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.

Overview Scientific Details Frequently Asked Questions

Jamie, you have **one** of the two 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.

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.

Ethnicity Considerations

- 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.

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.

At least one of your parents is also expected to have this variant. In addition, each of your siblings has at least a 50% chance of having this variant, and each of your children has a 50% chance of inheriting this variant from you.

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

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 18 and 46. In people with celiac disease, symptoms occur after consuming gluten.

Typical signs and symptoms

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

How common is the condition?

Celiac disease affects people of all ethnicities. About 1 in 198 people worldwide has celiac disease.

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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Overview | Scientific Details | Frequently Asked Questions

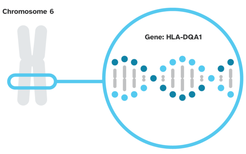
Celiac disease is associated with the HLA-DQ2.5 and HLA-DQ8 haplotypes.

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 one of the two 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]

*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 39% 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 96% 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%-26%, 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 or Williams 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.

Test Performance Summary

Clinical Performance [4, 10, 17, 20]

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. Greater than 99% of test results were 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. *
4. Gujral N et al. (2012). "Celiac disease: prevalence, diagnosis, pathogenesis and treatment." World J Gastroenterol. 18(42):6036-59. *
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 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?
- What does this test do?
- What does this test **not** do?
- Where can I learn more about celiac disease, support groups, and other resources?
- My report says **one variant** was detected. What does this mean?
- My report says **one variant** was detected. What are some things I could do?
- What does **slightly increased risk** mean?
- How could my result affect my family?

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