PTEN WOMEN

The medical management guidelines from the National Comprehensive Cancer Network (NCCN) for patients with a PTEN pathogenic / likely pathogenic variant are listed in this document.

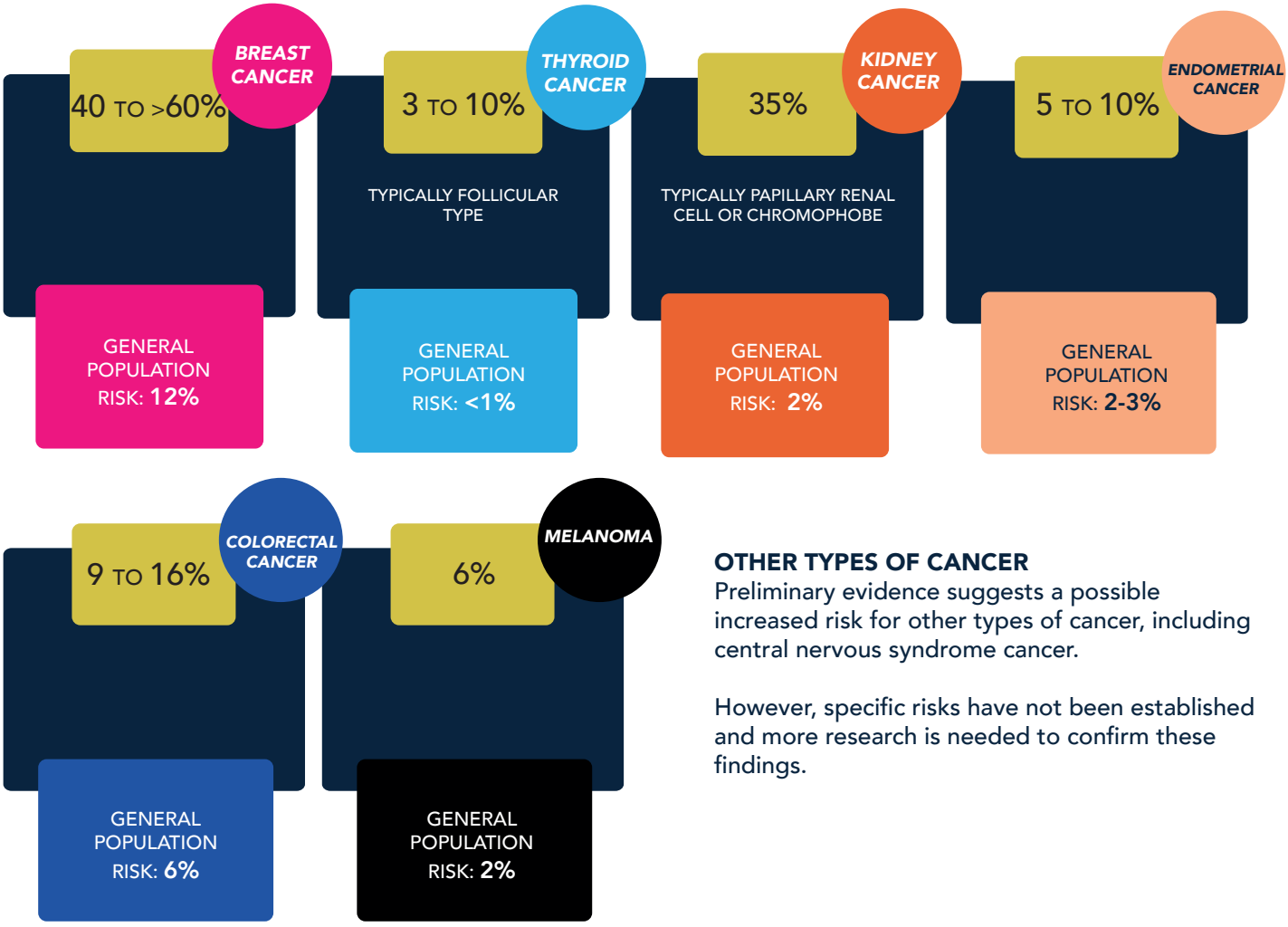
This overview is for informational purposes and does not constitute a personalised recommendation. Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another. The specific references should be consulted for more details before developing a treatment plan.

In addition, the information available on hereditary cancer susceptibility genes is constantly evolving and it is recommended to check this information annually as the management guidelines may change in the future.

LIFETIME ASSOCIATED RISKS

(UP TO AGE 75)

PTEN Hamartoma Tumor syndrome is rare and it includes Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, PTEN-related Proteus syndrome and Proteus-like syndrome. An individual who is a carrier of a PTEN pathogenic / likely pathogenic variant has an elevated risk of developing benign tumors (called hamartomas) as well as certain cancers during their lifetime.



OTHER TYPES OF CANCER

Preliminary evidence suggests a possible increased risk for other types of cancer, including central nervous syndrome cancer.

However, specific risks have not been established and more research is needed to confirm these findings.

