



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

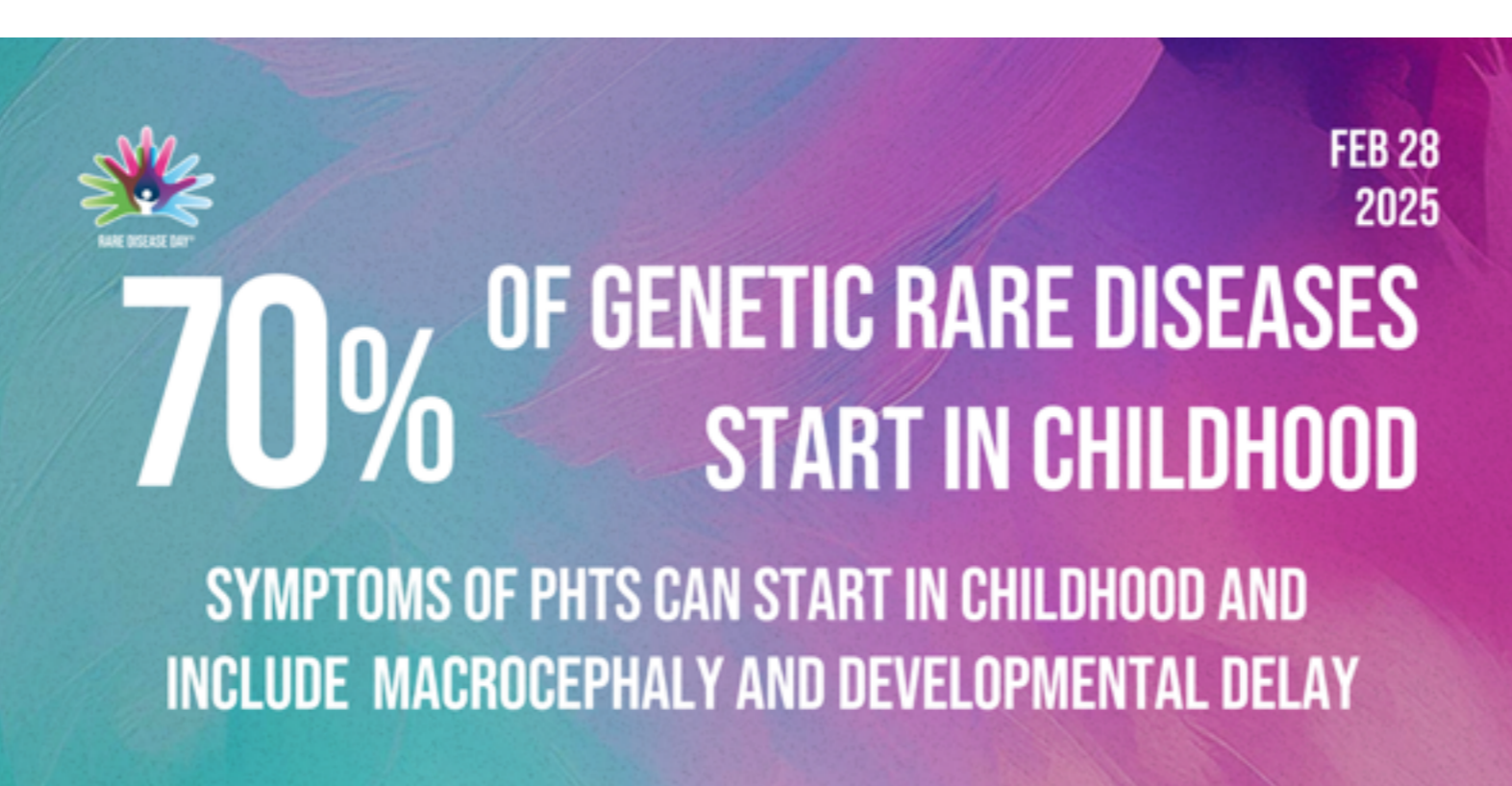
We are publishing our latest newsletter on 28 February to celebrate Rare Disease Day.

PHTS is a rare disease, although the exact prevalence is not known. Older studies suggest the disease is as rare as 1 in 200,000 individuals, but newer data points to it being less rare. More data is needed to refine these estimates, which we hope will be published in the future.

There are currently no health authority-approved treatments for PHTS. As part of our mission, we strive to raise awareness of PHTS, fund research that will lead to new and better treatments for the condition and bring together experts in the field. Our work would not be possible without our wide range of stakeholders, including academic researchers, expert clinicians, industry collaborators and, most importantly, the individuals 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](#)



PTEN Research Foundation update

To date, PTEN Research Foundation has committed over £23M to research in support of our mission to develop better treatments and improve outcomes for individuals with PHTS.

