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Coexistence of Infantile Crohn and Humoral Immunodeficiency: A Case Report

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1. Abstract

Infantile inflammatory bowel disease (IBD) is a kind of IBD that begins in early childhood. However, this subgroup is rare, it is important to detect these patients early. Infantile IBD patients may have more surgical interventions and have a greater failure rate of standard therapy. In this disorder, monogenic mutations are essential. We represent an 8-month-old patient with an anal fistula, failure to thrive, and systemic infection. IL-10 pathway mutation is considered with IBD. The patient had refractory illness despite standard therapies, surgical procedures, and antitumor necrosis factor-alpha therapies. The patient is waiting for hematopoietic stem cell transplantation.

2. Introduction

IBD in children is an uncommon kind of inflammatory bowel disease. Patients with an age of onset less than 10 years are classed as A1a in the pediatric Paris modification of the Montreal classification (early-onset IBD). However, according to some recent research, infantile-IBD is defined as an early-onset illness diagnosed before the age of six, and very-early-onset IBD (VEO-IBD) diagnosed before the age of two [1-3]. The onset of IBD in childhood is quite rare. It is believed that the pathogenesis of IBD starts with altered intestinal responses depending on various external stimuli in genetically susceptible hosts. In individuals with infantile-onset enterocolitis, mutations in genes producing interleukin-10 (IL10) or interleukin-10 receptors (IL10r) component proteins have been discovered, frequently within the first three months of life. Infants with these mutations typically have a severe perianal illness as well as extra-intestinal symptoms, including folliculitis and arthritis. [4]. In our case, we present an infantile onset-IBD patient who is diagnosed with Crohn's disease due to suspicion of IL10 receptor mutation.

3. Case

An 8-month-old male patient applied to Ankara City Hospital / Children Hospital with diarrhea and flaky, peeling skin since he was five months. He was born in the 4th pregnancy of a 29-year-old Afghan mother 4th pregnancy, had no antenatal follow up, and was born C/S section at 38 weeks of gestation. The patient was hospitalized due to bacterial and fungal sepsis when he was 45 days old. He had oral moniliasis, four times otitis media, and rectal prolapsus for two months. In his physical findings, his height: 66 cm (-1.9 SDS) weight:6400 g (-2.5 SDS). He had hyperemia in the anal region, fistula, maculopapular, and small pustular rash (Figure 1). He had a family history of one sister being ex due to bloody diarrhea. She was two months old, and he has an 8-year-old sibling diagnosed with IL-10 receptor mutation. As a consequence, patient was pre-diagnosed with IBD related to immunodeficiencies. To clarify the diagnosis, further tests and colonoscopy are planned. In patients' laboratory findings, has findings compatible with iron deficiency biochemical test results within normal limits, the c-reactive protein (CRP) level was high at 153 mg/l(N:0-5 mg/l). In abdomen ultrasonography (USG) of the patient, rectosigmoid, ascendant, and descendant colon was reported with increased wall thickness. Surface USG has been done due to the formation of fistulas. It was observed that loculated liquid thickness of 3 millimeters (mm) at 8-9 o'clock in the anus might be related to the accompanying inflammatory disease. The colonoscopy reported that normal histology of the colon mucosa was lost (Figure 2). Many inflammatory polyps and large deep white membrane-covered blooded ulcers were observed, which were associated with serious perianal disease. Differential diagnosis CMV and tuberculosis PCR from the tissue has been evaluated. In the biopsy report, the integrity of the surface epithelium is destroyed in the focal area, and there

