PTEN Research is committed to ensuring your data is safe and secure. For more information please see our Privacy Policy.

You can see our Rare Disease Day Zyer below:

28th of February.

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

PTEN Research Appoints Chief Executive

The clinical part of the study has been completed and the project team expects results to be recorded. After 6 months subjects who were found to have received the placebo were offered a further 6 months of treatment with active everolimus.

We know the whole PHTS community eagerly awaits the results of the phase II clinical trial being developed and validated will support their use in future clinical trials.

The Foundation are partnering with both groups to setup a discussion with the US Food and Drug Administration and further clinical trials with everolimus for PHTS.

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 are measured accurately and sensitively.

The problem with existing assessment protocols is that they are often not developed to assess the unique needs of this population. As a result, they can be less relevant, consistent, sensitive and less intensive. 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 straightforward methods, such as monitoring blood pressure at home. However, it is much more complex to measure changes in other people or their symptoms, as many of these symptoms or traits might include coordination, anxiety levels, how a subject interacts with others or the presence of restricted and repetitive patterns of behaviors and traits.

• Improve understanding of PHTS

• Improve patient outcomes, treatment and symptom management for PHTS

• Provide evidence to support the design of potential future studies.

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 this paper, although it is not in its final form. However, it supports the hope that we can use this to design the future clinical trials.

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.

In April 2019 Cambridge University secured funding from the PTEN Research Foundation to investigate the use of a novel assessment tool called the Person Specific assessment of Neurodevelopmental Disorders (PS-ASD/NDD) in people with PHTS. The tool is being adapted to measure the impact of treatments on people with PHTS and tested against a standard measure of ASD/NDD symptoms, the Autism Diagnostic Observation Schedule (ADOS). The study results are being published in the future.

In parallel to this grant, Dr. Thomas Frazier (John Carroll University, Ohio, USA) is being supported to develop a mobile phone app. This will allow patients to easily complete questionnaires from home, ensuring PHTS patients and other scientists benefit from the work which we fund. Further, in line with this, we aim to share the research we fund with the wider public.

Although very technical in nature, the paper is hoped to support the design and implementation of future clinical trials.