

# Diagnosis of Gluten-Sensitive Enteropathy (Celiac Disease)

A Summary of the NASPGHAN, AGA and WGO Guidelines

Celiac disease is an immune medical condition that is caused by ingestion of gluten in genetically susceptible individuals. The damage to the absorptive surface of the small intestine by gluten results in an inability to absorb nutrients. The prevalence is between 1:100 and 1:300 in the North American population. Celiac disease may present with a wide variety of symptoms (Table) at any point in life. The prevalence is higher in specific associated conditions (Table)

## PRACTICE POINT

The classic form of celiac disease can manifest at any age with weight loss, diarrhea, abdominal distention, and occasionally, severe malnutrition. Older children may present with constitutional short stature, delayed puberty or dental enamel defects. Children and adults may present with iron or folate-deficiency anemias.

Many symptoms (e.g., anemia, weight loss, bone pain, paresthesia, edema, skin disorders) are secondary to deficiency states. If intestinal symptoms (e.g., diarrhea, abdominal discomfort, distention) do not occur, the diagnosis celiac disease may not be suspected.

**TABLE 1 (Symptoms include but are not limited to one or more of the following)**

### Classic Symptoms

Abdominal distension  
Abdominal pain  
Chronic diarrhea  
Anorexia  
Irritability  
Weight loss or failure to thrive in children  
Muscle wasting  
Dermatitis herpetiformis

### Associated Conditions (% affected)

Relative of individual with celiac disease (8-15%)  
Type 1 diabetes mellitus (4-8%)  
Autoimmune thyroiditis (2-5%)  
Trisomy-21 (Down syndrome) (2-5%)  
Turner syndrome (2-5%)  
IgA deficiency (1-4%)

### Non-classic Symptoms and Signs

Unexplained iron or folate deficiency anemia  
Persistent vomiting  
Chronic constipation  
Irritable bowel syndrome  
Aphthous stomatitis  
Dental enamel defects  
Arthritis  
Osteoporosis  
Delayed puberty  
Short stature  
Abnormal liver enzymes (ALT/AST)  
Infertility  
Neurological presentations  
Unexplained ataxia or peripheral neuropathy  
Epilepsy with occipital calcifications  
Depression

