

# GlutenID®

## Celiac Genetic Health Risk Report

First Name/Last Name/DOB/GID-Number

### Purpose Of The Test

The GlutenID test determines **your risk for celiac disease** based on DNA testing.

Celiac disease is an autoimmune condition. It can develop when a person **with a genetic risk for celiac** eats gluten in their food. It causes digestive and non-digestive symptoms. The treatment is a gluten-free diet.

This test cannot tell you if you have celiac disease. It cannot tell you if you will develop it. It **can** tell you if you have a genetic risk for celiac disease or if you don't have a genetic risk. **Celiac disease rarely develops in people without a genetic risk. (6)**

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#### Your Result in More Detail

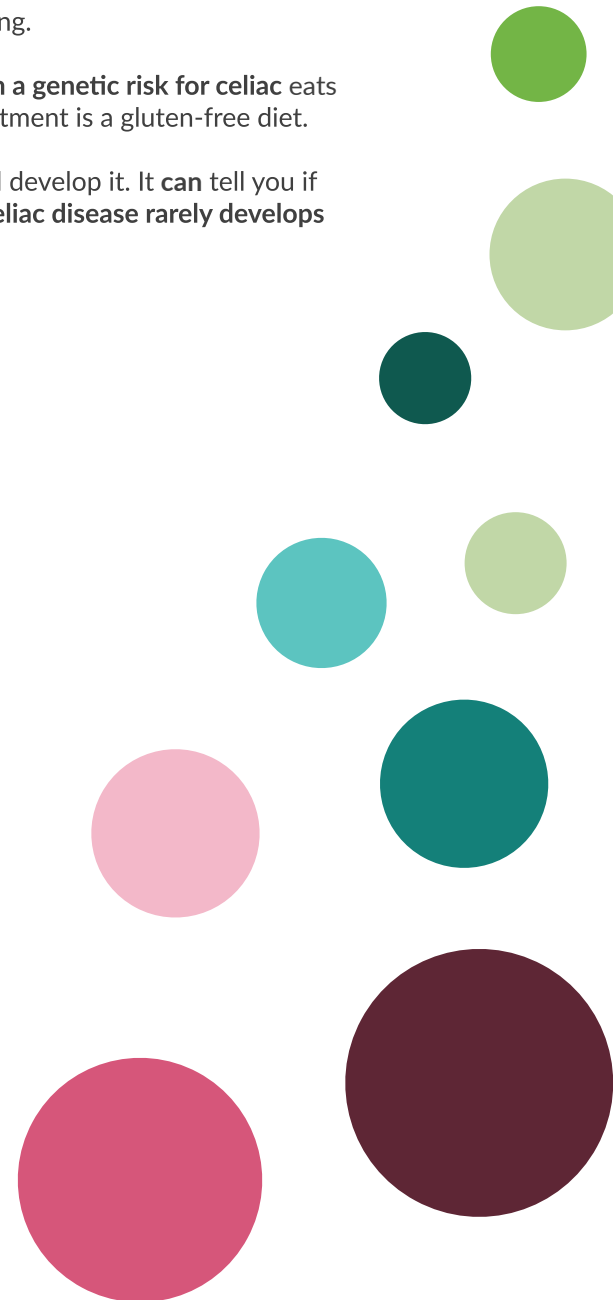
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# Your Report At-A-Glance

## Your Result

**2 Celiac Risk Variants  
Were Detected**

You have **TWO** genetic risk variants for celiac disease. Your variants are called DQ2(cis) and half-DQ2 (DQ2.2.)

### What does this mean?

You have increased genetic risk for celiac disease. Symptoms of celiac disease can be triggered when a person with a genetic risk eats gluten in their diet.

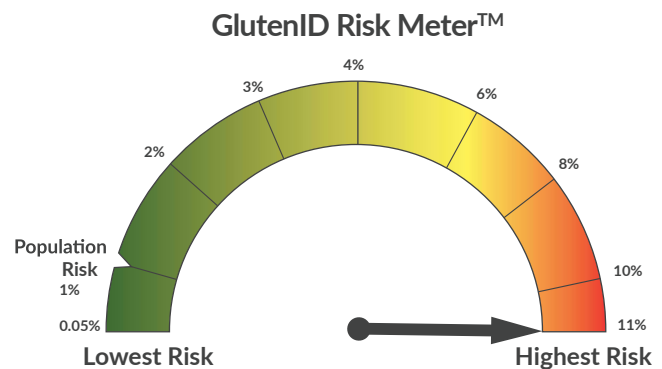
This result does not mean you have or will develop celiac disease.

## Your Risk

### Increased Risk

Based on DNA testing, you have an increased risk for celiac disease.

If you have symptoms of celiac disease or a family history, your actual risk may be higher.



## Important Next Steps

### Talk to a genetic counselor or your healthcare provider

Share your result with them. Some people may feel anxious about getting genetic results. A genetic counselor or healthcare provider can help. They can discuss next steps for you based on your health and family history. They may recommend other tests for celiac disease. For people who have celiac disease, removing gluten from their diet is usually an effective treatment. Talk to a genetic counselor or healthcare provider before making any changes to your diet or lifestyle.

### Talk to your family

**Celiac disease runs in families.** There is a chance your parents, siblings, and children also have the same variant. Your result might be important for their health too.

## Limitations of the Test

### Important things to know

It **does not diagnose** celiac disease, gluten or wheat-related sensitivities, or any other condition.

It **cannot predict** if someone will develop celiac disease in the future.

It **does not replace** a visit with a genetic counselor or healthcare provider.

It **does not detect** all possible genetic risk factors for celiac disease.

It is based on **current scientific knowledge**. This will continue to improve over time.

It does not rule out the presence of other genetic variants that may be related to celiac disease.

Other companies offering celiac genetic risk testing may detect different genetic variants.

# Your Result In More Detail

## About the Test

The GlutenID test looks at **two genes**. They are called **HLA-DQA1** and **HLA-DQB1**.

These genes sit beside each other. They are inherited together, as a set, one copy from each parent. They come in different versions (or variants) called haplotypes.

Four specific haplotypes are associated with a risk for celiac disease. These are the celiac risk variants. These variants can affect how your immune system responds to gluten. For some, eating gluten will trigger an abnormal immune response which damages the small intestine. This causes the symptoms of celiac disease.

**The GlutenID test looks for the presence or absence of the four celiac risk variants.**

When no celiac risk variants are found, the result will say “non-celiac genetics” (NCG). NCG results are not associated with a risk for celiac disease.

## About Your Risk

### Your GlutenID

The GlutenID test estimates your genetic risk for celiac disease based on your DNA results. Your genetic risk depends on the pair of variants you inherited from your parents. We call this your GlutenID. There are 15 possible combinations. Each is associated with a different risk for celiac disease.

Variant 1:  
Celiac risk  
variants found  
**DQ2(cis)**

Variant 2: Celiac  
risk variants found  
**DQ2.2 (half-DQ2)**

- You have 2 celiac risk variants. Your variants are called DQ2 and half-DQ2.
- This means you have a genetic risk for celiac disease.
- Your chance to develop celiac disease is increased when compared to people with no celiac risk variants.

## For Genetic Counselors or Healthcare Providers

- This test is not meant to diagnose celiac disease.
- The test provides users with their genetic risk for celiac disease.
- Any diagnosis or treatment should be based on the patient's clinical and laboratory findings.
- For people who have celiac disease, removing gluten from their diet is usually an effective treatment.

**!** Important: if you have symptoms of a health condition please speak with your doctor.

### Race/Ethnicity

**The GlutenID test is relevant for people with all different backgrounds.**

Celiac disease affects people all over the world. The celiac risk variants have been studied the most in people with European ancestry but are found in people of all races/ethnicities. This means if you have Hispanic, African, Asian or other non-European ancestry, this genetic test result still applies to you.



