GoPI3Ks

NEWSLETTER

Genetic Overgrowth PI3K Support

www.gopi3ks.com

December 2025

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 big thank you goes Dr Ralitsa Madsen & her team: Alex Musk, Sweta Swaminathan, Mark Bekala & Oliwia Mruk for their research update & continued support for the PROS community & their dedication to PROS research.

2025 PROS COMMUNITY UPDATE from Dr Ralitsa Madsen & Team

Dear GoPI3Ks readers,

As we approach the festive season, I am once again excited to share this year's update with you. Writing this newsletter is always one of my favourite responsibilities of the year. It is a chance to step away from the endless to-do-list and reflect on the meaningful progress we *have* made.

For those who may be reading one of my updates for the first time: I am Dr. Ralitsa Madsen, and my research group studies the PI3K signalling pathway – the biochemical system inside our cells that is disrupted in PROS. Our mission is to understand and predict how *PIK3CA* mutations change the behaviour of cells and tissues, and to use this knowledge to guide safer and more effective treatment strategies.

This year, I am especially excited to share that we have reached the final stage of what has truly been a *tour de force* of disease modelling since I started the lab. After several years of hard work and continuous support from my

mentor and collaborator, Prof. Robert Semple, we now have a comprehensive collection of human induced pluripotent stem cells (iPSCs) carrying a wide range of activating ***PIK3CA* mutations**. Thanks to Prof. Semple, we have also revived precious patient-derived cells from my time in Cambridge – samples originally donated by individuals in the PROS community. These are incredibly valuable resources, and we are eternally grateful to the patients who made this possible.

What makes this milestone transformative is that, for the first time, we are able to model **PIK3CA mutations in the correct human cell type** – and to do so at scale using the iPSCs we have generated. We began with **endothelial cells**, which form the inner lining of blood vessels and are central to the vascular malformations so often seen in PROS. Over the past three months in particular, our team has seen first-hand just how critical it is to study PIK3CA mutations in *precisely engineered, human venous endothelial cells*. Without the right cell type, some mutations may look completely “silent”, even though we know they cause debilitating disease. I experienced this myself during my PhD, when one of the less potent *PIK3CA* mutations showed no measurable effect in the iPSC lineage itself. Conversely, if one uses the non-human model systems or does not control carefully for the **exact dosage** of each mutation, there is a risk that conclusions are drawn that do not reflect the true biology of the human condition. Unfortunately, this remains a challenge in the field, and it is one of the reasons why I of caution against over interpreting scientific studies that do not model PROS in a sufficiently realistic way, especially if the authors do not acknowledge such limitations upfront. Ultimately, no model is perfect but we have got to be honest in our reporting about it.

Now that we have the appropriate endothelial models, we can study the true biology of the many PROS mutations. We can see clear, quantifiable differences that closely recapitulate key aspects of the human pathology. This is an incredibly important step forward. And the timing could not be better. In fact, the first week of

December, we will be running our **first full drug targeting experiment** using these disease-relevant models. We will be testing clinically approved medicines, as well as the Relay Therapeutics drug currently in clinical trials. Our goal is to directly compare how different *PIK3CA* mutations respond in the correct cell context – something that has never been done systematically before.

To give you a sense of what we now have available:

- engineered iPSCs with **H1047R** and **E545K**, each linked to fluorescent reporters so we can track when they become endothelial cells
- patient-derived iPSCs carrying **H1047R**, **E418K**, and others that are yet to be brought out of storage
- the ability to produce arterial and venous endothelial cells reliably and at scale; funding permitting, this will be expanded upon in coming years to capture a wider range of PROS-relevant cell types

We hope to share the first results publicly early next year. This will open the door to many follow-on studies, and, we believe, a clearer path toward more precise and personalised therapeutic strategies.

A major transition for our lab

Another significant update is that our entire research group will be relocating to the **CRUK Scotland Institute and the University of Glasgow**, with the move planned for mid-January 2026. Although this comes at a difficult moment – the University of Dundee has been facing severe financial challenges – the team has handled the uncertainty with unwavering resilience. The move will undoubtedly be stressful, but it also brings enormous opportunity. In Glasgow, we will be joining a thriving research environment with outstanding facilities and collaborators.

One idea, I am particularly keen to explore, together with partners in the UK and internationally, is the possibility of establishing a secure and ethically governed **PROS patient tissue biobank**. Many of you have expressed interest in contributing biopsy material to support research, and we want to develop a practical pathway that ensures such donations are meaningful, well-preserved, and maximally valuable for downstream studies. This cannot be done without careful infrastructure and will likely require long-term planning and investment, but I am hopeful that it is possible and that Glasgow will be the place to build it.

Looking ahead: more engagement and involvement

Once we have settled into the CRUK Scotland Institute, I am keen to offer **quarterly live lab updates** for anyone in the PROS community who would like to learn more about our work. These will be held online and will include the option to see the lab virtually, meet team members, and hear what we are working on in real time. I hope this will make the research process feel less distant, and offer a window into why rigorous science – especially science that aims to be clinically meaningful – often takes longer than we all wish it would. I would be keen to hear from the PROS Community whether this would be of interest.

Closing remarks

In closing, I would like to thank you – sincerely – for your patience, your trust, and your continued interest in our research. The work we are doing is slow by necessity, but this year we have crossed a critical threshold. We now have the right models, in the right cell types, and we are finally able to ask the mechanistic and therapeutic questions that matter most.

We look forward to sharing much more progress with you in 2026. Until then, we wish you and your loved ones a very Merry Christmas and a Happy New Year.

With warmest regards, **Dr. Madsen and Team**

This is us with our lab mascot, Oreo. We adopted Oreo from a nearby farm here in Scotland and visit him regularly for a catch-up! From left-to-right (excluding Oreo): Alex Musk (PhD student), Sweta Swaminathan (PhD student), Dr. Ralitsa Madsen (group leader), Mark Bekala (research technician), Oliwia Mruk (PhD student).

Dr Ralitsa Madsen who works researching PROS has won the RSE Dame Anne McLaren Medal for outstanding achievements to scientific research.

Ten individuals from diverse disciplines, including medicine, disease research, marine biology, law, astrophysics, and machine learning, attended a reception at the RSE on George Street to receive their awards.

The RSE's medals are nominated by RSE Fellows and awarded to individuals to recognise their exceptional contributions and achievements to their field. These medallists impact lives all over the world and highlights the talent that exists in Scotland's research sector: Full article

RSE Dame Anne McLaren Medal - Dr Ralitsa Madsen
Principal Investigator, Round 8 Future Leaders Fellow, University of Dundee

The RSE Dame Anne McLaren Medal is awarded to Dr Ralitsa Madsen, Principal Investigator, Round 8 Future Leaders Fellow, University of Dundee, for her advancing knowledge in the quantitative understanding of the growth-promoting phosphoinositide 3-kinase (PI3K) signalling pathway, and her endeavour of making knowledge accessible and useful through public and patient involvement in research activities. Beyond her formal appointment to serve on the UK Committee on Research Integrity, Dr Madsen is also an internationally recognised figure in Open Science and research integrity, advocating for the latter as a driver for research excellence.

Dr Ralitsa Madsen said, *"It is a tremendous honour to be awarded the RSE Dame Anne McLaren Medal 2025. This recognition would not have been possible without the support of my mentors, students and family along the way – I am deeply grateful to them all. I hope this recognition will also inspire many other early-career researchers to pursue science with passion, integrity, and a commitment to public benefit. For me personally, it is a huge motivation to keep pursuing my mission to crack the quantitative and highly context-dependent PI3K signalling "code" in health and disease."*

OUR 2025 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 adults & families.

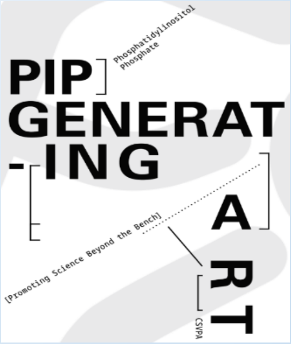
BIRTHDAY FUNDRAISER

Mandy (our chairperson) 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 as it was a big 50th birthday we were overwhelmed by the kindness of people who helped us raise over £1,000

We are also very grateful to other supporters who were kind enough to make GoPI3Ks their charity choice for their birthday Facebook fundraiser

PIPGEN DONATION

Last year we had the pleasure of working with a team at PIPGen: PIPgen on their PIPGenerating art project. As part of the event people were able to donate to us & we received nearly £90



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.

PAYPAL GIVING FUND

Thank you to those who choose to support us when using PayPal. Your kind gesture donated nearly **£50** to the charity. If you would like a sign up to donate as you shop: PayPal Giving Fund

MONTHLY DONATIONS

Did you know you are able to donate monthly to us at GoPI3Ks. We are lucky to have a monthly donation from one of our supporters.

If you would like to do this, then please click on the link to our donation site and you can set this up: https://donorbox.org/gopi3ks

DONATIONS

A huge **THANK YOU** goes to those who wanted to support us when they heard of the work we do to support those living with PROS. We received over **£500**

NEW BANK ACCOUNT DETAILS

We recently changed our banking details. For anyone that would like to donate directly these are our new details:

Account name: GOPI3KS
Account number: 17811341
Sort code: 01-00-04
IBAN: GB15NWBK01000417811341
BIC: NWBKGB2L

Thomas & friends fundraiser

Thank you Thomas & friends for organising such a fantastic fundraiser, you are all amazing ☺

Hi, I am Thomas. Me and my friend raised **over £600** for GoPI3Ks at my mum's birthday party in under 4 hours. We put temporary tattoos on guests for a donation. And my hairdresser gave me a gift card for a wash and blow dry. I ran an auction for this an it raised £500 because my mum has crazy friends ☺

Vicky's Loch Ness Cycle Challenge

My good friend Vicky took part in a 66-mile bike ride around Loch Ness in April. It was a challenging route which included a 4.8-mile-long hill right in the middle of it! There was also a time limit on the ride as the roads are closed to traffic for 5 hours and 38 minutes and re-opened after this time, which meant an average speed of 13mph was required. She smashed it like we knew she would **raising over £850** for us.

THANK YOU SO MUCH VICKY

Susan's Kilt walk for GoPI3Ks

Susan raised an amazing £1,647.80 by doing an 18mile Kilt-walk in Aberdeen.

Susan said "as some of you may know, my granddaughter was diagnosed with "PROS" "Through this trying time GOPI3KS supported Annabelle & her family greatly & they will continue to do so over the years". "I would like to give something back as GOPI3KS is a small charity".

Annie’s knitted gnomes!

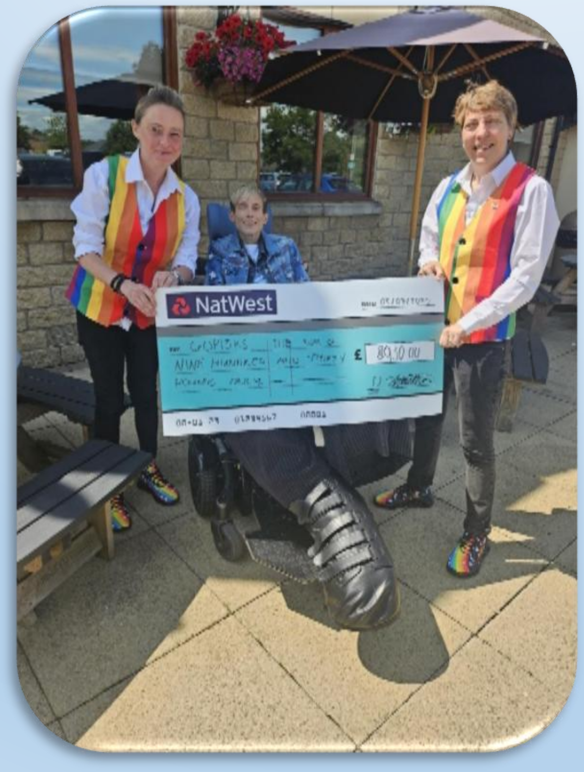
A supporter of the charity, Annie Boothman has been knitting gnomes to sell to raise money for GoPI3Ks.
So far she has raised a fab £35. THANK YOU ANNIE

Abigail and Davina’s Pride night

Earlier in the year my friends Abigail and Davina held a night of entertainment, selling pride related items and a raffle.

With only two weeks of organising they hosted this event that raised an **amazing £960** for us.

Thank you so much to you both for such a wonderful fundraising event.



HELPING GoPIKs
CHARITY
SUPPORTING MANDY
SELLARS AND MANY
OTHERS WITH
segmental overgrowth
CONDITION

Well we have 3 fantastic Artists for our first ever charity event on 27th June 2025.

BUPSI BROWN As seen on Xfactor along with our very own Black Country's finest, MISS LIVI MAE and SARAH MAC

..And with tickets selling fast, it would be a shame to miss out, as theres only 130 seats available. so get in touch, get your tickets and come on down to Summerhill social club, St John's Rd, Tipton DY4 9RZ

£5 per ticket, there will also be a buffet and a Raffle, so what are you waiting for?

Doors open 7.30pm till late

TICKETS FIVER IF IT'S NOT FOR
YOU .. YOU CAN STILL SEND THAT
FIVER
IF YOU CANT SEND THAT FIVER
SHARE THE HELL OUT OF THIS
POST
SEND IT FLYING HIGH
EVERY LITTLE INPUT GOES WAY
FURTHER THAN ANYONE WILL
EVER KNOW

Amelia’s Swimming Challenge


SUMMER SWIMMING CHALLENGE 5000M

"Swimming is my happy place"

This summer, Amelia will be swimming 5000m to raise money for GoPI3Ks and Great Ormond Street Hospital

Please help support Amelia by commenting below, sharing the link and making a small donation using the link attached.
All support will be greatly appreciated and help raise awareness of two amazing charities.

I'm raising £500 to support two amazing charities; GoPI3Ks and Great Ormond Street Hospital by completing a 5000m swimathon.

My name is Amelia and I am almost 10 years old and I was diagnosed with PIK3CA-related overgrowth spectrum (PROS) when I was just one years old. I have had over 40 appointments at GOSH since my diagnosis and I have a great team of consultants who help and monitor my health.

GoPI3Ks is a special charity that support me and my parents as living with a rare condition can be really hard sometimes. Amelia raised a **fantastic £652**

What an amazing achievement, thank you so much Amelia, you are a star

Linda’s Christmas Fayre Fundraiser

A huge thank you to Linda Robson who’s granddaughter has PROS.

She organised 2 Christmas fundraisers where she made homemade gifts for people to buy.

They made over £200

GRANTS GIVEN TO PROS FAMILIES IN 2025

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

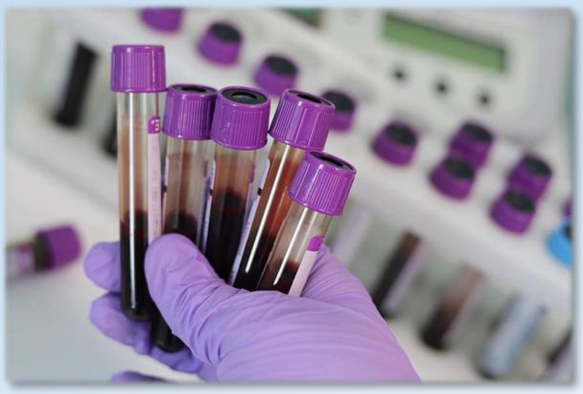
This year we were able to help a family living outside of the U.K meet a shortfall in their insurance so that their child could continue with medication that was helping with their PROS symptoms.

We helped several number of families over this year with travel to & from vital hospital appointments.

Earlier this year we were able to fund for a new pair of bespoke shoes to be made for one of our younger members. Megan was over the moon with these & was happy to show you her brand new shoes

GRANTS GIVEN TO PROS FAMILIES CONT:

With the cost of living continually increasing, we have been able to help one of our families with their rising costs. This gave them some breathing space during a difficult time.



We have been able to help a family from Singapore with medical bills, including: blood tests, doctors consultation's, injections & medication,

We helped one of our members who struggles to buy clothes due to how PROS affects them. So we were able to help them purchase a sewing machine so they can now make their own clothes that fit more comfortably.

We were also able to help some of our adults with PROS & families with children who have PROS by offering a small monetary grant to help with those extra costs during the festive season.

If anyone would like to apply for financial assistance please use this link to fill out our application form: GoPI3Ks funding application form

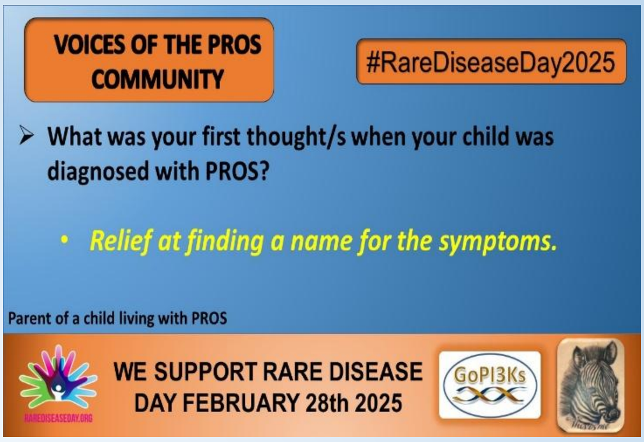
February 2025

In February 2025 as always we took part in Rare Disease Day by sharing information on the different aspect of PROS. We also asked the PROS community:

"What was your first thought/s when you/your child was diagnosed with PROS?"

"What are your thought/s now of living with PROS or as a parent to a child living with PROS".

