NEWSLETTER

Genetic Overgrowth PI3K Support

www.gopi3ks.com

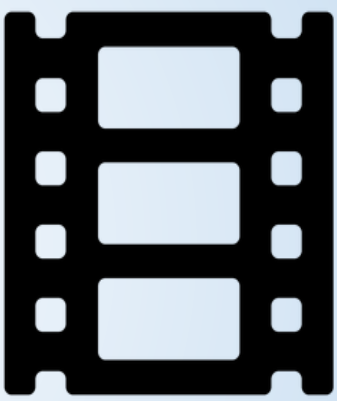
December 2023

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.

GoPI3Ks Family Weekend

A huge heartfelt thank you goes to all the doctors & Lucy Bridge who gave up their precious time to contribute to our family weekend this year. Your contribution means so much & we couldn't have done this without you.

Under every doctors information we have videos on our You Tube channel of their presentations or you can go directly to our channel: GoPI3Ks You Tube Channel



GoPI3Ks Family Weekend

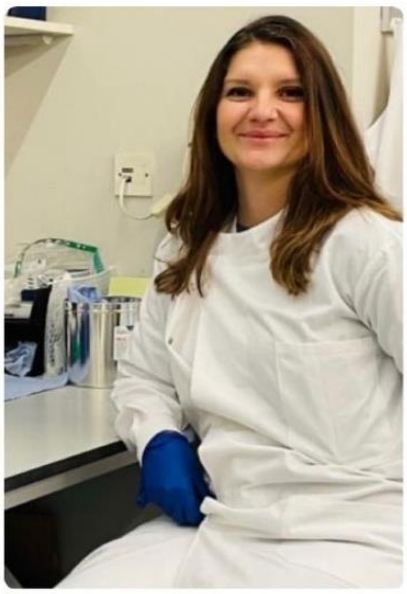
PROFESSOR SEMPLE'S PRESENTATION WILL BE:

AN OVERVIEW OF PROS - PIK3CA RELATED OVERGROWTH SPECTRUM

[Professor Rob Semple's presentation - An Overview Of PROS]

GoPI3Ks Family Weekend

WHO IS DR RALTISA MADSEN?



Dr. Ralitsa Madsen

obtained my undergraduate degree (BSc) in Molecular Biomedicine from the University of Copenhagen (2010-2013), after which I moved to Cambridge, UK to complete an MPhil in Medical Science, under the supervision of Prof Susan Ozanne and Prof Kenneth Siddle at the Metabolic Research Laboratories - Institute of Metabolic Science (Wellcome Trust-MRC). My MPhil research focused on the contribution of microRNAs to the development of insulin resistance as a result of a suboptimal nutritional environment in utero.

I subsequently completed a four-year Wellcome Trust PhD Programme in Metabolic and Cardiovascular Disease (2014-2018), with an initial MRes year. During my PhD with Prof Robert Semple, I engineered the first human induced pluripotent stem cell models with endogenous expression of either one or two copies of the cancer-associated *PIK3CA*-H1047R variant. Our initial aim was to study the potential mechanisms whereby this mutation causes rare, developmental overgrowth disorders known as *PIK3CA*-

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WHO IS DR RALTISA MADSEN?

related overgrowth spectrum (PROS). In the course of this work, we discovered allele dose-dependent effects of genetic PI3Ka activation, suggesting that there are quantitative PI3K signalling thresholds that may determine the pathophysiological consequences of PIK3CA mutations in human diseases, most notably cancer.

In December 2020, I was awarded a Sir Henry Wellcome Postdoctoral Fellowship to study the systems biology of PI3K-dependent phenotypic plasticity, with primary basis at UCL Cancer Institute and the CellSig laboratory of Prof Bart Vanhaesebroeck. I developed novel cellular systems for quantitative, single-cell PI3K signalling studies and applied them to studies of growth factor-

After a short postdoc in the Semple Lab upon its move to the University of Edinburgh (2018-2019), I joined the laboratory of Prof Bart Vanhaesebroeck at University College London. From 2019-2020, I worked on developing highly robust, cell-based quantitative assays for studies of small molecule-mediated *PIK3CA* activation. In a separate project, I also used computational approaches to identify evidence for dose-dependent PI3K signalling activation in human breast cancers with one or multiple copies of activating *PIK3CA* mutations.

specific PI3K signalling fingerprints in different genetic contexts. During this time, I also benefited from a collaboration with Prof Alex Toker at Beth Israel Deaconess Medical Center, in which we focused on the discovery of novel aspects of AKT biology through a multiomic characterisation of a second-generation AKT degrader.

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DR MADSEN'S PRESENTATION WILL BE:

WHAT ARE THE LATEST RESEARCH DEVELOPMENTS WITHIN PROS?

[Dr Ralitsa Madsen's presentation - PI3K Research Update]

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WHO IS ZUBYDA “RARE MINDS”?

Zubyda

Counsellor & Psychotherapist

Zubyda is trained as a psychotherapist and is accredited by the UKCP. She has worked as a therapist in a variety of settings including the NHS and a residential therapeutic community. She has also worked in prisons, and also with migrant refugees in temporary camps in mainland Europe. She was also a volunteer with the Samaritans for over 10 years.

She also works in private practice in central London and offers therapy in both Arabic and English.

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ZUBYDA’S PRESENTATION WILL BE:

Mental Health & Living With PROS

[Mental Health and Living With PROS - Zubyda Azzam]

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WHO IS PROFESSOR MANSOUR?

Sahar Mansour

Clinical interests

Prof Mansour is a Consultant in clinical genetics, whose specialist interests include primary lymphoedema, dysmorphology, skeletal dysplasias, prenatal diagnosis and the genetics of haematological disorders. She is also an honorary professor in clinical genetics at St George's, University of London.

Professional profile

Dr Mansour qualified as a doctor in 1987 at Nottingham University.

- Member: Royal College of Physicians (Paediatrics) - 1993
- Fellow: Royal College of Physicians (London) - 2005

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PROFESSOR MANSOUR'S PRESENTATION WILL BE:

WHAT IS HAPPENING IN THE RECENT ALPELISIB TRIAL IN LONDON?

Unfortunately we aren't able to bring you this presentation at this time

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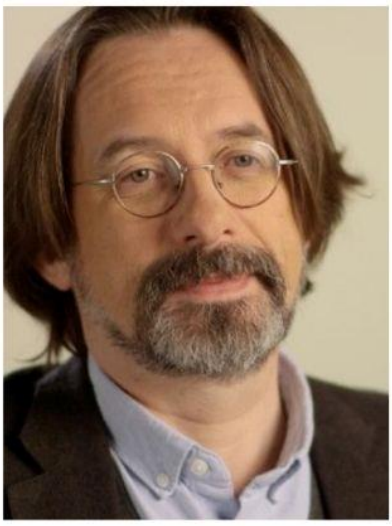
WHO IS PROFESSOR PIERRE VABRES?



Pierre Vabres, MD, has been Professor of Dermatology since 2005, and former Chair of the Department of Dermatology - University Hospital, Dijon, France. His expertise is in Pediatric Dermatology and Genodermatology, particularly in the diagnosis and translational medicine for rare genodermatoses. He manages a national reference center for rare genetic skin diseases (MAGEC-Dijon), specialised in skin mosaic syndromes.

His research activity at INSERM unit UMR1231 focuses primarily on innovative translational and personalised approaches in genetic skin conditions : identification of new genes involved in skin mosaic syndromes through whole genome and next generation sequencing, clinical delineation and deep phenotyping of skin mosaic conditions. He has discovered and characterized new developmental syndromes related to somatic mosaicism. He has been a principal investigator for clinical trials of drug repurposing in the PIK3CA-related overgrowth spectrum. He is also involved in a multidisciplinary research project on the psychosocial impact of cosmetic impairment related to congenital skin anomalies.

He has authored over 200 articles in peer-reviewed journals and has given numerous conferences as an invited speaker at international dermatology meetings. He

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PROFESSOR VABRES'S PRESENTATION WILL BE:

THE MANAGEMENT OF PROS - ALPELISIB, NON-SURGICAL TREATMENT, PREVENTION OF BLEEDING & MORE

[Professor Pierre Vabres - The management of PROS]

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WHO IS DR MAANASA POLUBOTHU?

Maanasa Polubothu MD PhD is an academic paediatric dermatologist, working both as a Paediatric Dermatology Consultant in Great Ormond Street Hospital for Children and as a Principal Investigator in the Molecular Biology of Rare Disease Section, Genetics and Genomic Medicine Programme, UCL GOS Institute of Child Health. She trained in Medicine at the University of Glasgow, and in Paediatrics and Paediatric Dermatology in London. She undertook a laboratory research PhD in Molecular Genetics at University College London. She specialises in the clinical management and genetic investigation of complex high-flow vascular anomalies (arteriovenous malformations) and complex lymphatic anomalies.

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DOCTOR POLUBOTHU'S PRESENTATION WILL BE:

THE RARE DISEASE COLLABORATIVE NETWORK EXPLAINED

[Dr Maanasa Polubothu - Rare Disease Collaborative Network]

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DOCTORS DISCUSSION PANEL

Doctors Discussion Panel

GoPI3Ks Family Weekend

MANDYS PRESENTATION WILL BE:

MY JOURNEY WITH THE DRUG ALPELISIB

Mandy's journey with Alpelisib 2022-2023

THE FAMILY WEEKEND NOVEMBER 2023

On the 18th of November we held our first ever dedicated PROS family weekend at the Holiday Inn at Heathrow in London. This was a chance for adults, children and families to come together & meet others with the same/similar conditions.

As well as listen to various talks from those with expertise in PROS, from research, current drug trials, an overview of PROS, management of PROS, as well as a talk on the Rare Disease Collaborative Network for mosaic conditions. Also for the first time a dedicated talk on mental health & living with PROS.

SCHEDULE:	GoPI3Ks family weekend 2023
9.45 am	Registration
10.15 am	Welcome & Introductions
10.30 am	Professor Rob Semple, overview of PROS
11.00 am	Q & A with Professor Rob Semple
11.15 am	**COFFEE BREAK**
11.35 am	Dr Ralitsa Madsen – Research in to PROS
12.00 pm	Q & A with Dr Ralitsa Madsen
12.10 pm	Lucy – Talk by parent with a child with PROS
12.25 pm	Q & A with Lucy
12.30 pm	**LUNCH**
14.00 pm	Zubyda Azzam from Rare Minds – Mental Health & living with PROS
14.30 pm	Q & A & discussion with Zubyda Azzam
14.45 pm	Doctor Maanasa Polubothu - RDCN
15.15 pm	Q & A with Doctor Maanasa Polubothu
15.30 pm	**COFFEE BREAK**
15.50 pm	Professor Sahar Mansour U.K Alpelisib trial & trial participant
16.20 pm	Q & A with Professor Sahar Mansour
16.30 pm	Doctors' discussion panel
17.00 pm	**END OF DAY**
18.00 pm	**EVENING MEAL**

As the adults listened to the talks, the children were kept entertained & had loads of fun with the wonderful child care service: All Events Child Care run by Lisa Roberts. I would highly recommend them for any event where little ones need taking care of. Even Lisa commented on how she had never seen children who hadn't met before play together so well.

All our children thoroughly enjoyed it and it was so helpful for them to see other children with visible differences. They were already asking when the next family weekend will be!

From one parent: "My child won't usually play with other children, but this weekend played with all the other children."

From an attendee: "It was so lovely to see the children getting along so well & even making sure they had their own table at dinner."

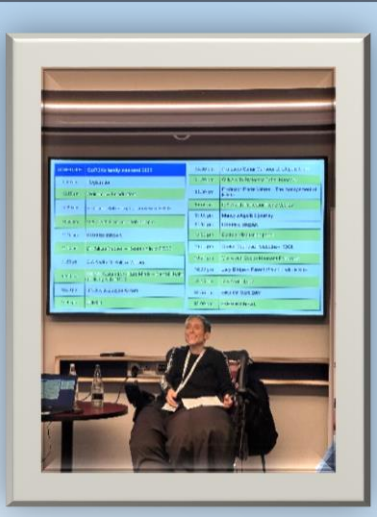
I highly recommend them 2 lady's they did an incredible job feeding and entertaining the kids.

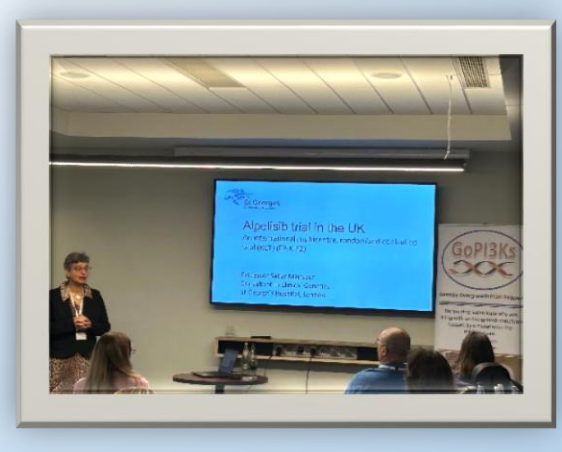
Huge benefit mixing with children with similar condition.

Amazing, my daughter had an awesome time. Those ladies were awesome!

Excellent. Lovely, friendly and inclusive

THE FAMILY WEEKEND NOVEMBER 2023 PHOTOS





MORE FAMILY WEEKEND PHOTOS & FEEDBACK

Was really beneficial sharing experiences with other people. Also for our child to see other people with visible differences. We now have a much clearer medical understanding of what to expect from different drugs.

It was very informative and it helped us appreciate that we are not alone and can empathise with others in the same position

This was the first time we had spoken to doctors that knew the condition and met other people who had similar experiences. I would have loved this as a child, to see others like myself and to know it was okay. We learnt more in one weekend then we had my whole life living with this. I feel like we now have a support network which is wonderful.

We 110% benefitted from the weekend To now officially understand research and meet families in the same situation it was incredible, we will be forever grateful.

I feel this event has helped my mental health so much, but also gave me the courage to help make sure my child is reviewed by the specialists in the area. I honestly cannot thank you enough

Meeting new families and my daughter seeing she wasn't the only child living with pik3ca and she now understand how it effects all of us differently.

I really enjoyed Saturday, all the specialist Drs talks on new medication and treatments. It's so nice to know there is help for the children who are born with PROS.

meeting people and seeing my little boy running round happy

GoPI3Ks Family Weekend – Scientific summary

Disclaimer

This summary was written by Koen Nijbroek (one of the attendees) and is not scientifically verified. It contains the highlights of the scientific part and may contain personal views and interpretations.

10.30 – 11.00 Professor Rob Semple, overview of PROS

* In Mandy's case; H1047L in her leg (heterozygous/mosaic).
* Mosaicism is a common thing, around in nature everywhere.
* We don't know:

What explains the selective pattern of abnormal growth
Which cells are affected
When growth is changed and when it is harmful
Whether/how affected cells influence neighbors
Which problems are reversible
The best way to design drug trial

* Sirolimus does a little bit of good (trials), but mild/significant side effects may occur.
* Taselisib hits the PIK3CA gene itself, but study had to be stopped due to nasty side effects caused by a related gene causing problems in the immune system.
* In 2023 results published by Dr. Canaud and can now be used in the US. Follow-up study is underway.

11.30 – 11.50 Dr Ralitsa Madsen – PI3K Research Update

* Researchers community; PIK3CA Roundtable (share before it's out in the public domain).
* 8-11-23; preliminary results of European multicentric phase III (sirolimus) slow-flow VMs.
* 16-11-23; FDA approves capivasertib with fulvestrant for breast cancer (not yet PROS – could be?).
* You need to tweak the PI3K pathway, you cannot turn it off completely because of all the good things it does, too.
* Madsen lab; focuses on the signals of the PI3K pathway with the goal 'to turn it back to normal'.

PIK3CA variant causes signal misinterpretation, like a 'blurred vision'.
Systematic mapping of the PI3K code across contexts (work to be started).

- Different cell types
- More signals
- Different mutations (now: H1047R)

Predictive computational models.

* Wilms Tumor only found in kidney related PROS. No higher chances of cancer (Rob Semple).
* The PIK3ca affected tissues are usually not prone to cancer compared to PTEN eg.

12.00 – 12.30 Zubyda Azzam from Rare Minds – Mental Health and living with PROS

* Service Provision
* Training & Support
* Research & Awareness
* There is no health without your mental health. You cannot pull them apart.
* Notice feelings, don't push them away (they will come back).
* You can control how you behave; not how other people behave.

GoPI3Ks Family Weekend – Scientific summary

14:30 – 14:50 Professor Pierre Vabres – The management of PROS

* PROS is like the 'Blind man and the elephant' story, in various aspects.
* Due to the overlap in cancers, there is a variety of drugs out there applicable for PROS.
* It is not that straightforward to move drugs developed for cancers to PROS (e.g.) as primary outcome, secondary outcome et cetera are completely different. Treatment times and drug dosages are also completely different.
* From the phase II sirolimus study; definite improvement in pain.
* From the PERFORMUS trial: volume not much changed, improvement in pain reduction / QoL.
* Quite a high number of adverse events observed in their own studies.
* Few results (not trial) from patients treated with Alpelisib:
Patient 1: small decrease of lesion, after 3-6 months: no further improvement.
Patient 2: no response in terms of volume, no real improvement.
* TOTEM: Trial of Taselisib in Overgrowth: assess safety of the drug (30 patients).
The trial stopped after only 16 patients, due to severe adverse side effects.
* SESAM? Trial: Alpelisib in MCAP.
* General issue: risk of cancer in PROS. Theoretically it could be a predisposition.
Retro perspective study: low risk in PROS throughout life.
In study of literature: cases of Wilms tumor (although in PROS very low, <5%).
Other individual cases observed, unclear if related to PROS, no evidence.
* Re-analysis of patients with PROS, half of them were PIK3CA+, half PIK3CA- (PIK3R1+ mutations observed in 15 patients). PIK3R1 may cause KTS-type overgrowth.

15:30 – 16:00 Doctors' discussion panel (on the spot audience questions not noted down)

GoPI3Ks Family Weekend – Scientific summary

Question about the occurrence of double mutations.
* Pierre: In a large panel of various diseases; extremely low frequency observed. Their effect is not yet known whatsoever – as well as how they occur. Rob comments on that as well, also observed in a very low frequency. It seems that it's not shared among the whole area. Also, Sahar reacts that they observed a few.

Question about occurrence in prevalence of twins.
* The general consensus is that no correlation has been observed.

Drug trials for children with segmental overgrowth syndromes?
* This was covered in the talks.

