BRCA1/BRCA2 (Selected Variants)

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing certain cancers, including breast cancer (in women and men) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer and certain other cancers. This test includes three genetic variants in the BRCA1 and BRCA2 genes that are most common in people of Ashkenazi Jewish descent.

play+06c0554a05, we could **not determine** if you have the three genetic variants we tested.

This test is intended to detect two variants in the BRCA1 gene and one variant in the BRCA2 gene, but your result could not be determined.

This can be caused by random test error or other factors that interfere with the test. **If you have a personal or family history of cancer, you should talk to a healthcare professional about other testing options.**

How To Use This Test

This test does not diagnose cancer or any other health conditions and should not be used to make medical decisions. Results should be confirmed in a clinical setting before taking any medical action.

Please talk to a healthcare professional if cancer runs in your family, you think you might have cancer, or you have any concerns about your results.

Review the BRCA1/BRCA2 (Selected Variants) tutorial

See Frequently Asked Questions

See Scientific Details for complete Indications for Use statement and full list of Warnings, Precautions, and Limitations

Intended Uses

- Tests for three specific genetic variants: the **185delAG** and **5382insC** variants in the BRCA1 gene and the **6174delT** variant in the BRCA2 gene. These variants are associated with an increased risk of developing certain cancers.
- Provides information on whether a person's genetic result is associated with an increased risk for breast and ovarian cancer and may be associated with an increased risk for prostate cancer and certain other cancers.

Limitations

- Does **not** test for all possible variants in the BRCA1 and BRCA2 genes. More than 1,000 variants in these genes are known to increase cancer risk. Only three of those variants are included in this test.
- Does **not** test for variants in other genes linked to hereditary cancers.
- Does **not** account for non-genetic factors, like environment and lifestyle, that influence overall cancer risk.
- The interpretation of your genetic result depends on the sex you reported in your account settings.

Important Ethnicities

- The variants included in this test are most commonly found in people of **Ashkenazi Jewish** descent. In 23andMe customers of other ethnicities, between 0% and 0.1% of individuals has one of the three variants in this report.
- This test does **not** include the majority of BRCA1 and BRCA2 variants found in people of other ethnicities. Therefore, a "variants not detected" result is less informative for people with no Ashkenazi Jewish ancestry.

We could **not determine** if you have any of the three variants we tested linked to hereditary breast and ovarian cancer.

If you have a personal or family history of cancer, consider talking with a healthcare professional about additional testing.

We could not rule out any of the three variants we tested.

These three variants are most common in people of **Ashkenazi Jewish** descent and do not account for the majority of BRCA1 and BRCA2 variants in people of other ethnicities.

See Scientific Details

In the general population, about **1 in 8 women** develops breast cancer during her lifetime, and about **1 in 80 women** develops ovarian cancer.

Only a small percentage of these cancers are caused by the three genetic variants in this report. Your risk is influenced by many other factors, including lifestyle, family history, and other genetic factors.

See Scientific Details

If you have a personal or family history of cancer, talk to a healthcare professional about other testing options.

A genetic counselor can help you assess your overall cancer risk. Learn more about genetic counseling.

Lifestyle, family history, and other factors can also influence the chances of developing breast and ovarian cancer.

Consult with a healthcare professional before making any major lifestyle changes.

Age

In general, the chances of developing breast and ovarian cancer increase as a woman gets older. Most cases of breast and ovarian cancer are diagnosed after the age of 55.

See Scientific Details for more information

About BRCA1/BRCA2-Related Cancers

BRCA1 and BRCA2 variants are associated with an increased risk for several different cancers, including breast cancer (in women and men) and ovarian cancer. Variants in these genes may also be associated with an increased risk for prostate cancer, pancreatic cancer, and melanoma. The risk estimates below apply to BRCA1 and BRCA2 variants in general, including the three variants in this report.

Lifetime cancer risks

- Women with a **BRCA1** variant have a 45-85% chance of developing breast cancer by age 70 and a 39-46% chance of developing ovarian cancer. They may also have an increased risk for pancreatic cancer.
- Women with a **BRCA2** variant have a 45-85% chance of developing breast cancer by age 70 and a 10-27% chance of developing ovarian cancer. They may also have an increased risk for pancreatic cancer and melanoma.
- Men with a BRCA1 or BRCA2 variant have an increased risk for male breast cancer and may have an increased risk for prostate cancer, pancreatic cancer, and melanoma.
- See Scientific Details to learn more about these risks

When these cancers develop

In general, the chances of developing cancer increase as a person gets older. However, women with a BRCA1 or BRCA2 variant have an increased risk for early-onset breast cancer (before age 45) and multiple breast cancers. Women with a BRCA1 variant may also develop ovarian cancer at an earlier age. Men with a BRCA1 or BRCA2 variant may develop earlier and more aggressive prostate cancer.

