

PTENUKI

Summer Newsletter

August 2019



Fundraising update

Kelly Kearley (kelly@ptenuki.org)

Trustee Fundraising

Welcome

Ian Stock (ian@ptenuki.org)

Charity chair

Since our last newsletter there has been so much going on it's hard to know where to start. Our patient day in November was a great success. With money raised earlier in the year we were able to assist with travel expenses enabling some people to attend who had never been before, which I'm personally really proud of. We've published anti-bullying resources on the website which was a topic raised during the day, and we made two new videos, one to help with awareness of the charity, and one of Katherine's PTEN Overview presentation, which I'm pleased to say we've launched through the newsletter today and you can find [here](#).

I'd like to say a big thank you to Colette who has stepped up to become a trustee for the charity responsible for Care and Support. She's making great progress in this area having recently launched a buddy scheme and local meet-up groups. I've been busy working with other PTEN groups in Europe reviewing EU wide screening guidelines and also meeting with people to understand how a new PTEN Patient Registry in the UK will work and ensuring patient needs are being considered.

Our members continue to grow, we continue to raise money, we are publishing information on the website regularly and pleased to say we have agreed the date for our next patient day which will be on Saturday 16th November 2019, please put it in your diaries and sign up [here](#).

Finally, Ellie Collins (Trustee Communications) is now Ellie Challis, having got married to the very lucky James - congratulations to you both!!

2019 has been a busy year in the world of PTENUKI fundraising! We want to say a HUGE thank you to everyone who has fundraised for us this year, with sporting events, school events and corporate events around the UK.

On top of fundraising activities, we have been busy applying to Major Trusts in the UK that specifically help causes like our own and we are keeping our fingers crossed on the outcome. We will keep you posted!

Our online fundraising platform, BT My Donate page, has had to close due to the closure of the platform. We have therefore rebranded and opened a Just Giving page from July 2019. If you would like to raise money for our charity through a sporting or community event and would like to use an online platform, then please do now head over to www.justgiving.com/ptenuki for all of the details.

We want to remind all of our supporters that they can also help us to raise money with their on-line shopping by using the Smile Amazon account for the PTEN UK and Ireland Patient support Group. For a direct link to the account please visit our donation page on our [website](#). We can also post out a branded charity money collection tin. If you would like one for your event, local shop or school, then please do get in touch and we can post one out to you.



Meetup Groups

Colette Knight (colette@ptenuki.org)
Trustee Care and Support

Since we launched our Buddy and Meetup group initiatives after last year's Patient Day, we have been busy setting up and implementing the new schemes.

Meetup Groups

PTENUKI now has four regional meet up group reps; Zoe Read in the South, Sarah Plummer in the North West, Yvonne Forrester in Scotland and Sinead O'Hare in Northern Ireland. We are very pleased to announce that Zoe is hosting the first PTENUKI regional meet up in Southampton on the 7th September, at St Marks Church, see <https://ptenuki.org/south-west-support-group/>. Details of all our reps can be found on the PTENUKI website. It's really important that local knowledge and relationships can flourish in our rare community, and our reps are a great place to start.

Buddy Scheme

So far we have had 14 responses; for both those seeking and offering support. Introductions have already been made, and hopefully flourishing. All those requesting support have been matched with a Buddy. As we received an overwhelming response from those offering support, we have created a 'skillset' database, so if we receive requests for help and advice in the future we can refer to our buddy support record and match accordingly!!

We have recently updated the website with all the details of our schemes, <https://ptenuki.org/useful-resources/> including how to sign up and take part. Please take a look and if you require any further information or have some good ideas get in touch with Colette.

