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Clinical Image

Trisomy 8 Associated Behçet's Like Disease

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Behçet's disease (BD) is an autoinflammatory disease of unknown etiology presenting by recurrent oral aphthous and genital ulcers, uveitis, and gastrointestinal lesions. The disease is characterized by a neutrophil dysfunction and overexpression of inflammatory cytokines triggered by exogenous factors in genetically susceptible individuals. The association of Behçet's like disease with hematologic disorders is rarely reported. The prevalence of hematologic malignancy ranges from 0.4% to 4% in a single-center cohort from Korea and China [1,2].

A 29-year-old male presented with a 3-year history of intermittent fever, abdominal pain, and perianal swelling. Laboratory tests showed a hemoglobin level of 8.6 g/dL, platelet count of 3.3×10^9/L, white blood cell count of 1.7×10^9/L, and neutrophil count of 850/μL. Computed tomography showed enhancing wall thickening with ulceration in the ileocecum and terminal ileum (Figure 1A). Endoscopy revealed few oval-shaped deep ulcers with well-demarcation in the ileocecal valve (Figure 1B) and perianal abscesses (Figure 2). Bone marrow biopsy revealed $<5\%$ cellularity, which is compatible with hypoplastic myelodysplastic syndrome (MDS)-EB-2. Chromosomal analysis showed 47 XY with trisomy 8 (Figure 3). The prevalence of trisomy 8 is reported to be 7%~9% among patients with MDS. The presence of trisomy 8 may play an important role in the upregulation of immunologic inflammatory genes, including transforming growth factor-β, interferon-β2, and receptors for interleukin-6 and -7. Recent studies have described the presence of various gastrointestinal manifestations with Behçet's-like disease in patients with MDS and trisomy 8 [3]. Administration of immunosuppressive agents is mostly not sufficient to control Bechet's like disease symptoms in MDS [4]. Although our patient showed temporary improvement of intestinal ulcer and perianal abscesses with infliximab, the lesions worsened after several months.

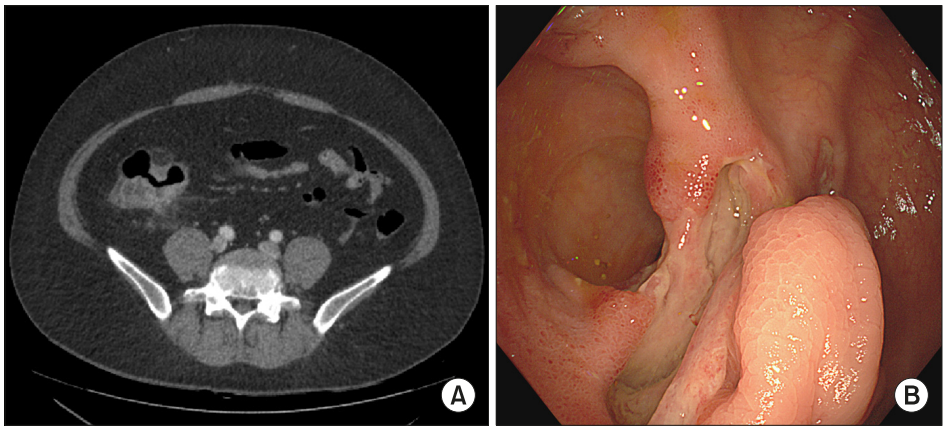

Figure 1. (A) Computed tomography finding showed layered enhancing wall thickening with perilesional fat infilteration and ulceration in terminal ileum. (B) Colonoscopic finding revealed large well defined deep oval ulcers in ileocecal valve.

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Treatment of the underlying hematologic condition should be considered [5]. We considered autologous hematopoietic stem cell transplantation for the treatment of refractory Behçet's-like disease with MDS. In patients presenting with Behçet's-like disease features and hematologic abnormalities, the possibility of chromosomal disorders such as trisomy 8 should be considered. Written informed consent was obtained from the patient who participated in this study.

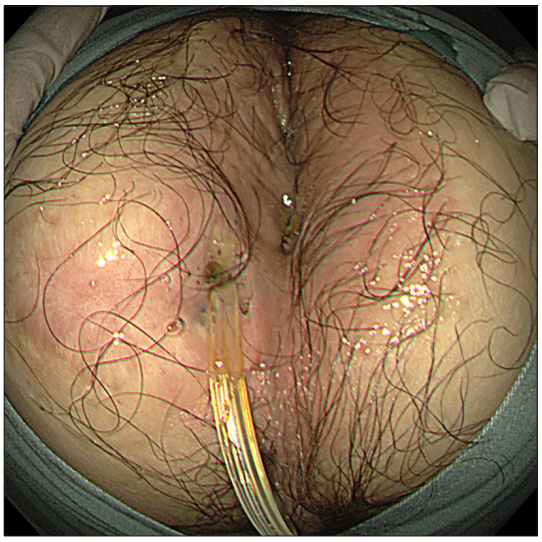

Figure 2. Perianal fistula was observed and seton tube drainage was performed.

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CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

AUTHOR CONTRIBUTIONS

Conceptualization: S.M.L., H.S.K. Data curation: W.H.C., K.A.L., S.R.J. Writing-original draft: S.M.L., J.S.K. Writing-review & editing: K.A.L., H.S.K.

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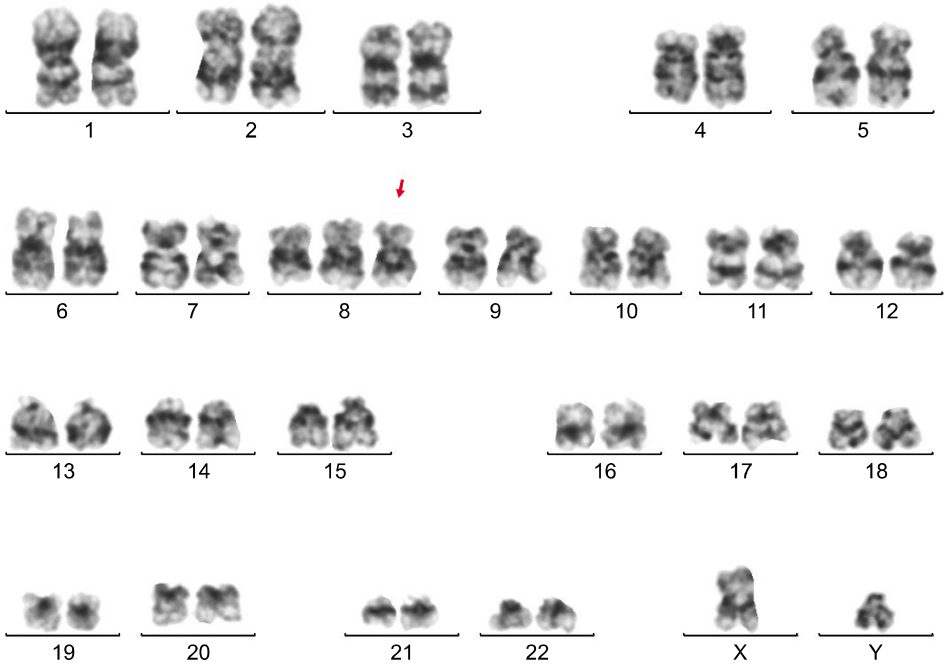

Figure 3. Karyotyping of this patient revealed trisomy 8 (arrow).

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