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Atypical celiac disease with IgA deficiency presenting as Plummer–Vinson syndrome: a case report

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Abstract A 40-year-old man presented with insidious onset dysphagia for both solids and liquids for 4 years with recurrent oral ulcerations. On examination he was anemic, and barium swallow demonstrated a web in the postcricoid region. As part of the workup for unexplained iron deficiency anemia, a duodenal biopsy was taken that revealed moderate flattening of villi with increased intraepithelial lymphocytes consistent with the diagnosis of celiac disease. However, the serological tests for celiac disease (IgA antendomysial antibody, IgA antitissue transglutaminase antibody, and IgA antigliadin antibody) were all negative. Serum level of IgA was markedly low. A diagnosis of atypical celiac disease with severe selective IgA deficiency was made. After the institution of a gluten-free diet (GFD), his general condition as well as anemia improved. Histological recovery was documented on repeat duodenal biopsy 6 months after GFD.

Key words Plummer–Vinson syndrome · Esophageal web · Dysphagia · Atypical celiac disease

Introduction

The classical presentation of celiac disease is with chronic diarrhea and malabsorption syndrome. During the past two decades, atypical celiac disease that presents without diarrhea is increasingly being recognized [1]. Atypical celiac disease may manifest as refractory anemia, short stature, metabolic bone disease, or even obesity. The introduction of serological assays in the last two decades has partly been responsible for the increase in the diagnosis of atypical cases. One of the limitations of antibody testing is the occurrence of false-negative tests in the setting of selective IgA deficiency. We report a case of atypical celiac disease associated with selective IgA deficiency. The unusual feature in this case was initial presentation with dysphagia.

Case study

A 40-year-old man presented with insidious onset dysphagia for solids and liquids for 4 years with recurrent oral ulcerations. The dysphagia was episodic and had worsened over the past 2–3 months. At the time of presentation, there was no history of diarrhea, anorexia, caustic ingestion, or weight loss. However the patient could recall an episode of loose stools at the age of 12 years that lasted for 12 months and subsided spontaneously. There was no similar family history, and the patient was not a known diabetic. For this symptom, the patient had undergone repeated bougie dilatation of the esophagus elsewhere.

Physical examination was unremarkable except for moderate pallor. His weight and body mass index (BMI) were 55 kg and 19.3 kg/m², respectively. Investigations revealed a hemoglobin of 6.2 gm% with a hypochromic microcytic picture: mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), and MCH concentration (MCHC) were 56.3 fL, 15.00 pg, and 26.7 g/dl respectively. At the time of diagnosis, serum bilirubin was 0.9 mg%, total serum proteins were 7.7 gm%, serum albumin was 3.9 gm%, aspartate aminotransferase (AST) and alanine aminotransferase (ALT) were 29 and 32 IU/l, respectively, and serum alkaline phosphatase was 124 U/l. His serum iron and serum ferritin were 36.00 µg% (normal, 59–158 µg%) and 9.00 ng/ml (normal, 22–322 ng/ml) respectively. Total iron-binding capacity (TIBC) was 404.00 µg% (normal, 228–428 µg%). Thus, low serum iron and ferritin and increased TIBC confirmed the presence of iron deficiency anemia. Barium swallow demonstrated the presence of a web in the postcricoid region (Fig. 1). Upper gastrointestinal endoscopy showed narrowing at 20 cm through which the scope was negotiated with difficulty. Endoscopic examination of the rest of the upper gastrointestinal tract was unremarkable.
As part of the evaluation for unexplained iron deficiency anemia, a deep duodenal biopsy was taken, which showed severe flattening of villi with increased intraepithelial lymphocytes consistent with the diagnosis of celiac disease (Fig. 2). Subsequently, a urine  \( \delta \)-xylose excretion test done with a 5-g loading dose showed a urine excretion of 0.5 g (normal, >1 g/5 g/day), confirming malabsorption. Serological tests for celiac disease (IgA antiendomysial antibody, IgA antitissue transglutaminase antibody, and IgA antigliadin antibody) were all negative. Serum IgA, IgG, and IgM levels done subsequently were 6 mg/dl (normal, 100–490 mg/dl), 1877 mg% (normal, 800–1800 mg%), and 195 mg/dl (normal, 60–280 ng/dl), respectively.

A diagnosis of atypical celiac disease with selective IgA deficiency was considered, and the patient was started on a strict gluten-free diet (GFD). Over the next 6 months, the patient had complete disappearance of dysphagia and no recurrence of oral ulcerations. His weight increased by 6 kg, and his hemoglobin increased by 5.5 gm% over the baseline values. His follow-up hemoglobin and MCV at 6 months were 11.7 gm% and 92 fl respectively. Urine  \( \delta \)-xylose test done with the same loading dose was 1.0 g in 5 h. Serum albumin was 4.1 gm% at follow-up. Follow-up IgA levels were still low (20 mg/dl; reference, 100–490). Repeat duodenal biopsy 6 months after institution of GFD revealed nearly normal tissue (Fig. 3).

**Discussion**

This patient presented with dysphagia resulting from a postcricoid web with severe anemia. Workup for anemia in this patient revealed an iron deficiency pattern characterized by low serum iron and ferritin. Investigation of the iron deficiency anemia (IDA) led us to perform a duodenal biopsy, which was compatible with celiac disease. He had subclinical malabsorption, as was evident by abnormal  \( \delta \)-xylose test. Keeping a strong possibility of celiac disease in mind, the markers for celiac disease were done. Serum IgA antitissue transglutaminase antibody, antigliadin and endomysial antibodies were all negative. However, the serum IgA done subsequently was found to be markedly reduced. Inability to produce antibodies of the IgA1 and IgA2 subclasses occurs in approximately 1 in 600 individuals of European origin. IgA deficiency is much less common in Asians. In Japan, for example, the incidence is approximately 1 in 18,500 [2]. No such data are available from South Asia. Selective IgA deficiency has been found to be more common in patients with celiac disease. Data from the United States have shown that 2%–3% of patients with celiac disease may have associated IgA deficiency [3]. Kumar et al. [4] also highlighted that IgG antigliadin may be

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**Fig. 1.** Esophageal web seen on barium swallow in the region of cervical esophagus

**Fig. 2.** Pretreatment duodenal biopsy shows complete flattening of villi, with increase in intraepithelial lymphocytes and crypt hyperplasia, Marsh III stage. Hematoxylin and eosin, ×100