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## BRCA1/BRCA2 (Selected Variants)

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing breast cancer (in females and males) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer, pancreatic cancer, and potentially other cancers. This test includes 44 out of more than 4,000 variants in the BRCA1 and BRCA2 genes that are known to increase cancer risk.

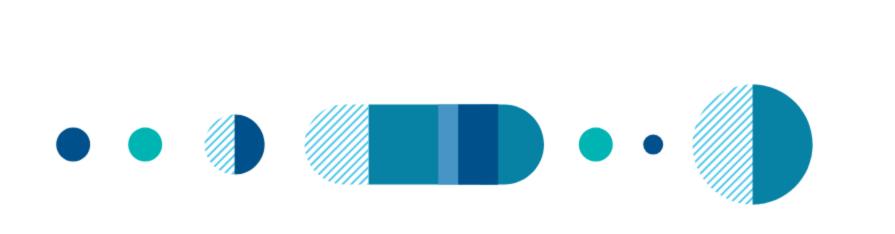
**Overview** 

**Scientific Details** 

**Frequently Asked Questions** 

# Jamie, you have a **greatly increased risk** of developing breast and ovarian cancer.

You have one of the genetic variants we tested. Females with this variant have a much higher than average risk of developing breast and ovarian cancer. Risk for pancreatic cancer may also be increased.



1 variant detected





Please share your result with a healthcare professional. Results should be confirmed by an independent genetic test prescribed by your own healthcare provider before taking any medical action.

#### 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 by an independent genetic test prescribed by your own healthcare provider before taking any medical action.

Please talk to a healthcare professional about additional testing to confirm this result and to better understand your potential cancer risks.

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 44 variants in the BRCA1 and BRCA2 genes. 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, pancreatic cancer, and potentially other cancers.

# Limitations

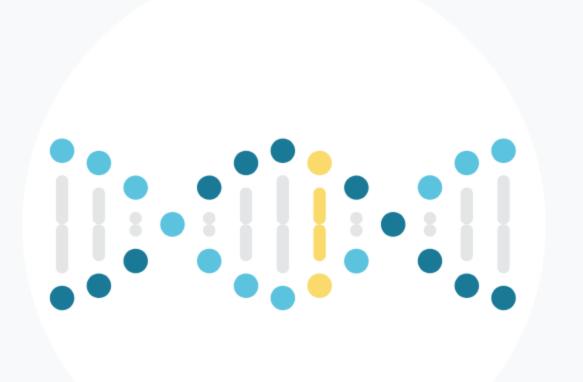
- Does **not** test for all possible variants in the BRCA1 and BRCA2 genes. More than 4,000 variants in these genes are known to increase cancer risk. Only 44 of those variants are included in this test.
- Does **not** test for variants in other genes linked to hereditary cancers.
- Does **not** account for non-genetic factors, like environment and lifestyle, that influence overall cancer risk.
- Does **not** report if someone has two BRCA1 or two BRCA2 variants (due to technical limitations).
- The interpretation of your genetic result depends on the birth sex you reported in your account settings.

# Ethnicity Considerations

- This test does **not** include the majority of BRCA1 and BRCA2 variants found in people of most ethnicities.
- Of the variants included in this test, the majority are most commonly found in people of Ashkenazi Jewish, African American, European, and Hispanic/Latino descent.

# You have a **greatly increased risk** of developing breast and ovarian cancer based on your result.

People with this result may also have an increased risk for pancreatic cancer. It is important to talk with a healthcare professional about options for cancer screening and prevention. It is also important to confirm this result with an independent genetic test prescribed by your own healthcare provider before taking any medical action.

