



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](#)*

Chromosome 17



Gene: BRCA1



You have one of the three genetic variants we tested.

Variants Detected		View All Tested Markers
Marker Tested	Genotype*	Additional Information
6174delT Gene: BRCA2 Marker: rs80359550	(–) Variant copy from one of your parents	T Typical copy from your other parent
		- 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]



Other Factors

Many factors are known to influence the risk of developing breast and ovarian cancer in the general population. In women with your genetic result, the effects of most of these factors on cancer risk are not as well understood.

This is not a complete list of other factors. Except for age and family history, the effects of these factors on cancer risk in women with your genetic result are not as well understood.

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 In general, the chances of developing breast and ovarian cancer increase as a woman gets older. However, women with a BRCA1 or BRCA2 variant have a higher risk of developing early-onset breast cancer (before the age of 45) and multiple breast cancers. Women with a BRCA1 variant may also develop ovarian cancer at an earlier age.	[11, 75, 87, 91, 111]
Family history Women with a BRCA1 or BRCA2 variant whose mothers or sisters have had breast or ovarian cancer are more likely to develop these cancers themselves. The risk is even greater in families with more than one affected family member. This increased risk is likely due to other shared genetic and non-genetic factors. A family history of pancreatic cancer or melanoma is also expected to increase a woman's risk for those cancers.	[11, 78]
Obesity In general, being overweight after menopause 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 In general, 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 In general, drinking alcohol increases the chances of developing 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 is not associated with an increased risk for ovarian cancer.	[2, 3, 21, 49, 95, 110]
Other genetic variants Other genetic variants seem to influence breast and ovarian cancer risk in women with a BRCA1 or BRCA2 variant. In combination, these other variants may increase or decrease a woman's risk for cancer compared to other women with a BRCA1 or BRCA2 variant. Scientists are still working to understand how these other variants modify cancer risk.	[75]
Hormone exposure Exposure to external sources of the hormones estrogen and progesterone affect the risk for breast and ovarian cancer in the general population. For example, certain types of hormone replacement therapy after menopause are 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 In general, 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 had a lower risk for 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 and pancreatic cancer risk are not yet well understood.	[3, 35, 52, 69, 73, 77, 119]
Smoking In the general population, 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 and Prevention 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 a BRCA1 or BRCA2 variant. These guidelines may help you and your doctor decide on the best screening and prevention plan for you.



Women

Women with a BRCA1 or BRCA2 variant should be screened for breast cancer earlier and more often than other women.

However, there are currently no ovarian cancer screening tests that have been proven safe and effective. Women with a BRCA1 or BRCA2 variant may also consider preventive surgery or medication to reduce the risk for breast and ovarian cancer. No specific screening guidelines exist for pancreatic cancer and melanoma, but your doctor may have specific screening recommendations for you. Learn more about breast and ovarian cancer screening and prevention options from [Facing Our Risk of Cancer Empowered](#).



Men

Men with a BRCA1 or BRCA2 variant should be screened for male breast cancer. Screening guidelines for prostate cancer vary. No specific screening guidelines exist for pancreatic cancer and melanoma, but your doctor may have specific screening recommendations for you. Learn more about screening for male breast cancer and prostate cancer from [Facing Our Risk of Cancer Empowered](#).

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 Genetic 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 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

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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.



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Overview

Scientific Details

Frequently Asked Questions



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 one variant called 6174delT was detected in the BRCA2 gene. What does this mean?

This means you have one of the three genetic variants we tested.

Women with a BRCA2 variant have a greatly increased risk of developing breast cancer and ovarian cancer. They also have an increased risk for early-onset breast cancer (before age 45) and multiple breast cancers. In addition, they may have an increased risk for pancreatic cancer and melanoma.

However, this result does not mean you have developed or definitely will develop any of these cancers.

It is important to discuss this result with a healthcare professional. Results should be confirmed in a clinical setting before taking any medical action.

The variant we detected is also associated with a condition called Fanconi anemia group D1. Consider talking with a genetic counselor if you are thinking about having children. [Learn more about genetic counseling.](#)

Is this answer helpful?

Yes

No

What does greatly increased risk mean?

A "greatly increased risk" means that, based on your genetic result for this test, your chances of developing breast and ovarian cancer are much higher than average. Studies have found that 45-85% of women with a BRCA2 variant develop breast cancer, compared to 12% for the general population. Similar studies have found that about 10-27% of women with a BRCA2 variant develop ovarian cancer, compared to 1-2% for the general population. [See Scientific Details for more information.](#)

Your risk for pancreatic cancer and melanoma may also be higher than average. We cannot provide numerical risk estimates because risks for these cancers are not as well understood in people with your genetic result.

