



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.

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.

Boston Children' Hospital – Generation of PHTS iPSC lines to discover novel treatment options

Professor Mustafa Sahin and Dr. Elizabeth Buttermore at the Rosamund Stone Zander Translational Neuroscience Center at Boston Children's Hospital in Boston, Massachusetts are undertaking a project to develop cell lines, derived from individuals with PTEN mutations, that will be a crucial resource for the research community to better understand how treatments developed in the laboratory may translate into humans.

To generate the patient-specific cell lines, known as induced pluripotent stem cells (iPSCs), this research project will involve the collection of samples of cells (either from blood or a small sample of skin) and the conversion of these samples into iPSCs. iPSCs are a powerful research tool that can be changed, or differentiated, into many different cell and tissue types, including cells of the brain or blood vessels. These 'differentiated' cells can then be used as a patient-derived model of PHTS to test the effect of treatments and to better understand the fundamental biology of the syndrome.



2021 CALL FOR RESEARCH APPLICATIONS

We have recently opened an opportunity for new scientific research applications focused on an underexplored area of PHTS disease mechanism. The PTEN gene controls many processes in cells. Many studies over the years have established how the PTEN protein functions and why mutations in PTEN lead to changes in biochemical signalling within cells which results in the multiple different symptoms of PHTS.

These studies have also found less well understood aspects of PTEN function but how these contribute to the development of PHTS is not clear. To study the importance of these so-called 'non-canonical functions' of PTEN in PHTS, the Foundation has launched a call for new research applications. By funding research into the basic mechanisms of PTEN, we aim to improve understanding of the underlying molecular mechanisms that lead to various symptoms PHTS and possibly uncover future avenues for treatment development.

This so called 'targeted call' is open to researchers globally and following our formal review and assessment process, successful applications will receive funding from the Foundation to undertake their research programmes.

The deadline for submitting applications is 7 January 2022. More information and criteria on who is eligible to apply can be found on our website:

[Click here for more information and to apply](#)

NEWS AND EVENTS

New Foundation Website

PTEN Research Foundation is pleased to announce the update of its website PTENresearch.org.

Over the past few months, we have been working to improve the content of our website to make it more useful and relevant to different users.

The website now has two distinct sections:

• 'For families living with PHTS': gives an overview of the condition, sources of further information and support, back copies of the PTEN Research Newsletter, and a frequently asked questions section - all written in family friendly language.

• 'For researchers and professionals' has been extensively updated to provide an overview of the Foundation's strategy as well as more details on the type of projects currently supported through Foundation grants. It also outlines how suitability qualified researchers and health care professionals may apply for grants. We hope this information will stimulate additional high-quality grant applications and support further vital research into PHTS.

[Click here to access to visit the new look website](#)

UK PHTS Patient Registry

Recruitment to the PHTS Registry is going is progressing well 39 participants now enrolled. Many thanks to all those that have consented so far.

PTEN UK and Ireland together with the lead investigator Marc Tischkowitz have recently made a You Tube video which we hope will answer many of the questions that patients and families have about taking part in in the Registry.

More information is available on our website or by emailing add-tr.phts@nhs.net

[Click here to view the YouTube videos](#)

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

PHTS and COVID-19 Vaccination

At the current time PTEN Research is not aware of any specific guidance regarding the use of Covid-19 vaccines in people with PHTS. However, in February 2021 some general guidance was issued in Europe by the European Reference Network specialising in rare diseases, including PHTS.

This guidance provides advice on two things, both relating generally to adults with rare diseases:

• Which particular diagnoses, symptoms and treatments which would mean that patients should be **prioritised** for vaccination;

• The circumstances where vaccination would be "**contra-indicated**", which means it is not recommended for safety reasons.

