CELIAC DISEASE UPDATE

How Genetic Markers Aid Diagnosis

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This article describes celiac disease and the genetic markers that confer susceptibility to its development. This article includes an overview of symptoms and potential complications, and it explains how testing for genetic markers can aid with diagnosis.

Gluten Sensitivity

Celiac disease, which is also known as gluten sensitivity enteropathy or GSE, is an autoimmune disorder characterized by intolerance to wheat gluten or related proteins found in rye and barley. Gluten is a fraction of wheat flour, which is composed of two groups of proteins, the glutenins and the gliadins. Individuals with celiac disease are intolerant to the gliadin fraction of gluten and to equivalent proteins found in rye and barley.

Environmental Triggers

Celiac disease is unique in that the environmental factor that causes disease in persons with specific immune system disease has clearly been identified and is required for disease expression. Celiac disease is triggered by gluten protein found in wheat, rye, and barley. In some forms of gluten sensitivity, patients react to only specific subtypes of gluten.

Signs and Symptoms

Even in the absence of symptoms, individuals with celiac disease who continue to ingest gluten develop changes in their small intestines. These changes include villous atrophy, a destruction of the intestinal villi that absorb nutrients, invasion of the gastrointestinal tract by lymphocyte white blood cells, and crypt hyperplasia, a form of cellular overgrowth. Intestinal symptoms include abdominal pain, distension, bloating, vomiting and diarrhea.

In children, growth may be stunted. While the intestines are primarily affected, celiac disease also causes systemic changes affecting the skin, liver, bones, joints, heart, brain, and other organs. Long-term complications of untreated celiac disease include osteoporosis, increased risk of bone fractures, infertility, and an increased risk for small bowel malignancy. The only treatment for celiac disease is adherence to a gluten-free diet.

Incidence and Diagnosis
Recent studies show that the risk for celiac disease in North America and Europe ranges from 0.4 to 1 percent or about 1 in every 250 people. Traditionally, antibody tests for gliadin, endomysial, and tissue transglutaminase antibodies, and intestinal biopsy have been used to diagnose celiac disease.

In addition, tests for HLA genotypes DQ2 and DQ8 found on chromosome six are available for diagnosing celiac disease. More than 90 percent of patients with celiac disease have DQ2 and most remaining patients, about 8 percent, have the DQ8 allele. The molecules formed by these genetic markers present gluten to the immune system, which drives the autoimmune disease process. However, these markers are present in individuals who do not develop celiac disease. Thus, the test is more suited for determining the genetic risk or probability of developing celiac disease or for ruling out the possibility of developing celiac disease (negative results for these genetic markers).

**Disease Susceptibility**

Individuals with other autoimmune diseases such as type 1 diabetes, Addison's disease, and autoimmune thyroid disease are more likely to have these genetic markers, making people with these disorders more susceptible to celiac disease. Celiac disease is also more common in certain genetic diseases such as Down's, Turner's and William's syndrome.


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