Pulled application of Alpelisib in Europe – and resubmission. Any further information on the long-term outcome?
* Rob: No, it's not completely surprising. It's a very large trial and they are slightly adjusting it, taking some of the learnings. Manaasa: agrees with Rob's view. Approval systems differ slightly between UK/US. There is no extra information.

Sahar mentions that there's already options available that does something already – we shouldn't forget about that as well.

16:00 – 16:20 Doctor Maanasa Polubothu – RDCN Improving the patient pathway for PROS
* Genetic diagnosis of 600 patients.
* Overarching aims;
To increase knowledge and understanding of mosaic disorders.
To progress research.
To improve patient experience.
* Specific aims;
To reduce the mean time to first seeing a specialist.
To reduce the number of trips to the specialist centre.
To improve access to accurate clinical and genetic diagnosis.
To improve the transition from pediatric to adult services, and to provide new adult access to specialist opinion.
To improve coordination of care between the RDCN and local hospitals.
* Opened an adult clinic, reduce mean referral age.
* They're seeing skin diseases + vascular conditions.
Improve speed of access to MDT specialist care in childhood which includes clinical and genetic diagnosis.
Improve access to MDT specialist care in adulthood for clinical and genetic diagnosis and management of new problems.
Improve the transition between the two.

A huge thank you to Koen for this excellent summary of the doctors presentations during our family day.

Mental Health and living with PROS by Zubyda Azzam from Rare Minds

Mental Health and Living with PROS

You and your mental health and emotional wellbeing

Zubyda Azzam, UKCP Accredited
Specialist Counsellor

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Who We Are

- Our Founder has been working as a psychotherapist for those impacted by rare diseases since 2013
- Increasing evidence of, and assessment of, need BUT lack of service provision
- We designed the first specialist post-qualification Training Programme for 'Rare Disease Counselling'
- Launched in October 2020, founded in professional and lived experience
- Team of 9 fully qualified counsellors and psychotherapists
- Our services are mainly Online

To our knowledge, we are the first specialist mental health third sector organisation for the rare disease community

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What We Do

Service Provision	Training & Support	Research & Awareness
Couples/individual counselling.	Clinical supervision for front-line workers.	Presenting at conferences, stakeholder, partners & industry events.
Group programmes.	Rare leader training, group supervision & consultancy.	Contributing case studies & expert opinion.
Workshops.	Workshops, training & courses for HCP.	Build data - initiating & contributing to research.
Self-help psycho-educational resources.	Partnership working.	Inform policy and practice

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There Is No Health Without Mental Health

The relationship between physical and mental health is well established

⇒ Poor mental health > can impact on physical health, the capacity to selfcare, resilience etc.

⇒ Poor physical health > higher rates of depression, anxiety etc.

www.mentalhealth.org.uk/explore-mental-health/mental-health-statistics/physical-health-conditions-statistics

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Life can be busy enough.....

Living with PROS adds another dimension, and it's own challenges:

⇒ Physically

⇒ Practically

⇒ Emotionally

⇒ Relationships

The good news is - it is possible to cope well ☺

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Living with a rare condition like PROS

Lengthy/traumatic diagnosis > Reduced trust in HCP's

Low disease awareness among professionals / public> Isolation, frustration

Multiple uncertainties > anxiety

High logistical and financial burden of disease care / management

Emotional impact of symptoms, treatments, monitoring > Trauma

Feelings of difference / isolation

Complex interacting emotional and physical symptoms

Stress and strain on family life / couple relationships

Visibility / invisibility

The impact on identity, and life choices > Grief and loss

Feelings you might recognise living with PROS

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What feelings do you recognise?

What feelings do you find hard to accept?

You don't have to do anything about the feelings, just acknowledge them

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'Re-visiting' feelings around your condition can happen when:

- ⇉ Disease progression
- ⇉ Child's growing awareness
- ⇉ Re - visiting hospital (and memories)
- ⇉ Having treatments, surgeries, scans and medical appointments
- ⇉ Life stages and choices (relationships, having children, going to university, leaving home)
- ⇉ Comparing life-styles
- ⇉ Noticing limitations and difficulties (in yourself/your child)

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Accepting difficult stuff

- ⇉ **Notice**, and try to name the feeling/s
- ⇉ **Don't judge** the feeling - or yourself
- ⇉ **Decide what helps** ie Talk to a friend/partner? Walk/ Exercise? Draw/paint/write? Music? Distractions?
- ⇉ Learn how to **let the feeling pass**, or reduce
- ⇉ Notice **what – and who - helps** when a feeling is 'visiting'

Remember that feelings 'ebb and flow'

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Disability

According to the Disability Unit of the Cabinet Office, a visible disability is:

"A disability or health condition that is immediately apparent to others. This can include mobility aids, such as wheelchairs or crutches or a visible physical impairment."

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Feelings around the word 'disability'

✓ **Positive:** acknowledged, better understood

✗ **Negative:** lacking, different

? **Both:** positive and negative feelings?

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Living with a visible disability

- ⇉ Wanting to be visible / invisible
- ⇉ Being different, feeling alone, isolated, misunderstood.
- ⇉ Fearful - people can be cruel

Remember - you cannot control how other people behave but you can decide how you want to respond

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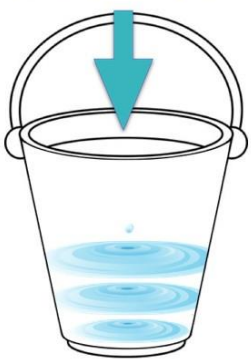
The Stress Bucket:
ordinary life can be demanding enough

Ongoing demands
Big life events
Day to day / week to week issues
Global world events

'Ordinary life'

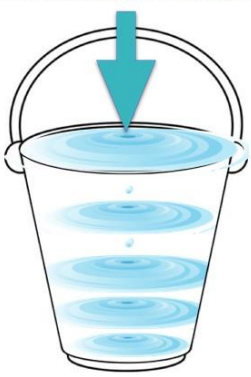


Based on an original concept by Brabben and Turkington (2002

Adding in the demands of living with a rare condition

Uncertainty (micro and macro)
Managing symptoms and side effects
Coping with difficult thoughts / feelings
Family / friends
Healthcare team
Fears about the future

Remember:
Managing the impacts of a rare condition like PROS takes 'emotional energy'



Looking after your stress bucket!

How much room PROS takes up..

....affects how much 'emotional capacity' you have available...

for managing life's 'ordinary' stresses and demands
(and vice versa)

Dynamic relationship

Know when your bucket is filling up

How we behave

How we think / feel

Our body shows us

Managing difficult feelings

⇒ Don't try and push away feelings, or be critical of yourself

⇒ Notice 'what you are like'

⇒ Self knowledge can inform and enable choices

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Knowing how much you need

- ⇉ Feelings around being helped (dependence / independence)
- ⇉ Be mindfully mindless at times
- ⇉ Recognising your need and who is good for what

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7 Healthy Habits

1. Self awareness
2. Practical self-care
3. Acknowledging and allowing difficult feelings
4. Learning about your condition
5. Working with your team
6. Finding your community!
7. Helping others help you

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Looking after yourself and your family

- ⇉ Living with PROS takes physical and emotional energy in its own right
- ⇉ Accept your feelings
- ⇉ Recognise and know your 'stress signs' and who is good for what
- ⇉ Looking after yourself emotionally is *integral* to your journey

Don't struggle alone!

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An important part of me

"I wouldn't have chosen to have all this, but I do see life differently now. I don't 'sweat the small stuff' as much. I used to be stressed out all the time, but I think I'm probably nicer to be around now"

(Mike, 48 Dad to Polly age 10)

"It's weird, it's part of who I am now. I wouldn't be me without it - I'd be someone else, and I don't mind who I am. Even my scars (which I never thought I'd say.)"

(Maddie, 18)

"I felt really frightened at first and couldn't cope. But I've learned to accept the ups and downs, and take things a bit at a time. My counsellor says 'just do the next thing, that's all.' I feel good about realising I'm stronger than I thought I was."

(Aredi , 35 – Mum to India age 4)

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Personally identifiable information erased.

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General mental health support organisations

Every Mind Matters self-help resources and advice on common issues such as anxiety, sleep problems, coping with worry and managing low mood. It also has information on how to access **free NHS talking therapy** **www.nhs.uk/every-mind-matters**

Anxiety UK a charity providing support and information on managing anxiety, stress, anxiety-based depression and phobias **www.anxietyuk.org.uk**

The British Association for Counselling and Psychotherapy provides information about counselling, and a list of private counsellors and therapists by location and speciality (for example physical illness / cancer / relationships) **www.bacp.co.uk**

Carers UK provides advice, information and support for the partners, spouses and other family members of those living with a health condition. **www.carersuk.org**

Crisis Support:

Samaritans **116 123** contact by phone, text or email 24 hours **www.samaritans.org**

Text SHOUT to 85258 - a free, confidential 24-hour text messaging support service

Papyrus support for young people under 35 who are experiencing thoughts of suicide, as well as people concerned about someone else. **0800 068 4141 - (9am-midnight)**

Helpful resources

For General Anxiety & Mood:

Smartphone/tablet Apps:

BUDDHIFY Meditation done differently · headspace · Calm

Mindfulness Courses:
for specific rare conditions, pain and fatigue, or anxiety

Rareminds runs 8-week Mindfulness Programmes for both specific patient communities, or generically.