Clinical trial update

David Ross

Member of the PTENUKI community

I, with my daughter, attended the 2019 Patient Symposium whilst I was taking part in a clinical trial at Boston Children's Hospital. I heard talks from Kristin Anthony, Claudio Ales about the achievements within the PTEN community over the past year. They talked about the international care guidelines for people with PHTS and the patient registry. Dr Eng gave an explanation of the studies which Cleveland Clinic are involved with. Ramon Parsons, Director of the Tisch Cancer Institute at Mount Sinai spoke about Pten treatment pathways and cancer. Joanne Ngeow, Head of Clinical Cancer Genetics at the National Cancer Center in Singapore gave a talk on the role of extended testing for Cowden and Cowden like Syndrome in the Asian Context. Rabi Hanna, director of the Pediatric Bone Marrow Transplantation at Cleveland Clinic Children's Hospital, gave an update on the multi centre Clinical Trials, including the Everolimus trial. This trial is looking at how people with PHTS are at risk in developing cognitive and behavioural problems as well as autism. And to learn how the new drug affects symptoms. There was a discussion around the risk/reward ratio of taking drug with Rabi stating that safety is their main priority with very little toxicity. As of yet there is not enough evidence to show positive or negative effects of taking the drug.

Professor Pandolfi, discussed Pten reactivation for tumour prevention therapy. His studies found that cruciferous, which are green vegetables can keep Pten levels up, which would be beneficial to those affected by PHTS. Thom Frazier from Autism Speaks spoke about resources and support for people with Autism and Pten Mutations. Throughout the day I listened to leading experts discuss all things relating to PHTS. All the speakers came across as genuinely approachable people, committed to the care and support that our community needs. I feel that my trips to Boston have been a great gateway to meet people in the wider Pten community, increasing my understanding about our condition and leading me to have a sense of empowerment, awareness, hope and optimism for the future.



PTEN Research Foundation Latest news

PTEN Research charity (www.ptenresearch.org)

It has been an exciting and busy year for the Foundation. The 2017/2018 grant awards have been issued and we are delighted to be supporting a range of research projects. They include a project to better understand the clinical presentation of PHTS (Dr. Nicoline Hoogerbrugge and Dr. Janet Vos, Radboud University), the frequency of PTEN mutations in children with ASD and macrocephaly (Dr. Hande Çelebiler, Istanbul Bilim University), research to help identify new treatment opportunities for PHTS (Dr. Pier Paolo Pandolfi, Beth Israel Deaconess Medical Center) and the creation of a UK PHTS patient registry (Dr. Marc Tischkowitz and Dr. Katherine Lachlan).

The Foundation is very pleased to have set up a clinical development working group bringing together PHTS clinical specialists (doctors and geneticists) and patient representatives from across the world. This group is working together to identify the research priorities for PHTS needed to develop new treatments.

Internally we are building up our team and are delighted to welcome Dr. Paul Elvin as the Interim Director of Preclinical Research and Kirsty Stenhouse as the Administrative Officer. We are also in the process of updating our website to incorporate new content on PHTS, updates on our mission, strategy and current work, along with interviews with our grant recipients and partners. This will be available later this year. In the meantime please visit our website for more information on PTEN Research and our work.

Patient Registry update

Funding has recently been secured from the PTEN Research Foundation to create the UK's first PTEN Hamartoma Tumour Syndrome (PHTS) Patient Registry. This project will be led by clinicians at the Universities of Cambridge, Southampton and Edinburgh. The Registry aims to improve understanding of the PTEN gene mutation by analysing clinical data from study participants. The planned outcome of the Registry will be to enhance patient surveillance and improve the medical management for this condition. The project will compliment international studies the Universities already collaborate with. Study participation will be straightforward, and more details will be available soon. Recruitment for the study is expected to open later this year. The contacts for the study are Dr Marc Tischkowitz and Katie York (Cambridge) Dr Katherine Lachlan (Southampton) and Professor Robert Semple (Edinburgh). Further information on the project will be available at the PTENUKI patient day in November.

IMPORTANT DATE Annual Patient Day

- Saturday 16th November 2019
- 10am to 5pm
- Heron Tower, 29th Floor, 110 Bishopsgate, London, EC2N 4AY

[REGISTER NOW HERE](#)

Please let us know if you have any suggestions for the agenda!

