THE SUMMER ISSUE

THE LATEST FROM PTEN RESEARCH

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 our activities please visit our website or email us at contact@ptenresearch.org

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 publication of the work to ensure that individuals and families affected by PHTS, as well as other scientists, benefit from the work which we fund. Further, our formal application and external review processes ensure we only fund work which we believe is of high scientific value.

The Foundation is happy to announce that a research project has been recently initiated.

PHTS-associated PTEN variants and molecular mechanisms of disease

Although PHTS is caused by a germline mutation of the PTEN gene, the way in which different PTEN mutations disrupt cell function is not fully understood. Through its Targeted Call for Proposals, PTEN Research has awarded funding to Prof Kurt Haas at the University of British Columbia, Canada, to investigate how a large number of PHTS-associated PTEN variants impact molecular networks in cells. Building on previous work by Prof Haas, the current project aims to understand more about the molecular mechanisms underlying PHTS, which could help to identify future drug targets.

If you would like to find out more about how we fund research or apply for a research grant, please click here.

NEW PUBLICATIONS

Neurobehavioral Evaluation Tool (NET) study

Professor Tom Frazier from John Carroll University, Ohio, USA, together with several collaborators have recently published the initial results of a study describing the development and initial validation of an online survey tool called the Neurobehavioral Evaluation Tool (NET) which is being developed to assess neurodevelopmental disorders, including autism spectrum disorder, in people with genetic syndromes and specifically PHTS.

Existing measures have not been developed specifically with individual with PHTS in mind. Further, existing surveys for parents and caregivers often take a long time to complete and performance measures require travel to the clinic or hospital where expert clinical staff are needed to administer the tests. The objective of the NET is to overcome these challenges and it can be undertaken online at home. These new tools will be particularly important for future clinical trials assessing if new treatments are helping individuals with PHTS.

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.

For more information see NCT05671107 or contact project coordinator Katie Huba khuba@jcu.edu

Click here for more details on the publication in the American Journal of Medical Genetics

Genetic modifiers in PHTS

Professor Charis Eng and her team at Cleveland Clinic have reported the first results from a Foundation funded study investigating genetic modifiers in PHTS.

PHTS is caused by germline mutations in the PTEN gene, but the phenotype of the syndrome is highly variable, even between siblings who share the same PTEN mutation.

Therefore, it is likely that genomic and metabolomic factors may act as modifiers. Professor Eng and her team are investigating these genetic modifiers using whole genome sequencing in a large cohort of individuals with PHTS.

The recent publication from this study shows that the mitochondrial genome may modify the PHTS phenotype, as individuals with PHTS and autism spectrum disorders (ASD) or developmental delay (DD) (but not cancer) have significantly higher mitochondrial DNA copy number variant burden compared to individuals with PHTS and cancer (without ASD/DD).

Click here for more details on the publication in Human Genetics and Genomics Advances

NEWS AND EVENTS

International Society for the Study of Vascular Anomalies - Debates and Updates meeting

The Foundation attended the International Society for the Study of Vascular Anomalies (ISSVA) Debates and Updates meeting in Boston in April 2023.

As vascular anomalies are part of the symptoms some individuals with PHTS experience, the Foundation team found it very helpful to learn about the latest progress in the field. The meeting also gave an opportunity for the leadership team of the Foundation funded project aiming to better understand the vascular anomalies in PHTS to meet in person for the first time since the project was initiated during the pandemic.

The Foundation would like to thank ISSVA and all key stakeholders for a very successful and interactive meeting.

PTEN UK & Ireland Patient Day

The PTEN UK & Ireland Patient and Family Day took place on Saturday June 24th in Tubney, Oxford, in beautiful sunshine. The Foundation was honoured to attend to provide an update on our progress over the last year.

The Foundation would like to thank PTEN UKI and all participating families for a very successful day and for the opportunity to engage with the UK PTEN community.

GET INVOLVED

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

If you would like to make a donation to support our work please click here

PTEN RESEARCH

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