Please contactus@rareminds.org to register your Interest for upcoming programmes

For Fatigue:

Spoon Theory

https://www.bjchealth.com.au/blog-fitness/the-spoon-theory-managing-fatigue-with-chronic-illness

From Fatigue to Energy: Oxford University NHS Trust

https://www.ouh.nhs.uk/patient-guide/leaflets/files/45345Pfatigue.pdf

Helpful Books:

Bogosian, A. (2020) *Living Well with a long-term health condition* (2020) Routledge. London

Berhard, T *How to Live Well with Chronic Pain and Illness*

Burch , V (2008) *Living Well with Pain and Illness.* Piatkus. UK

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Mental Health for the Rare Disease Community

OUR 2023 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.

FEBURARY 2023

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 donated £52.18 THANK YOU. Although this scheme has now ended we're grateful for your support.

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

Thank you to those who choose to support us when using PayPal. Your kind gesture donated over £127 to the charity.

FUNDRAISER FROM A GOPI3KS FAMILY

One of the GoPI3Ks members Lucy Bridge who is a great support to the charity held a Go Fund Me fundraiser to raise money so that Lucy, GoPI3Ks & Dr M Polubothu could host a PROS Day at Great Ormond Hospital, London to highlight PROS to as many doctors as we could. From GP's to consultants to those working with PROS children, e.g teachers, social workers etc

Lucy raised an amazing £2111.00

Thank you

FUNDRAISER FOR THE FAMILY WEEKEND

We held a fundraiser on social media to help with the costs of the family weekend & the generosity of people we raised over £380 which was a big help.

PENDLE POWERFEST FUNDRAISER

In May we attended the annual PENDLE POWERFEST car show where we ran a tombola & sweet stall. Along with Mandy & Sue we had some fabulous volunteers, Vicky, Anne, Caz & Jackie who we couldn't have done it without, so a HUGE thank you to them. We raised over £190

GRANTS GIVEN TO PROS FAMILIES IN 2023

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

Just before the end of last year we were able to help out one of our adult members with her heating bill by giving a grant of £1000.

In early 2023 we helped out one of our members with the cost of accommodation during a hospital visit to London.

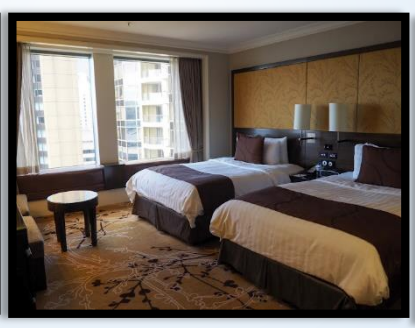
We were also able to help one of our younger members who needs to use equipment to help them breath & a newer more lightweight version was needed.

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. So we were able to help with trainers & shoes for school & everyday wear.

As we hosted the family weekend this year, we were able to help some of our families that would have struggled financially to attend the event. Providing them with overnight stays, travel and food expenses.



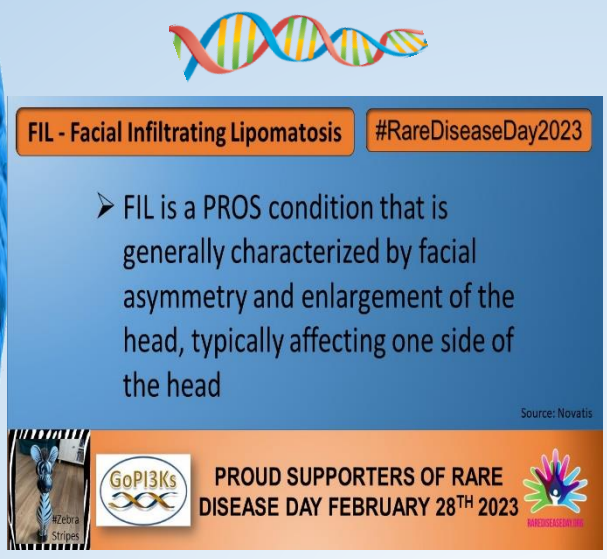
We are here to help you with those extra costs that living with PROS can bring. Help to heat your home, hospital appointment costs (travel & accommodation) Help with equipment/aids such as shoes, wheelchairs, bespoke clothing & much more. Contact us for more info: gopi3ks@yahoo.com

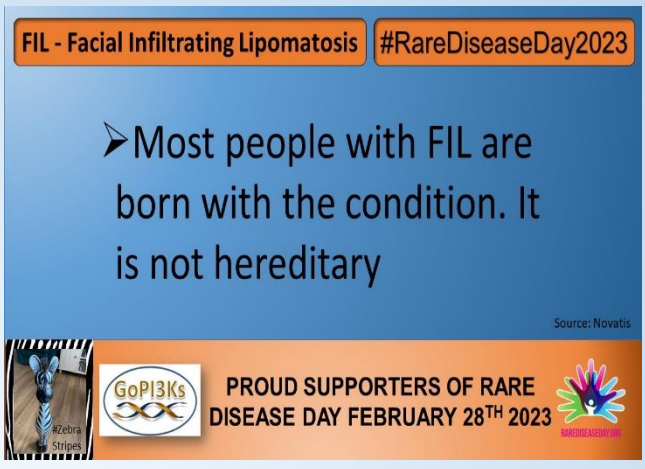
To apply for financial assistance please use this link to fill out our application form: GoPI3Ks funding application form

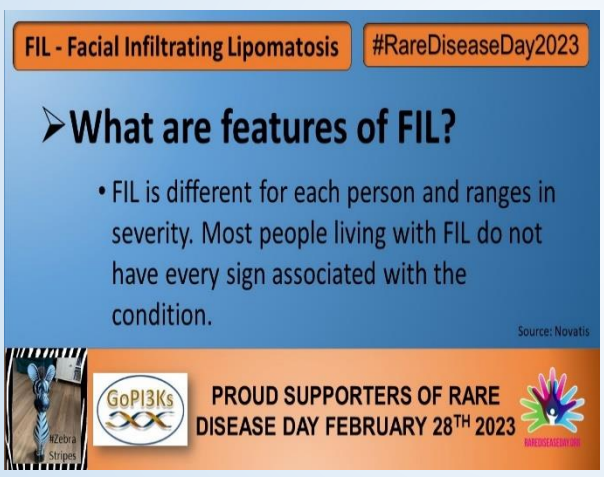
February 2023

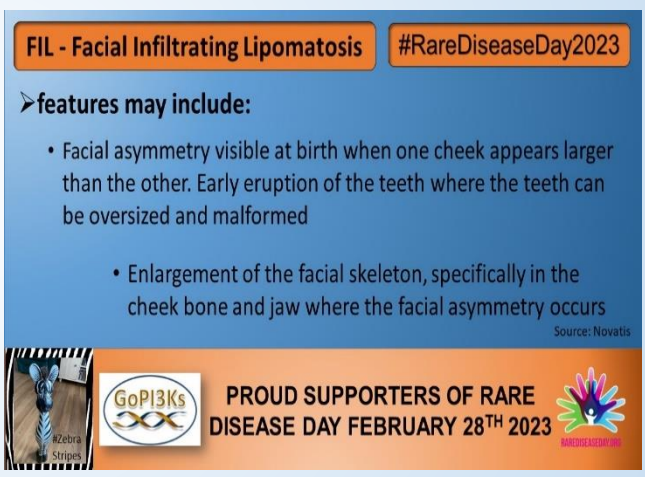
In February 2023 we took part in Rare Disease Day by sharing information on PROS, as well as what PROS individuals & families thought the PROS & CONS of having PROS is.