Voices of parents and carers



VOICES OF THE PROS COMMUNITY

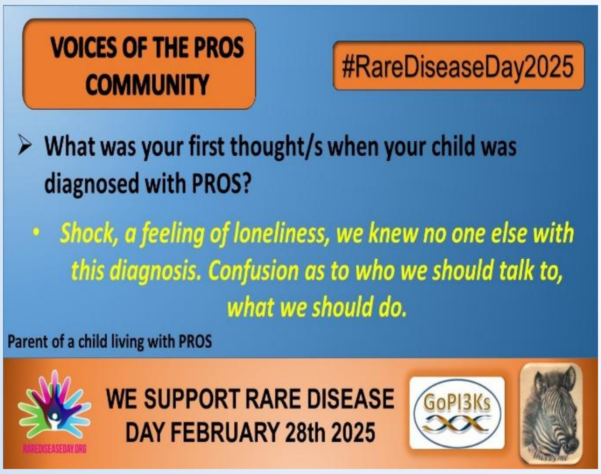
#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- *Relief at finding a name for the symptoms.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

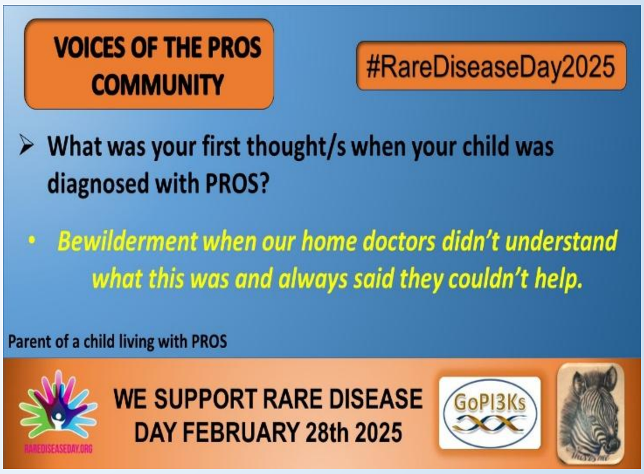
#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- *Shock, a feeling of loneliness, we knew no one else with this diagnosis. Confusion as to who we should talk to, what we should do.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- *Bewilderment when our home doctors didn't understand what this was and always said they couldn't help.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

VOICES OF THE PROS COMMUNITY

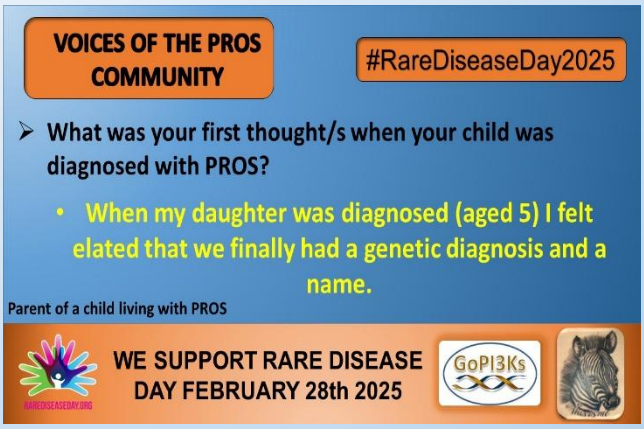
#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- *When my daughter was first diagnosed it was a feeling of fear and the unknown. Lots of what ifs and many tears.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- When my daughter was diagnosed (aged 5) I felt elated that we finally had a genetic diagnosis and a name.

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

VOICES OF THE PROS COMMUNITY

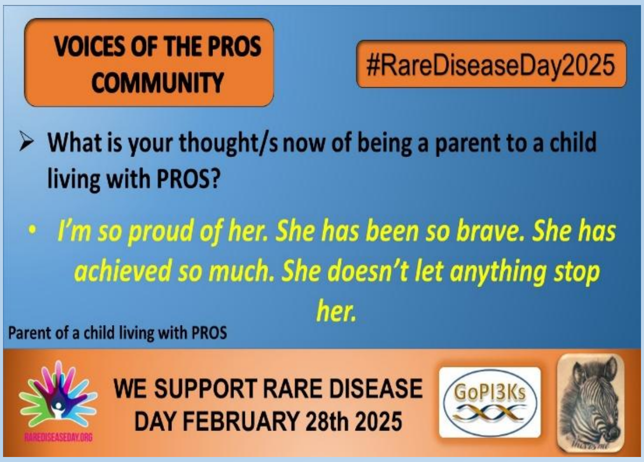
#RareDiseaseDay2025

➢ What was your first thought/s when your child was diagnosed with PROS?

- Until the diagnosis I had been going crazy googling all sorts.

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

➢ What is your thought/s now of being a parent to a child living with PROS?

- *I'm so proud of her. She has been so brave. She has achieved so much. She doesn't let anything stop her.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

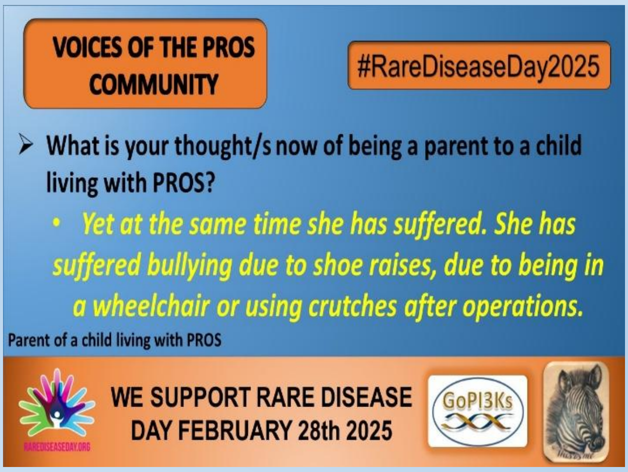
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *Now there is still a worry of changes and progression but a lot of pride. I always see her and what she's overcome and now we turn these feelings into strength and advocate for betterment in care and have new mums get in touch with us for support.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



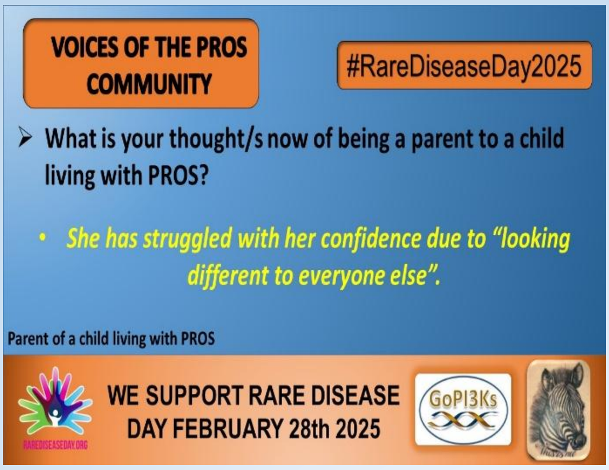
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *Yet at the same time she has suffered. She has suffered bullying due to shoe raises, due to being in a wheelchair or using crutches after operations.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



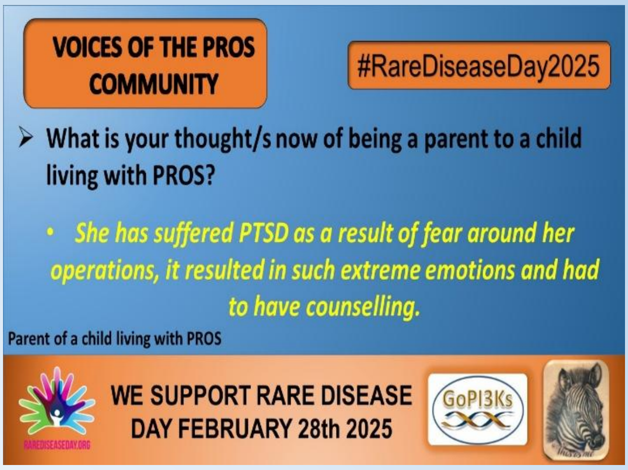
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *She has struggled with her confidence due to "looking different to everyone else".*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



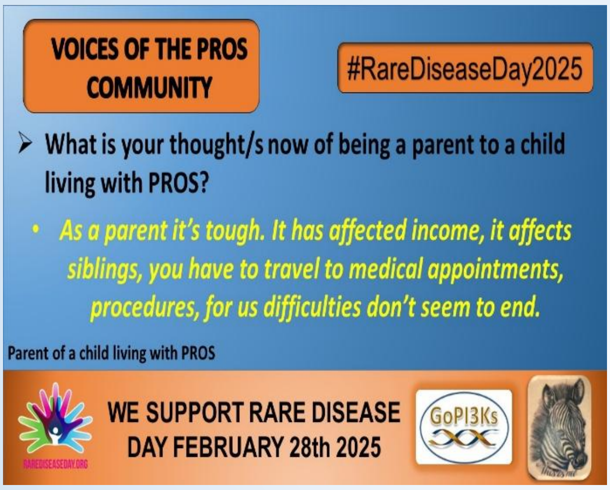
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *She has suffered PTSD as a result of fear around her operations, it resulted in such extreme emotions and had to have counselling.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



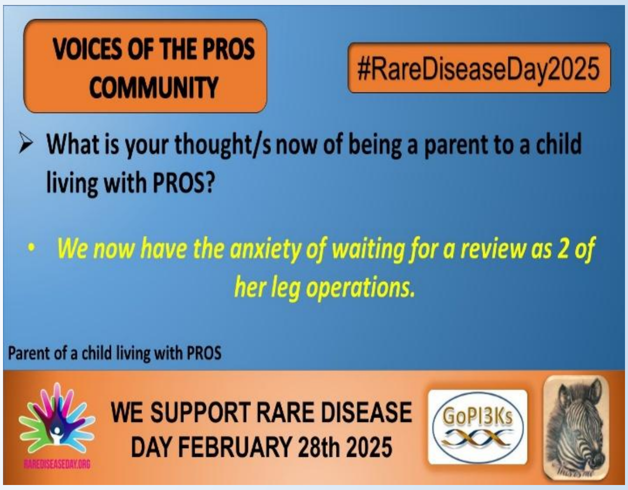
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *As a parent it's tough. It has affected income, it affects siblings, you have to travel to medical appointments, procedures, for us difficulties don't seem to end.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



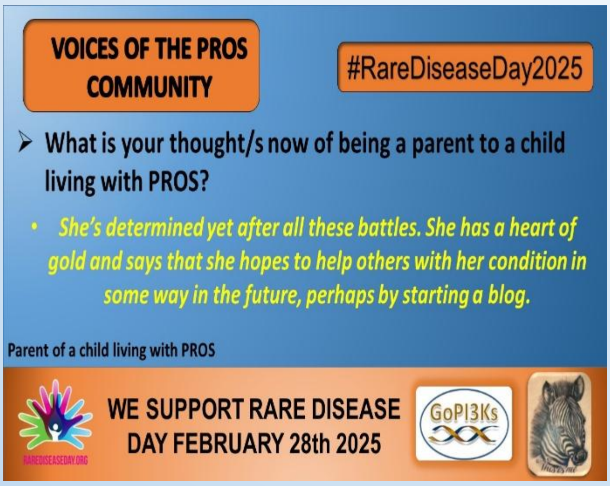
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *We now have the anxiety of waiting for a review as 2 of her leg operations.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



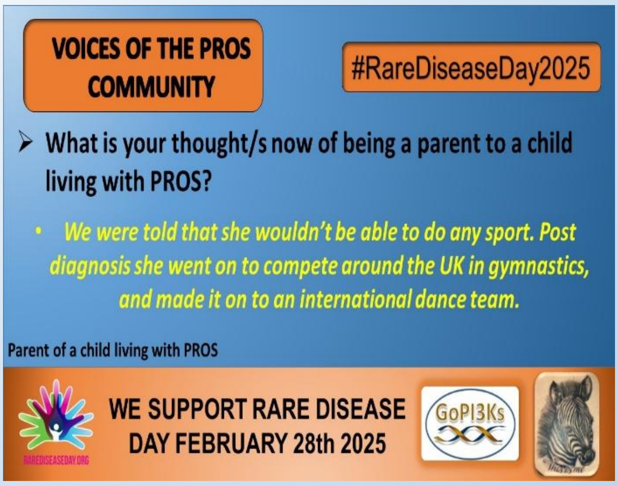
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *She's determined yet after all these battles. She has a heart of gold and says that she hopes to help others with her condition in some way in the future, perhaps by starting a blog.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

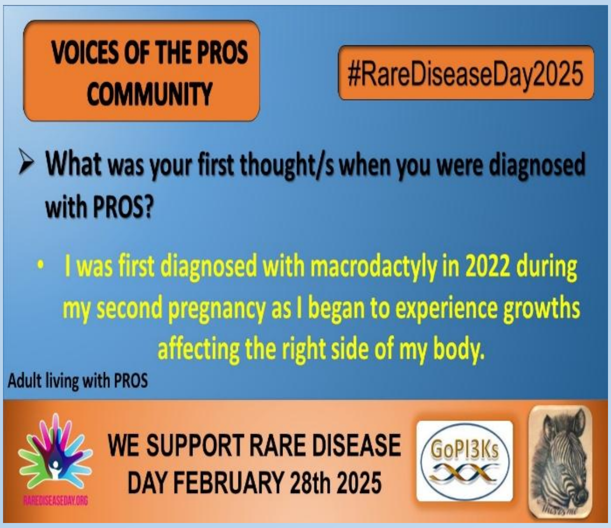
#RareDiseaseDay2025

- What is your thought/s now of being a parent to a child living with PROS?
  - *We were told that she wouldn't be able to do any sport. Post diagnosis she went on to compete around the UK in gymnastics, and made it on to an international dance team.*

Parent of a child living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

Voices of those living with PROS



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - I was first diagnosed with macrodactyly in 2022 during my second pregnancy as I began to experience growths affecting the right side of my body.

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?

Being given a medical term to be able to describe my fingers, gave me so much power back and really positively impacted my self-esteem.

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

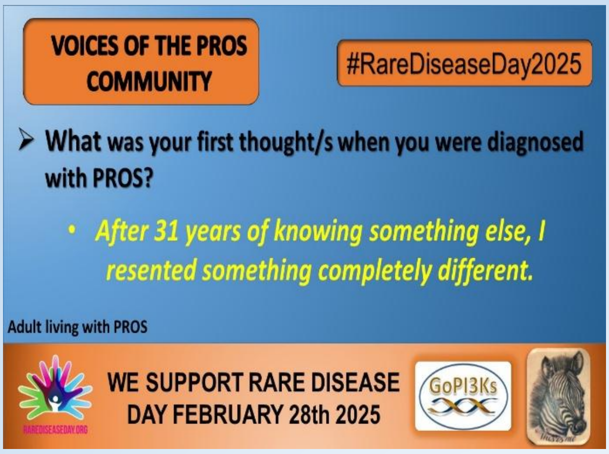
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *I was excited as I never thought a diagnosis would be found*

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



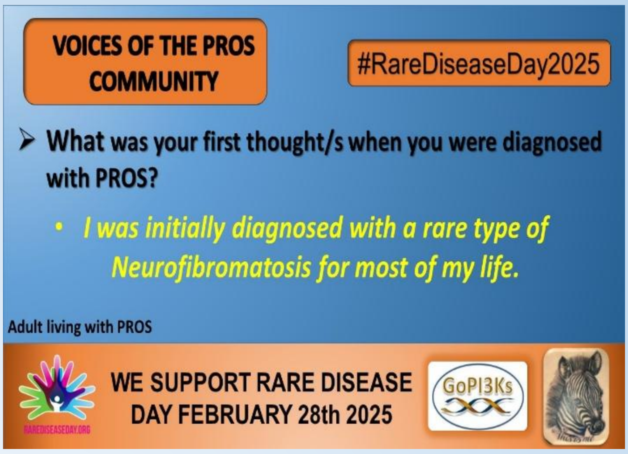
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *After 31 years of knowing something else, I resented something completely different.*

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



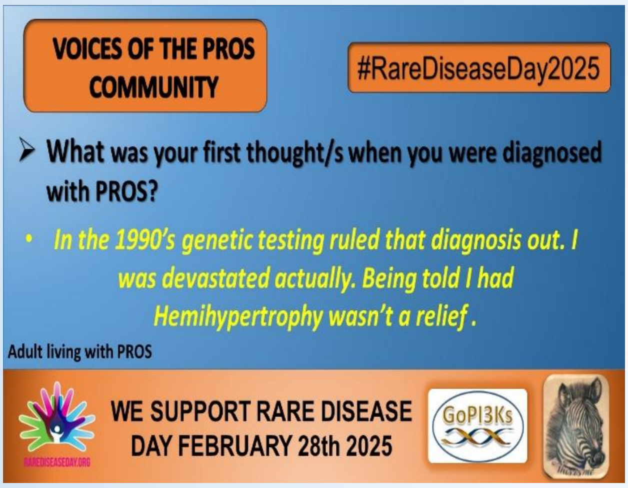
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *I was initially diagnosed with a rare type of Neurofibromatosis for most of my life.*

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



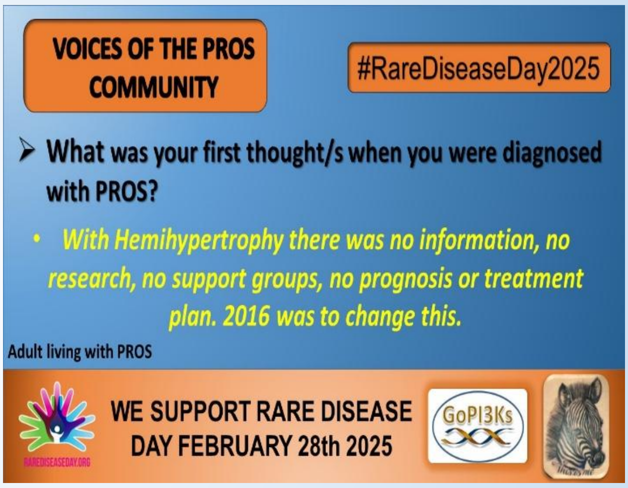
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *In the 1990's genetic testing ruled that diagnosis out. I was devastated actually. Being told I had Hemihypertrophy wasn't a relief .*

