



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)

## 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 we fund. Further, our formal application and external review processes ensure we only fund work which we believe is of high scientific value.

We are proud to introduce two recently initiated Foundation funded research projects:

### **Inclusion of UK Families in the Development of a PHTS Neurobehavioral Evaluation Tool**

PTEN Research is funding the work of Dr Tom Frazier at John Carrol University in Ohio, USA, to develop a PHTS specific [Neurobehavioral Evaluation Tool](#) (NET) which can be used to help diagnose people with PHTS as well as monitor whether treatments used in clinical trials result in changes to the symptoms an individual is experiencing. To date this work has included families in the US. To ensure that the tool has applicability to families outside of the US, PTEN Research is funding the inclusion of UK families in the study, led by Dr Katherine Lachlan at University Hospital Southampton NHS Foundation Trust. The study is conducted completely online and therefore doesn't require any visits to hospital making it possible for families all over the UK to take part. If you are interested in learning more, please see [ClinicalTrials.gov](#) or contact the research team directly on [netstudy@uhs.nhs.uk](mailto:netstudy@uhs.nhs.uk).

### **Improving Understanding of PTEN Function in the Brain**

Following the Foundation's first Targeted Call for Applications in 2021/2022 (our competitive funding mechanism to address specific research questions), PTEN Research is funding a new grant led by Dr Bryan Luikart at Dartmouth College USA. The project, which is a collaboration with the groups of Dr Jeremy Barry (University of Vermont USA) and Dr Matthew Weston (Virginia Tech USA), aims to investigate how cognitive processes are disrupted in individuals with PHTS and autism spectrum disorder. The study will use laboratory models of PTEN loss in the brain. Investigation of these processes at a molecular level is hoped to improve understanding of how behavioural changes seen in PHTS develop. It is hoped that data from this project can help develop more efficient treatments for PHTS in the future.

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 Collaborators Meeting**

The second PTEN Research Collaborators Meeting was held on 8 March. This year the focus was on PTEN Research funded projects which have been initiated within the last year, in addition to some projects which are nearing completion.

The aim of the event was to share information between expert researchers and clinicians treating PHTS about the projects supported by the Foundation with the goal to increase collaboration and accelerate developments within the field.

We are very grateful to all the participants who agreed to present data or took the time to attend the virtual event.

### **PTEN UK&Ireland Power Hour on Drug Repurposing for PHTS**

Paul Elvin, our Director of Translational Medicine, was invited to present at the PTEN UK&Ireland Power Hour on 22 March together with Priyanka Tibarewal, researcher at University College London and PTEN UK&Ireland Trustee, about repurposing of existing drugs for use in PHTS.

The Foundation is working with both academic and contract laboratories to test existing drugs to assess if they could be used as future PHTS treatments. The main objective of this work is to create the rationale for future PHTS clinical trials. Drug repurposing is an established approach and has the ability to find new uses of existing drugs in a way which can be faster, lower risk and lower cost than traditional drug discovery approaches.

The Power Hour is a monthly online event covering items of interest to the PHTS community. Recordings of some of the Power Hours are available on the [PTEN UK&Ireland YouTube channel](#). Thank you to PTEN UK&Ireland for the opportunity to be involved in this important event.

[Click here to visit the PTEN UK&Ireland website for more information and future events.](#)



## NEW PUBLICATIONS

Professor Charis Eng's research group at Cleveland Clinic recently published a review of different hamartomatous polyposis syndromes, of which PHTS is one. The review reports that gastrointestinal polyps or growths in the gut are found in more than 90% of individuals with PHTS who underwent at least one upper or lower endoscopy (where a tiny camera is used to look at the digestive system).

To understand whether the polyp burden can be reduced in individuals with PHTS using treatment with sirolimus (a drug targeting the molecular pathway which is activated by a PTEN mutation), PTEN Research is providing funding for a study led by Dr Peter Stanich at Ohio State University. More information about the study can be found on [ClinicalTrials.gov](#).

[Click here for more details on the publication in Gastroenterology.](#)

## 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](#) in aid of our work

Thank you for your interest in and support of PTEN Research and for taking the time to read our newsletter.



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