



We hope that everyone is safe and well and our thoughts go out to all our contacts across the world.

At PTEN Research it is our goal is to improve the lives of the patients and their families living with PHTS (PTEN Hamartoma Tumour Syndrome) by funding research into better understanding of the condition, and into 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

THE FOUNDATION IN 2022

2022 has been a busy year for the Foundation and all its collaborators. We have tried to summarise the key activities in the image below. Further, the Foundation has initiated several new research projects, while others finished and reported results, and we hope to see continued growth into 2023. As 2022 ends we wanted to thank all our collaborators across the globe for their ongoing help, support and commitment to individuals living with PHTS.

19 active projects from basic laboratory research to clinical trials				
17 projects with hospitals and universities in Europe, the US and Australia		Active interactions with industry	Two partnerships with contract research laboratories	
28 acknowledgments in scientific journals and counting	Three PHTS multidisciplinary working groups of international experts	Multiple PHTS specific experimental models established	Two sets of PHTS specific change measures for clinical trials in development	PHTS specific diagnostic code approved by the Centers for Disease Control (US) and application ongoing with the World Health Organization

OUR RESEARCH

At PTEN Research we fund projects with leading experts in the field of PHTS at institutions around the world. It is through this approach that we generate the most valuable scientific research with the leading scientists in the field. Our formal application and external review processes ensure that we only fund work 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!](#)

NEWS AND EVENTS

PHTS is granted an International Classification of Diseases ICD-10 Code

The Foundation have been working closely with Charis Eng and David Flannery (Cleveland Clinic) and Marc Tischkowitz (University of Cambridge) and other experts to establish an International Classification of Diseases (ICD) code specific for PHTS, and are delighted to share that in October 2022 this code was granted for ICD-10-CM in the US.

In parallel the team have also applied for a code for

PHTS for the successor system ICD-11 which is managed by the World Health Organisation (WHO).

Having such a code is important as it can be recorded in an individual's medical records as a clear and unambiguous identification of their PHTS diagnosis. This supports not only individuals with PHTS directly, but also their doctors and researchers in several ways including:

- It supports access to appropriate medical care for PHTS patients including cancer screening and surveillance programmes.
- It helps to better understand the number of patients who have a PHTS diagnosis.
- It helps promote greater awareness of PHTS by doctors which in turn should lead to improved levels of diagnosis and better care.
- Supports PHTS research and potential future clinical trials.

[Click here to read the full press release](#)

2022 Scientific Advisory Board Meeting

In October, the Foundation held its annual Scientific Advisory Board (SAB) meeting by video conference. The objective of this annual meeting is to review the Foundation's scientific strategy and progress on research activities. The SAB was in alignment with the updated Foundation strategy and commended the team on progress made on research activities. The Foundation is very grateful to all our SAB members for their valuable contribution in shaping the work we do.

[Click here to find out more about our Scientific Advisory Board members](#)

Pharma Integrates Conference

One of our Trustees, Renata Crome, was a panellist at the

Pharma Integrates Conference which was held in London on the 17th of November. The expert panel focussed on the latest developments in cancer treatment and early diagnosis, and the opportunities of precision medicine. The panel, as well as several other panels on the day, included representation from patient groups highlighting the importance of the pharmaceutical industry, the NHS, and patient representatives working together to develop better treatments. Other key topics of the day included the challenge of drug development in rare disease, the role of artificial intelligence (AI) in modern drug development, the latest clinical trial designs and the importance of diversity, equity, and inclusion.

Two members of our team , Maria Whitehead and Karola Rehnstrom also participated in this event. It was an excellent conference bringing together key stakeholders across the pharmaceutical pipeline to share insights and create debate on crucial topics that influence the future of patient outcomes. It was also energising to return to a face-to-face format after several years of online conferences.

[Click here for more information about the event](#)

NEW PUBLICATIONS

Updated cancer risk estimates in PHTS

A large collaborative European study led by Dr Janet Vos and Prof Dr Nicoline Hoogerbrugge, Dutch scientists at Radboud university medical center, and funded by the PTEN Research Foundation has recently published updated cancer risk estimates in PHTS (ref Hendricks et al JNCI 2022). The study included data from medical records of 455 PHTS patients across Europe and is believed to be the largest published study in PHTS of its kind.

Critically the data will help better understand the risk of PHTS associated cancers and will be important for

informing future enhancements to cancer surveillance guidelines for PHTS. With ongoing funding from the PTEN Research Foundation, the collaborative research group is now working on understanding how those patients with PHTS who go on to develop cancer are treated and how they respond to treatment. This information is expected to be published in 2023.

[Click here to access the full publication in the Journal of the National Cancer Institute](#)

Rapamycin delays tumours in laboratory models

A study conducted by Professor Bart Vanhaesebroeck's team at University College London and funded by the PTEN Research Foundation has recently published encouraging results in non-human PHTS models. The data suggest that long-term treatment with a low dose of a drug called rapamycin (or sirolimus) delays the development of some tumours in laboratory models of PHTS. Rapamycin is a medicine currently used for treatment of some non-inherited cancers and to prevent organ rejection in transplant patients.

As rapamycin targets the biological pathway which is disrupted in PHTS, the researchers wanted to investigate if it could prevent cancer development in a laboratory setting.

These results are a promising start to further explore the role of rapamycin, as well as other drugs working in the same biological pathway as PTEN, in preventing cancers in PHTS. **However, it is important to stress that use of rapamycin for individuals with PHTS is not approved and more research including clinical trials are needed before rapamycin can be routinely used in people with PHTS.**

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

Despite COVID-19 related restrictions lessening during the year, it goes without saying that 2022 has not been without its significant challenges for people with PHTS, their families as well as our academic, clinical and other external collaborators alike. We are very proud and appreciative of all the hard work and continued progress that has been made across our supported projects during the year.

On behalf of everyone at PTEN Research we would like to extend our thanks and appreciation for the ongoing partnerships and collaboration.



If you would like to make a donation to support our work please [click here](#)

**PTEN
RESEARCH**

PTEN Research
Registered charity number: 1173589
St James House, St James Square
Cheltenham GL50 3PR

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