NEWSLETTER

Genetic Overgrowth PI3K Support

www.gopi3ks.com

December 2022

Welcome to the newsletter for GoPI3Ks – Genetic Overgrowth PI3K Support. Here we will share updates from PROS medical experts, information on fundraising, how we have helped those living with PROS, as well as rare events & information from the rare community.

A huge thank you to Dr Ralitsa Madsen for her contribution to this years newsletter. Thank you also for your continued dedication, research & support of the PROS community.

Thank you also to Professor Rob Semple for your tireless work in the scientific research of PROS.

You both give hope to all of us living with & affected by this condition.

Research and Treatment Update – Dr Ralitsa Madsen

Dear GoPI3Ks/CLOVES community,

Here we go again – another year has taken a turn, and Christmas is waiting around the corner. I would lie if I said it is not somewhat stressful to think about how time just flies. On the other hand, it is also rewarding to look back over the last year, which brought several bits of good news and positive experiences with it.

As of scientific news, one of the first items to mention is the development of novel types of pharmacological agents that appear to hit the faulty product of the *PIK3CA* gene (that causes PROS) without affecting the remaining normal component (a brief recap on PROS biology: the *PIK3CA* gene defect causing PROS is present on only one of the two DNA copies that you inherited from your parents; the other copy remains normal and gives rise to a product with important functions in a cell, so really we only want to hit

the faulty one!). Until now, available pharmacological agents have been hitting both the good and the bad *PIK3CA* gene products, in other words they were not selective. As with all new research – and by now, many of you may also know my aversion to hyping results up – the road is long until any of the new agents get to a stage where they may be considered for clinical testing. It is however exciting that such developments are taking place as key issues with a drug like Piqray/Vijoice (or Alpelisib) relate to the side effects arising from lack of discrimination between the altered and the normal product of the *PIK3CA* gene. And while drug development efforts in this space usually start off with a cancer-heavy focus, we know already that the shared biology affords subsequent opportunities for so-called drug repurposing. You can read more about such repurposing, including important ways in which cancer and PROS differ from one another, in An open access piece which I recently co-authored with Professor Robert Semple.

Earlier this year, I spent three months in my collaborator's, Prof Alex Toker's, laboratory at Harvard Medical School in Boston. Here, we focused on completing a particularly – for us – rewarding project focused on a chemical agent that targets a cellular component known as AKT. Some of you will know that rare genetic variants in AKT can give rise to a PROS-like disease known as Proteus Syndrome. Alex Toker is an expert on AKT and while he, too, mainly works on cancer, he is now also involved in a recently established initiative by CLOVES Syndrome Community that allows scientists focusing on PIK3CA, AKT and related components to meet virtually once every two months to discuss and advise each other on ongoing research. Such discussions, well in advance of formal scientific publications, are really important as they help speed up the research progress that is urgently needed by all patients. I am therefore pleased that prominent cancer researchers with the relevant scientific expertise are beginning to familiarise themselves with PROS and are actively considering ways in which they may be able to contribute to ongoing research in this space.

Prof Toker is also a highly trusted mentor of mine, who has been immensely supportive of my scientific career. It is partly thanks to his support with the above research and efforts to introduce me to relevant senior scientists at a recent meeting in Canada that I have a final piece of good news to share. This news is somewhat more personal, though I hope you will rejoice at the opportunity for continued basic research of relevance to PROS in the UK and internationally. The news is that I have been given the opportunity to start my own, fully independent research group at the MRC-PPU in Dundee, Scotland. It is difficult to convey how important this is, not only for my ability to continue academic research in general but also for my passion to pursue novel solutions in the realms of human diseases caused by genetic *PIK3CA* defects. Remember that PI3K radio I wrote about in a newsletter three years ago now? I am actively working on understanding exactly how "it" is engineered, and this has resulted in substantial progress which I hope to share as part of a scientific article in Spring 2023. In a nutshell, what I can reveal already is the notion of a much more nuanced understanding of how components like PIK3CA and AKT enable critical communication signals to be received and interpreted by our cells. This, in turn, has allowed me to discern ways in which this communication gets "corrupted" or "blurred" in the presence of a genetic PIK3CA defect. The key question of course is – how do we correct it, and will the correction factor differ depending on the affected tissue or the specific *PIK3CA* defect? To use a simple analogy, it appears that cells with a *PIK3CA* defect need correction glasses to "see" particular signals. While this riddle remains to be solved, I have much hope in the power of modern computers and algorithms to help me with the solution. It will take time and it will take a lot of hard work, but the will has always been there – and now also the opportunity of full research independence.

So, in summary, I look forward to contributing many more years of Christmas newsletter writings to the PROS community. Who knows, 20 years from now, the above riddle may be solved and translated into therapeutic improvements, allowing us to look back at this and older pieces of research news with much joy and even more hope for the future. Thus, let me close off here with my own version of President Obama's inspirational slogan: "Yes, we can, and we certainly will!".

Happy Holidays to all of you!

Dr Ralitsa Madsen (UCL Cancer Institute, London, UK)

OUR 2022 FUNDRAISING FOR GOPI3KS

As another year goes by, we must say a big THANK YOU to those who have donated. Your generosity allows us to continue to be able to help the GoPI3Ks families

FEBURARY 2022

Mandy again ran a birthday online fundraiser.