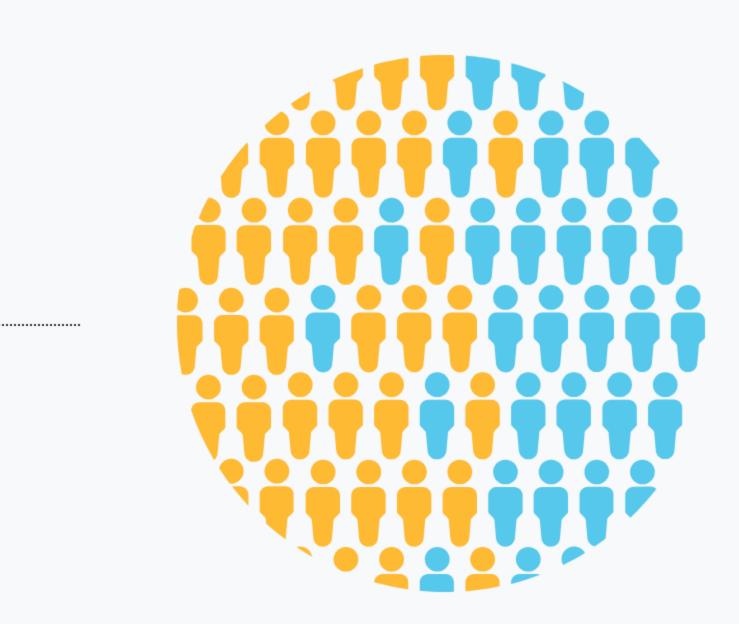


We detected the c.68\_69del variant in the BRCA1 gene.

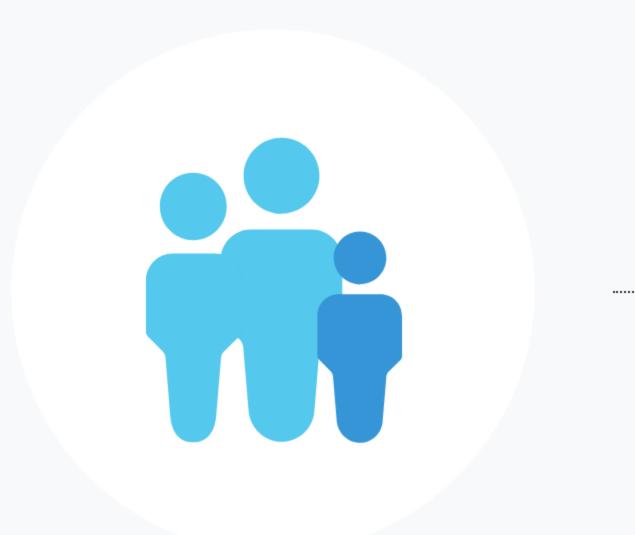
**See Scientific Details** 

Estimates vary, but studies suggest that **45-85%** of females with a BRCA1 variant develop breast cancer by age 70, and **39-46%** develop ovarian cancer.

This is much higher than the risk for the general population. Females with a BRCA1 variant who develop breast cancer also tend to do so at an earlier age, and they may develop ovarian cancer at an earlier age. They may also have an increased risk for pancreatic cancer. Exact risks depend on family history and other factors.



**See Scientific Details** 

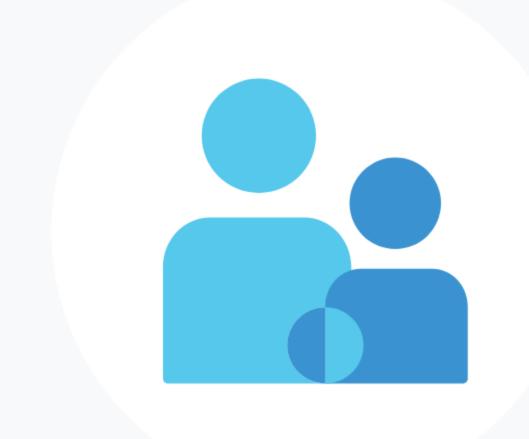


Since you share DNA with your family members, they may also wish to learn more about their cancer risk.

Your parents, siblings, and children each have a 50% chance of having the variant we detected. If you are thinking about sharing your results with family members, **see this article** for a discussion about things to consider before having the conversation. **Genetic counselors** can help your adult family members decide about genetic testing.

# People with one BRCA1 variant are also carriers for a genetic condition called Fanconi anemia group S.

They do not have the condition themselves, but their children may be at risk if both parents are BRCA1 carriers. Consider talking with a **genetic counselor** if you're thinking about having children. You can learn more about Fanconi anemia group S on our **Frequently Asked Questions** page.



# There are things you can do to reduce your risk for breast and ovarian cancer.

Females with a BRCA1 or BRCA2 variant have a greatly increased risk of developing breast and ovarian cancer. They may also have an increased risk for pancreatic cancer. But there are options to consider to help manage your risk for these cancers, so it's important to talk with your doctor about your result. Genetic counseling can also help you understand your results and options. For more information about what to think about and



#### **Know your family history**

Females with your genetic result who have a family history of breast, ovarian, or pancreatic cancer have a higher risk of developing cancer than those without a family history. By knowing your family history, a healthcare professional can better assess your risk.



# Understand your options for screening and prevention

National guidelines recommend earlier and more frequent breast cancer screening for females with your genetic result, to help catch potential cancers at an early stage. However, there are currently no ovarian cancer screening tests that have been proven safe and effective. Your doctor can also explain options to reduce your risk for breast and ovarian cancer, which may include surgery or medication. If you have a family history of pancreatic cancer, your doctor may also recommend pancreatic cancer screening.



#### Maintain a healthy lifestyle

In general, maintaining a healthy weight, engaging in physical activity, limiting alcohol consumption, and avoiding smoking can reduce the risk for breast and ovarian cancer. However, more research is needed to fully understand the impact of these and other lifestyle factors on cancer risk in people with your genetic result.

Results should be confirmed by an independent genetic test prescribed by your own healthcare provider before taking any medical action.

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 females and males) and ovarian cancer. Variants in these genes may also be associated with an increased risk for prostate cancer, pancreatic cancer, and potentially other cancers.



#### ifetime cancer risks

- Females 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.
- Females with a BRCA2 variant have a 45-85% chance of developing breast cancer by 70 and a 10-27% chance of developing ovarian cancer. They also have an increased risk for pancreatic cancer.
- Males with a BRCA1 or BRCA2 variant have an increased risk for male breast cancer and may have an increased risk for prostate cancer and pancreatic cancer.
- 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, females with a BRCA1 or BRCA2 variant have an increased risk for early-onset breast cancer (before age 45) and multiple breast cancers. Females with a BRCA1 variant may also develop ovarian cancer at an earlier age. Males with a BRCA1 or BRCA2 variant may develop earlier and more aggressive prostate cancer.



#### How common are BRCA1 and BRCA2 variants?

About 1 in 200 people in the general population has a BRCA1 or BRCA2 variant linked to hereditary breast, ovarian, and pancreatic cancer, although most of those variants are not included in this report. BRCA1 and BRCA2 variants are more common in people of certain ethnicities. For example, among people of Ashkenazi Jewish descent, about 1 in 40 has a variant (usually one of three specific variants in this report: BRCA1 c.68\_69del, BRCA1 c.5266dup, or BRCA2 c.5946del).



#### Screening and prevention

- Guidelines recommend that females 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 females 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.
- Males with a BRCA1 or BRCA2 variant should be screened for male breast cancer. They should discuss their result with a doctor to determine whether prostate cancer screening is appropriate.
- People with a BRCA1 or BRCA2 variant and a family history of pancreatic cancer may also be offered pancreatic cancer screening.

Read more at: National Cancer Institute GeneReviews

## It is important to discuss this result with a healthcare professional.



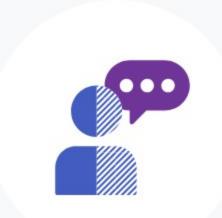
It's important to consult with a healthcare professional to confirm your result and discuss options for cancer screening and prevention.

**Print report** 



If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help.

Find a genetic counselor



People with one BRCA1 variant are also carriers for a condition called Fanconi anemia group S. If you're considering having children, a genetic counselor can help you understand if additional testing may be appropriate.

Find a genetic counselor



See our Frequently Asked Questions for more information.

**FAQs** 



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## BRCA1/BRCA2 (Selected Variants)

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing breast cancer (in females and males) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer, pancreatic cancer, and potentially other cancers. This test includes 44 out of more than 4,000 variants in the BRCA1 and BRCA2 genes that are known to increase cancer risk.

Overview

**Scientific Details** 

**Frequently Asked Questions** 

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

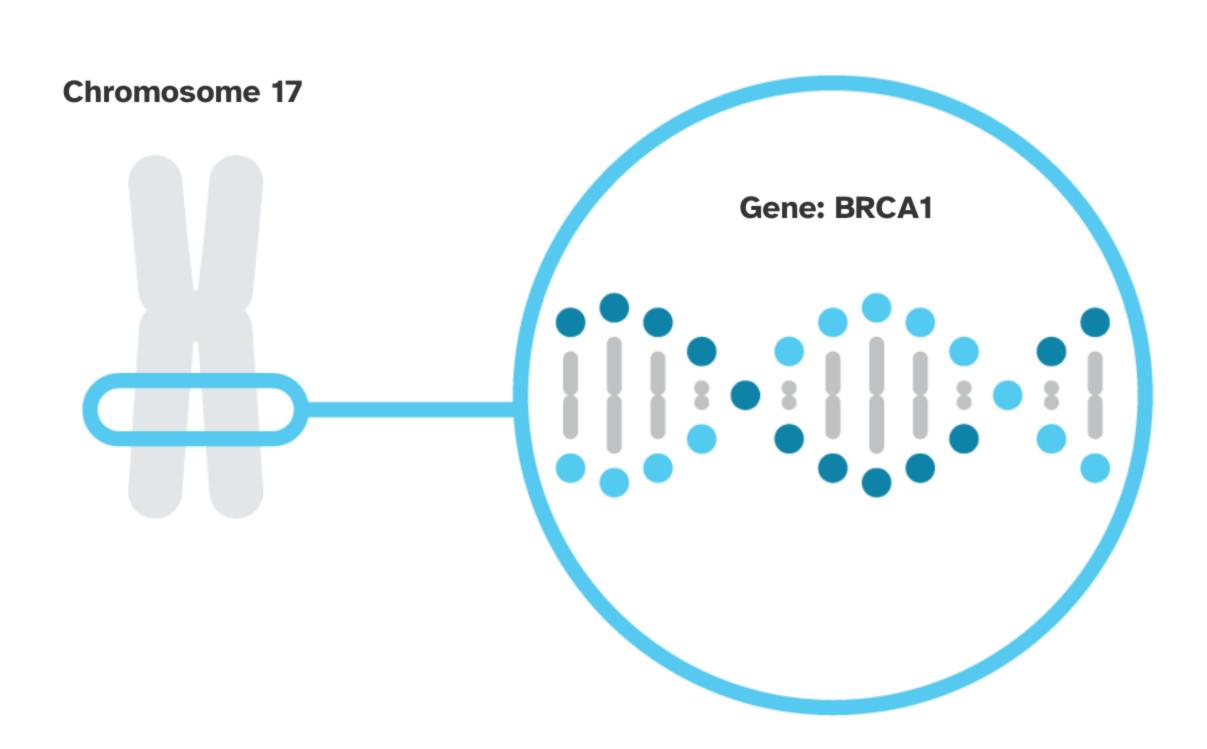
This report includes 24 variants in the BRCA1 gene and 20 variants in the BRCA2 gene. These variants do not account for the majority of the BRCA1 and BRCA2 variants in people of most ethnicities. More than 4,000 variants in these genes are known to increase cancer risk.

BRCA1

BRCA2

The BRCA1 gene contains instructions for making a protein that helps repair damaged <u>DNA</u>. The BRCA1 protein also helps control the process of <u>cell</u> division. Through both of these functions, the BRCA1 protein acts as a <u>tumor suppressor</u>, preventing cells from growing and dividing too rapidly. Certain <u>variants</u> 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 MedlinePlus



#### You have one of the genetic variants we tested.

<u>Variants</u> Detected				View All Tested Markers
Marker Tested	Genotype*			Additional Information
c.68_69del Gene: BRCA1 Marker: rs80357914	(—)  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 [ 9, 34, 39, 43, 77, 102 ]   ClinVar

<sup>\*</sup> 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. 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 [ 19, 27, 46, 92, 93 ]

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 people 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 females) or during their lifetime (for males).

Cancer type	General population	BRCA1 variant	BRCA2 variant
Breast (female)	12.9%	45-85%	45-85%
Ovarian	1.1%	39-46%	10-27%
Breast (male)	0.1%	1-2%	7-8%
Prostate	12.6%	May have an increased risk (1)	Increased risk
Pancreatic	1.7%	May have an increased risk	Increased risk
Melanoma	2.1%	Research ongoing	Research ongoing

See risk estimates by race and ethnicity for the general population

#### Other Factors

Many factors are known to influence the risk of developing breast, ovarian, and pancreatic cancer in the general population. In people 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 people with your genetic result are not 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	[ 71, 88, 92 ]
In general, the chances of developing breast, ovarian, and pancreatic cancer increase with age. However, females with a BRCA1 or BRCA2 <u>variant</u> have a higher risk of developing early-onset breast cancer (before the age of 45) and multiple breast cancers. Females with a BRCA1 variant may also develop ovarian cancer at an earlier age.	
Family history	[ <b>46</b> , <b>56</b> ]
People 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 nongenetic factors. A family history of pancreatic cancer is also expected to increase a person's risk for that cancer.	
Weight	[ 3, 4, 5, 49 ]
In the general population, being overweight after menopause increases the 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	

[ **3**, **4** ]

levels, insulin signaling, and inflammation in females who are overweight.

**Reproductive history** 

In general, females 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 person is exposed to estrogen are often associated with an increased risk for these cancers.

#### Alcohol consumption

[ **1**, **3**, **5** ]

In general, drinking alcohol increases the chances of developing breast cancer. The risk increases with greater alcohol consumption, but even one drink per day may increase risk. Risk 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 may also increase risk for pancreatic cancer. It has not been associated with an increased risk for ovarian cancer.

The American Cancer Society states that it's best not to drink alcohol at all. For those who choose to drink, experts recommend limiting alcohol consumption to  $\leq 1$  drink per day for females and  $\leq 2$  drinks per day for males.

#### Other genetic variants

[ 45, 53 ]

Other genetic variants seem to influence breast and ovarian cancer risk in people with a BRCA1 or BRCA2 variant. In combination, these other variants may increase or decrease the risk for cancer compared to other people with a BRCA1 or BRCA2 variant. Scientists are still working to understand how these other variants modify cancer risk.

#### Hormone exposure

[ **3**, **4** ]

Exposure to external sources of the hormones estrogen and progesterone affect the chances of developing breast and ovarian cancer in the general population. For example, certain types of menopausal hormone therapy are associated with an increased risk for breast cancer. Current or recent use of menopausal hormone 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 and may be associated with a slightly increased risk for breast cancer in people who are currently taking them.

#### **Breast density**

[ **3** ]

In the general population, having dense breast tissue increases the risk of developing breast cancer. Dense breast tissue also makes cancers harder to detect with a mammogram.

#### **Physical activity**

[**3**]

In the general population, engaging in regular physical activity is associated with a lower risk of developing breast cancer, and higher levels of activity are associated with bigger reductions in risk. The links between physical activity and ovarian cancer risk are not yet well understood.

#### Smoking

[ 3, 4, 5 ]

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 people who have smoked heavily for many years. Smoking is also a strong risk factor for pancreatic cancer, accounting for about 25% of all cases.

### 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.





#### **Females**

Females with a BRCA1 or BRCA2 variant should be screened for breast cancer earlier and more often than other females. However, there are currently no ovarian cancer screening tests that have been proven safe and effective. Females with a BRCA1 or BRCA2 variant may also consider preventive surgery or medication to reduce the risk for breast and ovarian cancer. If they have a family history of pancreatic cancer, these individuals may also be offered pancreatic cancer screening. Learn more about breast, ovarian, and pancreatic cancer screening and/or prevention options from Facing Our Risk of Cancer Empowered.