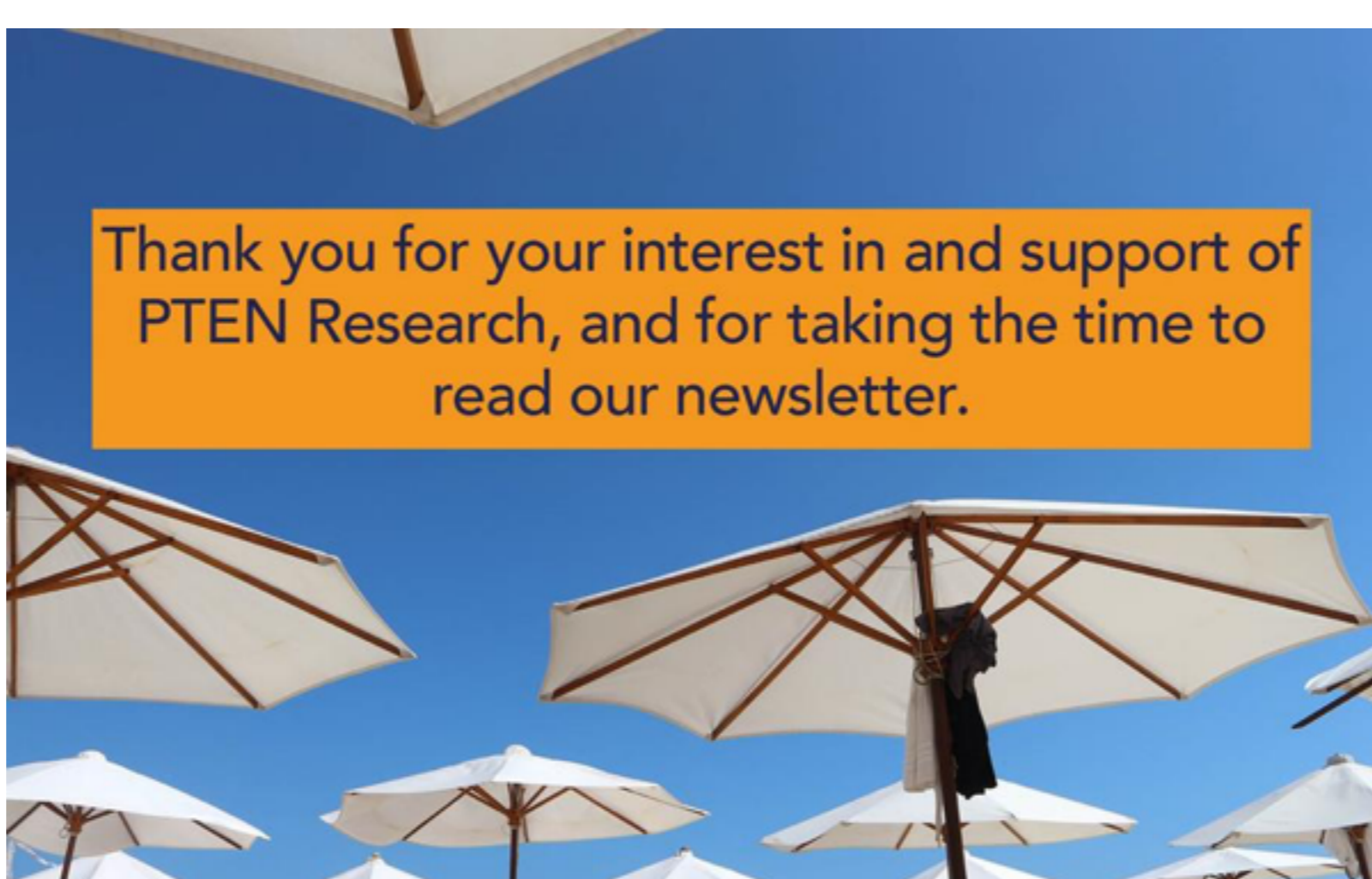
PHTS as a condition is *not* listed as either a reason to *prioritise* for vaccination, or as a *contra-indication*.

This means that, in general, people with PHTS should be vaccinated in line with the plans for the general population.

However, due to the wide variation in symptoms, characteristics and treatments associated with PHTS, it is important to seek advice from your doctor to check what is most appropriate for you.

In addition, experience with Covid-19 vaccinations is growing rapidly, including the approval of vaccines for use in children in some countries, **underlining the need to seek up-to-date information from your doctor.**

[Click here to access the online COVID-19 guidance](#)



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