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CASE REPORT

A long way to a simple diagnosis – a case report of an overlooked juvenile polyp

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ABSTRACT

Lower gastrointestinal bleeding (also known as rectal bleeding) is a common symptom reported to a pediatrician. It can be caused by a variety of medical conditions across different age groups, including in the majority, benign and common, such as annal fissure, rectal polyps or gastrointestinal infection. We report the case of a 10-year-old boy with an isolated juvenile polyp. The multi-symptoms and inappropriate/incorrect initial diagnosis (eosinophilic colitis) led to unnecessary medical tests and interventions, including a highly restrictive diet. This case highlights the need for higher awareness of the most common causes of rectal bleeding which should be firstly excluded. It also shows a challenging diagnosis in case of several gastrointestinal symptoms that may be associated with more than one possible disorder. Highly restrictive elimination diet in children is related to some risks and should be used only in conditions with some evidence of its efficacy.

KEY WORDS:
juvenile polyp, gastrointestinal bleeding, bloody stools, differential diagnosis.

INTRODUCTION

Lower gastrointestinal bleeding (LGIB), defined as bleeding distal to the ligament of Treitz and also referred as rectal bleeding, is a common condition in children, affecting 0.3% of children in the emergency department [1]. Two main types of LGIB are distinguished: hematochezia (the passage of fresh blood per anus) and melena (passage of black, tarry stools per anus) [2]. Rectal bleeding can be associated with a range of conditions, from mild and self-limiting to life-threating diseases.

The causes of rectal bleeding vary by age group. In children over 5 years old, anal fissures, often associated with constipation, infectious enterocolitis, and polyps, are the three most common causes of rectal bleeding. Other, less frequent but still possible causes include inflammatory bowel disease, lymphonodular hyperplasia, Henoch-Schönlein purpura, angiodysplasia, hemolytic-uremic syndrome, bleeding diathesis, and rare eosinophilic gastrointestinal disorders (EGIDs). These should also be considered later in the differential diagnosis [2]. The variety of clinical conditions that can present with rectal bleeding, combined with non-specific symptoms, can make the diagnosis challenging.

We present the case of a 10-year-old boy with a 2-year history of rectal bleeding, who endured ineffective treatment and a prolonged diagnostic process due to an incorrect initial diagnosis. The unnecessary introduction of a 'hypoallergenic' diet, compounded by his food selectivity, led to a prolonged period on a highly restricted diet.

CASE REPORT

The preferred reporting item for case report guidelines were followed [3].

We present a 10-year-old boy who was admitted to our department in January 2024 due to a 2.5-year history of periodic LGIB. His main symptom was bloody stools

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with mucous, occurring every 3–4 days. The boy also reported symptoms of constipation, including lower abdominal pain relieved after defecation, painful defecation every 2 days, hard stools, and straining. Additional symptoms included dysphagia (particularly when swallowing solid foods) and nausea. The boy was under the care of a psychological counseling center due to anxiety disorders and autism spectrum disorder.

PATIENT HISTORY

In early childhood (at 4 month of age), due to recurrent rashes and symptoms suggesting infant colic, the boy was diagnosed with cow's milk allergy. As a result, a cow's milk-free diet using extensively hydrolyzed formula, was prescribed, with subsequent improvement in symptoms. Although the diagnosis was not confirmed by an oral food challenge, the diet was maintained until he was 14 months old. At the age of 8, component-resolved diagnostics (ALEX®) identified only inhaled sensitivities/allergies, specifically to grass and tree pollen. At 8 years of age, he was first hospitalized due to lower intestinal bleeding, characterized by bloody stools occurring every 3–4 days. An elevated calprotectin level of 2388 μg/g (normal: 50 μg/g) was noted, while results of other laboratory tests were normal, including peripheral blood count, ionogram, aminotransferases, pancreatic enzymes, blood coagulogram, and thyroid hormones.

During the initial gastro- and colonoscopy, samples were taken from the esophagus, cardia, antrum, duodenum, ileum, cecum, ascending, transverse, and descending colon, as well as the sigmoid and rectum. A polyp in the cardia was identified and removed; however, apart from the cardia polyp, the macroscopic evaluation was normal. In the histopathological assessment, the number of intraepithelial eosinophils per high-power field (eos/hpf) was elevated, with 23 eos/hpf in the duodenal bulb, ranging from 11 to 63 eos/hpf in the other colon samples, and 4 eos/hpf in an erosion (not reported macroscopically) from the cardia. No other abnormalities were found. Based on the increased eosinophil count, the patient was diagnosed with allergic proctitis (proctitis allergica). This diagnosis was considered misleading due to his age and ongoing symptoms, including persistent constipation.