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *With Hemihypertrophy there was no information, no research, no support groups, no prognosis or treatment plan. 2016 was to change this.*

Adult living with PROS

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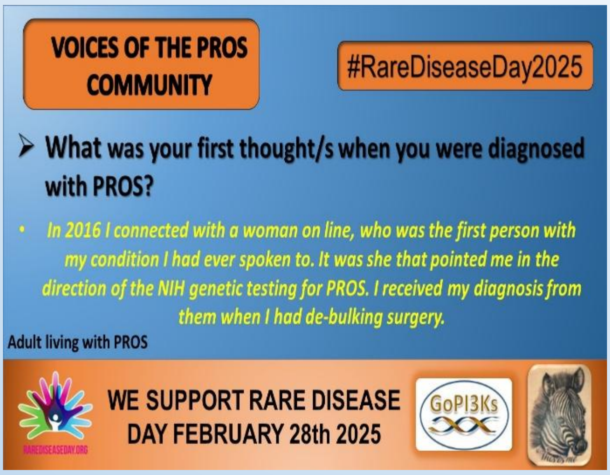

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *In 2016 I connected with a woman on line, who was the first person with my condition I had ever spoken to. It was she that pointed me in the direction of the NIH genetic testing for PROS. I received my diagnosis from them when I had de-bulking surgery.*

Adult living with PROS

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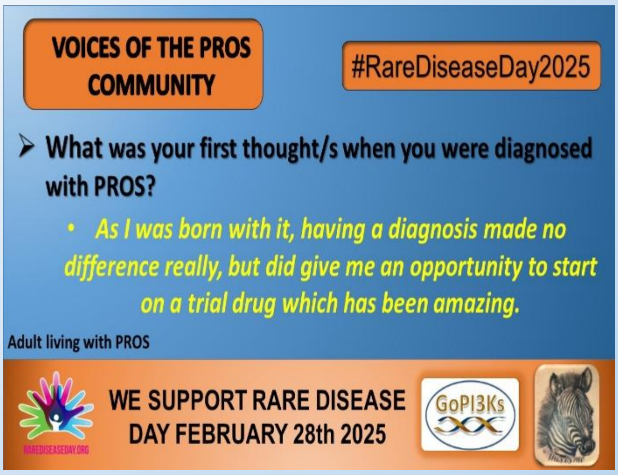

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *As I was born with it, having a diagnosis made no difference really, but did give me an opportunity to start on a trial drug which has been amazing.*

Adult living with PROS

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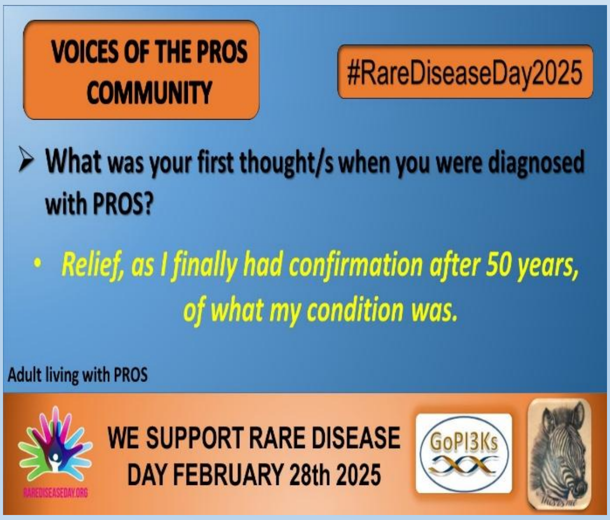

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *Relief, as I finally had confirmation after 50 years, of what my condition was.*

Adult living with PROS

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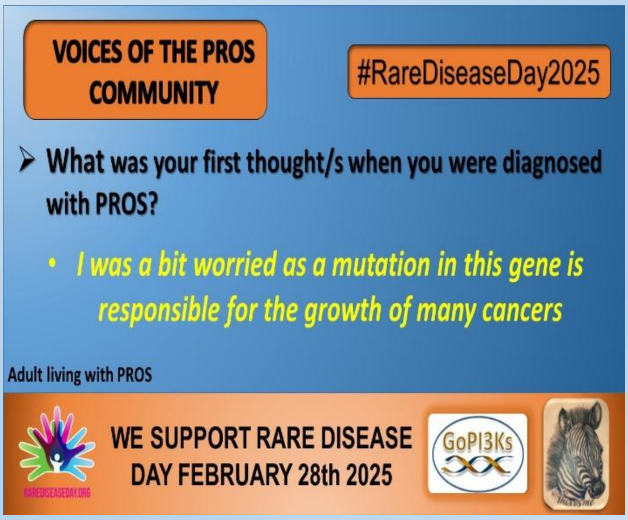

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *I was a bit worried as a mutation in this gene is responsible for the growth of many cancers*

Adult living with PROS

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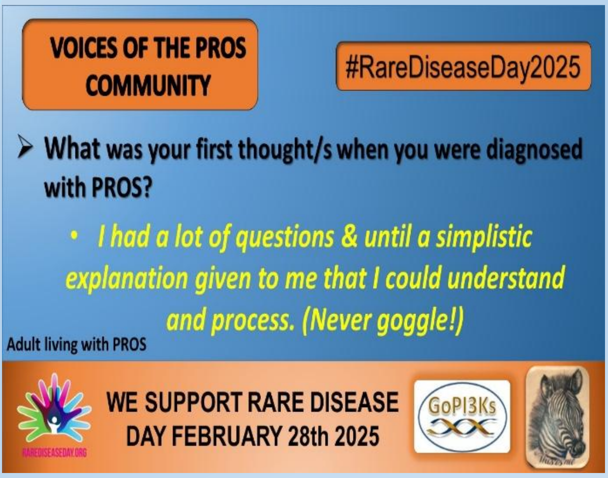

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *I had a lot of questions & until a simplistic explanation given to me that I could understand and process. (Never goggle!)*

Adult living with PROS

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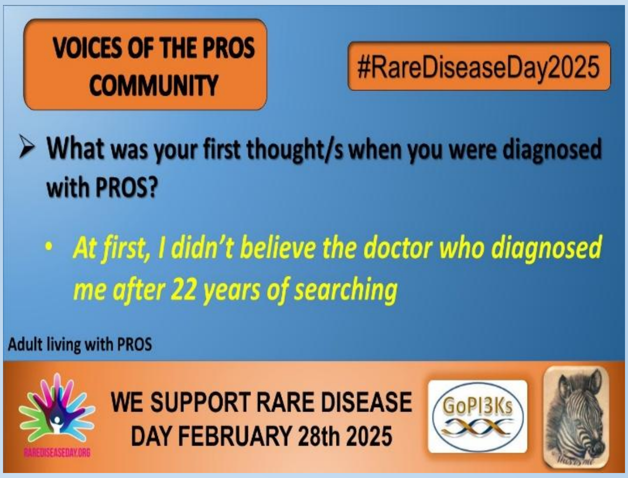

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *At first, I didn't believe the doctor who diagnosed me after 22 years of searching*

Adult living with PROS

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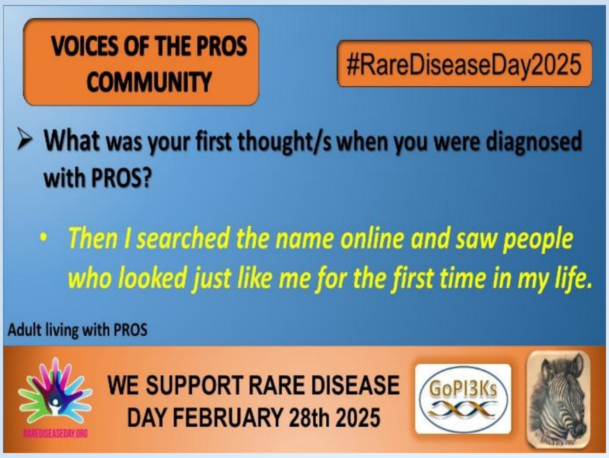

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *Then I searched the name online and saw people who looked just like me for the first time in my life.*

Adult living with PROS

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What was your first thought/s when you were diagnosed with PROS?
  - *It was a surreal moment of realization.*

Adult living with PROS

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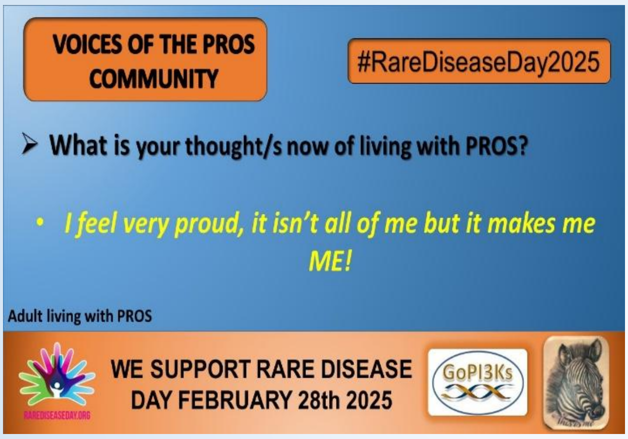

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *I feel very proud, it isn't all of me but it makes me ME!*

Adult living with PROS

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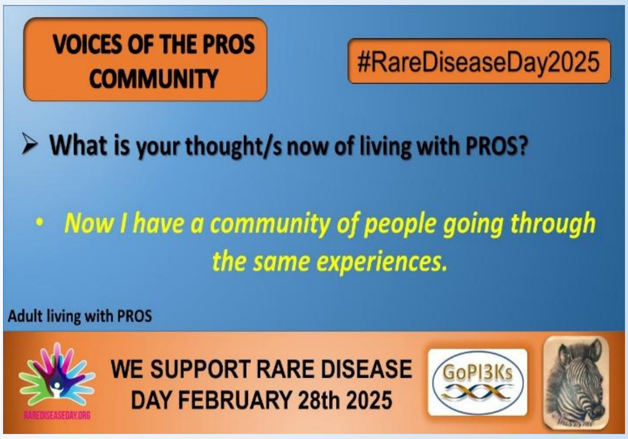

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *Now I have a community of people going through the same experiences.*

Adult living with PROS

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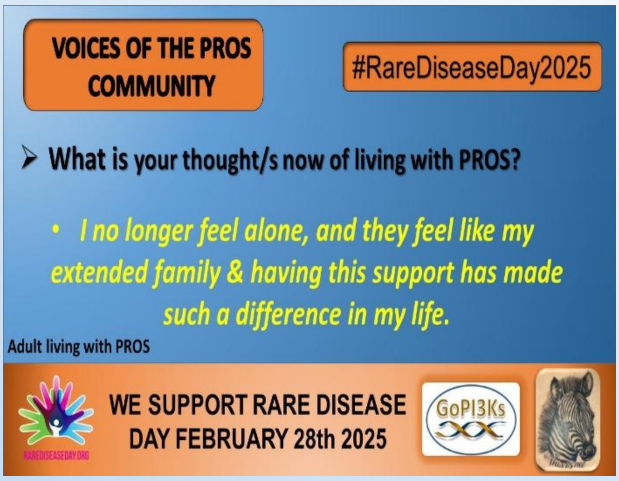

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *I no longer feel alone, and they feel like my extended family & having this support has made such a difference in my life.*

Adult living with PROS

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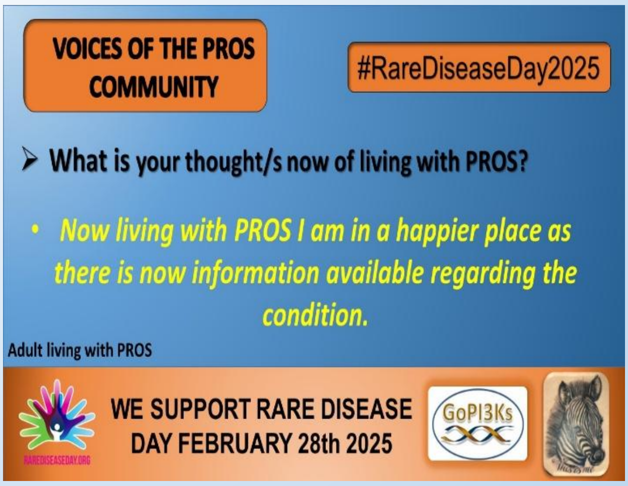

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *Now living with PROS I am in a happier place as there is now information available regarding the condition.*

Adult living with PROS

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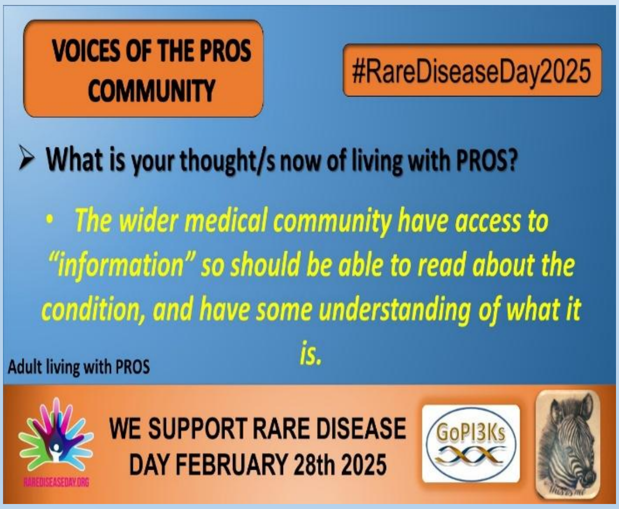

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *The wider medical community have access to "information" so should be able to read about the condition, and have some understanding of what it is.*

Adult living with PROS

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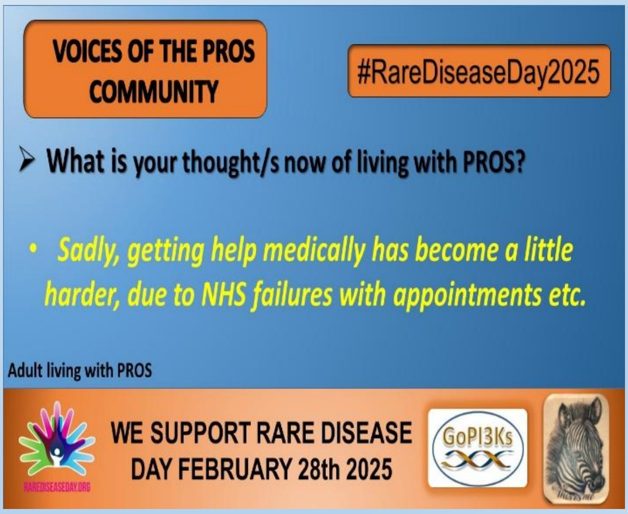

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - *Sadly, getting help medically has become a little harder, due to NHS failures with appointments etc.*

Adult living with PROS

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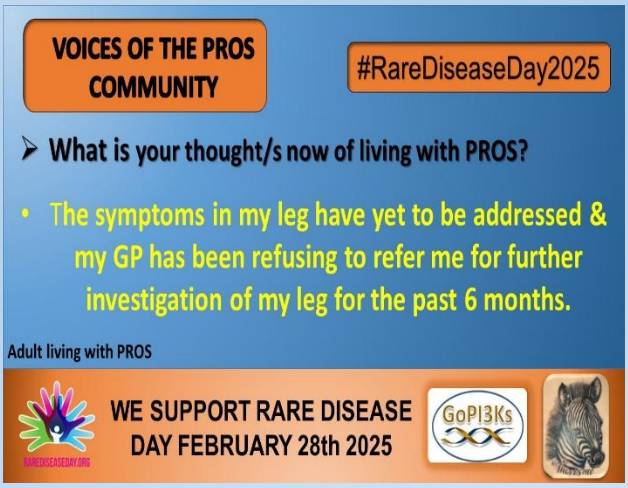

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - The symptoms in my leg have yet to be addressed & my GP has been refusing to refer me for further investigation of my leg for the past 6 months.

Adult living with PROS

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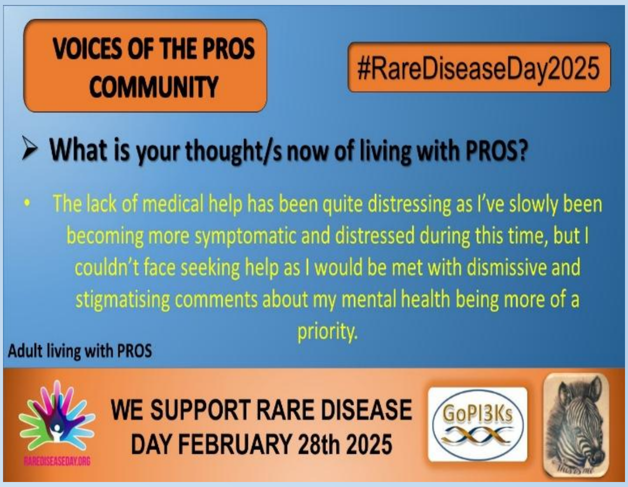

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - The lack of medical help has been quite distressing as I've slowly been becoming more symptomatic and distressed during this time, but I couldn't face seeking help as I would be met with dismissive and stigmatising comments about my mental health being more of a priority.

