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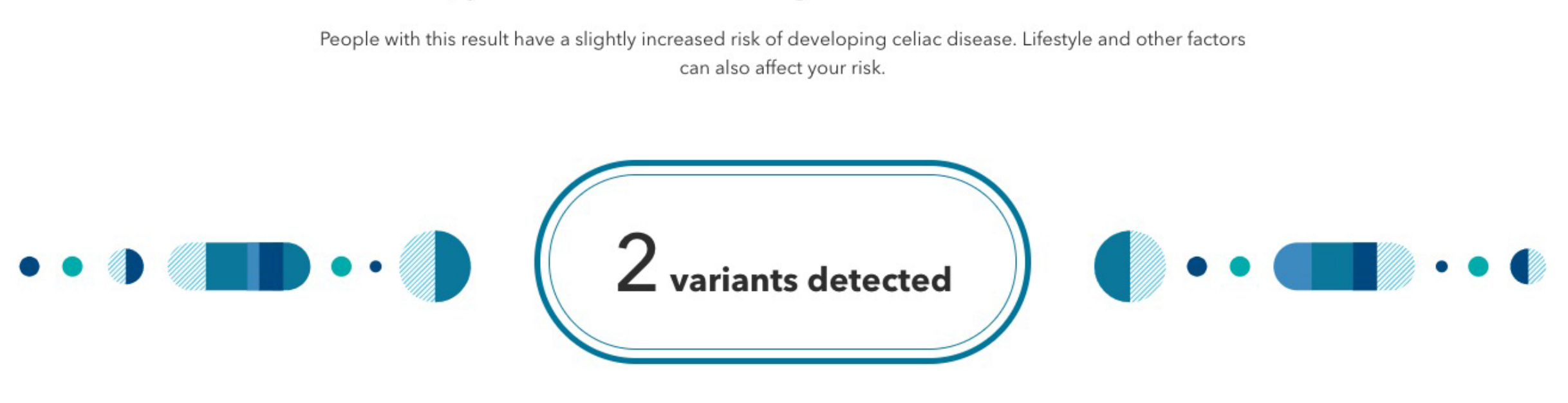
Celiac Disease

Celiac disease is an autoimmune condition in which the consumption of gluten (found in wheat, barley, and rye) can result in damage to the small intestine. Celiac disease can lead to both digestive and non-digestive problems. This test includes two common variants associated with an increased risk of developing this condition.

Basic, you have **both** of the genetic variants we tested.

People with this result have a slightly increased risk of developing celiac disease. Lifestyle and other factors can also affect your risk.



One variant detected in the HLA-DQA1 gene and one variant detected in the HLA-DQB1 gene.

How To Use This Test

This test does not diagnose celiac disease or any other health conditions.

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

Review the Genetic Health Risk tutorial

See Scientific Details

See Frequently Asked Questions

Intended Uses

- Tests for variants near the HLA-DQA1 and HLA-DQB1 genes linked to the **HLA-DQ2.5** and **HLA-DQ8** haplotypes. These haplotypes are associated with celiac disease.

Limitations

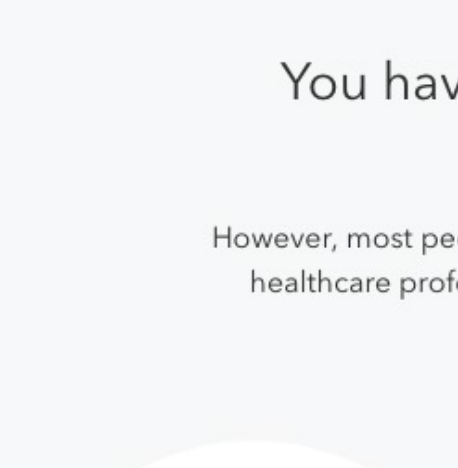
- Does not test for all possible variants, genes, or haplotypes associated with celiac disease.
- Does not cover other potential gluten- or wheat-related conditions.

Important Ethnicities

- The variants included in this test are common in many ethnicities, but are best studied in people of **European** descent.

You have a **slightly increased risk** of developing celiac disease based on your genetic result.

