MASSACHUSETTS GENERAL HOSPITAL
CANCER CENTER

BRCA1 and *BRCA2* genes: What You Need to Know

What does it mean to test positive for a *BRCA1* or *BRCA2* gene mutation?

Mutations in the *BRCA1* and *BRCA2* genes cause 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* mutation?

If you have a *BRCA1* or *BRCA2* 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	*BRCA1* or *BRCA2* Gene Mutation
Female breast cancer	10-12%	50-85%
Second primary breast cancer	up to 15%	40-60%
Ovarian cancer	1-2%	10-60%
Male breast cancer	<1%	Up to 10%
Prostate cancer	16%	Increased [a]

[a] Limited data available.

In certain families with *BRCA1* or *BRCA2* mutations there is an increased risk for pancreatic cancer.

How will the laboratory identify *BRCA1* or *BRCA2* gene mutations?

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

BRCA1 and BRCA2 gene sequencing and deletion/duplication testing: This complete testing of both *BRCA1* and *BRCA2* detects the majority of mutations. Deletion/duplication testing is also known as rearrangement analysis.

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

Single-site testing (known familial mutation): If a relative has already tested positive for a *BRCA1* or *BRCA2* gene mutation, single-site testing is typically the most appropriate test. Single-site testing only detects the known familial mutation in either *BRCA1* or *BRCA2*.

What is the chance that my family members will have a *BRCA1* or *BRCA2* 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.