## Other Risk Factors

Celiac disease is caused by a combination of factors. Genetics is just one of them. Some other factors which can influence the risk for celiac disease include:

### Gluten

In people with a genetic risk, eating gluten is the main factor which triggers celiac disease.

### Family History

Having close relatives with celiac disease can significantly increase your risk.

### Other Conditions

The risk for celiac is higher for people with Down syndrome, Williams syndrome, and autoimmune conditions, like type 1 diabetes. (3)

### Biological Sex

While celiac disease can affect anyone, more biological females are diagnosed with the condition. The reason is not well understood. (3)

The more risk factors you have, the higher your overall chance for celiac disease.

This test cannot predict your overall risk for celiac disease. A genetic counselor or your healthcare provider can help you understand your personal risk for celiac disease.

There are other risk factors (both genetic and non-genetic) that are not yet well understood. Research is on-going and knowledge will continue to improve over time.

## Risk to Family

You share your genes with your biological family members. Some of your relatives will have the same celiac risk variant. **This result might be important for their health.**

### Recommended Next Steps.

**Share this report with your close relatives if they are interested.** It can be medically important to learn if a person has a genetic risk for celiac disease or not. It can help a doctor determine next steps for them based on their health.

The only way for a person to know if they have a genetic risk for celiac disease is to have DNA testing. Relatives (including children) who want to clarify their own genetic risk can do GlutenID testing.

To learn more about what these results mean for you and your family, speak with a genetic counselor.

## More About Celiac

Celiac disease is an autoimmune condition. It can be triggered when genetically at-risk people consume dietary gluten. In response to gluten, the person's immune system attacks and damages the lining of the small intestine.

### What are the symptoms?

Celiac disease causes both digestive (e.g., diarrhea, weight loss, abdominal pain) and non-digestive (e.g., anemia, chronic fatigue, vitamin deficiencies) symptoms. The complete list of potential symptoms is very long and variable. For this reason, **celiac disease can be difficult to diagnose just based on symptoms.**

### What is the treatment?

The treatment is a gluten free diet. Removing gluten from the diet repairs the intestinal damage caused by gluten ingestion.

### What is the cause?

The cause of celiac is complex. It involves both genetic and non-genetic factors. The genetic risk alone does not cause celiac disease. The genetic risk for celiac disease is necessary but not enough to cause the condition.

If you are concerned about potential symptoms of celiac disease, share this report with a genetic counselor or healthcare provider. They can determine if you need further medical tests or if you should consider a gluten free diet.

## Additional Information

### ***National Celiac Association***

<https://nationalceliac.org>

### ***Beyond Celiac***

<https://beyondceliac.org>

### ***Celiac Foundation***

<https://celiac.org>

### ***American Academy of Allergy, Asthma & Immunology***

<https://aaaai.org/Tools-for-the-Public/Conditions-Library/Allergies/celiac-disease>

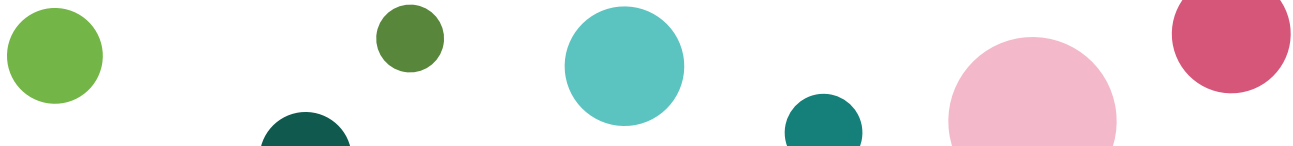
### ***University of Chicago Medicine***