How common are BRCA1 and BRCA2 variants?

About 1 in 400 people in the general population has a BRCA1 or BRCA2 variant linked to hereditary breast and ovarian cancer, although most of those variants are not included in this report. Among people of Ashkenazi Jewish descent, about 1 in 40 has a variant (usually one of the three variants in this report).

Screening and prevention

Guidelines recommend that women with a BRCA1 or BRCA2 variant should be screened for breast cancer earlier and more often. However, there are currently no ovarian cancer screening tests that have been proven safe and effective. For women with a BRCA1 or BRCA2 variant, surgery and medication have been shown to be effective in reducing the risk of developing breast and ovarian cancer.

Men with a BRCA1 or BRCA2 variant should be screened for male breast cancer. Screening guidelines for prostate cancer vary.

Always consult with a healthcare professional before taking any medical action.

Read more at: National Cancer Institute* GeneReviews*

Learn more about BRCA1/BRCA2-related cancers.

See our Frequently Asked Questions for more information.

FAQs

If you have a personal or family history of cancer, consult with a healthcare professional.

Print report

Learn more about cancer screening to help you and your doctor create a screening plan that's right for you.

Learn more

Print

BRCA1/BRCA2 (Selected Variants)

Scientific Details

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing certain cancers, including breast cancer (in women and men) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer and certain other cancers. This test includes three genetic variants in the BRCA1 and BRCA2 genes that are most common in people of Ashkenazi Jewish descent.

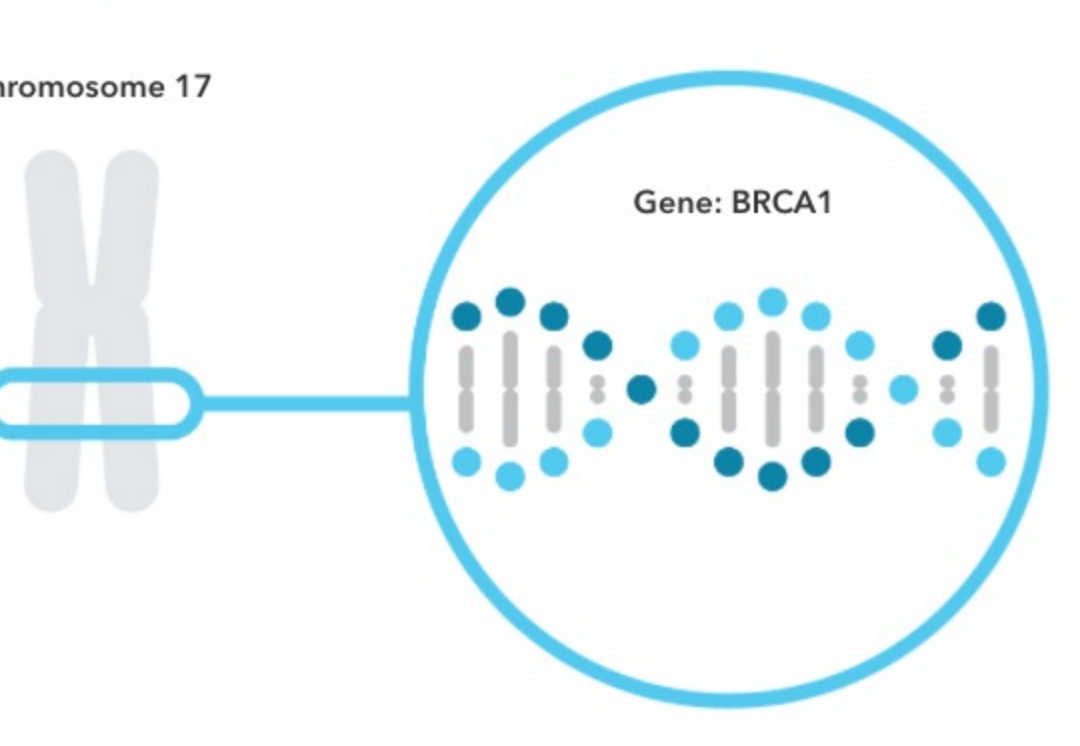
Genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk for certain hereditary cancers.

