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This Man's Increased Psychiatric Issues Masked an Undiagnosed Disease

— The long delay to diagnosis and treatment caused frightening symptoms

by Kate Kneisel, Contributing Writer, MedPage Today
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A 25-year-old man presents to an emergency department (ED) in Illinois, with a dramatic behavioral disturbance. He has been brought in by his mother, who reports that he has just broken into a

neighbor's house. The patient, already under the care of a psychiatrist, has a medical history of autism spectrum disorder and schizophrenia.

The mother notes that over the past few months, the patient has become increasing combative, and frequently fights with his family. She tells the admitting clinician that the family has noticed the patient's stomach beginning to protrude and that his legs have become swollen.

He also has started complaining constantly of being hungry all the time, and in fact, out of concern about the patient's increasing girth, the family has installed locks on the cabinets and refrigerator to limit his eating.

The aggressive behavior appears to have started with the onset of gastrointestinal symptoms about 5 months earlier, his mother notes, when he reported having mild abdominal pain and severe and uncharacteristic behavioral problems. Notably, even with his frequent eating, he has lost about 14-18 kg (31-40 lbs) in the past year.

Over the past 1-2 months, the patient's symptoms have worsened, with vomiting and diarrhea that is so severe that he is frequently incontinent, the mother notes. As a result, over the past 2 months, the patient was evaluated by gastroenterologists, nephrologists, and his primary care provider, and has also visited another ED, all without a definitive diagnosis.

He has become significantly weaker, and when his extreme hunger caused him to break into a neighbor's house for food, his mother decides to bring him to this ED.

The patient's psychiatrist had diagnosed him, in addition to autism spectrum disorder, with episodic mood disorder and an unspecified psychosis. He is taking lamotrigine, lithium carbonate, risperidone, and topiramate.

Physical examination finds evidence of malnutrition with bitemporal wasting, abdominal distention, diffuse anasarca, and 4+ bilateral lower-extremity pitting edema.

Laboratory investigations reveal significant metabolic derangements:

- Potassium: 2.8 mmol/L
- Magnesium: 1.1 mg/dL
- Albumin: 1.8 g/dL
- Phosphorus: 1.6 mg/dL

Computed tomography of the abdomen and pelvis reveals small bowel enteritis. The patient says he has had no travel or exposure risk to suggest an infectious cause. A stool sample is collected for culture, ova, and parasite examination, white blood cell lactoferrin, and *Clostridium difficile* polymerase chain reaction analysis.

Evaluation for celiac disease shows elevated tissue transglutaminase immunoglobulin (Ig) A antibody above the laboratory threshold of 250 U/mL. A gastroenterology consultant performs an esophagogastroduodenoscopy, which reveals villous blunting of the duodenum, which is microscopically confirmed in a biopsy.

Esophagogastroduodenoscopy showing villous blunting of the duodenum.

Based on these findings, clinicians diagnose the patient with celiac disease, Marsh type 3b.

Which of the following factors during the first year of life may affect the risk of developing celiac disease? (Please select all that apply)

- [ ] Antibiotics use
- [ ] Gluten introduction in the diet
- [ ] Gastrointestinal infections
- [ ] Maternal breastfeeding

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Further nutritional evaluation revealed deficiencies of fat-soluble vitamins, micronutrients, and essential fatty acids:

- Vitamin A: 28.2 mcg/dL (normal range 32.5-78.0 mcg/dL)
- Folate: <1.6 ng/mL (7.0-31.4 ng/mL)
- 25-hydroxy Vitamin D: <4 ng/mL (~12.5 ng/mL)
- Vitamin E: 2.3 mg/L (5.5-17.0 mg/L)
- Copper: 0.63 mcg/mL (0.75–1.45 mcg/mL)
- Selenium: 147 ng/mL (150–241 ng/mL)
- Zinc: 0.23 mcg/mL (0.66–1.10 mcg/mL)
- Triene: Tetraene ratio 0.071 (0.010-0.038)

Clinicians put the patient on a gluten-free and lactose-free diet, and his symptoms improve. His severe malnutrition requires immediate repletion and in consideration of the time it takes for the villous architecture to recover, clinicians start him on total parenteral nutrition (TPN) via a Groshong catheter.

