

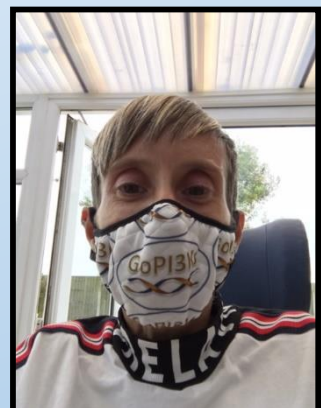
Donations from some generous people who recommended GoPI3Ks through Charitable programs on PayPal, eBay, GoFundMe, Facebook, Humble Bundle etc raised a total of: **£95**

RAISING AWARENESS



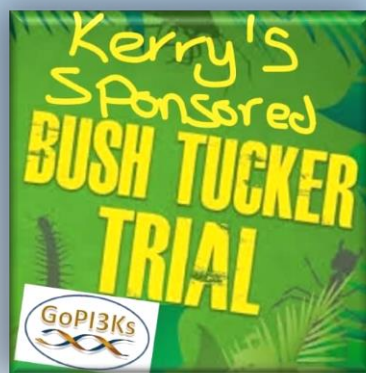
2020 has meant new ways of highlighting GoPI3Ks.

The latest GoPI3Ks masks are available to buy, please email: gopi3ks@yahoo.com



DECEMBER 2020: KERRY'S BUSH TUCKER EATING TRIAL RAISING £760

on FRIDAY 4th DECEMBER just before the I'm a Celebrity final, my friend Kerry decided she wanted to raise money for GoPi3Ks. So Kerry's sponsored Bush Tucker Trial was born.



With her target's set, how much would people pay to see her eat an array of dried bugs & yummy dishes:



£50 raised saw her chomping on *MEALWORMS*!

£100 raised & next came the *CHILLI*!

£150 raised & it was on to the *CRICKETS*!

£200 raised & it was time for a yummy *LOCUST*!



£300 some BLENDED VOMIT FRUIT EXTRACT with water was drunk!

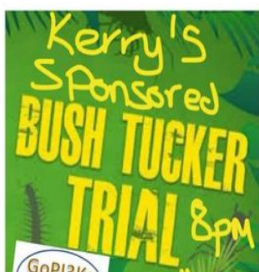
£400 she ate a CENTURY EGG (fermented egg)

£500 Lastly was FISH EYE

Extra money raised during the trial meant eating BRAINS.



 **Kerry Leigh Kavannah**
5 December at 19:41 · 📺
£744!! I'm so happy with this amount. Especially as the initial target was much lower! Thank you so much for all your donations. You're all amazing! I managed to keep it all down somehow and felt pretty normal today (as normal as can be I suppose 🥰). This will be a big help to [Mandy](#) & GoPi3Ks. Thank you again ❤️❤️



Kerry's fundraiser for GoPi3Ks charity
Fundraiser for GoPi3Ks charity by Kerry Leigh Kavannah

£760 of £500 raised

Ended



THANK YOU SO MUCH TO KERRY FOR RAISING AN AMAZING £760. For anyone who may want to watch the trial, (if you dare!) click here: [KERRY'S BUSH TUCKER TRIAL](#)

FUNDRAISING, COULD YOU HELP?

Covid-19 has meant that this year it has been even more difficult to fundraise.

However, there are several ways you can help by shopping from your home.

Here are 3 ways you can shop & raise money for GoPI3Ks.

Amazon Smile

amazonsmile
You shop. Amazon gives.

Download or update to the latest version of the **Amazon Shopping app** on your phone. You **can** find it in the **App Store** for iOS or Google Play for **Android**. Tap on '**AmazonSmile**' and follow the on-screen instructions to turn on **AmazonSmile** on your phone.

Here's how to sign up for AmazonSmile

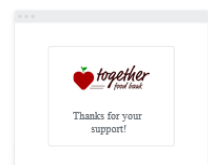
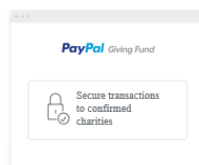
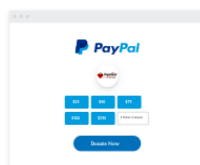
<https://smile.amazon.co.uk/ch/1176289-0>

1. Visit **smile.amazon.com**.
2. Sign in with your **Amazon.com** credentials.
3. Choose a charitable organization to receive donations, or search for the charity of your choice.
4. Select your charity.