This report includes two variants in the BRCA1 gene and one variant in the BRCA2 gene. These three variants do not account for the majority of the BRCA1 and BRCA2 variants in the general population. More than 1,000 variants in these genes are known to increase cancer risk.

BRCA1 | BRCA2

The BRCA1 gene contains instructions for making a protein that helps repair damaged DNA. The BRCA1 protein also helps control the process of cell division. Through both of these functions, the BRCA1 protein acts as a tumor suppressor, preventing cells from growing and dividing too rapidly. Certain variants in the BRCA1 gene disrupt the protein's function. This can lead to a buildup of DNA errors, and can cause normal cells to become cancer cells.

Read more at Genetics Home Reference



Your result for the three genetic variants we tested could not be determined.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*	Additional Information
185delAG Gene: BRCA1 Marker: rs386833395	**Not determined**	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 12, 23, 36, 40, 42, 59, 60, 67, 97, 98, 103, 104, 116] | ClinVar
5382insC Gene: BRCA1 Marker: rs80357906	**Not determined**	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 12, 40, 42, 53, 59, 79, 97, 98, 99, 104, 108, 116, 120] | ClinVar
6174delT Gene: BRCA2 Marker: rs80359550	**Not determined**	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 12, 18, 23, 38, 40, 42, 43, 59, 82, 85, 97, 98, 102, 104, 116] | ClinVar

* The percent of 23andMe customers with a variant may not be representative of the general population.

This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides risk estimates for several cancers associated with BRCA1 and BRCA2 variants. In most cases, these estimates represent a general risk for individuals with any BRCA1 or BRCA2 variant, not the specific risk estimates associated with the three variants in this report. This test does not take into account non-genetic factors that influence a person's overall risk for these cancers.

Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Numerical risk estimates are not available for people who have both a BRCA1 and a BRCA2 variant. An interpretation of "increased risk" is provided to people with this result. It is likely that their risk is at least as high as the risk for people with just one variant. More research is needed to understand the risk for people with this result.

For some cancers, numerical risk estimates are not available.

Consider talking to a healthcare professional if you have any concerns about your results.

References [30, 54, 59, 61, 111, 112]

Lifetime risk | Risk by age

The risk estimates shown below represent the proportion of people expected to develop a given cancer during their lifetime. Estimates for the general population are based on observed cancers among people in the United States. Estimates for men and women with a BRCA1 or BRCA2 variant are based primarily on studies of people of European and Ashkenazi Jewish descent. Estimates for people with a BRCA1 or BRCA2 variant represent the risk of developing cancer by the age of 70 (for women) or during their lifetime (for men).

Cancer type	General population	BRCA1 variant	BRCA2 variant
Breast (female)	12.4%	45-85%	45-85%
Ovarian	1.3%	39-46%	10-27%
Breast (male)	0.12%	1-2%	7-8%
Prostate	11.6%	May have an increased risk	Increased risk
Pancreatic	1.6%	May have an increased risk	May have an increased risk
Melanoma	2.2%	Research ongoing	May have an increased risk

See risk estimates by ethnicity for the general population

Other Factors

The three genetic variants in this report are associated with a greatly increased risk for breast and ovarian cancer. They may also be associated with an increased risk for pancreatic cancer and melanoma. However, other factors besides the genetic variants in this report can influence your chances of developing these cancers.

This is not a complete list of other factors.

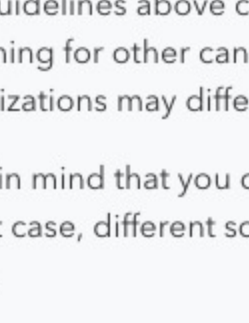
People with multiple risk factors may have a higher risk of developing cancer.

Consult with a healthcare professional before making any major lifestyle changes.

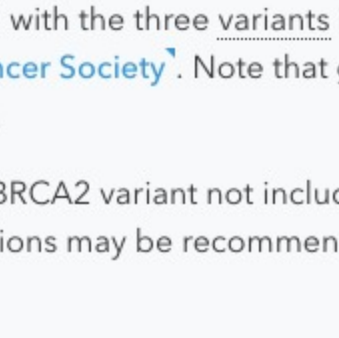
Other Factors	References
Age Like most cancers, the risk of developing breast and ovarian cancer generally increases with age. For the average woman in the U.S., the risk of developing breast cancer by age 40 is 1 in 200. That number rises to 1 in 8 by age 80. For ovarian cancer, the risk is 1 in 1,000 by age 40 and 1 in 100 by age 80. In general, the risks for pancreatic cancer and melanoma also increase with age.	[54, 101, 111]
Family history Most women who develop breast and ovarian cancer don't have a family history of these cancers. However, women whose mothers or sisters have had breast or ovarian cancer are more likely to develop these cancers themselves. For both cancers, the risk is even greater in families with more than one affected family member. The risk is also greater in families with members diagnosed with cancer at an earlier age. This increased risk is likely due to shared genetic and non-genetic factors. A family history of pancreatic cancer or melanoma also increases a person's risk for those cancers.	[3, 63, 68, 76, 88, 90, 118]
Obesity After menopause, being overweight increases a woman's chances of developing breast cancer. Weight gain during adulthood is also associated with an increased risk. In addition, obesity is associated with a higher risk for pancreatic cancer and may be associated with a higher risk for ovarian cancer. These increased risks may be due to differences in estrogen levels, insulin signaling, and inflammation in women who are overweight.	[3, 24, 34, 58, 63, 64, 74, 80, 113]
Reproductive history Women who started menstruating at a young age or who experience menopause at an older age have a higher risk of developing breast and ovarian cancer. Conversely, having children and breastfeeding are associated with a lower risk for these cancers. Scientists think that reproductive history affects breast and ovarian cancer risk by altering estrogen levels in the body. Factors that increase the amount of time a woman is exposed to estrogen are often associated with an increased risk for these cancers.	[3, 10, 28, 29, 32, 46, 63, 74, 105, 109]
Alcohol consumption Drinking alcohol increases the chances that a woman will develop breast cancer. The risk increases with greater alcohol consumption and does not seem to vary by type of alcohol consumed. Scientists think this increased risk may be due to changes in hormone levels caused by drinking alcohol. Alcohol consumption has not been associated with an increased risk for ovarian cancer.	[2, 3, 21, 49, 95, 110]
Other genetic variants More than 1,000 variants in the BRCA1 and BRCA2 genes have been linked to hereditary breast and ovarian cancer. Variants in other genes can also increase a woman's risk for breast and ovarian cancer. In some cases, risk is increased to levels similar to the risk conferred by BRCA1 and BRCA2 variants.	[76, 88, 100]
Hormone exposure Exposure to external sources of the hormones estrogen and progesterone affect a woman's chances of developing breast and ovarian cancer. For example, certain hormone replacement therapy after menopause is associated with an increased risk for breast cancer. Current or recent use of hormone replacement therapy has also been associated with an increased risk for ovarian cancer. The use of oral contraceptives is linked to a decreased risk for ovarian cancer.	[3, 22, 25, 27, 63, 72]
Physical activity Women who regularly engage in physical activity have a lower risk of developing breast cancer than women who rarely or never do. In one study, women who walked more than seven hours per week were less likely to develop breast cancer compared to women who walked less than three hours per week. Moderate and vigorous exercise can also decrease breast cancer risk. The links between physical activity and ovarian cancer risk are not yet well understood.	[3, 35, 52, 69, 73, 77, 119]
Smoking Smoking may be associated with an increased risk of developing breast cancer and certain types of ovarian tumors. The strongest effect is observed in women who have smoked heavily for many years. Smoking is also a strong risk factor for pancreatic cancer, accounting for about 25% of all cases.	[3, 26, 45, 47, 68, 110]

Cancer Screening Guidelines

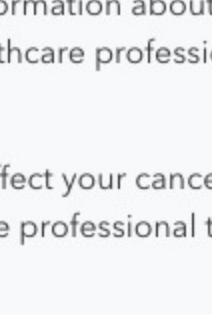
Cancer screening can help detect certain cancers at an earlier stage, when they may be more treatable. The guidelines below apply to people with an average risk of developing cancer. These guidelines may help you and your doctor create a screening plan that's right for you.