Subsequently, an antihistamine, proton-pump inhibitors, macrogols (for symptoms recurrence), and a 'hypoallergenic' diet – excluding milk, nuts, soy, eggs, citrus fruits, and cacao were introduced. However, after 9 months, no improvement was observed despite the treatment. Therefore, it was modified to sodium cromoglycate and a multi-strain probiotic (*Streptococcus thermophilus* DSM24731, *Bifidobacterium longum* DSM24736, *Bifidobacterium breve* DSM24732, *Bifidobacterium infantis* DSM24737, *Lactobacillus acidophilus* DSM24735, *Lactobacillus plantarum* DSM24730, *Lactobacillus paracasei* DSM24733, *Lactobacillus delbrueckii ssp. bulgaricus* DSM24734), along with suppositories containing hydrocortisone; the hypoallergenic diet was continued. After 22 months from the initial diagnosis, no improvement was reported in the boy's condition.

When the boy was admitted to our department, he was receiving ketotifen, vitamin D (2500 IU), and probiotics. His current diet was extremely selective, consisted mainly of meat (chickens, sausage), bread, sweets, and a limited number of vegetables and fruits. He particularly avoided lactose, citrus fruits and food coloring E133 as his parents believed these were triggers for his symptoms. The child had never been consulted by a dietician.

DIAGNOSTIC ASSESSMENT AND TREATMENT

Upon admission to our clinic, no significant deviations were found during examination. The patient's growth and physical development were normal. Laboratory tests, including blood count, urinalysis, aminotransferases, inflammation indicators, tissue transglutaminase IgA, and Clostridium difficile toxins, revealed no abnormalities. The only notable finding was a slightly elevated fecal calprotectin level (164.5 ug/g; normal range [NR < 50 μg/g]). Additionally, the follow-up histopathological analysis of tissue samples from previously collected gastrointestinal sections, performed by the consulting pathomorphologist, also revealed no significant abnormalities.

Due to inconsistent results and a lack of consensus regarding the patient's final diagnosis, a decision was made to conduct a more thorough diagnostic assessment by repeating the gastro- and colonoscopy. During the interim period until the next planned hospitalization, he was only prescribed macrogols (10 g per day).

FOLLOW-UP

After one month, the patient reported improvement in some symptoms, including less frequent lower intestinal bleeding (only once in the last month) and no constipation or abdominal pain. The patient also enriched his diet and became more willing to try new foods. However, nausea and dysphagia persisted. Considering the above symptoms, the gastro- and colonoscopy were repeated.

During the colonoscopy, a large polyp in the rectum was successfully identified and removed. This polyp, approximately 10 mm in size, was histologically classified as a juvenile polyp (Figure 1). Due to its location, it may have been overlooked during earlier examinations. Gastroscopy revealed only mild signs of esophageal trachealization; however, the histopathological examination did not show the presence of eosinophils. All other tissue samples taken during the endoscopy were histologically assessed as normal.

Since none of EGIDs was confirmed, no additional treatment was required beyond continuing the regular diet and vitamin D supplementation. Due to the lack

of an organic explanation for some of the patient's persistent symptoms (nausea, dysphagia) and his positive psychiatric history (anxiety disorder), somatization and obsessive-compulsive disorder were suspected. Psychiatric consultation and intensified psychological care were recommended.

DISCUSSION

This case highlights two issues often observed in the management of pediatric patients. The first issue relates to the ease with which highly restrictive elimination diets are introduced without sufficient clinical justification. The second concerns the diagnostic difficulties associated with the broad spectrum of common gastrointestinal symptoms reported by the patient. Additionally, this case also highlights the challenging diagnosis of children with several gastrointestinal symptoms that may be not limited to one condition, and this may delay an appropriate diagnosis.

The patient was initially diagnosed with proctitis allergica, a condition typically affecting infants, also known as eosinophilic proctocolitis [4]. This condition is characterized by inflammatory changes in the distal colon caused by an immune response to one or more food proteins [5]. In most cases, eosinophilic proctocolitis can be diagnosed based on an elimination diet followed by an oral food challenge with the suspected triggering food allergens. The treatment involves an elimination diet excluding the triggering food allergen(s).

Proctitis allergica typically resolves by late infancy in most patients [5]. In this case, we can presume that, despite the terminology used, a different diagnosis was intended. According to the nomenclature employed for adults and older children, and based on the histopathological examination results, our patient may have been more accurately diagnosed with eosinophilic colitis (EC). Eosinophilic colitis is a very rare manifestation of EGIDs [4]. It usually presents with abdominal pain, changes in bowel movements, diarrhea, rectal bleeding, and failure to thrive. However, these symptoms are nonspecific and should not be considered characteristic clinical features [6].

