Celiac Disease Updated: Diagnostic Challenge and Gluten Sensitivity

October 23, 2012
By Heidi Anne Duerr, MPH [1]

Here: an update on the challenges of detection, diagnosis, and management of celiac disease and non–celiac disease and the importance of gluten sensitivity.

Only one-fourth of patients with celiac disease present with the classic symptomology and about half of patients present monosymptomatic, Joseph A. Murray, MD, told colleagues at the American College of Gastroenterology 77th Annual Scientific Meeting in Las Vegas.

In his presentation, Dr Murray shared updates on the challenges of detection, diagnosis, and management of celiac disease and non–celiac disease and discussed the importance of gluten sensitivity. Dr Murray is Professor of Medicine and Consultant in the division of gastroenterology and hepatology and the department of immunology at the Mayo Clinic, Rochester, Minnesota.

Concerns over celiac disease and gluten sensitivity may be traced back centuries, Dr Murray noted. The condition was first described about 2000 years ago as a “disease” of diet and belly pain; the word celiac came from the Greek meaning “belly ache.” Now celiac disease is well-defined as an inflammation of the small intestines with an environmental and genetic pathogenesis; active disease usually has a trigger, such as neonatal infection or bacterial infection.

The classic symptoms of celiac disease seen in one-fourth of patients include malabsorption, diarrhea, weight loss, abdominal distention, steatorrhea, and multiple deficiency states, Dr Murray said. The single symptom seen in about half of patients may be simple to chronic diarrhea, iron deficiency anemia, or another non-GI symptom. The remaining one-fourth of patients present with such non-GI symptoms as infertility, chronic fatigue, and premature bone disease.

A common diagnostic clue for celiac disease is anemia, Dr Murray added, noting that 5% to 8% of adults with the condition present with unexplained anemia that is resistant to oral iron administration. The most common complaint among presenting patients is acute abdominal pain. Age, sex, and race play a role in celiac disease susceptibility, according to the World Health Organization; adults, women, and Caucasians are at greatest risk. Dr Murray added that patients with other autoimmune disorders (eg, diabetes mellitus, thyroid disease) and those with a family history also are at increased risk for celiac disease.

Patients often live with celiac disease for a long time before receiving the diagnosis, Dr Murray noted. On average, patients have symptoms for 10 to 15 years before a diagnosis is made. One study reported that only 17% of the estimated 1.8 million patients with celiac disease are being treated for the illness.

As such, it is the physician’s role to help detect celiac disease and then make the diagnosis, Dr Murray said.

In detection, he explained, clinicians are trying to ensure that they do not miss any cases (sensitivity) and, in the diagnosis, they want high specificity so that they do not make an erroneous diagnosis. Diagnosis usually involves serological tests, but the tests have varying sensitivity and specificity and therefore varying degrees of usefulness.

For instance, tissue transglutaminase antibody testing has both high sensitivity and high specificity, but endomysial antibody testing has medium sensitivity and high specificity. Dr Murray reported that gliadin TgA testing, which has variable sensitivity and specificity, should be avoided, but its new counterpart, deamidated gliadin, has medium sensitivity and specificity and may provide some adjunctive benefit, particularly in IgD-deficient patients.

Guidelines for detection and diagnosis of Celiac disease may be found on the European Society for Paediatric Gastroenterology Web site (http://www.celiachia.it/public/bo/upload/eventi_progetti%5Cdoc/S-Husby.pdf). The current diagnostic criteria are expected to be updated within the next 6 months, Dr Murray noted.

New pediatric guidelines for diagnosis of celiac disease are controversial, Dr Murray said, because
they aim to avoid biopsy. Even though tTG-IgG is a good indicator of disease, he noted that he does not think they are ready for prime time. He commented that a single test is a big thing to rest on for a lifelong diagnosis. As such, he said biopsy is important to help confirm the diagnosis of celiac disease.

Dr Murray noted that management of celiac disease involves the following important steps:

- Properly explain the disease to the patient.
- Refer the patient to a dietician.
- Include calcium and vitamin D supplementation.
- Check the patient’s bone density.
- Provide support resources.

The last step is important, Dr Murray explained, because anxiety and depression are common among patients with celiac disease—they struggle with their diagnosis and the resulting limitations and lifestyle changes.

Diet change is key to managing celiac disease, Dr Murray added. When patients adhere to these limitations, symptoms may improve in 1 to 3 months and some patients note improvement in a matter of days. However, Dr Murray suggests follow-up every 1 to 2 years with biopsy. A lack of healing as noted in biopsy can mean increased morbidity and even mortality.

With that in mind, Dr Murray also warned of patients not adhering to their regimens. Because histology healing takes longer than symptom healing, patients need to continue their regimens. Yet, Dr Murray noted that about 10% of patients may be nonresponsive to a gluten-free diet, sometimes because of inadvertent contamination of their diet. Thorough evaluation of patients who do not seem to respond adequately should be completed, he added, advising expert management for those with refractory celiac disease.

Dr Murray suggested that patients with non–celiac disease gluten sensitivity also are of interest. They see functional improvement of their symptoms when gluten is removed from their diets, but they do not test positive for celiac disease, he explained. Although these patients originally were thought to possibly have mild or potential celiac disease, new evidence suggests that non-celiac gluten sensitivity does not share immunological or genetic features of celiac disease. In addition, the etiology and management of non-celiac gluten sensitivity are uncertain.

What is certain, Dr Murray concluded, is that gluten can cause inflammation and can do so in various ways. When celiac disease is suspected, he added, clinicians should follow guidelines to ascertain whether the patient has celiac disease or non-celiac gluten sensitivity.

Source: Celiac disease and gluten sensitivity: New tests and approaches. Presented at the American College of Gastroenterology 77th Annual Scientific Meeting.

Source URL:

Links: