



# 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.

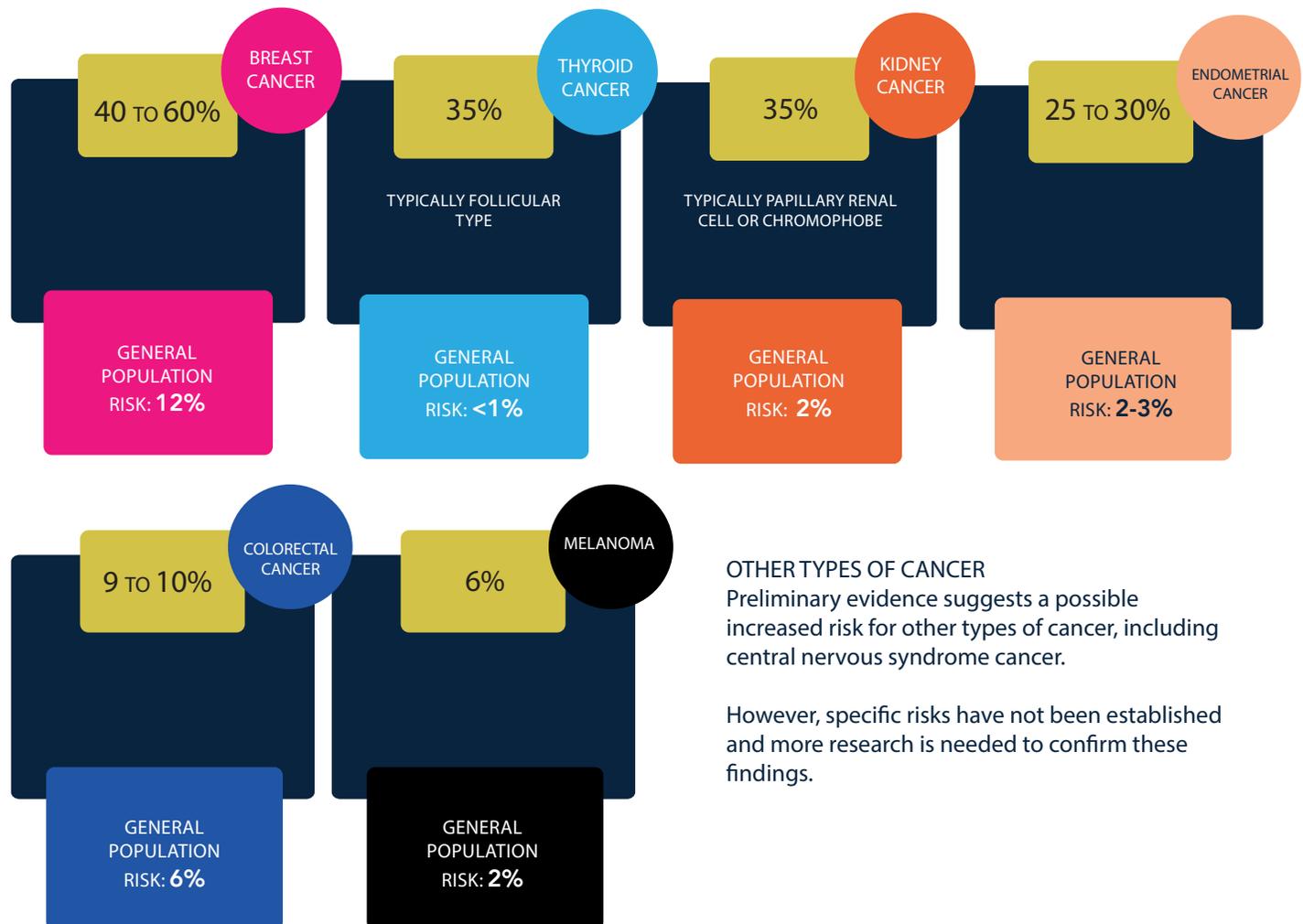
This overview is for informational purposes and does not constitute a personalised recommendation. **Recommended options may vary based on your personal and family history. Access to some options may also vary from one medical center to another.** The specific references should be consulted for more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





# PTEN WOMEN



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.

## BREAST CANCER

## ENDOMETRIAL CANCER

### SCREENING

#### STARTING AT AGE 18

- BREAST AWARENESS
- ◊ PROMPTLY REPORT ANY CHANGES TO YOUR HEALTHCARE PROVIDER

18

#### 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

25

#### STARTING AT AGE 30

OR 10 YEARS BEFORE THE EARLIEST BREAST CANCER IN THE FAMILY

- MAMMOGRAM **AND** BREAST MAGNETIC RESONANCE IMAGING (MRI) WITH AND WITHOUT CONTRAST EVERY 12 MONTHS

30

35

#### STARTING AT AGE 75

- MANAGEMENT SHOULD BE DETERMINED ON AN INDIVIDUAL BASIS

75

#### STARTING AT AGE 35

- CONSIDER SCREENING VIA ENDOMETRIAL BIOPSY EVERY 1 TO 2 YEARS

FOR POSTMENOPAUSAL WOMEN:

- AT THE CLINICIAN'S DISCRETION, CONSIDER TRANSVAGINAL ULTRASOUND EVERY 12 MONTHS
- ◊ THIS SCREENING IS NOT RECOMMENDED IN PREMENOPAUSAL WOMEN

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

## THYROID CANCER

## KIDNEY CANCER

## COLORECTAL CANCER

## OTHER TYPES OF CANCER

### SCREENING

#### STARTING AT AGE 7

- THYROID ULTRASOUND EVERY YEAR

7

#### 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

18

#### 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

35

#### STARTING AT AGE 40

- CONSIDER RENAL ULTRASOUND EVERY 1-2 YEARS

40



## BREAST CANCER

### RISK REDUCTION

#### SURGERY

DISCUSS THE OPTION OF PROPHYLACTIC BILATERAL MASTECTOMY (REMOVAL OF THE BREASTS BEFORE CANCER DEVELOPS), WITH OR WITHOUT RECONSTRUCTION. THIS OPTION HAS BEEN SHOWN TO SIGNIFICANTLY REDUCE THE RISK OF DEVELOPING BREAST CANCER.

## 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

DISCUSS 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.



## 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.

### 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/>