
prevalence in Europe, where subsequent small intestine biopsies have confirmed celiac disease in all patients testing positive for antiendomysial antibody (positive predictive value: 99 percent).⁵ The authors of the U.S. study⁴ conclude that data suggest that celiac disease may be greatly underdiagnosed and is relatively common in this country. ...

Pathogenesis: Normally, ingested food does not elicit a local or systemic immune response. Ingestion of protein down-regulates the intestinal immune response to that protein. This phenomenon is known as oral tolerance.⁷ In patients with celiac disease, the immune system is abnormally activated by gluten, specifically the gliadin portion of wheat protein, and prolamines (insoluble proteins) in rye, barley and oats.⁸ Thus, celiac disease is a genetic, immunologically mediated, small intestine enteropathy in which mucosal villi are destroyed by cellular and humoral-mediated immunologic reactions to gliadin protein.⁹ The loss of functioning villi limits the ability of the small intestine to absorb nutrients, thus adversely affecting all systems of the body. The immunologic response to gluten may also occur secondarily in other bodily tissues, an example being dermatitis herpetiformis.

Studies of patients with celiac disease using molecular techniques demonstrate a strong association with specific HLA class II genotypes. Approximately 95 percent of patients with celiac disease have a particular type of HLA DQ alpha and beta chain encoded by two genes, HLA-DQA1 0501 and HLA-DQB1 0201.¹⁰ If people genetically predisposed to celiac disease do not ingest gluten, they have no manifest illness. Delaying ingestion of gluten products through breast feeding or dietary habits may change or delay the onset of disease.¹¹ Viral exposures may trigger an immunologic response in persons genetically susceptible to celiac disease; this occurs with adenovirus type 12, which shares a sequence of eight to 12 amino acids with the toxic gliadin fraction.¹²

Clinical Presentation

Infancy: During the first year of life, an infant may manifest celiac disease with intermittent vomiting, diarrhea, growth delay and failure to thrive. The incidence of this early classic presentation in infants has decreased. However, to prevent significant growth problems in infants, confirmation of celiac disease is important¹³ (*Figure 3*).

Childhood: Children with celiac disease may present with short stature, anemia, hepatitis, epilepsy and other extra-gastrointestinal conditions. With age, these presentations become more subtle. In one study¹³ of a group of school children screened for IgA anti gliadin antibodies, positive titers were found in 19 of the children. Endoscopic biopsies were performed in 18, and villous atrophy was found in 12. None of these children had shown characteristic symptoms of celiac disease. The most frequent of their symptoms were abdominal pain, aphthous stomatitis and atopic dermatitis (*Figures 4 and 5*).

Angular cheilitis (*Figure 6*) and recurrent aphthous ulcers (*Figure 7*) are frequent in children and adults with celiac disease.¹³ These clinical findings should prompt the physician to consider the diagnosis of celiac disease.

Young Adults: The initial presentation of celiac disease in patients in their 20s and 30s may be dermatitis herpetiformis. This condition usually appears as clear or blood-tinged vesicles symmetrically distributed over the extensor areas of the elbows, knees, buttocks, shoulders and scalp (*Figure 8*). Intense pruritus and/or burning sensations in the area occur hours before the onset of the vesicle. Dermatitis herpetiformis flares after consumption of foods containing a high amount of gluten.

Small intestine biopsies from patients with dermatitis herpetiformis reveal features identical to those found in patients with celiac disease.¹⁴ In a study¹⁵ of 212 patients with dermatitis herpetiformis who were managed over a period of 25 years with a gluten-free diet, several benefits of dietary therapy were found, including (1) the patients' need for medication was reduced or abolished, (2) the enteropathy resolved and (3) patients experienced a feeling of well-being after beginning the diet.

In a study¹⁶ of the occurrence of malignancies and the survival of 305 patients with dermatitis herpetiformis from 1970 to 1992, it was indicated that the incidence of non-Hodgkin's lymphoma is significantly increased in patients with dermatitis herpetiformis. The results also confirmed no increase in mortality in patients with dermatitis herpetiformis who are treated with a gluten-free diet.

Adults : Malabsorption. The varied signs and symptoms of malabsorption may be caused by celiac disease or many other diseases. Mild malabsorption may be asymptomatic. With its gradual onset, the classic manifestations of flatulence and bulky, greasy and foul-smelling stools may not be recognized by the patient as signs of celiac disease. Malabsorption should be suspected in any patient with weight loss and diarrhea, and the signs and symptoms of specific vitamin or nutritional deficiencies. The latter include visual disturbances, neuropathy, anemia, osteopenic bone disease, tetany, hemorrhagic diathesis or infertility.

In celiac disease, the clinical symptoms are determined by the severity and the proximal-to-distal extent of the intestinal lesions. Symptoms often manifest in childhood and then disappear, only to recur in adulthood. In some patients, the disease presents initially in their 60s and beyond. ...

(Full text, charts and illustrations available at <http://www.aafp.org/afp/980301ap/pruessn.html>)
