

## Clinician Management Resource for *PTEN* (*PTEN* hamartoma tumor syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a *PTEN* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)<sup>1</sup> in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS <sup>1</sup>	AGE TO START	FREQUENCY
<b>Female Breast Cancer</b>		
Breast awareness <ul style="list-style-type: none"> <li>Women should be familiar with their breasts and promptly report changes to their healthcare provider</li> </ul>	18 years old	Periodic and consistent
Clinical Breast Exam	25 years old, or 5-10 years before the earliest known breast cancer in the family (whichever is first) >75 management should be considered on an individual basis	Every 6-12 months
Breast Screening <ul style="list-style-type: none"> <li>Mammography with consideration of tomosynthesis</li> <li>Breast MRI with contrast</li> </ul>	30-35 years old, or 5-10 years before the earliest known breast cancer in the family (whichever is first)  Women treated for breast cancer, and have not undergone bilateral mastectomy: follow screening as described above.	Every 12 months
	>75 years old: individualized management	Individualized
Discuss option of risk-reducing mastectomy	Individualized	N/A
<b>Endometrial Cancer</b>		
Encourage prompt response to symptoms (e.g., abnormal bleeding)	35 years old	Individualized
Patients are encouraged to keep a calendar in order to identify irregularities in their menstrual cycle	35 years old	Periodic and consistent
Consider endometrial biopsies	35 years old	Every 1-2 years
Transvaginal ultrasound may be considered in postmenopausal woman	35 years old	Clinician's discretion
Discuss option of hysterectomy upon completion of childbearing	35 years old	N/A
<b>Thyroid Cancer</b>		
Comprehensive physical exam, with particular attention to thyroid exam	18 years old, or 5 years before the youngest age of diagnosis of <i>PTEN</i> hamartoma tumor syndrome-related cancer in the family (whichever is first)	Every 12 months
Thyroid ultrasound	7 years old	Every 12 months

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<b>Colorectal Cancer</b>		
Colonoscopy	35 years old unless symptomatic, or if close relative with colorectal cancer before age 40, then start 5-10 years before the earliest known colorectal cancer in the family	Every 5 years, or more frequently if patient is symptomatic or polyps found
<b>Kidney Cancer</b>		
Renal ultrasound	40 years old	Every 1-2 years
<b>Melanoma</b>		
Dermatologic examinations	At time of diagnosis	Annual
<b>Other Cancers</b>		
Psychomotor assessment in children and brain MRI if there are symptoms	In childhood (at diagnosis)	Clinician's discretion

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V1. 2021. © National Comprehensive Cancer Network, Inc. 2020. All rights reserved. Accessed October 1, 2020. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.