



**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 it is our fundamental goal is to improve the lives of the patients and their families living with PHTS by funding research into better understanding of the disease and into the development of potential 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 we expect some of our activities to be impacted by the outbreak of 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 our strategy please visit our website or email us at contact@ptenresearch.org

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 grants for two additional research projects in the last quarter and are excited to be able to support this important work.



Professor Mustafa Sahin and his research team at Boston Children's Hospital are developing new preclinical models to study the effects of PTEN loss in cells of the brain.

It is hoped that this model will further develop the understanding of the neurological manifestations of PHTS and provide a means for testing new treatments that could support future clinical trials and treatment options for PHTS patients.



Dr Mariona Graupera and her team at the Institut d'Investigacio Biomedica de

Bellvitge (IDIBELL) in Spain are undertaking research to better understand the cause of vascular malformations in PHTS.

This research will generate and use model systems of vascular malformations in PHTS to improve the understanding of the molecular and cellular events that lead to the development of vascular malformations. In addition, the Graupera Lab will test potential drug candidates in these models to generate data to support future therapy options for PHTS-related vascular malformations.

NEWS AND EVENTS



February 29th 2020 was Rare Disease Day

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.

As PHTS is one of over 6000 rare diseases, we created an information poster to explain

a more about it.

[Click here to view our poster on PHTS for Rare Disease Day 2020](#)

Whilst the core mission of PTEN Research is working towards developing treatments to improve the lives of PHTS patients around the world, we also take an interest in other activities that are taking place within the global Rare Disease Community.

On Feb 26th 2020 the UK government published their 2020 update to the implementation plan for the UK Strategy for Rare Disease. A significant action that has been taken is that the government announced in Jan 2020 that whole exome sequencing would be provided to critically ill children and babies which may more than double the chances of successful diagnoses.

[Click here to read more about the February 2020 Policy Update](#)

Members of the PTEN Research Team attended a number of events during the last Quarter, including:



The inaugural Rare Disease Film Festival that was held on Feb 10th at Regent Street Cinema. All the films that were screened were exceptionally moving, and really highlighted the reality of living with a rare disease.

[Click here to visit the Rare Film Festival Website](#)



The Find-A-Cure conference on Repurposing Drugs for Rare Diseases. Speakers from Charities, Industry & Rare disease patients provided some key insights into the challenges and rewards of this

venture.

[Click here to find out more about Find-a-cure](#)



The Rare Disease Reception at Westminster organised by Rare Disease UK . Speakers included: Liz Twist MP; Dr Jayne Spink, Chair of Rare Disease UK; Dr Jenny Harries, Deputy CMO for England; as well as Amanda Brodie and Kirsty Blakeborough-Wesson who are both patients affected by rare diseases.

[Click here to find out more about the Rare Disease Reception](#)

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](#)

NEXT ISSUE: An update on the UK PHTS
Patient Registry as it approaches its launch
date in 2020



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