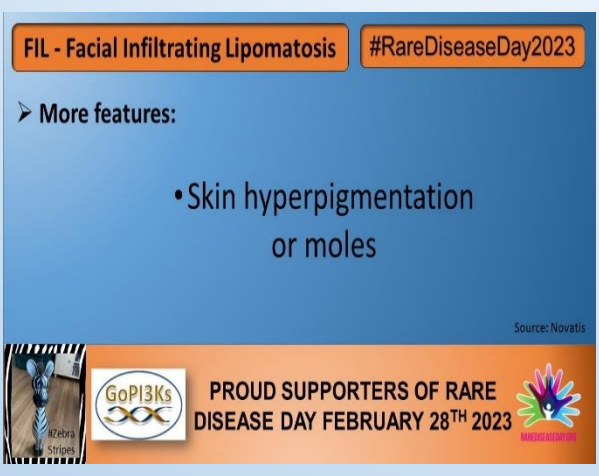
FIL – Facial Infiltrating Lipomatosis

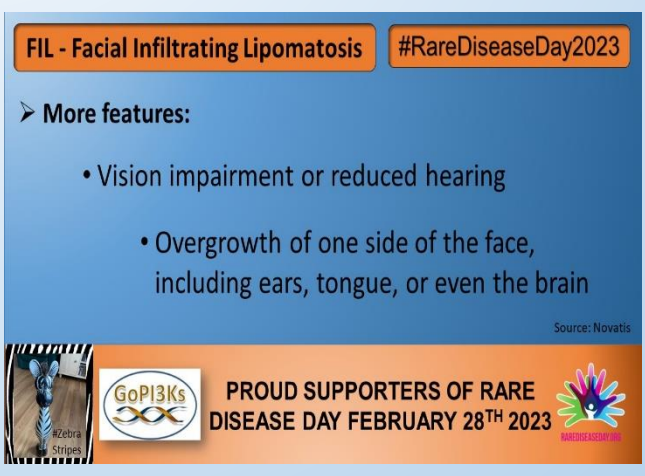


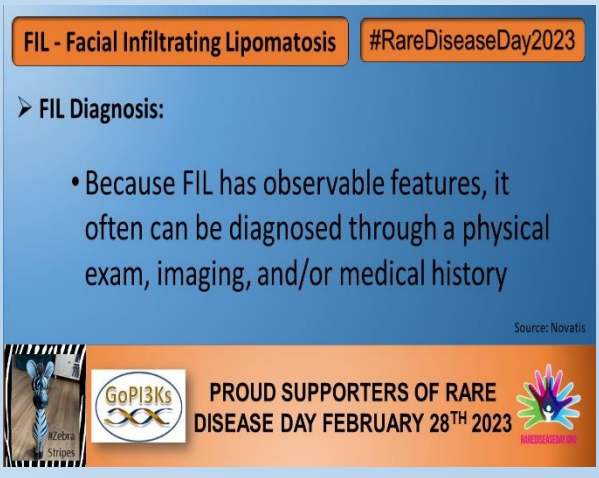


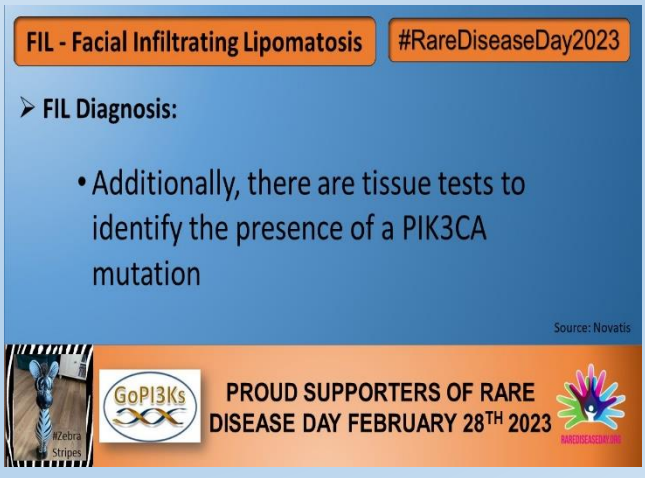












FIL - Facial Infiltrating Lipomatosis | #RareDiseaseDay2023

FIL Diagnosis:

- However, a negative test result does not rule out having a PROS condition as mutations can be difficult to detect

Source: Novatis

FIL - Facial Infiltrating Lipomatosis | #RareDiseaseDay2023

FIL Management:

- Managing FIL can be challenging and typically requires collaboration from a multidisciplinary team.

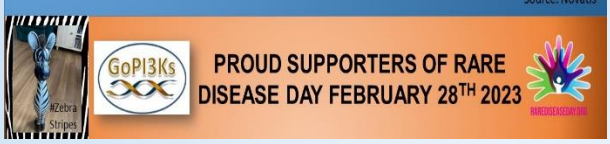
Source: Novatis

FIL - Facial Infiltrating Lipomatosis | #RareDiseaseDay2023

FIL Management:

- Like other PROS conditions, there are few management options for FIL

Source: Novatis



FIL - Facial Infiltrating Lipomatosis | #RareDiseaseDay2023

FIL Management:

- Some options, such as surgery, address symptoms and manifestations rather than the root cause of the disease

http://www.wonderfilsmiles.com/

Source: Novatis

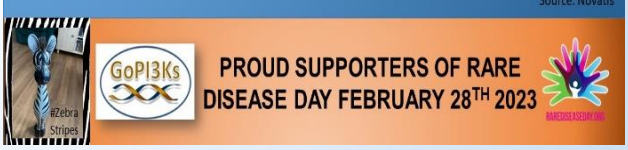
FAVA – FibroAdipose Vascular Anomaly

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

What is FAVA?

- 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

Source: Novatis



FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

What are features of FAVA?

- 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

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

These 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

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

These 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

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

FAVA Diagnosis:

- Because FAVA has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history

Source: Novatis

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

➢ FAVA Diagnosis:

- Additionally, there are tissue tests to identify the presence of a PIK3CA mutation

Source: Novatis

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

➢ FAVA Diagnosis:

- However, a negative test result does not rule out having a PROS condition as mutations can be difficult to detect

Source: Novatis

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

➢ FAVA Management:

- Managing FAVA can be challenging and typically requires collaboration from a multidisciplinary team

Source: Novatis

FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

➢ FAVA Management:

- Like other PROS conditions, there are few management options for FAVA

Source: Novatis

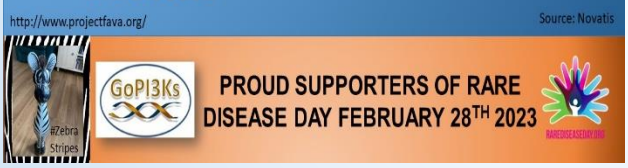
FAVA - FibroAdipose Vascular Anomaly | #RareDiseaseDay2023

➢ FAVA Management:

- Some options, like physical therapy and surgery, address symptoms and manifestations rather than the root cause of the disease

http://www.projectfava.org/

Source: Novatis



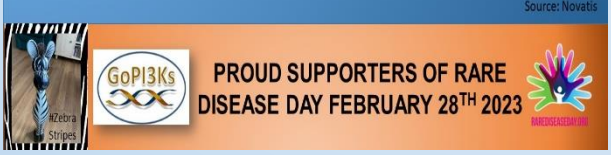
MCM – Megalencephaly -Capillary Malformation

Megalencephaly Capillary Malformation (MCAP or M-CM) | #RareDiseaseDay2023

➢ What is MCAP/M-CM?

- MCAP, sometimes referred to as M-CM, is a PROS condition that is generally characterized by overgrowth throughout the body and brain, developmental delays, and low muscle tone, known as hypotonia

Source: Novatis

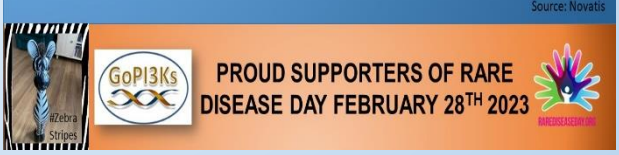


Megalencephaly Capillary Malformation (MCAP or M-CM) | #RareDiseaseDay2023

➢ What is MCAP/M-CM?

- Most people with MCAP/M-CM are born with the condition or have features of MCAP/M-CM during prenatal scans. MCAP/ M-CM is not hereditary

Source: Novatis

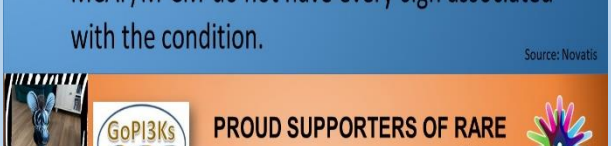


Megalencephaly Capillary Malformation (MCAP or M-CM) | #RareDiseaseDay2023

➢ What are features of MCAP/M-CM?

- MCAP/M-CM is different for each person and ranges in severity. Most people living with MCAP/M-CM do not have every sign associated with the condition.

Source: Novatis



Megalencephaly Capillary Malformation (MCAP or M-CM) | #RareDiseaseDay2023

➢ These features may include:

- Brain irregularities due to irregular widening of the sac-like spaces (or ventricles) of the brain, which contain cerebrospinal fluid (CSF)

Source: Novatis

Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

These features may include:

- Excessive accumulation of CSF may lead to hydrocephalus, one of the potentially serious complications of MCAP or M-CM

Source: Novatis

Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

These 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

Source: Novatis

Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

These features may include:

- Vascular anomalies, like skin mottling spread over the trunk, limbs, and face, known as capillary malformations

Source: Novatis

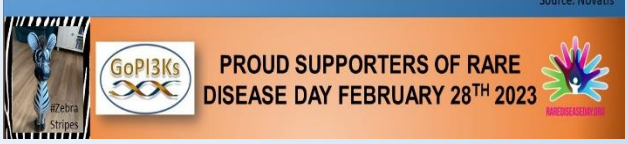
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

These features may include:

- Body asymmetry, such as webbed toes or fingers

Source: Novatis



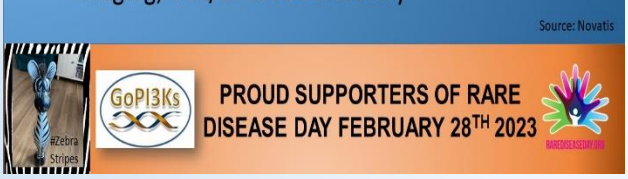
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Diagnosis

- Because MCAP/M-CM has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history

Source: Novatis



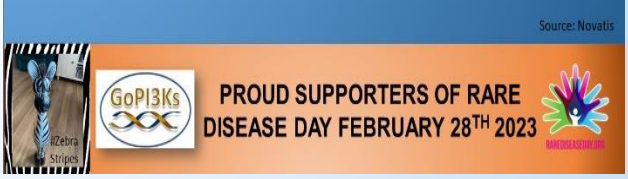
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Diagnosis

- Additionally, there are tissue tests to identify the presence of a PIK3CA mutation

Source: Novatis



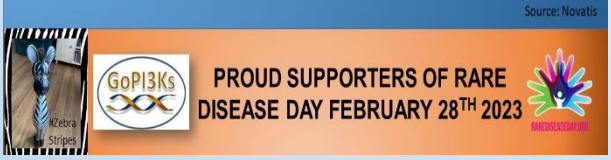
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Diagnosis

