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CASE REPORT

Case report: celiac disease associated with gastric trichobezoar - Rapunzel syndrome

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Abstract

Objectives: Report a case of a patient with celiac disease and psychologic disturbance associated with gastric tricobezoar - Rapunzel syndrome. **Case report:** E.N.F., a 7-year-old girl, appeared on ambulatory with her mother referring a history of abdominal pain, episodes of diarrhea, weight loss associated with sadness. On physical examination, patient presented good general condition, globose abdomen, bowels sounds presence and distended and painful abdomen at superficial palpation. Gastrointestinal endoscopy revealed presence of gastric trichobezoar. The biopsy of esophagus, stomach and duodenum fragments showed celiac disease patterns. She was submitted to a laparotomy for bezoar removal. The patient had a good clinical evolution after the surgical procedure and followed the treatment with gluten-free diet and pshychological/psychiatrist follow-up. **Conclusions:** It is believed that celiac disease has relation with psychologic disorders, trichotillomania and trichophagia, which influenced on the development of Rapunzel syndrome.

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INTRODUCTION

Celiac disease (CD) is a systemic inherited autoimmune immune-mediated disorder triggered by the ingestion of gluten and wheat, rye, or barley prolamins that affects genetically susceptible children and adults.[1] CD is currently underdiagnosed on account of the broad clinical manifestations tied to the condition. Rapunzel syndrome is a severe condition affecting patients suffering from trichotillomania and trichophagia that causes severe abdominal pain, nausea, and weight loss. It is commonly seen in children and adolescents with mental disorders.[2,3]

Our search found only seven papers investigating the concurrent occurrence of CD and trichobezoar.[2-8] Our paper describes the case of a patient with celiac disease and a gastric trichobezoar suspected for Rapunzel syndrome.

CASE REPORT

A seven-year-old girl was taken to our service with abdominal pain, a distended abdomen, and low weight and short stature for her age. When asked about symptoms, her mother cited apathy, weakness, despondency, and diarrhea. No other complaints were mentioned. She had taken all vaccines in the immunization schedule. The girl had not taken antiparasitic agents for more than six months. Her mother reportedly had a history of digestive disorders.

She was pale during physical examination and looked distressed and sad. The girl had a distended abdomen and a waist circumference of 60 cm (Figure 1). She felt pain during superficial and deep palpation of the abdomen; her liver could be palpated and she had decreased gastric and bowel sounds; no additional significant alterations were seen in her physical examination. Biochemical tests were ordered and she was prescribed an anthelmintic drug.

Complete blood count revealed she had iron-deficiency anemia. Total abdominal ultrasound examination found a mildly enlarged liver, focal hepatic steatosis, and bowel loops filled with significant amounts of fluid and gas. Upper gastrointestinal (UGI) endoscopy found a trichobezoar and erosive antral gastritis with focal bleeding. Her serology tests were positive for tissue transglutaminase (tTG) IgA antibodies (>128 IU/mL) and tTG IgG antibodies (>1.5). Wheat-specific, omega 5-gliadin, and gluten IgE were negative. The levels of immunoglobulin A and E were 413.8 IU/ mL and 544.0 IU/mL, respectively. The findings of core biopsies of the esophagus, stomach, and duodenum were consistent with lymphocytic enteritis with subtotal villous atrophy (Marsh IIIC).

The patient was referred to gastrointestinal surgery and underwent a laparotomy to have the gastric trichobezoar removed (Figure 2). She progressed well after surgery. The child has been provided psychological/psychiatric support. She was prescribed a gluten-free diet, iron replacement therapy, and has since resumed growing and developing.



Figure 1. Child with a distended abdomen.



Figure 2. Gastric trichobezoar removed with surgery.

DISCUSSION

Differently from what was known in past decades, celiac disease (CD) is a frequent and globally disseminated autoimmune enteropathy. Research estimates the global prevalence of CD at 1% including children and adults, with proportions

varying between countries.[1] CD is not a rare condition in Brazil, with trends showing increases in prevalence and studies reporting an incidence of 2.11:1000 adults and 5.44:1000 children aged 14 or younger, although the scarcity of epidemiology studies and the absence of standardization in serology testing compromise the accuracy of research findings.[9,10] CD affects individuals of all ages, but is more frequently seen in females and children aged between six months and five years.[11]

Celiac disease (CD) is a systemic inherited autoimmune immune-mediated disorder triggered by the ingestion of gluten and wheat, rye, or barley prolamins that affects genetically susceptible children and adults. This T-cell-mediated chronic enteropathy affects the small bowel and is characterized by multiple gluten-dependent clinical manifestations associated with HLA-DQ2 and HLA-DQ8.[1,4,9-11]

