

Home blood testing for celiac disease

Recommendations for management

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Case description

A 35-year-old mother was diagnosed with celiac disease several years ago. She is on a gluten-free diet. Her brother informed her of the recently marketed, Health Canada-approved, over-the-counter, self-administered home blood test for celiac disease. She purchased the kit from the local pharmacy and tested her 12-year-old son. The test result was positive. The boy has been in generally good health. She wants to know whether or not the child should start a gluten-free diet.

Sources of information

Home blood testing for celiac disease is a recent phenomenon and no published guidelines are available on this issue. As this form of testing mimics serologic laboratory testing, guidelines for such diagnostic testing were sought. Articles published in English from January 1985 to April 2008 were identified using the subject headings *diagnosis of celiac disease* and *management or treatment of celiac disease* in PubMed and the Cochrane Database of Systematic Reviews. Case reports, letters to the editors, editorials, and nonsystematic reviews were excluded.

In PubMed 14 articles were found. Four articles were duplicated in the 2 categories. Two articles were identified in the Cochrane database. Guidelines for serologic testing and confirmation of diagnosis of celiac disease developed by the American Gastroenterological Association¹ and the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition² were selected, as they represented the most recent, comprehensive, and systematic reviews pertaining to adult and pediatric populations, respectively (level 1 evidence). The recommendations presented in this article are based on the guidelines developed by these 2 professional gastroenterological organizations.

Main message

Celiac disease (gluten-sensitive enteropathy) is a common disorder affecting about 1% of the population.³⁻⁵ It is a chronic gastrointestinal disorder in which ingestion of gluten—a protein present in wheat, rye, and barley—leads to damage of the small intestinal mucosa by an autoimmune mechanism in genetically susceptible individuals. This can lead to a variety of symptoms and

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Cet article a fait l'objet d'une révision par des pairs.

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Abstract

OBJECTIVE To provide recommendations for the management of patients who inquire about the Health Canada-approved, self-administered home blood tests for celiac disease or who present with positive test results after using the self-testing kit.

SOURCES OF INFORMATION PubMed and the Cochrane Database of Systematic Reviews were searched from January 1985 to April 2008, using the subject headings *diagnosis of celiac disease* and *management or treatment of celiac disease*. Guidelines for serologic testing and confirmation of diagnosis of celiac disease by the American Gastroenterological Association and the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition are used in this review. Level 1 evidence was used.

MAIN MESSAGE Although blood tests are helpful for screening purposes, the confirmatory test for celiac disease is a small intestinal biopsy.

CONCLUSION Patients whose blood tests for celiac disease provide positive results should have endoscopic small intestinal biopsies to confirm the diagnosis before starting a gluten-free diet.

Résumé

OBJECTIF Faire des recommandations concernant le traitement des patients qui s'informent sur l'autotest sanguin approuvé par Santé Canada pour la maladie cœliaque ou qui se présentent avec un résultat positif à ce test.

SOURCES DE L'INFORMATION Une recherche intensive a été faite dans PubMed et dans Cochrane Database of Systematic Reviews, entre janvier 1985 et avril 2008, à l'aide des rubriques *diagnosis of celiac disease* et *management et treatment of celiac disease*. Dans cette revue, on a utilisé les directives de l'American Gastroenterological Association et de la North American Society for Pediatric Gastroenterology, Hepatology and Nutrition pour le dépistage sérologique et la confirmation du diagnostic de la maladie cœliaque. Les preuves de niveau 1 ont été retenues.

PRINCIPAL MESSAGE Quoique le test sanguin soit utile pour le dépistage de la maladie cœliaque, l'examen qui confirme le diagnostic est une petite biopsie du grêle.