Everyone who donated was entered in to a draw to win 1 of 2 £25 Amazon vouchers that we could then email directly to the winner.

This year we were again amazed by the kindness of people who helped us raise over £1,000

AMAZON SMILE DONATIONS

amazonsmile
You shop. Amazon gives.

For those who shopped at Amazon you have donated £59.35. THANK YOU. If you'd like donate when you shop please see how to later in this newsletter.

CHARITABLE GIVING / PAYROLL GIVING

Payroll Giving is a simple and tax efficient way of donating to #charity! Donations are taken from your pre-tax pay so a £5 donation only costs a standard tax-payer £4. For more information please see: CHARITABLE GIVING

To a donor who kindly donates £25 each month. Your support is truly appreciated.

2022 FUNDRAISING FOR GOPI3KS CONT:

In May we attended the annual PENDLE POWERFEST car show where we ran a tombola & sweet stall. Along with Mandy were 2 of the other trustees, Sue & Amanda. We raised over £150.

2022 FUNDRAISING FOR GOPI3KS CONT:

Mandy was able to share information online as to what it is like to live with PROS, which led to a kind donation to GoPI3Ks of over £500

In November we received a donation of £250 from @MADLcharity: Making A Difference Locally after our chairperson Mandy Tweeted about the work we do supporting those living with PROS

SO HOW DOES YOUR MONEY HELP THOSE LIVING WITH PROS?

SEE OUR PAGE BELOW

GRANTS GIVEN TO PROS FAMILIES IN 2022

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

Remember, we are always here to help, here is our grant application form for anyone who would like to apply: GoPI3Ks Application form

As the cost of living rises we were happy that we were able to help one of our families by giving them a grant for £1,000 to help with their energy bills.

One of our families had their holiday cancelled due to Covid. They received a voucher to book another trip, but this wasn't enough to cover a "once in a lifetime" holiday. So GoPI3Ks were happy to pay the £597 that they needed to secure their holiday.

We were also able to pay for £392 for accommodation for one of our younger PROS members, so she could take part in a short documentary focusing on how disabled people are represented & treated in todays' society. This also gave her a chance to spend time with other young people with rare conditions, helping her to feel more included & accepted.

I feel so lucky to have had this experience, it was a total dream come true! It was so lovely to meet other people with different conditions and to celebrate it and create awareness. I made some friends for life and found a huge love for acting and film. Thank you for making this possible for me!

GRANTS GIVEN TO PROS FAMILIES CONT:

We were able to also help one of our younger PROS members.
With overgrowth of the lower limbs & feet, it can mean that families need to buy 2 pairs of shoes/trainers to fit comfortably. We were more than happy to help.

As winter sets in & energy prices rise, we don't want anyone to struggle. So we were able to help one of our families to keep their home warm during this winter with a grant of £800

GoPI3Ks

GoPI3Ks "Let's Chat"

Come along to our small informal group chat on **Thursday 5th May 2022 at 19.00 (GMT) via Zoom.**

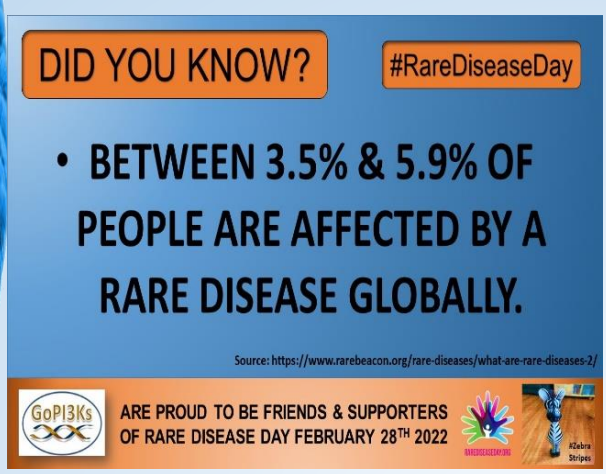
Be in the company of **likeminded people**, people living with PROS, caregivers & parents.

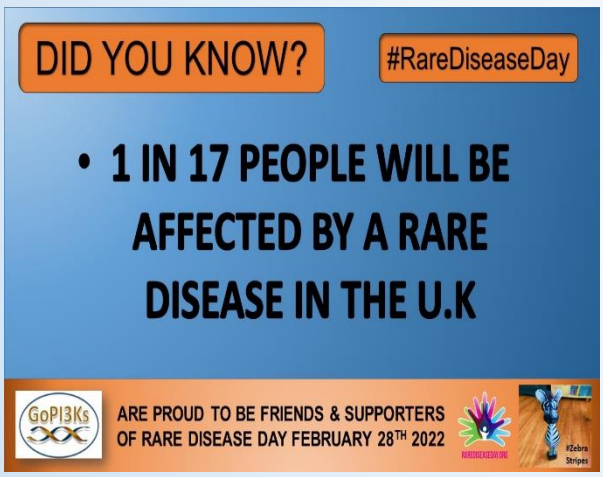
A place to **share experiences** or just a general chit chat.

