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Case series

Unusual Presentations of Celiac Disease in the Pediatric Population: A Case Series

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Introduction

Celiac disease (CD) is an immune-mediated small bowel enteropathy precipitated in genetically susceptible individuals by the ingestion of gluten. Gluten is a term used to encompass the alcohol-soluble storage proteins called prolamines that are present in wheat (gliadin), rye (secalin), and barley (hordein). CD is the most common genetically based food intolerance in the world [1].

Patients with CD can be grouped according to presentation: (1) Classical cases presenting early in life with signs of intestinal malabsorption; (2) Non-classical cases with predominantly extra-intestinal manifestations; (3) Silent cases with the presence of histopathologic features of celiac disease in the absence of symptoms; and (4) Latent cases with patients who manifest serologic evidence of celiac disease with no histologic signs.

CD is not just a condition limited to the gastrointestinal tract and awareness of its large clinical spectrum by physicians in all fields of medicine is essential for a timely diagnosis and initiation of treatment. Non-classical symptoms are becoming more common than the classical ones [2].

Non-classical CD is mostly diagnosed in older children and adolescents. Amongst gastrointestinal presentations abdominal distention, flatulence, irregular bowel habits and chronic constipation may be the atypical presentations. In particular constipation is often not responsive to standard therapy.

Non-classical CD often presents with extra-intestinal symptoms associated with the disease, such as puberty delay, unexplained chronic or iron deficient anemia non-responsive to iron supplementation, decreased bone mineralization (osteopenia/osteoporosis), dental enamel defects, irritability, chronic fatigue, neuropathy, arthritis/arthralgia, amenorrhea, unexplained increased liver enzymes [3].

We describe 3 extremely unusual presentations of CD with gastrointestinal and neurological presentations even atypical for the described presentations of non-classical CD to date.

Case 1

The patient is a 19-year-old female with medical history of asthma who presented to pediatric rheumatology for evaluation of a 6-month history of bilateral upper extremity numbness and tingling and years of neck pain. Prior evaluation did reveal disc herniations in her neck for which she was referred for physical therapy which partially helped. Most recent MRI of her neck was reported as normal. The numbness in her extremities was occurring daily and self-resolving after varying amounts of time. She also had reports of knee and hip pains. She denied abdominal pain, diarrhea, constipation, oral ulcerations, weight loss, vomiting, fevers. She does have a cousin with celiac disease. Screening labs revealed anemia with Hgb 8.4g/dL with MCV 67fL, iron 14ug/dL, ferritin 3ng/mL. Celiac serologies revealed TTG IgA >100U/mL with normal total immunoglobulin A. She underwent upper endoscopy with biopsies which confirmed the diagnosis of celiac disease with Marsh stage 3 criteria fulfilled in the duodenal and duodenal bulb biopsies. She was referred to our celiac center to meet with the dietician for initiation of a gluten free diet. Follow up 3 months after initiation of a gluten free diet showed resolution of her iron deficiency anemia while on iron supplementation. Her TTG IgA improved to 13U/mL. Her symptoms of upper extremity numbness and tingling and neck pain completely resolved. She was lost to follow up when she went away to college.

Case 2

The patient is an 18-year-old female who presented to pediatric gastroenterology (GI) with 1 month of dysphagia with solids. She denied abdominal pain, diarrhea, vomiting, fever, weight loss, rashes, joint pains. Given concern for eosinophilic esophagitis she was started on a proton pump inhibitor (PPI). Celiac serologies obtained and were within normal limits. Upper endoscopy (EGD) on PPI showed normal esophageal, gastric and duodenal biopsies. With continued dysphagia, her PPI was increased to twice daily for 1 month with no improvement. Esophageal manometry showed a normal lower esophageal sphincter pressure, but on nine of ten swallows, she had incomplete clearance of her fluid bolus. A 24-hour impedance probe was recommended but refused. Her dysphagia continued to worsen but she was lost to follow up. She presented 1 year later with dysphagia with soft foods and gurgling in her chest. Upper GI series obtained and was unremarkable. Rheumatologic evaluation revealed elevated CRP 8.7 mg/L, positive ANA and an elevated anti-centromere antibody 3.3 AI (normal 0-0.9 AI). Repeat esophageal manometry showed ineffective motility. Eight of ten swallows were not followed by peristalsis. Trial of Bethanechol was initiated to increase smooth muscle tone and promote esophageal motility. Two months later, her symptoms

improved, but were still present. Repeat EGD was performed which showed villous blunting and focally increased intraepithelial lymphocytes in the duodenum, concerning for celiac disease. Gastric and esophageal biopsies were normal. Celiac serologies remained negative (serum IgA 142, tissue transglutaminase IgA <2U/mL, tissue transglutaminase IgG <2U/mL, deamidated gliadin IgA 4 units, deamidated gliadin IgG 5 units, endomysial IgA negative) and celiac genetics showed her to be DQ2 positive/DQ8 negative. CD was considered. As a therapeutic trial, the patient was started on a strict gluten free diet. After 8 months of a gluten free diet and bethanechol, dysphagia has resolved and she rarely complains of "gurgles." Her repeat endoscopy revealed normalization of her duodenum on histopathology.

