

# BRCA1/BRCA2 Analysis



## Features of Hereditary Breast and Ovarian Cancer

*BRCA1/BRCA2* analysis may be appropriate if your personal and/or family history is suggestive of a hereditary predisposition to breast and ovarian cancer. This includes:

- Breast cancer diagnosed under 50 years of age or triple negative breast cancer diagnosed under the age of 60
- Multiple cancers in one person, either of the same origin (such as two separate breast cancers) or of different origins (such as breast cancer and ovarian cancer)
- Ovarian cancer or male breast cancer at any age
- Multiple relatives diagnosed with the same or related cancers (including breast, ovarian, pancreatic and/or metastatic/aggressive prostate) on the same side of the family and spanning multiple generations
- Ashkenazi Jewish ancestry with a history of breast, ovarian or pancreatic cancer
- Metastatic/aggressive prostate cancer

## The *BRCA1* and *BRCA2* Genes

Many genes have been associated with an increased risk of breast and/or ovarian cancer. *BRCA1* and *BRCA2* are categorized as High-Risk genes and are associated with an increased risk of certain types of cancer, including breast and ovarian cancer. Your healthcare provider may order additional testing depending on your personal and/or family history of cancer.

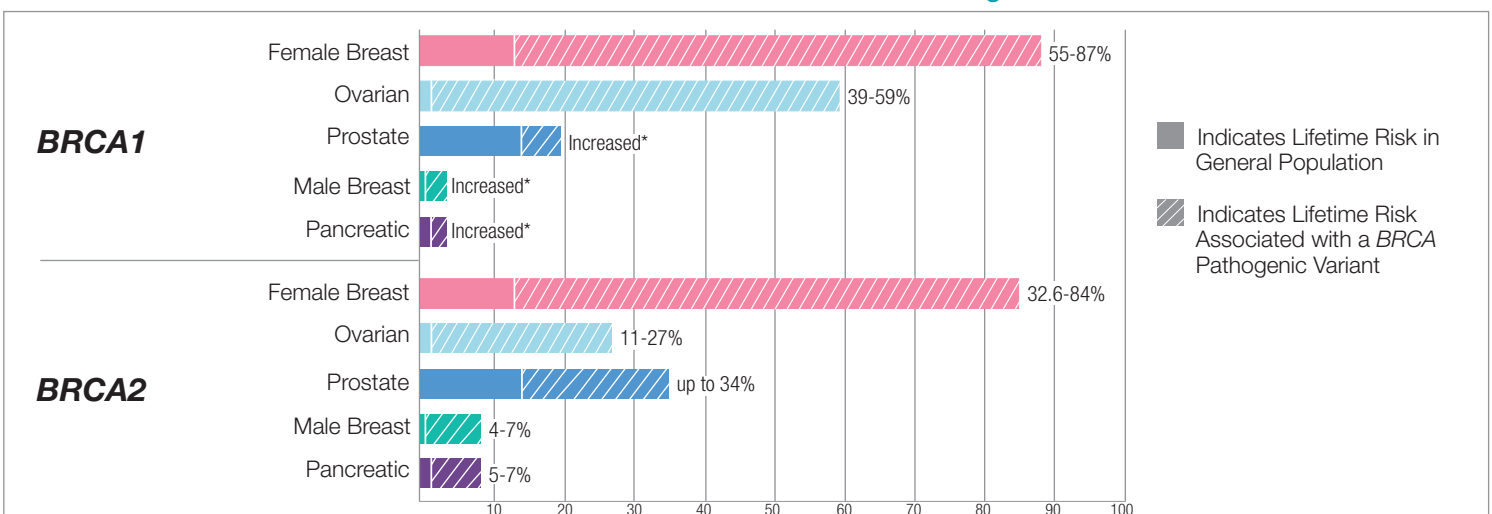
**High-Risk Genes** Well-studied • Greater than 4-fold risk of developing one or more cancers • Can cause a moderate risk for other cancers • National or expert opinion guidelines for screening and prevention are established

## Lifetime Cancer and/or Tumor Risks

Gene	Lifetime Cancer and/or Tumor Risks*
<i>BRCA1</i>	Female breast (55-87%), Ovarian (39-59%), Prostate, Male breast, Pancreatic, Fallopian tube, Primary peritoneal, Endometrial
<i>BRCA2</i>	Female breast (32.6-84%), Prostate (up to 34%), Ovarian (11-27%), Pancreatic (5-7%), Male breast (4-7.1%), Melanoma, Fallopian tube, Primary peritoneal, Endometrial

\*Most commonly associated cancers/tumors listed; lifetime risks provided when available. Risks relate to carriers of a single pathogenic variant.

