What you need to know BRCA1 and BRCA2 genes

What does it mean to have a BRCA1 or BRCA2 pathogenic variant?

Having a *BRCA1* or *BRCA2* pathogenic variant, also known as a mutation, causes a cancer predisposition condition called Hereditary Breast and Ovarian Cancer (HBOC) syndrome.

What is my risk for cancer if I have a BRCA1 or BRCA2 pathogenic variant?

If you have a *BRCA1* or *BRCA2* 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	BRCA1 or BRCA2 Pathogenic Variant
Breast cancer (assigned female at birth)	10-12%	>60%ª
Ovarian cancer	1-2%	13-58%
Breast cancer (assigned male at birth)	<1%	Up to 10%
Pancreatic cancer	1.6%	Up to 5-10%
Prostate cancer	11.6%	Up to 61%

*Estimated risk to develop a second primary breast cancer within 20 years is 26-40%.

How will the laboratory identify BRCA1 or BRCA2 pathogenic variant?

Your genetic counselor will determine the most appropriate test(s) for you based on your personal and family history.

<u>BRCA1 and BRCA2 gene sequencing and deletion/duplication testing</u>: This complete testing of both BRCA1 and BRCA2 detects most pathogenic variants. Deletion/duplication testing is also known as rearrangement analysis.

<u>Ashkenazi Jewish panel testing</u>: This test screens for the three founder mutations that account for the majority of *BRCA1* and *BRCA2* pathogenic variations among individuals of Ashkenazi Jewish (Eastern European) ancestry.

If I have a *BRCA1* or *BRCA2* pathogenic variant, what is the chance my family members will have it?

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 have a 50% chance to have the pathogenic variant. Additionally, other family members are at risk to have the pathogenic variant.