## Screening and Diagnosis of Celiac Disease

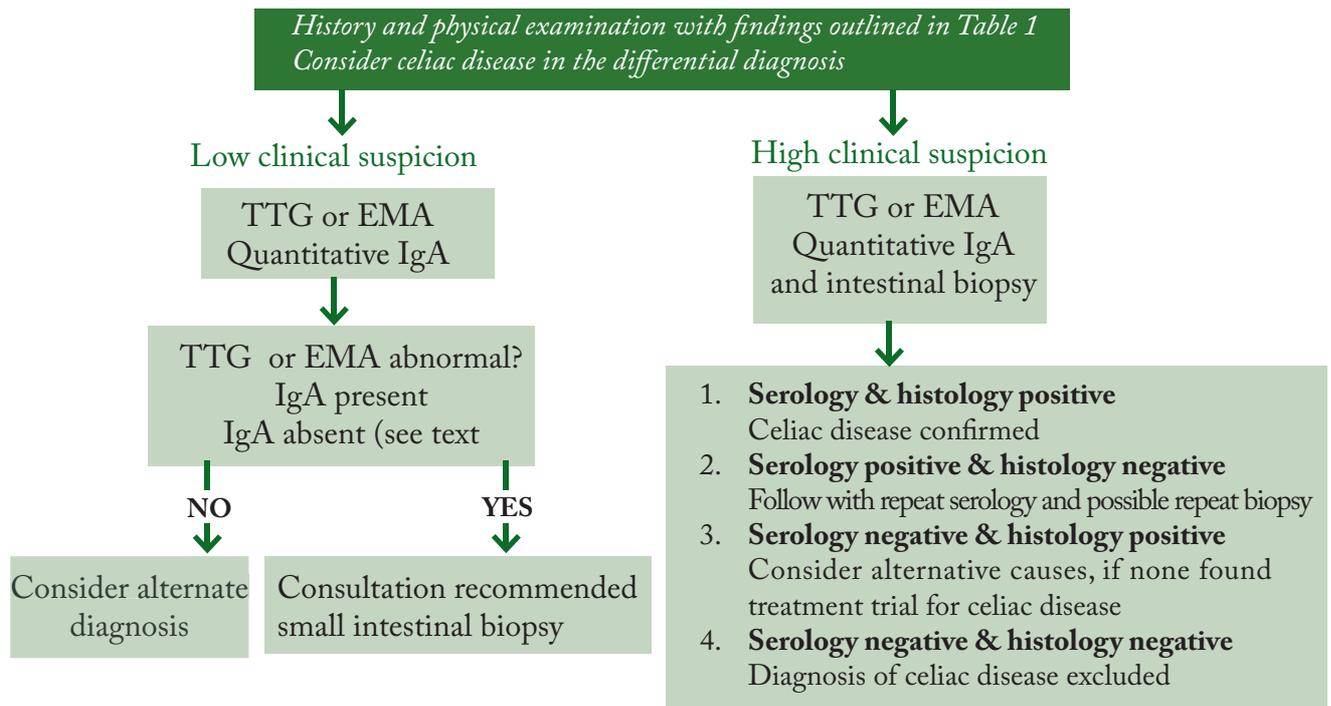
- Diagnosis of celiac disease may be suspected in individuals with one or more of the above symptoms, signs or associated conditions. Screening serologic tests can be utilized to identify individuals at risk for celiac disease BUT the diagnosis MUST be confirmed with small bowel biopsy. It is recommended that the biopsy be done BEFORE starting the patient on a gluten-free diet in order to confirm the diagnosis. **Celiac disease requires lifelong treatment with a gluten-free diet.** The diet is complicated and expensive. Initiation of the diet requires counseling by a qualified and knowledgeable dietitian.

## PRACTICE POINT

Screening tests and intestinal biopsy need to be performed while the patient is on a **gluten-containing diet.**

- IgA antibody human recombinant tissue transglutaminase (IgA-TTG) or endomysium (IgA-EMA) are recommended for initial testing by experienced laboratories. The choice of test depends on laboratory preference. Both tests display positive and negative predictive values of about 90% in IgA sufficient patients. Both tests require the presence of IgA and will be falsely negative in IgA deficient patients. The prevalence of IgA deficiency is increased in individuals with celiac disease and therefore screening for IgA deficiency should be performed at the same time as the serology tests.
- IgA or IgG anti-gluten antibody tests are no longer recommended as a screening test for celiac disease because of their very poor positive and negative predictive values.

# Algorithm for the Evaluations of Celiac Disease



**Note:** Individuals with CD who are also IgA deficient will not have elevated levels of TTG or EMA. Celiac disease occurs in 1-4% of people with IgA deficiency. All symptomatic patients should be referred for intestinal biopsy regardless of their serology results because false negative serological tests can occur. In asymptomatic individuals with IgA deficiency, the laboratory may be able to perform IgG-TTG.

## Management

1. Prescribe gluten free diet for life
2. Treat specific nutrient deficiencies if identified e.g.. Iron, Vitamin D, calcium, folate
3. Refer to dietitian with expertise in management of celiac disease
4. Refer to Canadian Celiac Association, an excellent resource and support group. National office in Mississauga , ON (905-507-6208; 800-363-7296; www.celiac.ca ).
5. Advise bone mineral density if suspect osteoporosis
6. Monitor for symptom resolution and repeat serology in ~ 6 mo.
7. Screen 1st degree relatives for celiac disease

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Bai, J, Zeballos E, Fried GR et al; WGO-OMGE practice guideline: Celiac disease February 2005 Available at url: [www.worldgastroenterology.org/globalguidelines/guide13/guideline13.htm](http://www.worldgastroenterology.org/globalguidelines/guide13/guideline13.htm) Accessed Jan 15, 2006.

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