

Celiac Disease (EGD-1.18)

The diagnosis of celiac disease (CD) incorporates both serologic and histologic data. Serologic testing consists of measuring tissue transglutaminase (TTG) IgA while on a gluten-containing diet with concomitant or prior measurement of total IgA levels to ensure that patient is not IgA deficient. Patients with an elevated TTG IgA should then undergo an esophagogastroduodenoscopy (EGD) with duodenal biopsy to confirm the diagnosis. Due to the fact the TTG IgA in patient without IgA deficiency has a high negative predictive value when the pretest probability is low moderate, CD can be considered as adequately ruled out in this scenario without undergoing an EGD.

The specificity of the TTG IgA ranges between 96-100% with sensitivity variable between 63-93%. Therefore, in patients who carry a >5% chance of having celiac disease, EGD with duodenal biopsy should be considered even with a negative serology. This includes certain genetic and autoimmune conditions as well as first-degree relatives of individuals with CD. This is based on the sensitivity of TTG IgA serology, risk of verification bias on studies assessing celiac testing, the possibility of seronegative celiac disease, and differential diagnosis with other enteropathies. If a patient is IgA deficient, then IgG serology with either deamidated gliadin peptide (DGP) and/or TTG should be measured.

Genetic testing is not required for diagnosis in all cases, but in select circumstances can be helpful, such as in the context of serologic-histologic discrepancy and in individuals who have already started a gluten-free diet. If negative, celiac disease can be considered as effectively ruled out.

Since histologic abnormalities in celiac disease can be patchy, at least four biopsies should be obtained in the post-bulbar duodenum and at least 1-2 from the duodenal bulb. This results in approximately 96% sensitivity in diagnosing celiac disease.

A gluten-free diet (GFD) is the only effective therapy for CD, and intestinal biopsies are the only way to document mucosal healing of the intestine. This takes time after starting a GFD, and does not always correlate with either serologies nor symptoms. Given that the lack of mucosal healing can be associated with increased risk of lymphoproliferative malignancy, bone disease, and refractory celiac disease (RCD). In the United States, a study demonstrated that the median time from implementing a GFD to achieving mucosal healing was three years; as such, a follow-up biopsy after two years of GFD to assess for mucosal healing appears reasonable.

References

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