Six-year-old Girl with a Rare Cause of Bloody Diarrhea

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Introduction

A previously healthy, 6-year-old, girl presented with a 2-month history of bloody diarrhea, associated with abdominal pain, anorexia, weight loss, and intermittent fever. She has not travelled recently, and reported no contact with sick people or animal exposure. Physical examination revealed a well looking child with mild conjunctival pallor. Abdominal examination revealed tenderness in the left iliac fossa with no palpable masses or organomegaly. Perianal examination was declined. Her initial colonoscopy demonstrated severe ulcerating rectal mucosa with fungated lesions extending into sigmoid colon (Figure 1). Histological examination of the rectal biopsy revealed distorted crypt architecture with crypt branching and increased inflammatory cells, dominantly eosinophils. The clinical, biochemical and histological findings were suggestive of inflammatory bowel disease (IBD), so she was commenced on 5-aminosalicylic acid and oral prednisolone(1mg/kg/day). Her condition deteriorated after starting prednisolone and she developed a fever with severe perianal pruritus and pain. Therefore, her immunosuppressive therapy was escalated to intravenous methylprednisolone, mercaptopurine (6-MP) and infliximab. She also received intravenous metronidazole and cefuroxime. Laboratory investigations revealed a high C- reactive protein of 66 mg/l, hypocromic microcytic anemia (Hb 8.7 g/dL (11.5-15.5) with thrombophilia (840 X 10^9) and eosinophilia at 2.8 (0.1-0.8 X 10). She had low iron of 2 umol/L (6-35) with high ferritin of 266 ug/L (4-67).

