

# Result Navigator

## ⊕ Positive Test Result: BRCA1

Positive test results identify a change, or “misspelling,” of DNA that is known or predicted to cause an increased risk for cancer. DNA is the blueprint of life—it carries all of the genetic information needed for our bodies to function. A section of DNA with a specific job is called a gene. A pathogenic variant is a change in a gene that is known to cause increased risk for cancer. A likely pathogenic variant is a change in a gene that is very likely to cause increased risk for cancer.

*After a positive test result, there can be many questions about what to do next.*

This guide is a supplement to Riscover® Hereditary Cancer test results, helping patients and healthcare providers understand the results and together determine the best path forward.

## Navigate Your Results



### *Learn* | page 2

Review the test results along with this guide, and discuss the meaning of these results with your healthcare provider.



### *Plan* | page 3

Develop a healthcare plan for early detection and prevention of specific cancers.



### *Share* | page 4

Understand the risks for other family members, and consider how to inform relatives of their risks.



### *Connect* | page 5

Reach out to medical experts and find local and national groups that can provide more information and support.

## Glossary of Terms

**Breast tomosynthesis:** Advanced three-dimensional (3-D) breast imaging.

**CA-125:** A substance that can indicate the development of ovarian cancer. Testing for CA-125 is performed on a blood sample.

**Chemoprevention:** The use of certain medications to lower the risk or prevent cancer in healthy people.

**Gene:** A section of DNA with a specific job.

**Mastectomy:** Surgical removal of the breast tissue.

**MRI:** An abbreviation for magnetic resonance imaging, a test that provides detailed pictures of a specific part of the body.

**Pathogenic:** Causing or capable of causing disease.

**Risk:** The chance, or possibility, of developing a disease.

**Salpingo-oophorectomy:** Surgical removal of the ovaries and fallopian tubes.

**Transvaginal ultrasound:** An imaging test performed by inserting an ultrasound probe into the vagina to take detailed images of the ovaries and fallopian tubes.

**Variant:** A change, or “misspelling,” in the DNA sequence.

Variants can be benign (not associated with disease), pathogenic (associated with disease), or of uncertain significance.

