Case 1: Going forward in lack of movement

A 12-year-old girl was referred to the paediatric gastroenterology service with a history of chronic constipation and fecal incontinence. The symptoms were present for seven years, with frequent passage of stools in the clothes. She also experienced intermittent nocturnal enuresis. There was no abdominal pain, bloating, nausea, vomiting, diarrhea, rectal bleeding, dysuria, chronic cough, fatigue or weight loss. Her history was otherwise unremarkable, with no problem passing meconium after birth and a normal stool pattern in infancy. Her growth and development were normal. The child was in foster care and a family history was not available.

The patient was tried on a variety of laxative therapies including lactulose, oral polyethylene glycol and repeated rectal enemas. The patient had a difficult and protracted course over several years, with frequent stool incontinence. Cognitive behavioural therapy was provided by a professional psychologist. Due to uncertain compliance with oral therapy, lavage with polyethylene glycol via a nasogastric tube had been tried several times for fecal disimpaction.

Investigations included normal thyroid function tests, serum calcium and urinalysis. An abdominal radiograph had revealed fecal loading in the distal colon.

On examination, she was obese with a weight between the 90th and 95th percentiles, and a height between the 10th and 25th percentiles. Her body mass index was 29.8 kg/m^2 (greater than the 97th percentile). There was no pallor, clubbing or thyromegaly. The abdomen was soft and nondistended, with no tenderness, organomegaly or masses. (A previous rectal examination conducted by the referring physician revealed stool in the rectum.) Examination of the nervous system and spine was normal.

A diagnostic laboratory test was performed, which revealed the diagnosis.

Correspondence (Case 1): Dr Mohsin Rashid, Department of Paediatrics, Dalhousie University, IWK Health Centre, 5850 University Avenue, Halifax, Nova Scotia B3K 6R8. Telephone 902-470-8746, fax 902-470-7249, e-mail mohsin.rashid@iwk.nshealth.ca

Correspondence (Case 2): Dr Allison Bahm, Department of Pediatrics, The Hospital for Sick Children, 555 University Avenue, Room 1447, Toronto, Ontario M5G 1X8. Telephone 416-371-3772, e-mail abahm2009@meds.uwo.ca

Case 1 accepted for publication March 11, 2010. Case 2 accepted March 12, 2010

CASE 1 DIAGNOSIS: CELIAC DISEASE

Serological screening for celiac disease was obtained. The tissue-transglutaminase antibody (TTG) was positive. Endoscopic small intestinal biopsies revealed patchy villous atrophy, crypt hyperplasia and a marked increase in intraepithelial lymphocytes, confirming celiac disease.

The patient was started on a gluten-free diet. Over the next several months, the stool difficulties improved and the laxatives were discontinued. The enuresis also resolved. A year later, the TTG antibody was negative. On regular follow-up for five years, the patient has remained completely asymptomatic on a strict gluten-free diet, and has not required any laxative therapy.

Constipation is defined as a delay or difficulty in defecation, present for two or more weeks, and is a common problem in children. Frequency of defecation in healthy infants and children varies considerably, and changes with age. The pattern is even more unpredictable in breastfed infants. However, most healthy children will have at least three stools weekly. The presence of symptoms (pain and/or soiling), rather than frequency of stooling, better defines constipation.

In most children (90% to 95%) who experience constipation, the problem is functional in nature (ie, without objective evidence of a pathological condition). This is often caused by painful stooling with resultant voluntary fecal withholding by a child who wants to avoid unpleasant defecation. Functional constipation may be triggered by coercive toilet training, toilet phobia, school bathroom avoidance, stressful events, intercurrent illness, changes in routine or diet, or a child's postponing defecation because he/she is too busy. Organic causes of constipation in children are less common and include anatomical problems (eg, anal stenosis), neuropathic causes (eg, Hirschsprung's disease or spinal cord lesions), metabolic and systemic disorders (eg, hypothyroidism, cystic fibrosis, hypercalcemia or celiac disease), and medications (eg, opioids or anticholinergics).

Because most cases of constipation are functional, investigations are not required. An algorithm for the management of constipation in children has been developed by the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (1). The treatment of constipation includes education of the family regarding appropriate stooling patterns, fecal disimpaction with laxatives, dietary advice on adequate fluid and fibre intake, and behaviour modification. Ongoing monitoring of response to therapy is important.