Adult living with PROS

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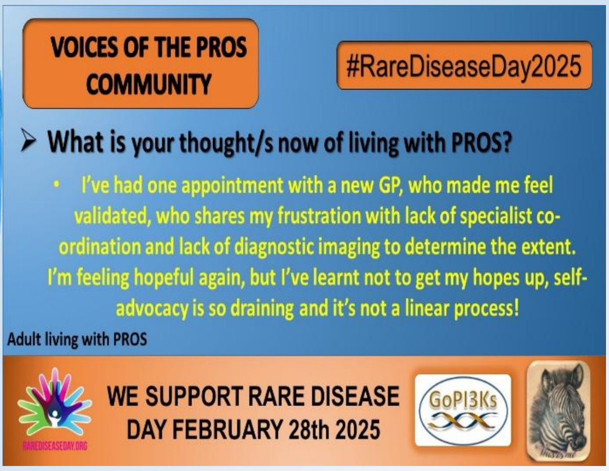

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay2025

- What is your thought/s now of living with PROS?
  - I've had one appointment with a new GP, who made me feel validated, who shares my frustration with lack of specialist co-ordination and lack of diagnostic imaging to determine the extent. I'm feeling hopeful again, but I've learnt not to get my hopes up, self-advocacy is so draining and it's not a linear process!

Adult living with PROS

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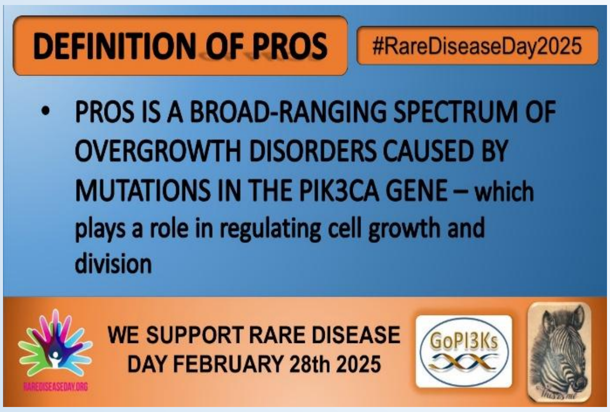
WHAT IS THE DEFINITION OF PROS?



DEFINITION OF PROS

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- PROS IS A BROAD-RANGING SPECTRUM OF OVERGROWTH DISORDERS CAUSED BY MUTATIONS IN THE PIK3CA GENE – which plays a role in regulating cell growth and division

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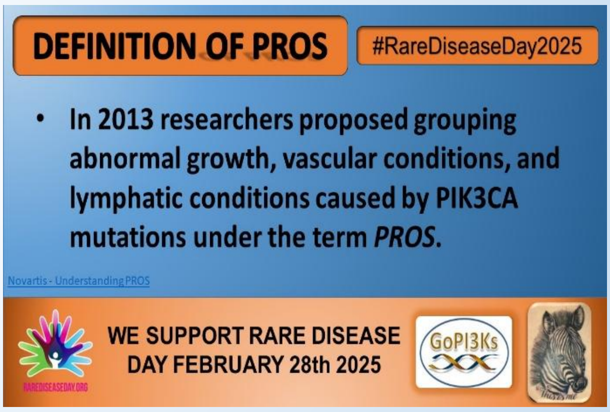

DEFINITION OF PROS

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- In 2013 researchers proposed grouping abnormal growth, vascular conditions, and lymphatic conditions caused by PIK3CA mutations under the term *PROS*.

Novartis - Understanding PROS

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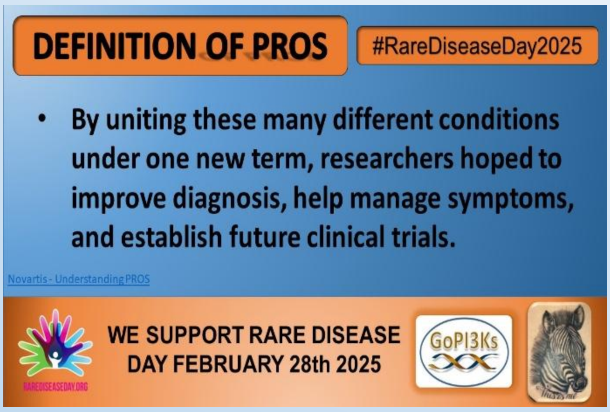

DEFINITION OF PROS

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- By uniting these many different conditions under one new term, researchers hoped to improve diagnosis, help manage symptoms, and establish future clinical trials.

Novartis - Understanding PROS

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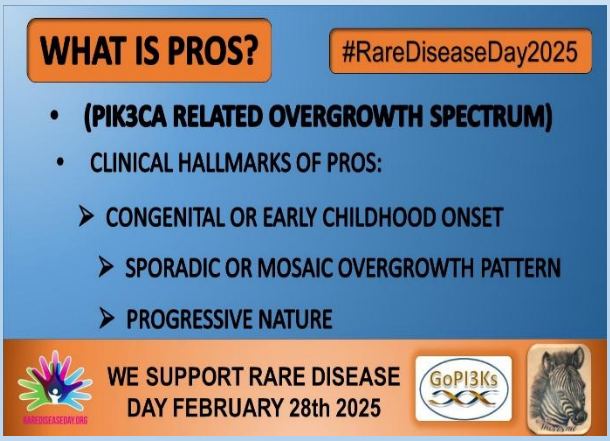
WHAT IS PROS?



WHAT IS PROS?

#RareDiseaseDay2025

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

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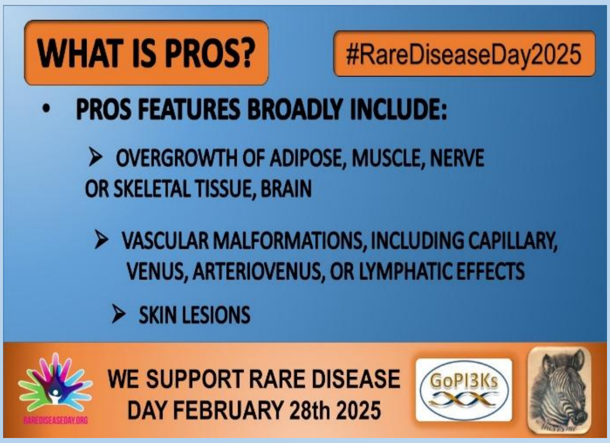

WHAT IS PROS?

#RareDiseaseDay2025

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

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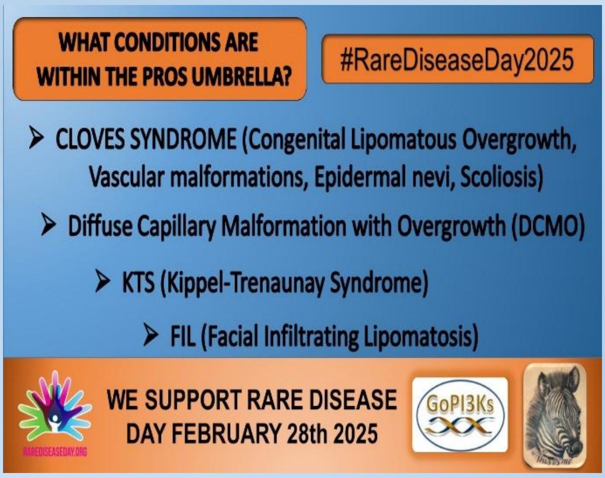
What conditions are found within PROS?



WHAT CONDITIONS ARE WITHIN THE PROS UMBRELLA?

#RareDiseaseDay2025

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

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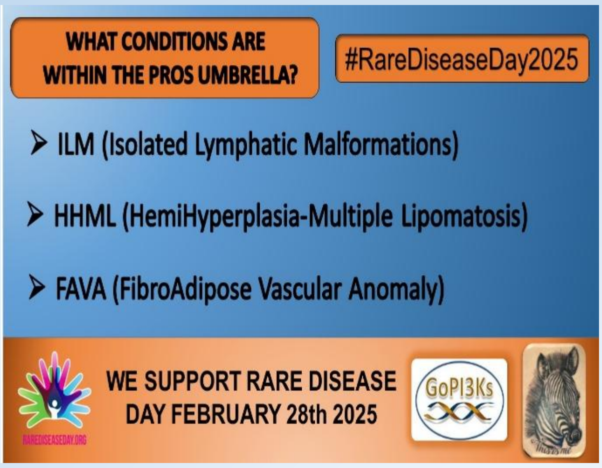

WHAT CONDITIONS ARE WITHIN THE PROS UMBRELLA?

#RareDiseaseDay2025

- ILM (Isolated Lymphatic Malformations)
- HHML (HemiHyperplasia-Multiple Lipomatosis)
- FAVA (FibroAdipose Vascular Anomaly)

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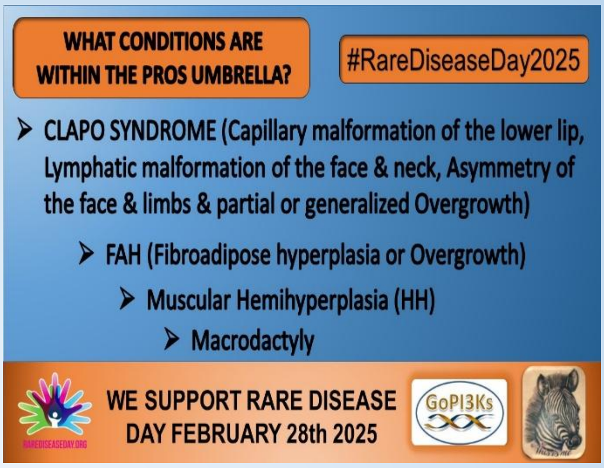

WHAT CONDITIONS ARE WITHIN THE PROS UMBRELLA?

#RareDiseaseDay2025

- CLAPO SYNDROME (Capillary malformation of the lower lip, Lymphatic malformation of the face & neck, Asymmetry of the face & limbs & partial or generalized Overgrowth)
- FAH (Fibroadipose hyperplasia or Overgrowth)
- Muscular Hemihyperplasia (HH)
- Macrodactyly

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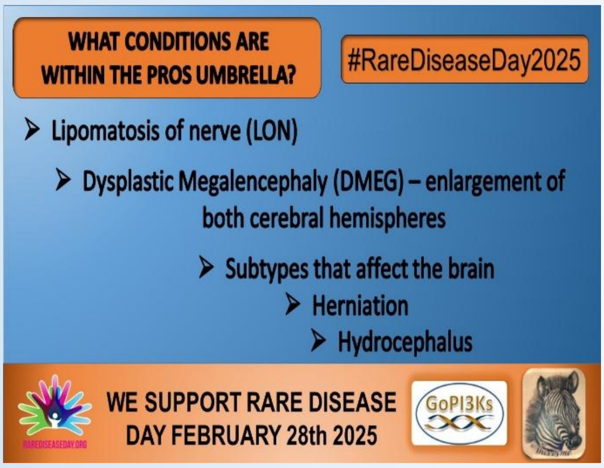

WHAT CONDITIONS ARE WITHIN THE PROS UMBRELLA?

#RareDiseaseDay2025

- Lipomatosis of nerve (LON)
  - Dysplastic Megalencephaly (DMEG) – enlargement of both cerebral hemispheres
    - Subtypes that affect the brain
      - Herniation
      - Hydrocephalus

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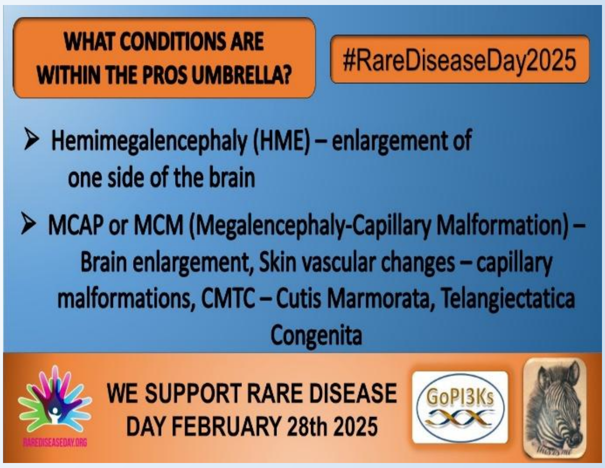

WHAT CONDITIONS ARE WITHIN THE PROS UMBRELLA?

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- Hemimegalencephaly (HME) – enlargement of one side of the brain
- MCAP or MCM (Megalencephaly-Capillary Malformation) – Brain enlargement, Skin vascular changes – capillary malformations, CMTC – Cutis Marmorata, Telangiectatica Congenita

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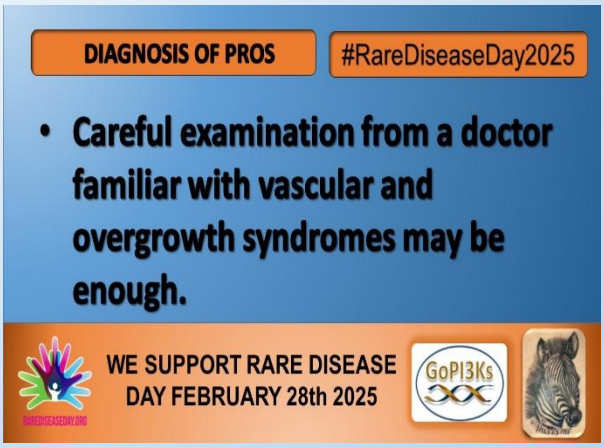
How is PROS diagnosed?



DIAGNOSIS OF PROS

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- Careful examination from a doctor familiar with vascular and overgrowth syndromes may be enough.

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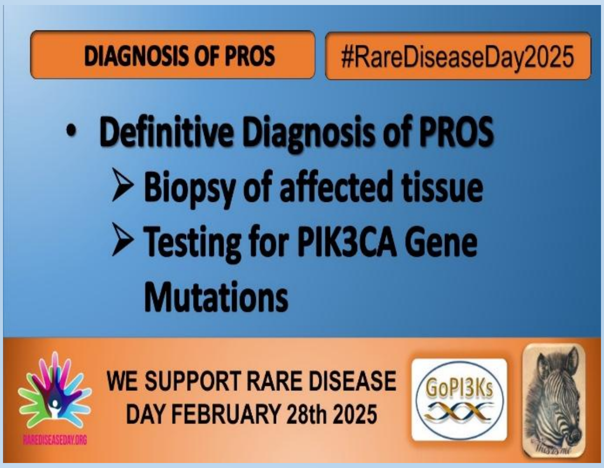

DIAGNOSIS OF PROS

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- Definitive Diagnosis of PROS
  - Biopsy of affected tissue
  - Testing for PIK3CA Gene Mutations

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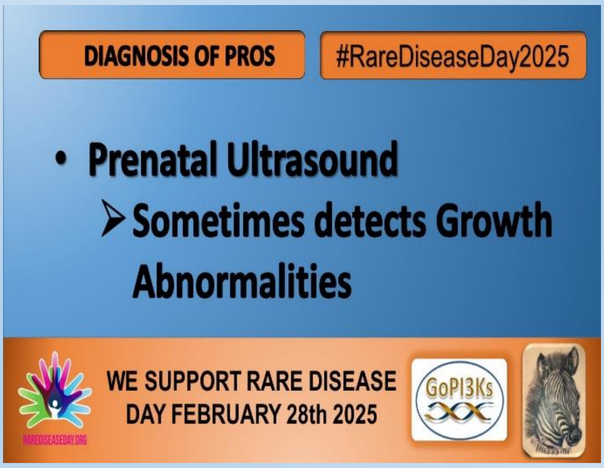

DIAGNOSIS OF PROS

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- Prenatal Ultrasound
  - Sometimes detects Growth Abnormalities

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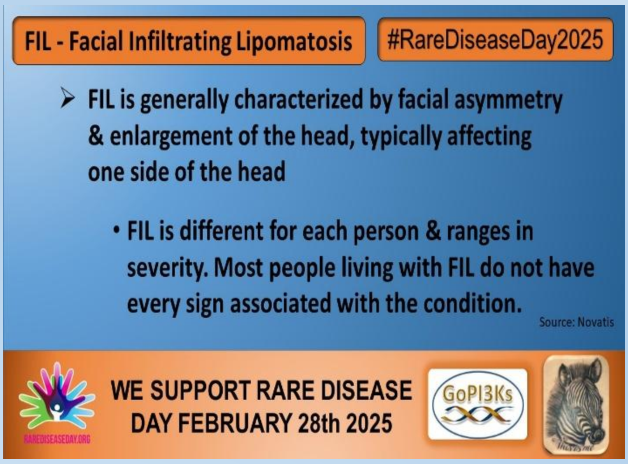
What is F.I.L? Facial Infiltrating Lipomatosis



FIL - Facial Infiltrating Lipomatosis

#RareDiseaseDay2025

- FIL is generally characterized by facial asymmetry & enlargement of the head, typically affecting one side of the head
  - FIL is different for each person & ranges in severity. Most people living with FIL do not have every sign associated with the condition.

Source: Novatis

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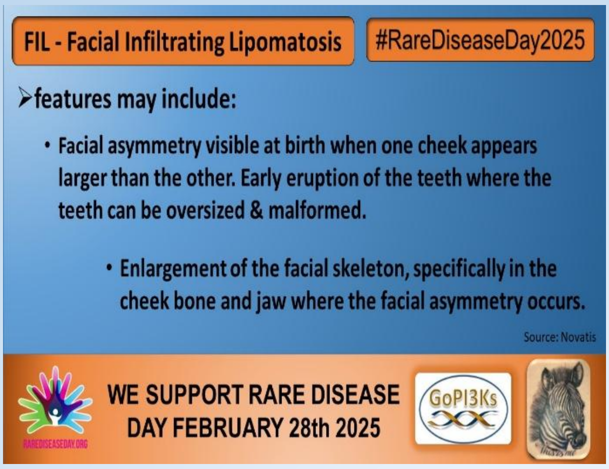

FIL - Facial Infiltrating Lipomatosis

#RareDiseaseDay2025

- features may include:
  - Facial asymmetry visible at birth when one cheek appears larger than the other. Early eruption of the teeth where the teeth can be oversized & malformed.
    - Enlargement of the facial skeleton, specifically in the cheek bone and jaw where the facial asymmetry occurs.

