



At PTEN Research it is our goal to improve the lives of individuals affected by PHTS (PTEN Hamartoma Tumour Syndrome) and their families by funding research into better understanding of the condition, improving outcomes and supporting the development of potential future treatments.

Thank you for continuing to support us.

- The PTEN Research Team

If you would like to read more about us please visit our website or email us at [contact@ptenresearch.org](mailto:contact@ptenresearch.org)

## NEWS AND EVENTS

### PTEN Hamartoma Tumour Syndrome Awareness Day

We are publishing our Autumn newsletter on October 23rd which is PHTS awareness day and are marking this important date with other organisations who support individuals with PHTS and their families.

The date of October 23<sup>rd</sup> (10/23) was selected as it reflects the location of PTEN on chromosome 10 (10q23).

Our website lists several other organisations supporting the PHTS community and we encourage you to follow them on their social media accounts as they will publish information about their activities and about PHTS.

If you have PHTS, or are a family member of someone with PHTS, you can find more information on our website, including

- [Links to PHTS/PTEN patient organisations](#)
- [Links to ongoing PHTS studies, trials and registries](#)
- [Making a donation](#) or [fundraising](#) to support our work

[Find out more on our website](#)

### Biochemical Society PI3K-AKT-mTOR-PTEN pathway meeting

In September, the Foundation was honoured to sponsor and attend a conference organised by the Biochemical Society in Barcelona, focused on the PI3K-AKT-mTOR pathway. The meeting brought together researchers, patient representatives, and companies for three days of exciting science and discussion, which covered basic research into PI3K pathway signalling as well as how new knowledge can help optimise therapeutic approaches targeting this complex pathway. The Foundation team had the opportunity to interact with existing grant recipients and made new connections which we hope will lead to future collaborations. The Foundation team would like to thank the Biochemical Society for a great meeting.

### 2023 Scientific Advisory Board Meeting

In October, the Foundation held its annual Scientific Advisory Board (SAB) meeting by video conference. This annual meeting is critical to the Foundation as it provides expert feedback from a multidisciplinary team of researchers, clinicians and pharmaceutical industry professionals with expertise in PHTS and drug development. The SAB supports the Foundation by helping to prioritise strategic aims, projects and focus areas.

The SAB provided valuable scientific insight and was in alignment with the updated Foundation strategy and commended the team on progress made on research activities. The SAB members also play an important part in the Foundation's grant review process. The Foundation is very grateful to all our SAB members for their valuable contribution in shaping our work.

### Screen4Care Forum on Newborn Screening

The Foundation team attended the Screen4Care Forum on Newborn Screening in early October. Screen4Care is an EU funded consortium aiming to shorten the path to rare disease diagnosis via newborn genetic screening and digital technologies. The meeting aimed to develop principles for selection of genes for inclusion in the Screen4Care Newborn screening pilot programme. The Foundation is grateful to Screen4Care for the opportunity to contribute to this important work. The following day, the team also attended the EURORDIS (European Organisation for Rare Diseases) workshop on Overcoming Barriers to Rare Disease Diagnosis which provided important insights into the challenges faced by patients and potential solutions to help reduce these challenges.

[Click here to learn more about Screen4Care](#)

## OUR RESEARCH

At PTEN Research we fund projects with leading experts in the field of PHTS at institutions around the world. A condition of our funding is the transparent publication of the work to ensure that individuals and families affected by PHTS, as well as other physicians and scientists working in field are aware of the latest data. Further, our formal grant application and external review processes ensure we only fund work which we believe is of high scientific value and expected to provide benefit to the PHTS community.

If you would like to find out more about how we fund research, or apply for a research grant please [click here!](#)

## NEW PUBLICATIONS

### Assessment of social and cognitive performance in PHTS using webcam measures

Professor Tom Frazier from John Carroll University, Ohio, USA, together with several collaborators including Cleveland Clinic, Ohio, USA, and University Hospital Southampton, UK, have recently published the initial results of a study leading to the development and initial validation of a webcam performance measures with the goal of assessing the social and cognitive performance of individuals with PHTS and which has the potential to be extended to other neurodevelopmental conditions in the future. This publication builds on the work that Professor Frazier recently published describing a survey tool called the Neurobehavioral Evaluation Tool (NET) which we featured in our previous newsletter.

Work is still ongoing to continue validation of the webcam measure, together with the NET survey, and the study is looking for participants in the UK and US with a PHTS diagnosis aged 3-45, with a parent, other family member or caregiver to support them. No clinic visits or travel is needed but participants will need access to a reliable internet connection and be fluent in English (although not necessarily as the primary language).

For more information see [NCT05671107](#) or contact project coordinator Katie Huba [khuba@jcu.edu](mailto:khuba@jcu.edu).

[Click here to access the full publication](#)

### New studies using PTEN antibodies to help better understand the biology of PTEN

Professor Rafael Pulido and his research group at the Biobizkaia Health Research Institute in Spain have published two studies using new and existing PTEN antibodies to help improve understanding of PTEN biology. The first study focused on assessing unique features of novel PTEN variants identified in individuals with PHTS, and the second study developed a new antibody that recognises a different part of the PTEN protein compared to existing antibodies. This new research tool adds additional value to the collection of antibodies widely available to researchers today.

[Click here to access the full publication about novel PTEN variants](#)

[Click here to access the full publication about the novel antibody](#)

Thank you for your interest in and support of PTEN Research, and for taking the time to read our newsletter.

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