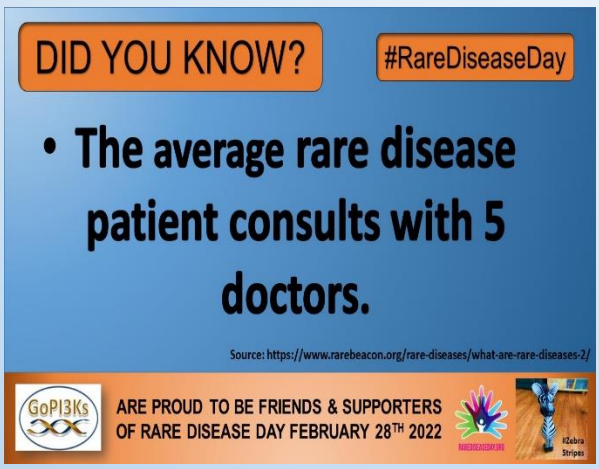
In May we held our first "Lets Chat." A place for those living with PROS, parents & carers to come together to chat. If this is something you would be interested in doing more of please let us know as this is your group.

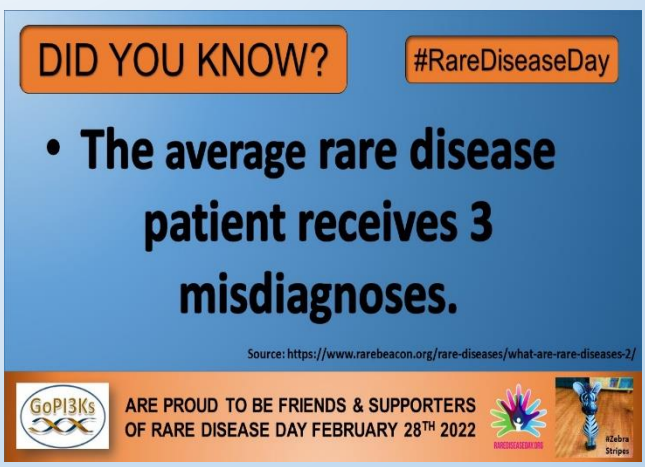
February 2022

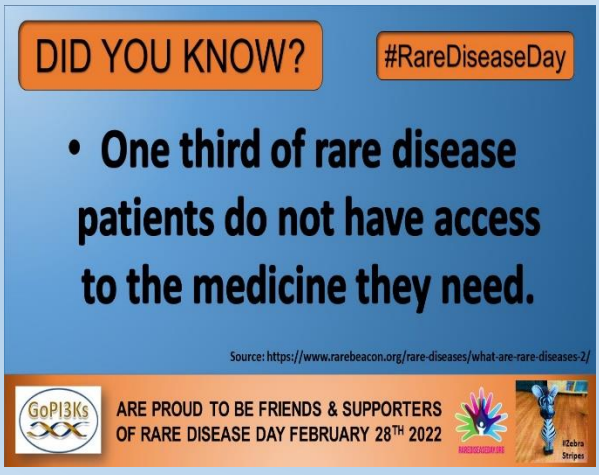
In February 2022 we again took part in Rare Disease Day by sharing information cards on rare disease statistics & PROS

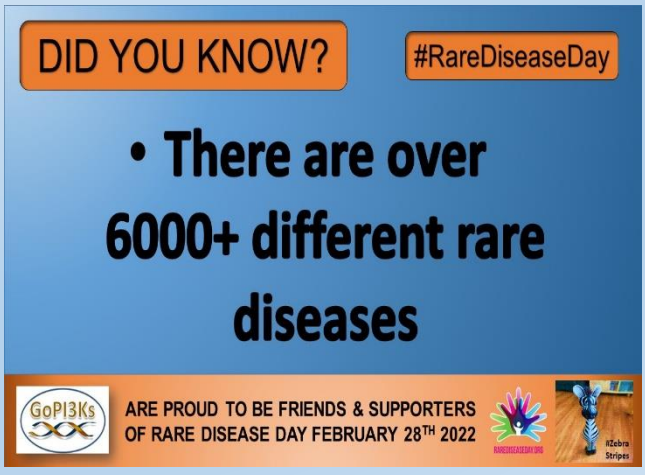












DEFINITION OF PROS

#RareDiseaseDay

- PROS IS A BROAD-RANGING SPECTRUM OF OVERGROWTH DISORDERS CAUSED BY PIK3CA MUTATIONS
- PROS WAS ESTABLISHED BY THE NIH IN 2015 TO UNITE A GROUP OF OVERGROWTH DISORDERS BASED ON THEIR SHARED GENOMIC CAUSE.

GoPI3Ks ARE PROUD TO BE FRIENDS & SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2022

WHAT IS PROS?

#RareDiseaseDay

- **(PIK3CA RELATED OVERGROWTH SPECTRUM)**
- CLINICAL HALLMARKS OF PROS:
  - CONGENITAL OR EARLY CHILDHOOD ONSET
  - SPORADIC OR MOSAIC OVERGROWTH PATTERN
  - PROGRESSIVE NATURE

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WHAT IS PROS?

