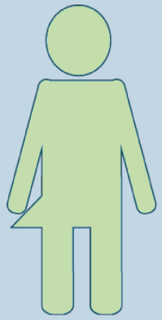
ERN GENTURIS patient journey for cancer risk in *PTEN* hamartoma tumour syndrome (PHTS)

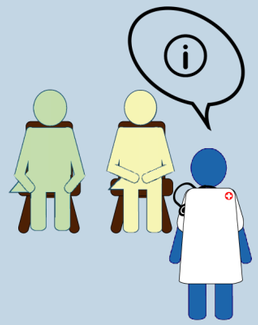
Affected person with cancer (and/or multiple PHTS symptoms) and unknown genetic status

Asymptomatic relative of a PHTS patient with unknown genetic status



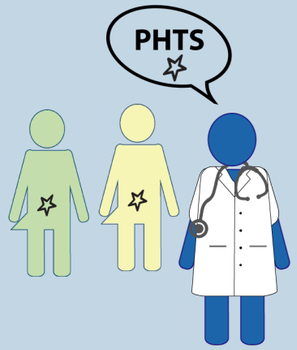
Suspicion of PHTS - Eligible for germline genetic testing

Genetic counselling

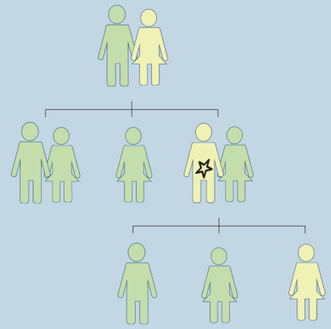


Germline genetic testing

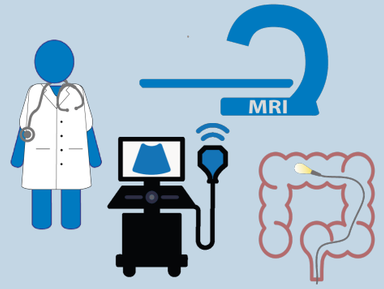
Receiving the results



Cascade testing (testing of blood relatives)

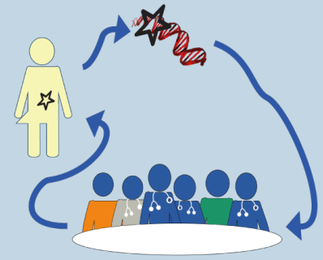


Surveillance for early cancer detection
breast, thyroid, endometrial, kidney, colorectal and skin

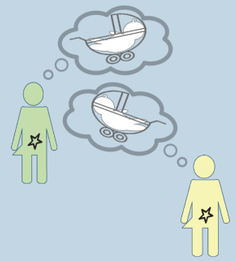


Prevention

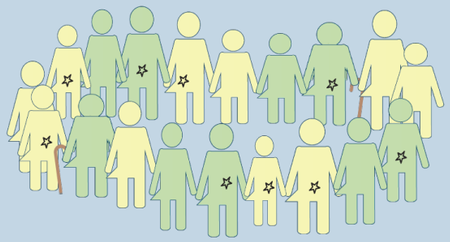
Personalised cancer treatment

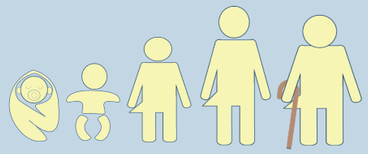


Family planning

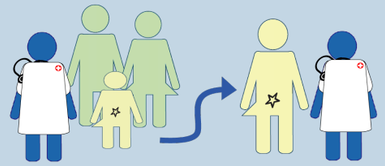


Peer support and emotional support





PHTS care throughout life with a focus on the transition from paediatric to adult care



European Reference Network

Genetic Tumour Risk Syndromes (ERN GENTURIS)

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25-08-2026 version 2.0
approved 08-09-2026

Disclaimer: This patient journey is intended as a general overview of the clinical and diagnostic pathway for *PTEN* hamartoma tumour syndrome (PHTS) or its suspicion, focussing on cancer risk. It should not replace individual clinical and genetic counselling at an expertise centre. It also does not intend to address all specific challenges of the complex condition of PHTS. Specific clinical guidelines, diagnostic criteria and nomenclature may change at short notice and therefore are only referred to in this patient journey.

Affected person with cancer (and/or multiple PHTS symptoms) and unknown genetic status

This refers to someone who has been diagnosed with cancer* and/or multiple PHTS symptoms (see eligible) but has not yet had genetic testing.

* The types of cancer associated with PHTS include breast, thyroid and endometrial (uterine) cancer and to a lesser extend kidney, and colorectal cancer, and skin melanoma.

Asymptomatic relative of a PHTS patient with unknown genetic status

This refers to someone who has not had cancer and/or multiple PHTS symptoms themselves, but has a family member with PHTS, or a family history that suggests PHTS may be present. They have not yet had genetic testing, so it is not known whether they carry a genetic change.

Suspicion of PHTS - Eligible for germline genetic testing

Germline genetic testing (inherited DNA testing) for PHTS should be considered for someone who has several of the following:

- breast, thyroid, endometrial (uterine), kidney, or colorectal cancer or skin melanoma, especially if they occur at a young age
- specific skin or mucosal features or certain non-cancerous growths, such as vascular malformations and/or gastrointestinal polyps
- neurodevelopmental conditions including autism spectrum disorders together with a larger-than-average head size.

Testing for PHTS may be relevant if:

- there is genetically diagnosed PHTS in the family

In selected cases, testing might be offered if there is a family history of a combination of previously mentioned symptoms (see above) that suggest PHTS may be present and affected relatives are not available for testing.

