



Two cases of a super mimic

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Case 1

History

Ching, a 38 year old school teacher, presented with a 14 day history of tiredness, weakness, anorexia and mouth ulcers. She said that for the past 12 months her health had been poor and that she had struggled to inject much energy into her work. In recent months she had suffered from flatulence, abdominal bloating and loose bowel actions of up to three or four times a day. Sometimes she had periods of constipation. In addition she had lost about 6 kg of weight in the past 6 to 8 months. She had visited a hospital clinic where she was prescribed a herbal remedy for 'bowel dysfunction'.

On physical examination she presented as a pale, thin, tall woman with no subcutaneous fat. Her vital parameters were normal. The only abnormal finding was abdominal distension with hyperactive bowel sounds. Rectal examination was normal.

Questions

1. What is your probability diagnosis?
2. What are the differential diagnoses?
3. What further questions would you ask Ching?

Answers

1. Coeliac disease (also known as coeliac sprue, gluten enteropathy, non-tropical sprue) is the most likely diagnosis. Although it presents in childhood it can present at any age. Apart from tiredness the classic tetrad of coeliac dis-

ease is diarrhoea, weight loss, abdominal bloating and iron/folate deficiency. Ming does have the mentioned symptoms of weakness, weight loss, bloating and diarrhoea.

2. Differential diagnoses include irritable bowel syndrome, carbohydrate intolerance (includes lactose, fructose, raffinose and galactans) this may be due to acquired lactase deficiency, and uncommon malabsorption syndromes such as Whipple's disease.

3. Ask about specific symptoms such as diarrhoea including bulky, offensive stools, difficulty flushing the stools down the toilet and abdominal cramps. Enquire about family history of malabsorption and autoimmune disease such as Type 1 diabetes, IgA deficiency, thyroid disease and sub fertility.

Further history

Ching confirmed that she did have symptoms of malabsorption in addition to a disinterest in food and occasional vomiting. She said that there was a family history of coeliac disease in an uncle and one first degree cousin

Questions

4. How do you confirm the diagnosis of coeliac disease?
5. How do you treat coeliac disease?

Answers

4. The serological diagnostic tests are IgA anti-endomysial and IgA transglutaminase antibodies but the gold standard diagnostic test

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is small-bowel biopsy which reveals the characteristic villous atrophy. A full blood examination should also be performed to investigate any iron or folate deficiency.

A common error in people with this type of presentation is to simply consider it as bowel dysfunction or as an irritable bowel syndrome

5. There is no cure for coeliac disease but it can be controlled through diet which is needed for the person's lifetime. It excludes gluten which is a type of protein found in most grains especially in wheat, rye, barley and oats.

Case 2

Sam, aged 15 months, was brought by his parents because of increasing diarrhoea with 6 large offensive stools daily. Four weeks earlier he had been seen by another doctor who considered an infective cause but stool microscopy and culture showed no parasites and no growth of pathogenic organisms. Despite a trial of tinidazole he continued to be unwell, anorexic, miserable and irritable with abdominal bloating and failure to put on weight. He was eventually diagnosed as coeliac disease.

Feedback

Children usually develop coeliac disease between the ages of 9 and 18 months following the introduction of solids into the diet. They usually present as diarrhoea or failure to thrive. Coeliac disease may affect people of all ages and is just as likely to be recognised in those over 60 years as it is in infancy and the first two decades of life. It is a true mimic or masquerade and many remain undiagnosed after several visits to medical services.

Conflict of interest

The authors declare no conflict of interest.

.RELATED GUIDELINES .

ACG CLINICAL GUIDELINES: DIAGNOSIS AND MANAGEMENT OF CELIAC DISEASE

This guideline presents recommendations for the diagnosis and management of patients with celiac disease.

Celiac disease is an immune-based reaction to dietary gluten (storage protein for wheat, barley, and rye) that primarily affects the small intestine in those with a genetic predisposition and resolves with exclusion of gluten from the diet. There has been a substantial increase in the prevalence of celiac disease over the last 50 years and an increase in the rate of diagnosis in the last 10 years. Celiac disease can present with many symptoms, including typical gastrointestinal symptoms (e.g., diarrhea, steatorrhea, weight loss, bloating, flatulence, abdominal pain) and also non-gastrointestinal abnormalities (e.g., abnormal liver function tests, iron deficiency anemia, bone disease, skin disorders, and many other protean manifestations). Indeed, many individuals with celiac disease may have no symptoms at all. The prevalence of celiac disease is increasing worldwide and many patients with celiac disease remain undiagnosed, highlighting the need for improved strategies in the future for the optimal detection of patients.

(Source: Rubio-Tapia A, Hill ID, Kelly CP, Calderwood AH, Murray JA; American College of Gastroenterology. ACG clinical guidelines: diagnosis and management of celiac disease. *Am J Gastroenterol* 2013;108(5):656-676)