is inflammatory debris on the surface and granulation tissue below. Distortion in crypts, branching, cryptic, regenerative changes, and mixed inflammation were observed in the lamina propria. With these findings, the patient was diagnosed with IBD. Prednisolone and azathioprine treatments were started. Genetic analysis was sent for the IL10 receptor mutation due to family health history. Fluconazole, Sulfamethoxazole Trimethoprim, and Intravenous immunoglobulin (IVIG) treatments were started because underlying immunodeficiency was considered. Infliximab treatment was started when the still patient had diarrhea under current therapy, and after treatment, the number of stools in a day was decreased. The patient is still suffering from perianal abscesses under Infliximab (IFX) treatment. He had multiple time of hospitalization because of pneumonia and perianal abscesses. He had been operated on for colostomy for recurrent abscess and bladder fistula after 15 months as he was first prediagnosed as IBD. His current body weight is 11 kg (-1.77 SDS) and height is 86 cm (-1.44 SDS). After the patient's genetic analysis, he can be a candidate for hematopoietic stem cell transplantation.

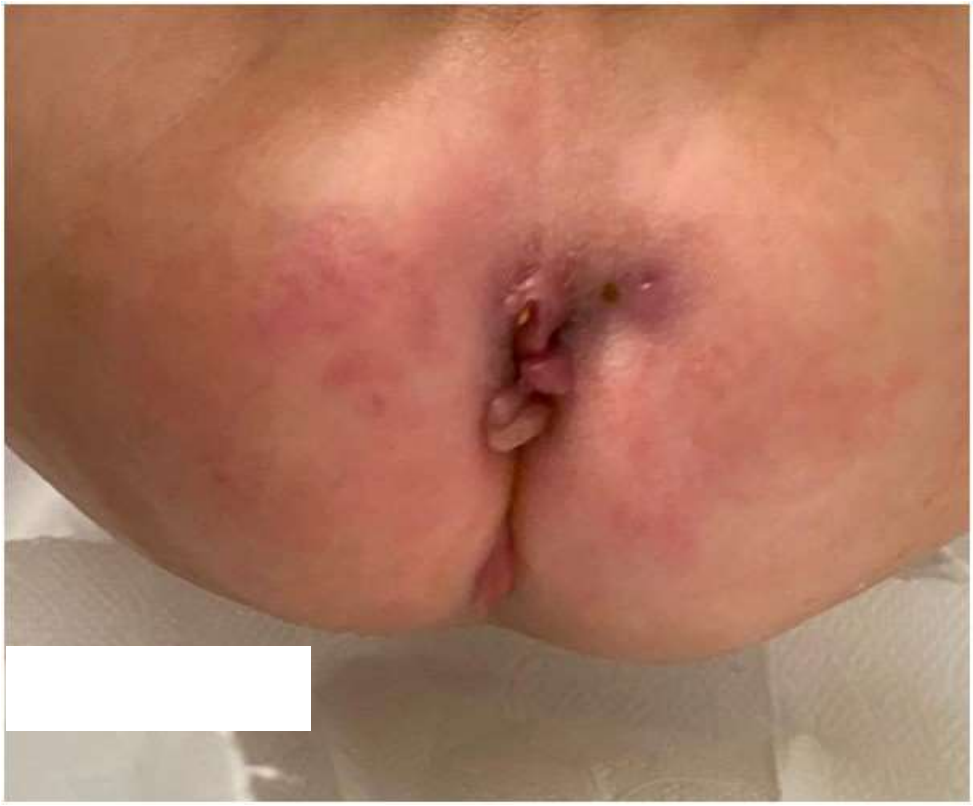

Figure 1. Anocutaneous fistulas.

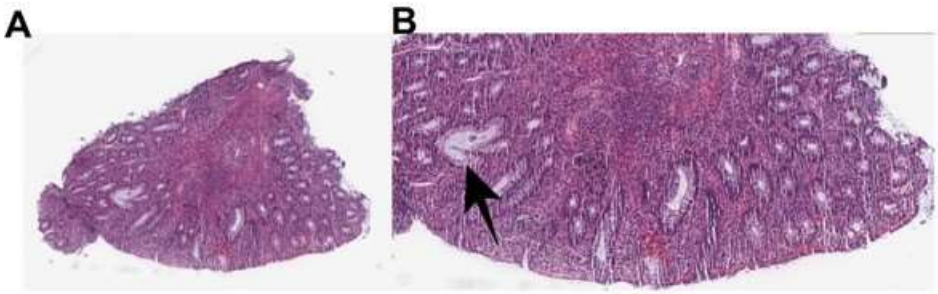

Figure 2. Histological findings of the colon (A) 10x zoom, Chronic inflammation in the crypts, crypt loss, and hyperplastic changes (B) 100x zoom (Hematoxylin and Eosin Stain).