#### Males

Males with a BRCA1 or BRCA2 variant should be screened for male breast cancer. They should discuss their result with a doctor to determine whether prostate cancer screening is appropriate. People with a BRCA1 or BRCA2 variant and a family history of pancreatic cancer may also be offered pancreatic cancer screening. Learn more about screening for male breast cancer, prostate cancer, and pancreatic cancer from <a href="#Facing Our Risk of Cancer Empowered">Facing Our Risk of Cancer Empowered</a>.

#### 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 44 variants in the BRCA1 and BRCA2 genes. The report describes if a person's genetic result is associated with an increased risk of developing breast cancer and ovarian cancer and may be associated with an increased risk for prostate cancer, pancreatic cancer, and potentially other cancers. The variants included in this report do not represent the majority of the BRCA1/BRCA2 variants in people of most ethnicities. 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 cancerrelated. This report is for over-the-counter use by adults over the age of 18, and provides genetic information to inform discussions with a healthcare professional. 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. The full list of variants included in this report is: BRCA1: c.68\_69del, c.213-11T>G, c.427G>T, c.815\_824dup, c.1556del, c.1687C>T, c.1960A>T, c.1961del, c.2681\_2682del, c.2864C>A, c.3481\_3491del, c.3598C>T, c.3627dup, c.3756\_3759del, c.3770\_3771del, c.4035del, c.4065\_4068del, c.4327C>T, c.4357+1G>A, c.4964\_4982del, c.4986+6T>G, c.5123C>A, c.5177\_5180del, c.5266dup. BRCA2: c.658\_659del, c.771\_775del, c.1929del, c.2808\_2811del, c.2957\_2958insG, c.3170\_3174del, c.3264dup, c.3545\_3546del, c.3847\_3848del, c.4471\_4474del, c.5542del, c.5576\_5579del, c.5682C>G, c.5946del, c.6037A>T, c.6275\_6276del, c.7024C>T, c.7480C>T, c.7934del, c.8904del.

#### **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. In addition, new research in the future may determine that other
  cancers besides those mentioned in this report may be associated with BRCA1 and BRCA2 variants.
  Please talk to a healthcare professional if you have questions about new research related to BRCAassociated cancers.

#### **Test Performance Summary**

Clinical Performance [ 12, 15, 34, 35, 37, 39, 40, 47, 51, 52, 62, 72, 74, 75, 77, 79, 80, 96, 101, 102 ]

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

- Inherited variants in the BRCA1 and BRCA2 genes account for approximately 5-10% of breast cancers, 10-15% of ovarian cancers, 15-20% of male breast cancers, 1-6% of prostate cancers, and 1-10% of pancreatic cancers. Among people with a family history, these percentages are expected to be higher.
- The variants in this report account for more than 90% of cancer-related BRCA1 and BRCA2 variants among people of Ashkenazi Jewish descent. These variants account for a much smaller proportion of

# 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 by an independent genetic test prescribed by your own healthcare provider 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

cancer-related BRCA1 and BRCA2 variants found in people of other ethnicities, including about 30-40% among African Americans, people of European descent, and people of Hispanic or Latino descent; about 5-25% among people of East Asian descent; and up to 35% among people of South Asian descent.

 About 1 in 40 people of Ashkenazi Jewish descent is expected to have one of three specific variants in this report (BRCA1 c.68\_69del, BRCA1 c.5266dup, or BRCA2 c.5946del). Among people of other ethnicities, about 1 in 200 has a BRCA1 or BRCA2 variant, but most of those variants are not included 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 widest 95% confidence interval was 47.3% to 100% for the heterozygous BRCA1 c.2864C>A genotype. 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.

