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REFERENCES

1. Purtilo DT, Cassel CK, Yang JP, *et al. X*-linked recessive progressive combined variable immunodeficiency (Duncan's disease). Lancet. 1975;1:935-40.
2. Henter JI, Horne A, Aricó M, *et al*. HLH-2004: diagnostic and therapeutic guidelines for hemophagocytic lymphohistiocytosis. Pediatr Blood Cancer. 2007;48:124-31.
3. Seemayer TA, Gross TG, Egeler RM, *et al*. X-linked lymphoproliferative disease: twenty-five years after the discovery. Pediatr Res. 1995;38:471-8.
4. Booth C, Gilmour KC, Veys P, *et al. X*-linked lymphoproliferative disease due to *SAP/SH2D1A* deficiency: A multicenter study on the manifestations, management, and outcome of the disease. Blood. 2011;117:53-62.
5. Marsh RA, Bleesing JJ, Chandrakasan S, *et al.* Reduced intensity conditioning hematopoietic cell transplantation is an effective treatment for patients with SLAM-associated protein deficiency/X-linked lymphoproliferative disease type 1. Biol Blood Marrow Transplant. 2014;20:1641-5.

Perianal Abscess With Stellate Lacerations in a 3.5-year-old Previously Healthy Boy

Perianal abscesses are soft tissue infections of the perianal region and are common in infants [1-3]. Most of them are idiopathic, although there may be an association with congenital abnormalities of the crypts of Morgagni or an infection of the cryptoglobular glands [1-3]. They occur mainly in males, which may be due to androgen excess in cases of androgen-estrogen imbalance or to abnormal development of androgen-sensitive glands *in utero* [1]. In older children, the etiology shifts to underlying diseases, such as inflammatory bowel disease, immune deficiency syndromes, trauma, infected mass lesions and other immunodeficiencies [4]. The most common organisms isolated are mixed aerobic and anaerobic bacteria from gastrointestinal tract flora [5]. The appropriate management of perianal abscess is incision, drainage and antimicrobial treatment [3].

A 3.5-year-old boy presented with a three day history of pain, skin irritation and discharge of pus around the anus. Notably, fifteen days prior to admission, he developed an upper respiratory tract infection treated with oral second-generation cephalosprin. Five days later, while on antimicrobial treatment, he complained of pain during defecation and his mother noticed mild redness around the anus. The patient was afebrile. Laboratory investigations revealed severe neutropenia (absolute neutrophil count: 0.14×10^9/L). The patient was treated with topical corticosteroids, but showed no improvement. The child continued to complain of perianal pain and the inflammation worsened with purulent discharge. Three days prior to admission, he received oral metronidazole, without improvement.

Past medical history was unremarkable, and there was no history of constipation before or during the preceding viral illness. Physical examination demonstrated notable swelling, redness and tenderness in the rectum area with concomitant laceration of the anus leading to stool incontinence. His physical examination was otherwise unremarkable. A rectal examination revealed painful inflammation purulent discharge and stellate lacerations of the anal mucosa and skin (***Fig. 1a***).

Laboratory investigation upon admission revealed white blood cell count of 14.9×10^9/L (neutrophils: 35.4%, lymphocytes: 55.8%, monocytes: 8.3%) with normal hemoglobin and increased platelet count. Both C-reactive protein and ESR were mildly raised. Liver and renal function tests were normal. There was a family history of recurrent abscesses in mother and maternal aunt raising the suspicion of immunodeficiency disorder, but all immunological investigation came out to be normal including classes and subclasses of the immunoglobulins, immunophenotyping, dihydrorhodamine (DHR) test and cell adhesion molecules (CAMs). Physical examination findings also raised the possibility of sexual abuse, which was further ruled out after behavioral and psychological assessment of child and his parents. The family was daily reviewed by the pediatric team who found no evidence of child abuse or family conflicts. There was no evidence of any behavioral changes or psychological problems in the child during hospitalization and the follow-up consultation for the next 2 years showed no indication of psychological problems or any other changes in the behavior of the child.