## Lifetime Risk of Select Cancers Associated with *BRCA1* and *BRCA2* Pathogenic Variants



Data pulled from: Surveillance, Epidemiology, and End Results (SEER) Program of the National Cancer Institute. SEER Cancer Statistics Review, 1975-2012: Lifetime Risk Tables (URL: <http://surveillance.cancer.gov/devcan>) [October 2017 accessed]

\*Lifetime risks of cancer are known to be significantly increased above the general population risk although a precise lifetime risk is unknown.

## Possible Outcomes of Genetic Testing:

There are four possible outcomes of genetic testing: positive (pathogenic variant), likely pathogenic variant, variant of uncertain significance (VUS), and negative. Genetic counseling is recommended prior to genetic testing to understand the benefits and limitations of testing.

A **positive** result indicates a genetic variant (change) was identified in a specific gene and that variant is pathogenic (harmful). With a **positive** test result, the risk to develop a particular disease (in this case, cancer and/or tumors) is increased.

A **likely pathogenic variant** result indicates that there is a variant in a specific gene for which there is significant, but not conclusive, evidence of an increased risk to develop a particular disease (in this case, cancer and/or tumors).

A **variant of uncertain significance (VUS)** result means that a change in a specific gene was identified, however the effect of the variant cannot be clearly established. There may be conflicting or incomplete information in the medical literature about this variant and its association with an increased risk of cancers and/or tumors is unknown. In other words, it cannot be determined yet whether this variant is associated with an increased risk of cancer and/or tumors or it is a harmless (normal) variant.

A **negative** result means that no reportable variants were identified.

## Medical Management Based on Genetic Test Results

Clinical guidelines are available which provide options and recommendations for patients who have a **positive** (pathogenic variant) test result indicating an increased risk for cancer and/or tumors. Guidelines and recommendations for early detection and/or risk reduction are specific to the gene in which the pathogenic variant was found.

### Recommendations may include:

- Breast awareness, including breast self-examination for both men and women
- Clinical exams, such as skin and/or breast exams
- Imaging exams, such as a mammogram, MRI, and/or ultrasound
- Risk-reducing medications and/or surgery

If you have a **positive** or a **likely pathogenic variant** result, your test report will include additional information regarding the available medical management options.

If you have a **negative** or a **variant of uncertain significance (VUS)** test result, medical management should be based upon your personal and/or family history of cancer and/or tumors.

Once your test results are available, a discussion with your healthcare provider is recommended to determine the most appropriate medical management options for you and your family.

Regardless of the test results, consider sharing them with your family members so that they may discuss them with their healthcare providers. If you have a **positive** or a **likely pathogenic variant** result, family members are at risk to have the same variant and should consider genetic testing to best understand their chance of developing cancer and/or tumors.

## Resources

### General

American Cancer Society

[www.cancer.org](http://www.cancer.org)

GeneDx

[www.genedx.com/oncology](http://www.genedx.com/oncology)

National Cancer Institute

[www.cancer.gov](http://www.cancer.gov)

### Breast Cancer

Bright Pink

[www.brightpink.org](http://www.brightpink.org)

Facing Our Risk of Cancer Empowered (FORCE)

[www.facingourrisk.org](http://www.facingourrisk.org)

### Find a Genetic Counselor

Canadian Association of Genetic Counsellors

[www.cagc-accg.ca](http://www.cagc-accg.ca)

National Society of Genetic Counselors

[www.nsgc.org](http://www.nsgc.org)



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