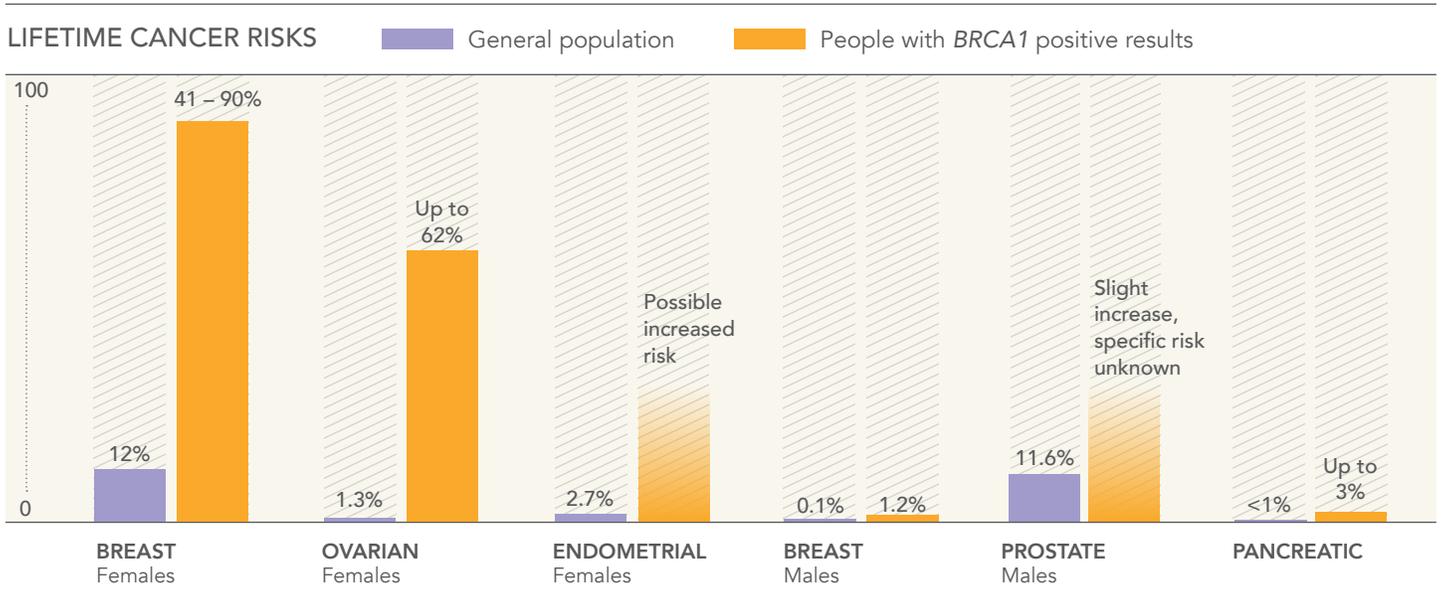


## Your Diagnosis: Hereditary Breast and Ovarian Cancer Syndrome

Hereditary breast and ovarian cancer syndrome (HBOC) is caused by a pathogenic/likely pathogenic variant in the *BRCA1* or the *BRCA2* gene. HBOC is an inherited disorder associated with an increased lifetime risk of breast, ovarian, pancreatic, prostate, and possibly other types of cancer. Women with a *BRCA1* variant may have a slightly increased risk of serous uterine cancer. Of all the genes associated with hereditary breast and ovarian cancer, *BRCA1* and *BRCA2* are the most common. About 1 in 400 people in the general population will test positive for a *BRCA1* or *BRCA2* variant. About 1 in 40 people of Ashkenazi Jewish ancestry will test positive for a *BRCA1* or *BRCA2* variant. Three specific variants (called *BRCA1* c.68\_69delAG, *BRCA1* c.5266dupC, and *BRCA2* c.5946delT) account for >98% of pathogenic variants in the Ashkenazi Jewish population.

## Your Risk

Not all people with positive test results will develop cancer, but the chance for cancer is higher than the general population risk. The graphs below show how much the risk for cancer increases when a pathogenic variant is found. The cancer risks are presented in ranges based on multiple studies of families who have a pathogenic variant. There is not enough evidence available to give a specific risk to a person or family.



## Your Screening and Management Options

Listed below are the intervention options that are recommended by medical experts for individuals who carry this cancer gene variant. Your healthcare provider can explain these options in more detail, and together you can make a plan for cancer prevention and/or early detection.

### BREAST CANCER (FEMALES)

Breast self-exams	Periodic, consistent breast self-exams at the end of your period may be considered.
Clinical breast exam	A clinical breast exam performed by your healthcare provider is recommended every 6 – 12 months, starting at age 25.
Breast MRI	An annual breast MRI is recommended starting at age 25, or earlier if you have a family history of breast cancer diagnosed before age 30. These are usually performed until age 75, and can be considered on an individual basis after that.
Mammogram	Annual mammograms with consideration for breast tomosynthesis are recommended starting at age 30 or earlier based on family history. These are usually performed until age 75, and can be considered on an individual basis after that.
Chemoprevention (i.e., tamoxifen)	Your healthcare provider may prescribe a medication to help prevent breast cancer, based on your personal and family history.
Risk-reducing mastectomy	Risk-reducing mastectomy can be considered on an individual basis.

