

# Complete Celiac Disease Profile

*Encompasses antibody and genetic testing for superior accuracy in CD diagnosis*

*New comprehensive serology panel detects celiac cases with near 100% specificity and sensitivity. Genetic testing indicates the absence of disease risk with approximately 95% certainty.*

## INTRODUCTION

Celiac disease (CD) is an autoimmune gastrointestinal disorder that may occur in genetically susceptible individuals triggered by ingestion of gluten containing grains such as wheat, barley and rye. The advent of serological methods for the detection of antibodies to endomysium, tissue transglutaminase and gliadin have enabled large scale screening studies for CD in both Europe and the United States. These studies suggest that CD is far more prevalent than previously thought. Recent serological studies demonstrate similar incidences of CD between one in 130 and one in 500.

IMMCO Diagnostics, a leader in the field of immunopathology, has a history of pioneering research in diagnosis of CD. Our lab was the first to describe endomysial antibodies in 1984. Since then, IMMCO has continued development of various antibody assays for detecting

CD using patented and proprietary technologies with enhanced specificity and sensitivity for accurate diagnosis, even in IgA deficient patients.

## CELIAC ANTIBODY TESTING

The most common serological tests for the screening of CD are (1) the indirect immunofluorescence (IFA) method of detecting endomysial antibodies and (2) enzyme-linked immunosorbent assay (ELISA) method of detecting antibodies to tissue transglutaminase (tTG) and gliadin. The detection of endomysial antibodies aids in the diagnosis of gluten sensitive enteropathy, i.e. CD and *dermatitis herpetiformis* (DH). Patients with CD and DH are reported to have antibodies to endomysium, tissue transglutaminase and gliadin. Of the various antibody markers of CD and DH, endomysial antibodies of the IgA class employing indirect immunofluorescence seem to be the most sensitive and specific marker. Several studies have shown that endomysial



Endomysial antibody reaction in primate smooth muscle

antibodies have 100% specificity and sensitivity in diagnosing CD.

Anti-tissue transglutaminase IgA antibodies were identified later as the antigen in the endomysium specific for CD. The identification of the specific antigen led to the development of tissue transglutaminase ELISAs for both ease of use and the reduction in subjectivity associated with IFA. Tissue transglutaminase antibody levels are also useful in monitoring gluten compliance of the patients.

## Utility of serum antibody tests in CD

	Endomysial antibodies	Tissue transglutaminase	Gliadin
Sensitivity	100	100	96
Specificity	100	94	97
Positive predictive value	100	93	97
Negative predictive value	100	100	97

IgA is the major immunoglobulin class of sero-mucous secretions, part of the defense system for external body surfaces. About 1-2% of the population is IgA deficient. Measurement of IgG levels with deamidated gliadin is helpful for establishing diagnosis of CD in IgA deficient patients.

## ■ CELIAC GENETIC TESTING

Prevalence of CD is much higher in first and second degree relatives of patients with CD. CD has been associated with many other autoimmune disorders such as type 1 diabetes, thyroid autoimmunity and other autoimmune disorders. Approximately 5 percent of the patients with type 1 diabetes have CD. It has been proposed that early detection of CD may be beneficial in such cases as it is believed that adherence to a gluten-free diet may delay the onset of diabetes. This further emphasizes the need for serum antibody and genetic tests in the screening of general population.

Susceptibility to CD is associated with the inheritance of tandem alleles of the HLA-DQ genetic locus, DQA and DQB. The mode of inheritance may be in the “cis” or “trans” configurations (i.e., both alleles inherited from a single afflicted parent or one from each non-afflicted parent). Therefore, accurate assessments of genetic risk require analysis of both DQA and DQB genes.

- Testing entails high resolution typing for HLA-DQA and HLA-DQB alleles
- Most common risk factor is DQA1\*0501 in tandem with DQB1\*0201
- Less frequent risk factor is DQA1\*0301 in tandem with DQB1\*0302
- Absence of susceptibility genes suggests the absence of disease risk with approximately 95% certainty.

## ■ SPECIMEN REQUIREMENTS

**IMMCO Test Name:**  
Complete Celiac Profile

**IMMCO Test Code:** 130

**Methodology:** ELISA

**Schedule/Turnaround Time:** Assay performed daily Mon.-Fri. Report availability is within 48 hours from the time of specimen receipt.

**For antibody testing:** Collect 2-5 mL of blood in a red top or serum separator tube.

**For genetic testing:** Collect 5-10mL of uncoagulated whole blood in EDTA (purple top tubes) or in ACD (yellow top tubes).

**Sample Stability:** Specimen need not be refrigerated or frozen. Sample is stable at ambient temperature during shipment. If sample is stored prior to shipment, it is stable refrigerated (2-8°C) up to five days and frozen (-20°C or lower) up to one year.

## ■ SAMPLE SUBMISSION

Specimen collection kits are available free of charge by calling 1-800-537-8378 or e-mail request to [service@immco.com](mailto:service@immco.com).

Specimen can be shipped by courier services, U.S. Postal service and overnight carriers free of charge. Results are reported within two business days of the receipt of the specimen via mail, fax and at [immco.com](http://immco.com), a HIPAA compliant patient tracking system.

## ■ SELECTED REFERENCES

- Hill ID et al., J Pediatr Gastroenterol Nutr. 2002; 35:s78-s88.
- Fasano A et al., Arch Intern Med. 2003;163:286-92.
- Bansal et al., Ann. N.Y. Acad. Sci. 2009;1173: 36-40.
- Heap GA, van Heel DA. Semin Immunol. 2009

### Test Details and Sample Requirements

Tests	Methodology	Sample Requirements <i>Volume, Tube Color, Type</i>	Turnaround Time <i>from receipt of sample</i>	CPT Codes
<b>Genetics</b> HLA DQ	Luminex	5-10 mL lavender top, EDTA Whole Blood	48 Hours	83891 83896 83900 83912
<b>Antibodies</b>				
Endomysial	Indirect Immunofluorescence	2-5 mL red top Serum	24-48 Hours	86255
Tissue Transglutaminase	ELISA			83516
Deamidated Gliadin (Celiac G+)	ELISA			83516

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