4. Results and Discussion

At the time of presentation, most patients with infantile-onset IBD had isolated colonic illness. Still, disease presentation may change over time, which may cause trouble in classifying IBD as ulcerative colitis or Crohn's disease. Diagnosis of the disease is important because it may affect the operative strategies. Previous studies demonstrated that children diagnosed with IBD at earlier ages used fewer health services and had a lower rate of surgery [5]. Therefore infantile-onset IBD patients should undergo immunologic and genetic evaluation. The pathophysiology of infantile-onset IBD (IO-IBD) is compounded by an inherited genetic abnormality that causes immunological dysregulation. In newborns with severe IO-IBD, which may frequently be present with perianal fistula and respond poorly to pharmacological therapy and early surgical procedures, clinicians must rule out mutations in IL10 and IL10r [6]. IBD caused by IL10 and IL10r loss has responded well to hematopoietic stem cell therapy. However, not every IO-IBD carries the IL10 and IL10r mutation; many monogenic disease mutations have been discovered in previous research, and primary immunodeficiencies can cause IBD [1]. Patients with IO-IBD typically require aggressive treatment., which may include immunosuppression of biologics such ah infliximab, and these therapies do not induce remission in patients with IL10r mutation. Stem cell transplantation is the current curative therapy [1,4,7,8].

5. Conclusion

In a nutshell patient's clinical findings, family history, and response to treatment support the diagnosis of IO-IBD related to IL10 deficiency. Patients with chronic diarrhea, fistula, and family history of immunodeficiency must be examined considering IO-IBD. Diagnosis may cause unnecessary surgeries and ineffective medical therapies [1,9].

References

1. Hung HH. Importance of early detection of infantile inflammatory bowel disease with defective IL-10 pathway: A case report. Medicine, 2021; 100(21): e25868-e25868.
2. Kelsen JR. The Unique Disease Course of Children with Very Early onset-Inflammatory Bowel Disease. Inflamm Bowel Dis. 2020; 26(6): 909-918.
3. De Bie CI. Diagnostic workup of paediatric patients with inflammatory bowel disease in Europe: results of a 5-year audit of the EUROKIDS registry. J Pediatr Gastroenterol Nutr. 2012; 54(3): 374-80.
4. Lee WS. Variable outcome in infantile-onset inflammatory bowel disease in an Asian cohort. World journal of gastroenterology, 2016; 22(48): 10653-10662.
5. Benchimol EI. Incidence, outcomes, and health services burden of very early onset inflammatory bowel disease. Gastroenterology. 2014. 147(4): 803-813.e7.
6. Kotlarz D. Loss of interleukin-10 signaling and infantile inflammatory bowel disease: implications for diagnosis and therapy. Gastroenterology. 2012; 143(2): 347-55.
7. Ye Z. Phenotype and Management of Infantile-onset Inflammatory Bowel Disease: Experience from a Tertiary Care Center in China. Inflamm Bowel Dis. 2017; 23(12): 2154-2164.
8. Yeh PJ. High initial bowel resection rate and very-early-onset inflammatory bowel disease - A challenge in a low-prevalence area. J Formos Med Assoc. 2021; 120(1 Pt 3): 720-727.
9. Glocker EO. Inflammatory bowel disease and mutations affecting the interleukin-10 receptor. N Engl J Med. 2009; 361(21): 2033-45.