

## What is PTEN Hamartoma Tumour Syndrome?

PTEN Hamartoma Tumour Syndrome, (PHTS or PTEN syndrome for short) is a rare condition caused by a change in the genetic code in the PTEN gene. You might hear other names including Cowden Syndrome (CS) and Bannayan Riley Ruvulcaba Syndrome (BRRS) being used. These were described in the years before genetic testing. They were brought under the umbrella term PHTS after the genetic cause in the same gene was found.

PHTS is thought to affect around 200-300 people in the UK, although the exact number of people with this diagnosis is not known.

The change in the genetic code can be inherited from parent to child, or it can happen spontaneously for the first time in a person. This is called a de novo variant.

People with PHTS usually have a big head (macrocephaly). They sometimes have developmental delay, autism, problems affecting their skin including unusual pigmentation and wart-like lesions, fatty lumps called lipomas or overgrowth of blood vessels (vascular growths or malformations).

People with PHTS are at risk of developing cancers of the breast, thyroid, kidney (renal), bowel, womb lining (endometrial) and skin earlier in their lifetime than the general population. For this reason, regular surveillance tests to try to identify these different cancers is recommended.

## Contact us

Please do feel free to contact us if you have any queries:

 [www.ptenuki.org](http://www.ptenuki.org)

 [www.facebook.com/ptenuki](https://www.facebook.com/ptenuki)

 [info@ptenuki.org](mailto:info@ptenuki.org)

## Donate or Fundraise for us

We could not provide the support to our community without the vital fundraising efforts of our supporters.

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**Events**

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We would like to take this opportunity to say thank you to everyone that has contributed to our fundraising efforts and is thinking of doing so in the future. We could not provide the support to our community without your generous support.



# PTEN UK and Ireland Patient Group

*The support group for all patients, family members and carers in the UK and Ireland affected by a PTEN gene mutation*



## Our Vision

The patient group's purpose is to improve the lives of patients, parents and carers of all ages, in the United Kingdom and Ireland, who are affected by PTEN genetic alterations, PTEN Hamartoma Tumour Syndrome (PHTS), which includes Cowden Syndrome (CS), and Bannayan-Riley-Ruvalcaba Syndrome (BRRS); through better patient support, increased awareness, more accurate and accessible information, earlier diagnosis and intervention, greater research into treatment and prevention, and improved coordination of care.

## Our Official Charitable Objectives

### Patient Support

To promote and protect the health of persons affected by PTEN Hamartoma Tumour Syndrome and related syndromes in the UK and Ireland by the provision of support, information and practical advice for such persons, their families and carers.

### Improved Healthcare

To improve the healthcare of persons affected by PTEN Hamartoma Tumour Syndrome and related syndromes in the UK and Ireland through advancing the education and awareness of medical professionals and the general public.

### Research

To support and promote research, especially but not exclusively into the diagnosis, treatment and prevention of PTEN Hamartoma Tumour Syndrome and related syndromes in the UK and Ireland.

## The support we offer

Our first, and priority, mission is to support all those affected by a PTEN gene variant. We do this through providing useful resources on our website, annual patient meetups, patient grants and online wellness events for example.

We have lots of information on our website, including useful resources for you to download.

Please visit: [www.ptenuki.org](http://www.ptenuki.org)



You can access and download various PTEN screening guidelines via our website:

<https://ptenuki.org/screening-guidelines/>

eventbrite

For more information on upcoming events visit:



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## PTEN Counselling service

We are pleased to be working in partnership with Rareminds to offer free, confidential counselling for those impacted by PTEN Hamartoma Tumour Syndrome (PTEN), be it patients, family members or carers. Up to 12 sessions can be provided online or by telephone.

Living with PTEN happens within the context of your everyday life, it's inevitable that the 'ups and downs' of both impact on each other. You can talk to our counsellors about anything that you are struggling with and which may influence your ability to live as well as possible with the impact of PTEN.

This might include:

- Your family relationships
- Anxiety or low mood
- Feeling different, lonely or isolated
- Your relationship with health care professionals, or navigating health and social care systems
- Dealing with difficult, unpredictable or increasing symptoms
- The psychological burden of being the 'PTEN expert'
- Coping with multiple unpleasant tests, surgery or treatment
- Worries and uncertainty about the future
- The psychological and practical implications of inheritance

It's not always straightforward to know why you feel as you do. PTEN Counsellors provide a safe, confidential relationship to work through difficult thoughts or feelings, think about options that are open to you, and explore new ways of approaching your difficulties.

To apply for the FREE PTEN counselling service please email [info@ptenuki.org](mailto:info@ptenuki.org)