

# CELIAC DISEASE



## HLA-DQA/DQB genotyping to aid in excluding the diagnosis of **celiac disease** and for risk assessment

### Background

- Celiac disease is estimated to affect 1 out of 100 people, but fewer than 5% of patients are diagnosed as having the disease.<sup>1,3</sup>
- Early diagnosis and lifelong treatment with a gluten-free diet is critical to relieve symptoms and reduce the risk of complications such as secondary autoimmune disorders.<sup>1,3</sup>
- Diagnosis is challenging due to variable, nonspecific symptoms and varying age of onset.<sup>3,4</sup>
- Tissue transglutaminase (tTG) and endomysial (EMA) IgA antibody testing are the most commonly used screening tests.<sup>1,3</sup>

### Clinical Utility

- The HLA-DQ2 allele has been found in 90% to 95% of celiac cases, and HLA-DQ8 has been identified in most remaining cases.<sup>1</sup>
- A negative HLA-DQA/DQB genotyping result essentially excludes celiac disease as the diagnosis.<sup>1,3,5</sup>
- A positive result is not diagnostic but indicates a higher probability for celiac disease.<sup>3</sup>

### Indications for Testing

- **When diagnosis of celiac disease is unclear**
  - Ambiguous antibody test result (tTG IgA or EMA IgA)<sup>3</sup>
  - Equivocal small bowel biopsy results<sup>3</sup>
  - Discrepancy between antibody and biopsy findings<sup>6</sup>
- **When patient is on a gluten-free diet**
  - Antibody testing is not helpful in this setting.<sup>3</sup> HLA-DQA/DQB genotyping is unaffected by diet and is thus a useful alternative.<sup>3</sup>
  - If patient has maintained a gluten-free diet with no reduction in symptoms, HLA DQA/DQB genotyping can aid in excluding celiac as the source of symptoms.<sup>3,6</sup>
- **Evaluation of asymptomatic relatives**
  - Presence or absence of celiac-associated HLA alleles can help assess celiac risk in first-degree relatives of affected patients.<sup>3</sup>

## Scientific Excellence

- HLA-DQA/DQB genotyping provides detection of DQ2 (DQA1\*0501, DQA1\*0505, and DQB1\*0201/\*0202) and DQ8 (DQB1\*0302)
- Report includes DQ2, DQ8, half DQ2, homozygosity for DQB1\*02, and complete DQA and DQB genotypes
- Easy-to-interpret risk assessment
- Assay performed using FDA-cleared HLA test kits

## Superior Service

- Comprehensive test menu for celiac disease, including antibody testing
- Consultative services from our scientific staff
- Broad network of managed care health plans
- Nationwide network of patient service centers

## Estimated Celiac Risk from Associated HLA-DQA/DQB Genotypes<sup>7-8</sup>

Genotype	Risk
DQ2+DQ8	1:7 (14.3%)
DQ2+DQ2 or DQ2 Homozygous DQB1*02	1:10 (10%)
DQ8+DQ8	1:12 (8.42%)
DQ8+DQB1*02	1:24 (4.2%)
Homozygous DQB1*02	1:26 (3.8%)
DQ2 alone	1:35 (2.9%)
DQ8 alone	1:89 (1.1%)
General population risk (genotype unknown)	1:100 (1%) <sup>1,3</sup>
½ DQ2: DQB1*02	1:210 (0.5%)
½ DQ2: DQA1*05	1:1842 (0.05%)
No HLA-DQA/DQB susceptibility alleles	1:2518 (<0.04%)

**Note:** Actual risk for celiac disease may be greater than shown above when there are symptoms of celiac disease, positive results for celiac antibody tests or small bowel biopsy, or if there is a family history of celiac disease.

<b>Test Name</b>	Celiac disease HLA DQ association
<b>Test Number</b>	167082

Visit the online Test Menu at [www.LabCorp.com](http://www.LabCorp.com) for full test information, including CPT codes and specimen collection requirements.

### References

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2. Green PHR. Mortality in celiac disease, intestinal inflammation, and gluten sensitivity. *JAMA*. 2009;302(11):1225-1226.
3. Snyder CL, Young DO, Green PHR, Taylor AK. Celiac disease. GeneReviews. Available at: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=celiac>. Created July 3, 2008. Accessed January 20, 2011. PMID 20301720 (PubMed).
4. Green PHR. The many faces of celiac disease: clinical presentation of celiac disease in the adult population. *Gastroenterol*. 2005;128(4):S74-S78.
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6. Rashtak S, Murray JA. Tailored testing for celiac disease. *Ann Intern Med*. 2007;147:339-341.
7. Megiorni F, Mora B, Bonamico M, et al. HLA-DQ and risk gradient for celiac disease. *Hum Immunol*. 2009;70:55-59.
8. Pietzak MM, Schofield TC, McGinniss MF, and Nakamura RM. Stratifying risk for celiac disease in a large at-risk United States population by using HLA alleles. *Clin Gastroenterol Hepatol*. 2009;7:966-971.



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For more information or celiac genetic consultation, please call HLA customer service at **800-533-1037**.