Megan Ryan, BSN, RN
Sheila Grossman, PhD, RN, APRN, FNP-BC

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Celiac Disease

Implications for Patient Management

ABSTRACT

Celiac disease is an autoimmune disorder that is known specifically for causing inflammation of the mucosa in the small intestine. Through multiple diagnostic and screening tools such as small intestinal biopsy sample, serological testing, and human leukocyte antigen testing, healthcare providers can diagnose this disease that contains components related to genetic predisposition and intake of gluten proteins found in wheat, barley, and rye. There are some who believe that having an autoimmune disease may predispose one to acquiring another disease. With patients experiencing mostly diarrhea, abdominal pain, and weight loss, the implementation of a gluten-free diet is the treatment that healthcare providers recommend. Through monitoring gluten intake and providing nutritional supplementation, those diagnosed with celiac disease can lead a relatively normal life without complications. With celiac disease affecting all age ranges in the population, and with a documented higher frequency, there is a growing awareness in society that can be easily seen in grocery stores, restaurants, and food manufacturers.

Celiac disease is an autoimmune disorder that can occur at almost any age with cases ranging from childhood to adulthood. The development of this disease can be linked to consumption of gluten proteins and genetic predisposition with most patients expressing the human leukocyte antigen (HLA), HLA-DQ2, or HLA-DQ8 (Presutti, Cangemi, Cassidy, & Hill, 2007). This disease is known especially for causing inflammation of the mucosa in the small intestine; however, it can also affect the skin, joints, uterus, brain, heart, and other organs (Martin, 2007).

Background

Celiac disease usually presents with gastrointestinal symptoms including, but not limited to, diarrhea, emaciation, stomatitis, dyspepsia, and abdominal pain (Green & Cellier, 2007). Diagnosis of celiac disease can be difficult and in the past was a diagnosis of exclusion. Now there are several tests available that include small intestinal biopsy sample, serological testing, and HLA typing. The purpose of this article is to describe the pathophysiology, signs and symptoms, diagnostics, and treatment of celiac disease, and to offer suggestions as to how to facilitate patients in managing the disease.

Pathophysiology

The pathophysiological process that occurs during celiac disease is related to the gluten proteins that cannot be fully digested by gastric, pancreatic, and intestinal brush-border membrane proteases (Green & Cellier, 2007). Gluten proteins can be found in many processed foods, but specifically in rye, wheat, and barley products. When the proteins are unable to be digested because of a deficiency of enzymes, they stimulate an inflammatory process in the intestine. This inflammatory process usually occurs in the upper small intestine where the lamina propria and the epithelium are infiltrated by interferon-y, which is produced by intestinal CD4 T cells, which cause the villous atrophy (Green & Jabri, 2003).

Ingesting gluten proteins in one's diet is not the only reason for celiac disease to occur. Research has shown that there is a genetic predisposition to its development. The immune response against gluten proteins

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About the authors: *Megan Ryan, BSN, RN, is at Yale New Haven Hospital, New Haven, Connecticut.*

Sheila Grossman, PhD, RN, APRN, FNP-BC, is Professor and Coordinator, FNP Specialty Track, Fairfield University School of Nursing, Fairfield, Connecticut.

Correspondence to: *Sheila Grossman, PhD, RN, APRN, FNP-BC, FNP Specialty Track, Fairfield University School of Nursing, 1073 North Benson Road, Fairfield, CT 06824 (e-mail: sgrossman@fairfield.edu).*

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has been linked to HLA-DQ2 or HLA-DQ8 (Presutti et al., 2007). The presence of these genes is necessary but not sufficient to the development of the disease. HLA-DQ2 is expressed in more than 90% of the people with celiac disease (Green & Jabri, 2003). If HLA-DQ2 or HLA-DQ8 is absent, there is little likelihood that the patient will have celiac disease.

Signs and Symptoms

Many patients present with diarrhea, abdominal pain, and weight loss, but various presentations can be experienced depending on the patient's age. Infants and toddlers can present with symptoms as early as 6 months when gluten is first introduced into their diet. This population usually presents with the typical symptoms of diarrhea, abdominal distention, impaired growth, decreased appetite, muscle wasting, weight loss, lethargy, and irritability (Martin, 2007). The adolescent population can present with short stature, iron-deficiency anemia, or neurological symptoms such as peripheral neuropathy, ataxia, or epilepsy. If celiac disease is neglected by the patient, it can have serious results such as lymphoma if the intestines are inflamed and allowed to progress to an irreversible state (Murray, 1999).