It is important to share this result with a healthcare professional.

Is this answer helpful?

Yes

No

My report says that women with a BRCA2 variant have a 45-85% chance of developing breast cancer and a 10-27% chance of developing ovarian cancer. What do those percentages mean? And why is there such a large range?

A 45-85% chance of developing breast cancer means that, out of 100 women with a BRCA2 variant, between 45 and 85 women will develop breast cancer by the age of 70.

A 10-27% chance of developing ovarian cancer means that, out of 100 women with a BRCA2 variant, between 10 and 27 women will develop ovarian cancer by the age of 70.

Many studies have looked at variants in the BRCA1 and BRCA2 genes, and these studies report somewhat different risk estimates. Some of these differences may be due to other factors besides the BRCA1 and BRCA2 variants. For example, women with a BRCA1 or BRCA2 variant who have a family history of breast or ovarian cancer have a higher chance of developing these cancers themselves. Because the group of women included in each study is different, the risk estimates may be different as well.

Your exact risk of developing breast and ovarian cancer depends on many factors, including family history, lifestyle, and genetic factors not included in this test. A healthcare professional can help you get a more precise estimate of your risk.

Is this answer helpful?

Yes

No

What does it mean that the variant detected is associated with a condition called Fanconi anemia group D1?

This result does not mean that you have Fanconi anemia group D1. But your result may be relevant for your family.

Fanconi anemia group D1 is a condition characterized by birth defects, a decreased production of blood cells, and an increased risk for infections and cancer.

People with one copy of the BRCA2 variant we detected are carriers for Fanconi anemia group D1. They do not have the condition themselves, but they can pass a variant on to their children. A child must inherit a BRCA2 variant from each parent in order to have this condition. If your partner has a BRCA2 variant, each of your children may have a 25% chance of having Fanconi anemia group D1. Learn more about Fanconi anemia group D1 at [Facing Our Risk of Cancer Empowered.](#)^{*}

Is this answer helpful?

Yes

No

My report says one variant called 6174delT was detected in the BRCA2 gene. What are some things I could do?

This result is associated with a greatly increased risk of developing breast and ovarian cancer. Women with your result may also have an increased risk for pancreatic cancer and melanoma. It is important to share this result with a healthcare professional, such as a doctor or genetic counselor.

Professional guidelines recommend that women with your genetic result undergo more rigorous cancer screenings and consider certain medications and surgeries that can reduce the risk for cancer. [Learn more about cancer screening and prevention.](#)

For more information about what to think about and possible next steps, see this [help article](#).

It is important to discuss your result with a healthcare professional. **Results should be confirmed in a clinical setting before taking any medical action.**

Is this answer helpful?

Yes

No

How could my result affect my family?

Since you share DNA with your family members, they may also be interested in your result. If you are thinking about talking to family members about your results, [see this article](#) for a discussion of things to consider before having the conversation.

Because you have one variant, it is expected that:

- Each of your children has a 50% chance of inheriting this variant from you.
- One of your parents has this variant.
- Each of your siblings has a 50% chance of having this variant.

Because the variant we detected is associated with an increased cancer risk in both men and women, your adult family members may wish to learn more about their cancer risk. They can talk with a healthcare professional, such as a doctor or genetic counselor, to help them decide if genetic testing is right for them. [Learn more about genetic counseling.](#)

The variant we detected is also associated with a condition called Fanconi anemia group D1. People with one copy of this variant do not have Fanconi anemia group D1, but they could pass the variant on to their children. A child must inherit a BRCA2 variant from each parent in order to have this condition. If your partner also has a BRCA2 variant, each of your children may have a 25% chance of having Fanconi anemia group D1. A genetic counselor can help you determine if additional testing may be appropriate.

Is this answer helpful?

Yes

No

I have questions about my results. Who should I talk to?

It's normal to have questions or concerns after viewing this report. Some people feel anxious, upset, or worried about their risk or risk for their family members. Others simply want to understand their results better or talk to someone about what they can do. Genetic counselors can help. Genetic counselors are healthcare professionals with special training in genetics and genetic testing. [Learn more about genetic counseling.](#)

For more information about what to think about and possible next steps, see this [help article](#).

Since you have a variant detected, it is also important to talk with a healthcare professional about your result and options.

Is this answer helpful?

Yes

No

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