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 <u>Package Insert</u> 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. American Cancer Society. "Alcohol Use and Cancer." Retrieved Dec 6, 2022, from https://www.cancer.org/cancer/cancer-causes/diet-physical-activity/alcohol-use-and-cancer.html \(^{\bar{1}}\)
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- 3. American Cancer Society. "Breast Cancer Facts & Figures." Retrieved Dec 6, 2022, from https://www.cancer.org/research/cancer-facts-statistics/breast-cancer-facts-figures.html \rightarrow
- 4. American Cancer Society. "Ovarian Cancer Risk Factors." Retrieved Jun 9, 2023, from https://www.cancer.org/cancer/types/ovarian-cancer/causes-risks-prevention/risk-factors.html \rightarrow
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- 6. American Cancer Society. "Prostate Cancer Risk Factors." Retrieved Jun 9, 2023, from https://www.cancer.org/cancer/types/prostate-cancer/causes-risks-prevention/risk-factors.html \( \)
- 7. American Cancer Society. "Risk Factors for Breast Cancer in Men." Retrieved Jun 9, 2023, from https://www.cancer.org/cancer/types/breast-cancer-in-men/causes-risks-prevention/risk-factors.html \(^3\)
- 8. American Cancer Society. "Risk Factors for Melanoma Skin Cancer." Retrieved Jun 9, 2023, from https://www.cancer.org/cancer/types/melanoma-skin-cancer/causes-risks-prevention/risk-factors.html \rightarrow

  1. \*\*Concer\*\* | Cancer\*\* | Cancer
- 9. Antoniou AC et al. (2005). "Breast and ovarian cancer risks to carriers of the BRCA1 5382insC and 185delAG and BRCA2 6174delT mutations: a combined analysis of 22 population based studies." J Med Genet. 42(7):602-3.
- 10. Baudi F et al. (2001). "Evidence of a founder mutation of BRCA1 in a highly homogeneous population from southern Italy with breast/ovarian cancer." Hum Mutat. 18(2):163-4. \[ \]

See all references  $\vee$ 

#### Change Log

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

Date	Change
Oct. 16, 2023	41 genetic variants were added to the report for customers on the most recent version of the 23andMe genotyping platform (V5). If any of these variants were detected, customers will see this reflected in their result.
	For customers with a BRCA2 variant detected, the interpretation of the genetic result was changed from "may have an increased risk for pancreatic cancer" to "increased risk for pancreatic cancer" based on updated clinical

guidelines.

For customers with a BRCA1 variant detected, information about Fanconi anemia group S was added.

Variant names were updated to align with the naming conventions used in the scientific literature. The names of the three Ashkenazi Jewish founder variants have been updated as follows: 185delAG is now c.68\_69del, 5382insC is now c.5266dup, and 6174delT is now c.5946del.

Nov. 5, 2020

Information about pancreatic cancer screening was added for people with a BRCA1 or BRCA2 variant and a family history of pancreatic cancer.

April 9, 2018

BRCA1/BRCA2 (Selected Variants) report created.



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Health > Health Predisposition



# BRCA1/BRCA2 (Selected Variants)

Specific genetic variants in the BRCA1 and BRCA2 genes are associated with an increased risk of developing breast cancer (in females and males) and ovarian cancer. These variants may also be associated with an increased risk for prostate cancer, pancreatic cancer, and potentially other cancers. This test includes 44 out of more than 4,000 variants in the BRCA1 and BRCA2 genes that are known to increase cancer risk.

Overview

**Scientific Details** 

**Frequently Asked Questions** 

### BRCA1/BRCA2 (Selected Variants)

What does this test do?	~
What does this test <b>not</b> do?	~
Who is at risk for breast, ovarian, prostate, and other cancers?	~
The report says this report does <b>not</b> include the majority of BRCA1 and BRCA2 variants found in people of most ethnicities. What does this mean?	~
Where can I learn more about cancer, support groups, and other resources?	~
My report says <b>one variant</b> was detected in the BRCA1 gene. What does this mean?	~
What does <b>greatly increased risk</b> mean?	~
My report says that females with a BRCA1 variant have a <b>45-85% chance</b> of developing breast cancer and a <b>39-46% chance</b> of developing ovarian cancer. What do those percentages mean? And why is there such a large range?	~
My report says that BRCA1 and BRCA2 variants may potentially be associated with an increased risk for other cancers. What does this mean?	~
What does it mean that people with one BRCA1 variant are carriers for a condition called Fanconi anemia group S?	~
My report says <b>one variant</b> was detected in the BRCA1 gene. What are some things I could do?	~
How could my result affect my family?	~
I have questions about my results. Who should I talk to?	~

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



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Family Tree

**Your Connections** 









**Traits** 











