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

Rare Disease Day

We are pleased to share our latest newsletter in celebration of Rare Disease Day on 28 February.

PTEN Hamartoma Tumour Syndrome (PHTS) is a rare genetic condition, caused by genetic changes in the *PTEN* gene. The exact number of people affected is still unknown. Older studies suggest that a related condition called Cowden syndrome may occur in as few as 1 in 200,000 people. However, a new study published in 2025 by researchers at the University of Colorado looked at genetic information from two research cohorts consisting of more than 700,000 people and found the types of changes in the *PTEN* gene that cause PHTS in about 1 in 9,000 to 1 in 13,000 individuals. This means that PHTS may be 10-20 times more common than previously thought. It is important to note that we do not yet know whether these individuals experienced any symptoms or have a diagnosis of PHTS and understanding this better will be an important focus for future research.

These new results mean that more individuals and families may be living with PHTS without knowing it. PTEN Research is funding several projects aimed at improving diagnosis so that people receive the right medical care, particularly access to cancer surveillance that has been shown to identify cancers early when treatments are known to be more effective.

While there are currently no health authority-approved treatments for PHTS, we are committed to supporting research that will lead to new and better treatments. Our work would not be possible without our wide range of stakeholders, including academic researchers, expert clinicians, industry collaborators and, most importantly, the people and families living with PHTS. We want to extend a huge thank you for your support of, and interest in, our work.

To find out more, our website includes information for families living with PHTS, including:

- Information about PHTS and the PTEN gene
- Information about the research we fund
- Links to PHTS patient organisations across the world
- Links to ongoing research seeking participants with PHTS

Dr Phil Ambery joins the PTEN Research Scientific Advisory Board

We are delighted to announce that Dr Phil Ambery has joined the PTEN Research Scientific Advisory Board.

The Board consists of a group of international experts in the fields of PHTS, genetics and drug development.

Dr Phil Ambery is a Global Clinical Head within the late Cardiovascular, Metabolic and Renal research group in AstraZeneca Gothenburg and a Consultant Physician with Cambridge University Hospitals. As well as being medically trained, he holds an eMBA from University of Gothenburg. Over nearly 25 years Phil has led medical affairs and clinical development teams across multiple projects, first in GlaxoSmithKline and for the past 11 years in AstraZeneca. He has a particular interest in obesity and associated comorbidities and has developed both incretin agonists and SGLT-2 inhibitor combination products in recent times. A particular interest of Phil's is how changing the metabolic profile in patients with cell growth disorders can alter mTOR activation and how this can positively impact both complications of weight gain and cancer risk.

PTEN Research Foundation annual update

To date, PTEN Research Foundation has committed over £25 million to research that supports our mission to develop better treatments and improve outcomes for people living with PHTS.

We also continue to make progress with testing existing medicines to see whether any can be repurposed to treat PHTS. We hope to announce details of new clinical trial(s) soon.

In 2025, our Trustee Board approved funding for seven new research grants. We received nine proposals through our Innovation Awards, several of which are still under review. In addition, we launched a new Targeted Call in memory of the late Professor Charis Eng, which seeks to honour Professor Eng's remarkable contributions to people with PHTS by investigating potentials biomarkers that are relevant for PHTS. (A biomarker is a measurable sign in the body, like a protein, gene, or blood test result, that gives information about a normal process, a disease, or how well a treatment is working.)

Fourteen project proposals from researchers across three continents were received and the final project selection for this Targeted Call will be made in the last quarter of 2026.

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 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.

Project to detect early clues of cancer

PTEN Research is pleased to announce the initiation of a new grant led by Dr Raheleh Rahbari at the Wellcome Sanger Institute and Dr Joseph Christopher at the University of Cambridge. The project aims to study genetic changes that could help scientists better understand early cancer development in PHTS.

People with PHTS have a higher risk of developing certain types of cancer, but not necessarily others. To improve outcomes, it is important to detect cancer at its earliest stages when treatments are known to be more effective.

