

## Frequently Asked Questions

### How long does testing take?

The turnaround time for testing is 7 to 14 days.

### Is there an age limit?

No, the DNA sample can be obtained from individuals at any age, even newborns.

### How is the DNA sample collected?

The DNA sample is collected using cheek swabs provided in the test kit. The swabs are rubbed gently inside the amount against the cheek to collect buccal cells. Buccal cells contain an abundance of DNA which is used for the genetic test. The collection is fast, simple and painless. Ingestion of gluten is not required.

### How will I receive the results?

Once the test is complete, the final test report will be sent to you by mail, email or both.

### Which loci are detected in this test?

This test will detect the gene variants HLA-DQA1\*05, HLA-DQB1\*02, and HLA-DQB1\*0302 which are associated with celiac disease.

### What will the results tell me?

Detection of celiac disease associated alleles in a symptomatic individual is supportive of a diagnosis of celiac disease. Identification of celiac disease-associated alleles implies an increased risk for celiac disease but is not diagnostic of celiac disease as only a subset of individuals with these genes will develop celiac disease. However, individuals who test negative for HLA-DQA1\*05, HLA-DQB1\*02, and HLA-DQB1\*0302, can exclude a diagnosis of celiac disease and have almost no lifetime risk of developing celiac disease (well less than 0.01% chance), regardless of whether or not they ingest gluten.

Visit [www.celiacdiseasedna.com](http://www.celiacdiseasedna.com) for more

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### How do I take the test?

The DNA test for celiac disease involves a few simple steps:

- 1 Order the DNA test kit.** The kit can be ordered online at [www.celiacdiseasedna.com](http://www.celiacdiseasedna.com) or by phone. Once you place the order, the testing kit will be shipped directly to you.
- 2 Collect a DNA sample using the swabs included in the kit.** The kit contains swabs called "buccal swabs". DNA is collected quickly and easily by rubbing the swab inside your mouth against the cheek for 15 seconds. Once the DNA is collected, the swabs are placed into the specimen container provided in the kit and returned to the laboratory for testing using the return package included in the testing kit.
- 3 Receive Results Report.** Once your samples arrive at the laboratory, testing begins immediately and results are available in 7 to 14 days.

For more information, visit

[www.celiacdiseasedna.com](http://www.celiacdiseasedna.com)

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Medical DNA Testing Services • Legal Biomedical Services

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## Celiac Disease DNA Test

Knowledge is power

**1 in 100** have it.  
**Are you at risk?**



**1 in 100 people are affected with Celiac Disease, but only 3% to 5% are diagnosed**

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## A dangerous reaction to Gluten

### What is Celiac Disease?

Celiac disease is an autoimmune disorder which causes inflammation of the small bowel and villous damage when a genetically susceptible individual ingests gluten containing grains (wheat, rye, or barley)



### Genetics of Celiac Disease

Almost all individuals with celiac disease have detectable genetic variants in their HLA-DQ genes.

The HLA-DQ protein consists of two subunits, an alpha chain encoded by the HLA-DQA1 gene and a beta chain encoded by the HLA-DQB1 gene. HLA-DQ has an important function in the immune system.

Celiac disease is strongly associated with two inherited variants of the HLA-DQ protein: DQ2 and DQ8, which are thought to increase the risk of celiac disease by causing receptors in the immune system to preferentially bind to gliadin (a component of gluten) and initiating an autoimmune response.

#### HLA-DQ2

- Found in more than 90% of individuals with celiac disease and in 20% to 30% of the general population.

- Confirmed by positive genetic test results for the following gene variants: HLA-DQA1\*05 and HLA-DQB1\*02

#### HLA-DQ8

- Found in 5% to 10% of individuals with celiac disease and 10% of the general population.

- Confirmed by positive genetic test results for the following gene variant: HLA-DQB1\*0302 (HLA-DQB1\*0302 is always inherited together with HLA-DQA1\*03)

### What are the Symptoms?

Clinical diagnosis of celiac disease is extremely difficult because it can be associated with a wide range of both gastrointestinal and non-gastrointestinal symptoms, or may even have no clinical symptoms at all.

#### Gastrointestinal Symptoms

- Pale, loose and malodorous diarrhea
- Weight loss
- Abdominal pain, cramping, bloating
- Anorexia
- Lactose intolerance

#### Non-Gastrointestinal Symptoms

- Chronic fatigue
- Anemia
- Joint pain/inflammation
- Migraines
- Depression
- Attention-deficit disorder
- Osteoporosis/osteopenia
- Vitamin and mineral deficiency
- Infertility and recurrent miscarriage
- Epilepsy
- Neuropsychiatric conditions
- Failure to thrive
- Dental enamel defects
- Dermatitis herpetiformis (skin blistering)
- Increased risk of infections
- Autoimmune disorders

### Celiac Disease is Under-diagnosed

Due to the difficulty in diagnosing celiac disease symptomatically and a lack of clinician familiarity with the disease, only 5% to 10% of individuals with celiac disease are diagnosed.

Untreated celiac disease leads to an increased risk of adenocarcinoma, lymphoma, esophageal and oropharyngeal squamous carcinoma

A diagnosis of celiac disease can be excluded if you do not have any of the celiac disease associated HLA alleles.

### DNA Testing for Celiac Disease

Detection of celiac disease associated alleles in a symptomatic individual is supportive of a diagnosis of celiac disease. Since 30% of the population have one of the celiac disease-associated HLA alleles, but only 3% of these individuals will develop celiac disease in their lifetime, identification of celiac disease-associated alleles implies an increased risk for celiac disease but is not diagnostic of celiac disease as only a subset of individuals with these genes will develop celiac disease.

However, individuals who test negative for HLA-DQA1\*05, HLA-DQB1\*02, and HLA-DQB1\*0302, can exclude a diagnosis of celiac disease and have almost no lifetime risk of developing celiac disease (well less than 0.04% chance), regardless of whether or not they ingest gluten.

### How does the Test Work?

The celiac disease DNA test determines the presence or absence of HLA gene variants which are known to be associated with celiac disease susceptibility, including the DQ2 alleles (HLA-DQA1\*05, HLA-DQB1\*02) and the DQ8 allele (HLA-DQB1\*0302).

### Treatment of Celiac Disease

The only known effective treatment for celiac disease is a life-long adherence to a strict gluten free diet. Even ingesting foods which may be tainted with small amounts of gluten during processing must be avoided, as even small amounts of gluten can be dangerous for individuals with celiac disease. Additional treatment may be required to manage vitamin deficiencies and osteoporosis associated with celiac disease.

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