

Screening: Importance of Early Diagnosis in Celiac Disease

Key Points

- Symptoms of celiac disease can be easily confused with the symptoms of other gastrointestinal disorders such as irritable bowel syndrome, inflammatory bowel disease,¹ and small intestinal bacterial overgrowth. In some cases, symptoms may even appear unrelated to the gastrointestinal tract (foggy head, arthritis, fractures, fatigue, infertility, headaches).
- Children can present with delayed puberty, short stature or poor weight gain. Symptoms may not be easily evident in some children if they are unable to identify or communicate them effectively.
- Since celiac disease causes damage to the villi of the small intestine, further health disorders can arise from malabsorption of proper nutrients, such as weak bones, iron deficiency, and vitamin deficiencies. Timely diagnosis of celiac disease allows these manifestations to be assessed and treated before further damage occurs.^{2,3}
- Untreated celiac disease can lead to serious health concerns, such as infertility, end stage liver disease or lymphoma (a type of white blood cell cancer). The risk of these diseases returns to normal when following a strict gluten-free diet.

Screening Recommendations:

- Immediate family members (1st degree relatives) of a person with celiac disease have ~8 percent chance of testing positive for celiac disease.⁴ Therefore, it may be advisable for family members to be tested if they have symptoms.
- It is widely accepted that children with close relatives with celiac disease should be tested regularly. All children with a first degree relative with celiac disease should be tested, whether they have symptoms or not. Children with a second or third degree relative with celiac disease should also be tested if they have gastrointestinal or non-gastrointestinal symptoms consistent with celiac disease.
- The standard screening tests for children are total IgA and tissue transglutaminase IgA (IgA-tTG). If a child has a selective IgA deficiency, a deamidated gliadin IgG (DGP) and tissue transglutaminase IgG should be sent. Repeat testing should occur every 3-5 years or sooner if

new symptoms develop. All children with positive blood tests for celiac disease should see a pediatric gastroenterologist before starting a gluten-free diet. The next steps may involve an upper endoscopy. In adults, every one with positive bloodwork should have an upper endoscopy.⁵

- There is concern that celiac blood testing can be less reliable in patients under the age of two years. So, if a child is asymptomatic, testing should be delayed until a child turns two years old. However, if symptoms suggestive of celiac disease are present, testing should be done earlier and a pediatric gastroenterologist should be consulted.
- As in adults, all testing for celiac disease in children should be done on a **gluten-containing diet**. Unfortunately, the tests available today are not accurate if a child is on a gluten-free diet. Daily gluten ingestion equal to 1-2 slices of bread a day for at least 8 weeks is recommended before endoscopy testing. If a child has significant symptoms eating gluten, a pediatric gastroenterologist should be consulted for further recommendations.
- Experts are divided as to whether adults who display no symptoms should be screened for celiac disease. Some believe that everyone should be screened since there is a clear treatment (the gluten-free diet) and untreated celiac disease can lead to serious complications.⁵ Others believe that we should not screen asymptomatic people because we do not know enough about celiac disease when symptoms are not apparent, and whether symptoms will ever appear.⁶ It is also not uncommon that individuals who thought they were asymptomatic report symptoms when asked specifically about them⁸
- After reviewing the available literature, guidelines from US Preventive Services Task Force found that the current evidence is insufficient to assess the balance of benefits and harms of screening for celiac disease in asymptomatic persons not at risk for celiac disease.⁷ The resources listed below, and talking to your doctor, can help you learn more about this controversy and decide for yourself if you should be tested.

Take Home Messages:

1. Symptoms vary and can be similar to other diseases. The only way to know the cause of your symptoms is to see your doctor and request a tTG blood test.
2. Being diagnosed and starting the gluten-free diet can prevent further damage in the body and allow it to heal once on a strict gluten-free diet.
3. If you do NOT have symptoms but think you might have celiac disease, consider getting screened for celiac disease if you are at risk, based on family history.
4. All children with a close relative with celiac disease should be tested for celiac disease while on a gluten containing diet.

References:

1. NIH Publication No. 08–4269 September 2008.
<http://digestive.niddk.nih.gov/ddiseases/pubs/celiac/> Accessed July 9, 2018..
2. Celiac Disease Center at Columbia University: Diagnosis of Celiac disease.
<http://celiacdiseasecenter.columbia.edu/celiac-disease/diagnosis> . Accessed July8, 2018.
3. Corrao G, Corazza GR, Bagnardi V, Brusco G, Ciacci C, Cottone M, Guidetti CS, Usai P, Cesari P, Pelli MA, Loperfido S, Volta U, Calabro A, Certo M. Mortality in patients with coeliac disease and their relatives: a cohort study. *Lancet* 2001; 358:356-61.
4. Singh P, Arora S, Lal S. Risk of celiac disease in the first- and second-degree relatives of patients with celiac disease: a systematic review and meta-analysis. *Am J Gastroenterol*. 2015;110:1539-48.
5. Rubio-Tapia A, Hill ID, Semrad C, Kelly CP, Greer KB, Limketkai BN, Lebwohl B. American College of Gastroenterology Guidelines Update: Diagnosis and Management of Celiac Disease. *Am J Gastroenterol*. 2023 Jan 1;118(1):59-76. doi: 10.14309/ajg.0000000000002075. Epub 2022 Sep 21. Erratum in: *Am J Gastroenterol*. 2024 Jul 1;119(7):1441.
6. Catassi C, Fasano A. Coeliac disease. The debate on coeliac disease screening--are we there yet? *Nat Rev Gastroenterol Hepatol*. 2014;11:457-8.
7. Stahl MG, Geno Rasmussen C, Dong F, Waugh K, Norris JM, Baxter J, Yu L, Steck AK, Frohnert BI, Liu E, Rewers MJ; ASK Study Group. Mass screening for celiac disease: the autoimmunity screening for kids study. *Am J Gastroenterol*. 2021 Jan 1;116(1):180-187.
8. Rubio-Tapia A, Murray JA, Kelly, C. Celiac Disease: Should there be screening for all? *AGA Perspect*. Apr/May 2012; 8(2):1-8.
9. US Preventive Services Task Force, Grossman DC, Curry SJ. Screening for Celiac Disease: US Preventive Services Task Force Recommendation Statement. *JAMA*. 2018;319:165-172.