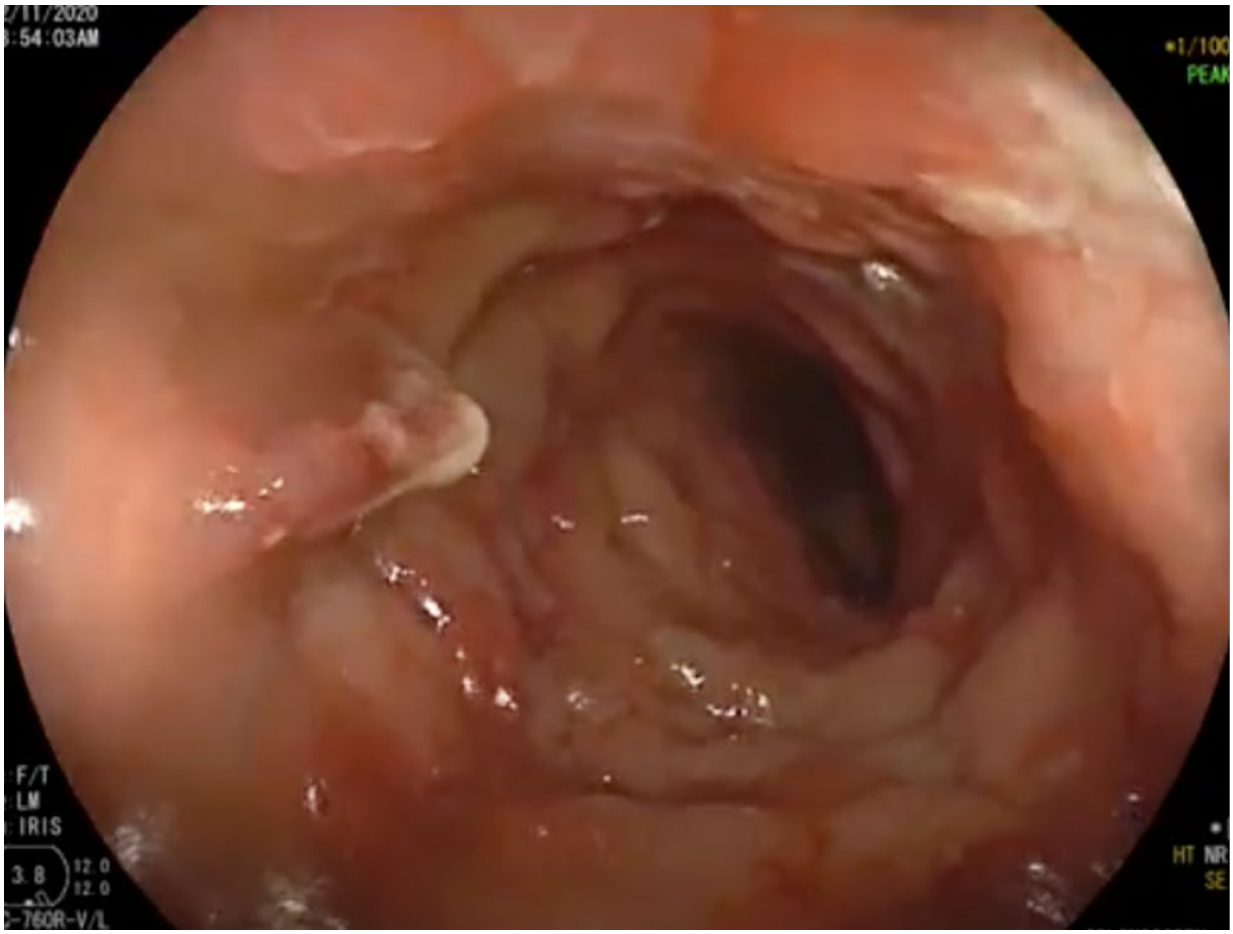

Figure 1: Endoscopy image reveals severe ulcerating rectal mucosa with fungated lesions extending into sigmoid colon.

After the second dose of infliximab, her symptoms worsened, and she developed urinary retention. Magnetic resonant image (MRI) pelvis was done and revealed severe irregular thickening of the rectal wall extending up to the sigmoid. There was evidence of inflammatory mass (rectal wall thickness measures 1.8 cm extending for 10 cm in length) which was compressing the surrounding structures. (Figure 2). Parents' consent was taken for publication.

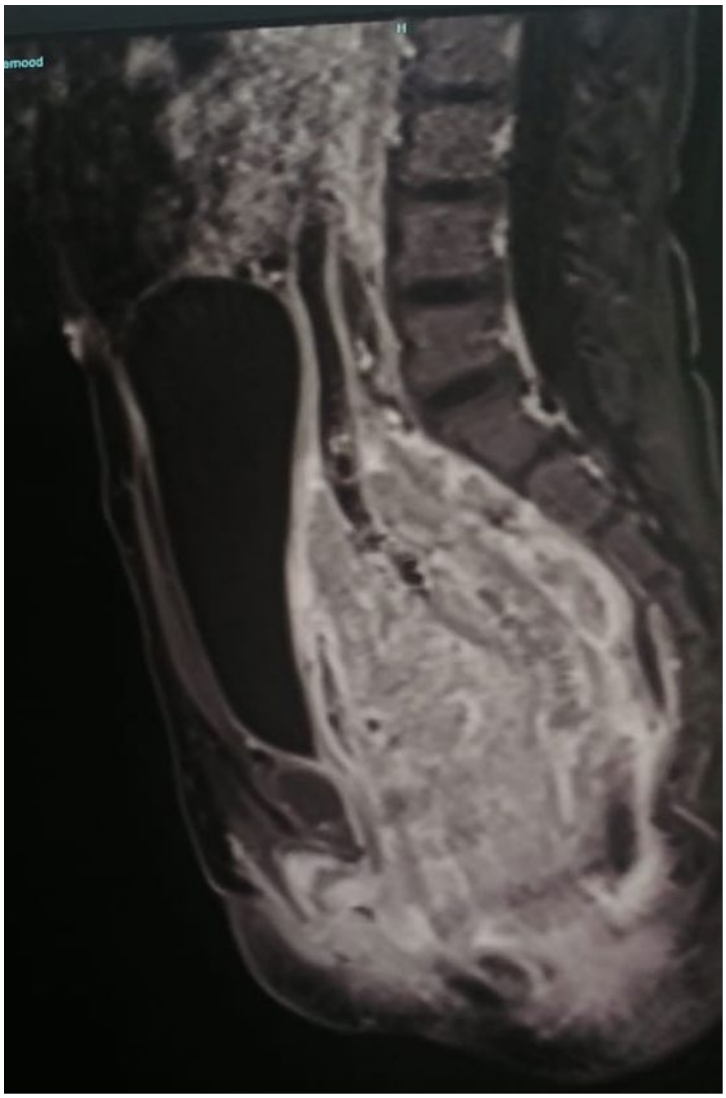

Figure 2: Sagittal view of magnetic resonant image of the patient's pelvis at the initial presentation showing extensive thickening and inflammation of the whole rectum including rectosigmoid junction (10cm in length). The inflammatory mass is compressing the surrounding structure including the uterus, urinary bladder and subcutaneous fat and facia.