However, most people with this result do not develop celiac disease. Consider discussing your risk with a healthcare professional, especially if you have a family history or other risk factors for this condition.



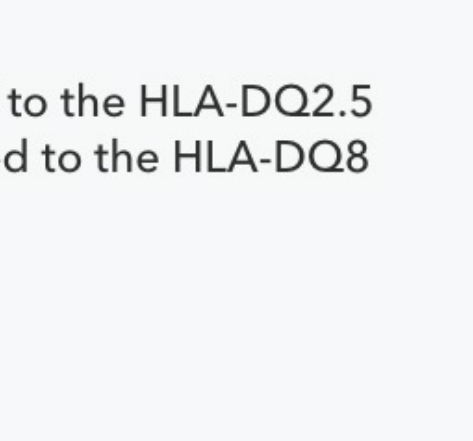
We detected a variant linked to the HLA-DQ2.5 haplotype and a variant linked to the HLA-DQ8 haplotype.

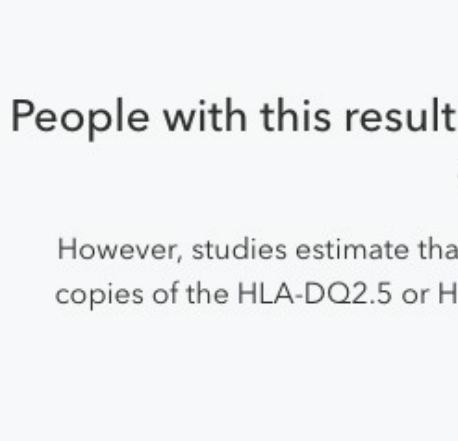
See Scientific Details

People with this result have a slightly increased risk of developing celiac disease.

However, studies estimate that only about 3% of people with one or more copies of the HLA-DQ2.5 or HLA-DQ8 haplotypes develop celiac disease.

See Scientific Details





Since you share DNA with your family members, they may also be interested in this result.

One or both of your parents is expected to have at least one of these variants. In addition, each of your children likely has a 75% chance of having one or both variants. Your siblings may also have one or both variants.

Lifestyle and other factors can also influence the chances of developing celiac disease.

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

Gluten

Gluten (found in wheat, barley, and rye) is the main non-genetic factor that triggers the development of celiac disease in people with increased genetic risk.

See Scientific Details for more information

Family history

Other conditions

About Celiac Disease

Also known as: Coeliac disease, celiac sprue, CD, gluten-sensitive enteropathy, nontropical sprue

When it develops

Celiac disease can develop anytime from infancy to adulthood, most commonly between the ages of 10 and 40. In people with celiac disease, symptoms occur after consuming gluten.

How common is the condition?

Celiac disease affects people of all ethnicities. About 1 in 100 people worldwide has celiac disease.

Typical signs and symptoms

- Diarrhea, gas, and bloating
- Poor appetite
- Skin rashes
- Fatigue
- Anemia
- Headache

How it's treated

Celiac disease can be effectively treated by removing all sources of gluten from the diet. This includes foods and drinks made with wheat, barley, and rye.

Read more at: Mayo Clinic National Institute of Diabetes and Digestive and Kidney Diseases GeneReviews Genetics Home Reference

Consider sharing this result with a healthcare professional, especially if you have other risk factors.

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

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.

Learn more

See our Frequently Asked Questions for more information.

FAQs

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Celiac Disease

Scientific Details

Celiac disease is an autoimmune condition in which the consumption of gluten (found in wheat, barley, and rye) can result in damage to the small intestine. Celiac disease can lead to both digestive and non-digestive problems. This test includes two common variants associated with an increased risk of developing this condition.

