A Pediatric Patient With Recurrent Abdominal Pain and Enamel Hypoplasia

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Case Report

A 4.3-year-old boy was referred to a pediatric clinic for recurrent abdominal pain with formed stools without hematochezia. The cramps were in the periumbilical region without vomiting and fever. No weight loss was reported.

He was born at term after an uneventful pregnancy with a birth weight of 2.930 g and a length of 51.0 cm. The Apgar score was not available, but spontaneous breathing, without cyanosis and jaundice, was documented. He received breast milk from birth until the age of 7 months when gluten was introduced into the diet without any adverse gastrointestinal effect.

At the time of the first referral (4.3 years), he showed a height of 101.2 cm (standard deviation score [SDS] = −0.93), and a weight of 14.5 kg (body mass index = −1.34 SDS). The target height of the boy was 169.5 cm (SDS = −1.20). Both parents were healthy and unrelated, without endocrinological or autoimmune diseases and had normal pubertal development. A physical examination revealed a distended abdomen without tenderness, defense, or masses. Bowel sounds were present without organomegaly. The remaining systemic examination showed no pathological findings. Biological parameters including white blood cell count, hemoglobin value, C-reactive protein, thyroxin, and thyroid-stimulating hormone were within the normal ranges. Furthermore, serum anti-tissue transglutaminase (tTG) antibodies (immunoglobulin [Ig]A-tTG = 0.1 U/mL, normal values <10 U/mL) and anti-deamidated gliadin peptide (DGP) antibodies were negative (IgA-DGP = 0.5 U/mL, IgG-DGP <0.0 U/mL, normal value <10 U/mL). No total circulating IgA value was available.

An ultrasound of abdomen was performed without any pathological findings.

As lactose breath hydrogen test is hard to apply in children, a lactose-free diet ex adiuvantibus was started on the basis of suspicion of lactose intolerance.

Hospital Course

After 3 years (at the age of 7.1 years), he returned to our department complaining of recurrent abdominal pains without diarrhea or weight loss. He showed a height of 122.0 cm (SDS = −0.06) and a weight of 21.0 kg (body mass index = −1.62 SDS) with a Tanner stage 1. Clinical examination was normal except for dental enamel hypoplasia with multiple white and cream opacities with clearly defined margins.

Due to clinical suspicion of celiac disease (CD), a second screening test was repeated and high IgA-tTG (58 U/mL) and IgG-tTG (1.7 U/mL) values were observed with positive endomysial antibodies in the presence of normal for age circulating IgA values.

HLA-DQ2 and HLA-DQ8 genotyping was performed and showed DQA1*03:03, 05:01 and DQB1*03:02, 02:01 haplotypes, determinants of genetic susceptibility for CD.

A duodenal biopsy was performed and revealed severe villous atrophy (Marsh 3b) with crypt hyperplasia and increased intraepithelial lymphocytes (>40 intraepithelial lymphocytes/100 epithelial cells).

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The final diagnosis was CD, and a gluten-free diet was consequently started. HLA genotyping screening in first-degree relatives revealed DQA1*05:01, 05:05 and DQB1*02:01, 03:01 in his father, who also showed positive serology for CD. The intestinal biopsy confirmed the diagnosis of CD in the father, too.

Discussion

Recurrent functional abdominal pain has a significant impact on clinical practice as a disorder that affects 10% to 15% of school-aged children.[1] The primary evaluation should include a complete history and physical examination to rule out the presence of blood in the stools and anal fissures, which require a referral in order to make a correct diagnosis.[2]

Children with recurrent abdominal pain need celiac screening as the first clinical approach.[3] After the exclusion of CD, appropriate dietary and nutritional counselling should be started to exclude lactose-containing products ex adiuvantibus from the diet.[2] In fact, lactose intolerance is a common disease in pediatric patients and can be divided into 2 main forms: congenital lactase deficiency (a rare autosomal recessive disease) and secondary lactase deficiency (a transient condition deriving from intestinal damage).[4]

CD is an immune-mediated systemic disease triggered by dietary gluten in genetically predisposed individuals carrying HLA-DQ2 and/or HLA-DQ8, characterized by a variable degree of intestinal villous damage and by a variable combination of clinical manifestations.[3] CD-specific antibodies include anti-tTG antibodies, endomysial antibodies, and antibodies against DGP.[3] The incidence of CD in the general population is estimated to be about 1%.[5] The clinical spectrum is broad and includes typical forms usually presenting in childhood with signs of intestinal malabsorption, including recurrent abdominal pain, or in atypical forms with only extraintestinal manifestations such as short stature, delayed puberty, anemia, and particular enamel mineralization defects on permanent teeth and oral aphthous ulcers.[6] Silent forms of CD may also occur and are usually detected in first-degree relatives of celiac patients who undergo serological screening.[3]

This clinical case represents an example of difficulty in the diagnosis of CD, since, at the first examination, the child had recurrent abdominal pain but negative serology for CD. During the follow-up, only dental enamel lesions induced us to repeat the anti-tTG evaluation, which was positive.