#RareDiseaseDay

- **PROS FEATURES BROADLY INCLUDE:**
  - OVERGROWTH OF ADIPOSE, MUSCLE, NERVE OR SKELETAL TISSUE
  - VASCULAR MALFORMATIONS, INCLUDING CAPILLARY, VENUS, ARTERIOVENUS, OR LYMPHATIC EFFECTS
  - SKIN LESIONS

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WHAT OVERGROWTH DISORDERS ARE FOUND WITHIN PROS?

#RareDiseaseDay

- CLOVES SYNDROME (Congenital Lipomatous Overgrowth, Vascular malformations, Epidermal nevi, Scoliosis)
- KTS (Kippel-Trenaunay Syndrome)
- FIL (Facial Infiltrating Lipomatosis)

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WHAT OVERGROWTH DISORDERS ARE FOUND WITHIN PROS?

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- MCAP or MCM (Megalencephaly-Capillary Malformation)
- ILM (Isolated Lymphatic Malformations)
- HHML (HemiHyperplasia-Multiple Lipomatosis)
- FAVA (FibroAdipose Vascular Anomaly)

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WHAT OVERGROWTH DISORDERS ARE FOUND IN PROS?

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- CLAPO SYNDROME (Capillary malformation of the lower lip, Lymphatic malformation of the face & neck, Asymmetry of the face & limbs & partial or generalized Overgrowth)
- FAO (FibroAdipose hyperplasia or Overgrowth)
- MUSCULAR HH (HemiHyperplasia)
- Macrodactyly

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SELECT COMPLICATIONS OF PROS

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- **ABNORMAL GROWTH CAN CAUSE:**
  - SPINAL COMPRESSION
  - REDUCED MOBILITY
  - SCOLIOSIS
  - PAIN

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SELECT COMPLICATIONS OF PROS

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- VASCULAR MALFORMATIONS CAN CAUSE:
  - BLOOD CLOTS (EG, DVT OR PE)
  - INTERNAL BLEEDING
  - VARICOSE VEINS

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SELECT COMPLICATIONS OF PROS

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- LYMPHATIC MALFORMATIONS CAN CAUSE:
  - CELLULITIS

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SELECT COMPLICATIONS OF PROS

#RareDiseaseDay

- OTHER COMPLICATIONS INCLUDE:
  - URINARY INCONTINENCE
  - INTELLECTUAL DISABILITY
  - ABNORMAL KIDNEY'S
  - SEIZURES

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MANAGEMENT OF PROS

#RareDiseaseDay

- This varies dependent on the individual
  - SURGERY: Removing areas of overgrowth. Having this type of de-bulking surgery can cause regrowth in the same area, meaning the procedure needs repeating.

GoPI3Ks ARE PROUD TO BE FRIENDS & SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2022

RAREDISEASEDAY.ORG #Zebra Stripes

MANAGEMENT OF PROS

#RareDiseaseDay

- This varies dependent on the individual
  - INTERVENTIONAL RADIOLOGY: Used for those with vascular malformations by applying various techniques that cut off blood flow from the vessels & surrounding tissue so affected cells die.

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MANAGEMENT OF PROS

#RareDiseaseDay

- This varies dependent on the individual
  - MONITORING: Monitoring of your symptoms maybe the most appropriate way forward for yourself & the medical team.

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MANAGEMENT OF PROS

#RareDiseaseDay

- This varies dependent on the individual
  - COMPRESSION & ELEVATION: Raising limbs to reduce inflammation & swelling caused by lymphedema. As well as applying pressure to control vascular bleeding & venous insufficiency.

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IS THERE A TREATMENT FOR PROS?

#RareDiseaseDay

- RECENT RESEARCH IN TO CANCER HAS LED TO A PROMISING BREAKTHROUGH WITH A DRUG CALLED ALPELISIB

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THE PROS COMMUNITY WORKING TOGETHER

#RareDiseaseDay

- Be part of the AllStripes program for PIK3CA-related overgrowth spectrum
  - Contribute to multiple research efforts for CLOVES, FIL and other PROS conditions without leaving home.
    - https://www.allstripes.com/program/pros

GoPI3Ks ARE PROUD TO BE FRIENDS & SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2022

RAREDISEASEDAY.ORG #Zebra Stripes

MENTAL HEALTH & WELL-BEING

We understand that living with a rare condition &/or being part of a wider family network, where someone is rare isn't always easy.

This year we have updated our resource page to include many mental health websites, pages & information. Here are a few examples, but please visit our resources page for more:GoPI3Ks resource page

ADVICE FROM RARE DADS

TOP TIPS FOR RARE PARENTS

GUIDANCE FROM DAVID ROSS AND ADAM JOHNSON:
HOW TO NAVIGATE PARENTHOOD AND LIFE WITH A RARE DISEASE

1 Time as a parent

Having a rare disease can take over your life in many ways, things can be challenging but you are still you. It is important to make quality time to just be a parent with your child and remember they don't see your disease they see their Dad or Mum.

2 Feel the feelings

There are so many corresponding emotions and feelings that go along with chronic illness and rare diseases. Give yourself permission to feel all of them when they arise. Sometimes writing down what you are going through can help to express your feelings.

