

# Update on Diagnosing, Managing Celiac Disease

*Short stature and dental enamel defects are added to the existing list of extraintestinal symptoms.*

BY DAMIAN McNAMARA  
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MIAMI — Ataxia, peripheral neuropathy, and epilepsy are among the extraintestinal manifestations of celiac disease when it presents in a child over age 15 months. The bowel symptoms of diarrhea, nausea, and abdominal pain are more common in younger children.

Other extraintestinal or atypical symptoms include dermatitis herpetiformis and other skin disorders, osteopenia and osteoporosis, iron-deficient anemia resistant to oral iron treatment, liver and biliary tract disease, and delayed puberty. Also, short stature can be a presenting sign. "In fact, 10% of children who walk into endocrinologist's office for short stature have celiac disease," Dr. Stefano Guandalini said during a pediatric update sponsored by Miami Children's Hospital.

A meeting attendee asked Dr. Guandalini if celiac serology should be routine in the work-up of patients with failure to thrive. "Yes, that is a big presentation for celiac disease, as long as the child has gluten in their diet," he said. "Residents sometimes will suggest this in a 3-month-old who has never had solid food."

There is even growing recognition that dental enamel defects in permanent teeth are a sign of the condition, Dr. Guandalini said.

"All the specialties are touched by this condition, even general pediatrics," said Dr. Guandalini, pediatric gastroenterologist and professor of pediatrics at the University of Chicago.

Celiac disease also can present with other autoimmune conditions, Dr. Guandalini said. For example, an esti-

mated 4%-10% of patients with type 1 diabetes mellitus also have celiac disease, as do 4%-8% of those with thyroiditis, and 2%-8% of those with arthritis.

"We tested one individual who had type 1 diabetes and found six relatives in the extended family with celiac disease," Dr. Guandalini said. "Celiac disease is much more prevalent if you are related to a celiac disease patient."

A child will, on average, visit eight pediatricians before being diagnosed, according to the Celiac Disease Center at the University of Chicago Web site, [www.celiacdisease.net](http://www.celiacdisease.net).

A meeting attendee asked if screening asymptomatic relatives of people with celiac disease is worthwhile. "Yes," Dr. Guandalini said. "The prevalence in first-degree relatives is between 10% and 25%. [These are] very high numbers."

A negative genetic test rules out celiac disease for life because it features a 100% negative predictive value. However, the test only has a 5% positive predictive value, so "a positive test means nothing," Dr. Guandalini said.

This "fascinating autoimmune disease" is triggered in genetically susceptible individuals by ingestion of gluten, the protein in wheat, rye, and barley. "We think that many, if not all celiac disease individuals, might have a problem with how they regulate their intestinal permeability," Dr. Guandalini said. Increased intestinal permeability allows more gluten to enter, attach to receptors, and present as antigens (non-self) to the immune system. This process can lead to damage of intestines, primarily the small intestines, including duodenal mucosa. Multiple childhood infections also are implicated in the possible etiology, he added.

In the early 1980s, diagnosis primarily was based on GI symptoms such as diarrhea, vomiting, and weight loss,

but currently, only about 8% of patients are diagnosed with these symptoms alone, Dr. Guandalini said. Diagnosis has changed over time toward more asymptomatic adults being detected through screening (*Am. J. Med.* 2006;119:e9-14).

"Every time you screen for celiac disease, test for [tissue transglutaminase] antibodies and total serum IgA, and confirm with a biopsy," Dr. Guandalini said. Some suggest that direct visualization of intestinal damage, including villous atrophy or blunting, is diagnostic. However, he said, "You cannot distinguish by visual findings if someone has celiac disease or not. You need to take a biopsy to an experienced pathologist—changes can be subtle." Dr. Guandalini had no relevant disclosures.

**'You cannot distinguish [celiac disease] by visual findings ... take a biopsy to an experienced pathologist.'**

DR. GUANDALINI

Early introduction of gluten into the diet of children at risk might be protective. For example, the timing of gluten introduction made a difference in a prospective study of 1,560 children at risk (*JAMA* 2005;293:2410-2). Specifically, the researchers found a protective window between 4 and 6 months of age.