Currently, there is a general trend of delayed onset of symptomatic CD involving older children between 5 and 7 years. These children frequently experience extraintestinal manifestations such as short stature or dental enamel defects.[8] In fact, the oral cavity is one of the areas of CD manifestation. These findings are highly suggestive of CD, as dental enamel lesions are observed in 60% of such patients.[6] According to some studies, impairment of dental crown mineralization appears symmetrically and chronologically in the same anatomical groups of teeth in all 4 quadrants of dentition. These defects may range from discoloration to pitting, grooving, and total loss of enamel.[3,7,8]

The atypical and oligosymptomatic clinical presentation of CD stresses the importance of considering dental enamel defects as a complementary tool for the diagnosis of atypical forms of CD or for surveillance of first-degree relatives.[6]

CD and lactose intolerance belong to a spectrum of adverse food reactions with different pathogeneses (immune mediated vs enzymatic defects), with both of them responsible for the same clinical symptoms such as distension of the small bowel, nonfocal abdominal pain associated with bloating and flatulence, nausea, increased gut motility, and sometimes diarrhea.[9-12]

The hydrogen breath test for the diagnosis of lactose intolerance is difficult to apply in children younger than 5 years, due to the patients' low compliance.

The routine exclusion of a diagnosis of CD for all patients with recurrent abdominal pain is currently recommended by the European Society for Paediatric Gastroenterology Hepatology and Nutrition (ESPGHAN) guidelines for CD.[3] However, CD is often atypical or even clinically silent and many patients remain undiagnosed and are exposed to the risk of long-term complications, such as osteoporosis, infertility, or cancer.[13]

According to the new ESPGHAN guidelines for CD diagnosis, HLA-DQ testing is recommended only in seronegative cases with severe symptoms and a strong clinical suspicion of CD or in individuals with an increased genetic risk for CD (first-degree relatives). When HLA-DQ2/DQ8 is positive, surveillance should continue and serology for CD should be repeated every 3 years.[3,14]

Conclusion

In accordance with other previous studies,[15] this case report highlights the importance of an in-depth investigation of all nonspecific symptoms in order to understand the real cause of recurrent abdominal pain. We suggest paying particular attention when dealing with children with abdominal pain because symptoms of CD overlap with those of irritable bowel syndrome. Furthermore, a meticulous examination of the oral cavity with particular

attention directed to dental enamel defects, aphthae, and other mucosal disorders, as well as to delayed teeth eruption, may contribute to an early diagnosis of CD.

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Author Contributions

PM: Contributed to acquisition, analysis, or interpretation of data; drafted manuscript; gave final approval; agrees to be accountable for all aspects of work ensuring integrity and accuracy.
FV: Contributed to acquisition, analysis, and interpretation; critically revised the manuscript for important intellectual content; gave final approval; agrees to be accountable for all aspects of work ensuring integrity and accuracy.
CM: Critically revised manuscript; gave final approval; agrees to be accountable for all aspects of work ensuring integrity and accuracy.
AV: Carefully read the intestinal biopsy; improved the Discussion section from an anatomopathological point of view.
MB: Contributed to conception and design; critically revised manuscript; gave final approval; agrees to be accountable for all aspects of work ensuring integrity and accuracy.

Declaration of Conflicting Interests

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References

1. Nurko S. The tip of the iceberg: the prevalence of functional gastrointestinal diseases in children. *J Pediatr*. 2009;154:313-315.
2. Gomez-Suarez R. Difficulties in the diagnosis and management of functional or recurrent abdominal pain in children. *Pediatr Ann*. 2016;45:e388-e393.
3. Husby S, Koletzko S, Korponay-Szabó IR, et al. European Society for Pediatric Gastroenterology, Hepatology, and Nutrition guidelines for the diagnosis of coeliac disease. *J Pediatr Gastroenterol Nutr*. 2012;54:136-160.
4. Berni Canani R, Pezzella V, Amoroso A, Cozzolino T, Di Scala C, Passariello A. Diagnosing and treating intolerance to carbohydrates in children. *Nutrients*. 2016;8:157.
5. Hill ID, Dirks MH, Liptak GS, et al; North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. Guideline for the diagnosis and treatment of celiac disease in children: recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. *J Pediatr Gastroenterol Nutr*. 2005;40:1-19.
6. Krzywicka B, Herman K, Kowalczyk-Zając M, Pytrus T. Celiac disease and its impact on the oral health status—review of the literature. *Adv Clin Exp Med*. 2014;23:675-681.
7. Fasano A. Clinical presentation of celiac disease in the pediatric population. *Gastroenterology*. 2005;128:S68-S73.
8. Aine L. Dental enamel defects and dental maturity in children and adolescents with coeliac disease. *Proc Finn Dent Soc*. 1986;82(suppl 3):1-71.
9. Hammer HF, Hammer J. Diarrhea caused by carbohydrate malabsorption. *Gastroenterol Clin North Am*. 2012;41:611-627.
10. Raithel M, Weidenhiller M, Hagel AF, Hetterich U, Neurath MF, Konturek PC. The malabsorption of commonly occurring mono and disaccharides: levels of investigation and differential diagnoses. *Dtsch Arztebl Int*. 2013;110:775-782.
11. Latulippe ME, Skoog SM. Fructose malabsorption and intolerance: effects of fructose with and without simultaneous glucose ingestion. *Crit Rev Food Sci Nutr*. 2011;51:583-592.
12. Born P, Sekatcheva M, Rosch T, Classen M. Carbohydrate malabsorption in clinical routine: a prospective observational study. *Hepatogastroenterology*. 2006;53:673-677.
13. Fasano A, Catassi C. Current approaches to diagnosis and treatment of celiac disease: an evolving spectrum. *Gastroenterology*. 2001;120:636-651.
14. Bozzola M, Bozzola E, Pagani S, Mascolo A, Porto R, Meazza C. Late diagnosis of celiac disease in an asymptomatic infant with growth failure. *Ital J Pediatr*. 2014;40:4.
15. Kansu A, Kuloğlu Z, Demir A, Yaman A; Turkish Celiac Study Group. Yield of coeliac screening in abdominal pain-associated functional gastrointestinal system disorders. *J Paediatr Child Health*. 2015;51:1066-1070.