Questions

1. What is your diagnosis?

2. How to rule out other possible differential diagnosis?

3. What is the gold standard test to make the diagnosis?

4. How would you manage this patient?

Answers

1. Gastrointestinal basidiobolomycosis

2. Work up for primary immunodeficiency, tuberculosis, malignancies and monogenic IBD molecular testing

3. Tissue culture, but histopathology can help in making the diagnosis,

4. Voriconazole or itraconazole for a minimum of 9-12 months

Discussion

The possibility of gastrointestinal basidiobolomycosis was raised in our patient given the deterioration on IBD therapy, the presence of peripheral eosinophilia, abdominal mass and increased eosinophilic infiltration in the colonic biopsy. Deep biopsy to confirm the diagnosis was deferred due to the potential high-risk complications raised by the surgeons. Her immunosuppressive medications were ceased, and she was commenced on empirical itraconazole (5mg/kg/ dose) twice daily. The patient's condition improved clinically and biochemically significantly within few days. Her inflammatory markers and eosinophils count normalized within two weeks from initiating itraconazole. Her 6-weeks pelvis MRI follow-up showed significant interval improvement of the rectal wall thickness with reduced perirectal oedema and inflammation. Her subsequent clinic follow-ups showed significant improvement in her symptoms.

Inflammatory bowel diseases are chronic inflammatory disorders of the gastrointestinal tract. There are many conditions that can mimic IBD. Lack of response to standard IBD therapy should prompt consideration of an alternative diagnosis. IBD mimics can be classified broadly into two main categories including infectious and noninfectious etiologies. Rarely fungal infection like *basidiobolomycosis* can mimic IBD in presentation.[1]

Basidiobolomycosis is a rare fungal infection caused by *Basidiobolus ranarum* which is an environmental saprophyte. It is found in soil, dung of amphibians, reptiles and decaying vegetable.[2] It is one of the zygomycosis that can cause skin, soft tissue and gastrointestinal infections in immunocompetent patients in tropical and subtropical parts of the globe.[3] Gastrointestinal basidiobolomycosis (GIB) is an emerging fungal infection that can affect immunocompetent patients including children.[2,3] There have been more than 120 reported cases of GIB worldwide. Most reported cases were in men with > 50% of cases reported in children.[3] Previous reports suggest that such infection can be acquired after a minor trauma to the skin[2] or via ingestion of contaminated food with fungus from soil or animal dung.[2,4]

In Oman, Al-Masqari et al. reported a case series of 5 patients with GIB managed at Royal Hospital, a paediatrics tertiary center, recently. The majority of these patients were children from Al Dhakhylia region.[3] *Basidiobolus omanensis* is a novel species of Basidiobolus that was just reported recently from four Omani patients by Al-Hatmi and his colleagues.[5]

Due to the rarity of the disease and unspecific presenting signs and symptoms, the diagnosis of this condition was challenging. Clinical presentation of GIB includes abdominal pain, fever, weight loss, diarrhoea or even constipation, abdominal distention or abdominal mass which can mimic IBD.[4]

The gold standard to diagnose GIB is tissue culture, but histopathology can help in making the diagnosis, where typical features like chronic granulomas with high eosinophils infiltrate and the Splendore–Hoeppli phenomenon are noted.[3] Despite all the challenges in reaching the diagnosis, the best diagnostic clue of this rare infection is considering this disease in the differential diagnosis of patients presenting with abdominal pain and fever, associated with intestinal or colonic mass and/or wall thickening, and concomitant high ESR and eosinophilia.[3,4] In our patient, the lack of awareness of this infection as a IBD's mimicker has led to misdiagnosis and deterioration of her clinical condition. Delaying GIB treatment might cause disseminated disease and poor outcome.[3] Prompt treatment by antifungal therapy with or without surgery is recommended to eradicate the infection and prevent recurrence.[3,4]

Disclosure

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