3 Ask for help

Asking family and friends for help can feel like you are failing or you may feel you don't want to burden them, but it is really important to reach out if you are struggling physically or mentally. If you want to find support outside of your family unit there are charities and communities to help you. Check out David's Ross's blog post here for signposting: bit.ly/MensMentalHealthMeetings

4 Accept the help

Accepting help when it is offered is not always easy. But if a family member or one of your children is offering you help with a physical task or with emotional support accept it. They are offering it because they want to!

5 Communicate

Don't be afraid to let your children know about your condition. Children are often more aware than we realise and talking things through with them in child-friendly way can really help them process what is going on. Start a dialogue and be honest and as they get older they will appreciate being involved in your journey bringing you closer together.

6 Allow time to adjust

Just as you have needed time to adjust to living with your rare condition let your children process the information you give them. Don't be worried if they don't express their thoughts straight away, it can be a lot to take in and time is needed to process this.

7 Go with the flow

If you can't do the activities you used to do with your children, go with the flow and find ways to tweak things so you can still have fun and spend time with them even if this time looks different to how it once did.

8 Pace yourself

Somedays may be tougher than others. Don't try to do too much, your family will understand that you need some time to recoup and rest before jumping back into the full swing of family life. They won't judge you so don't judge yourself.

9 Make connections

Reach out to other parents who are going through the same thing, they might have advice and strategies for coping that you haven't heard of before! Take the time to build relationships with people who understand your process.

10 Time for play

Living with a rare condition means that serious issues and appointments take priority in your life. Try and find the time to enjoy yourself and your journey as a parent. Have fun with your children and cherish the good times.

RARE REVOLUTION MAGAZINE

Produced by **David Ross and Adam Johnson** - **A Rare Disease Collaboration** on behalf of Rare Revolution Magazine
www.rarerevolutionmagazine.com @RareRevolutionMagazine

MENTAL HEALTH CONT:

A RARE MUM'S ADVICE

TOP TIPS FOR RARE PARENTS

GUIDANCE FROM REBECCA PENDER ON HOW TO NAVIGATE PARENTING A CHILD WITH A RARE DISEASE

1 It's ok to feel relieved

If you have fought a hard battle to get to this point you may be overwhelmed with relief. You trusted your own voice and kept going until you got answers. It is ok to have more questions after receiving a diagnosis. You now have a direction you can research in and connect to find answers.

2 It's ok to grieve

You are not grieving for your child but for the picture you initially had in your head of what your life would look like. It's ok to have those feelings just don't let them weigh you down and carry them forward. They can prevent you from seeing all the joy that is to be had.

3 Become the expert

Don't expect professionals to have all the answers – it's not their fault, the likelihood is, especially with rare conditions, they have simply never come across the condition before. Become the expert on both the condition but also what it means for your child. Work WITH professionals, not against them. It's important to form a coalition with them and redress any imbalance of power.

4 Connect

Join Facebook groups and patient support groups and connect with other families with the same condition, they are a treasure trove of information, lived experience, love support and guidance. It is so important to find your tribe and feel that you can go to them for advice and anytime you are worried or just need a chat.

5 Research

By all means, research the condition and get involved in research and connect with others with the same condition and share stories. BUT don't define your child's future by others' development. Guides, while useful, don't account for love, tenacity, stubbornness, nurturing, encouragement, happiness, strength and determination.

6 Learn your no

Summon your inner strength. It's not a straight road, there are twists and turns, conserve your energy for the battles that really count. Don't waste time on prisoners. If a path or treatment doesn't align with your family's best interest, look for alternatives. Stay true to yourself.

7 Communicate

You may find that certain friendships end but also that your life becomes completely enriched by your new friendships and connections. Talk to your child like they understand everything. They're listening and feeling everything you do together. Make your world a better, more inclusive place for your child and speak your truth.

8 Involve your family

Letting children know their sibling has a rare condition is important. They will be the ones who exude acceptance automatically as they grow into the world. Use child-friendly language and encourage them to share with their peers. I love the book "Chromosome Kids like me" by Annette Fournier, it's easy to follow rhyme makes explaining complex genetics easy for children.

9 Ask and accept help

People tell me that I am supermum and they couldn't do what I do daily. While it comes from a good place, all this does is perpetuates the myth that we all need to be strong and 100% in control of our mental health at all times. It's unhealthy and leads to emotions and inner turmoil. Feeling like we can't express or we may appear weak. It's OK to not be OK.

10 Find breathing space

No one ever truly prepares you for sleep deprivation and fatigue in the first few years of parenthood. Add a child with a rare condition into the mix and this can be overwhelming. Make time for you. It doesn't have to be outlandish or kept to a strict routine. A quiet coffee, a guided meditation on your phone or a day away. It all helps reenergise and refocus us.

RARE REVOLUTION MAGAZINE

Produced by Rebecca Pender and the Rare Revolution Team - **A Rare Disease Collaboration** on behalf of Rare Revolution Magazine www.rarerevolutionmagazine.com @RareRevolutionMagazine

[5 Mental Health Resources to Help Rare Patients & Families Feel Less Alone]

As stated by All Stripes: "It is not surprising that having or caring for someone with a rare disease may have an impact on one's mental health." "A glimmer of hope, however, is that mental health is becoming a greater part of the conversation among the rare disease community and its supporters. ."
"To help contribute to this important conversation, compiled five of these rare disease-specific mental health resources that may help you feel less alone."