The vast majority of children with constipation will improve with therapy and the problem will resolve. Laboratory investigations are, therefore, not needed. A plain radiograph of the abdomen is helpful in assessing the degree of fecal impaction when a rectal examination is not possible. However, some children respond poorly to treatment or they relapse once therapy is withdrawn. For children who remain constipated despite adherence to the treatment program, laboratory investigations are indicated (1). These screening investigations should include thyroid function tests with thyroxine, thyroid-stimulating hormone, serum calcium and celiac serology with TTG. The currently available TTG is an immunoglobulin (Ig) A-based test, and total serum IgA must be measured to avoid a false-negative result. IgA deficiency is more common in patients with celiac disease (2). A sweat test should be obtained in infants younger than one year of age with chronic constipation. A barium enema for anatomical abnormalities and magnetic resonance imaging of the lumbosacral spine for intraspinal abnormalities may also be required. The barium enema performed to demonstrate the transition zone in Hirschsprung's disease must be unprepped without a colonic clean out. All of the abovementioned tests can be completed by the primary care physician before a referral is made to a paediatric gastroenterology service. More specialized investigations, including full-thickness rectal biopsy for myenteric abnormalities (including Hirschsprung's disease) and anorectal manometry for myopathy or neuropathy, are best left to the gastroenterologists.

Celiac disease is a very common disorder that affects 1% of the paediatric population (2). It is now becoming increasingly evident that some children (and adults) with celiac disease do not present with diarrhea but with quite the opposite, ie, constipation (2-5). The present patient was constipated and overweight, which is not what most people would associate with a patient with celiac disease. In a recent Canadian study (4), constipation was the presenting symptom in five of 199 children diagnosed with celiac disease.

It is important to emphasize that not all children with constipation require screening for celiac disease. Serological screening should be considered if one or more of the following is present:

- The patient has chronic, refractory constipation despite adequate therapy.
- The patient exhibits other clinical features suggestive of celiac disease (2-4). These include vomiting, weight loss, delayed puberty, short stature, fatigue, iron-deficiency anemia, recurrent oral aphthous ulcers and dental enamel defects.
- The patient belongs to a high-risk group for celiac disease. This includes children with Down syndrome, type 1 diabetes, thyroiditis and those with a first- or second-degree relative with celiac disease (2).

The exact cause of constipation in celiac disease remains unclear. Gastrointestinal motor abnormalities can occur in celiac disease and improve in most patients after starting a gluten-free diet (6). The dysmotility from intestinal inflammation is believed to be the primary cause of constipation seen in celiac disease. Abdominal distension is sometimes seen in these patients.

CLINICAL PEARLS

- Although in the vast majority of children with constipation the problem is functional in nature, organic causes should be considered in case of intractable symptoms or poor response to therapy.
- Celiac disease can sometimes present with constipation as the only symptom. Children with chronic constipation should be screened for celiac disease by serological testing. A timely diagnosis and treatment will help resolve the constipation and associated problems in these children.

REFERENCES

1. Constipation Guideline Committee of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. Evaluation and treatment of constipation in infants and children: Recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. J Pediatr Gastroenterol Nutr 2006;43:e1-13.
2. Hill ID, Dirks MH, Liptak GS, et al. Guidelines for the diagnosis and treatment of celiac disease in children: Recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. J Pediatr Gastroenterol Nutr 2005;40:1-19.
3. Fasano A. Clinical presentation of celiac disease in the pediatric population. Gastroenterology 2005;128:S68-73.
4. McGowan KE, Castiglione DA, Decker Butzner JD. The changing face of childhood celiac disease in North America: Impact of serological testing. Pediatrics 2009;124:1572-8.
5. Catassi C, Kryszak D, Louis-Jacques O, et al. Detection of celiac disease in primary care: A multicenter case-finding study in North America. Am J Gastroenterol. 2007;102:1454-60.
6. Cucchiara S, Bassotti G, Castellucci G, et al. Upper gastrointestinal motor abnormalities in children with active celiac disease. J Pediatr Gastroenterolo Nutr 1995;21:435-42.

Mohsin Rashid MBBS MEd FRCPC,
Division of Gastroenterology and Nutrition,
Department of Paediatrics,
Dalhousie University, IWK Health Centre,
Halifax, Nova Scotia