

## ERN GENTURIS Plain Language Summary:

### *Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome (PHTS)*

#### INTRODUCTION

PTEN hamartoma tumour syndrome (PHTS), is caused by a change in the PTEN (phosphatase and tensin homolog) gene. PHTS increase the risk of breast, thyroid, endometrial, renal and colorectal cancers. PHTS is rare and its diagnosis relies on genetic testing. Surveillance is considered key to detecting early cancers and being able to treat people.

#### GUIDELINE AIMS

The PTEN hamartoma tumour syndrome (PHTS) Guideline has been created to assist healthcare professionals give the most up-to-date surveillance for individuals with PHTS. This guideline has been drawn from the best available evidence and the consensus of experts in caring for people with PHTS and it is regularly updated to reflect changes in evidence. The expectation is that clinicians will follow this guideline unless there is a compelling clinical reason specific to an individual patient not to.

#### SCOPE & PURPOSE OF THE GUIDELINE

The guideline is intended for the cancer surveillance of individuals with PHTS. For each type of cancer, the guideline states what test should be used for surveillance, what age to start surveillance and how often to repeat investigations.

#### KEY RECOMMENDATIONS

	What Test	How often	Starting at
Thyroid cancer	Ultrasound	Every year	18 yrs.
Breast cancer	MRI Mammography	Every year Every 2 yrs.	30 yrs.
Renal cancer	Ultrasound	Every 2 yrs.	40 yrs.
Endometrial cancer	Not recommended	*if screened then: Ultrasound: yearly	(40)*
Colorectal cancer	Follow general population screening guidelines	-	-

In addition to the tests listed above the guideline recommends that risk reducing breast surgery can be offered to affected women.

**Disclaimer:** The content of this summary is based on the "GENTURIS Cancer surveillance guideline for individuals with PTEN Version 14 (05.09.2019)".