CHILDREN'S MENTAL HEALTH & WELL-BEING

Visit your hospital through your phone!
"Little Journey supports children and their families before, during and after healthcare interactions, reducing anxiety through the use of our multi-award winning smartphone App."
To take a look at their website & app please click here:
http://www.littlejourney.health/

Little Journey

For children

In these fun sessions for children, Diana Thornton will introduce children to what happens in their brain and body when they become anxious and will introduce some ways to deal with stress and anxiety drawn from her work as a counsellor and a mindfulness teacher. Parents must be there at the start to ensure that children access the appropriate video and should be nearby to offer support if required.

Mental Health videos for children

Great resource videos from MPSSocietyUK focusing on helping adults, children & young people with their mental health & wellbeing.

Mental Health videos for young people

Great resource videos from MPSSocietyUK focusing on helping adults, children & young people with their mental health & wellbeing.

For young people

Anxiety can sometimes be difficult to break away from. It can also be worrying when you experience it until you understand what is happening to your brain and to your body. In this series of videos, Diana Thornton will demystify what is happening to you physically when anxiety kicks in and will teach you some tried and tested techniques to help get it under control. Parents must be there at the start and should be nearby to offer support if required.

Helping Children Cope With Medical Examinations

Our Friends at Italian Macrodactyly and PROS Association:
https://www.associazione-nazionale-macrodattilia.org/english/ & https://www.aibws.org/

CHILDREN'S MENTAL HEALTH & WELL-BEING CONT:

MENTAL HEALTH & WELL-BEING FOR ADULTS LIVING WITH A RARE CONDITION:

Mental health and wellbeing videos for adults - dealing with anxiety

Updated: Nov 18, 2021

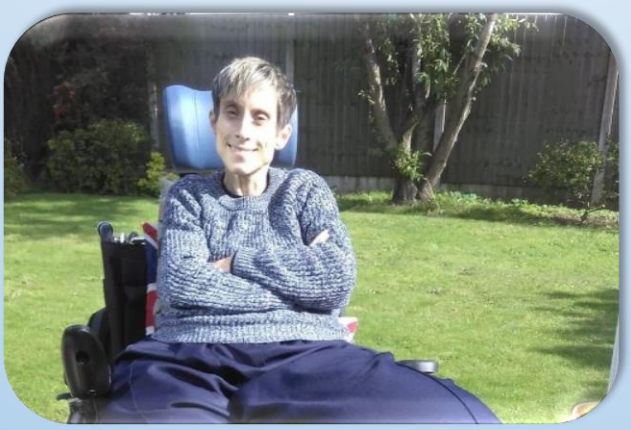
A series of seven videos aimed to help you deal with anxiety, stress and worry. Diana Thornton is a counsellor and mindfulness teacher and these videos will give you tips and techniques to use and help you when you feel anxious.

Great resource video from MPSSocietyUK focusing on helping adults with their mental health & wellbeing: Mental Health & Well-being Videos For Adults

Advice & guidance

This section is full of self-help and guidance for adults and young people living with a visible difference, as well as for parents of children who look different.

Self Help & Guidance For Adults This website has contains numerous sections for adults living with a rare condition. From confidence, self-esteem, relationships & social life, as well as coping with people's reactions.



WHEN PEOPLE POINT & STARE

When People Point and Stare Our chairperson Mandy opened up about how her mental health has been effected over the years by reactions from the general public to her physical differences.

HOW WE CAN HELP YOU

WE UNDERSTAND THAT IT ISN'T ALWAYS EASY TO ASK FOR HELP, BUT AS THE COST OF LIVING RISES FOR US ALL, WE WANT TO HELP YOU & YOUR FAMILY DURING DIFFICULT TIMES.

GoPI3Ks Grant Application Form

DID YOU KNOW THAT GoPI3Ks CAN OFFER WINTER GRANTS & ALSO HELP WITH THOSE FESTIVE EXPENSES.

We want to make sure that those living with PROS & their families are able to enjoy the festive season & get through the winter.

We can help with:

- **THE COST OF FESTIVE FOOD**
- **THE COST TO TRAVEL TO SEE FAMILY**
- **THE COST OF A FAMILY DAY OUT**
- **AND MORE**

AS THE COST OF LIVING RISES & WINTER STARTS, GoPI3Ks WANTS TO HELP YOU.

DID YOU KNOW?

We know how important it is to keep warm & that PROS can cause limited mobility which we know can mean you feel the cold.

GoPI3Ks will be happy to consider applications for help with rising energy bills.

GoPI3Ks is a charity that can help you with those extra costs that living with PROS can bring to your daily life.

