Summary of the cancer surveillance protocol for individuals with *PTEN* hamartoma tumour syndrome (PHTS)

This guideline has been drawn from the best available evidence and the consensus of experts in this area, and it is regularly updated to reflect changes in evidence.

Clinicians are encouraged to follow these recommendations, facilitating evidence-based follow-up for individuals with PHTS.

European Reference Network
for rare or low prevalence complex diseases

Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

www.genturis.eu

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	Surveillance	Interval	From age	Strength*
Breast cancer	MRI	as for *BRCA1***	25	Strong
	Mammography	as for *BRCA1***	40 **	Strong
	Risk reducing surgery offered	Yes	25	Strong
Thyroid cancer	Ultrasound (US)	Yearly, frequency decreases to every 2-3 years after 2 USs with no suspicious findings	10-18	Surveillance: strong, Age: weak, Interval: moderate
Endometrial cancer	Ultrasound (endometrial biopsy can be considered)	Yearly	35-40	Moderate
Renal cancer	Baseline imaging	No further surveillance unless required by baseline outcome	35-40	Moderate
Colorectal cancer (CRC)	Baseline colonoscopy preferably in expert centre	No further surveillance unless required by baseline outcome or CRC family history	35-40	Strong
Skin melanoma	Baseline skin examination by dermatologist or expert	No further surveillance unless required by baseline outcome	30	Moderate

* This grading is based on published articles and expert consensus, see chapter 8.1: strong – expert consensus AND consistent evidence, moderate – expert consensus WITH inconsistent evidence AND/OR new evidence likely to support the recommendation, weak – expert majority decision WITHOUT consistent evidence.

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