PayPal Giving Fund

How can I support GoPI3Ks using PayPal
Please click this link: <https://www.paypal.com/gb/fundraiser/hub>

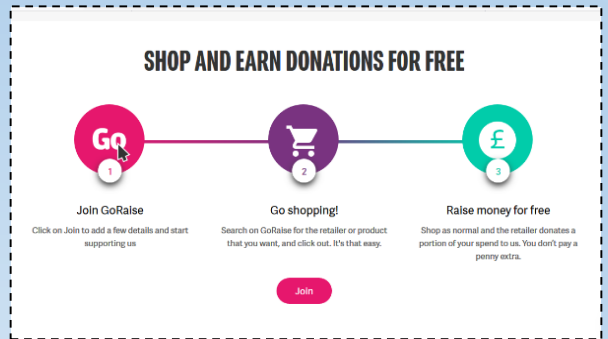
How donating to PayPal Giving Fund works



- 1 Find a charity you'd like to help. Your gift goes to PayPal Giving Fund, a registered charity (No. 1110538). PayPal will take care of all transaction fees.
- 2 We'll give 100% of the donation to your chosen charity, or in rare cases, a similar charity if necessary. Please read our [Donation Delivery Policy](#) for details.
- 3 PayPal Giving Fund will issue your donation receipt, with the option to [add Gift Aid](#). You can ask us to share your name and email with the charity you help so they can contact you or you can donate anonymously, if you prefer.

GO RAISE

You can help raise funds for GoPI3Ks by simply shopping online with Go Raise. Click on the link: www.goraise.co.uk/gopi3ks-genetic-overgrowth-pi3k-support & follow the instructions provided.



HOW THE GoPI3Ks CHARITY HAS HELPED IN 2020

THANK YOU to everyone that has donated, this is how **YOUR** money is helping & making a difference to people's lives.

MARCH 2020

In March we were able to help a family in America with the cost of their daughters' dental care, something their insurance wasn't able to cover.



My family and I would like to give GOPI3KS a heartfelt "Thank you!" for donating money towards our daughter's orthodontic bill. With your help, we are thrilled to announce, that her bill will be paid in full by this time next month! This is a huge weight off our shoulders! It really means a lot to us. Thank you so much!

MAY 2020

In May we had the pleasure of being able to help one of our families who had recently bought a dog.



Pugsley is to become an assistance dog to help their daughter who lives with PROS.



Initially we were able to help with the vaccinations for Pugsley before he started out on his training to become a therapy dog.

JUNE 2020

In June we were also able to help Pugsley with the start of his training to become a therapy / assistance dog to help one of our young GoPI3Ks members.



Here is Pugsley as he starts his training to become helpful to his little human owner.



JULY 2020



Due to Covid-19 holding a family weekend this year would have been impossible.

Instead we hosted a Zoom video call for GoPI3Ks members along with Professor Semple & Dr Madsen.

The GoPI3Ks members submitted questions prior to the call but were also able to ask questions on the day. This proved very successful & helpful to all involved.

AUGUST 2020



In August we were able to help one of our members who needed to purchase two pairs of trousers from America that are bigger in size.

Subsequently we were then able to help with the payment to alter the 2 pairs to be made into one pair of trousers to accommodate the overgrowth in his low limbs.



In August one of our younger members underwent an operation, but due to Covid-19 the family were only allowed to travel by car to the hospital.



This meant facing the cost of parking as well as congestion charges. GoPI3Ks was able to help with these.



SEPTEMBER 2020

zoom

In September we held two Zoom sessions with Tracey Whitewood-Neal who's son lives with the rare overgrowth condition (Proteus Syndrome) She kindly offered to chat with our GoPI3Ks families about being a parent of a child with an overgrowth & to answer any questions our families may have had.

OCTOBER 2020



In October we were contacted by Meggie's mum as Meg had recently undergone an operation on her leg. This meant a long road of recovery ahead of her with no guarantee she would still be able to dance.

GoPI3Ks were able to help by paying for specialised physio & so far Meg is definitely on the road to making a full recovery.

We asked Meggie why she loves dancing so much:

"I love dancing because you can express yourself in a way that everyone will understand without using any words. I can lose myself and just enjoy the feeling of freedom. All your problems go away, it's just you and dancing".



