



## RARE DISEASE DAY ISSUE

THE LATEST FROM PTEN RESEARCH

PTEN RESEARCH

At PTEN Research it is our goal to improve the lives of individuals affected by PHTS (PTEN Hamartoma Tumour Syndrome) 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 us please visit our website or email us at [contact@ptenresearch.org](mailto:contact@ptenresearch.org)

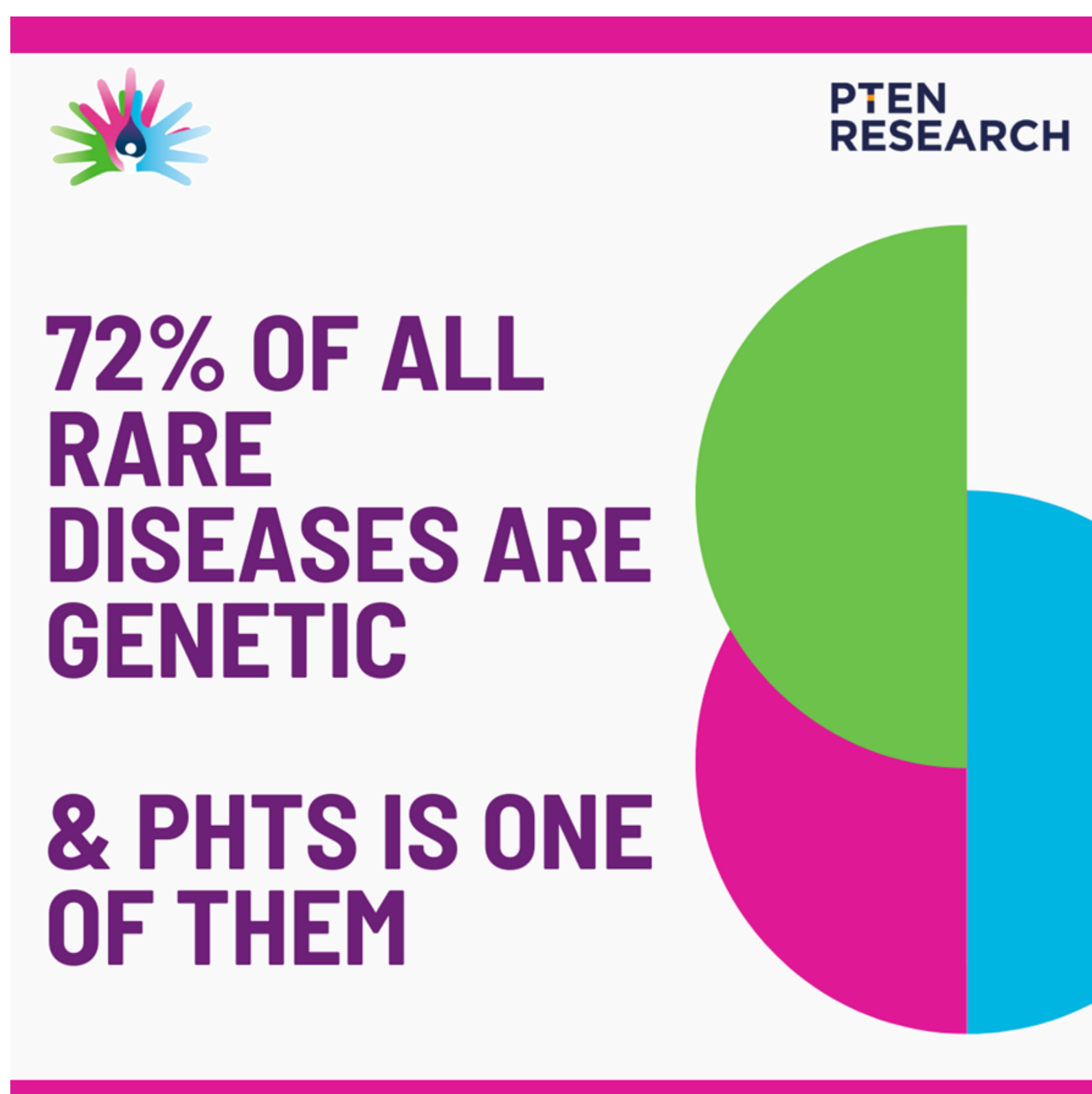
## NEWS AND EVENTS

### Rare Disease day

We are publishing the latest edition of our newsletter on Rare Disease Day, February 29<sup>th</sup>, the rarest of days.

As part of our mission, we strive to raise awareness of PHTS as well as funding research that will lead to new and better treatments for the condition. This would not be possible without our wide range of stakeholders including academic researchers, experts in the field, 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.

For more information about PHTS [see our website](#).



### PTEN Research Foundation update

Last year, the Foundation's Trustees approved funding for four new grants, that range from basic research to clinical studies, from a total of 14 project proposals and grant applications submitted to the Foundation. In addition, two grants funded via our first ever targeted call were initiated during 2023. We also continue to make significant progress in the testing of existing drugs with the goal of repurposing them for the treatment of PHTS. To date, PTEN Research Foundation has committed over £19.5M to research to support its mission of developing better treatments and improved outcomes for PHTS patients.

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		
44 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

## 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 provide benefit to 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

**At the Society for Neuroscience annual meeting in Washington DC in November 2023, data from two PTEN Research Foundation projects were presented.**

### Development of a new mouse model of PTEN loss in the brain

The Boston Children's Hospital team led by Professor Mustafa Sahin presented their work on the successful generation of a new and innovative mouse model for PTEN loss in the brain. It is hoped that the model will help better understand the fundamental biology of PHTS and support laboratory development and testing of potential new treatments in the future.

### Development of cell-based models of PHTS

PTEN Research Foundation presented work developed in collaboration with BridgeBio Pharma and our laboratory research partner Evotec.

The work explored several cell-based models of PHTS. These models, which can be used to support drug development efforts, are currently being used by the Foundation as part of our [drug repurposing project](#). The poster can be viewed on [our website](#).

**Elsewhere in the scientific literature, a recent publication investigated a new biomarker in individuals with PHTS and multiple cancers.**

### Cell-free DNA in individuals with PHTS and a second cancer

Professor Charis Eng and her team at Cleveland Clinic have investigated the role of cell-free DNA as a marker in PHTS cancer. Cell-free DNA are small pieces of DNA which are released from cells, including cancer cells, into the blood stream. By sequencing these small DNA pieces, Professor Eng's team showed that the profile of cell-free DNA could potentially, with further prospective evaluation and validation, be used to predict the risk for a second primary cancer in individuals with PHTS. This work was partly funded by the PTEN Research Young Investigator Award.

You can find the full publication in [Cell Reports Medicine](#).

## NEW PROJECTS

Building on the new mouse model of PTEN loss in the brain developed by the Boston Children's Hospital Group and presented at the Society of Neuroscience annual meeting in 2023, Professor Mustafa Sahin has now been awarded additional funding by PTEN Research to further utilise this model for a better understanding of the biological mechanisms underlying PHTS.

This work will combine multiple approaches to evaluate a class of drugs called FLT3 inhibitors to assess their potential future use a treatment of the neurological manifestations of PHTS.

## 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/PTEN patient organisations](#) around the world
- [Making a donation](#) or [fundraising](#) to support our work



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