- However, a negative test result does not rule out having a PROS condition as mutations can be difficult to detect

Source: Novatis



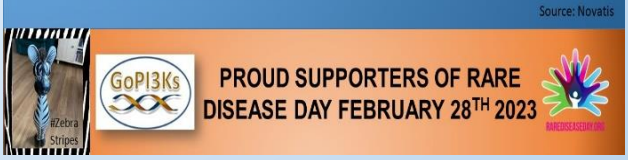
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Management

- Managing MCAP/M-CM can be challenging and typically requires collaboration from a multidisciplinary team

Source: Novatis



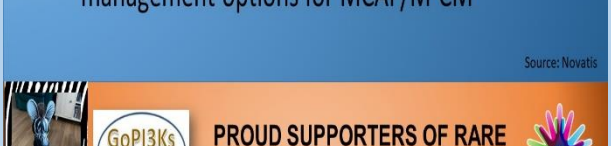
Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Management

- Like other PROS conditions, there are few management options for MCAP/M-CM

Source: Novatis



Megalencephaly Capillary Malformation (MCAP or M-CM)

#RareDiseaseDay2023

MCAP/M-CM Management

- Some options, like physical therapy and surgery, address symptoms and manifestations rather than the root cause of the disease

http://www.m-cm.net/

Source: Novatis

CLOVES – Congenital Lipomatous Overgrowth Vascular Malformations Epidermal Nevi Scoliosis

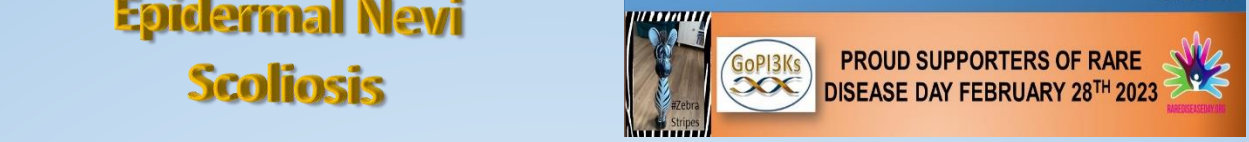
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What is CLOVES syndrome?

- CLOVES is a PROS condition that is generally characterized by overgrowths on the body and irregularities in blood vessels

Source: Novatis



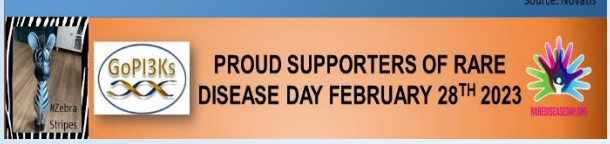
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What is CLOVES syndrome?

- Most people with CLOVES are born with the condition or have features of CLOVES during prenatal scans. It is not hereditary

Source: Novatis



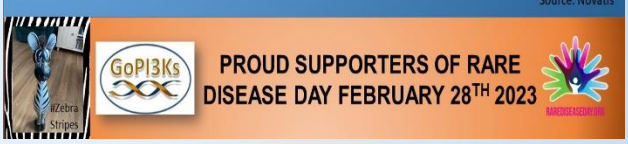
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are features of CLOVES?

- CLOVES is different for each person and ranges in severity. Most people living with CLOVES do not have every sign associated with the condition.

Source: Novatis



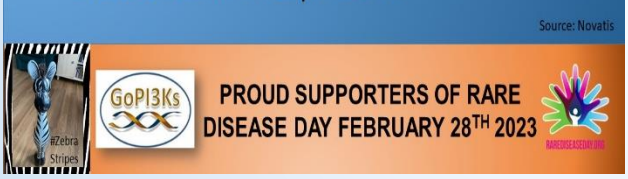
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are 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

Source: Novatis



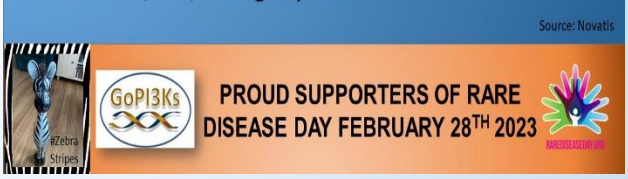
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are features of CLOVES?

- Skin lesions, port-wine stains, birthmarks, or other discolorations known as epidermal nevi that can be flat, tan, or slightly raised

Source: Novatis



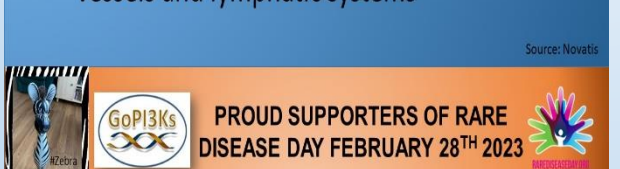
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are features of CLOVES?

- Vascular anomalies in the formation of blood vessels and lymphatic systems

Source: Novatis



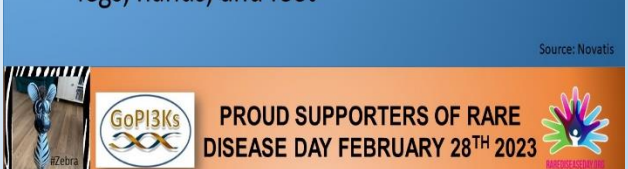
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are features of CLOVES?

- Overgrowth of extremities, including arms, legs, hands, and feet

Source: Novatis



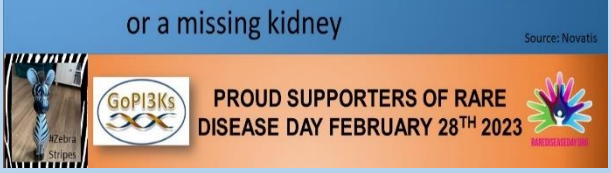
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

What are features of CLOVES?

- Scoliosis or curving of the spine
- Differences in the size of the kidney, or a missing kidney

Source: Novatis



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

#RareDiseaseDay2023

What are features of CLOVES?

- Atypical knee caps and joints of the hip and knee

Source: Novatis

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

#RareDiseaseDay2023

CLOVES Diagnosis

- Because CLOVES syndrome has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history

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

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CLOVES Diagnosis

- Additionally, there are tissue tests to identify the presence of a PIK3CA mutation

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- However, a negative test result does not rule out having a PROS condition as mutations can be difficult to detect

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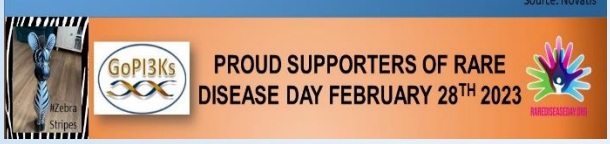

CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

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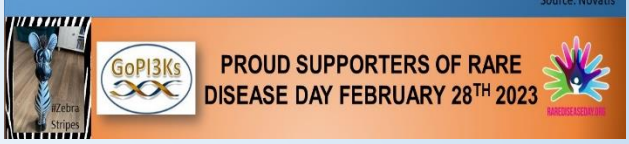
CLOVES Management

- Managing CLOVES can be challenging and typically requires collaboration from a multidisciplinary team

Source: Novatis



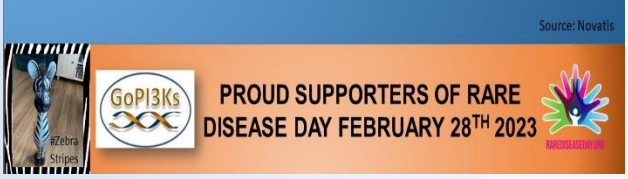
CLOVES - Congenital Lipomatous Overgrowth Vascular malformations Epidermal nevi Scoliosis/skeletal and spinal

#RareDiseaseDay2023

CLOVES Management

- Like other PROS conditions, there are few management options for CLOVES

Source: Novatis



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

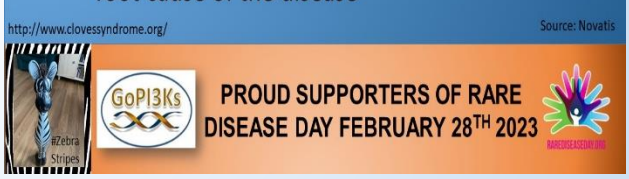
#RareDiseaseDay2023

CLOVES Management

- Some options, such as surgery, address symptoms and manifestations rather than the root cause of the disease

http://www.clovessyndrome.org/

Source: Novatis



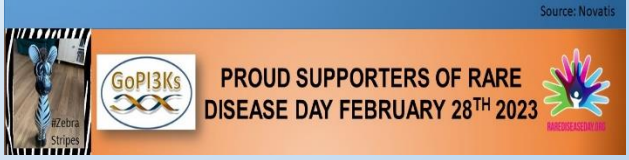
KTS – Kippel-Trenaunay Syndrome

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

What is KTS?

- 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

Source: Novatis



KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

What is KTS?

- Most people with KTS are born with the condition. It is not hereditary

Source: Novatis

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

What are features of KTS?

- KTS is different for each person and ranges in severity. Most people living with KTS do not have every sign associated with the condition.

