European Reference Network | Genetic Tumour Risk Syndromes (ERN GENTURIS)

European Reference Networks

PLAIN LANGUAGE SUMMARY OF ERN GENTURIS CANCER SURVEILLANCE GUIDELINE FOR INDIVIDUALS WITH *PTEN* HAMARTOMA TUMOUR SYNDROME (PHTS)

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Disclaimer: The content of this plain language summary is based on the "ERN GENTURIS cancer surveillance guideline for individuals with *PTEN* hamartoma tumour syndrome (PHTS)_final version_20251124 ".

INTRODUCTION

PTEN hamartoma tumour syndrome (PHTS) is caused by a change (pathogenic variant/"mutation") in the *PTEN* gene. Because this gene helps control cell proliferation, changes in *PTEN* can lead to a higher chance of developing certain cancers over a lifetime. People with PHTS have a high risk of breast, thyroid, and endometrial (uterine) cancer, and a moderate risk of kidney, bowel and skin cancer.

PHTS can also cause benign growths, for example in skin and connective tissue, as well as vascular malformations (abnormal blood vessels). Some people with PHTS also experience neurodevelopmental disorders, including autism spectrum disorders.

PHTS is rare, and genetic testing in DNA from blood or equivalent, is needed to confirm its diagnosis. Once a diagnosis is made, regular surveillance (monitoring) is very important, so cancers can be found early, when they are most treatable.

GUIDELINE AIMS

The PHTS guidelines were created to help healthcare professionals provide the most up-to-date and effective cancer surveillance for people with PHTS.

Since the previous European PHTS-guidelines were published in 2020, new research has become available about cancer risks and how helpful different surveillance tests are. This updated guideline (revised in 2024-2025 and published in 2026) brings together the best available scientific evidence and expert agreement, including input from PHTS patient representatives.

SCOPE AND PURPOSE

These guidelines describe which tests are recommended for surveillance, what age to start surveillance and how often each test should be repeated.

The goal is to support early detection and improve long-term health outcomes.

GUIDELINE SUMMARY (KEY RECOMMENDATIONS)

	Surveillance	Interval	From age
Breast cancer	MRI	as for national high-risk guidelines*	25
	Mammography	as for national high-risk guidelines*	40
	Risk reducing surgery offered	Yes	25
Thyroid cancer	Ultrasound	Yearly, frequency decreases to every 2-3 years after 2 ultrasounds with no suspicious findings	10-18
Endometrial (uterus) cancer	Ultrasound (endometrial biopsy can be considered)	Yearly	35 - 40
Renal (kidney) cancer	Baseline imaging	No further surveillance unless required by baseline outcome	35-40
Colorectal cancer (bowel)	Baseline colonoscopy preferably in expert centre	No further surveillance unless required by baseline outcome or colorectal cancer family history	35-40
Skin melanoma	Baseline skin examination by dermatologist or expert	No further surveillance unless required by baseline outcome	30

* For instance, *BRCA1* guidelines. Most national guidelines for *BRCA1* surveillance recommend yearly surveillance with MRI and mammography every 1-2 years.

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