Recent advances in technology allow the research team to detect very early genetic changes linked to cancer, even in normal, healthy tissues. Research has shown that small patches of cells with genetic changes are common throughout the body, and many of these genetic changes are also found in advanced cancers. However, it remains unclear why certain tissues in individuals with PHTS are more prone to cancer while other tissues remain unaffected.

This study will use cutting-edge technology to analyse these early genetic changes in normal tissues of individuals with PHTS to help understand any patterns that lead to increased cancer risk in certain individuals and tissues.

Understanding brain changes in PHTS to guide better treatments

PTEN Research is pleased to announce the start of a new grant led by Dr Yun Li at The Hospital for Sick Children in Toronto, funded through our 2024 Targeted Call.

This project aims to understand how changes in the *PTEN* gene affect brain development and function in people with PHTS. Some people with PHTS experience neurological challenges such as learning difficulties, autism or seizures.

Whilst existing research has provided valuable clues, effective treatments for these brain-related symptoms are still lacking, largely because we do not yet fully understand how *PTEN* changes specifically affect human brain cells.

To address this question, Dr Li's team will use advanced stem cell models created from blood or skin samples donated by volunteers. In the laboratory, these stem cells will be turned into different types of brain cells carrying *PTEN* changes. This will allow researchers to examine how these cells interact and how communication between them is disrupted in PHTS.

The study will also investigate specific pathways that become overactive in PHTS, with the goal of identifying possible future treatment approaches.

New grant to support UK national PHTS multidisciplinary team

PTEN Research is pleased to announce a new grant awarded to Dr Katherine Lachlan of University Hospital Southampton NHS Trust (UHS), to develop a coordinated national framework for PHTS care within England and the wider UK as part of the National Health Service (NHS).

Dr Lachlan is also the medical trustee for PTEN UK and Ireland.

Funding will support the newly established PHTS national virtual multidisciplinary team (MDT). The key activities of the MDT include review of clinical cases and care recommendations by a group of physicians experienced in managing the diverse range of manifestations experienced by people with PHTS. The MDT will also support standardisation of care through sharing guidelines and best practice. Longer term, it is hoped that the MDT will help support future PHTS research in the UK.

The grant will also support development of a patient-facing digital tool to support family education and PHTS specific screening compliance. A secure clinical database will also be developed to better record and understand the problems people with PHTS experience and to support future research.

The project will work closely in collaboration with the UK PHTS Patient Registry led by Prof Marc Tischkowitz (University of Cambridge). More information for individuals and families who would like to participate in the registry can be found at https://www.phts.org.uk/.

Completion of grant to assess the prognosis of cancer patients with PHTS

A PTEN Research Foundation-funded project led by Prof Nicoline Hoogerbrugge at Radboud University Medical Centre has recently been completed. The study aimed to better understand cancer outcomes in people with PHTS.

This large, collaborative effort brought together over 20 European centres with PHTS expertise and resulted in eight peer-reviewed publications (for a full list of publications, see our website). By studying the treatment and outcomes of hundreds of individuals with PHTS who had developed cancer, the research team was able to gather valuable insights into how cancers progress in this group.

One key finding was that the prognosis for cancers linked to PHTS was similar to that for cancers not related to PHTS. The study also found that a healthy lifestyle such as being physically active, avoiding smoking, and limiting alcohol is associated with a reduced breast cancer risk in people with PHTS, which is comparable to findings in the general population.

NEW PUBLICATIONS

Management recommendations published for treatment of the neurodevelopmental and neurologic symptoms of PHTS

New recommendations for managing neurodevelopmental and neurologic symptoms in people with PHTS have recently been published. PTEN Research is proud to have provided funding support for this important work.