<https://cureceliacdisease.org>

### ***National Society of Genetic Counselors***

<https://findageneticcounselor.nsgc.org/?reload=timezone>

### ***Link to Scientific Terms***



# Recap of Important Information

## Test Limitations

- The GlutenID test **does NOT diagnose celiac disease**, gluten (or wheat) sensitivity, or any health condition. Only a healthcare professional can diagnose conditions.
- The GlutenID test is **NOT a substitute for a medical appointment**. Any diagnostic or treatment decisions should be based on discussions with a healthcare provider.
- Many people with a celiac risk variant will never develop celiac disease. This test cannot predict who will or will not develop the condition.
- This test **does not describe a person's overall risk to develop celiac disease**. It is a multifactorial condition, and many other factors play a role.
- The GlutenID test only detects four HLA haplotypes associated with celiac disease risk. Having no copies of those haplotypes is a non-celiac genetics (NCG) result. The variant details for NCG results are not reported. There are many other HLA haplotypes that may not be relevant to celiac disease risk.
- The GlutenID test **does not detect all possible genetic variants associated with celiac disease**. It does not test for emerging celiac-associated genetic markers with insufficient level of evidence (LOE) to be classified as clinically relevant.
- While unlikely, this test may provide false positive or false negative results.
- Genetic tests for celiac disease offered by other companies may be looking at different genetic variants. It is possible to receive different celiac genetic risk results from a different company.
- The GlutenID test is **based on current knowledge**. Scientific understanding will continue to improve over time. Additional relevant variants may be established in the future that are not covered by this test.

## Important Next Steps

1. To learn more about what these results mean for you and your family, **speak with a genetic counselor**.
2. If you are having signs and symptoms of celiac disease, it is important to share your GlutenID results with your healthcare provider. Your healthcare provider may wish to use this result to make decisions about your care.
3. Click here to **review our Frequently Asked Questions (FAQs)** and learn more about the GlutenID test.
4. Click here for glossary of genetic terms.

# Scientific Details

## Your DNA Results

Haplotype	Other Names for Haplotype	Tag SNP	Variant Genotype	Your Genotype	Number of Copies
HLA-DQ2	DQ2(cis) DQ2.5	rs2187668	T	C ✖ T	1
HLA-DQ8	DQ8	rs7454108	C	T ✖ T	0
HLA-DQ2.2	half-DQ2	rs7775228	C	T ✖ C	1
HLA-DQ7	DQ7	rs4639334	A	G ✖ G	0

Two additional SNPs rs2395182 and rs4713586 are included in DQ2.2 analysis.

### What is the GlutenID Test Looking For?

The GlutenID test detects the presence or absence of celiac risk haplotype variants. It looks for markers (or tag SNPs) associated with celiac risk haplotypes located close to the HLA-DQA1 and HLA-DQB1 genes.

Four specific haplotypes are associated with a risk for celiac disease, commonly called DQ2, half-DQ2, DQ7, and DQ8

### About these variants:

- 95% of people with celiac disease have at least one copy of DQ2 or DQ8
- 30% of people in the general population have at least one copy of DQ2 or DQ8.
- However, only 3% of people with at least one copy of DQ2 or DQ8 will develop celiac disease-most people with DQ2 or DQ8 will not develop celiac disease.
- The celiac risk variants have been studied most in people with European ancestry but have been found in people of all races/ethnicities.

## Your Risk Estimate

Risk estimates are based on clinical studies. This table summarizes the estimated genetic risk and risk category associated with each GlutenID result.