Clinical manifestations vary depending on individual sensitivity, gluten intake levels, and time of onset. The disease has been described in three different forms: classical, atypical, and asymptomatic.[9,12] Classical symptoms stem from malabsorption and include diarrhea, weight loss, bloating, nutrient and vitamin deficiency, fatigue, edema, irritability, and depression, the last three being more common in children. Patients with atypical CD develop oligosymptomatic disease, with weaker or no digestive symptoms, short stature, iron-deficiency anemia, pernicious anemia, osteoporosis, arthralgia or arthritis, refractory constipation, delayed puberty, recurrent miscarriage, neurologic symptoms such as headaches and ataxia, and mental disorders including depression, autism, and schizophrenia. Individuals with asymptomatic CD do not suffer from clinical manifestations despite the existence of bowel damage, but may experience improvements in quality of life by eating gluten-free diets.[1,9-11] There is no correlation between clinical manifestation severity and the degree of bowel mucosal damage.[10]

The gold standard diagnostic method for CD is UGI endoscopy and small bowel biopsy, although serology tests have been used to detect gluten intolerance and are useful in monitoring patients with the condition.[11] The most commonly used serology markers are anti-gliadin antibodies, anti-endomysial antibodies, and anti-tissue transglutaminase antibodies. Laboratory tests are indicated to patients suspected for CD, including relatives of individuals with CD, and must be performed while the tested individual is on a diet containing gluten.[11,12] Early diagnosis and observance of a proper diet is required to avoid severe malignant complications such as neurologic disorders and cancer.[1]

Individuals with CD are prescribed a gluten-free diet and, depending on the degree of involvement of the villi causing disaccharide production deficiency, are also required to temporarily refrain from eating lactose and sucrose.[11,12] Nutrient deficiencies stemmed from the malabsorption of micro and macromolecules must be tackled with replacement measures and a multidisciplinary care team involving physicians, nutritionists, and psychologists.[9,11]

The patient described in this case presented with classical CD symptoms including diarrhea, weight loss, gluteal wasting, weakness, apathy, iron deficiency, and low weight and short stature for her age, probably due to impaired bowel mucosa permeability leading to malabsorption. She also experienced psychiatric symptoms characteristically seen in oligosymptomatic CD.

Psychiatric and neurological symptoms such as cerebellar ataxia, trichophagia, cognitive impairment, neurodegenerative disorders, epilepsy, and schizophrenia have been tied to celiac disease, although the pathophysiology of the condition remains unclear. These symptoms may be triggered by serotonergic system disorders caused by CD and the ensuing effects on the central nervous system.[4]

Trichotillomania (hair-pulling disorder) is a chronic obsessive-compulsive disorder generally associated with trichophagia (compulsive eating of hair) more commonly seen in girls in the first two decades of life.[2,3,5] The two have been associated with mental disorders in children and may lead to the formation of a gastric trichobezoar, a concretion built from the continuous deposition of hair on the stomach lining. In rare cases, trichobezoars may extend into the duodenum in a condition known as Rapunzel syndrome.[2,13] The most common symptoms arising from trichobezoars are abdominal pain, diarrhea, vomiting, edema, anemia, and bowel obstruction and perforation in more advanced cases. The treatment of choice is the removal of the concretion via laparotomy, in some cases also involving the resection of portions of the bowel.[2,3,13]

Trichobezoars are a complication of trichophagia, a condition associated with iron deficiency and mental disorders.[4,6,7]

The rare combination of celiac disease and Rapunzel syndrome has been described in only seven case reports published in the literature. Few studies have looked into the association of pica and celiac disease, but the cumulative effect of nutritional deficits and immune and inflammatory factors linked to CD may significantly affect clinical manifestations.[4] Two of the seven papers described cases of trichophagia independent from anemia in individuals with CD, a fact supported by the finding that in one of them iron replacement was ineffective and mental disorders persisted.[2-8] Our patient improved from the symptoms tied to malabsorption and mental disorder after she was prescribed a gluten-free diet. Therefore, trichotillomania and trichophagia may be associated with psychiatric symptoms triggered by celiac disease, which occur more commonly in oligosymptomatic CD.[2]

Patients with CD must be examined and interviewed thoroughly to enable the identification of supplementary investigation needs. Familial predisposition to mental illness, in addition to stress and socialization factors, must be considered in the detection of symptoms of depression or anxiety.[6]

CONCLUSION

The history of psychological distress presented by the patient indicates that celiac disease may cause mental disorders, trichotillomania, and trichophagia, which ultimately led to the development of Rapunzel syndrome. This is further corroborated by the fact that the patient improved significantly from clinical symptoms after she was prescribed a gluten-free diet and psychological support.