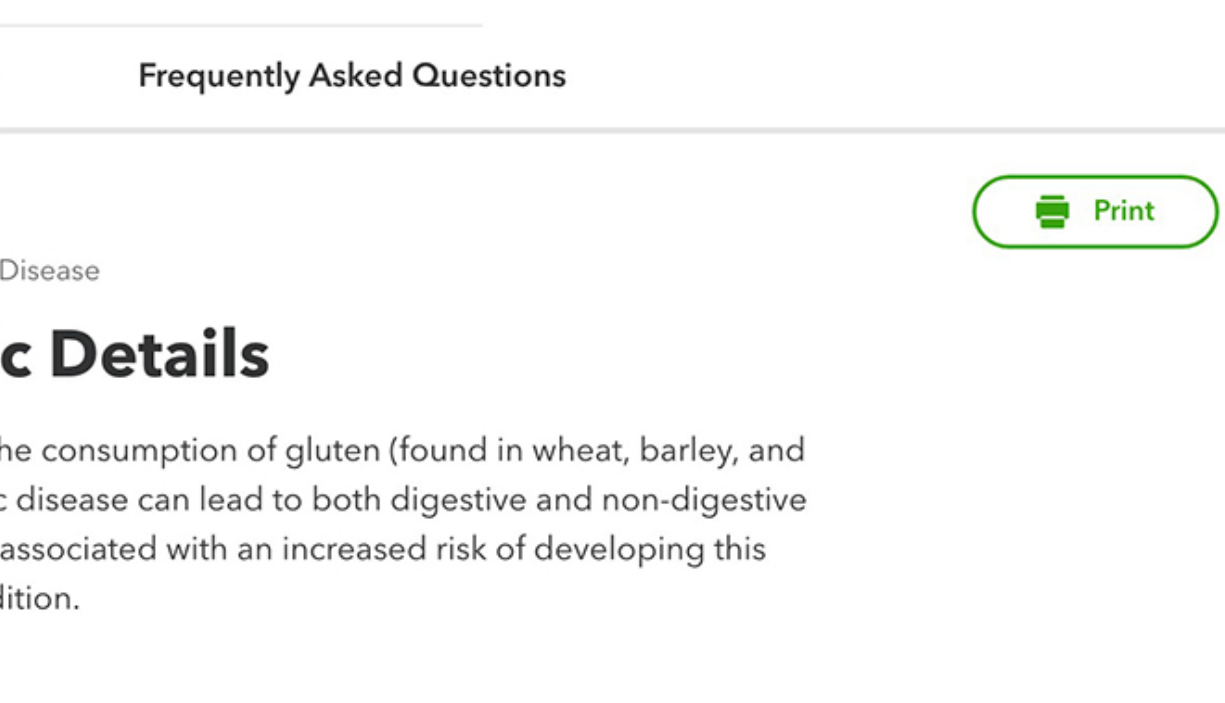
Celiac disease is associated with the HLA-DQ2.5 and HLA-DQ8 haplotypes.

These haplotypes are associated with variants in the HLA-DQA1 and HLA-DQB1 genes.

HLA-DQA1 HLA-DQB1

The HLA gene family contains instructions for making a group of proteins called the human leukocyte antigen (HLA) complex. The HLA-DQA1 gene provides instructions for making one half of a protein complex that is present on the surface of certain immune cells. The other half of the protein complex is made by the HLA-DQB1 gene. This protein complex presents foreign peptides to the immune system to trigger the body's immune response. Certain variants in the HLA-DQA1 gene can trigger an inappropriate immune response to gluten.

Read more at Genetics Home Reference



You have both of the genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*		Additional Information
HLA-DQ2.5 **Gene:** HLA-DQA1 **Marker:** rs2187668	C Typical copy from one of your parents	T Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 9, 11, 16, 21, 22, 23]
HLA-DQ8 **Gene:** HLA-DQB1 **Marker:** rs7454108	C 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 [5, 6, 10, 11, 12, 13, 16, 18]

*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

Health Risk Estimates

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

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

References [7, 20]

Risk estimates

- The HLA-DQ2.5 and HLA-DQ8 haplotypes included in this test are common in many ethnicities. About 15% to 30% of the general population have either or both of these haplotypes.
- Most people with the haplotypes detected by this test do not develop celiac disease. Studies estimate that only about 3% of people with one or more copies of the HLA-DQ2.5 or HLA-DQ8 haplotypes develop celiac disease.
- Among people who develop celiac disease, about 90% have one or more copies of the HLA-DQ2.5 haplotype and about 5% have one or more copies of the HLA-DQ8 haplotype. The remainder may have other haplotypes not covered by this test.
- People without either of the two tested variants are not likely at risk of developing celiac disease.

