

THE SUMMER ISSUE

THE LATEST FROM 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 UK and Ireland patient day

The PTEN Research team attended the [PTEN UK and Ireland](#) patient day on 29 June. It was great to meet families and expert clinicians and hear the latest developments for people with PHTS and their families within UK and Ireland. The PTEN Research team was pleased to make some new connections which we hope in the future may lead to new studies and will ultimately improve the quality of life for people with PHTS and their families.

Thank you to PTEN UK and Ireland for the invitation to attend the day and for continued positive collaboration. Most of all, we want to thank all the families who shared their thoughts and questions throughout the day.



OUR RESEARCH

At PTEN Research, we fund projects with leading experts in the field of PHTS at hospitals and universities 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 the 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](#).

Two new studies funded by PTEN Research have recently started with the aim to find out why and how unusual growth of blood vessels and related tissues, known as vascular anomalies, develop in people with PHTS. It is estimated that between a third and half of individuals with PHTS have one or more vascular anomaly. Improving care for vascular anomalies in PHTS is of key interest to PTEN Research as they can cause significant pain and other problems.

Study to better understand PHTS associated vascular anomalies

PTEN Research is pleased to announce the start of a new study led by Professor Michael Hildebrand and colleagues at The University of Melbourne. This study will assess tissue samples from 42 individuals with PHTS and vascular anomalies or other types of overgrowth.

Researchers at the University of Melbourne are studying whether extra genetic changes in the affected blood vessel tissue, known as "second hits," play a role in causing vascular anomalies in people with PHTS. In PHTS, changes in a gene called PTEN are present in every cell of the body. However, the blood vessel problems only show up in some people with PHTS and then only in certain tissues. This suggests they might be triggered by an additional, local genetic change - either in the second PTEN gene copy or in another gene in that tissue.

The findings from this research could lead more personalised treatment plans, and potentially new treatment options in the future.

Study of the onset and progression of vascular anomalies in PHTS

PTEN Research is pleased to announce the start of a new study led by Dr. Mariona Graupera at the Institut Josep Carreras (IJC) in Barcelona, in collaboration with Dr. Sandra Castillo, at the Institut de Recerca Sant Joan de Déu (IRSJD). People with PHTS often develop different types of vascular anomalies. Previous research from Dr. Graupera's team, also funded by PTEN Research, suggests that these vascular anomalies may have distinct origins and patterns of progression, highlighting the need to better understand their development.

This study aims to fill an important gap in knowledge by looking closely at the different types of blood vessel cells and how they may contribute to the development of vascular problems in people with PHTS. The researchers will also study how outside factors (like the environment) might influence when these problems appear, and explore for the first time how vascular problems and tumour growth may be linked.

As part of the project, the team will create a patient registry together with Dr. Eulalia Baselga, a leading doctor in vascular anomalies. This registry will help gather and study tissue samples and allow researchers to test their findings using cells from people with PHTS.

The results could open up new directions for research and support the development and early testing of possible new treatments. In the long term, this work aims to improve care and quality of life for people living with PHTS.

Acceptance of Neurobehavioural Evaluation Tool (NET) into competitive FDA programme

We are excited to share that the Neurobehavioural Evaluation Tool (NET) and webcam measures developed by Professor Tom Frazier and his team at John Carroll University (with funding from PTEN Research) have been accepted into an evaluation programme with the US health regulator the Food and Drug Administration (FDA). A big thank you to all the families who have participated in the now completed study of the NET and webcam measures. Without you this would not have been possible.

Only 13 projects will be included in the 3-year life of the programme, so this is an important achievement.

The NET and webcam measures are designed to assess the key behaviours and thinking skills that are especially relevant for people with PHTS. All assessments are undertaken from home on a regular computer, laptop, or tablet. Compared to existing approaches this is faster, easier to use and more specific to PHTS. It also does not require in-clinic assessment with a healthcare professional, reducing the need for clinic visits at specialist centres with specially trained staff.

Being accepted into the FDA's programme means that Professor Frazier's team can meet with FDA experts up to four times to get advice on how to further develop and test the NET so that it can eventually be used in future clinical trials.

In addition to supporting clinical trials, the NET could also make routine clinical assessments more useful, by giving doctors reliable information gathered at home.

Finally, the FDA selection also helps raise awareness about PHTS at a major regulatory level.

Targeted Call for a new grant launching today

To date, PTEN Research has committed £25 million to projects at institutions across three continents, supporting work that ranges from basic science aimed at understanding the molecular mechanisms of PHTS through to clinical studies. Our funding is currently supporting 23 active projects, and our work has been acknowledged in 63 peer-reviewed publications.

PTEN Research is now pleased to announce that the Targeted Call for a new grant honouring the life, work and legacy of Prof Charis Eng will open for submission of project proposals on 2nd September 2025.

To build on the work of Prof. Charis Eng, we invite proposals from hospital and universities to investigate markers (in blood or other tissues) that improve diagnosis, prognosis, or treatment decisions, and to explore how different changes in the PTEN gene affect people with PHTS, helping to explain the wide range of symptoms seen in the condition.

This call is open to both preclinical and clinical research proposals. All applications must clearly show how the research could lead to future clinical benefits for individuals living with PHTS.

The deadline for submitting Project Proposals is **25 November 2025**.

More information for researchers on eligibility and how to apply is available on our website

NEW PUBLICATIONS

Results of a clinical trial of sirolimus for PHTS associated colon polyposis

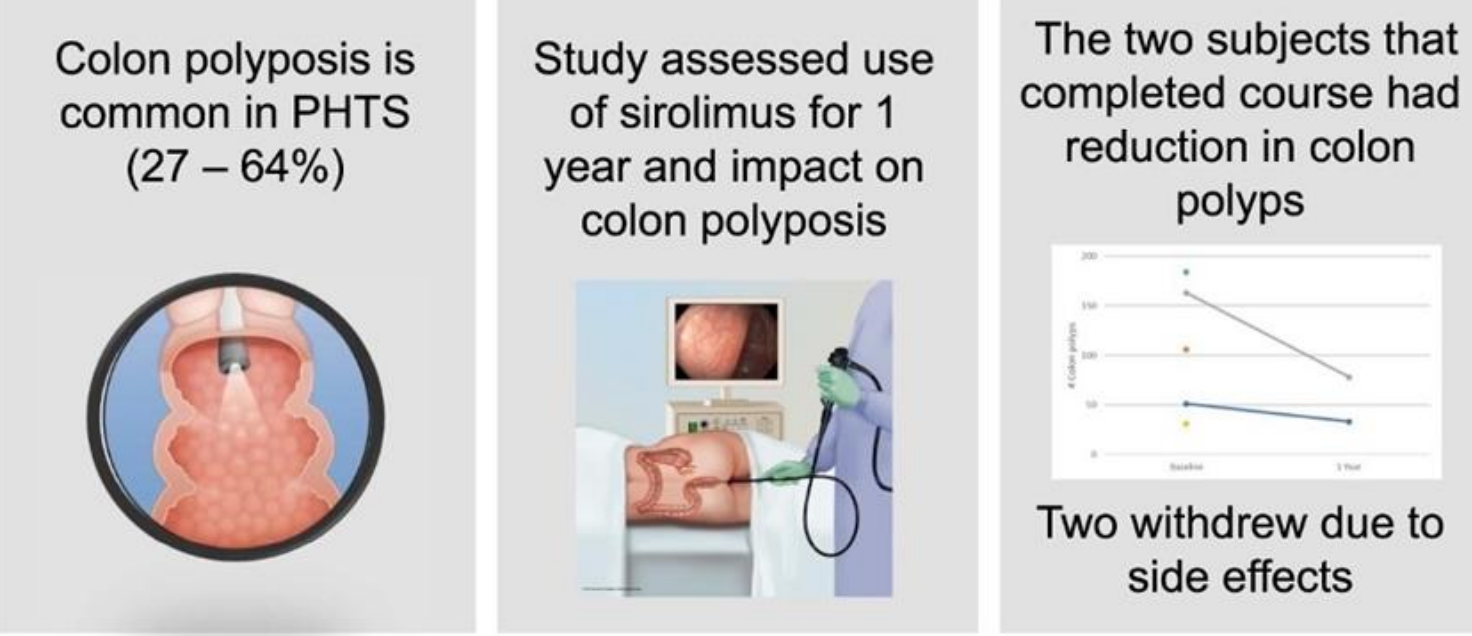
The results of the PTEN Research Foundation funded clinical trial assessing the effects of sirolimus on colon polyposis in PHTS have been published. This trial was led by Dr Peter Stanich from The Ohio State University.

Colon polyposis refers to the presence of multiple small, abnormal growths in the bowel which are common in people with PHTS.

While most polyps don't cause symptoms, it is important to monitor and remove them due to the risk of developing colorectal cancer. For individuals with PHTS this may be challenging due to the large number of polyps.

The study involved treatment of individuals with PHTS and many colon polyps for up to a year with the drug sirolimus to see if the number of polyps could be reduced by the treatment. Five individuals took part, with two completing the study and three withdrawing due to health issues (two withdrawals were likely related to the medication). The study suggests that sirolimus could help reduce the number of colon polyps in individuals with PHTS, but side effects may limit usage, and larger studies are needed to see if the results can be repeated.

[Sirolimus for Colon Polyposis in PTEN Hamartoma Tumor Syndrome (PHTS)]



Stanich et al. *Clin Trans Gastroenterol*. 2025. doi:10.14309/ctg.00000000000000871
All icons above are from Clinical and Translational Gastroenterology. [CLINICAL AND TRANSLATIONAL GASTROENTEROLOGY](#)

For more details, see the full publication in Clinical and Translational Gastroenterology

Cancer prognosis and treatment results in people with PHTS

The results of a large European collaborative study, funded in part by PTEN Research Foundation and led by Prof Nicoline Hoogerbrugge from Radboud University Medical Center in the Netherlands, have been published. The study looked at cancer outcomes in 513 people with PHTS from 30 medical centres in 16 European countries.

Encouragingly, people with PHTS who develop cancer usually do just as well as those in the general population, and the data showed standard treatments work effectively. Just like for everyone else, outcomes are better when cancer is found early - which is why timely diagnosis of PHTS and access to regular cancer screening is so important.

For more details, see the full publication in BJC Reports

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](#) 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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