Your next steps

Find a specialist
All patients should be seen in a clinical genetics department or consultation.

Follow medical guidelines
A surveillance should be undertaken by a specialist according to the guidelines.

Monitor symptoms
Keep track of new or changing health issues.

Consider treatment options
Discuss available therapies and potential surgical interventions.

Stay updated on PHTS research
Clinical trials may offer new treatment possibilities.

Find a patient organisation
They can provide guidance and support.

Visit **www.genturis.eu** for medical guidelines and expert centres.

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

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European Reference Network

Funded by the European Union

Patient information

PTEN hamartoma tumour syndrome (PHTS)

If you or a family member has been diagnosed with *PTEN* hamartoma tumour syndrome (PHTS) this guide provides clear medical information and practical next steps.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

What is PTEN hamartoma tumour syndrome (PHTS)?

PHTS is a rare genetic condition caused by a pathogenic variant in the *PTEN* gene. *PTEN* helps control cell growth. When it does not work properly, people may develop benign growths (hamartomas) and have an increased risk of certain cancers, particularly of the breast, thyroid, endometrium, kidney, bowel and skin.

PHTS can affect different parts of the body and the features can vary considerably from one person to another. Common features may include:

- a larger-than-average head size (macrocephaly);
- skin and mucosal changes, such as nodules or papillomas in the mouth, or benign polyps in the gut;
- thyroid nodules or thyroid cancer;
- breast, kidney, bowel or endometrial cancer;
- vascular (blood-vessel) abnormalities; and
- in some children, developmental delay, intellectual disability or autism spectrum disorder.

How is PHTS inherited?

PHTS is usually inherited in an autosomal dominant pattern. A person with a *PTEN* pathogenic variant has a 50% chance of passing it on to each child. *PTEN* pathogenic variants can also arise spontaneously in a child, even if neither parent has PHTS (this is called de novo).

What causes PHTS?

PHTS is caused by change in a gene called *PTEN*. This gene normally acts as a "tumour suppressor", helping to control cell growth and prevent tumours.

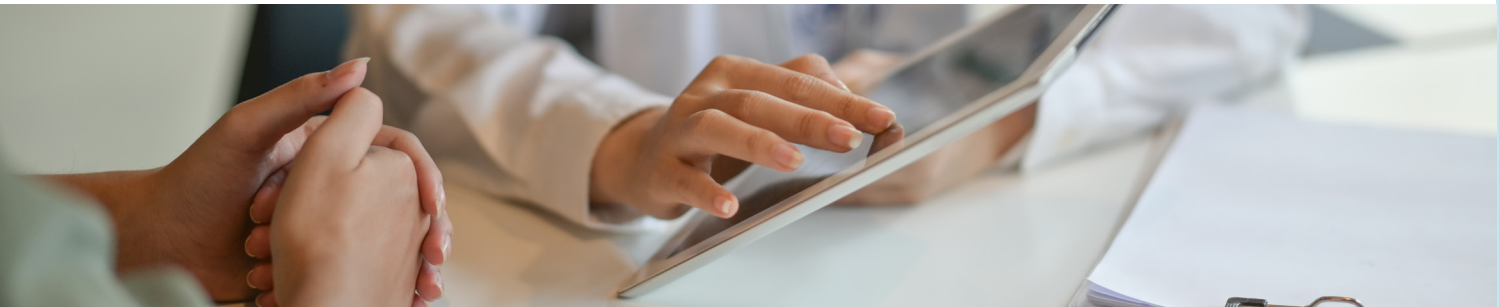
What are the diagnostic/ testing options?

PHTS is confirmed by genetic testing that identifies a pathogenic or likely pathogenic *PTEN* variant. Testing is usually performed from a blood sample. Depending on the person's features, testing may include *PTEN* analysis alone or as part of a broader genetic panel. When a *PTEN* variant is identified, genetic counselling and testing of relatives is recommended.

What are the surveillance recommendations?

Current ERN GENTURIS recommendations include:

- Breast: yearly breast MRI from around 25 years and yearly mammography from around 40 years.
- Thyroid: yearly thyroid ultrasound, starting in childhood/adolescence from around 10-18 years.
- Endometrium: yearly gynaecological surveillance from around 35–40 years.
- Kidneys: baseline kidney imaging from around 35–40 years.
- Bowel: baseline colonoscopy from around 35–40 years, with follow-up depending on findings.
- Skin: baseline dermatological examination from around 30 years.



How this diagnosis affects you?

Receiving a genetic diagnosis requires careful medical supervision. Here are key points to consider:

How will PHTS and the associated tumour risk impact your health?

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need a specialist evaluation over time.
- Since PHTS is genetic, family members should also consider genetic testing and surveillance.

What medical steps should I take?

- **Step 1:** Discuss the topics with your General Practicioner (GP) and schedule an appointment with a specialist in PHTS syndrome.
- **Step 2:** Discuss options for genetic testing of relatives
- **Step 3:** Develop a personalised long-term surveillance plan with your doctor.

ERN GENTURIS provides information and resources to help you understand this condition and guide your medical team toward the best possible care.

More information on **PHTS**: