The latest updates from PTEN Research



At PTEN Research it is our goal to improve the lives of individuals affected by PTEN Hamartoma Tumour Syndrome (PHTS) 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

Our clinical collaborators and the PTEN Research team are busy developing new research projects. We hope to share more details in the coming months about several new clinical studies planned in the US and UK.

We are very grateful to PHTS advocacy groups and organisations for helping the respective study teams share further information when these projects open so that people with PHTS and their families can consider, following discussion with the respective study medical team, if participation may be right for them.

OUR RESEARCH

At PTEN Research, we fund and facilitate projects with leading experts in the field of PHTS, run in hospitals and universities around the world. Our grant funding process and rigorous reviews ensure that we only fund work of high scientific value which is expected to benefit the PHTS community. A condition of our funding is that the work we support must be published to allow individuals and families affected by PHTS, as well as other physicians and scientists working in field, to be aware of the latest developments and data.

If you would like to find out more about how we fund research or apply for a research grant, please click here.

Development of a test to screen existing medicines for neurological symptoms in PHTS

PTEN Research is pleased to announce a new grant led by Dr Elizabeth Buttermore at Boston Children's Hospital, working with Dr Jed Hubbs.

This project aims to identify existing medicines that could, in the future, potentially be repurposed to treat neurological symptoms (problems linked to the brain and nervous system) associated with PHTS.

Research suggests that neurons (a type of nerve cell) carrying genetic *PTEN* changes can be more active than regular neurons. This level of activity may disturb the balance of signals in the brain, affecting how the brain develops and functions, and contribute to the neurological symptoms seen in some people with PHTS. However, there are currently no approved treatments that directly address these underlying changes in PHTS.

To investigate possible treatments, Dr Buttermore's team will use stem cells. These were created from blood or skin samples donated by people with PHTS in an earlier project supported by PTEN Research. Stem cells have the special property that they can be turned into other cell types found in the body, in this case neurons, which will be used to develop a test that measures unusual patterns of brain cell activity.

The researchers will then use this test to screen approximately 4,000 compounds, focusing particularly on medicines that are already approved to treat other conditions. Promising compounds will undergo further testing to confirm whether they can reduce unusual neuronal activity without harming the cells and thereby provide the rationale for further evaluation as a future treatment for people with PHTS.

Because existing drugs that are already approved by health authorities to treat other conditions have a large body of data about how they work and their safety, this so-called repurposing approach could help accelerate the search for potential future PHTS treatments. Any promising medicines identified through this project will require further research to ensure they are safe and effective before they could be tested in clinical trials involving people with PHTS.

New case studies available on the PTEN Research website

To highlight new projects we are funding, we have updated the 'Case studies' section of our website. Here you can find detailed information about select research grants, their aims and the expected or achieved outcomes. We fund research at all stages, from early pre-clinical laboratory research to human clinical trials, and our research portfolio is carefully considered to only support projects that we believe will have a high possible impact for people with PHTS in the future.

For more information, see our new and improved Case Studies.

NEW PUBLICATIONS

New European PHTS cancer surveillance guidelines published

A European expert group has recently published updated PHTS cancer surveillance recommendations. It brings together research evidence, specialist expertise and input from patient representatives and aims to help detect cancers at an earlier, more treatable stage.

PHTS is associated with an increased chance of breast, thyroid and endometrial (lining of the womb) cancers, and a more moderate increase in the chance of kidney, bowel and skin melanoma cancers. The guidelines, developed in the context of European health systems, recommends how PHTS surveillance should be tailored to the individual, taking into account their medical and family history, age and sex.

The guidelines also stress the importance of review by a clinical geneticist at diagnosis, access to a multidisciplinary PHTS team where available, and prompt assessment of new symptoms. The advice is meant to be personalised with the individual's clinical team and may vary by country.

See the full guidelines published in the European Journal of Human Genetics here.

New guidelines for dermatological manifestations in PHTS published

New guidelines for managing dermatological (skin) symptoms in people with PHTS have been published. These guidelines were developed by a group of dermatology and genetics experts, with input from patient advocates.

People with PHTS can develop a range of skin and mouth changes. These may include small, flesh-coloured bumps on the face, rough areas on the hands and feet, growths in the mouth, and other skin changes. Some changes become more noticeable with age. Recognising these signs can help doctors identify PHTS and ensure that people receive appropriate health monitoring.

The guidance is based on published research and expert agreement. It aims to help healthcare professionals provide more consistent advice to children and adults living with PHTS.

The publication highlights the value of coordinated care involving dermatologists, genetic specialists and other members of the healthcare team. It also identifies the need for further research to understand skin-cancer risks and determine which treatments work best for people with PHTS.

See the full guidelines in the Journal of the American Academy of Dermatology here.

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 ongoing PHTS studies, trials and registries
- Links to PHTS patient organisations around the world
- Learn more about our research, funding, and making a donation



PTEN RESEARCH
Funding research to support people with PHTS

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