Source: Novatis

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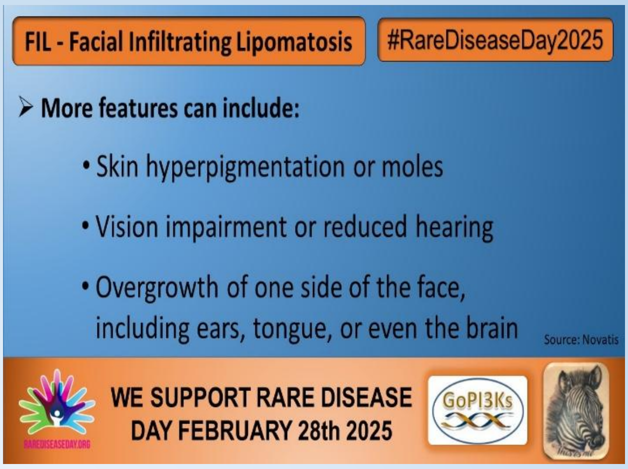

FIL - Facial Infiltrating Lipomatosis

#RareDiseaseDay2025

- More features can include:
  - Skin hyperpigmentation or moles
  - Vision impairment or reduced hearing
  - Overgrowth of one side of the face, including ears, tongue, or even the brain

Source: Novatis

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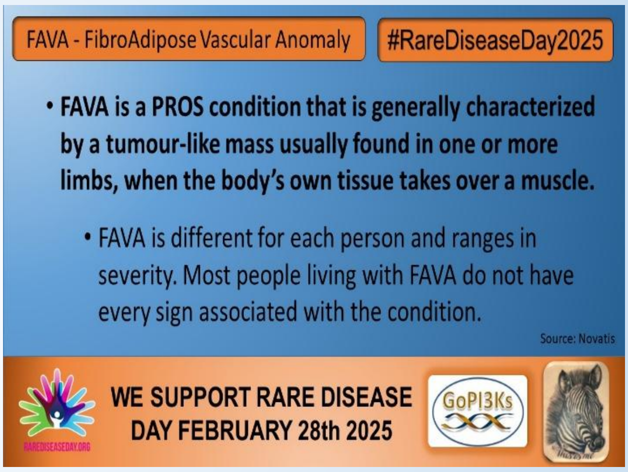
What is FAVA? FibroAdipose Vascular Anomaly



FAVA - FibroAdipose Vascular Anomaly

#RareDiseaseDay2025

- FAVA is a PROS condition that is generally characterized by a tumour-like mass usually found in one or more limbs, when the body's own tissue takes over a muscle.
  - FAVA is different for each person and ranges in severity. Most people living with FAVA do not have every sign associated with the condition.

Source: Novatis

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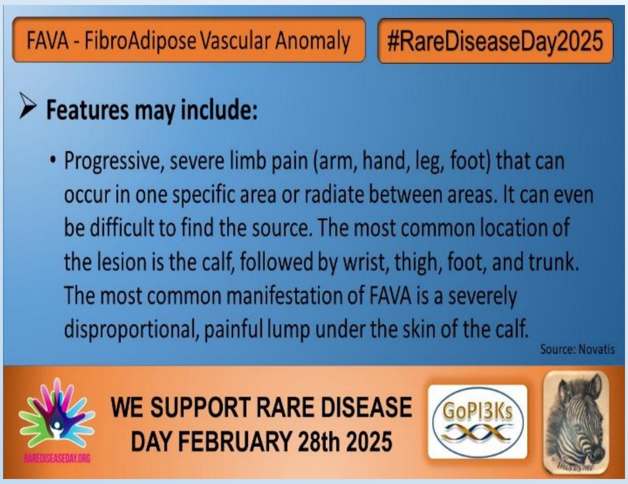

FAVA - FibroAdipose Vascular Anomaly

#RareDiseaseDay2025

- Features may include:
  - Progressive, severe limb pain (arm, hand, leg, foot) that can occur in one specific area or radiate between areas. It can even be difficult to find the source. The most common location of the lesion is the calf, followed by wrist, thigh, foot, and trunk. The most common manifestation of FAVA is a severely disproportional, painful lump under the skin of the calf.

Source: Novatis

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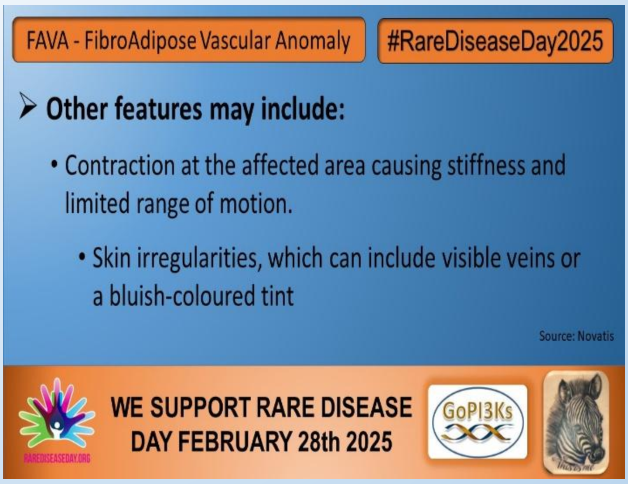

FAVA - FibroAdipose Vascular Anomaly

#RareDiseaseDay2025

- Other features may include:
  - Contraction at the affected area causing stiffness and limited range of motion.
    - Skin irregularities, which can include visible veins or a bluish-coloured tint

Source: Novatis

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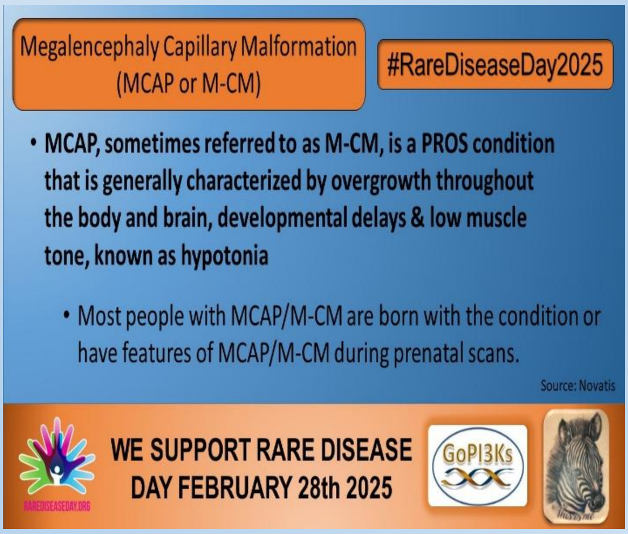
WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

GoPI3Ks

What is MCAP Or M-CM? Megalencephaly Capillary Malformation



Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2025

- MCAP, sometimes referred to as M-CM, is a PROS condition that is generally characterized by overgrowth throughout the body and brain, developmental delays & low muscle tone, known as hypotonia
  - Most people with MCAP/M-CM are born with the condition or have features of MCAP/M-CM during prenatal scans.

Source: Novatis

RAREDISEASEDAY.ORG

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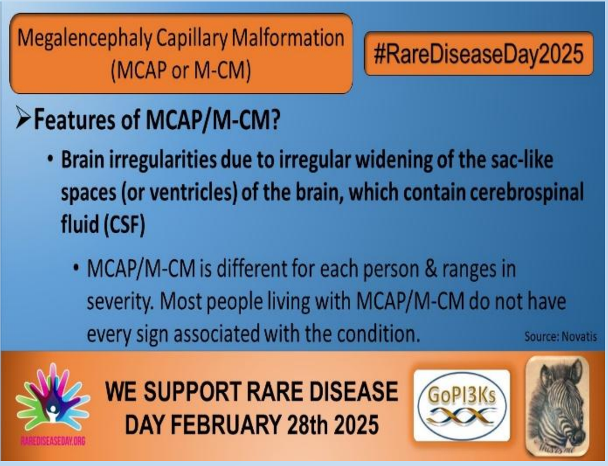

Megalencephaly Capillary Malformation (MCAP or M-CM)

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➢**Features of MCAP/M-CM?**

- **Brain irregularities due to irregular widening of the sac-like spaces (or ventricles) of the brain, which contain cerebrospinal fluid (CSF)**
  - MCAP/M-CM is different for each person & ranges in severity. Most people living with MCAP/M-CM do not have every sign associated with the condition.

Source: Novatis

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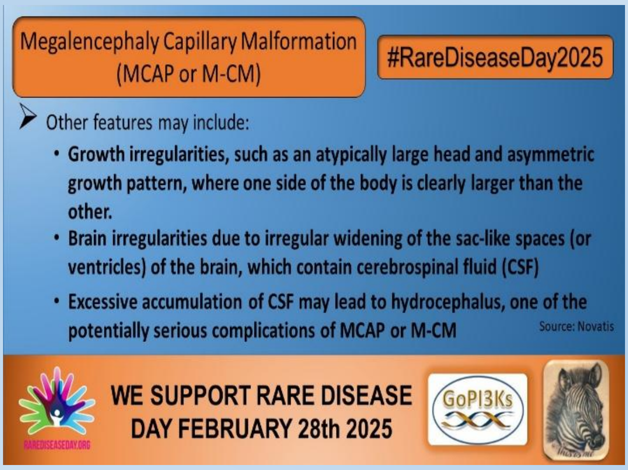

Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2025

➢ Other features may include:

- **Growth irregularities, such as an atypically large head and asymmetric growth pattern, where one side of the body is clearly larger than the other.**
- **Brain irregularities due to irregular widening of the sac-like spaces (or ventricles) of the brain, which contain cerebrospinal fluid (CSF)**
- **Excessive accumulation of CSF may lead to hydrocephalus, one of the potentially serious complications of MCAP or M-CM**

Source: Novatis

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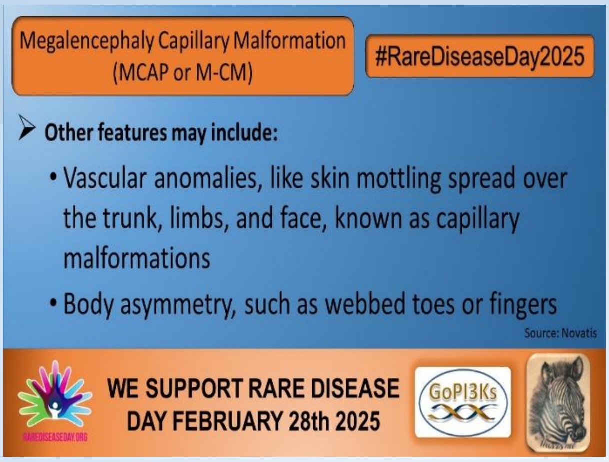

Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2025

➢ **Other features may include:**

- Vascular anomalies, like skin mottling spread over the trunk, limbs, and face, known as capillary malformations
- Body asymmetry, such as webbed toes or fingers

Source: Novatis

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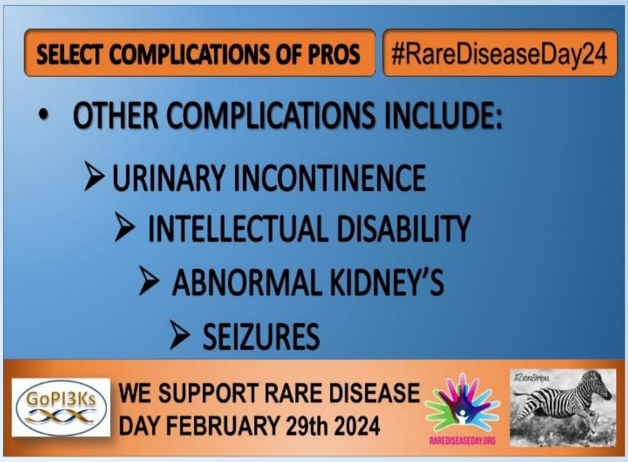

SELECT COMPLICATIONS OF PROS

#RareDiseaseDay24

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

GoPI3Ks

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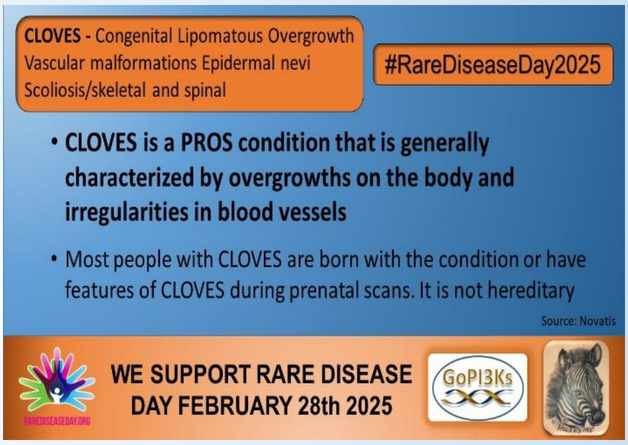
#ZebraStripes

What is CLOVES?

Congenital, Lipomatous, Overgrowth, Vascular Malformations Epidermal nevi Scoliosis/Skeletal & spinal



CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2025

- **CLOVES is a PROS condition that is generally characterized by overgrowths on the body and irregularities in blood vessels**
- Most people with CLOVES are born with the condition or have features of CLOVES during prenatal scans. It is not hereditary

Source: Novatis

RAREDISEASEDAY.ORG

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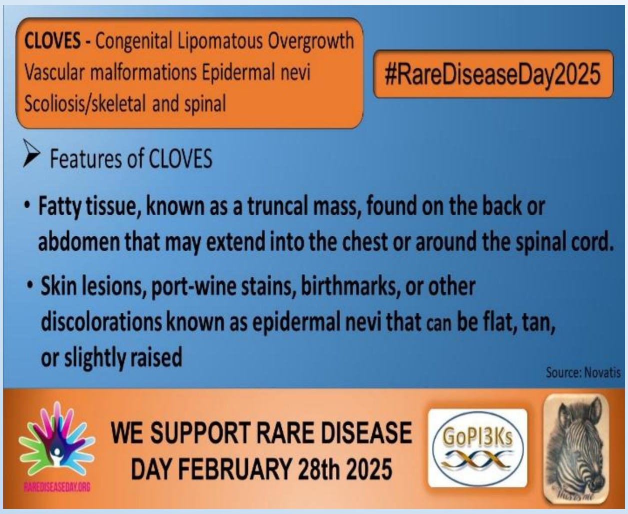

CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2025

➢ Features of CLOVES

- **Fatty tissue, known as a truncal mass, found on the back or abdomen that may extend into the chest or around the spinal cord.**
- **Skin lesions, port-wine stains, birthmarks, or other discolorations known as epidermal nevi that can be flat, tan, or slightly raised**

Source: Novatis

RAREDISEASEDAY.ORG

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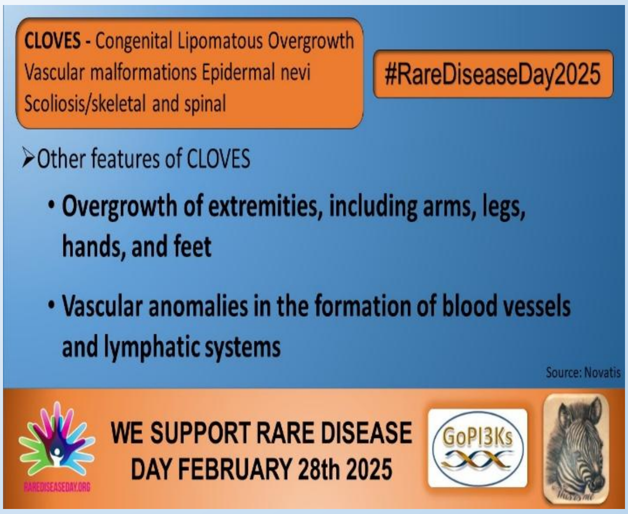

CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2025

➢Other features of CLOVES

- **Overgrowth of extremities, including arms, legs, hands, and feet**
- **Vascular anomalies in the formation of blood vessels and lymphatic systems**

Source: Novatis

RAREDISEASEDAY.ORG

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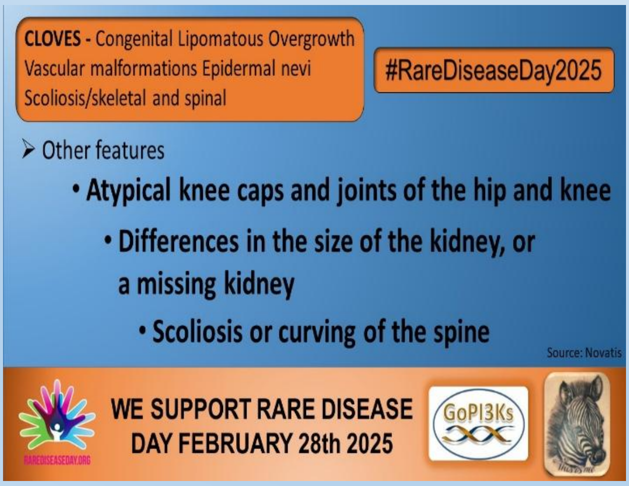

CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2025

➢ Other features

- **Atypical knee caps and joints of the hip and knee**
  - **Differences in the size of the kidney, or a missing kidney**
    - **Scoliosis or curving of the spine**

Source: Novatis

RAREDISEASEDAY.ORG

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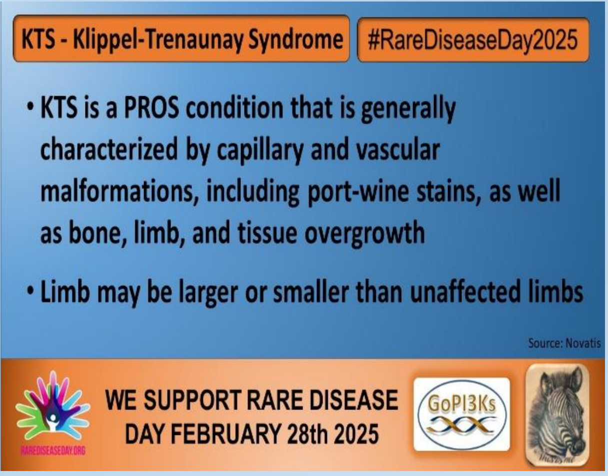
GoPI3Ks

What is KTS?