Other Factors

Other factors besides the variants included in this test can influence your chances of developing celiac disease.

This is not a complete list of other factors.

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

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

Other Factors	References
Gluten In people with a genetic predisposition for celiac disease, consuming gluten (found in wheat, barley, and rye) can trigger an immune response that attacks the lining of the small intestine. This damages the surface of the small intestine, such that the body cannot properly absorb nutrients. As a result, people with celiac disease can experience both digestive and non-digestive symptoms. However, most people with a genetic predisposition never experience any problems from eating gluten.	[20]
Family history Parents, siblings, and children of a person diagnosed with celiac disease have a higher chance of developing the condition themselves. This may primarily be explained by genetic factors, but could also be related to family members sharing a similar lifestyle. For a person with unknown genetic risk and an affected first-degree relative, the chance of developing celiac disease is about 5%-20%, compared to 1% for the general population.	[3, 10, 19]
Other conditions People with certain health conditions are more likely to develop celiac disease. The prevalence of celiac disease increases to 3-12% among people with Down syndrome, and to approximately 5% for people with other autoimmune conditions like type 1 diabetes, arthritis, or Sjögren's syndrome. This is compared to the 1% prevalence of celiac disease in the general population.	[3, 20]
Sex Some studies have shown that women are more likely to be diagnosed with celiac disease than men, but other studies did not find a difference between men and women. Scientists are still trying to figure out if there truly is a difference in risk between the two sexes.	[1, 8, 10, 24]
Other genes There are other genes and variants that have been associated with celiac disease. However, many of these variants may have only a small effect on risk.	[14, 15]

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for Celiac Disease is indicated for reporting of one variant associated with the HLA-DQ2.5 haplotype and one variant associated with the HLA-DQ8 haplotype. The report describes if a person has a variant linked to a haplotype that is associated with a higher risk of developing celiac disease, but it does not describe a person's overall risk of developing celiac disease. This report is most relevant for people of European descent.

Special Considerations

- Genetic testing for celiac disease is recommended under certain circumstances by several health professional organizations, including the American College of Gastroenterology.

[4, 10, 17, 20]

Test Performance Summary

Clinical Performance

About 95% of all cases of celiac disease are associated with the HLA-DQ2.5 and HLA-DQ8 haplotypes.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. All test results were at least 99% correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

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

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

References

1. Ciacci C et al. (1995). "Gender and clinical presentation in adult celiac disease." Scand J Gastroenterol. 30(11):1077-81.
2. Dubois PC et al. (2010). "Multiple common variants for celiac disease influencing immune gene expression." Nat Genet. 42(4):295-302.
3. Fasano A et al. (2003). "Prevalence of celiac disease in at-risk and not-at-risk groups in the United States: a large multicenter study." Arch Intern Med. 163(3):286-92.
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5. Khosravi A et al. (2016). "The likelihood ratio and frequency of DQ2/DQ8 haplotypes in Iranian patients with celiac disease." Gastroenterol Hepatol Bed Bench. 9(1):18-24.
6. Koskinen L et al. (2009). "Cost-effective HLA typing with tagging SNPs predicts celiac disease risk haplotypes in the Finnish, Hungarian, and Italian populations." Immunogenetics. 61(4):247-56.
7. Leonard MM et al. (2017). "Celiac Disease and Nonceliac Gluten Sensitivity: A Review." JAMA. 318(7):647-656.
8. Liu E et al. (2014). "Risk of pediatric celiac disease according to HLA haplotype and country." N Engl J Med. 371(1):42-9.
9. Lundin KE et al. (1993). "Gliadin-specific, HLA-DQ(alpha 1*0501,beta 1*0201) restricted T cells isolated from the small intestinal mucosa of celiac disease patients." J Exp Med. 178(1):187-96.
10. Megiorni F et al. (2009). "HLA-DQ and risk gradient for celiac disease." Hum Immunol. 70(1):55-9.

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
Oct. 19, 2017	Celiac Disease report created.

