



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 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. During this first quarter we have been busy planning with this in mind and hope to have some exciting updates to share with you later in the year.

Whilst some of our activities are still impacted by COVID-19, we continue to work hard towards achieving our vision and mission.

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

NEW SCIENTIFIC PROJECTS INITIATED

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 is of high scientific value.

Developing PHTS specific tools

A major consideration when developing any new treatment for a medical condition is to ensure that treatment effects are measured accurately. In some cases, this might be quite straightforward. For example, for a patient in a clinical trial for a new medicine to control blood pressure, it is easy to directly measure changes to blood pressure using simple and accurate measuring device.

For people with autism spectrum disorder (ASD) or neurodevelopmental disorders (NDD) measuring changes as result of therapy is more complex. This is because therapy may affect multiple different behaviours and traits which are harder to measure directly. Such behaviours or traits might include coordination, anxiety levels, how a subject interacts with other people or the presence of restricted and repetitive patterns of behaviours and interests, among many other possibilities.

Also, it might not be possible to know ahead of time whether only one or several changes might be caused by a new treatment even though a treatment difference in any one behaviour or trait might make a big difference to the quality of life of the affected person and their family.

Generally in ASD/NDD, changes are assessed through questionnaires completed by parents and caregivers and by asking subjects to do specifically designed tasks which are observed and rated by expert doctors or psychologists.

At the moment there are no tools that have been developed specifically for people with PHTS. This is important as the type of symptoms experienced by people with PHTS and ASD/NDD may differ from those caused by other conditions. Therefore, to ensure that any potential positive effects of treatments are accurately and sensitively measured in this population, it is important to see if the type of symptoms experienced by people with PHTS differ from other types of ASD/NDD.

Dr. Mirko Ujcarevic (University of Melbourne, Australia) and Dr. Antonio Hardan (Stanford University, USA) are exploring how existing measures designed for ASD/NDD populations perform with people with PHTS and whether these tools need to be modified for this patient population. It is hoped that this work will also allow existing tools to be shortened, making this type of assessment more relevant, accurate, sensitive, and less intensive on parents and caregivers to complete.

In parallel to this grant, Dr. Thomas Frazier (John Carroll University, Ohio, USA) is being supported to develop brand new survey tools for PHTS subjects that can be administered quickly at home over the internet. In addition, this project will also use a webcam to assess how individuals with PHTS respond to different stimuli before and after treatment, without the need for patients and families to visit their doctor in-person.

The Foundation are partnering with both groups to setup a discussion with the US Food and Drug Administration. The goal of this review is to ensure that the way the tools are being developed and validated will support their use in future clinical trials.

NEWS AND EVENTS

Everolimus Study Data

We know the whole PHTS community eagerly awaits the results of the phase II clinical trial which assessed the use of everolimus in PHTS subjects with difficulties with neurocognition.

In this study, patients received either treatment with everolimus or placebo (a dummy pill) for 6 months. During treatment neither the families nor the treating doctors knew the treatment that had been assigned so that this information did not influence the way study results were recorded. After 6 months subjects who were found to have received the placebo were offered a further 6 months of treatment with active everolimus.

The study was led by a group from Boston Children's Hospital with support from the US National Institute of Health, Novartis Pharma and PTEN Research. In addition to Boston Children's Hospital, the Cleveland Clinic and Stanford University also enrolled patients into the study.

The clinical part of the study has been completed and the project team expects study results to be published in a scientific journal later in the year.

This was the first time a placebo controlled randomised trial was conducted in a PHTS population and is a significant milestone for PHTS research. The team therefore published a scientific paper of the study design and methods that are planned to be used to analyse the study data. Although very technical in nature, the paper is hoped to support the design of potential future studies.

[Click here to access the full text article at Elsevier](#)

UK PHTS Patient Registry

In April 2019 Cambridge University secured funding from the PTEN Research Foundation to create the UK's first PTEN patient registry alongside the University of Edinburgh and University Hospitals Southampton.

By collecting clinical data, the PHTS registry aims to:

- Improve understanding of PHTS
- Improve patient outcomes, treatment and symptom management for PHTS
- Develop multidisciplinary approaches to predict PHTS complications for people
- Provide evidence to support treatment guidelines

We are delighted that this registry is now open! Please visit the website for more information via the link below.

[Click here to visit the PHTS Registry website](#)

PTEN Research Appoints Chief Executive

The Trustees are pleased to announce the appointment of Thomas Pepper as the first Chief Executive of PTEN Research. The creation of this new role results from the growth of activity since the Foundation was formed in 2017.

Tom joined PTEN Research in 2019 as Director of Clinical Development. He brings 25 years of drug development experience to the role and has previously worked in industry leading projects both in rare diseases and in oncology. Tom will also continue as Director of Clinical Development within the Foundation.

Rare Disease Day 2021

Rare Disease Day takes place on the last day of February each year and the main objective of this Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives. This year Rare Disease Day was the 28th of February.

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

You can see our Rare Disease Day flyer below:



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



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