

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.

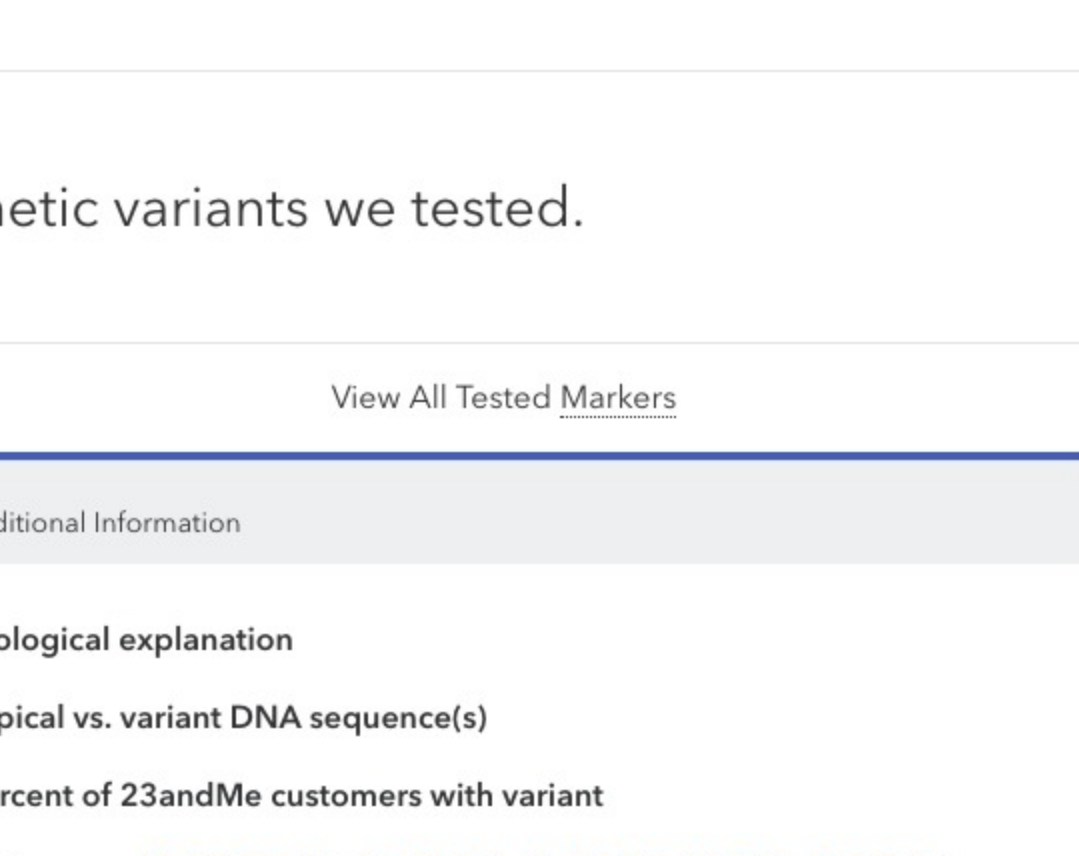
BRCA1

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



You have one of the three genetic variants we tested.

Variants Detected				View All Tested Markers
Marker Tested	Your Genotype*			Additional Information
185delAG Gene: BRCA1 Marker: rs386833395	(-) Variant copy from one of your parents		CT Typical copy from your other parent	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	(-) Typical copy from one of your parents		(-) Typical copy from your other parent	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	T Typical 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]

Lifetime risk			
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 i	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

Many factors are known to influence the risk of developing male breast cancer and prostate cancer in the general population. In men 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 men with your genetic result are not as well understood.

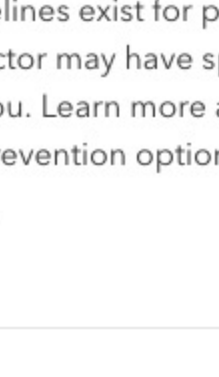
People with multiple risk factors may have a higher risk of developing cancer.