Klippel-Trenaunay Syndrome



KTS - Klippel-Trenaunay Syndrome

#RareDiseaseDay2025

- KTS is a PROS condition that is generally characterized by capillary and vascular malformations, including port-wine stains, as well as bone, limb, and tissue overgrowth
- Limb may be larger or smaller than unaffected limbs

Source: Novatis

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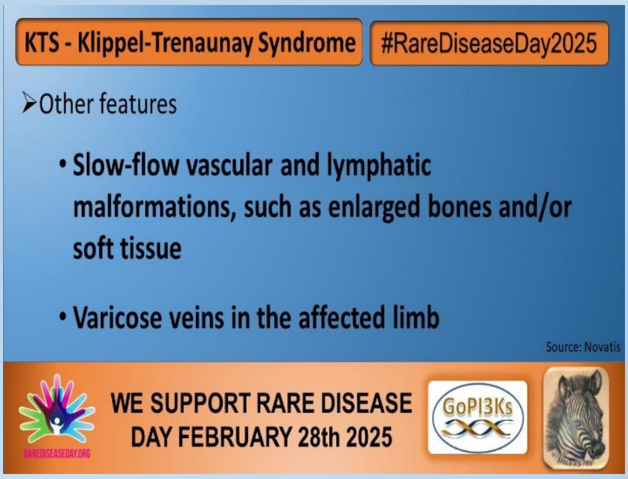

KTS - Klippel-Trenaunay Syndrome

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➢Other features

- Slow-flow vascular and lymphatic malformations, such as enlarged bones and/or soft tissue
- Varicose veins in the affected limb

Source: Novatis

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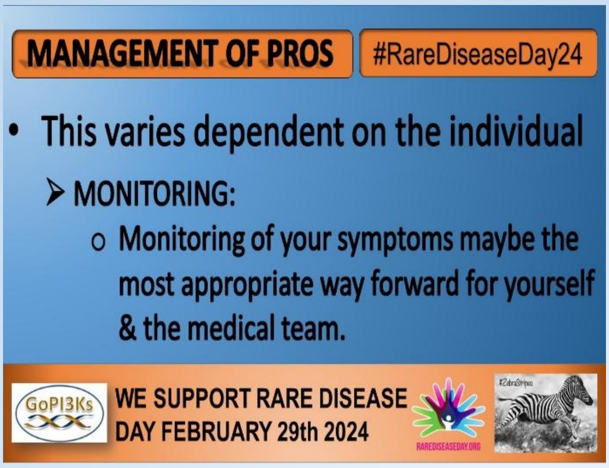

MANAGEMENT OF PROS

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

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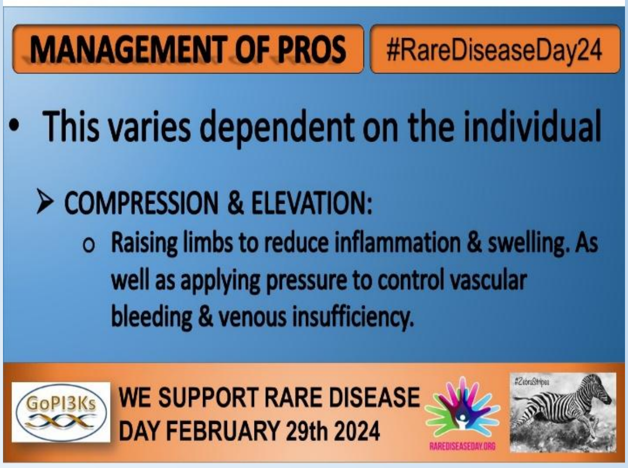

MANAGEMENT OF PROS

#RareDiseaseDay24

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

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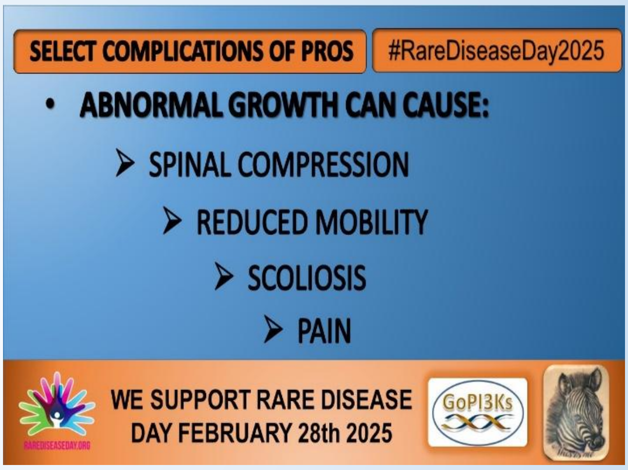
What are possible complications of PROS



SELECT COMPLICATIONS OF PROS

#RareDiseaseDay2025

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

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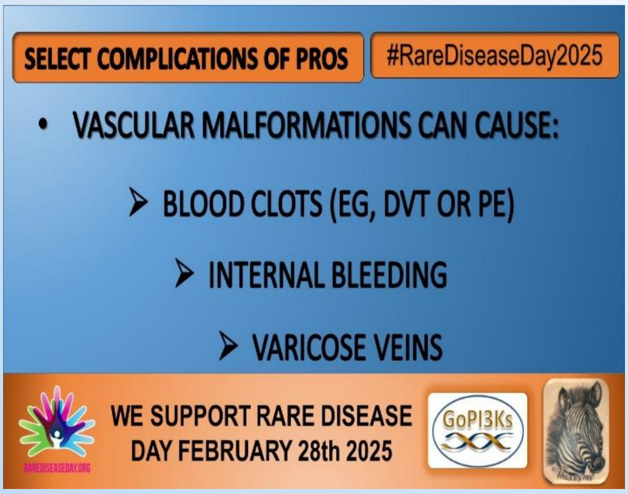

SELECT COMPLICATIONS OF PROS

#RareDiseaseDay2025

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

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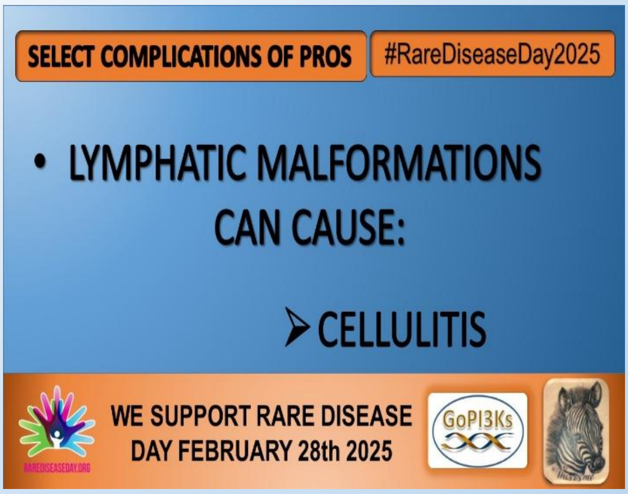

SELECT COMPLICATIONS OF PROS

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- LYMPHATIC MALFORMATIONS CAN CAUSE:
  - ➢CELLULITIS

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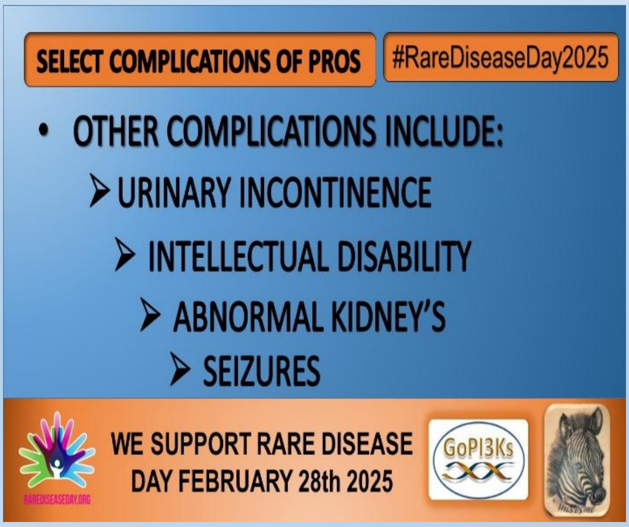

SELECT COMPLICATIONS OF PROS

#RareDiseaseDay2025

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

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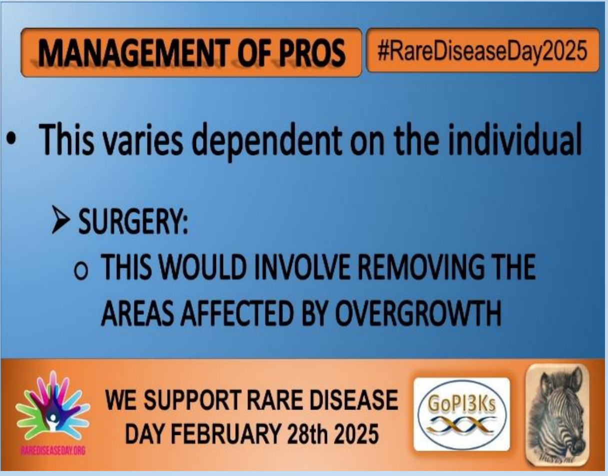
How is PROS managed?



MANAGEMENT OF PROS

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- This varies dependent on the individual
  - SURGERY:
    - THIS WOULD INVOLVE REMOVING THE AREAS AFFECTED BY OVERGROWTH

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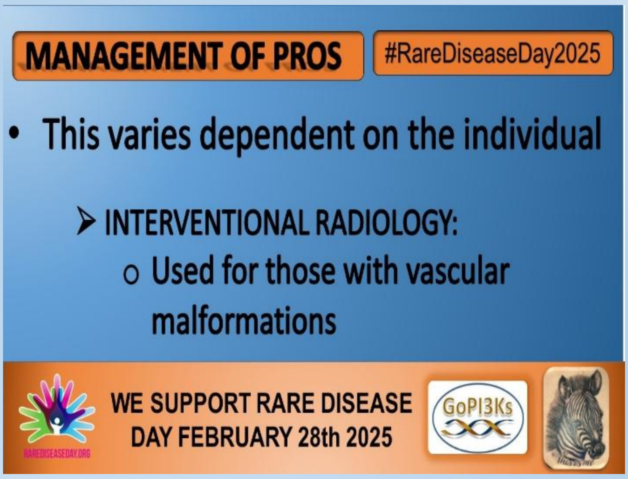

MANAGEMENT OF PROS

#RareDiseaseDay2025

- This varies dependent on the individual
  - INTERVENTIONAL RADIOLOGY:
    - Used for those with vascular malformations

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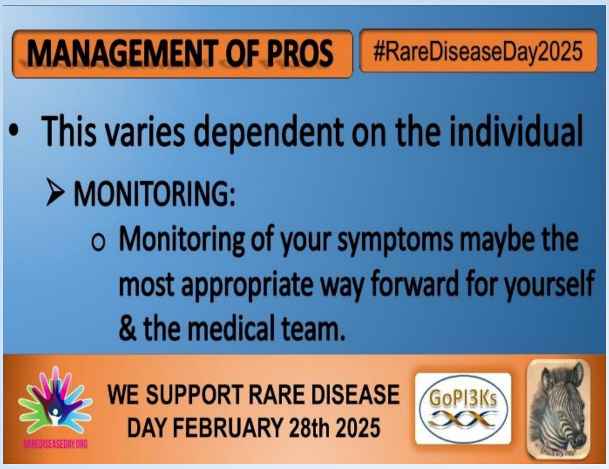

MANAGEMENT OF PROS

#RareDiseaseDay2025

- 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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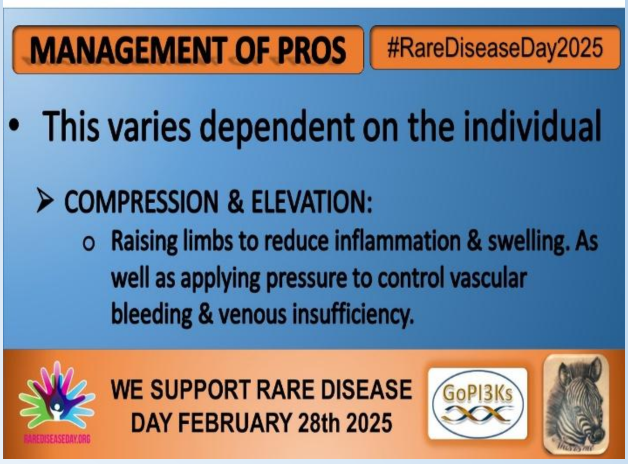

MANAGEMENT OF PROS

#RareDiseaseDay2025

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

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Is there a treatment for PROS?

IS THERE A TREATMENT FOR PROS?

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- A multi-disciplinary evaluation & monitoring by experts familiar with PROS.
- "Although for many of us, this is very difficult to find" – adult with PROS

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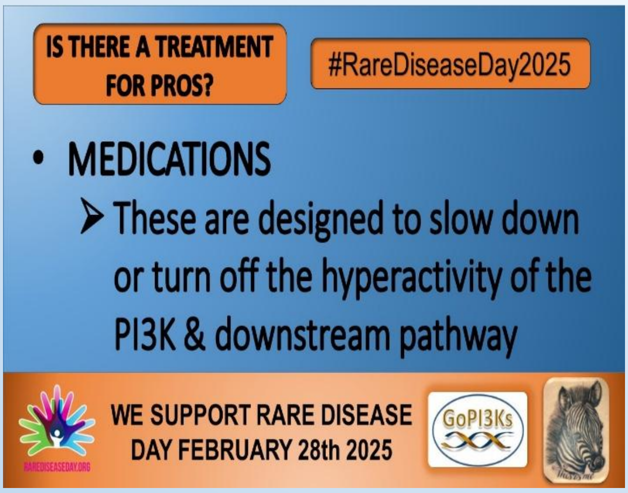

IS THERE A TREATMENT FOR PROS?

#RareDiseaseDay2025

- MEDICATIONS
  - These are designed to slow down or turn off the hyperactivity of the PI3K & downstream pathway

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IS THERE A TREATMENT FOR PROS?

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- **SUPERFICIAL VASCULAR MALFORMATIONS**
  - Laser Ablation – lighten colour, stop leaking & bleeding.
    - Sclerotherapy & Embolization
- **INDIVIDUALS WITH MOTOR &/OR INTELLECTUAL PROBLEMS:**
  - Physical, Occupational & Speech therapies

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IS THERE A TREATMENT FOR PROS?

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- HYDROCEPHALUS:
  - **Placing a shunt to drain excess CSF**
- BRAIN STEM HERNIATION:
  - **Decompression Surgery**

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IS THERE A TREATMENT FOR PROS?

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- SIMPLE LOCALISED LESIONS:
  - **Surgical Excision**
- LARGER LESIONS:
  - **De-bulking to be repeated if & when needed**

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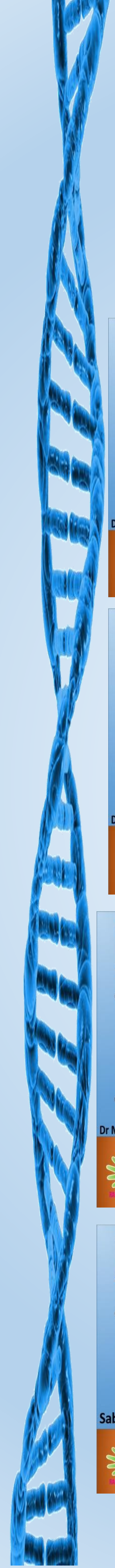
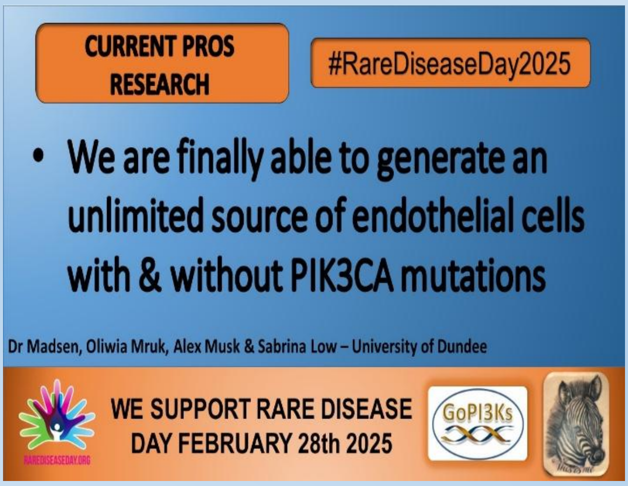


Current research – Madsen lab, Dundee



CURRENT PROS RESEARCH

#RareDiseaseDay2025

- We are finally able to generate an unlimited source of endothelial cells with & without PIK3CA mutations

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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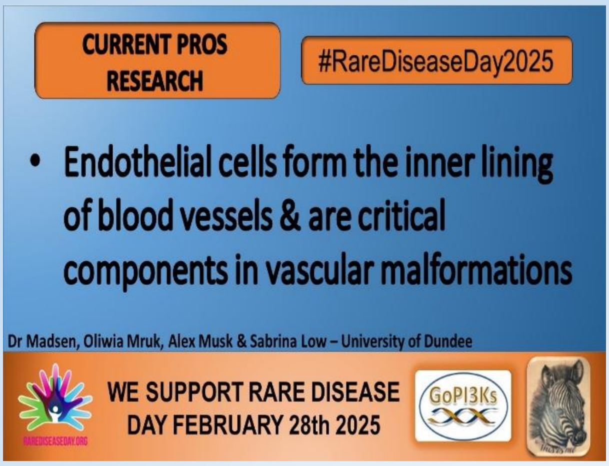

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Endothelial cells form the inner lining of blood vessels & are critical components in vascular malformations

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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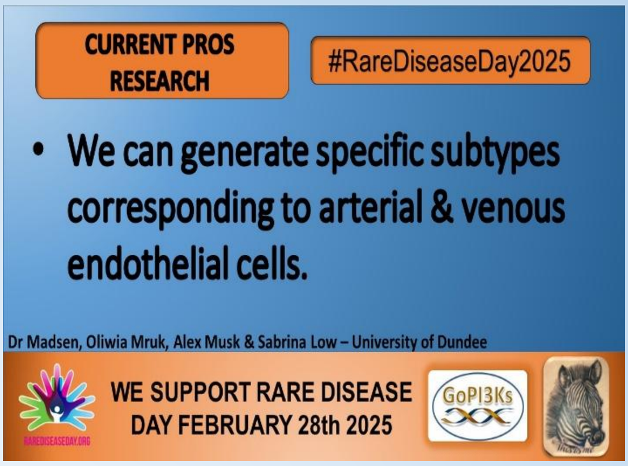

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- We can generate specific subtypes corresponding to arterial & venous endothelial cells.