### OVARIAN CANCER (FEMALES)

Risk-reducing salpingo-oophorectomy (RRSO)	Removal of the ovaries and fallopian tubes, typically performed between 35 – 40 years of age, and after a woman has had her last child.
Transvaginal ultrasound and serum CA-125 (at clinician discretion)	If RRSO is not performed, your healthcare provider may consider a transvaginal ultrasound to screen for ovarian cancer along with a blood test for cancer antigen 125 (CA-125) starting at 30 – 35 years of age.
Risk-reducing medications (birth control pills)	Your healthcare provider may prescribe medication such as hormonal birth control pills to reduce risk for ovarian cancer, based on your personal and family history.

### ENDOMETRIAL CANCER (FEMALES)

Hysterectomy	Although data is limited for this cancer, consider hysterectomy at the time of RRSO.
--------------	--

### BREAST CANCER (MALES)

Breast self-exam	A clinical breast exam performed by your healthcare provider is recommended every 12 months, starting at age 35.
Clinical breast exam	A physical exam performed by your healthcare provider is recommended every 12 months, starting at age 35.

### PROSTATE CANCER (MALES)

PSA blood test and digital rectal exam	Consider a regular blood test for prostate-specific antigen (PSA) along with a digital rectal exam performed by your healthcare provider starting at age 45.
--	--

### PANCREATIC CANCER

No specific guidelines	Your healthcare provider can recommend screening options based on your personal and family history of pancreatic cancer.
------------------------	--

