Mass General Brigham
Mass General Cancer Center

PTEN gene: what you need to know

What does it mean to have a *PTEN* pathogenic variant?

Having a *PTEN* pathogenic variant, also known as a mutation, causes a cancer predisposition condition called *PTEN* hamartoma tumor syndrome (PHTS). Cowden syndrome is the most common subtype of PTHS.

What is my risk for cancer if I have a *PTEN* pathogenic variant?

If you have a *PTEN* pathogenic variant, you have an increased risk of developing certain types of cancer. However, not everyone who has a pathogenic variant will develop cancer.

Lifetime Cancer Risks

	General Population	*PTEN* Pathogenic Variant
Breast cancer (assigned female at birth)	10-12%	25-60%
Endometrial (uterine) cancer	2-3%	5-10%
Thyroid cancer (typically follicular subtype)	<1%	3-10%

Some patients with Cowden syndrome have a benign tumor in the cerebellum (brain), known as Lhermitte-Duclos disease (LDD). There are limited data suggesting some families with a *PTEN* pathogenic variant have an increased risk to develop colon cancer, melanoma and renal cell cancer.

Are there other symptoms of Cowden syndrome?

Other features of Cowden syndrome include thyroid nodules, macrocephaly (large head size), specific skin findings (trichilemmomas and papillomatous papules), lipomas, hamartomatous colon polyps, vascular anomalies, and autism spectrum disorders.

If I have a *PTEN* pathogenic variant, what is the chance my family members will have it too?

There is a 50% chance that a person with a pathogenic variant will pass it on to each of their children. In most cases, siblings of a person with a pathogenic variant each have a 50% chance to have the pathogenic variant. Additionally, other family members are at risk to have the pathogenic variant.