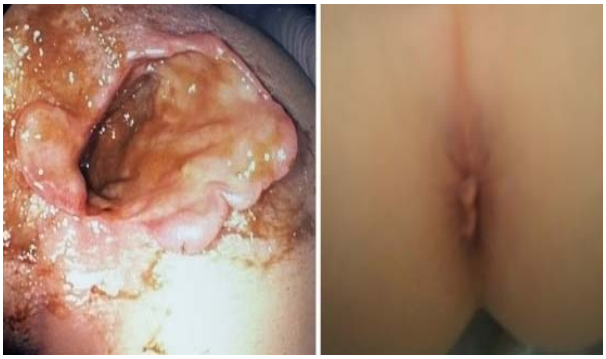

Fig. 1 *Perianal abscess of the 3.5-year-old child (a) on admission and (b) 10 days after treatment.*

Though the stool culture was negative, purulent discharge culture revealed *Klebsiella oxytoca* and *Proteus mirabilis* and the culture of perianal skin revealed *Klebsiella oxytoca, Enterobacter cloacae* and *Escherichia coli*. The patient was seronegative for HSV 1 and 2. Rectosigmoidoscopy was unremarkable and pathology did not reveal any evidence of underlying inflammatory bowel disease; colonic biopsy revealed only moderate alterations suggestive of active focal erosive rectitis. Additional investigations with polymerase chain-reaction in the blood, skin lesion and rectal tissue for Herpes viruses (HSV1, HSV2, VZV, EBV, CMV) were also negative. Empiric antimicrobial treatment with cefotaxime, clindamycin, metronidazole and acyclovir was initiated and continued for 14 days. The clinical course was favorable with complete clinical resolution (***Fig.* 1b**). In the follow-up period for next two years, child continued to remain well with no stool incontinence.

We report this unusual case because of the clinical presentation mimicking lesions associated with sexual abuse, as stellate lacerations were present. We elected to treat with broad spectrum antibiotics and provide antiviral treatment. The complete resolution of his lesions and anal incompetence was remarkable. Since the investigation did not identify any underlying disease, we concluded that the most likely pathogenetic cause was the development of severe neutropenia post viral infection. This highlights the importance of a complete blood count and a peripheral blood smear in the initial evaluation of perianal abscess upon presentation. Moreover, although his family history strongly suggested possible phagocytic dysfunction, the investigation failed to diagnose such an immune deficiency.

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REFERENCES

1. Fitzgerald RJ, Harding B, Ryan W. Fistula-in-ano in childhood: Congenital etiology. J Pediatr Surg. 1985; 20:80-1.
2. Chang HK, Ryu JG, Oh JT. Clinical characteristics and treatment of perianal abscess and fistula-in-ano in infants. J Pediatr Surg. 2010;45:1832-6.
3. Ezer SS, Oguzkurt P, Ince E, Hiçsönmez A. Perianal abscess and fistula-in-ano in children: Etiology, management and outcome. J Paediatr Child Health. 2010;46:92-5.
4. Whiteford MH. Perianal abscess/fistula disease. Clin Colon Rectal Surg. 2007;20:102-9.
5. Brook I, Frazier EH. The aerobic and anaerobic bacteriology of perirectal abscesses. J Clin Microbiol. 1997; 35:2974-6.

Early Onset Predominantly Diffuse Lung Disease in an Infant of Combined Methylmalonic Acidemia With Hyperhomocysteinemia Cobalamin C Type

Elevated blood methylmalonic acid (MMA) levels combined with elevated homocysteine is called combined methylmalonic acidemia with hyperhomocysteinemia [1,2]. It is found that MMA may damage the central nervous system, retina, liver, kidneys and blood cells. It also causes macular coloboma, thrombotic microangiopathy [3], and sometimes pulmonary arterial hypertension (PAH) [4,5], but an association between combined methylmalonic acidemia with hyperhomocysteinemia and diffuse lung disease (DLD) has rarely been reported in infants [6].

A 7-month-old boy was admitted with complaints of pallor for 30 days. It was followed by cough 8-10 days later. Personal history showed delayed motor development. The child was hospitalized in a local hospital for respiratory distress. Investigations showed white blood cell count of 9.78 $X10^9$/L, hemoglobin of 6 g/L, platelet count $319X10^9$/L, and reticulocytes of 9%. High resolution computed tomography (HRCT) scan of the lungs revealed diffuse lesions in both lungs. Cytomegalovirus DNA detection revealed $5.08x10^5$ copies/mL in sputum. Injection meropenem, azithromycin, voriconazole and ganciclovir were administered. In spite of the above treatment, child continued to have progressively worsening respiratory difficulty. He was intubated transferred to our hospital.

We added trimethoprim-sulfamethoxazole with a possibility of *Pneumocystis carinii* infection. Further investigations were non-contributory for bacterial, fungal and tuberculosis infection, and liver and renal function tests were