THE SPRING ISSUE

THE LATEST FROM PTEN RESEARCH

PTEN RESEARCH

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 our activities, please visit our website or email us at contact@ptenresearch.org

NEWS AND EVENTS

Two New Members Join the Foundation's Scientific Advisory Board

PTEN Research are delighted to welcome two new members to our Scientific Advisory Board (SAB) Dr Donald Ogilvie and Prof Sir Mene Pangalos. Both Donald and Mene bring enormous experience and expertise in the field of drug development and complement our existing board members' leadership in PHTS, genetics, rare disease drug development, registries and bio-banking.

By strengthening its scientific oversight, the Foundation hopes to further progress and accelerate its mission to fund and facilitate research that will lead to better treatments and improved outcomes for people with PHTS.

Dr Donald Ogilvie MA DPhil

Donald is an independent scientific consultant providing expert advice on cancer drug discovery to academic, industrial, and venture capital organisations.

Donald obtained an MA in Biochemistry at the University of Oxford, UK, in 1980, before working at the John Radcliffe Hospital, Oxford, for eight years on the cell processes in cancer, then on inherited connective tissue disorders. The latter was the basis of his DPhil degree. In 1988, Donald joined ICI which subsequently became Zeneca then AstraZeneca. For most of his twenty-year career in the pharmaceutical industry, he worked on cancer drug discovery and early clinical development projects. He was directly responsible for the delivery of ten novel cancer development compounds, several of which have progressed to phase II & III clinical trials and two, to US regulatory approval. Between 2009 and 2017, Donald set up and led the academic Drug Discovery Unit at the Cancer Research UK Manchester Institute (University of Manchester, UK). Under his leadership, the team built a drug discovery infrastructure and a leading cancer project portfolio. During this period, three projects, including one with a development compound, were successfully licensed to downstream partners. Donald is a co-author on more than ninety peer-reviewed academic publications.

Prof Sir Mene Pangalos DSc PhD FBSB FMedSci HonFRSPS FRS

Until Spring 2024 Mene held the role of Executive Vice President, Research and Development at AstraZeneca being responsible for R&D from discovery through to late-stage development covering Cardiovascular, Renal, Metabolism, Respiratory, Immunology, Microbial Science and Neuroscience areas. Prior to this, he served as Executive Vice-President of AstraZeneca's Innovative Medicines & Early Development Biotech Unit and Global Business Development.

During his time with AstraZeneca, Mene led the transformation of R&D productivity through the development and implementation of the "5R" framework resulting in a greater than four-fold increase in success rates compared to industry averages. In parallel, he has championed an open approach to working with academic and other external partners, changing the nature of academic-industry collaboration.

In addition, Mene led and oversaw AstraZeneca's R&D response to COVID-19; maintaining existing clinical trials and delivery of medicines to patients, responding to the UK government's call for supporting to national testing effort, and discovering and developing new preventative and treatment approaches to the disease. This work involved partnering with the Oxford University in the global development of a vaccine and ensuring broad and equitable access at no profit during the pandemic, the discovery and development of a long-acting antibody combination for those who could not be vaccinated, as well as exploring AstraZeneca's existing portfolio as potential treatment options against the disease. The team were awarded the Copley Medal by the Royal Society in 2020.

Mene previously held senior R&D roles at Wyeth and GSK.

Mene holds Honorary Doctorates from Glasgow University and Imperial College, London, is a Fellow of the Academy of Medical Sciences, the Royal Society of Biology and Clare Hall, University of Cambridge and is a Visiting Professor at The Wolfson Centre at King's College. He co-chairs the UK Life Sciences Council Expert Group on Innovation, Clinical Research and Data and is a member of the Life Sciences Industrial Strategy Implementation Board. He is also on the Boards of The Francis Crick Institute and The Judge Business School, Cambridge University, and is a member of the Life Sciences Vision Advisory Group. Mene was awarded the 2019 Prix Galien Medal, Greece for his scientific research and named Executive of the Year at the 2019 Scrip Awards. In 2019, Mene was awarded the honour of a Knighthood by Queen Elizabeth II for his services to UK science. In 2021 Mene was awarded an Honorary Fellowship of the British Pharmacological Society. In 2022 Mene was elected as a Fellow of The Royal Society and an honorary professor of Cambridge University Medical School.

PTEN Research Collaborators' Meeting

The third PTEN Research Collaborators' Meeting was held on March 12th. This virtual event brought together PHTS researchers and expert clinicians from around the world to share the latest updates from Foundation funded research projects and to increase collaboration.