Breast cancer
Women should receive regular mammograms depending on their age and other factors. Learn more from the American Cancer Society.



Prostate cancer
Men should talk with their doctor about the benefits and risks of prostate cancer screening. Learn more from the American Cancer Society.



There are currently no specific screening guidelines for male breast cancer, pancreatic cancer, or melanoma. Although there are some conflicting recommendations, guidelines advise against screening for ovarian cancer in the general population. If you have a personal or family history of one of these cancers, please talk with a healthcare professional.

The guidelines above cover the cancers associated with the three variants in this report. You can find information about screening for other cancers from the American Cancer Society. Note that guidelines from different healthcare professional organizations may differ in their recommendations.

Keep in mind that you could still have a BRCA1 or BRCA2 variant not included in this report that could affect your cancer risk. In that case, different screening and prevention actions may be recommended. Consult with a healthcare professional to learn more.

Test Details

Indications for Use

The 23andMe Personal Genome Service (PGS) uses qualitative genotyping to detect select clinically relevant variants in genomic DNA isolated from human saliva collected from individuals ≥18 years with the Oragene Dx model OGD500.001 for the purpose of reporting and interpreting genetic health risks, including the 23andMe PGS Genetic Health Risk Report for BRCA1/BRCA2 (Selected Variants). The 23andMe PGS Genetic Health Risk Report for BRCA1/BRCA2 (Selected Variants) is indicated for reporting of the 185delAG and 5382insC variants in the BRCA1 gene and the 6174delT variant in the BRCA2 gene. The report describes if a woman is at increased risk of developing breast and ovarian cancer, and if a man is at increased risk of developing breast cancer or may be at increased risk of developing prostate cancer. The three variants included in this report are most common in people of Ashkenazi Jewish descent and do not represent the majority of the BRCA1/BRCA2 variants in the general population. The test report does not describe a person's overall risk of developing any type of cancer, and the absence of a variant tested does not rule out the presence of other variants that may be cancer-related. This test is not a substitute for visits to a healthcare provider for recommended screenings or appropriate follow-up and should not be used to determine any treatments.

Special Considerations

- Genetic testing for BRCA1 and BRCA2 variants in the general population is not currently recommended by any healthcare professional organizations.
- Cancer risk associated with a BRCA1 or BRCA2 variant varies from person to person. Exact risk depends on family history and other factors.

Test Performance Summary

Clinical Performance [48, 50, 56, 57, 62, 71, 89, 93, 94, 108, 115, 116]

The variants included in this report represent a very small subset of all those associated with breast, ovarian, and prostate cancer. The three variants tested are associated with an increased risk of developing these cancers. However, some people who have these variants do not develop cancer. In addition, most cases of these cancers are not caused by inherited genetic variants.

- Approximately 5-10% of breast cancer cases, 10-15% of ovarian cancer cases, and 15-20% of male breast cancer cases are known to be caused by inherited variants in the BRCA1 and BRCA2 genes. This number is expected to be higher among individuals of Ashkenazi Jewish descent.
- The three variants in this report account for more than 90% of cancer-related BRCA1 and BRCA2 variants among people of Ashkenazi Jewish descent. These three variants account for a much smaller proportion of cancer-related BRCA1 and BRCA2 variants found in people of other ethnicities.
- About 1 in 40 people of Ashkenazi Jewish descent is expected to have one of the three variants in this report. These three variants are much less common in people of other ethnicities. In 23andMe customers of other ethnicities, between 0% and 0.1% of individuals (up to 1 in 1,000) has one of the three variants in this report.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. The 95% confidence interval was 83.9% to 100%. While unlikely, this test may provide false positive or false negative results. It is possible that the presence of certain mutations in your sample may interfere with the performance of this test. The effects of the interfering mutations on the performance of this test have not been studied. For more details on the analytical performance of this test, refer to the package insert.