We also continue to make progress in the testing of existing drugs with the goal of repurposing them for the treatment of PHTS and we hope to be able to announce details of new clinical trial(s) soon.

In 2024, the PTEN Research Trustee Board approved funding for three new grants. A total of 9 project proposals and grant applications were received as part of our Innovation Awards, of which several are in the process of being reviewed in early 2025. In addition, a new Targeted Call was launched in 2024, receiving 13 project proposals from researchers in 3 continents. The goal of the call is to better understand the biology of PHTS by using the valuable resource of induced pluripotent stem cells (iPSCs) generated from sample donations from families with PHTS. The iPSCs were created as part of an earlier grant from PTEN Research to Boston Children's Hospital. The final project selection for the Targeted Call will be made during the first quarter of 2025.

In 2024 we also launched the [PHTS research tools](#) on our website. The purpose of this resource is to collate information about useful tools for PHTS researchers, such as information about clinical trials, patient registries and laboratory models, in one location which is easy to find and navigate. We hope this resource will stimulate new ideas and collaborations and accelerate future research into PHTS. We continue to update this resource as new additions become available.

20 active projects from basic research through to clinical trials

Current grants with 14 leading academic institutions in Europe, North America and Australia	2 preclinical partnerships with laboratory contract partners	Active interactions with multiple pharma and biotech collaborators
54 acknowledgments in scientific papers	3 multidisciplinary working groups of international experts	Multiple PHTS-specific laboratory models established in academic and partner laboratories
		2 sets of PHTS-specific clinical trial endpoints in development
		PHTS-specific international classification of disease codes assigned in 2022

Completion of grant to develop new outcome measures for PHTS

The PTEN Research Foundation funded grant to develop and validate new outcome measures for PHTS, led by Prof Tom Frazier at John Carroll University has been successfully completed. This work developed the neurobehavioral tool (or NET) as a brief, freely available set of survey scales that evaluate key behaviours and thinking skills relevant for people with PHTS. The tool also includes the ability to collect information on an individual's behaviour and thinking skills through a webcam system that can be completed at home on a standard computer, laptop or tablet. These two measures could in the future be used to improve research, including clinical trials for PHTS, as they take less time to complete compared to currently existing measures. The ability to complete the measures from home could reduce the need for individuals to travel long distances when taking part in research. Having reliable and accurate online measures could also enhance clinical visits by providing supplemental information to healthcare professionals.

We want to thank individuals and families who generously contributed to this research both in the US and UK by participating in the development of the scales and taking part in the research study to test how well these new tools work. These tools could not have been developed without your contribution!

The work has resulted in four publications describing the development and validation of the survey and webcam tools:

- [Development of informant-report neurobehavioral survey scales for PTEN hamartoma tumor syndrome and related neurodevelopmental genetic syndromes](#)
- [Development of webcam-collected and artificial-intelligence-derived social and cognitive performance measures for neurodevelopmental genetic syndromes](#)
- [Quantifying neurobehavioral profiles across neurodevelopmental genetic syndromes and idiopathic neurodevelopmental disorders](#)
- [Remote monitoring of social attention in neurogenetic syndromes and idiopathic neurodevelopmental disability](#)

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

International recommendations published for the management of cancer and overgrowth in PHTS

New international recommendations for the management of cancer and overgrowth in PHTS have recently been published. PTEN Research is proud to have provided funding support for this important effort.

The recommendations were developed collaboratively by an international team of 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. There was also input from an independent external panel to establish the final consensus recommendations.

These guidelines will support improved care for individuals and families living with PHTS, aligned with our mission to fund and facilitate research that will lead to better treatments and improved outcomes in PHTS.

For more information see the full publication in Clinical Cancer Research

A new non-human model for neurological symptoms of PHTS provides insights into therapeutic approaches

As a result of a grant funded by PTEN Research, Professor Mustafa Sahin and his team at Boston Children's Hospital have published their work on the development of a new model that is focused on the neurobehavioral features of PHTS.

Findings from the study showed that whilst the drug everolimus restored some of the functioning of affected brain cells (neurons), other biological mechanisms are also important in this model of PHTS.

Building on this research, the team is now expanding their work with support from a new PTEN Research grant to explore another potential drug target which may one day be a drug treatment for neurobehavioral features of PHTS.

For more information see the full publication in Molecular Psychiatry (paywall)

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



PTEN Research
Registered charity number: 1173589
4th Floor
St James House
St James Square
Cheltenham
GL50 3PR

PTEN Research is committed to ensuring your data is safe and secure. For more information please view our [privacy policy](#).

You are receiving this email because you signed up to receive our newsletter. If you would like to be removed from our mailing list, please click the 'unsubscribe' link below.

[Unsubscribe](#)