Source: Novatis

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- These features may include:
  - Varicose veins in the affected limb
  - Limb may be larger or smaller than unaffected limbs

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- These features may include:
  - Slow-flow vascular and lymphatic malformations, such as enlarged bones and/or soft tissue

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- KTS Diagnosis
  - Because KTS has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- KTS Diagnosis
  - Additionally, there are tissue tests to identify the presence of a PIK3CA mutation

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- KTS Management
  - Managing KTS can be challenging and typically requires collaboration from a multidisciplinary team

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- KTS Management
  - Like other PROS conditions, there are few management options for KTS

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

KTS - Klippel-Trenaunay Syndrome #RareDiseaseDay2023

- KTS Management
  - Some options, such as surgery, address symptoms and manifestations rather than the root cause of the disease

www.k-t.org

Source: Novatis

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

Voices Of The Community

VOICES OF THE PROS COMMUNITY #RareDiseaseDay23

- Are there any positives from living with PROS?
  - Getting to meet, even if it's virtual the rare community, especially my PROS family.

Adult living with PROS

PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

VOICES OF THE PROS COMMUNITY #RareDiseaseDay23

- Are there any positives from living with PROS?
  - Being able to appear on TV, radio shows & written media to talk about my condition & educate others that rare doesn't mean scary, weird & that we just like anyone else.

Adult living with PROS

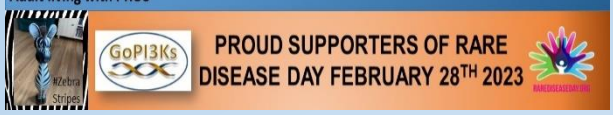
PROUD SUPPORTERS OF RARE DISEASE DAY FEBRUARY 28TH 2023

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - To have met some amazing doctors who took a chance on me to investigate my condition.

Adult living with PROS

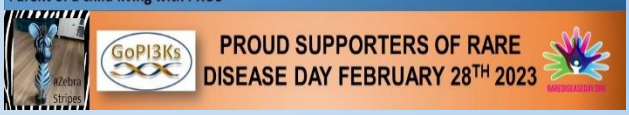


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - I feel like you enjoy milestones more when they come, joy in little things your child does because they aren't guaranteed.

Parent of a child living with PROS

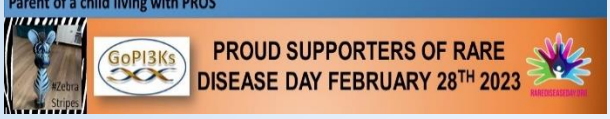


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - I've bought star shaped lights for my house because it was the only word my daughter said for ages. They mean so much to me & they wouldn't if there wasn't any uncertainty about my daughter.

Parent of a child living with PROS

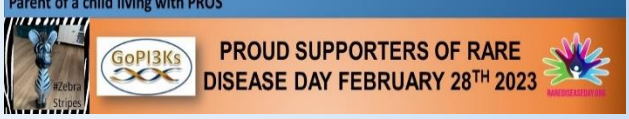


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - I love my child to bits just as they are.

Parent of a child living with PROS

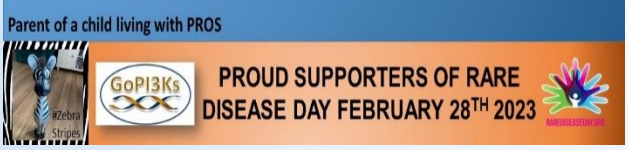


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - The experience of medical help we have received is brilliant, our confidence has grown especially with great Ormond Street and have come to trust them more, over the years things have changed for the better with help of research.

Parent of a child living with PROS

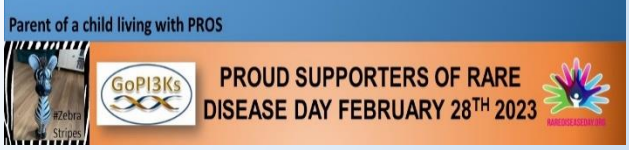


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - Positives to living with PROS, great network of friends online

Parent of a child living with PROS

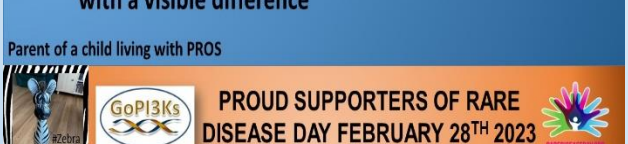


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - Pros has thought us a lot as a family, about disabilities and how easy it is to personally offend someone who's struggling mentally, physical and emotional with a visible difference

Parent of a child living with PROS



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?
  - This has helped my daughter comfort friends who are having a hard time too.

Parent of a child living with PROS

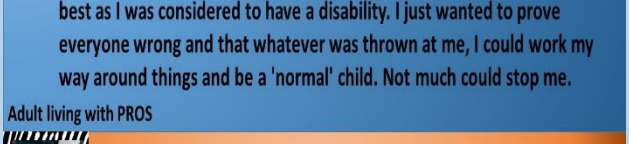
VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any positives from living with PROS?

Having PROS has made me who I am today? It has made me more caring, more determined to succeed. It has certainly made me competitive. I think I have always felt, especially when younger, that I needed to be the best as I was considered to have a disability. I just wanted to prove everyone wrong and that whatever was thrown at me, I could work my way around things and be a 'normal' child. Not much could stop me.

Adult living with PROS

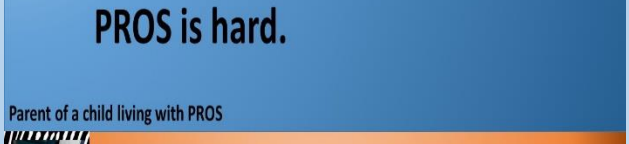


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - The worry and unpredictability of PROS is hard.

Parent of a child living with PROS

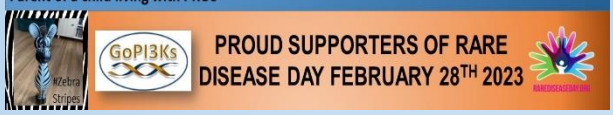


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - The one negative that probably stands the most out and is most present in our day to day lives are the stares and the comments

Parent of a child living with PROS



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - With that comes a deep concern of how my child will handle this during the coming years. Will they have the strength to withstand and persevere.

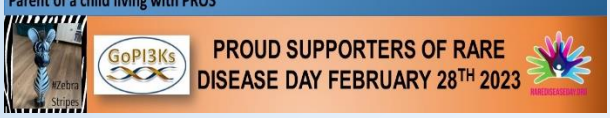
Parent of a child living with PROS

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - It does take a lot of a person to handle this, especially getting through those difficult teenage years.

Parent of a child living with PROS

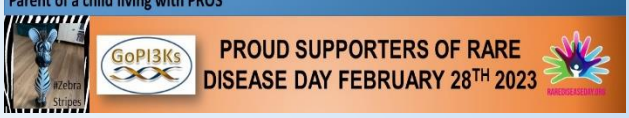


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - Being looked at funny by medical students, children bullying.

Parent of a child living with PROS



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - Having to find coping ways to handle the stares & verbal abuse & knowing these are going to continue all your life.

Adult living with PROS

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - Losing parts of my independence, so that life becomes limited & reliance on others for simple things, such as going to the bathroom.

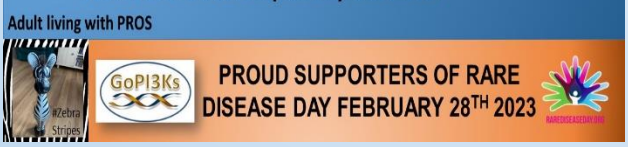
Adult living with PROS

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - Not many people medically have heard of PROS. It becomes difficult even going to my GP as I have to explain what issues I have with my hand and arm. Even though PROS is mentioned somewhere in my medical file, a Dr will just give me a blank look. I then have to explain my condition.

Adult living with PROS

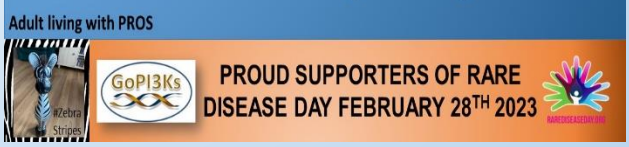


VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - As I get older osteoarthritis is causing havoc to my joints and previous surgeries. Where medical intervention used to correct and prevent things, this tends not to be the case anymore as I get older.

Adult living with PROS



VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - Getting funded help is also proving difficult. I no longer work, but again, it has proved to be difficult to get someone reading an assessment form to engage in what PROS is, and how it is now impacting my daily living, such a cooking, reaching up into cupboards, sometimes dressing & showering, driving, etc.

Adult living with PROS

VOICES OF THE PROS COMMUNITY

#RareDiseaseDay23

- Are there any negatives from living with PROS?
  - The heaviness of my enlarged arm has given me slight scoliosis and secondary backache, as my body is out of alignment on one side.

Adult living with PROS

PROS EDUCATIONAL DAY 2023

On April 24th 2023 GoPI3Ks along with Lucy Bridge & Dr Maanasa Polubothu hosted a meeting at GOSH entitled: PROS - PIK3CA Related Overgrowth Spectrum meeting – updates & advances in MDT care & navigating the patient pathway.

Those attending were specialist physicians and researchers in PROS, physicians and other healthcare professionals treating PROS patients and members of the patient community.