Warnings, Precautions, and Limitations

- This test does not diagnose cancer or any other health conditions and cannot determine your overall risk of developing cancer in the future.
- This test should not be used to make medical decisions. Results should be confirmed in a clinical setting before taking any medical action.
- This test does not cover all variants that could increase risk for cancer.* The absence of a variant tested does not rule out the presence of other genetic variants that may impact cancer risk.
- Other factors, such as environmental and lifestyle risk factors, may affect your risk of developing cancer. This test does not account for those factors, and does not test for variants in other genes linked to hereditary cancers.
- Your ethnicity may affect how relevant this test is for you.
- This test is intended to provide you with genetic information to inform conversations with your doctor or other healthcare professional.
- This device is not intended for prenatal testing.
- This test should not be used to assess the presence of genetic variants that may impact response to medications.
- This test is not intended to detect the presence of deterministic variants in autosomal dominant diseases or conditions.
- This test is not a substitute for visits to a healthcare professional for recommended screenings. Consult with a healthcare professional if you have any questions or concerns about your results or your current state of health.
- Some people feel a little anxious after getting genetic health risk results. This is normal. If you feel very anxious, you should speak to your doctor or a genetic counselor.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. Abeliovich D et al. (1997). "The founder mutations 185delAG and 5382insC in BRCA1 and 6174delT in BRCA2 appear in 60% of ovarian cancer and 30% of early-onset breast cancer patients among Ashkenazi women." Am J Hum Genet. 60(3):505-14.
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4. American Cancer Society. "Breast Cancer in Men." Retrieved March 5, 2018, from https://www.cancer.org/cancer/breast-cancer-in-men.html.
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10. Anderson KN et al. (2014). "Reproductive risk factors and breast cancer subtypes: a review of the literature." Breast Cancer Res Treat. 144(1):1-10.

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
April 9, 2018	BRCA1/BRCA2 (Selected Variants) report created.

Print

BRCA1/BRCA2 (Selected Variants)

Frequently Asked Questions

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing certain cancers, including breast cancer (in women and men) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer and certain other cancers. This test includes three genetic variants in the BRCA1 and BRCA2 genes that are most common in people of Ashkenazi Jewish descent.

BRCA1/BRCA2 (Selected Variants)

What does this test do?

What does this test not do?

The report says the variants included in this test are most common in people of Ashkenazi Jewish descent. What if I'm not of Ashkenazi Jewish descent?

Where can I learn more about cancer, support groups, and other resources?

My report says my result could not be determined. What does this mean?

This means we could not tell if you have or do not have the three genetic variants we tested. This can be caused by random test error or other factors that interfere with the test.

It is still possible that you have one of these three variants, or another variant associated with an increased risk of developing breast and ovarian cancer. In the general population, about 1 in 8 women develops breast cancer during her lifetime, and 1 in 80 develops ovarian cancer. Women with a BRCA1 or BRCA2 variant have a greatly increased risk of developing these cancers.

Other factors can also affect your risk of developing breast and ovarian cancer, even if you do not have any genetic variants. Learn more about other factors.

Is this answer helpful? Yes No

What does it mean if the result for a variant could not be determined?

This means we could not tell if you have or do not have the tested genetic variant. This can be caused by random test error or other factors that interfere with the test.

Is this answer helpful? Yes No

My report says my result could not be determined, but I have a personal or family history of breast or ovarian cancer. What does this mean for me?

Women with a family history of breast or ovarian cancer have a higher risk of developing these cancers themselves.

We could not determine whether you have the three tested variants, so we can't tell you whether these variants are contributing to your family history.

There are more than 1,000 variants in the BRCA1 and BRCA2 genes associated with a greatly increased risk for breast and ovarian cancer. Our test only includes three of those variants. Variants in other genes have also been linked to hereditary breast and ovarian cancer, and non-genetic factors also influence a woman's risk of developing these cancers. Learn more about other factors.

It is important to discuss your personal or family history of cancer with a healthcare professional, who can help you determine if additional genetic testing is appropriate. Genetic counseling can also help you understand your results and your options for additional testing. Learn more about genetic counseling.

Is this answer helpful? Yes No

My report says my result could not be determined. What are some things I could do?

Because we could not determine your result, it is still possible to have one of the genetic variants tested or another genetic variant not tested. So your result doesn't give you any new information about your risk for breast and ovarian cancer.

There are many other genetic and non-genetic factors that can affect your risk, which this test does not take into account. Learn more about other factors.

It is important to continue with any cancer screenings your healthcare provider recommends. Learn more about cancer screening.

Talk to a healthcare professional if:

- You have a personal or family history of breast cancer, ovarian cancer, or any other type of cancer.
- You think you might have breast cancer, ovarian cancer, or any other type of cancer.
- You have questions about other risk factors you may have.

Is this answer helpful? Yes No

Have more questions? Check out our Customer Care Help Center.