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Celiac Disease

Frequently Asked Questions

Celiac disease is an autoimmune condition in which the consumption of gluten (found in wheat, barley, and rye) can result in damage to the small intestine. Celiac disease can lead to both digestive and non-digestive problems. This test includes two common variants associated with an increased risk of developing this condition.

Celiac Disease

What is celiac disease and how is it different from non-celiac disease gluten sensitivity?

Celiac disease is an autoimmune condition in which the consumption of gluten (found in wheat, barley, and rye) can result in damage to the small intestine. This damage occurs because the body has an inappropriate immune reaction to gluten and attacks its own intestinal cells. Celiac disease can lead to both digestive and non-digestive symptoms.

Other gluten- or wheat-related conditions, such as non-celiac gluten sensitivity, have also been described. Although people with these conditions have reported symptoms similar to those with celiac disease, the underlying cause is thought to be different. If you think you have digestive symptoms, or other symptoms that could be the result of a gluten- or wheat-related condition, talking to a healthcare provider may help determine if more testing is appropriate.

You can learn more about celiac disease from the following resources:

- Celiac Disease Foundation
- National Celiac Association
- Beyond Celiac
- Association of European Coeliac Societies

Is this answer helpful? Yes No

What does this test do?

This test looks for two specific genetic variants, one in the HLA-DQA1 gene and one near the HLA-DQB1 gene, that are linked to the HLA-DQ2.5 and HLA-DQ8 haplotypes. These two haplotypes are associated with celiac disease.

People with the HLA-DQ2.5 and HLA-DQ8 haplotypes are more likely to develop celiac disease. However, most people with these haplotypes will not develop the condition.

This test does not include all possible genetic variants, genes, or haplotypes that may increase your risk of developing celiac disease.

Is this answer helpful? Yes No

What does this test not do?

This test does not diagnose celiac disease. Only a healthcare professional can do that.

This test does not tell you if you have celiac disease or if you will definitely develop the condition in the future.

This test does not take into account other risk factors for celiac disease, such as personal and family health history. Thus, this test cannot provide a complete assessment of your risk of developing the condition.

This test does not include all possible genetic variants, genes, or haplotypes that may increase your risk of developing celiac disease.

This test does not cover other potential gluten- or wheat-related conditions.

Is this answer helpful? Yes No

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

You can learn more about celiac disease from the following resources:

- Celiac Disease Foundation
- National Celiac Association
- Beyond Celiac
- Association of European Coeliac Societies

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

Is this answer helpful? Yes No

My report says **two variants** were detected. What does this mean?

This means you have one copy of each of the two genetic variants we tested.

People with this result have a slightly increased risk of developing celiac disease compared to the general population. However, most people with this result do not develop celiac disease. Studies estimate that only about 3% of people with one or more copies of the HLA-DQ2.5 or HLA-DQ8 haplotypes develop celiac disease, compared to approximately 1% in the general population.

Your risk of developing celiac disease is influenced by other factors, such as family history, lifestyle, and other genetic variants not covered by this test. Learn more about other factors.

Is this answer helpful? Yes No

My report says **two variants** were detected. What are some things I could do?

People with this result have a slightly increased risk of developing celiac disease.

Lifestyle and other factors may also influence your risk. Learn more about other factors.

Consider talking to a healthcare professional if:

- Celiac disease runs in your family.
- You think you might have celiac disease.
- You have questions about other risk factors you may have.
- You have concerns about your results.

Is this answer helpful? Yes No

What does **slightly increased risk** mean?

A "slightly increased risk" means that, based on your genetic result for the test, your chances of developing celiac disease are slightly higher than average. See Scientific Details for more information.

Consider sharing this result with a healthcare professional, especially if you have other risk factors, such as a family history of celiac disease.

Genetic variants not covered by this test and non-genetic factors may also influence your risk of developing celiac disease. Learn more about other factors.

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 two genetic variants, it is expected that:

- Each of your children has a 75% chance of inheriting one or both variants from you.
- One or both of your parents has at least one of these variants.
- Each of your siblings may have one or both of these variants.

Is this answer helpful? Yes No

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

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