Quebec Breast Cancer Foundation
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PTEN WOMEN

BREAST CANCER | ENDOMETRIAL CANCER

SCREENING

STARTING AT AGE 18
- BREAST AWARENESS
  - ◊ PROMPTLY REPORT ANY CHANGES TO YOUR HEALTHCARE PROVIDER

STARTING AT AGE 25
OR 5-10 YEARS BEFORE THE EARLIEST BREAST CANCER IN THE FAMILY
- CLINICAL BREAST EXAM BY A PHYSICIAN EVERY 6 TO 12 MONTHS

STARTING AT AGE 30-35
OR 5-10 YEARS BEFORE THE EARLIEST BREAST CANCER IN THE FAMILY
- MAMMOGRAM WITH CONSIDERATION OF TOMOSYNTHESIS **AND** BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH CONTRAST EVERY 12 MONTHS (ALTERNATING EACH EXAM EVERY 6 MONTHS)

STARTING AT AGE 75
- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

18 — 25 — 30 — 35 — 75

STARTING AT AGE 35
- CONSIDER SCREENING VIA ENDOMETRIAL BIOPSY EVERY 1 TO 2 YEARS

***NOTE:* THESE SCREENING HAVE NOT BEEN SHOWN TO BE SUFFICIENTLY SENSITIVE OR SPECIFIC IN THE DETECTION OF UTERINE CANCER.**

FOR POSTMENOPAUSAL WOMEN:
- AT THE CLINICIAN'S DISCRETION, CONSIDER TRANS-VAGINAL ULTRASOUND EVERY 12 MONTHS
  - ◊ *THIS SCREENING IS NOT RECOMMENDED IN PREMENOPAUSAL WOMEN*

THYROID CANCER | KIDNEY CANCER | COLORECTAL CANCER | OTHER TYPES OF CANCER

SCREENING

STARTING AT AGE 7
- THYROID ULTRASOUND EVERY YEAR

STARTING AT AGE 18
OR 5 YEARS BEFORE THE EARLIEST CANCER DIAGNOSIS IN THE FAMILY
- COMPREHENSIVE PHYSICAL EXAM, WITH PARTICULAR ATTENTION TO THYROID AND SKIN EXAM EVERY YEAR

7 — 18 — 35 — 40

STARTING AT AGE 35
OR 5-10 YEARS EARLIER THAN THE YOUNGEST COLORECTAL CANCER DIAGNOSIS IN THE FAMILY
- COLONOSCOPY EVERY 5 YEARS OR MORE FREQUENTLY BASED ON SYMPTOMS OR PRESENCE OF POLYPS

STARTING AT AGE 40
- CONSIDER RENAL ULTRASOUND EVERY 1-2 YEARS

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BREAST CANCER

RISK REDUCTION

SURGERY

PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION, HAS BEEN SHOWN TO REDUCE THE RISK OF DEVELOPING BREAST CANCER BY UP TO 90%.

ENDOMETRIAL CANCER

SCREENING

PROMPTLY REPORT ANY ABNORMAL BLEEDING OR POSTMENOPAUSAL BLEEDING TO YOUR HEALTHCARE PROVIDER

- EVALUATION OF BLEEDING SHOULD INCLUDE ENDOMETRIAL BIOPSY.

RISK REDUCTION

SURGERY

CONSIDER THE OPTION OF PROPHYLACTIC HYSTERECTOMY (REMOVAL OF THE UTERUS BEFORE CANCER DEVELOPS) AFTER CHILDBEARING IS COMPLETE.

***NOTE:* OOPHORECTOMY (REMOVAL OF THE OVARIES) IS NOT INDICATED FOR PTHS BUT MAY BE INDICATED FOR OTHER REASONS.**

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MELANOMA

SCREENING

- CONSIDER REFERRAL TO A DERMATOLOGIST FOR WHOLE-BODY SKIN EXAMINATION AND EYE EXAM

RISK REDUCTION

- IT IS RECOMMENDED TO LIMIT EXPOSURE TO UV RADIATION BY AVOIDING EXCESSIVE SUN EXPOSURE, BY WEARING A HAT, SUNGLASSES AND LONG PROTECTIVE CLOTHES; BY APPLYING SUNSCREEN WITH A SUN PROTECTION FACTOR (SPF) OF 30 OR MORE; BY AVOIDING SUN TANNING BEDS AND LAMPS.
- REPORT TO YOUR HEALTHCARE PROVIDER ANY UNUSUAL OR CHANGES IN BEAUTY MARKS OR MOLES.

OTHER TYPES OF CANCER

SCREENING

CONSIDER PSYCHOMOTOR ASSESSMENT IN CHILDREN AND BRAIN MRI IF THERE ARE SYMPTOMS.

References:

Daly M et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 2.2021. November 20, 2020. http://www.nccn.org

Eng C. PTEN Hamartoma Tumor Syndrome. 2001 Nov 29 [Updated 2016 Jun 2]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1488/

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