



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

NEWS AND EVENTS

In memory of Charis Eng

The whole of PTEN Research Foundation were greatly saddened to hear of the recent death of Charis Eng.

Prof Eng was a true leader and pioneer in the field of PHTS. She was a key contributor to the first publication that identified *PTEN* mutations in individuals with Cowden Syndrome and showed that the *PTEN* protein acts as a tumour suppressor. She was also well known to many PHTS families all over the world via her clinical work at the Cleveland Clinic PTEN Multidisciplinary Clinic.

Prof Eng also played a pivotal role in shaping the scientific strategy of PTEN Research and was always generous with her time and wise counsel. She acted as chair of our scientific advisory board from 2019-2022 and we were privileged to continue to actively collaborate with her up to her death.

Speaking for the whole PHTS community, Prof Eng will be greatly missed. Our thoughts are with her family, friends, and colleagues across the globe.

2024 Scientific Advisory Board Meeting

On 3rd October, PTEN Research held its annual Scientific Advisory Board (SAB) meeting.

The SAB has a critical role in steering the Foundation's scientific strategy and we continue to be privileged that senior leaders from pharma and biotech, as well as academic physicians and scientists with a deep knowledge of PHTS, have been willing to join the Board and guide the Foundation's mission.

This year the PTEN Research team was pleased to welcome two new members, Dr Donald Ogilvie and Prof Sir Mene Pangalos, joining the SAB.

During the meeting, the SAB provided valuable scientific insight and helped steer the PTEN Research Foundation strategic priorities. The SAB also supported the PTEN Research strategy and commended the team on progress made on activities over the past years. In addition to providing valuable insight at the annual meeting, the SAB members also play an important part in our grant review process. They provide expert feedback and thereby help to ensure that all projects funded by PTEN Research are scientifically impactful and, most importantly, serve the PHTS community.

The PTEN Research team is very grateful to all our SAB members for their invaluable contribution in shaping our work.

World PHTS day

On 23rd October [PTEN Research Foundation](#) and other organisations who support individuals with PHTS and their families are proud to mark PHTS Awareness Day.

The date of October 23rd (10/23) was selected as it reflects the location of *PTEN* on chromosome 10 (10q23) and reminds the Foundation team of our **mission** to fund and facilitate research that will lead to better treatments and improved outcomes for PHTS and our **vision** to have transformative therapies available for people with PHTS in the next ten years.

PIPGenerating art exhibition

PTEN Research was delighted to attend the PIPgenerating ART exhibition opening in Cambridge on the 26th September. This innovative project is part of the EU Horizon 2020 funded [PIPGen](#) network, which funds PhD training in PI3K/PTEN-related diseases, thereby creating the next generation of researchers of multiple conditions including PHTS.

In this project, artists from the Cambridge School of Visual and Performing arts created art inspired by the PIPGen research projects and the voices of patients and families from [PTEN UK and Ireland](#) and [GoPI3Ks](#) patient advocacy groups.

The art exhibition was an exciting mix of original art ranging from videos to sculptures to superhero comics to clothing. The passion of the scientists and the artists for this collaboration was evident, showcasing how art and science do not have to be juxtapositions but rather can be used to celebrate connections.

The Foundation team congratulates the artists, students, and patient organisations for an inspiring collaboration, thanks the organisers for the invite, and wishes all the best to the PIPGen PhD students as they finish their research and prepare for their dissertations.

[To find out more, see here](#)

OUR RESEARCH

At PTEN Research we fund projects with leading experts in the field of PHTS at institutions around the world. A condition of our funding is the publication of the work to ensure that individuals and families affected by PHTS, as well as other scientists, benefit from the work which we fund. Further, our formal application and external review processes ensure we only fund work which we believe is of high scientific value.

If you would like to find out more about how we fund research, or apply for a research grant please click here!

Tissue samples from individuals with PHTS may help improve diagnosis in a new project

We are pleased to announce the initiation of a new collaborative research project led by Dr Peter Stanich at Ohio State University (US), working together with colleagues at Cleveland Clinic (US) and Radboud University (Netherlands) funded by PTEN Research.

The study will analyse almost 1000 samples from benign overgrowth in the colon, breast and thyroid. The study will assess if there is a difference in the level of *PTEN* protein in these tissues in individuals with PHTS compared to individuals who do not have PHTS. Previous smaller studies suggest that individuals with PHTS have much lower levels of *PTEN* in these types of tissue samples.

In the future, this test could help identify people with PHTS who have not yet been diagnosed and who would benefit from a referral for genetic testing. If the genetic testing confirms PHTS, these individuals can receive cancer surveillance to improve their health outcomes. This approach has been successfully used to detect other hereditary cancer conditions, such as Lynch syndrome (another genetic condition that increases the lifetime risk of certain types of cancer), where specific tissue markers have led to improved diagnosis.

New PHTS research 'toolbox' now available on the PTEN Research website

The PTEN Research team has recently published a new, freely available collection of resources for PHTS research on our website. This includes information about projects funded by PTEN Research as well as other funders.

The resource is aimed at PHTS researchers and includes information about clinical trials undertaken in PHTS, PHTS registries and patient cohorts published in the literature, and PHTS specific laboratory and animal models. The purpose of this 'toolbox' is to collate information about these multiple tools in one location which is easy to find and navigate. We hope this resource will stimulate new ideas, result in collaboration and accelerate future research into PHTS. The website will be updated as new resources become available.

[To find out more, see here](#)

NEW PUBLICATIONS

Literature review reveals a wide range of neurologic features in PHTS

The PHTS research team from Cleveland Clinic led by the late Prof Charis Eng have recently published a systematic literature review of neurologic features in PHTS in *Neurology*. This study was in part funded by PTEN Research.

This systematic literature review investigated 1,996 published articles describing various neurologic features in PHTS, i.e. features affecting the brain and the central nervous system. The study found that there is a wide range of neurologic features associated with PHTS and they vary by age. The publication also highlighted the importance of future clinical trials to identify new treatments but one of the key challenges for such trials is the lack of validated trial endpoints. The Foundation is highly active in this area of research to develop new trial endpoints.

[Click here to access the full publication](#)

Increased frequency of infections and autoimmune disease in PHTS

A study assessing the frequency of infections and autoimmune disease in individuals with PHTS by the Dutch PHTS research group led by Professor Nicoline Hoogerbrugge was recently published in the *European Journal of Medical Genetics*.

The study found that children and adults with PHTS have a range of infections. Children mainly experienced tonsillectomies or adenoidectomies and bacterial infections needing antibiotics. Adults reported more viral and bacterial infections and repeated fungal infections. Autoimmune diseases were more common in adults with PHTS than in individuals who do not have PHTS.

These findings highlight that individuals with PHTS experience many different immune-related issues which should be investigated further.

[Click here to access the full publication](#)

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 or fundraising](#) to support our work



PTEN RESEARCH

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](#)