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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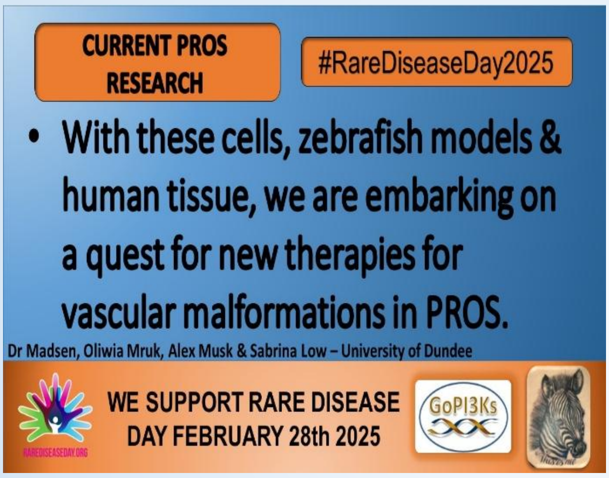

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- With these cells, zebrafish models & human tissue, we are embarking on a quest for new therapies for vascular malformations in PROS.

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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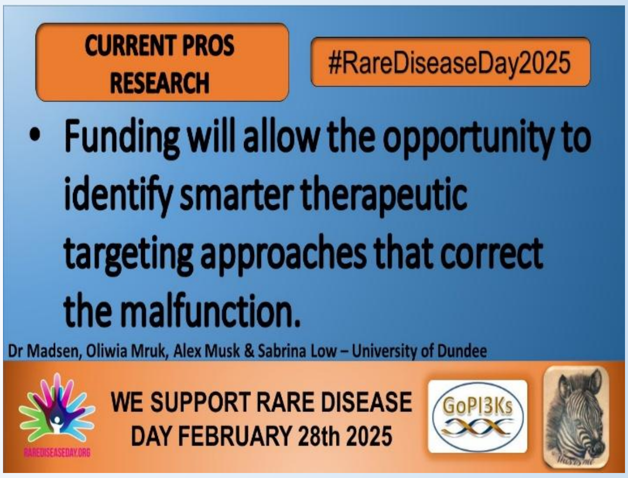

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Funding will allow the opportunity to identify smarter therapeutic targeting approaches that correct the malfunction.

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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CURRENT PROS RESEARCH

#RareDiseaseDay2025

- These are likely to be effective long term & cause minimal side-effects.
- Bare in mind research takes a long time.

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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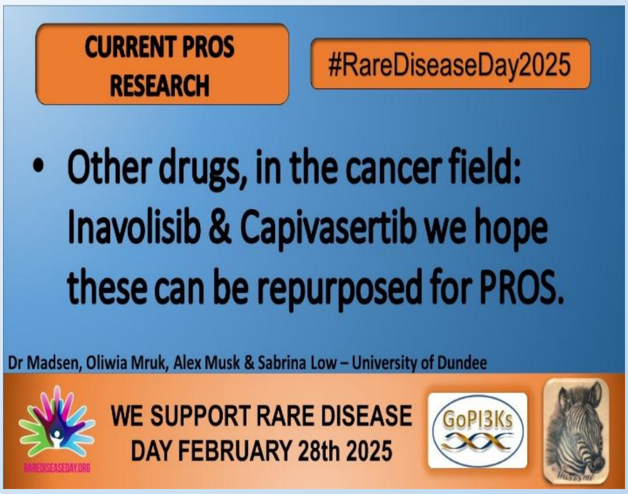

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Other drugs, in the cancer field: Inavolisib & Capivasertib we hope these can be repurposed for PROS.

Dr Madsen, Oliwia Mruk, Alex Musk & Sabrina Low – University of Dundee

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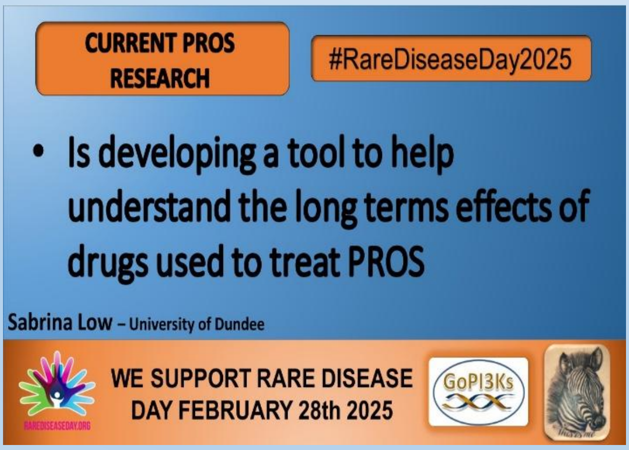

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Is developing a tool to help understand the long terms effects of drugs used to treat PROS

Sabrina Low – University of Dundee

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CURRENT PROS RESEARCH

#RareDiseaseDay2025

- This can help predict what may happen to patients who take these drugs long term & help scientists create safer & more effective treatments.

Sabrina Low – University of Dundee

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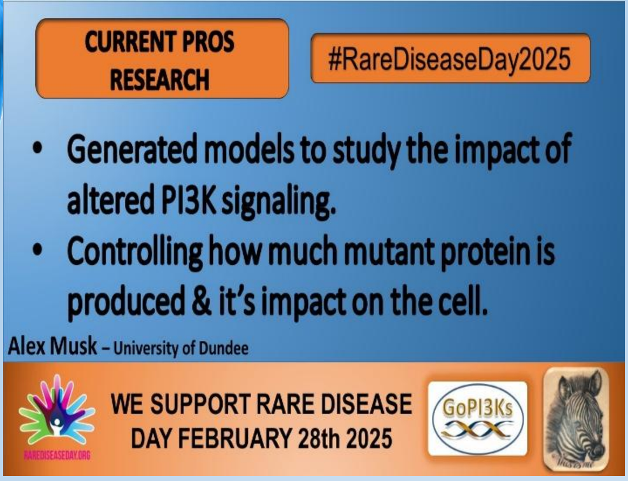

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Generated models to study the impact of altered PI3K signaling.
- Controlling how much mutant protein is produced & it's impact on the cell.

Alex Musk – University of Dundee

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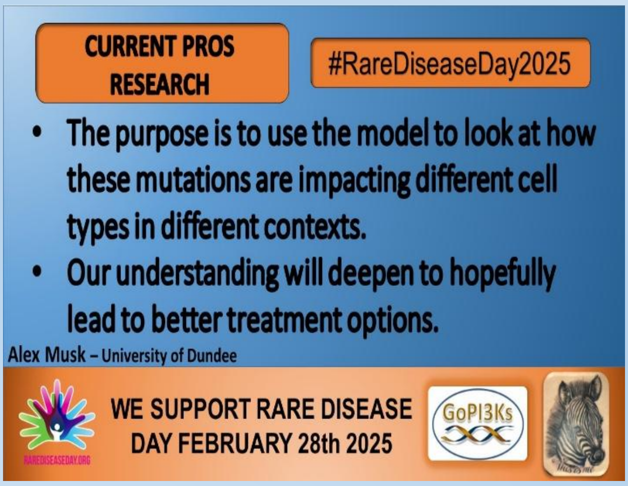

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- The purpose is to use the model to look at how these mutations are impacting different cell types in different contexts.
- Our understanding will deepen to hopefully lead to better treatment options.

Alex Musk – University of Dundee

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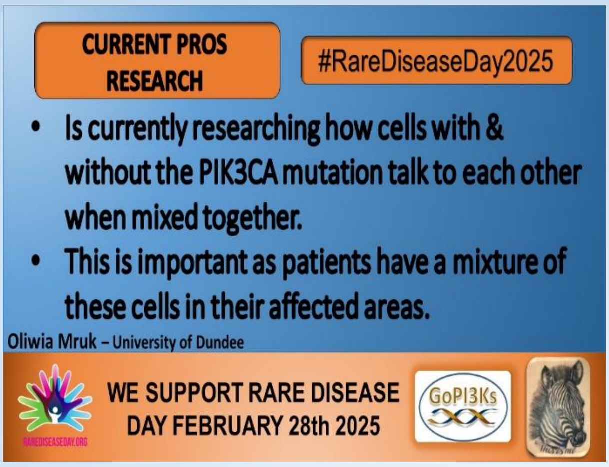

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Is currently researching how cells with & without the PIK3CA mutation talk to each other when mixed together.
- This is important as patients have a mixture of these cells in their affected areas.

Oliwia Mruk – University of Dundee

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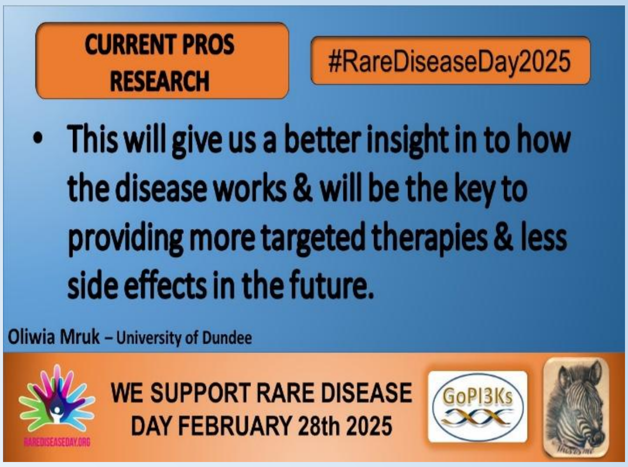

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- This will give us a better insight in to how the disease works & will be the key to providing more targeted therapies & less side effects in the future.

Oliwia Mruk – University of Dundee

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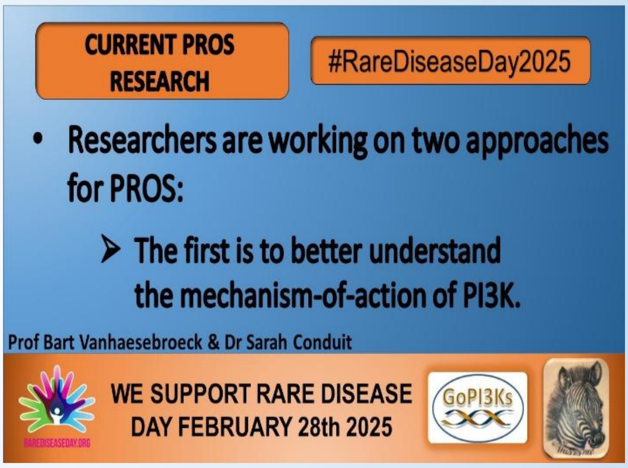
Professor Bart Vanhaesebroeck & Dr Sarah Conduit



CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Researchers are working on two approaches for PROS:
  - ➢ The first is to better understand the mechanism-of-action of PI3K.

Prof Bart Vanhaesebroeck & Dr Sarah Conduit

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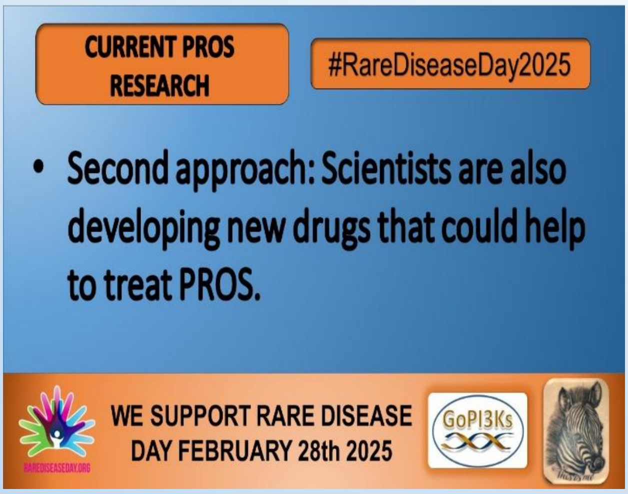

CURRENT PROS RESEARCH

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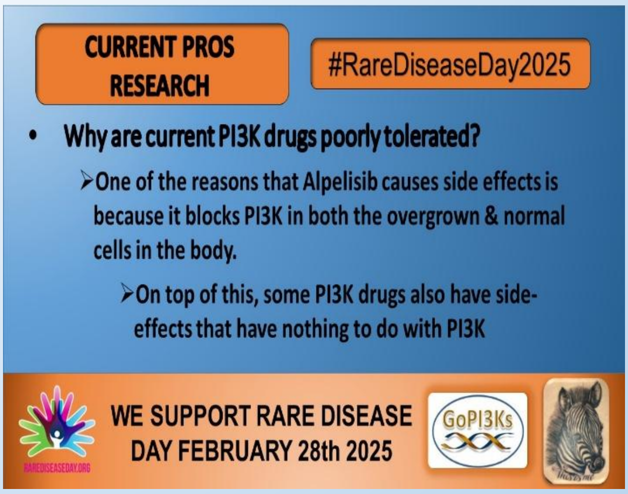
- Second approach: Scientists are also developing new drugs that could help to treat PROS.

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

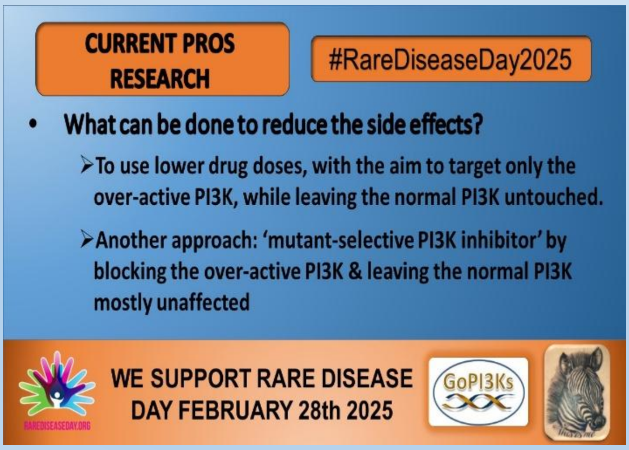


CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Why are current PI3K drugs poorly tolerated?
  - ➢ One of the reasons that Alpelisib causes side effects is because it blocks PI3K in both the overgrown & normal cells in the body.
    - ➢ On top of this, some PI3K drugs also have side-effects that have nothing to do with PI3K

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025



CURRENT PROS RESEARCH

#RareDiseaseDay2025

- What can be done to reduce the side effects?
  - ➢ To use lower drug doses, with the aim to target only the over-active PI3K, while leaving the normal PI3K untouched.
  - ➢ Another approach: 'mutant-selective PI3K inhibitor' by blocking the over-active PI3K & leaving the normal PI3K mostly unaffected

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

CURRENT PROS RESEARCH

#RareDiseaseDay2025

- Outlook:
  - ➢ We are hopeful that basic research to better understand PROS & together with the development of better tolerated PI3K targeting drugs will provide new treatment options for PROS in the coming years.

WE SUPPORT RARE DISEASE DAY FEBRUARY 28th 2025

IS THERE A TREATMENT FOR PROS?

#RareDiseaseDay24

- **HYDROCEPHALUS:**
  - ➢ Placing a shunt to drain excess CSF

GoPI3Ks

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RAREDISEASEDAY.ORG

IS THERE A TREATMENT FOR PROS?

#RareDiseaseDay24

- **BRAIN STEM HERNIATION:**
  - ➢ Decompression Surgery

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RAREDISEASEDAY.ORG

IS THERE A TREATMENT FOR PROS?

#RareDiseaseDay24

- **INDIVIDUALS WITH MOTOR &/OR INTELLECTUAL PROBLEMS:**
  - ➢ Physical, Occupational & Speech therapies

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RAREDISEASEDAY.ORG

IMPROVING PSYCHOSOCIAL SUPPORT FOR INDIVIDUALS WITH PIK3CA-RELATED OVERGROWTH SPECTRUM (PROS)

At the beginning of this year we received a grant to look into how PROS affects all aspects of people’s lives, such as the psychological aspect, working life, social life, medical & more. Here we included those who live with PROS, adults and young adults as well as parents and carers.