With celiac disease mostly affecting the proximal small intestine, iron, folic acid, calcium, and fat-soluble vitamins are malabsorbed leading to iron deficiency, folate deficiency, and reduced bone density that is often not seen until adulthood (Green & Jabri, 2003). The most common sign of celiac disease in adults is diarrhea, which may also be paired with abdominal pain or discomfort. In addition to the typical symptoms of celiac disease, the adult population can also present with dermatitis herpetiformis (a pruritic vesicular rash found mainly on the knees, elbows, and scalp), hypoproteinemia, hypocalcemia, elevated liver enzyme levels, and a previous diagnosis of irritable bowel syndrome (IBS) (Green & Cellier, 2007).

With IBS, patients experience a combination of chronic or recurrent gastrointestinal symptoms such as abdominal pain and irregular patterns of defecation with constipation or diarrhea (Wahnscaffe, Schulzke, Zeitz, & Ullrich, 2007). Despite research and various diagnostic tests, no pathological connection has been made between these symptoms. A significant number of IBS patients have wheat fiber intolerance; therefore, it has been noted that these patients benefit from a gluten-free diet due to the wheat fiber exclusion more than the actual gluten withdrawal (Lea & Whorwell, 2005).

Diagnosis

Patients who undergo an endoscopy can also reveal signs of villous atrophy that is common with celiac disease. The endoscopy can show the signs of villous atrophy including a reduction in the number of circular folds in the descending duodenum, scalloping of folds, mucosal fissures, and a mosaic or nodular appearance to the mucosa (Green & Jabri, 2003). The provider has to be careful of solely basing diagnosis on this sign because these abnormalities are not necessarily specific to just celiac disease.

The diagnosis of celiac disease can be formed through a multitude of testing including small intestinal biopsies, histological testing, HLA typing, and improvement of clinical symptoms with the gluten-free diet. A small bowel biopsy will allow the provider to view any mucosal changes in the form of villous atrophy, intraepithelial lymphocytosis, or crypt hyperplasia (Green & Cellier, 2007). These changes may be sporadic throughout the intestine, so it is important to obtain several samples in various spots for histological testing. It is also significant to recognize that celiac disease is not the only cause of villous atrophy, which may lead to the usage of other tests for diagnosis (Hill, 2005).

Serological testing for celiac disease is most commonly seen as serum immunoglobulin A (IgA) endomysial antibodies and IgA tissue transglutaminase (tTG) antibodies. Research has found the sensitivity and specificity of testing for IgA endomysial and tTG antibodies to be greater than 95% (Presutti et al., 2007). The titers of endomysial antibodies and anti-tTG antibodies correlate with the degree of mucosal injury, where the sensitivity of the tests declines when patients have a lesser amount of villous atrophy (Green & Cellier, 2007). Serological testing may result in false-positive or false-negative results, so it cannot be relied on for primary diagnosis of celiac disease.

While most patients have a genetic predisposition for celiac disease, HLA may prove significant in confirming diagnosis of celiac disease. HLA testing proves to be a reliable test for detecting those with HLA-DQ2 and HLA-DQ8. Those who do not possess this allele will most likely not have celiac disease.

Treatment and the Gluten-Free Diet

Because the consumption of gluten plays a major factor in celiac disease, it is usually advised that the newly diagnosed patient starts on the strict gluten-free diet. The foundation of this diet is to stay away from the grains wheat, rye, and barley. While it is easy to read these ingredients on the back of a food label, there are some other variations of the term "wheat" to look for such as "enriched flour," "self-rising flour," "white flour," or "semolina." Food products that are usually made from barley include beer, malt, malt syrup, and malt extract. Many processed foods contain grains that are contraindicated for those with celiac disease such as candy, cold cuts, hot dogs, french fries, gravies, sauces, soups, and vegetables in sauce (Thompson, 2006).

Even though there seems to be many limitations, there are other grains that are allowed in a gluten-free

diet. These include, but are not limited to, buckwheat, corn, millet, rice, and quinoa. If one cannot find a natural food store nearby, many supermarkets have a natural food section where gluten-free flour, bread, pasta, and breakfast cereal are sold. When one is not in a controlled cooking environment outside of the home, it is paramount that the chef or host is aware of the patron's dietary constraints. In restaurants, breads and sauces are always food concerns for those with celiac disease and cross-contamination is an issue during preparation (Thompson, 2006). The individual with celiac disease has to verbalize their issues and stay persistent to ensure that their food is gluten-free.

The newly diagnosed patient may have many questions, even after seeing a nutritionist and continue to have questions years later. There are support groups for celiac disease and many Web sites and books that offer information. If one has a question about a certain food and its ingredients, many food companies have Web sites and phone numbers that the consumer can call. Ultimately, more and more food manufacturers will recognize the growing need for developing more gluten-free food products.

It is important to recognize and treat the other nutritional deficiencies that are associated with malabsorption and celiac disease. Iron, folic acid, vitamin B_{12}, and fat-soluble vitamin levels should be monitored and replaced to their adequate levels. Thyroid dysfunction is also seen in celiac disease, so thyroid function tests should be monitored. Osteoporosis is a common result of celiac disease, so bone density values need monitoring for vitamin D deficiency and calcium malabsorption (Green & Jones, 2006). Research focusing on other treatments for celiac disease that do not revolve around the implementation of the gluten-free diet is also in progress. Currently, research is using recombinant enzymes that digest the toxic gliadin fractions in the stomach or upper small intestine (Gass, Bethune, Siegel, Spencer, & Khosla, 2007). There has also been the usage of treatment where the immune response is interfered with, such as blocking the binding of deaminated gliadin to HLA-DQ2 or HLA-DQ8 or by blocking the action of tTG (Green & Cellier, 2007); however, these therapies are more likely to have potential adverse effects.

Case Study of a Man With Celiac Disease and Multiple Sclerosis

The individual highlighted in this case study is a Caucasian man in his middle 50s who was diagnosed in 2007 with celiac disease. When asked about his signs and symptoms that led him to further question his health, he described consistently experiencing a stomachache after eating just about any type of food. In addition, he noticed a 10-lb weight loss in 5 weeks. His first thought was fear because these signs could mean cancer. The individual went to his primary care provider, who ordered endoscopic evaluation of his esophagus, stomach, and intestine. He never tested positive for the celiac disease allele but did have a medical history that included multiple sclerosis, which is considered an autoimmune disease; therefore, the medical team believed that this autoimmune disorder (e.g., multiple sclerosis) caused the development of the second autoimmune disorder (celiac disease). Patients with type 1 diabetes mellitus, Down syndrome, Turner syndrome, or an associated autoimmune disorder are at increased risk of celiac disease (Presutti et al., 2007).

After basic teaching from the provider, the individual was sent to a nutritionist who reviewed his diet and reinforced the importance of being hypersurveillant when he read the back of food labels. The individual went on the gluten-free diet and commented on its effectiveness. One has to look carefully at the labels, search for words such as "modified," and stay away from fast food. Even if a restaurant is gluten-free friendly, the person diagnosed with celiac disease has to have zero tolerance for shortcuts in preparing food. In some cases, the food needs to be prepared with completely different pans, utensils, and in a separate part of the kitchen. Once on the gluten-free diet, this individual felt more comfortable and had no symptoms unless he inadvertently ate gluten (Leffler, Saha, & Farrell, 2003). Many of his signs and symptoms of multiple sclerosis have been mitigated, and this helped to control his weight.

This individual was an ideal candidate for health teaching and management. Through good fitness and health, he believes that the diagnosis of celiac disease has improved his life. It takes an individual enormous commitment to understand the disease and the strength to stay strong to the diet limitations; however, once an individual develops a personalized plan that works, one can live with the disease on the gluten-free diet without any additional medications.

Conclusion

Celiac disease is a common disorder seen in a growing number of the population. Once the individual is diagnosed, there are multiple health-teaching suggestions to assist celiac disease patients with managing their diet and decreasing or even obliterating symptoms of the disease. There is an increased awareness toward celiac disease in society, which, it is hoped, will lead to an increase in research, testing, and endeavors to find new therapies for those diagnosed.

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