Genetic counselling

Individuals who are eligible for germline genetic testing should receive clear information both before and after testing. This helps them understand what the results may mean and what steps to take next. Genetic counselling typically covers:

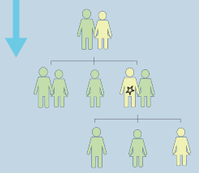
- what PHTS is, how it develops, and the symptoms or health issues it can cause over time
- what surveillance and follow-up are recommended and what to be careful about
- treatment options for any specific health problems related to PHTS
- how the genetic counselling and testing process works
- what the test results could mean for the patient and for their family members
- possible legal, social, insurance or financial considerations related to a diagnosis
- emotional support options including peer support (www.genturis.eu, section patient area)

Germline genetic testing and receiving the results

General information about genetic testing can be found on: https://www.coe.int/en/web/bioethics/information-brochure-on-genetic-tests-for-health-purposes

When genetic test results are available, they should always be discussed with the patient during a post-test genetic counselling session.

A diagnosis of PHTS can only be made if a disease-causing change (pathogenic variant / "mutation") is found in the *PTEN* gene.

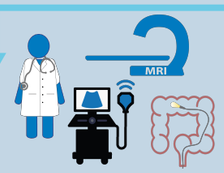


Cascade testing (testing of blood relatives)

Cascade testing is the process of providing genetic counselling and germline genetic testing to family members at risk of having inherited PHTS, after the diagnosis has been made in a patient. A clinical geneticist will look at the family history and decide for which family members germline genetic testing would be relevant. The clinical geneticist will also explain how these family members can arrange their own genetic counselling and testing. **Identifying relatives who carry the same genetic change allows them to receive the right follow-up care, which can improve health outcomes and, in some cases, save lives.**

PHTS follows an autosomal dominant pattern of inheritance. This means that a person with PHTS has a 50% chance of passing the condition on to each child.

Children (minors) may be eligible for genetic testing as in some countries, surveillance for patients with PHTS starts during childhood.



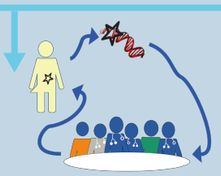
Surveillance for early cancer detection

Surveillance programmes for people with a genetic tumour risk syndrome aim to detect precancerous lesions or cancer as early as possible. Early detection usually means treatment can begin sooner, which often leads to better outcomes. For this reason, it is important to take part in the recommended surveillance programme on a regular basis.

There are specific surveillance programmes for people with PHTS, but these can vary from one country to another. Patients should ask their physician about recommendations in their country and expert centres can also provide information. European-level recommendations can be found on the ERN GENTURIS website (www.genturis.eu, section guidelines), in the ERN GENTURIS cancer surveillance guideline for individuals with PTEN hamartoma tumour syndrome and in the PHTS care pathway, including what to look for and when to refer for non-cancerous health issues.

Prevention

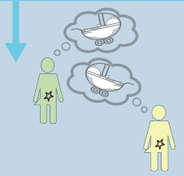
Women with PHTS may choose to have a risk-reducing mastectomy (operation to remove breast tissue) to lower their chance of developing breast cancer. This is a personal decision and should be discussed with healthcare professionals who can explain the potential benefits and risks.



Personalised cancer treatment

Outside of clinical trials (health studies), individuals with PHTS are generally treated with the same cancer treatments used for other patients, including systemic therapies, such as chemotherapy, targeted therapy, endocrine treatment, or immunotherapy. However, it is important that the medical team is aware of the PHTS diagnosis, as this may influence decisions about surgical planning and other aspects of care.

Because PHTS can affect different parts of the body, treatment is best managed by a multidisciplinary team. This means specialists from all the relevant fields work together to ensure that care is well-coordinated and tailored to the patient's needs.



Family planning

PHTS is an inherited condition, which means it can be passed on from parent to child. For some people, this may influence decisions about family planning.

When appropriate, individuals with PHTS should be offered a referral for reproductive counselling early in their patient journey (ERN GENTURIS guideline on counselling on reproductive options for individuals with a cancer predisposition syndrome (including genturis)).

Depending on the country there might be options for prenatal testing during pregnancy or preimplantation genetic testing as part of IVF. Financial coverage for reproductive options through the public healthcare system and the legal regulations can vary from one country to another.

Peer support and emotional support

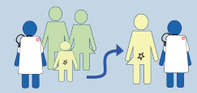
Individuals with PHTS and their family may need peer or emotional support at different points in their live, for example when: - deciding whether to have a genetic test - coping with stress while wait for test results, - talking with relatives about their possible genetic risk, - managing anxiety about untested children or feelings of guilt if a child is affected, - making decisions about family planning, - dealing with the ongoing risk of cancer or other health issues related to PHTS, - receiving a new cancer diagnosis, - coping with stress caused by delays in diagnosis, - choosing between different treatment options, - going through cancer treatment.

Patient organisations can offer information, shared experiences and emotional support. A list of organisations be found on: https://www.genturis.eu/l=eng/patient-area/patient-associations.html

PHTS care throughout life with a focus on the transition from paediatric to adult care

Individuals with PHTS have an increased risk of developing cancer. They can also be affected with other symptoms, such as learning difficulties, neuropsychiatric disorders, blood vessel malformations, skin lesions and low muscle tone. PHTS patients need regular, lifelong, medical follow-up.

As children with PHTS grow into teenagers and young adults, it is important that their care moves smoothly from paediatric services to adult healthcare. This works best when there is a dedicated clinician overseeing the process and when a structured healthcare transition programme is in place. The aim of this transition is to help young people become more confident in managing their own health, improve their quality of life, and reduce the risk of medical complications as they move into adulthood.



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