DON'T FORGET

GoPI3Ks can still help with:

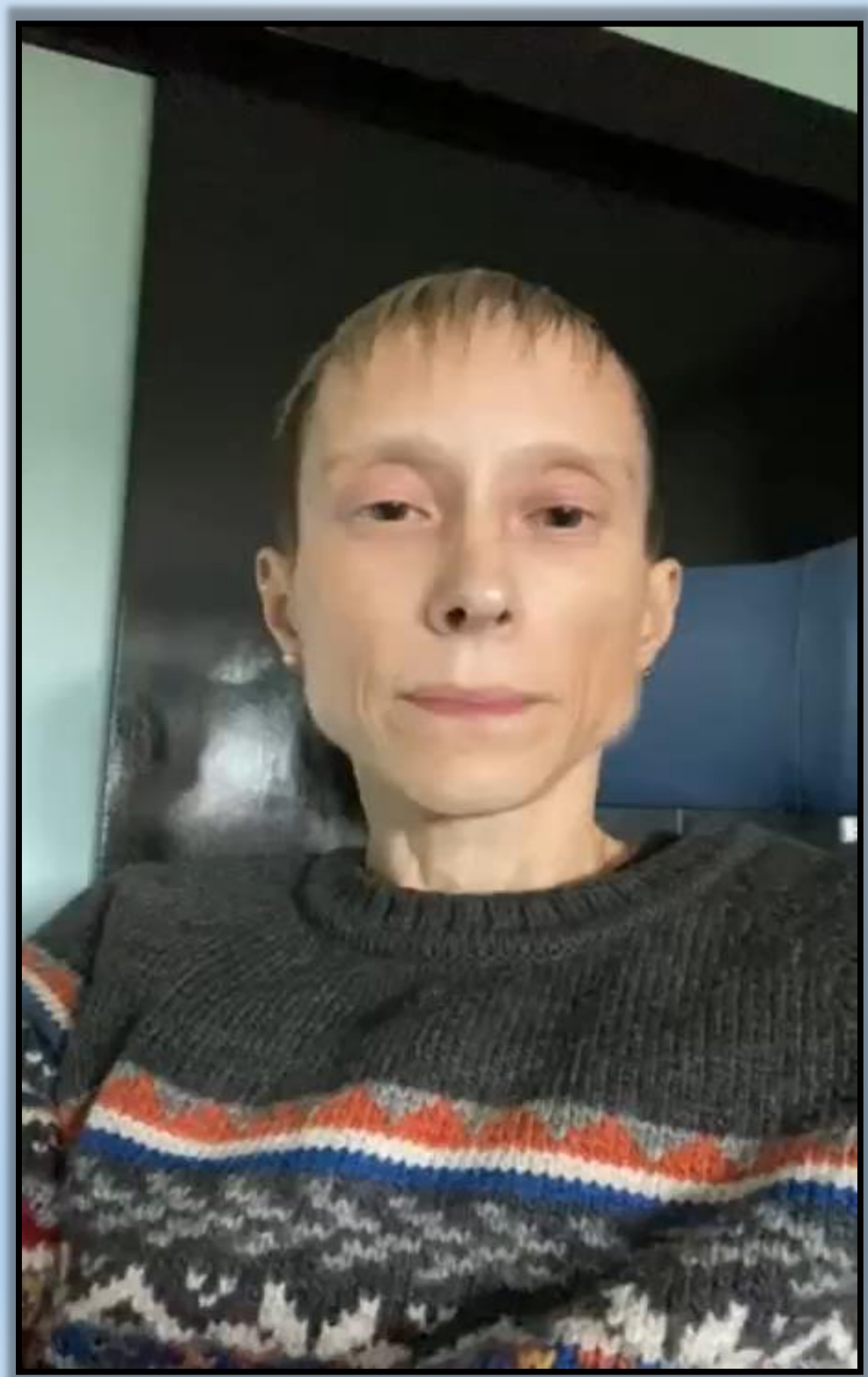
- **Hospital appointment expenses**, **travel & accommodation.**
- Help with equipment or aids: **Wheelchairs, altering of clothing, bespoke clothing &/or shoes, cost of driving lessons or aids needed in your vehicle to transport wheelchairs & more.**

WE WELCOME APPLICATIONS FROM PROS FAMILIES & INDIVIDUALS OVERSEAS.

ALPELISIB NEWS

I GET QUITE A LOT OF MESSAGES & EMAILS FROM PEOPLE ASKING HOW THINGS ARE GOING TAKING ALPELISIB. SO HERE IS MY LATEST VIDEO (12 MONTHS AFTER STARTING THE DRUG)

Mandy's Update On Taking Alpelisib December 2022



As some of you may know, GoPI3Ks have partnered with https://www.allstripes.com/program/pros to help advance research for PROS. If you would like to know more please see videos from some of the PROS ambassadors below answering some important questions about joining: AllStripes

AllStripes PROS Ambassador Kelsie, answers "How does AllStripes keep my child's data safe?"
Kelsie's AllStripes Video

AllStripes PROS ambassador Mandy answers: "Does it cost to be part of AllStripes?"
Mandy's Video

Lindsey a PROS AllStripes ambassador answers "Is it time consuming to take part in AllStripes?"
Lindsey's AllStripes Video

AllStripes PROS ambassador James answers: "What are the benefits to signing up to AllStripes?"
James' AllStripes Video

HOW YOU CAN HELP FUNDRAISE FOR GOPI3KS

Here are 3 ways you can raise money for GoPI3Ks, whilst you partake in some retail therapy.

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.

GoRaise

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

PayPal
Giving Fund

Up & coming events in 2023

EDUCATIONAL MEETING ON PROS FOR DOCTORS & NURSES – SUPPORTING THE PROS PATIENT PATHWAY.

We've been working closely with Lucy, who works in medical communications for rare diseases. Lucy also has two children, Felix & Lily, Lily was born in 2020 & has been diagnosed with PROS. As well as with Dr Maanasa Polubothu who is a Paediatric dermatologist at Great Ormond Street Hospital. Special thanks to Lucy for the Information about the meeting below:

On the **afternoon of 24th April 2023** GoPI3Ks will be collaborating with Dr Maanasa Polubothu at Great Ormond Street Hospital (GOSH) to host an educational meeting **'PIK3CA Related Overgrowth Spectrum (PROS): Navigating the patient journey'** for doctors and nurses. The meeting will be live at GOSH as well as virtual.

Why do we need this meeting?
One of the things that we hear time and time again in our community is that we struggle to access the right doctors to help us with our care. With this in mind we wanted to organise a meeting aimed at doctors from GPs, to oncologists, to orthopaedics to plastic surgeons. The aim will be to provide education on PROS including genetics, the spectrum of how patients can be affected and the treatments that can be used to help.

It is also timely because GOSH has recently been awarded **Rare Disease Clinical Network** Status which means that your GP can refer directly to them to create a care plan that will hopefully mean you or your family member can access the right specialists.
In addition to this, Guy's and St Thomas' Hospital will also be launching a much needed Adult clinic and hopefully all PROS patients will be able to access this service in time too! A specialist doctor from France will be leading this clinic.

The meeting will talk about these two new clinics and help to spread the word that people can be referred here as well as how doctors can help to refer to the most appropriate people.

What can you do?
We really need your help here for this! Please mention this to your doctors and with their permission pass on their email addresses to us so that we can invite them.
gopi3ks@yahoo.com

Can I attend the event?
Patients and their families are more than welcome to attend the meeting virtually and we will have limited in person places, however, please note that the meeting content will be aimed at doctors and nurses. After the meeting we will be preparing a plain language summary to circulate to the patient community so that you can all hear what has been discussed!
Please let us know if you would like to attend.

FAMILY WEEEKND – 2023

Due to Covid it has been difficult for us to host a family weekend, but we hope to be able to all come together again in 2023, stay tuned for more information next year.

For those who didn't attend or are new to the group, this involved talks from relevant doctors in the field of PROS, as well as time for patients, families & children to get to know each other & socialise, something that is invaluable for those in the rare community. Take a look at an exert from our previous family weekend here: Family Weekend 2018

YOUR GOPI3KS CHARITY TRUSTEES.

MANDY SELLARS - CHAIRPERSON

It seems like a blink of an eye since we all sat down last year to write our annual message for the newsletter. It has been wonderful to be able to help some of our GoPI3Ks families & get back to face to face with people to raise awareness of GoPI3Ks & PROS & I hope we can help more of you next year too. In 2023 we hope to bring you another Family Weekend as well as an educational meeting on PROS, bringing together doctors from various fields to help ease the journey for PROS patients & their families. We will let you know dates asap. To finish I want to wish you all a very Merry Christmas for those that celebrate & a healthy & happy start to 2023 for everyone. #StrongerTogether

SUE HARPER - TREASURER

As 2022 comes to an end I firstly want to say thank you to our Chairperson Mandy for raising over £1500 this year through fundraisers & a written blog. It has also been very rewarding to be part of GoPI3Ks & be able to help some of our GoPI3Ks families with financial grants & we hope to be able to help more of you in 2023.
We understand that with the cost of living rising that it isn't always easy to enjoy the festive season & holidays, but I hope whatever you do it brings you happiness, joy & laughter.

AMANDA KENYON - SECRETARY

Hi! The festive season is almost upon us and the dark nights are drawing in and the temperature is starting to drop. It has been another difficult year especially with the Cost of Living. Having a disability can come with additional costs and we can offer small grants if you meet the criteria . Please contact us for details. Your local council could also have a fund you could be eligible for. Please check the website of your local authority for details. There are other organisations and charities that can help through these challenging times whether that be financially or having someone to listen. There are links included below. However you spend the festive season, I hope it's in good health and happiness.
There are other organisations out there to help too:
https://www.mind.org.uk/donate/
https://england.shelter.org.uk/get_help
https://www.samaritans.org
https://refuge.org.uk

KATIE KAVANNAH - TRUSTEE

I'm baffled to be writing another festive message, as I'm sure it hasn't been a year since we all celebrated this joyous occasion. But what a 12 months it's been. We started the year full of uncertainty still surrounding Covid, and how or if we would return to normal. But thankfully we have all been able to adapt to a 'new normal' and relatively return to our normal lives. Which I for one am extremely grateful for. As such we have all been able to engage and share in experiences with family and friends across recent months. And this I hold onto as we go into the Christmas period with excitement and anticipation.

I feel privileged to be part of GoPI3Ks team, and grateful that we could help some of you throughout this year. And I know we all look forward to supporting many more of you throughout 2023.
I wish each and every one of you a wonderful Christmas, and sending best wishes for the new year. Here's to a happy and healthy 2023