



Celiac Disease DNA Test

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.

Celiac disease is due to the interactions of several HLA and non-HLA genes with gluten and other environmental factors. It is strongly associated with the human leukocyte antigen (HLA) molecule DQ2 (encoded by alleles DQA1*0501 or *0505 plus DQB1*0201 or *0202) and DQ8 (encoded in part by DQB1*0302). Approximately 92-98% of patients with celiac disease carry DQ2 while the remaining 2-8% of cases carry DQ8. 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. Tissue transglutaminase antibody and antiendomysial antibody test results can be equivocal depending upon diet adherence and stage of disease. However, celiac disease DNA test results are always reliable and need only be performed once in a lifetime.

Indications for Celiac Disease DNA Testing

- Relatives of individuals with celiac disease
- Negative or equivocal antibody results (tissue transglutaminase, antiendomysial, 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

Our Celiac Disease DNA Test Service Provides:

- PCR analysis for DQ2 alleles (DQA1*0501, DQA1*0505, and DQB1*0201/*0202) and DQ8 allele (DQB1*0302)
- Detailed reports with genetic interpretation, recommendations, and education
- Free genetic counseling for physicians, patients, and families
- Free shipping

Specimen Requirements

- Cheek cell sample OR 5 ml blood in an EDTA (lavender top) tube, room temperature

Turnaround Time

- 1 business day

Please call Kimball Genetics for more information

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