

Gene test information

CELIAC DISEASE
(HLA-DQA1 and HLA DQB1 GENE TESTS)

- **Background**

Celiac disease is a common enteropathy with a strong genetic risk. It is characterized by a permanent intolerance for gluten proteins present in dietary wheat, rye, and barley. It affects approximately 1:100–300 individuals, although only 1 person in 8 is aware of being affected because the symptoms may be mild or nonspecific.

Environmental, genetic, and immunologic factors are important in the pathogenesis of celiac disease. Celiac disease is strongly associated with specific HLA-DQ2 and HLA-DQ8 molecules, encoded by HLA-DQA1 and HLA-DQB1 genes.

A negative gene test for HLA-DQA1 and HLA-DQB1 variants associated with celiac disease effectively rules out a diagnosis of celiac disease. Nevertheless, the gene test on its own cannot diagnose celiac disease, only a small fraction of subjects with a positive gene test will develop celiac disease.

- **Possible test results**

Test result	Commentary
Negative	No HLA-DQA1 and HLA-DQB1 variants associated with celiac disease were detected. A diagnosis of celiac disease can be ruled out with very high probability (>99%).
Positive	Detection of HLA-DQA1 and HLA-DQB1 variants associated with celiac disease. A diagnosis of celiac disease can not be ruled out.

References:

Wolters VM. Genetic Background of Celiac Disease and Its Clinical Implications. Am J Gastroenterol 2008;103:190-5.