REFERENCES

1. Bai JC, Ciacci C, Corazza GR, Fried M, Olano C, Rostami-Nejad M, et al. Celiac disease. WGO [Internet]. 2016 Jul; [citado 2018 out 05]; 1-35. Disponível em: http://www.worldgastroenterology.org/guidelines/global-guidelines/celiac-disease/celiac-disease-english
2. Appak YÇ, Ertan D, Karakoyun M, Özyurt G, Özdemir T, Baran M. The cause of abdominal mass in a child with celiac disease: Rapunzel syndrome. A case report. Sao Paulo Med J [Internet]. 2018 Mai; [citado 2018 out 01]; 137(3):292-4. Disponível em: http://www.scielo.br/scielo.php?script=sci_arttext&pid=S1516-31802018005003101&lng=en
3. Larsson LT, Nivenius K, Wettrell G. Trichobezoar in a child with concomitante coeliac disease: a case report. Acta Paediatr [Internet]. 2007 Jan; [citado 2018 ago 10]; 93(2):278-80. Disponível em: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1651-2227.2004.tb00722.x
4. Irastorza I, Tutau C, Vitoria JC. A trichobezoar in a child with undiagnosed celiac disease: a case report. World J Gastroenterol [Internet]. 2014 Fev; [citado 2018 out 01]; 20(5):1357-60. Disponível em: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3921519/ DOI: https://doi.org/10.3748/wjg.v20.i5.1357
5. Al Lihabi AA. Trichotillomania in celiac disease. Case Rep Gastroenterol [Internet]. 2016 Set/Dez; [citado 2018 ago 06]; 10(3):613-6. Disponível em: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5121569/ DOI: https://doi.org/10.1159/000448877
6. Kalyoncu T, Çıldır DA, Özbaran B. Trichotillomania in celiac disease patient refractory to iron replacement. Int J Adolesc Med Health [Internet]. 2017 Ago; [citado 2018 ago 10]; 31(4):20170027. Disponível em: https://www.degruyter.com/view/j/ijamh.ahead-of-print/ijamh-2017-0027/ijamh-2017-0027.xml DOI: https://doi.org/10.1515/ijamh-2017-0027
7. Marcos AS, Bravo MM, Casanova AB, Belinchón PP, Corral LM. Gastric trichobezoar as an atypical form of presentation of celiac disease. An Pediatr (Barc) [Internet]. 2005 Jun; [citado 2018 ago 10]; 62(6):601-2. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/15927136
8. McCallum IJD, Zanten CV, Imran ZI, Craig W, Mahdi G, Thompson R. Trichobezoar in a child with undiagnosed coeliac disease. J Paediatr Child Health [Internet]. 2008 Set; [citado 2018 ago 01]; 44(9):524-5. Disponível em: https://www.ncbi.nlm.nih.gov/pubmed/18928474 DOI: https://doi.org/10.1111/j.1440-1754.2008.01363.x
9. Liu SM, Resende PVG, Bahia M, Penna FJ, Ferreira AR, Liu PMF, et al. Doença celíaca. Rev Med Minas Gerais [Internet]. 2014; [citado 2018 ago 10]; 24(Supl 2):38-45. Disponível em: http://www.rmmg.org/artigo/detalhes/622 DOI: http://www.dx.doi.org/10.5935/2238-3182.20140037
10. Sdepanian VL, Galvão LC. Doença celíaca. In: Campos Júnior D, Burns DAR, Lopez FA. Tratado de pediatria. 4a ed. Barueri (SP): Manole; 2017. p. 738-42.
11. Ministério da Saúde (BR). Portaria SAS/MS nº 1149, de 11 de novembro de 2015. Aprova o protocolo clínico e diretrizes terapêuticas da doença celíaca. Diário Oficial da União, Brasília (DF), 2015 nov 12; Edição 216: Seção 1: 65.
12. Batista GS, Carvalho SR. Doença celíaca: consenso. Rev Resid Pediatr [Internet]. 2014; [citado 2019 fev 10]; 4(1):36. Disponível em: http://residenciapediatrica.com.br/detalhes/98/doenca%20celiaca-%20consenso
13. Pereira PSC, Borges LGS. Bezoar - Relato de caso de um pré-escolar com síndrome de Rapunzel. Rev Resid Pediatr [Internet]. 2016; [citado 2019 fev 10]; 6(3):134-6. Disponível em: http://residenciapediatrica.com.br/detalhes/245/bezoar%20-%20relato%20de%20caso%20de%20um%20pre-escolar%20com%20sindrome%20de%20rapunzel DOI: https://doi.org/10.25060/residpediatr-2016.v6n3-07