This was a so called 'closed meeting' in order to allow the sharing of new and previously unpublished data with the PHTS research community as early as possible. This is hoped to both accelerate research and stimulate debate into new areas of potential PHTS research.

It should be stressed that in keeping with good practice, all results from Foundation funded research must ultimately be published so that the data will benefit all members of the PHTS community including researchers, clinicians, and most importantly, patients and their families.

Thank you to all the researchers and clinicians who joined to share their latest findings and contributed to the discussions.

International Society for the Study of Vascular Anomalies World Congress 2024

The Foundation attended the International Society for the Study of Vascular Anomalies (ISSVA) World Congress in Madrid, Spain in early May 2024.

It is estimated that vascular anomalies are experienced by 35-50% of individuals with PHTS, and in common with other manifestations of the condition, there are currently no health authority approved drug treatments for PHTS vascular anomalies. These symptoms associated with vascular anomalies can cause significant illness, pain and impact on quality of life.

The Foundation hosted a meeting for an international team of researchers, working on the Foundation funded project aiming to better understand and classify the vascular anomalies in PHTS. Developing the classification is an important supporting step in developing future treatments.

The Foundation would like to thank ISSVA and all key stakeholders for a very successful and interactive meeting.

OUR RESEARCH

At PTEN Research we fund projects with leading experts in the field of PHTS at institutions around the world. As noted above, a critical 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 which we support. Further, our formal application and external review processes ensure we only fund work which we believe is 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.

Targeted Call for Applications Launched

The Foundation has opened a Targeted Call for Applications for new research projects to be funded by the Foundation with a goal of further building the understanding of the underlying biology of PHTS and how symptoms develop.

This targeted call is supported by the availability of cell models called induced pluripotent stem cells, or iPSCs, that were developed as part of an earlier collaboration between the Foundation and Boston Children's Hospital. These cells have been created from small skin or blood samples, generously donated by individuals with PHTS and their families.

iPSCs are a valuable research tool as they can be grown in a laboratory into many types of cells, eg a neurone or brain cell, which can then be used to model the different symptoms that people with PHTS experience.

The deadline for submitting research proposals is 28 June 2024.

To find out more information and criteria on who is eligible to apply, please click here.

Natural History Study of Individuals with Autism and Germline Heterozygous PTEN Mutations

This PHTS natural history study is seeking volunteers with PHTS, 18 months of age or older, to participate at multiple sites across the United States.

A natural history study follows a group of individuals with PHTS over time and collects health information to better understand the problems they experience and how their condition changes. Such data is critical for development of future treatments.

More information can be obtained from the study coordinators listed in the information sheet below or from the Clinicaltrials.gov website.

This study is funded by the National Institutes of Health grant (U54NS092090) to the Developmental Synaptopathies Consortium, and the Foundation will be providing additional funding for the study in 2024-2025.

NEW PUBLICATIONS

Three recent publications report results from projects which have received funding from PTEN Research.

Lifestyle factors and breast cancer risk in females with PTEN Hamartoma Tumour Syndrome (PHTS)

A recent study from the research group of Professor Nicoline Hoogerbrugge at the Radboud University Medical Centre, Netherlands is the first of its kind to explore the role of lifestyle factors affecting the risk of breast cancer in women with PHTS. The study findings suggest that a healthier lifestyle could potentially decrease breast cancer risk in PHTS in a similar way as for the general population.

For more information about the study results, see Cancers.

Exploring the neurological features of individuals with germline PTEN variants: A multicentre study.

This study from Professor Charis Eng's laboratory at Cleveland Clinic, US, which is part of the PTEN Research Young Investigator Award to Dr Andrew Dhawan, systematically explored the neurological features of individuals with PHTS who have participated in the ongoing study Natural History Study of Individuals with Autism and Germline Heterozygous PTEN Mutations. See above for more details about the study). Thank you to all the PHTS patients and their families for supporting the natural history study and for making this research possible.

For more information about the study results, see Annals of Clinical and Translational Neurology.

Exploring the Prevalence of Oral Features for Early Detection of PTEN Hamartoma Tumour Syndrome

This publication from Professor Hoogerbrugge and collaborators at Radboud University Medical Centre, Netherlands, aims to raise awareness amongst dentists of the oral symptoms of PHTS to help identify people who may benefit from a PHTS genetic assessment. Particularly adults with PHTS have been noted to have at least two or more of gingival hyperplasity (gum enlargement), oral papillomas (growths on the lips or the mouth) and high oral palate (roof of the mouth).

For more information about the oral symptoms in individuals with PHTS, see the International Journal.

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 and support of PTEN Research and taking the time to read our newsletter

PTEN RESEARCH

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