THE AUTUMN ISSUE

THE LATEST FROM PTEN RESEARCH

We hope that everyone is safe and well and our thoughts go out to all of our contacts across the world during this difficult time.

At PTEN Research we are dedicated to the discovery and development of treatments for PHTS. We support, fund and facilitate research and bring together a global network of PHTS-focused experts.

Whilst we expect some of our activities to be impacted by the outbreak of COVID-19, we are actively supporting all our grant holders and researchers to progress their work where they can.

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

CLINICAL DEVELOPMENT

Over the past year the PTEN Research team have been partnering with PHTS experts from multiple different medical and research specialisms who are based in the UK, US and Europe to set up two Clinical Development Working Groups.

One group is focused on the people with PHTS who have cognition difficulties and the other group is focussed on PHTS patients who have vascular anomalies (a group of disorders that involve abnormal connections of blood vessels). The focus of both groups is to build better ways to more consistently develop treatments for people with PHTS in the future.

Some the of the key questions the groups are looking at include:

- What are the best clinical trial designs?
- What kind of tools are needed to measure changes in people who have been treated in a clinical trial?

Ultimately, we hope to publish the outputs of both groups so that this information is available to all researchers in the future.

MEET OUR GRANT RECIPIENTS!

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 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!

We are delighted to announce that we have awarded funding to two new research projects during the last quarter and are excited to be able to support this important work.

Professor Charis Eng and her team at the Genomic Medicine Institute of the Cleveland Clinic Lerner Research Institute in Cleveland, Ohio are undertaking research to better understand why different individuals with a germline *PTEN* mutation have different symptoms.

This research will sequence the entire DNA of hundreds of people with have a germline *PTEN* mutation, associated with PTEN hamartoma tumour syndrome (PHTS), to analyse the differences between individuals and how this relates to the presence or absence of symptoms. It is hoped that this improved understanding generated from the study will ultimately lead to better predictions of, if and when symptoms will develop in a specific individual. In addition, the information gained will be crucial to identify new potential treatments for PHTS in the future, at an individual basis, thus personalising medical care.

Professor Bart Vanhaesebroeck and his team at University College London in the UK are undertaking research to better understand the potential for drugs to be used to prevent cancer development in models developed to mimic PHTS in people.

It is hoped that the results from this work will inform about the potential for drugs to prevent cancer development in PHTS that may one day lead to the use of treatments for this purpose.

NEWS AND EVENTS

During the last quarter the team have been working on updating the Foundation's Strategy. While our vision and mission remain the same, we have been fine-tuning our approach to ensure we are being as focussed and productive as possible. We hope to be able to publish a summary of the updated Strategy on our website during the next quarter.

We are also pleased able to share the publication of a paper on "Cancer Surveillance Guidelines for individuals with PTEN Hamartoma Tumour Syndrome" by Dr Marc Tischkowitz *et al.*

Publication was partially supported by funding from the PTEN Research Foundation.

Click here to access the full publication

Members of the PTEN Research Team attended the PTEN Hamartoma Tumor Syndrome Foundation (USA) annual symposium which was held online on the 13th of August.

This was an excellent event and featured a number of interesting sessions on PHTS and the vital role of patients in rare disease research.

To find out more about the PTEN Hamartoma Tumour Syndrome Foundation and their work click here.

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

If you would like to make a donation to support our work please click here

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