Following initiation of TPN, the patient's potassium, magnesium, and phosphorus levels are monitored closely, with appropriate repletion to prevent onset of re-feeding syndrome.

After initiation of the diet and 10 days of TPN, the patient's symptoms improve significantly, including near-total resolution of his anasarca. He is discharged.

Which of the following comorbidities has been significantly associated with celiac disease? (Please select all that apply)

- [ ] Alopecia areata
- [ ] Down syndrome
- [ ] Lupus
- [ ] Psoriasis

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Two days after his admission, however, the patient has a partial traumatic removal of his Groshong catheter, which requires another visit to the ED. After a total of 12 days of TPN, he is re-evaluated and deemed sufficiently recovered to have the catheter removed.

Discussion

The authors of this case report note that it highlights the fact that celiac disease may also present primarily as behavioral disturbance in adults, particularly if there is already underlying psychiatric disease or intellectual deficits.

In celiac disease in infants, which can vary significantly from that in adults, in addition to genetic factors, possible risk factors include gastrointestinal infections and antibiotics use during the first year of life, which may be related to differences in the composition of gut microbiota between children with and without celiac disease, the case authors explain. On the other hand, the possible contribution of certain infant feeding practices such as the timing of the first gluten introduction in the diet and the presumed protective role of maternal breastfeeding have been recently shown to be irrelevant in relation to the development of the disease.

The case report authors note that increasing awareness of the disease has led to improved detection of patients with extra-intestinal, atypical, or mild presentations, with serologic testing allowing for easy testing. This has reduced the number of adults presenting with celiac crisis, a syndrome of profuse diarrhea and severe metabolic/nutritional disturbances, which is now almost completely confined to children with the disease.

Although recent evidence suggests that not all patients require a biopsy for diagnosis, this remains the criterion standard, showing villous atrophy, intraepithelial lymphocytosis, and crypt hyperplasia, three pathologic findings that have been standardized through the Marsh classification.

Guidelines from the American College of Gastroenterology cite evidence for type 1 diabetes, autoimmune thyroiditis, thyroid disorders (including non-autoimmune causes of hypothyroidism and hyperthyroidism), selective IgA deficiency, Down syndrome, psoriasis, inflammatory bowel disease, and dermatitis herpetiformis as significant co-morbidities. Meanwhile, Turner's syndrome, alopecia areata, attention-deficit hyperactivity disorder (ADHD), lupus, autoimmune hepatitis, and sarcoidosis were not found to be significantly associated with celiac disease.

The case authors explain that while electrolyte and metabolic derangements have been described in adults with celiac disease, previous case reports have involved shorter time periods and were often linked with an inciting event, such as infection or surgery. In this particular patient the signs and symptoms unfolded over a period of months to a year, with weight loss and diarrhea that progressed to peripheral edema and, ultimately, weakness.

Diagnosis was obfuscated by the patient's baseline function related to his autism spectrum disorder and episodic mood disorder, which not only masked gastrointestinal symptoms but interfered with his ability to adequately describe his symptoms. This delayed the diagnosis, despite his visits with several medical specialists.

The patient's extended period between the onset of symptoms and diagnosis resulted in a prolonged exposure to dietary gluten, leading to profound degradation via gluten-bound HLA-DQ2 and HLA-DQ8 molecules propagating a T-cell-mediated inflammatory response, which when prolonged, impairs the function of the brush border, producing metabolic derangements and nutritional deficiencies, the authors explain.

Conclusion

They conclude that although the data are conflicting and definite conclusions are difficult to draw, at a minimum physicians should be aware of possible associations between psychiatric symptoms/disorders and celiac disease, and that exacerbation of psychiatric symptoms, especially if accompanied by diarrhea, weight loss, or other indications of gastrointestinal disease, may be a reasonable indication to screen for celiac disease.

Kate Kneisel is a freelance medical journalist based in Belleville, Ontario.

Disclosures

The case authors reported having no conflicts of interest.

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