



# Celiac Disease DNA and Antibody Testing

Celiac disease, also known as gluten-sensitive enteropathy or celiac sprue, is a chronic, autoimmune disorder affecting approximately 1:100 individuals in the U.S. population. In genetically susceptible individuals with HLA-DQ2 and/or HLA-DQ8, ingestion of gluten-containing grains, especially wheat, causes inflammation of the small intestine and leads to malabsorption. Long believed to be a rare gastrointestinal disorder, celiac disease is now known to be very common and to have a wide range of both gastrointestinal and non-gastrointestinal symptoms. In addition, many individuals have silent celiac disease with no overt clinical symptoms, though damage to the gut mucosa may nevertheless be occurring. This wide range of clinical presentation combined with variable age of onset make celiac disease extremely difficult to diagnose. Early diagnosis by antibody testing, DNA testing, and small bowel biopsy is critical. Treatment by elimination of gluten from the diet is essential for preventing future tissue damage and avoiding increased risk of other autoimmune disorders in affected individuals.

## Indications for Celiac Disease Testing

- Relatives of individuals with celiac disease
- Negative or equivocal antibody results (antiendomysial, tissue transglutaminase, or antigliadin) or intestinal biopsy results in an individual with symptoms of celiac disease
- Gastrointestinal symptoms including diarrhea, malabsorption, recurrent abdominal pain, abdominal distention, weight loss, hepatitis, and/or irritable bowel syndrome
- Iron-deficient anemia
- Persistently elevated transaminases
- Dermatitis herpetiformis
- Osteoporosis/osteopenia
- Autoimmune disease including type-1 diabetes, thyroiditis, or Sjögren's syndrome
- Children with failure to thrive, short stature, delayed puberty, irritability, and/or attention-deficit disorder
- Infertility and/or recurrent fetal loss
- Recurrent aphthous stomatitis and/or dental enamel hypoplasia of permanent teeth
- Migraine headaches, peripheral neuropathy, cerebellar ataxia, epilepsy, anxiety, and/or depression
- Individuals with Down Syndrome, Turner syndrome, or Williams syndrome

## Special Aspects of our Service:

- Rapid turnaround time
- Detailed reports with interpretation, recommendations, and education
- Genetic consultation by board-certified genetic counselors and geneticists

## Celiac Disease Testing Services

### ■ Celiac Disease DNA Test

**Specimen requirements:** 5 ml whole blood in an EDTA (lavender top) tube (ambient) or cheek cell sample

**Turnaround time:** 1 business day

### ■ Celiac Disease Antibody Panel

Tissue transglutaminase (tTG) IgA  
Endomysial antibody (EMA) IgA  
Gliadin antibody IgA, IgG  
Total IgA

**Specimen requirements:** 4ml serum (ambient)

**Turnaround time:** 2-3 business days

### ■ Combined Celiac Disease DNA Test and Celiac Disease Antibody Panel

**Specimen requirements:** 5 ml whole blood in an EDTA (lavender top) tube (ambient) or cheek cell sample

and

4ml serum (ambient)

**Turnaround time:** 2-3 days

Note: Any of these tests may also be ordered individually.

Please call Kimball Genetics for more information.

**kimball genetics** ■ ●