Time	Topic	Speaker
12:00 – 13:00	Lunch and Arrivals	
13:00 – 13:10	Introduction	Maanasa Polubothu (GOSH) & Mandy Sellars (GoPI3Ks)
13:10 – 13:35	PROS: an overview	Maanasa Polubothu
13:35 – 14:20	Mosaic Disorders: The role of the MDT Plastic surgery Interventional Radiology Orthopaedics	Branavan Sivakumar, Plastic Surgeon (GOSH) Alex Barnacle, Interventional Radiologist (GOSH) Deborah Eastwood, Orthopaedic surgeon, GOSH
14:20 – 14:50	Novel Medical Therapies for PROS	Pierre Vabres (CHU Dijon-Bourgogne)
14:50 – 15:10	Coffee	
15:10 – 15:30	The patient experience Adults and paediatrics	Mandy Sellars (GoPI3Ks) & Lucy Bridge (parent)
15:30 – 16:00	Establishing an NHS England Rare Disease Collaborative Network	Veronica Kinsler (UCL GOS Institute of Child Health)
16:00 – 17:00	Facilitated discussion Next steps towards a patient pathway for PROS	All

What were our aims?

- Update the PROS community on the latest developments within the field
- Provide a forum for discussion about the PROS patient pathway including the role of GOSH following grant of rare disease clinical (RDCN) network status and adult and paediatric patient experiences
- Strengthen links amongst members of the PROS community for future collaboration

THE PATIENT AND CARER JOURNEY

As a parents & people living with PROS we hear many times from families & individuals how they struggle, who do they turn to when they need help.

We sent out a survey to gauge patients & families feelings on their medical care.

This meeting gave us a chance to let those within the medical field understand a little more about the PROS journey.

The summary presented here is a combination of themes from the survey and what we have seen discussed regularly.

Surgery

- There is a great deal of uncertainty amongst the community about surgery
- Will it all just grow back? Will it grow back worse? Will it cause more problems such as nerve damage?
- Who should be performing the surgery, is special expertise required?
- Poor expectation management about duration of aftercare and extent of pain (experience of leg length discrepancy surgery). Because surgery is discussed often, could it be that a level of knowledge is presumed?

"My daughters legs are now of the same length however. But we have both suffered so much. Memories of the 1st operation haunt me, not knowing that she should have been in a wheelchair after it, too ill to use her crutches, trying to carry a grown child to a toilet on the other side of a service station, her cries of agony in a public place."

"I felt extremely lucky when I found a surgeon who would perform my amputation, never had any doctor in that area performed this on someone with such large lower limbs. I had a lot of faith in this surgeon & he saved my life"

OUR ASK:
To have surgery covered within the RDCN training (if not already)

A group of surgeons with expertise to support patient materials

Mental health

- PROS is associated with significant mental health burden
- Anxiety around visual impact, mobility issues, worries about disease progression, financial worries
- Parents worry about their children and take on the burden of finding care
- New mums are particularly vulnerable and are experiencing grief, especially if they weren't aware of any problems before birth. What special attention could you give to them?

"I felt totally unsupported…the mental load on my daughter is huge, particularly now as a teen" Parent

"When my child was offered counselling, they refused as they felt the counsellor wouldn't know anything about PROS, so what was the point"

"When my daughter was diagnosed the doctor said it was great that she had been diagnosed so early (10 months or so), that was the longest 10 months of my life and I just wanted to know that she was in the right hands"

OUR ASK:

- **Point patients to the GoPI3Ks website: https://gopi3ks.com/resources/**
- **To advise people to speak to their GP, the patient community about their worries**
- **To work with doctors to create a "what happens next" page for website & leaflet for patients.**

Coordination of care

- Referrals are sent off and disappear into the ether, huge amounts of admin to track, follow-up – would be great to have someone explain the process upfront
- Need guidelines or a patient pathway, we don't know what should be happening
- Confusion and delays in getting the referral creates anxiety in patients and carers – it undermines the clinical care even if this is excellent
- Transition from pediatric to adult care

"To be prepared of the numerous hospital trips which need to be undertaken would be helpful to ease the reality of living with PROS."

"Moving from child to adult medical care is a minefield. The patient/family often feel on their own with no real guidance as to the process."

"Have more information to hand. I was totally reliant on Google searches etc, a leaflet with details or an explanation of the condition would be a start."

OUR ASK:
Fundraising for the coordinator at GOSH is essential
Publish a patient pathway, checklist for diagnosis and monitoring

Access to care

- Both adults and children found it difficult to access specialist care
- When they did get to the right person their experiences are generally very good
- Adults report being totally unable to access specialised care in many situations
- A&E is particularly challenging as doctors there don't know what to do, often don't listen to patients and can sometimes use it as a learning experience inviting other doctors to look or asking to take pictures (not the time or place)
- Orthotics is an area that many with foot problems struggle with, from ill fitting, to lack of shoes, to developing sores that become infected.

"Shoes are not provided in a timely enough manner, sometimes are incorrectly fitting, there are gaps where our daughter is without footwear."

"When visiting the GP we get told to, "see how it goes," then you end up back a week later with a severely sick child. They need to trust the parents."

Attending A&E can be tricky – the patient with PROS was asked about their condition but the nurse stated "Oh I have never heard of that, so neither will the doctor, so lets say you have elephantitis!."

OUR ASK:
Discharge letters from GOSH / Guys to include guidance for A&E (where possible)

Thank You

A huge thank you to Lucy & Dr Polubothu for all their hard work to make this meeting a success. Hopefully we will be able to organise more meetings like this to help the medical field understand more about the patient journey, so we can work together.

QUICK LINKS TO INFORMATION:

We know it can be a mindfield 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?

- What is PROS from NORD
- What is PROS from Dr Sanjay & Dr Garg
- PROS for doctors from Novartis
- Information on Alpelisib & Vijoice

EXPLAINING PROS TO CHILDREN

- Explaining living with a rare condition to children of different ages
- PROS leaflet for children from our lovely friends at WonderFIL Smiles

WHAT RESEARCH HAS THERE BEEN IN TO PROS

- Research in to PROS over the years

PSYCHOLOGICAL & WELL-BEING SUPPORT

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

RARE DISEASE COLLABORATIVE NETWORK (RDCN)

- RDCN – scroll down the page to see this

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.

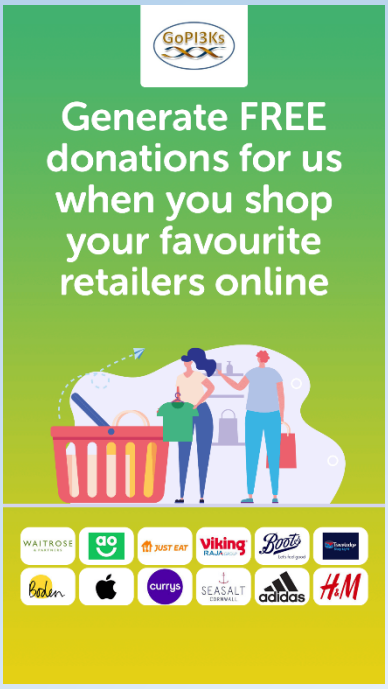
DON'T FORGET

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.

HOW YOU CAN HELP FUNDRAISE FOR GOPI3KS



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

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

PAYPAL GIVING

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

PayPal Giving Fund

YOUR GOPI3KS CHARITY TRUSTEES.

MANDY SELLARS - CHAIRPERSON

As another year ends, I want to take this opportunity to thank those that have donated, raised money, volunteered & supported us during this year. A special thank you to the doctors who gave up their precious time to attend our first ever dedicated PROS Family Weekend to present on all things PROS & what an amazing day it was. Also a special thank you to our other trustees who made the day run smoothly. To see the PROS community building & getting stronger, to see friendships being formed & children seeing others like them & playing together like they were old friends was a very special day that will always stay with me. Also thank you to the doctors who attended The PROS – PIK3CA Related Overgrowth Spectrum meeting –updates & advances in MDT care.

It has also been a pleasure to be able to help some of our GoPI3Ks members this year & we hope to be able to continue this in 2024. I hope that you are all able to have a wonderful festive season & remember we are always here to help: GoPI3Ks email address #StrongerTogether

SUE HARPER - TREASURER

Another year has passed and what a great year for GoPI3Ks! Mandy has again put lots of work into networking with other charities and doctors and processing grants for charity members as well as keeping everyone up to date via social media. She also organised the amazing family weekend where it was so great to meet so many of you. It was so nice to see so much positive feedback, thank you for taking time to do that for us. The family weekend obviously used a lot of Gopi3ks funds so if any of you would like to do any fundraising in 2024 please get in touch with your ideas. It would be great to have the funds to do another family weekend in the future. Wishing all our Gopi3ks families a very Merry Christmas and a Happy New Year.

AMANDA KENYON - SECRETARY

Well it's that time of year again! It's come round very quickly and I hope you've had a blessed one. I like to use this time of year for reflection. For me personally, it's been a great year. I travelled more and even joined a drama group where I was involved in an Agatha Christie play!

It was fantastic to be able to host events again and whilst I wasn't able to attend, I know how valuable this community is. Information sharing is the most vital element and no one knows what its like to live or care for someone with PROS like you do. However you spend the holidays, I hope its filled with warmth, laughter and love If this time of year is challenging for you in anyway, please reach out. There are many wonderful organisations that are here to help Amanda & Dot the Pug

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 can’t believe we’re back here again celebrating another Christmas and another year gone by. How quickly time flies. Recently I had the opportunity to meet many of you at the Family Weekend and share your experiences, which was very special indeed. As such this message means just a little bit more, as I know how important this charity is and the support it provides to so many of you.

I want to wish each and every one of you a wonderful Christmas, and to a healthy and happy 2024. I hope I get the opportunity to meet with you all again very soon.