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Diagnosis of coeliac disease

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The diagnosis of coeliac disease depends on the finding of characteristic, though not specific changes, of intraepithelial lymphocytosis, crypt hyperplasia and various degrees of villous height reduction identified in duodenal biopsies of individuals ingesting a gluten containing diet, together with symptomatic and histologic improvement on gluten withdrawal. Serologic testing has become the main mode of determining who will undergo biopsy. The IgA endomysial antibody and IgA tissue transglutaminase antibody have approximately 90% sensitivity and specificity, though there are reports of lower sensitivity and specificity in the clinical practice setting. This is due to lower titers of these antibodies in the presence of lesser degrees of mucosal damage. The widespread availability of serologic tests for coeliac disease allows the diagnosis to be considered by any physician. Gastroenterologists will be required to interpret the results of serologic tests and perform duodenal biopsies when indicated. Pathologists likewise need to be better acquainted with the more subtle changes of cell mediated immunity within the mucosa that are suggestive of underlying gluten sensitivity.

Key words: intraepithelial lymphocytosis; serologic tests; endomysial antibody; tissue transglutaminase antibody; selective IgA deficiency.

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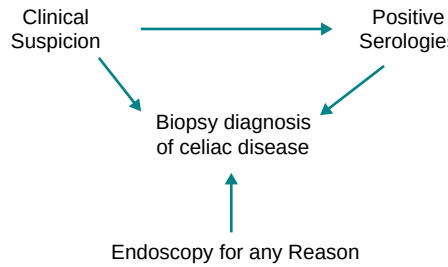

Figure 1. Diagnosis of coeliac disease.

Coeliac disease is an inflammatory condition of the small intestine with a varied clinical presentation. In its most characteristic form, coeliac disease is heralded by the presence of mucosal inflammation, absence of villi, and crypt hyperplasia, which occur upon exposure to gluten, and clinical and histological improvement with withdrawal of gluten from the diet.[1]

Clinically, coeliac disease represents a spectrum of clinical features and presentations. Although *classical* coeliac disease is the most commonly described presentation in text books, most individuals have a clinically occult or inapparent form. Other presentations may be atypical, as with joint pains, abdominal pains or abnormalities in hepatic transaminases.

The diagnosis of coeliac disease is usually made on the basis of an abnormal duodenal biopsy. Patients may come to biopsy (Figure 1) because of a clinical suspicion, positive serologies or because of 'routine' biopsies or abnormalities detected at the time of endoscopy.[2] Typical biopsy findings result in a presumptive diagnosis of coeliac disease and institution of a gluten-free diet. Definitive diagnosis is only established after demonstration of a response to the diet.[3] Issues discussed in this paper include who should be tested for coeliac disease and what is the most effective way to establish the diagnosis.

WHO SHOULD HAVE SEROLOGIC TESTING?

The clinical suspicion of coeliac disease may be based on the presence of gastrointestinal symptoms, i.e. those with classical, diarrhea-predominant syptoms with or without a malabsorption syndrome. The next most common group in whom the diagnosis should be considered are those with manifestations of malabsorption of nutrients or vitamins. This includes patients with iron deficiency anemia in the absence of gastrointestinal bleeding, folic acid deficiency, manifestations of calcium or vitamin D malabsorption including osteoprosis, unexplained hypocalcemia, vitamin D deficiency and secodary hyperparathyroidism. Vitamin E deficiency and vitamin K deficiency could also be secondary to coeliac disease. Unexplained weight loss should also prompt serologic screening. Serologic assessment of the presence of coeliac disease should be performed in the evaluation of abnormal liver function tests (elevated transaminases) (Table 1).[4]

Coeliac disease is associated with many autoimmune diseases.[5,6] In some of these conditions the association is very strong, as a result serologic screening for coeliac