Another study demonstrated that breast-feeding is beneficial to prevent or delay celiac disease (*Arch. Dis. Child.* 2006;91:39-43). Only one study included in this meta-analysis report showed no protective effect, Dr. Guandalini said.

"So you can recommend breast-feeding, and introduce gluten at 4-6 months along with breast-feeding in children at risk to minimize risk of celiac disease," he commented.

The only treatment for celiac disease is a diet free of gluten. However, "research is ongoing to find ways to remove gluten from wheat, degrade the enzyme, or to keep the gates tight," Dr. Guandalini said. "So there is hope that someday they will be able to eat at least some gluten." ■



## Colorectal Cancer Screening Rises, but Disparities Persist

BY MARY ANN MOON  
Contributing Writer

The use of sigmoidoscopy or colonoscopy to screen for colorectal cancer has increased in recent years, according to Dr. D.A. Joseph and his associates at the Centers for Disease Control and Prevention.

"Although this increase is encouraging, disparities persist in colorectal cancer test use," they said.

The researchers used data from the Behavioral Risk Factor Surveillance System to compare rates of colorectal cancer testing in 2002, 2004, and 2006. The BRFSS conducts telephone surveys of adults in all 50 states except Hawaii, and sampled between 100,000 and 200,000 adults in the 3 years included in this report.

The proportion of respondents aged 50 years and older who reported that they had undergone "lower endoscopy" within the preceding decade rose from 45% in 2002 to 50% in 2004 and to 56% in 2006.

However, the proportion that reported they had undergone fecal occult blood testing within the preceding year declined during the same

period, from 22% in 2002, to 18% in 2004, and to 16% in 2006, the investigators said (*MMWR* 2008;57:253-8).

Taking these colorectal cancer screens together, there was a net gain in the proportion of people aged 50 and older who underwent some form of screening.

Although the percentages increased for all races, the proportion of whites who underwent screening remained higher than that of all the other races. In addition, the proportions of people who underwent screening also increased with increasing education level and increasing household income.

In an editorial note accompanying this report, the CDC said that several factors might have contributed to the increase in colorectal cancer screening, including Medicare coverage of screening colonoscopy and increased coverage by private insurers.

"Previous studies have documented a greater prevalence of colorectal cancer screening among men than women. Data in this report suggest that the gap in prevalence between men and women is closing," the CDC said. ■

## Colonoscopy Greatly Underused In High-Risk African Americans

BY MARY ANN MOON  
Contributing Writer

African Americans who had multiple first-degree relatives with colon cancer were much less likely to undergo the recommended screening colonoscopy than their white counterparts, according to the findings of a large cohort study.

"Physicians and other health care providers need to elicit family history information for all patients and ensure that African Americans with affected relatives appropriately receive colon cancer screening," concluded Dr. Harvey J. Murff of Vanderbilt University, Nashville, Tenn., and his associates.

The investigators assessed colonoscopy screening rates in the Southern Community Cohort Study, an ongoing prospective observational study of disparities in cancer incidence and mortality across racial and geographic groups treated at 48 community health centers throughout 12 southeastern states. Their assessment included 41,830 subjects aged 40-79 years.

A total of 538 African Americans and 255

whites reported having multiple affected first-degree relatives or at least one first-degree relative diagnosed before age 50, putting them at increased risk for the disease.

Only 34% of these African Americans aged 50 and older reported that they had undergone colonoscopy within the preceding 5 years, compared with 50% of whites. Similarly, 21% of high-risk African Americans in their 40s had undergone colonoscopy, compared with 34% of high-risk whites in that age group.

This racial disparity persisted after the data were adjusted to account for participants' education status, income level, and insurance status, Dr. Murff and his associates said (*Arch. Intern. Med.* 2008;168:625-31).

For subjects of both races, the most often cited reason for not undergoing colonoscopy was that their health care providers never recommended the procedure. African Americans were much more likely to cite this reason than were whites. "However, it is unlikely that this difference completely explains the [racial] variation in the rate of colonoscopy procedures," the investigators said. ■

**In high-risk subjects aged 50 and older, 34% of African Americans had undergone colonoscopy in the preceding 5 years, compared with 50% of the whites.**