Case 3

The patient is an 11-year-old male who presented to the pediatric GI clinic with complaints of diarrhea, eye blinking and papilledema. Patient first reported developing eye blinking in the absence of headaches or changes in vision. Evaluated by an ophthalmologist and diagnosed with papilledema. Found to have elevated opening pressure on lumbar puncture. Tried Diamox without relief, discontinued after two weeks due to side effects of dizziness, fatigue, lightheaded sensation. He had developed frequent episodes of watery diarrhea around the same time as his eye blinking. Patient removed dairy from his diet with resolution of the diarrhea. He denied abdominal pain, joint pains, rashes. He had lost 8lbs around the onset of symptoms which occurred 6 months prior to presenting to the GI office. There is a family history of scleroderma in the mother and the maternal great grandmother has lupus. Screening labs were sent and revealed an elevated TTG IgA of 31U/mL with normal total IgA. His endomysial antibody was negative and TTG IgG was within normal limits. He was referred for upper endoscopy with biopsies which confirmed the diagnosis of celiac disease. He was referred to our celiac center to meet with the dietician for initiation of a gluten free diet.

Discussion

Celiac disease is an autoimmune, systemic disorder that occurs in genetically susceptible individuals exposed to gluten. The importance of early detection and treatment cannot be emphasized enough as untreated CD can lead to numerous complications including malnutrition secondary to malabsorption, vitamin/mineral deficiencies, osteopenia/osteoporosis, infertility, irritability, depression, fatigue, dental enamel defects, intestinal lymphoma, peripheral neuropathy.

The three non-classical presentations of CD outlined above could have gone unrecognized for a longer period of time were it not for their physician's astuteness in recognizing symptoms that may be attributed to non-classical CD. Upper extremity numbness/tingling and neck pain are not common symptoms that would typically clue you in to a diagnosis of CD. The patient in the second case had an unusual and tortuous course prior to diagnosis of CD. Dysphagia, in the absence of esophageal eosinophilia related to gluten ingestion, is an uncommon symptom associated with CD. The fact that her serologies were negative and biopsies were unremarkable on her first upper endoscopy made the diagnosis even more ambiguous at first. Her response to the gluten free diet in the presence of diagnostic biopsies with positive HLA DQ2 testing strongly suggests CD as the cause of her symptoms. Only rare case reports exist in the literature describing the potential association of papilledema with the presentation of CD proving the importance of publishing cases like our patient to promote increased awareness of this association within the medical community.

Non-classic celiac disease refers to a presentation of CD without classic gastrointestinal symptoms. It is now believed that non-classic CD is more common than the classic form [2]. Common non-classic presenting signs/symptoms include iron deficiency anemia, dermatitis herpetiformis, osteoporosis/osteopenia, dental enamel hypoplasia, infertility and/or recurrent fetal loss, vitamin deficiencies, abnormal liver function tests, fatigue, psychiatric symptoms, various neurologic conditions including peripheral neuropathy, ataxia, seizures, migraines, attention-deficit hyperactivity disorder and poor school performance [3]. Our 3 unusual case presentations of CD highlight the importance of keeping celiac disease in the differential diagnosis for patients with extra intestinal symptoms with or without gastrointestinal symptoms.

Some studies suggest older age at diagnosis for non-classical and subclinical types of CD. A retrospective review by Oliveira et al showed a higher mean age at diagnosis for these types and although the difference was not statistically significant it may suggest delayed recognition of the previously less common presenting symptoms [4]. Regardless of age it is important to recognize the potential diagnosis of CD in patients with non-classical symptoms and to work these patients up appropriately.

In conclusion there is a wide spectrum of clinical presentations of pediatric celiac disease. One must keep an open mind and consider CD in the differential diagnosis for patients presenting with both classical and non-classical symptoms suggestive of CD so as to not delay diagnosis and treatment for our patients.

References

1. Guandalin S, Assiri A. Celiac Disease A Review. JAMA Pediatrics 2014;168(3):272-278.
2. Ludvigsson JF, Leffler DA, et al. The Oslo definitions of coeliac disease and related terms. Gut 2013; 62(1):43-52.
3. Taylor AK, Lebwohl B, Snyder CL, et al. Celiac Disease. 2008 Jul 3 [Updated 2019 Jan 31]. In Adam P, Ardinger HH, Pagon RA, et al., editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.
4. Oliveira GN, Mohan R, Fagbemi A. Review of celiac disease presentation in a pediatric tertiary care centre. Arq Gastroenterol 2018; 55(1):86-93.

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