At PTEN Research it is our goal to improve the lives of individuals affected by PTEN Hamartoma Tumour Syndrome (PHTS) 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

NEWS AND EVENTS

PTEN Research Collaborators' Meeting

The 6th annual PTEN Research Collaborators' Meeting took place on 11 March, bringing together PHTS expert researchers and specialist clinicians to share progress from projects funded by PTEN Research and to discuss new ideas that could improve understanding and care for people with PHTS.

A key aim of the virtual meeting is to give researchers the opportunity to share early findings before they are formally published. This year's topics ranged from efforts to better understand vascular (blood vessel) changes seen in PHTS and using medical records to help improve diagnosis, to new research into the symptoms of the nervous system symptoms of the condition. There was also a presentation about plans for a large study that will screen existing medicines to see whether any could be repurposed to treat PHTS symptoms, potentially speeding up the path to new treatment options.

All the results from PTEN Research-funded projects will go on to be published in scientific journals, helping to ensure that knowledge is shared widely and benefits the whole PHTS community including researchers, clinicians and, most importantly, families and individuals with PHTS.

We are very grateful to everyone who presented and attended. Meetings like this help spark new ideas and collaborations, with the hopes of improved understanding of, and care for, PHTS.

Developmental Synaptopathies Consortium launch meeting

The PTEN Research team was excited to take part in the launch meeting of the Developmental Synaptopathies Consortium (DSC) in Boston in late March. This meeting marked the start of the third, and final, five-year phase of a major US National Institutes of Health (NIH) funded research programme, which includes work on PHTS alongside several related neurodevelopmental conditions.

There was a sense of momentum and optimism as researchers shared ideas and plans for the years ahead. A key part of this work is the PHTS natural history study, which has already been running for 10 years and will continue to recruit participants. This long-term study is helping clinicians and scientist build a clearer picture of how PHTS affects people over time.

The PTEN Research team is deeply grateful to every person and family who has already taken part. If you or a family member are interested in learning more about this study, and whether it might be right for you, you can find details on clinicaltrials.gov.

Society for the Study of Vascular Anomalies annual meeting

The PTEN Research team attended the annual International Society for the Study of Vascular Anomalies (ISSVA) annual meeting in Philadelphia this May. It was a valuable chance to meet collaborators in person, share updates on current and upcoming research and learn about the latest developments in the field.

One highlight for the team was the opportunity to present a poster on behalf of Dr Phil Ambery, a member of our Scientific Advisory Board who was unable to attend. The poster describes the case study of a woman with PHTS who has vascular anomalies (abnormal development of blood vessels and associated tissue) and showed improvement with a combination of a diabetes medication (called an SGLT-2 inhibitor) and intermittent fasting.

This is just a single case, so more research is needed to see if this approach could help others with PHTS. A review article discussing the potential of this approach in PHTS has been accepted by the peer-reviewed journal Metabologia and will be available online in due course.

You can see the poster here.

OUR RESEARCH

At PTEN Research, we fund projects with leading experts in the field of PHTS at institutions around the world. Our grant application and rigorous review processes ensure that we only fund work of high scientific value which is expected to benefit the PHTS community. A condition of our funding is that the work we support is published to allow individuals and families affected by PHTS, as well as other physicians and scientists working in field, to be aware of the latest developments and data.

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

NEW PUBLICATIONS

Case study of PHTS cancer treatment

PTEN Research is pleased to have provided support for a recent publication led by Dr Jordi Rodon at MD Anderson Cancer Center (Houston, USA), describing a promising treatment response in a person with PHTS and aggressive breast cancer.

The report describes a 34-year-old woman whose cancer was progressing quickly despite standard treatment. As part of a clinical trial (which has since closed) she received an investigational drug called TAS-117. This drug targets *AKT*, a protein that becomes abnormally activated in PHTS.

Encouragingly, since starting treatment in 2020 her cancer has remained stable and she has not experienced significant side effects. She continued treatment at the time the publication was submitted in November 2025.

Although more research is needed, this report together with a previous case report describing treatment of two women with PHTS associated breast cancer with another *AKT* inhibitor called capivasertib, add to the growing evidence that AKT inhibitors could be useful for treating cancers linked to PHTS.

PTEN Foundation's continued efforts to highlight the needs of the patient community and support clinical trial recruitment are recognized.

For more details see the full publication in BJC Reports.

Whole genome sequencing hints at role of many genes modifying PHTS symptoms

PTEN Research is proud to have supported a large study exploring why people with PHTS can experience such different symptoms, even within the same family. This suggests that other factors, including changes to other genes, may influence the presentation of PHTS.

In this study, researchers analysed the full genetic code (whole genome sequencing) of nearly 600 people with PHTS and their relatives.

This research, originally initiated by the late Professor Charis Eng from Cleveland Clinic (Ohio, USA), and completed by her team led by Drs Ying Ni and Lamis Yehia, identified several additional genes that may be linked to cancer risk and neurodevelopmental features seen in PHTS in addition to *PTEN*.

While these findings are an important step forward in understanding the variability of PHTS symptoms, more research is needed to understand how they might be used in clinical care in the future.

For more details see the full publication in NPJ Genomic Medicine.

Finding better treatments for PHTS neurobehavioral symptoms

PTEN Research has established a collaboration with Evotec, a leading drug discovery and development company, to explore whether existing medicines, already tested in humans for other conditions such as cancer, could be repurposed to treat the neurological symptoms of PHTS, including autism and learning difficulties. This collaboration has now resulted in the publication of a new study in the journal iScience.

Whilst a recent clinical trial of the mTOR inhibitor everolimus showed improvements in some aspects of neurocognitive function in people with PHTS, there remains a need for additional and better treatment options. To explore this, the team screened more than 60 compounds that target the PI3K/AKT/mTOR pathway, which is known to be overactive in PHTS, using a series of laboratory tests in cells and neurons. This is believed to be the largest drug repurposing effort to date for the neurological symptoms of PHTS.

Because these compounds have already undergone testing in the laboratory or clinical trials, or may even have health authority approval for other conditions, identifying possible candidates that may have activity in PHTS could shorten the path to clinical trials for people with PHTS, and this study paves the way for further investigation of the most promising treatments.

For more information, see the full publication in iScience.

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 ongoing PHTS studies, trials and registries
- Links to PHTS patient organisations around the world
- Learn more about our research, funding, and making a donation

PTEN RESEARCH
Funding research to support people with PHTS

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