THE SPRING ISSUE

THE LATEST FROM PTEN RESEARCH

PTEN RESEARCH

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

March 18 marked the fourth PTEN Research Collaborators' Meeting, a virtual event bringing together PHTS researchers and expert clinicians from around the world to share and discuss the latest updates from PTEN Research-funded projects. This year was our largest event to date with 80 attendees.

The goal of the meeting is to advance the mission of PTEN Research by giving our grantees and collaborators a forum to share new data with the PHTS research community, with a view to accelerate research, forge new partnerships and expand the PHTS research network.

Our hope is that sharing new and interim data amongst the PHTS research community will not only boost innovation but will also accelerate research. With that said, results from all PTEN Research funded projects must also be formally published in scientific journals to ensure the resulting data benefits the entire PHTS community including researchers, clinicians and, most importantly, patients and their families.

Thank you to all the attendees and especially those who shared their latest findings and whose expertise helps drive the field forward.

PHTS Vascular Anomalies Project Meeting

During the recent International Society for the Study of Vascular Anomalies (ISSVA) conference in Paris, PTEN Research hosted a satellite meeting for expert physicians as part of a project to better characterise the vascular anomalies (VAs) that are seen in PHTS.

This international collaboration led by Professor Denise Adams of Children's Hospital of Philadelphia and funded by PTEN Research included representatives from hospitals in the US, Netherlands, Belgium, Spain and France.

Vascular anomalies involve abnormal development and growth of blood vessels and other associated tissue. It is estimated that between a third and half of individuals with PHTS have one or more vascular anomaly. The severity of symptoms varies but can cause significant pain and other problems. The work to better characterise how vascular anomalies in PHTS differ from those related to other conditions is important to support future clinical trials of treatments.

PTEN Research is very grateful to all who took time out of their busy conference schedule to attend the meeting.

For more information see our LinkedIn post.

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.

Targeted Call in Honour of Professor Charis Eng

PTEN Research were proud to have presented at the scientific symposium honouring the life and legacy of Professor Charis Eng held at Cleveland Clinic on 31 March 2025.

Professor Eng was a true leader and pioneer in the field of PHTS and cared deeply for the people with PHTS that she treated. Professor Eng also played a pivotal role in shaping the scientific strategy of PTEN Research and was always generous with her time and wise counsel.

To honour the work and legacy of Professor Eng, PTEN Research plan to launch later in 2025 a targeted call in her name. A targeted call is a competitive funding mechanism inviting proposals from scientists to address a specific research question.

The scope of the call will be to build on previous work led by the Eng laboratory to characterise how different changes in the *PTEN* gene lead to a range of symptoms in PHTS and explore potential markers in blood or other tissues that could, in the future, support better ways of identifying or treating individuals with PHTS.

Researchers who are interested to find out more about this upcoming targeted call are encouraged to visit our website and may pre-register an interest at research@ptenresearch.org.

New Grant Exploring the Use of Medical Record Information to Improve Diagnosis of PHTS

We are pleased to announce a new collaborative research grant funded by PTEN Research and led by Professor Mustafa Sahin and Dr Siddharth Srivastava from Boston Children's Hospital, and Dr Paul Avillach from Harvard Medical School, working with colleagues at the Dana Farber Cancer Institute.

The goal of this study is to build an electronic algorithm to analyse medical record data to identify adults who have symptoms that are associated with PHTS detailed in their medical records but have not yet been diagnosed.

If successful, this approach could lead to the development of programs that, in the future, help identify individuals who may benefit from discussing with their doctor whether genetic testing for PHTS is right for them.

This study builds on the success of previous work by the same team, which successfully developed a similar approach for children.

Getting a timely diagnosis of PHTS is important because it can allow people to begin cancer surveillance that may detect cancer early, or even before it starts. Early cancer diagnosis has been shown to improve treatment outcomes.

In the past, getting a PHTS diagnosis has not always been timely. This is partly because some of the signs of PHTS, like non-cancerous breast lumps or skin growths, are also common in people without PHTS. In addition, many doctors are not familiar with the condition.

NEW PUBLICATIONS

New Research Provides New Information About the Prevalence of PHTS

PHTS is a rare disease, but very little information has been published about its prevalence, i.e., how many people have PHTS. A recent publication from Dr Nikita Pozdeyev and colleagues from the University of Colorado Anschutz Medical Campus reported that between 1 in 9,000 and 1 in 13,000 individuals have the types of changes in the *PTEN* gene that could lead to PHTS. This result was observed when studying the genetic information from over 700,000 individuals who have volunteered to take part in research through two large-initiatives in the US and UK, called the All of Us Research Program and the UK Biobank, respectively.

The results of the study suggest that PHTS is more common than reported by previously published data. The researchers only investigated the genetic changes in the *PTEN* gene, but did not investigate what symptoms of PHTS the identified individuals have, beyond noting that the number of individuals with thyroid cancer and a change in the *PTEN* gene was relatively low, as this was the main focus of the study.

For more details see the full publication in the Journal of Clinical Endocrinology & Metabolism.

New Research Sheds Light on How Vascular Anomalies Develop in PHTS

PTEN Research is proud to support the work of Professor Mariona Graupera (Josep Carreras Leukaemia Research Institute, Barcelona) and Dr Sandra Castillo (now at the Institut de Recerca Sant Joan de Déu, Barcelona), who are helping us better understand why some people with PHTS develop vascular anomalies.

Vascular anomalies affect around 30-50% of people with PHTS, often starting in early childhood, but the exact cause of these changes in blood vessels isn't fully understood. In their newly published study in Cancer Discovery, the researchers identified a specific genetic change in the cells that line blood vessels (called endothelial cells), which results in loss of the normal copy of the *PTEN* gene and abnormal blood vessel growth.

What's especially exciting is that the team confirmed their findings using real patient samples and created the first laboratory model of these vascular issues in PHTS. This model not only helps us understand how vascular anomalies form but also allows scientists to test potential treatments.

This discovery brings us closer to improved diagnosis, better treatment options, and ultimately, improved quality of life for children and adults living with PHTS.

For more details see the full publication in Cancer Discovery.

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
- Making a donation or fundraising in aid of our work

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

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