In adult patients, a value above 64 eos/hpf in the rectosigmoid is considered the cutoff for diagnosing EC [7]. However, it is important to note that these data concern adult patients, and the baseline count and distribution of eosinophils in the gastrointestinal tract of the pediatric population are even less well defined [8]. While some studies suggest that a count exceeding 50 eos/hpf should be considered normal [9], there is a lack of well-established guidelines, making the diagnosis and management of EC challenging. Some authors recommend steroid therapy as a first-line treatment [9].

Other recommended treatment methods include dietary therapy (preferably based on results of food allergy tests; the proposed treatment may range from dietary restriction of suspected allergens to an elementary diet), PPI, ketotifen, sodium cromolyn, immunosuppressive or biologic therapy [10]. Although no food allergies were diagnosed in this case, the patient was recommended to introduce a 'hypoallergenic' diet as the first-line treatment, in addition to ketotifen and PPI. Due to the patient's high food selectivity, both of these factors eventually resulted in a very restrictive and nutritionally poor diet. Unfortunately, this management approach did not improve the patient's well-being, so the therapy was partially modified. However, the dietary treatment remained unchanged, leading the patient to remain on a strict diet for almost two years, which turned out to be unnecessary.

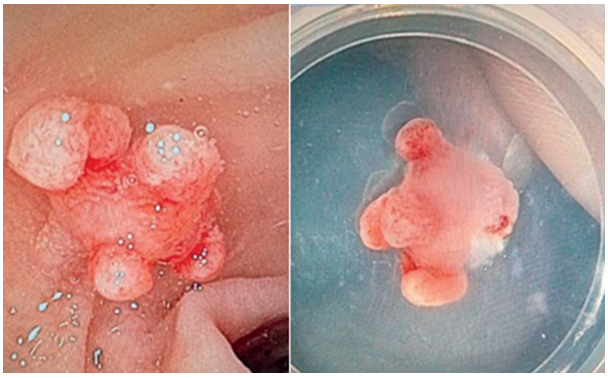

FIGURE 1. A) Colonoscopy showing the polyp located at the border of sigmoid colon and rectum measuring 10 mm in diameter. **B**) The excised specimen

After the second endoscopy, the patient's diagnosis was completely revised. Eosinophilic colitis was excluded and a juvenile polyp was detected. Isolated juvenile polyp is a condition frequently diagnosed in the pediatric population, affecting up to 2% of children [11]. It typically presents with rectal bleeding, usually related to defecation, and may occasionally be associated with abdominal pain [12]. Juvenile polyps are most commonly found during endoscopy in children aged 2–10 years and are slightly more prevalent in males (3 : 2) [13]. The majority of polyps are located proximal to the rectosigmoid junction, although those located distally are more often symptomatic. Isolated juvenile polyps rarely recur after removal and have virtually no malignant potential. Therefore, endoscopic extraction and histological confirmation of their benign nature allow the patient to be considered cured [13].

Although colonoscopy is the gold standard for the detection and removal of polyps, up to 22% of polyps may be missed [14]. In this case, a juvenile polyp was overlooked during the first endoscopy, leading to an inappropriate/incorrect diagnosis and treatment. Even though no improvement was observed, the doctor proposed only a modification of the previous treatment without reconsidering the primary diagnosis. Given the persistence of gastrointestinal bleeding, it seems appropriate to extend the diagnostics and repeat the endoscopic examination to verify the previous diagnosis.

As previously mentioned, juvenile polyps rarely cause abdominal pain. Although lower abdominal pain was present in this case, we cannot clearly attribute it to the juvenile polyp as the patient also suffered from constipation (painful defecation every two days, hard stools, and straining) [15]. Additionally, the constipation was not treated intensively enough at the beginning, but the symptoms improved after the intensification of macrogol treatment, coinciding with the removal of the polyp. Therefore, it is unclear whether the abdominal pain was directly related to the juvenile polyp. The associated rectal bleeding and its persistence, despite the eventual appropriate administration of macrogol doses, suggested another underlying cause of the bleeding—in this case, the juvenile polyp.

CONCLUSIONS

In conclusion, even multiple symptoms affecting the same system may be linked to a few common conditions (such as juvenile polyp and constipation) rather than to a single, extremely rare condition (eosinophilic colitis). Dietary restrictions, particularly the exclusion of multiple foods or entire food groups, should only be recommended when absolutely necessary and supported by evidence of efficacy. This case also highlights the importance of reintroducing a regular diet when no symptom improvement is observed despite dietary intervention.

DISCLOSURES

1. The Ethics Committee of the Medical University of Warsaw approved this case report (approval number: AKBE/278/2024). Informed consent was not required as this case was purely observational. Nonetheless, all patients treated at our hospital are informed that we operate under the auspices of the university, which includes notifying them and their caregivers that their data and treatment results may be used anonymously for research and publication purposes.
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4. Conflicts of interest: None.

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