**You have increased risk to develop celiac disease compared to people with no genetic risk.**

### GlutenID Risk Gradient™

GlutenID	Estimated Risk	Risk Category
DQ2+DQ2	1 in 9 (11%)	Increased Risk
DQ2+half-DQ2	1 in 9 (11%)	Increased Risk
DQ2+DQ8	1 in 12 (8%)	Increased Risk
DQ8+half-DQ2	1 in 25 (4%)	Increased Risk
DQ2+DQ7	1 in 35 (3%)	Increased Risk
DQ2.2+DQ7 (DQ2 trans)	1 in 35 (3%)	Increased Risk
DQ2(cis)	1 in 35 (3%)	Increased Risk
DQ8+DQ8	1 in 100 (1%)	Low Risk
DQ8	1 in 100 (1%)	Low Risk
DQ8+DQ7	1 in 100 (1%)	Low Risk
half-DQ2+half-DQ2	1 in 210 (0.5%)	Not Likely At Risk
half-DQ2 (DQ2.2)	1 in 210 (0.5%)	Not Likely At Risk
DQ7+DQ7	1 in 1842 (0.05%)	Not Likely At Risk
DQ7	1 in 1842 (0.05%)	Not Likely At Risk
Non-Celiac Genetics	1 in 2518 (0.05%)	Not Likely At Risk

Calculated risk is based on likelihood ratio data including both adults and children from Megiorni et al. 2009, Almeida et al. 2016, and Choung et al. 2020.

## Peer-Reviewed Journal Articles

1. Megiorni et al., (2009). "HLA-DQ and risk gradient for celiac disease." *Hum Immunol* 70:55-59.
2. Monsuur et al., (2008). "Effective detection of human leukocyte antigen risk alleles in celiac disease using tag single nucleotide polymorphisms." *PLoS ONE* 3(5): e2270. doi:10.1371/journal.pone.0002270  
by using HLA alleles." *Clin Gastroenterol Hepatol* 7:966-971.
3. Taylor et al., (2015). "Celiac disease." <https://www.ncbi.nlm.nih.gov/books/>
4. Pisapia et al., (2020). "Differential expression of pre-disposing HLA-DQ2.5 alleles in DR5/DR7 celiac disease patients affects the pathological immune response to gluten. *Nature Sci Rep* 10,17227 <https://doi.org/10.1038/s41598-020-73907-2>
5. Bajor et al., (2019). "Classical celiac disease is more frequent with a double dose HLA-DQB1\*02: A systematic review with meta-analysis." *PLoS ONE* 14(2): e0212329.
6. Pallav et al., (2014). "Clinical utility of celiac disease associated HLA testing." *Dig Dis Sci* 59:2199-2206.
7. Al-Toma et al., (2006). "Human leukocyte antigen-DQ2 homozygosity and the development of refractory celiac disease and enteropathy-associated T-cell lymphoma." *Clin Gastroenterol Hepatol* 4:351-319.
8. Vader et al., (2010). "The HLA-DQ2 gene dose effect in celiac disease is directly related to the magnitude and breadth of gluten-specific T cell responses." *Proc Natl Acad Sci USA* 100:12390-12395.
9. Singh et al., (2018). "Global prevalence of celiac disease: systematic review and meta-analysis." *Clin Gastroenterol and Hepatol* 16:823-83.
10. Liu et al., (2017). High incidence of celiac disease in a long-term study of adolescents with susceptibility genotypes. *Gastroenterology* 152:1329-1336.
11. Pietzak et al., (2009). "Stratifying risk for celiac disease in a large at-risk United States population. *Clinical Gastroenterol and Hepatol* 7:966-971.
12. Kavamme et al., (2020). Population-based screening for celiac disease reveals the majority of patients are undiagnosed and improve on a gluten free diet. *Scientific Reports* 12:12647.
13. Choung et al., (2020). Celiac disease risk stratification based on HLA-DQ heterodimer (HLA-DQA1~DQB1) typing in a large cohort of adults with suspected celiac disease. *Human Immunology* 81:59-64.
14. Almeida et al., (2016). Presence of DQ2.2 associated with DQ2.5 increases the risk for celiac disease. *Autoimmune Diseases*. Volume 2016, Article ID5409653; <http://dx.doi.org/10.1155/2016/5409653>.

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