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

Other Factors	References
Age In general, the chances of developing male breast cancer and prostate cancer increase as a man gets older. However, men with a BRCA1 or BRCA2 variant may develop prostate cancer at an earlier age and may develop more aggressive forms of prostate cancer.	[15, 17, 111]
Family history Men with a first-degree relative who has had breast, ovarian, or prostate cancer have a higher risk of developing male breast cancer and prostate cancer. This 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 man's risk for those cancers.	[33, 39, 44, 96]
Obesity In general, being overweight increases a man's chances of developing male breast cancer. The association between obesity and prostate cancer is less clear. However, some studies have found that men who are overweight are more likely to develop aggressive forms of prostate cancer. Obesity is also associated with a higher risk for pancreatic cancer. These increased risks may be due to differences in hormone levels in men who are overweight.	[16, 64, 70, 80, 92, 96, 117]
Ethnicity In general, African-American men have a greater risk of developing prostate cancer than men of other ethnicities. In the U.S., more than 1 in 7 African-American men develops prostate cancer by the age of 80, compared to 1 in 9 men of European descent. African-American men are also more likely to develop prostate cancer at an earlier age. This difference may be due to a combination of genetic and lifestyle factors.	[33, 44, 111]
Other genetic variants It is likely that other genetic variants influence cancer risk in men with a BRCA1 or BRCA2 variant. Scientists are still working to identify these variants.	[75]
Liver disease In the general population, conditions such as liver cirrhosis, which can be caused by drinking too much alcohol, can increase the chances of developing male breast cancer. The increase in cancer risk is thought to be due to a decrease in testosterone and an increase in estrogen in the body. Liver cirrhosis may also increase the risk for pancreatic cancer.	[13, 39, 117]
Smoking In the general population, smoking is associated with an increased risk of dying from prostate cancer. The effect of smoking on the risk of developing prostate cancer is less clear. Smoking is also a major risk factor for pancreatic cancer, accounting for about 25% of all cases. Smoking does not appear to increase the risk for male breast cancer.	[31, 55, 68]

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 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 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

- Abelovich 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.*
- Allen NE et al. (2009). "Moderate alcohol intake and cancer incidence in women." J Natl Cancer Inst. 101(5):296-305.*
- American Cancer Society. "Breast Cancer Facts & Figures 2017-2018." Atlanta: American Cancer Society, Inc. 2017.*
- American Cancer Society. "Breast Cancer in Men." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/breast-cancer-in-men.html>.*
- American Cancer Society. "Breast Cancer." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/breast-cancer.html>.*
- American Cancer Society. "Melanoma Skin Cancer." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/melanoma-skin-cancer.html>.*
- American Cancer Society. "Ovarian Cancer." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/ovarian-cancer.html>.*
- American Cancer Society. "Pancreatic Cancer." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/pancreatic-cancer.html>.*
- American Cancer Society. "Prostate Cancer." Retrieved March 5, 2018, from <https://www.cancer.org/cancer/prostate-cancer.html>.*
- 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.

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 185delAG was detected in the BRCA1 gene. What does this mean?

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

Men with a BRCA1 variant have an increased risk of developing male breast cancer and may also have an increased risk for prostate cancer and pancreatic cancer.

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.

Is this answer helpful?

Yes

No

What does increased risk mean?

An "increased risk" means that, based on your genetic result for this test, your chances of developing male breast cancer are higher than average. Studies have found that 1-2% of men with a BRCA1 variant develop male breast cancer during their lifetime, compared to 0.1% for the general population. [See Scientific Details for more information.](#)

Men with a BRCA1 variant may also have an increased risk for prostate cancer and pancreatic cancer. 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 men with a BRCA1 variant have a 1-2% chance of developing male breast cancer. What does this percentage mean?

A 1-2% chance of developing male breast cancer means that, out of 100 men with a BRCA1 variant, between 1 and 2 men will develop male breast cancer during their lifetime.

We aren't able to give you a numerical estimate of the risk for prostate cancer and pancreatic cancer because risks for these cancers are not as well understood in people with your genetic result.

Your exact risk of developing male breast cancer and other cancers 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

Why doesn't my report include numerical risk estimates for prostate cancer and certain other cancers?

We aren't able to give you a numerical estimate of the risk for prostate cancer and pancreatic cancer because risks for these cancers are not as well understood in people with your genetic result.

A healthcare professional can answer questions you may have about your risk for these cancers.

Is this answer helpful?

Yes

No

My report says one variant called 185delAG was detected in the BRCA1 gene. What are some things I could do?

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

Professional guidelines recommend that men with your genetic result undergo certain cancer screenings. [Learn more about cancer screening.](#)

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.

If your mother or any sisters or daughters have this genetic variant, they have a greatly increased risk of developing breast and ovarian cancer. These and other 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.](#)

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.