OUR AIM: To improve the provision of psychosocial support for individuals with PROS by assessing the impact of the condition on quality of life and psychosocial wellbeing and co-producing psychoeducational information.

Working alongside researcher Anna Zarola and Dr Ella Guest from UWE (University of West England) the project involved: **two elements:**

- Firstly a survey was designed with the help of those living with PROS & parents who’s child has PROS. This was then distributed throughout the PROS community.

- This allowed us to assess the impact PROS has on the quality of life, mental health, psychosocial wellbeing & what support needs there are.

- The second phase involved developing psychoeducational resources together with a group from the PROS community. Our hope is that these resources will help people now & in the future, including: patients, families, doctors & counsellors etc.

PROS, DANCING & ME – MEG BODOANO

Representing Great Britain

Hi! My name is Bonita Bodoano and I have PIK-3CA. It affects mostly my back (scoliosis) and legs (height and width discrepancies.) I was diagnosed at 5 years old and was told I would never dance or do sport again. Now at 17, I have danced for over 15 years of my life. I recently joined Sun-Rae dance school which specialises in inclusive dance and I was lucky to represent Great Britain in the World Para Dance Championships. It was an amazing experience and opportunity to meet so many wonderful dancers from across the world. It was a privilege to receive a Gold medal for the ambulant woman's solo category. It really emphasised that having a medical condition does not limit you from achieving whatever you put your mind to. If you want to follow my journey you can follow me on Instagram @bonitabod

Winning 1st prize

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.

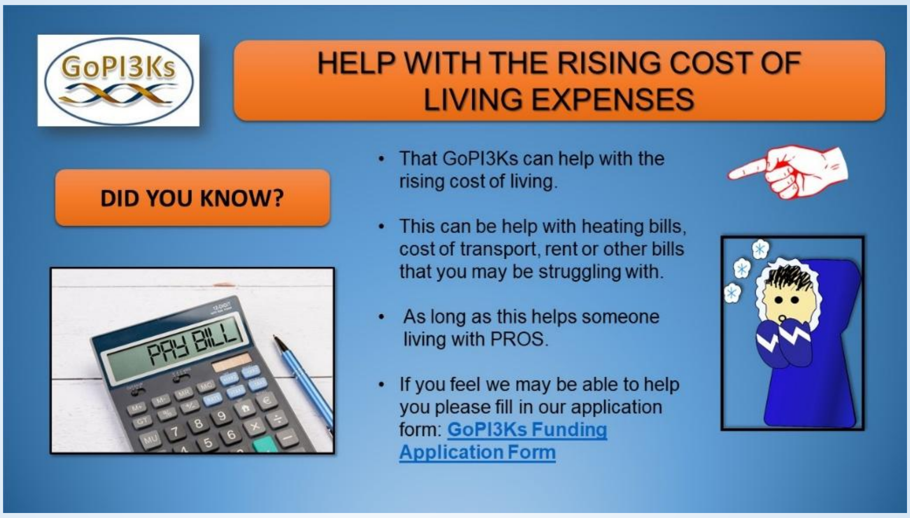
GoPI3Ks Grant Application Form

HELP WITH THOSE EXTRA COSTS LIVING WITH PROS BRINGS

DID YOU KNOW?

- At GoPI3Ks we understanding that living with PROS can bring added costs, we are here to help:
- This can be helping to towards:
  - Wheelchairs
  - Bespoke shoes or clothing
  - Travel & accommodation expenses to attend hospital appointments.
  - And much more

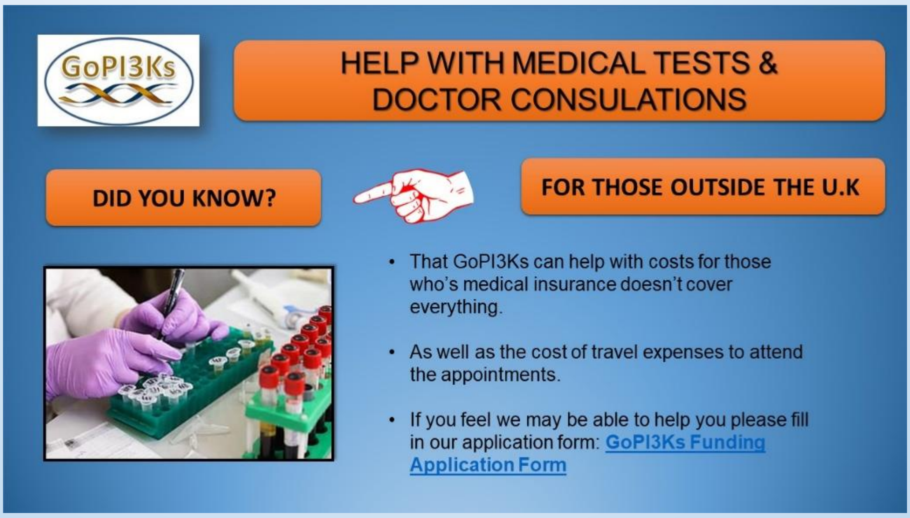
If you feel we may be able to help you, please fill in our application form: GoPI3Ks Funding Application Form



HELP WITH THE RISING COST OF LIVING EXPENSES

DID YOU KNOW?

- That GoPI3Ks can help with the rising cost of living.
- This can be help with heating bills, cost of transport, rent or other bills that you may be struggling with.
- As long as this helps someone living with PROS.
- If you feel we may be able to help you please fill in our application form: GoPI3Ks Funding Application Form

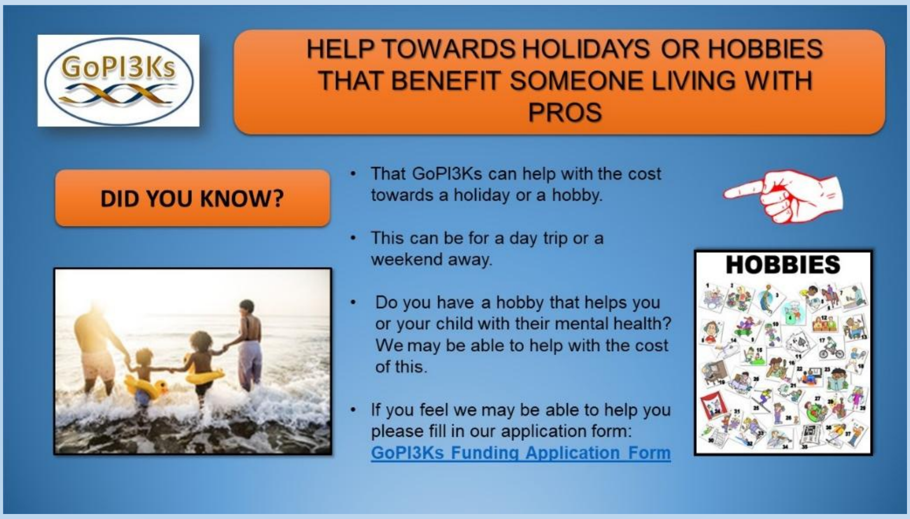


HELP WITH MEDICAL TESTS & DOCTOR CONSULATIONS

DID YOU KNOW?

FOR THOSE OUTSIDE THE U.K

- That GoPI3Ks can help with costs for those who's medical insurance doesn't cover everything.
- As well as the cost of travel expenses to attend the appointments.
- If you feel we may be able to help you please fill in our application form: GoPI3Ks Funding Application Form



HELP TOWARDS HOLIDAYS OR HOBBIES THAT BENEFIT SOMEONE LIVING WITH PROS

DID YOU KNOW?

- That GoPI3Ks can help with the cost towards a holiday or a hobby.
- This can be for a day trip or a weekend away.
- Do you have a hobby that helps you or your child with their mental health? We may be able to help with the cost of this.
- If you feel we may be able to help you please fill in our application form: GoPI3Ks Funding Application Form

HOW YOU CAN HELP FUNDRAISE FOR GOPI3KS

easyfundraising

https://www.easyfundraising.org.uk/causes/gopi3ks-genetic-overgrowth-pi3k-support

HOW IT WORKS:

Easyfundraising partners with over 7,000 brands who will donate part of what you spend to a cause of your choice. It won't cost you any extra. The cost is covered by the brand.

It is easy to use, just sign up & choose GoPI3Ks & add the app to your mobile & extension to your laptop & it will automatically enable a donation when you go to your shopping site.

To learn more about how you can help GoPI3Ks, check out this video from their website:

Video of how easy it is to raise money for GoPI3Ks

CHARITABLE GIVING / PAYROLL GIVING

CharitableGiving
we make giving easy

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

HOW YOU CAN HELP FUNDRAISE FOR GOPI3KS CONT:

PAYPAL GIVING

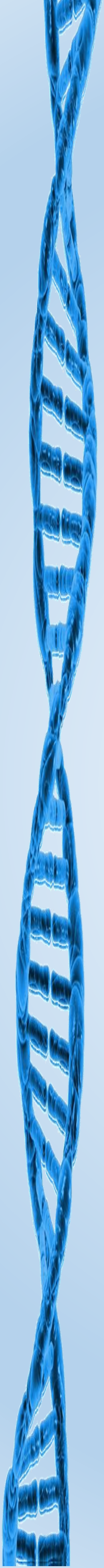
How can I support GoPI3Ks using PayPal
Please click this link:
https://www.paypal.com/gb/fundraiser/hub

PayPal
Giving Fund

GIVE AS YOU LIVE

Every time you shop online you could generate FREE donations for us! It takes 2 mins to sign up to @GiveasyouLive and their partner stores will donate a percentage of your spend back to us, at no cost to you. Sign up with the link below:

Give As You Live



HOW YOU CAN HELP FUNDRAISE FOR GOPI3KS CONT:

SOCIAL MEDIA FUNDRAISERS

Social media is an easy way to fundraise & many platforms offer this with a click of a button. If you are celebrating your birthday, why not choose GoPI3Ks as your nominated charity or even if you just want to help at any time, a fundraiser can be easily set up. Facebook & Instagram are the easiest. X you can post links to encourage followers to donate:

SPONSORED FUNDRAISERS

Are you thinking of challenging yourself in 2025?

Maybe a half marathon or even a full one – why not raise money for us whilst you run.

Maybe baking is your thing? Why not gather family & friends together or maybe co-workers fancy a yummy cake & raise money at the same time.

QUICK LINKS TO INFORMATION:

We know it can be a minefield at times trying to find out information about PROS, so here are a few links to pages on our website that you may find helpful.

WHAT IS PROS?

- PIK3CA-related overgrowth spectrum (NORD)
- What is PROS by Dr Sanjay & Dr Garg
- PROS for doctors by Novartis
- Information on Alpelisib / Vijoice

EXPLAINING PROS TO CHILDREN

- Understanding PROS
- PROS at home
- PROS at school
- PROS at the doctors
- Children's resources

WHAT RESEARCH HAS THERE BEEN IN TO PROS

- Research in to PROS

PSYCHOLOGICAL & WELL-BEING SUPPORT

- Mental Health support for adults
- Mental Health support for children
- Amputation – scroll to the bottom of the page
- Clothing, shoe & sock companies– at the bottom of the page

RARE DISEASE COLLABORATIVE NETWORK (RDCN)

- RDCN – Help for adults & children to receive a diagnosis & access treatment & teenagers moving to adult care

YOUR GOPI3KS CHARITY TRUSTEES:

(left to right) Sue, Mandy, Katie & Amanda

MANDY SELLARS - CHAIRPERSON

As 2025 ends, as always I want to say a huge thank you to all those who have supported us through this year. Whether that be volunteering your time or taking on various challenges to raise much needed funds, without you we wouldn't be able to continue to support those within the PROS community. Also to our fabulous trustees, you help this charity to keep on running. This year we held a year-long project with researchers at the University of West England to study the psychological burden living with & affected by PROS brings. We will soon be sharing the educational resources developed from this study which we hope will help patients, families & the wider medical field understand PROS a little more.

Again this year we have been able to help several families & those living with PROS by giving financial support when it's been needed. We understand that PROS can bring its challenges & extra financial worries, so we are always here to help when & where we can. Please send us an email if we can ever help: GoPI3Ks email address

In the meantime I wish everyone a fun, peaceful & happy festive season with loved ones, friends & family. #StrongerTogether

SUE HARPER - TREASURER

Another year has passed already and so much has happened with the charity this year. We are so lucky to have the amazing Dr Madsen working to understand the PIK signalling pathway. Her update makes fascinating reading.

We are also lucky to have had so many great fundraisers this year. Thank you so much to everyone who has put efforts into raising funds for the charity which is so important to it's continued success. Of course the final thanks is to Mandy for all she does for the charity, which involves many hours of work. She attends many meetings, keeping up to date on developments and sharing on social media. She also networks with the rare disease community as well as completing the annual report and accounts for the charity commission.

I'm excited to see what next year brings for Gopi3ks. Hopefully there will be a family weekend! Wishing you all a happy and healthy Christmas and New Year.

YOUR GOPI3KS CHARITY TRUSTEES:

**(left to right)
Sue, Mandy,
Katie & Amanda**

AMANDA KENYON - SECRETARY

Hi!
Here we are! It's that time again where we can't believe how fast the year has gone!
Did you set any goals or make any resolutions? Are you planning any for 2026?

I believe it's really important for everyone have a life outside of either being a carer or having a disability. Something that is just yours. An escape and a chance to be you. I know that that's not always easy or practical and if getting to a venue is a struggle, there are options online. BBC Maestro is fantastic and the skills you can learn are endless. It is a subscription. YouTube is also a great resource for learning new things. From makeup tutorials to cookery classes. It's all there.
I always wanted to act and study performing arts. I just didn't have the confidence to change my degree course. This actually turned out quite well for me as I got to meet one of my best friends and favourite people in the world. Our Mandy! I can't believe that was nearly 30 years ago!

Anyway, a couple of years ago I joined a drama workshop. I got the bug and this year I made my theatre debut and directorial debut! I am extremely proud of this! Now, acting isn't for everyone but there are many things you could try. It is never too late to chase your dreams even if the route there changes. I would love to hear if you have hobbies already or what you're going to try.
As always, we know how tough times are and it doesn't look like things are going to get better any time soon. Just know you're not alone. There is help available. Your local council is the one to go to first. We are always here too.
However you spend this time of year, stay safe, stay well and we will see you in 2026!

KATIE KAVANNAH - TRUSTEE

As we reach the end of another year, I want to take a moment to thank you for being part of this community. This season reminds us of the importance of connection, gratitude, and kindness as well as a sense of belonging.

Thank you to everyone who has contributed over the course of the last year with fundraising, your efforts are appreciated greatly. And for those of you who may be in need, please do reach out. The purpose of this Charity is to be able to give back and support each and every one of you, whether that's a listening and supportive ear or a financial contribution towards making your life a little easier.

May your holidays be filled with warmth, joy, and moments of peace with the people who matter most. Whether you're celebrating near a glowing tree, traveling to see loved ones, or enjoying a quiet winter evening, we hope this time brings you happiness. It's the people who stand beside us that matter the most, not about the presents under the tree. Remember to think about yourself too, and what makes you happy.
Wishing you a magical Christmas and wonderful New Year!

COPING AT CHRISTMAS.

We know that Christmas isn't always an easy time for some people. So we have put together a few resources that may help should you need them.

CALM - 0800585858

Mind - 03001233393

No Panic - 08449674848

Bereavement- 08088081677

PAPYRUS -08000684141

Samaritans - 116 123

SHOUT - Text SHOUT to 85258

Abuse - 0808 2000 247

Refuge -020 7395 7700

Christmas Self-Care Checklist

- ☐ Ensure all medication has been collected from the Chemist.
- ☐ Join an online community to help support you over the Christmas period.
- ☐ Try to stick to your regular routine as much as possible.
- ☐ Spend time with loved ones and seek support from them in times of need.
- ☐ Discover a new hobby - something to distract you if Christmas is a tough time.

COPING AT CHRISTMAS.

it's ok if

- you've never liked the holidays
- You're grieving
- You're not mentally doing well
- your holidays look a lot different
- you're alone or isolated from loved ones
- you need space from family

if you need help and support over Christmas please visit www.lawellbeing.co.uk

Mental Health Ireland

Healthy Coping Tools at Christmas

The holiday season can challenge routines and tempt us into unhealthy coping habits, like excessive drinking or neglecting self-care. These tools are designed to provide positive, structured ways to manage stress and maintain wellbeing.

Meal prep

Plan a few healthy meals or snacks and make a grocery list to keep nutritious options. Preparing easy meals ahead of time, like soups, salads or veggie wraps can help you reach healthier choices. Bring your own food.

Spend time in nature

Aim to spend at least 15 minutes outdoors each day, even if it is just stepping outside for fresh air. For a longer break, visit a park or take a walk where you can enjoy some greenery and unwind.

Connect with someone

Reach out to a friend or family member each day, even if it is just for a quick text or phone call. Schedule a coffee or walk with someone whose company you enjoy for a deeper connection.

Stick to a routine

Choose two or three core parts of your routine. Sticking to these, even if other parts of your days change, can help you feel grounded.

Bring non-alcoholic beverages

Bring a few 0% alcohol drinks or festive non-alcoholic beverages to gatherings to reduce any pressure to drink.

Do something you enjoy

Set aside a specific time each day to do something fun and enjoyable, whether that is reading, crafting or playing a game or listening to your favourite music.

Some useful websites if you are struggling this festive season

- [Top tips for looking after your mental health at Christmas]
- [Coping at Christmas from Mind]
- [Coping at Christmas with the Priory Group]
- [Coping with Grief at Christmas]