



## ***PTEN* gene: What You Need to Know**

### **What does it mean to test positive for a *PTEN* gene mutation?**

Mutations in the *PTEN* gene cause a cancer predisposition condition called *PTEN*-hamartoma tumor syndrome (PHTS). Cowden syndrome is one of the PHTS syndromes.

### **What is my risk for cancer if I have a *PTEN* gene mutation?**

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

### **Lifetime Cancer Risks**

	General Population	<i>PTEN</i> Gene Mutation
Female breast cancer	10-12%	25-85%
Endometrial (uterine) cancer	2-3%	5-30%
Thyroid cancer (typically follicular)	<1%	5-38%

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* mutation 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 macrocephaly (large head size), specific skin findings (trichilemmomas and papillomatous papules), lipomas, hamartomatous colon polyps, and autism spectrum disorders.

### **What is the chance that my family members will have a *PTEN* mutation if I test positive?**

There is a 50% chance that a person with a mutation will pass it on to each of his/her children. In most cases, brothers and sisters of a person with a mutation have a 50% chance to have the mutation. Additionally, other family members are at risk to have the mutation.