

Celiac Disease

Celiac Disease (CD) is a lifelong inherited autoimmune condition affecting children and adults. When people with CD eat foods that contain **gluten**, it creates an immune-mediated toxic reaction that causes damage to the small intestine and does not allow food to be properly absorbed. Even small amounts of **gluten** in foods can affect those with CD and cause health problems. Damage can occur to the small bowel even when there are no symptoms present.

Gluten is the common name for the proteins in specific grains that are harmful to persons with celiac disease. These proteins are found in **ALL** forms of **wheat** (including durum, semolina, spelt, kamut, einkorn and faro) and related grains **rye**, **barley** and **triticale** and **MUST** be eliminated.

Celiac Disease (CD) is unique in that a specific food component, gluten, has been identified as the trigger. When individuals with CD eat gluten, the villi (tiny hair-like projections in the small intestine that absorb nutrients from food) are damaged. This is due to an autoimmune reaction to gluten. Damaged villi do not effectively absorb basic nutrients – proteins, carbohydrates, fats, vitamins, minerals and, in some cases, water and bile salts. If CD is left untreated, damage to the small bowel can be chronic and life threatening, causing an increased risk of associated disorders – both nutritional and immune related.

Dermatitis Herpetiformis (DH) is the skin manifestation of celiac disease characterized by blistering, intensely itchy skin. The rash has a symmetrical distribution and is most frequently found on the face, elbows, knees and buttocks. DH patients can have intestinal damage without obvious gastrointestinal symptoms.

Dermatitis Herpetiformis (DH) is diagnosed by a biopsy of a skin lesion and staining for IgA in the tissues. More than 85% of DH patients have small bowel sensitivity to gluten. Everyone with DH needs to follow a gluten-free diet.

ASSOCIATED AUTOIMMUNE DISORDERS

Insulin-dependent Type 1 Diabetes Mellitus, Liver diseases, Thyroid Disease-Hashimoto's Thyroiditis, Lupus (SLE), Addison's Disease, Chronic Active Hepatitis, Rheumatoid Arthritis, Turner Syndrome, Sjögren's Syndrome, Raynaud's Syndrome, Alopecia Areata and Scleroderma

OTHER DISORDERS LINKED WITH CELIAC DISEASE

Down Syndrome, Fibromyalgia, Chronic Fatigue Syndrome, Williams Syndrome

THE CAUSE OF CELIAC DISEASE

The cause of Celiac Disease (CD), also known as celiac sprue or gluten sensitive enteropathy (GSE), is still a mystery. One out of 133 people in the United States is affected with celiac disease. CD occurs in 5-15% of the offspring and siblings of a person with celiac disease. In 70% of identical twin pairs, both twins have the disease. It is strongly suggested that family members be tested, even if asymptomatic. Family members who have an autoimmune disease are at a 25% increased risk of having celiac disease.

Celiac Disease is **not** a food allergy - it is an autoimmune disease. Food allergies, including wheat allergy, are conditions that people can sometimes grow out of. This is not the case with Celiac Disease

Celiac Disease can appear at any time in a person's life. In adults, the disease can be triggered for the first time after surgery, viral infection, severe emotional stress, pregnancy or childbirth. CD is a multi-system, multi-symptom disorder. Symptoms vary and are not always gastrointestinal (GI). GI symptoms can often mimic other bowel disorders.

Infants, toddlers and young children with CD may often exhibit growth failure, vomiting, bloated abdomen, behavioral changes and failure to thrive.

CLASSIC SYMPTOMS MAY INCLUDE

- Abdominal cramping, intestinal gas
- Distention and bloating of the stomach
- Chronic diarrhea or constipation (or both)
- Steatorrhea – fatty stools
- Anemia – unexplained, due to folic acid, B12 or iron deficiency (or all)
- Unexplained weight loss with large appetite or weight gain

OTHER SYMPTOMS

- Dental enamel defects
- Osteopenia, osteoporosis
- Bone or joint pain
- Fatigue, weakness and lack of energy
- Infertility – male/female
- Depression
- Mouth ulcers
- Delayed puberty
- Tingling or numbness in hands or feet
- Migraine headaches

SOME LONG-TERM CONDITIONS THAT CAN RESULT FROM UNTREATED CD

- Iron deficiency anemia
- Early onset osteoporosis or osteopenia
- Vitamin K deficiency associated with risk for hemorrhaging
- Vitamin and mineral deficiencies
- Central and peripheral nervous system disorders - usually due to unsuspected nutrient deficiencies
- Pancreatic insufficiency
- Intestinal lymphomas and other GI cancers (malignancies)
- Gall bladder malfunction
- Neurological manifestations

A person seeking diagnosis **MUST** be following a daily diet that contains gluten for at least 4 weeks in order for test results to be accurate. Specific antibody blood tests are the initial step in screening for CD. Patients should always consult with a physician to ensure proper diagnosis.

RECOMMENDED BLOOD TESTS:

- **Anti-tissue transglutaminase antibody (tTG – IgA and IgG)**
commonly used whether or not symptoms are present and the most sensitive test available
- **Anti-endomysial antibody (EMA-IgA)** – highly specific marker for celiac disease
- **Anti-deaminated gliadin peptide (DGP – IgA and IgG)**
used when tTG or EMA is negative and in cases where patient is IgA deficient
- **Total serum IgA** – used to check levels to exclude selective IgA deficiency that results in a false negative test
- **Anti-gliadin antibody (AgA – IgG and IgA)** not considered sensitive or specific enough for adults, but used for children under 2 because tTG and EMA antibodies may be absent. The anti-DGP test is sensitive in this group.

A patient with *positive* antibody tests and a patient with selective IgA deficiency are strongly advised to consult with their physician regarding a small bowel biopsy (which is performed endoscopically). A *positive* small bowel biopsy is required to confirm the diagnosis and assess the degree of damage to the villi in the intestinal lining. Antibody test results can only suggest the presence of Celiac Disease but cannot confirm it. When antibody results and biopsy are inconclusive, or when the patient is on a gluten-free diet, genetic testing of the HLA (human leukocyte antigen) DQ2/DQ8 genes may be helpful. The specific genes DQ2 and/or DQ8 are considered necessary for Celiac Disease to develop. Since one-third of the population also has these genes, the presence of DQ2 or DQ8 does not imply that the person will necessarily develop CD, rather, that they have a genetic predisposition to CD.

Genetic testing does not diagnose Celiac Disease - its largest benefit is that the absence of DQ2 and DQ8 essentially excludes CD.

The onset of Celiac Disease can occur at any time in a person's life. Once a person is diagnosed, family members should be urged to get tested as well.