CONCLUSION Les patients qui obtiennent un résultat positif au test sanguin pour la maladie cœliaque devraient subir une biopsie endoscopique du grêle pour confirmer le diagnostic avant de commencer un régime sans gluten.

nutritional deficiencies, including anemia and osteoporosis. Patients with celiac disease are at risk of developing serious complications, such as intestinal lymphoma.¹

Serologic testing

Highly sensitive and specific serologic tests, including tissue transglutaminase (tTG) antibody and endomysial antibody (EMA) tests, are available to screen for celiac disease.^{1,2} The anti-gliadin antibody (AGA) test is not recommended for screening because of its poor sensitivity and specificity. The tTG antibody and EMA tests detect antibodies of the immunoglobulin A (IgA) class. Immunoglobulin A deficiency is much more common in patients with celiac disease than in the general population.^{2,6} As such, serologic tests for celiac disease must include measurement of serum IgA in order to avoid false-negative test results.

Serologic tests are helpful in screening at-risk populations for celiac disease, including first- and second-degree relatives of patients with celiac disease, those with type 1 diabetes mellitus and other autoimmune endocrinopathies, and those with atypical symptoms. At the time of testing, the individual must be consuming a normal (gluten-containing) diet. Those already on a gluten-free or gluten-reduced diet might have invalid negative blood test results, thus confusing and delaying the diagnosis. A gluten-free diet rapidly heals the intestinal mucosa and most patients will have negative serologic test results approximately 6 months after beginning a strict gluten-free diet. To confirm diagnosis of celiac disease in these situations, an oral gluten challenge might be necessary to induce histologic changes before a small intestinal biopsy is attempted. How long gluten must be ingested before biopsy varies among individuals; some will relapse within a few weeks while for others it might take several years. Similarly, it can take several months to years for serologic tests to have positive results again, depending on the amount and frequency of gluten intake.¹ This can lead to prolonged uncertainty regarding the diagnosis.

Using blood tests to screen for celiac disease is also less reliable in children younger than 3 years of age.² Furthermore, a negative test result at a given time does not guarantee that an individual will not develop celiac disease in the future. The only confirmatory and definitive diagnostic test for celiac disease is a small intestinal biopsy.

Treatment

Treatment of celiac disease is a strict, lifelong adherence to a gluten-free diet.⁷⁻⁹ There is evidence that untreated celiac disease is associated with a substantial increase in morbidity and mortality.¹ Removal of gluten from the diet leads to improvement in symptoms and resolution of the intestinal damage. A gluten-free diet, however, is difficult and restrictive. Cross contamination of foods with offending grains is common, adding further challenges to everyday living with this diet.¹⁰⁻¹² Patients require counseling by registered dietitians with expertise

in this complex diet. A gluten-free diet is also more costly, and, as gluten sensitivity is permanent, the diet has to be followed for life.¹³ For these reasons, a gluten-free diet should be prescribed only when the diagnosis of celiac disease is confirmed using a small intestinal biopsy. (Dermatitis herpetiformis, the skin form of celiac disease, also requires a lifelong gluten-free diet.)

Self-testing

An over-the-counter home self-testing kit for celiac disease has recently been marketed in Canada. Using a tiny blood sample obtained by a pinprick of the fingertip, the home blood test identifies the tTG antibodies present in the blood of those with celiac disease.¹⁴ Endogenous tTG in the red blood cells is released by hemolysis and forms complexes with tTG-specific IgA-class antibodies. The complexes can be detected by binding tTG to a solid surface coated with tTG-capturing proteins. The bound antigen-antibody complexes can be seen by a colour reaction with the help of labeled antihuman IgA solution. The test takes about 10 minutes to read. These kits are an easy and attractive way to screen for celiac disease. The kit has been approved by Health Canada. The US Food and Drug Administration has not yet approved the kit and further information about and testing of the product is being sought.

The EMA and tTG antibody tests have a sensitivity of about 90% to 95%.¹ In a large European study, professional nurses screened school-aged children using rapid antibody testing of finger-prick blood. The sensitivity of rapid testing decreased to 78% when dealing with the general population.¹⁵ The authors of the study concluded that extra training is needed to improve sensitivity of the test. There is little data on how well this testing will perform when carried out by the general public.