COLLABORATION WITH OTHER PIK3CA GROUPS & RESEARCH COMPANIES

2020 has led to GoPI3Ks working on various projects with a number of other PROS groups. These groups include:

	https://www.m-cm.net/
	https://www.associazione-nazionale-macrodattilia.org/
	https://wonderfilsmiles.com/
	https://www.clovessyndrome.org/
	https://k-t.org/
	https://www.projectfava.org/
	https://www.lgdalliance.org/

Patient Registry

Along with the CLOVES Syndrome Community & WonderFil Smiles we have launched a rare disease patient registry for individuals with CLOVES and PIK3CA-Related Overgrowth Spectrum (PROS) syndromes.

With the help of the Backpack health app, patients can input & keep track of their medical records in an international data secure site.

WHY TAKE PART?

Research in to rare conditions is extremely challenging & developing a patient registry helps to provide information on treatments, patients experience all in one place. This information can then be shared with researchers etc. Information is only shared by authorisation from each patient.

For more information please see:
<https://my.backpackhealth.com/join/clovespik3caregistry>

2021 Scientific Meeting

International Scientific Meeting for PIK3CA Related Conditions
Thursday October 28 – Friday October 29, 2021

Save this date! We are proud to be working alongside the CLOVES Syndrome Community who are going to be hosting the first International Scientific Meeting of PROS & PIK Related conditions. This is part of their patient led research network development with the [Rare As One Network](#). This meeting will include both medical & scientific professionals as well as advocacy groups, this will be the first of its kind.

For more information please see the CSC website:
<https://www.clovessyndrome.org/about-csc/2021-scientific-meeting>

Novartis: This company is making big strides in potential treatment for PROS: <https://www.novartis.com/search-results?query=pik3ca> More info can be found on our website: <https://gopi3ks.com/research-in-to-pros/>

A few months ago I was contacted by the director of Global Patient Relations at Novartis to see if the GoPI3Ks families could help with a Quality Of Life questionnaire for their up & coming drugs trial for PROS. Thank you to those who responded, your help will pave the way for the up & coming research.

MEET YOUR GOPI3KS CHARITY TRUSTEES.



MANDY SELLARS - CHAIRPERSON

Hi, I'm Mandy & I was the first person to be diagnosed with my particular PIK gene mutation. I set up the GoPI3Ks charity in 2017 to try to help those like myself living with a PIK3CA condition. I'm a big tennis fan, love horror movies & reading about true life crimes. I also love live music, my fave's being Texas & Pink.

SUE HARPER - TREASURER

Hi, I am Sue & I am the Treasurer of the charity. I love animals, especially cats & share my life with 2 rescue cats. I spend my free time volunteering at a cat sanctuary, walking, gardening, listening to music & watching movies/TV series amongst other things. I am grateful to Mandy for asking me to play a part in the running of the charity as it is very rewarding to see the positive effect the charity has on it's members.

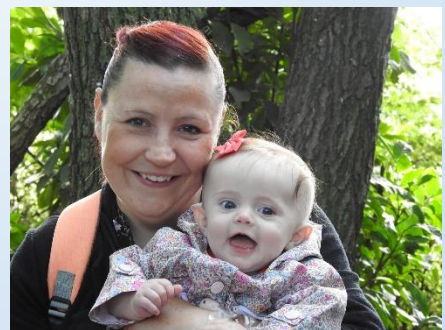


AMANDA KENYON - SECRETARY

Hi, my name is Amanda & I have been friends with Mandy since meeting at university back in the 90's. I enjoy reading & watching true crime documentaries. I love live music & have a great collection of vinyl. I'm the very proud owner of the most adorable pug by the name of Dottie.

KATIE KAVANNAH - TRUSTEE

Hi I'm Katie. I have been friends with Mandy for many years. She is one of my dearest friends. I'm here to help support her with this good cause and raise awareness. I've been very lucky this year as I welcomed a beautiful baby girl in April 2020. Now 7 months old she's keeping me on my toes.



FROM EVERYONE AT THE GOPI3KS TEAM WE WISH YOU A WONDERFUL FESTIVE SEASON, STAY SAFE & SEE YOU IN 2021



**Merry
Christmas**