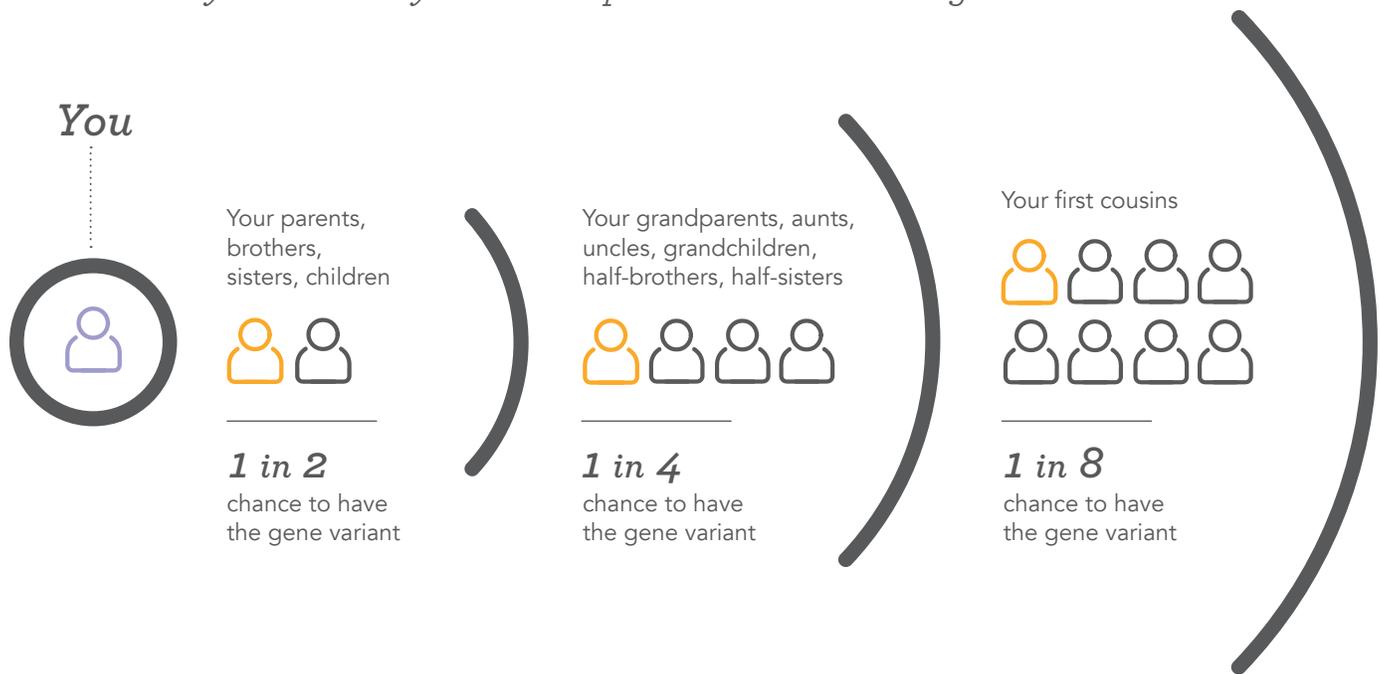
## Your Family Matters

Cancer gene variants are inherited in an autosomal dominant manner. This means that when a parent tests positive, each child has a 1 in 2 chance to inherit the variant. Some people who inherit the variant will develop cancer, and some will not, but the risks are significantly increased compared to the average person. People in the same family may develop different types of cancers at varying ages, and the pattern of cancer and age of onset can differ from family to family.

Gene variants can be passed through families for many generations. When a gene variant is identified in one person in the family, relatives can be tested for just that variant. Sharing information about test results with family members is important, because it allows them to make informed choices about their healthcare and to decide whether they want genetic testing.

Some people who have pathogenic and likely pathogenic variants are interested in learning how to avoid passing these variants on to their children. If you are planning to have a child, you may wish to consult with a reproductive medicine specialist to discuss all of your reproductive options, including use of an egg or sperm donor, preimplantation genetic diagnosis, and prenatal genetic testing.

*The likelihood for family members to carry the same gene variant depends on how closely related they are to the person with a known gene variant.*



## Your Expert Team

As you take your next steps, it can help to have a team of experts on your side. This team may include your primary care provider and a number of specialists who will help along the way.

If you haven't already done so, you may wish to speak with a genetic counselor. Genetic counselors are healthcare professionals with specialized training in medical genetics and counseling. They can help you understand your test results, discuss your healthcare plan, and connect to other resources in your area. Your healthcare provider may be able to provide a referral to a local genetic counselor. You can also find a genetic counselor through the National Society of Genetic Counselors at [www.nsgc.org](http://www.nsgc.org).

For questions about your test results, our genetic counseling team is available to help.  
To reach a genetic counselor, call +1 855-293-2639 and select option 3.

## Helpful Resources

### GeneReviews

[ncbi.nlm.nih.gov/books/NBK1247](http://ncbi.nlm.nih.gov/books/NBK1247)

A detailed clinical summary of hereditary breast and ovarian cancer written for healthcare providers.

### National Comprehensive Cancer Network (NCCN)

[nccn.org](http://nccn.org)

Expert guidelines for cancer screening and treatment. Information available for both healthcare providers and patients.

### Facing Our Risk of Cancer Empowered (FORCE)

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

A patient advocacy group whose mission is to improve the lives of individuals and families affected by hereditary breast, ovarian, and related cancers.

### Bright Pink

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

An organization targeting young women with a mission to save lives from breast and ovarian cancers.

### Sharsheret

[sharsheret.org](http://sharsheret.org)

An organization supporting Jewish women who are diagnosed with breast cancer or at increased risk to develop breast cancer due to their genetics.

### BRCAn't Stop Me

[brcantstopme.wixsite.com/brcantstopme](http://brcantstopme.wixsite.com/brcantstopme)

An organization targeting young adults whose mission is to raise awareness and provide support for people affected by BRCA gene mutations.

## REFERENCES

Hereditary Cancer. FORCE. <http://www.facingourrisk.org/understanding-brca-and-hboc/index.php>.

National Comprehensive Cancer Network. Genetic/Familial High-Risk Assessment: Breast and Ovarian (Version 2.2019). Available from: <http://www.nccn.org/>.

Petrucelli N, Daly MB, Feldman GL. BRCA1 and BRCA2 Hereditary Breast and Ovarian Cancer. 1998 Sep 4 [Updated 2016 Dec 15]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. Available from: <http://www.ncbi.nlm.nih.gov/books/NBK1247/>.