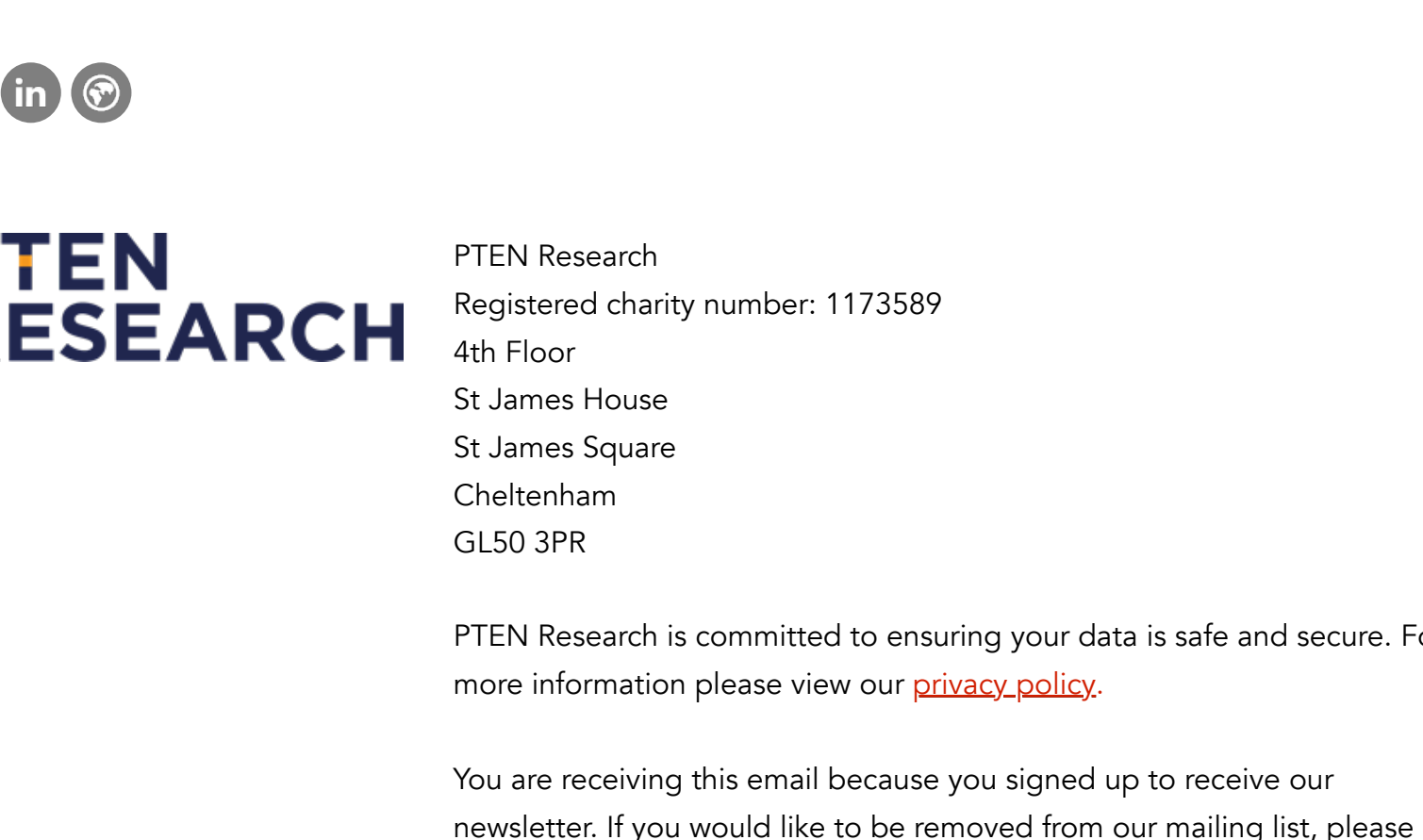
The recommendations were developed through a collaborative effort involving expert clinicians and patient advocates, led by the late Professor Charis Eng at Cleveland Clinic. The recommendations are based on a comprehensive review of the existing scientific literature, with input from an independent panel of multidisciplinary experts who helped shape the final consensus. These new guidelines aim to improve care for individuals and families living with PHTS. More information about these guidelines from Dr Andrew Dhawan, staff neuro-oncologist at Cleveland Clinic and the first author of the guidelines here.

For more information see the full publication in Neurology Genetics

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 to support our work



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