



**We hope that everyone is safe and well and our thoughts go out to all of 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](mailto:contact@ptenresearch.org)

## **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 PHTS patients and 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.

**Improved classification of vascular anomalies in PHTS**

Vascular anomalies are changes to veins, arteries and the lymphatic system which occur in 30-50% of patients with PHTS. These can cause significant problems for patients and may require either surgical or medical treatment.

The grant for this latest study, supported by PTEN Research Foundation, was signed in January 2022. The study is led by Dr Denise Adams from The Children's Hospital of Philadelphia and has brought together experts from multiple research centres in North America and Europe to provide a better classification system for vascular anomalies in PHTS.

The study will use a variety of data such as: information from medical imaging, a description of the location, characteristics, and cellular composition, of the vascular anomalies from multiple people with PHTS. An important application of the improved classification system is to help future clinical trials to better assess whether new treatments are effective for treating vascular anomalies in patients with PHTS.

As this project is being conducted using existing scan data and tissue samples, and information stored at the participating hospitals, it is not possible for patients to directly take part in this study. However, we will share the results of this study in our newsletter and on our website once the project has finished. The results will also be published in a peer-reviewed scientific journal to ensure the results are available to a wide audience of researchers and doctors.

**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**



### **PTEN Research Foundation appoints two new Scientific Project Managers**

The Trustees are pleased to announce the appointment of Cristina Gomes and Karola Rehnström as the Scientific Project Managers for the PTEN Research Foundation.

Cristina and Karola support funding processes and grant applications, external review processes, negotiation of legal contracts and the general day-to-day management of the Foundation's research portfolio.

Cristina facilitates basic and translational research following the Foundation's scientific strategy, by supporting academic partnerships and biotech collaborations. Before joining PTEN Research Cristina worked for more than 10 years as a research scientist, first in academia and later gaining translational research experience in industry. As a PhD student in Portugal and later as post-doctoral scholar at the University of Chicago, Cristina was dedicated to the development of

basic molecular and cellular research projects. Cristina studied the link between the mechanisms of protein synthesis fidelity and the stress response in human cells, while supporting several collaborations for advancements in the field of tRNA biology. More recently, Cristina specialised in non-clinical studies by supporting biotech companies and start-ups in the design and implementation of drug discovery programs.

Karola supports the Foundation's clinical project portfolio. Karola has 10 years' experience in scientific project and programme management, ranging from the NHS and academia to commercial health technology companies. Prior to joining PTEN Research Karola project managed academic and commercial genetics initiatives and clinical trials, such as delivery of the 100,000 Genomes Project in the East of England. The 100,000 Genomes Project was a national project creating infrastructure for improved access to genetic testing and whole genome sequencing for NHS patients with rare diseases and cancer. Most recently Karola worked for Genomics plc, a UK health-tech company leveraging genomic information for improved disease risk prediction, drug discovery and personalised medicine. Karola holds a PhD in medical genetics from the University of Helsinki, Finland.

### **Rare Disease Day 2022 – PTEN Research Collaborators Meeting**

The 28<sup>th</sup> of February 2022 was Rare Disease Day. The objective of this Day is to raise awareness amongst the public and decision-makers about rare diseases and their impact on patients' lives.

PHTS is one of over 6000 known rare diseases and it estimated that 300 million people worldwide live with a rare disease.

To mark this day for 2022, and to celebrate 5-year anniversary our formation, the Foundation hosted a scientific meeting for its current and past grant holders and collaborators. The aim of the event was to share information about the research projects supported by the

Foundation and to further increase collaboration within the field. The day brought together researchers from all over the world for a full day of interesting presentations and discussion. We were inspired by the fantastic attendance and level of enthusiasm shared by both speakers and attendees and hope this event will facilitate future research into PHTS.

### **PTEN UK & I Patient Day**

We are delighted to have been invited to give a presentation at this year's PTEN UK & Ireland Patient Day on Saturday the 2nd of July 2022.



[Click here to visit the PTEN UK&I website to sign up and secure your place.](#)



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PTEN Research  
Registered charity number: 1173589  
3rd floor, Paternoster House  
65 St Paul's Churchyard  
London EC4M 8AB

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