Management of positive test results

If a patient uses the home self-testing kit for celiac disease and has a positive test result, the following steps are recommended:

1. A serologic laboratory test, immunoglobulin A (IgA) tissue transglutaminase antibody or IgA endomysial antibody, is required. Total serum IgA should also be measured.
2. If the serologic test result is positive, the patient should be referred for an endoscopic small intestinal biopsy to confirm the diagnosis as soon as possible.
3. It is strongly recommended that the patient continue to consume a normal (gluten-containing) diet and *not* start a gluten-free or gluten-reduced diet before the biopsy is performed. A trial of a gluten-free diet before biopsy has the potential to promote intestinal mucosal healing, leading to difficulty in pathologic interpretation of the biopsy and additional delay in confirming the diagnosis.
4. Once celiac disease is confirmed using a biopsy, the patient should see a registered dietitian with expertise on gluten-free diets. The patient should be encouraged to join a support group like the Canadian Celiac Association (www.celiac.ca).

EDITOR'S KEY POINTS

- Celiac disease (gluten-sensitive enteropathy) is a common disorder, affecting about 1% of the population. It is a chronic gastrointestinal disorder, in which ingestion of gluten leads to damage of the small intestinal mucosa by an autoimmune mechanism in genetically susceptible individuals.
- Home blood tests for celiac disease are a cause for concern, as individuals who have positive test results might begin gluten-free diets before being further evaluated by their physicians.
- Patients who use the self-testing kit and present with positive test results need to be sent for serologic laboratory testing.
- Endoscopic small intestinal biopsies confirm the diagnosis of celiac disease; however, patients must be consuming normal diets, as a gluten-free diet before biopsy leads to difficulty in pathologic interpretation and delay in confirmation of diagnosis.

POINTS DE REPÈRE DU RÉDACTEUR

- La maladie cœliaque (entéropathie par intolérance au gluten) est une maladie fréquente qui touche environ 1% de la population. C'est une affection chronique du tube digestif qui fait en sorte que l'ingestion de gluten entraîne chez des sujets génétiquement prédisposés une atteinte de la muqueuse intestinale par un mécanisme d'auto-immunisation.
- L'autotest pour la maladie cœliaque est une source de préoccupations, parce que les sujets qui obtiennent un résultat positif risquent de commencer un régime sans gluten avant l'évaluation du médecin.
- Un examen sérologique doit être prescrit à tout patient qui obtient un résultat positif à l'autotest.
- C'est une biopsie endoscopique de l'intestin grêle qui confirme le diagnostic de maladie cœliaque: le patient ne doit toutefois pas modifier son régime avant la biopsie, parce qu'un régime sans gluten complique l'interprétation de l'examen sur le plan pathologique et retarde la confirmation du diagnostic.

Although serologic tests for celiac disease have been available for more than a decade, home blood testing for celiac disease is a new phenomenon. There is concern that individuals (and families) using this home test might self-diagnose celiac disease and treat themselves with a gluten-free diet based on the test alone, without intestinal biopsy. Furthermore, evaluations by physicians to identify any problems associated with celiac disease, such as anemia and osteoporosis, will not be carried out. Nutrition counseling by a dietitian might also be bypassed.

Although blood tests for the screening of celiac disease are fairly accurate, some individuals will have false-negative or false-positive test results. The blood tests for celiac disease are for screening purposes only. The diagnosis must be confirmed using a small intestinal biopsy before introduction of a lifelong dietary modification.

A false-negative blood test result can delay the diagnosis of celiac disease. Untreated, these individuals are at risk of developing potentially serious complications, including osteoporosis, infertility, miscarriages, lymphoma, and possibly other autoimmune disorders.¹

Case resolution

The patient's son should be sent for laboratory serologic testing. If the test result is positive, he will need a small intestinal biopsy to confirm the diagnosis of celiac disease before starting a gluten-free diet.

Conclusion

An over-the-counter, self-administered blood test for celiac disease is now available in Canada. This test is for screening purposes only and should not replace a medical diagnosis. A positive test result for celiac disease should be followed with serologic laboratory testing. A gluten-free diet should not be started until the diagnosis is confirmed with a small intestinal biopsy. 🌿

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Contributors

All authors contributed to the concept of the article. **Dr Rashid** performed the literature search and the review of selected articles. All authors participated in the development and review of the manuscript.

Competing interests

All authors are members of the Professional Advisory Board of the Canadian Celiac Association.

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