PTEN MEN

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.

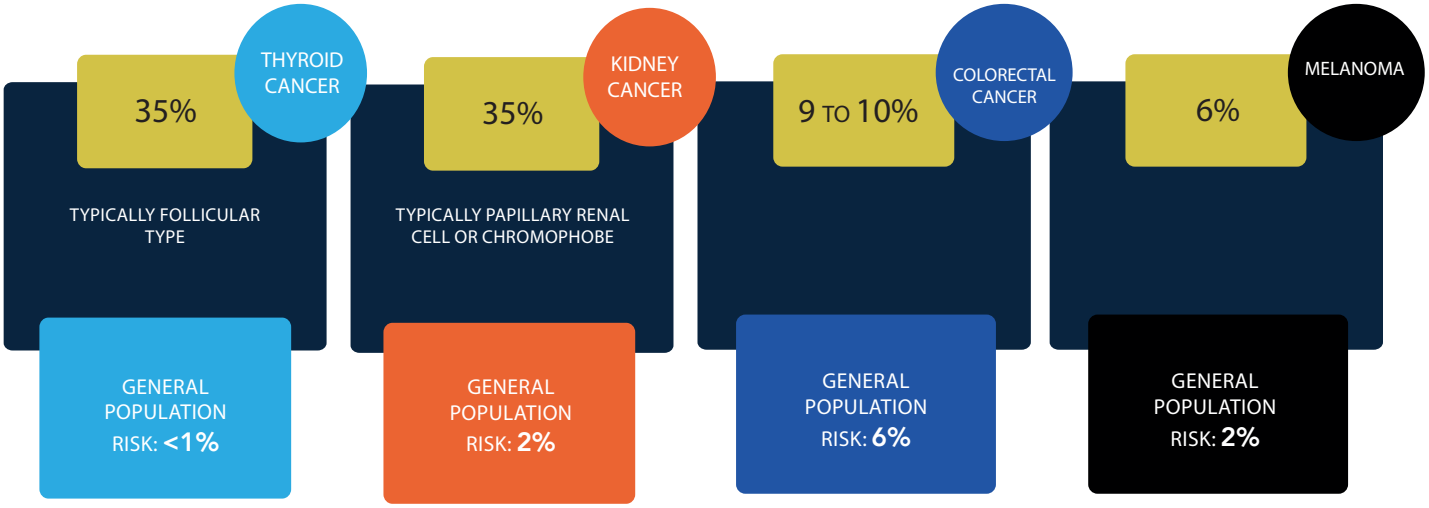
This overview is for informational purposes and does not constitute a personalised recommendation. Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another. The specific references should be consulted for more details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

References:

Daly M et coll. NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 3.2025-March 6, 2025. http://www.nccn.org

Yehia L, Eng C. PTEN Hamartoma Tumor Syndrome. 2001 Nov 29 [Updated 2021 Feb 11]. In: Adam MP, Everman DB, Mirzaa FM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. https://www.ncbi.nlm.nih.gov/books/NBK1488/

Quebec Breast Cancer Foundation
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PTEN MEN

BECAUSE THE MANAGEMENT OF INDIVIDUALS WITH COWDEN SYNDROME IS COMPLEX, IT IS PREFERRED THAT THEY BE FOLLOWED AT A CENTRE WITH EXPERTISE WITH THIS SYNDROME.

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

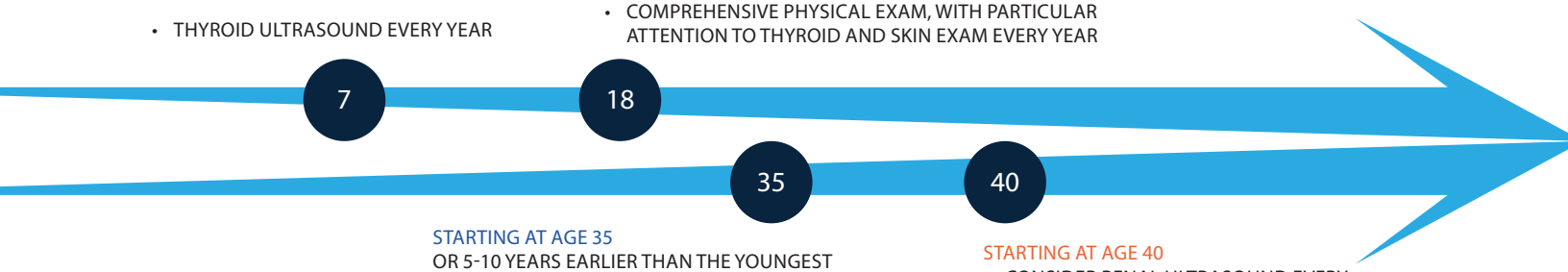
SCREENING

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



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

MELANOMA

SCREENING

- ANNUAL EVALUATION BY A DERMATOLOGIST FOR WHOLE-BODY SKIN EXAMINATION AND EYE EXAM IS RECOMMENDED.

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 AT DIAGNOSIS AND BRAIN MRI IF THERE ARE SYMPTOMS.

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