DOI: 10.16899/ctd.55801

Case Report / Olgu Sunusu

Behcet's disease in a patient with common variable immunodeficiency

Yaygın Değişken İmmun Yetmezlikli Bir Olguda Behçet Hastalığı

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Başvuru Tarihi/Received :
05-01-2015

Kabul Tarihi/Accepted:
27-02-2015

ABSTRACT

Common variable immunodeficiency (CVID) is one of the most common antibody deficiencies and about 20% of patients with CVID develop an autoimmune complication. Behçet's disease (BD) is a rare vasculitis diagnosed by the presence of recurrent oral and genital ulcers. Gastrointestinal involvement in Behçet's disease may be. Here, we report an uncommon case where a patient suffered from BD and CVID, together.

Keywords: Common variable immunodeficiency, Behcet's disease, autoimmunity, gastrointestinal involvement.

ÖZET

Yaygın değişken immun yetmezlik, yaygın yetersiz antikor yanıtı ile karakterize bir hastalıktır ve bu olguların yaklaşık %20'sinde otoimmün bir hastalık gelişebilir. Behçet hastalığı, ağızda ve genital bölgede tekrar eden ülserlerlerin birlikte bulunmasıyla teşhis edilen nadir bir vaskülittir. Behçet hastalığında gastrointestinal tutulum olabilir. Behçet hastalığı ve yaygın değişken immun yetmezlik hastalığının eşzamanlı birlikte seyreden bir olguyu sunmayı amaçladık.

Anahtar Kelimeler: Yaygın değişken immun yetmezlik, Behçet hastalığı, otoimmunite, gastrointestinal tutulum**.**

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INTRODUCTION

Common variable immunodeficiency (CVID) is one of the most common antibody deficiencies, is linked by a lack of immunoglobulin production, primary antibody failure and increased susceptibility to infection. However, about 20% of patients with CVID develop an autoimmune complication [1,2].

Behçet disease (BD) was first described in 1937 by Dr. Hulusi Behçet. Behçet's disease (BD) is a rare vasculitis with nopathognomonic test. The diagnosis of BH is based on clinical symptoms due to the lack of specific laboratory findings. In 1990, diagnostic criteria of BD were defined by the Working Group for Behçet's Disease [3]. According to the Working Group for Behçet's Disease**,** the patients with BD have recurrent oral aphthosis. At least two of the signs of recurrent genital ulcerations,eye symptoms and skin lesion addition to the recurrent oral aphthosis are required [3]. Diagnostic criteria of BD were proposed by the 2014 International Study Group from a multinational data from 27 countries. According to the International Criteria for BD (ICBD), ocular lesions, oral aphthosis and genital aphthosis are each assigned 2 points, while skin lesions, central nervous system involvement and vascular manifestations 1 point each. The pathergy test, when used, was assigned 1 point. A patient scoring ≥4 points is classified as having BD [4]. Althoug the cause of BD is unknown, it is likely due to an autoimmune reaction that is triggered by an infectious agent or other antigen in genetically predisposed individuals. Environmental and genetic factors can influence the incidence of autoimmune diseases. The prevalence of BD is high in Turkey, but low among Turkish patients in Germany. This may suggest an environmental trigger [5].

We present here an uncommon case who suffered from BD and CVID, together. This case is specific because of the two illnesses associated and only one case of an association of BD and CVID reported thus far.

Case presentation:

The patient was a 32 year-old female who admitted to the hematology department. General physical examination was had aphthous lesions of the oral mucosa and genital area. There were papulopustular lesions on back and legs of patients. In bilateral wrists of patient had pain. In medical history of the patient, she had recurret genital area lesions and oral aphthous lesions periodically. Arthritis of our patient had diagnosis. Colonoscopy showed aphthous lesions in all colonic segments. Aphthous lesions were seen in the patient's oropharynx by esophago-gastro-duodenoscopy. Pathology report is nonspecific, showing lymphocytic and neutrophilic infiltration. She had BD diagnosis based on this findings. Screening tests of serum immunoglobulins showed decreased concentrations of three types of immunoglobulins: Total IgG 3.65, IgA 0.42, IgM 0.25 g/L. Serum protein electrophoresis showed hipogammaglobuline with gammaglobuline fraction in 4.8% and albumin/globuline ratio 1.44. In lymphocyte immunophenotype examination, $CD3^+$ T cell ratio was 91.4%, $CD4^+/CD8^+$ 0.31, $CD19^+$ 5% and $CD16^+56^+$ 3.2%. The diagnosis was CVID. The patient received intravenously administered immunoglobulin (IVIG) and colchicine.

DISCUSSION

CVID is characterized by low serum levels of IgG, IgA and IgM, normal or decreased B cell numbers. Early diagnosis of antibody deficiency is important because delayed or inadequate treatment leads to more irreversible complications and increased mortality. CVID can present at any age. However, CVID is peak in childhood and early adulthood. The prevalence of CVID varied. In

the general population, it ranged from 1/500 to 1/500 000 [6]. IVIG as a replacement therapy is an effective way of management. Screening tests of serum immunoglobulins of our patient showed decreased concentrations of three types of immunoglobulins. Also, Serum protein electrophoresis showed hipogammaglobuline with gammaglobuline fraction in 4.8% and albumin/globuline ratio 1.44. We detected a reduction in both the number and function of antibodies in lymphocyte immunophenotype examination.

Autoimmune disorders occur with most frequently in CVID patients. Autoimmune complications may be the first manifestation of CVID in patients without severe infections. According to the Working Group for Behçet's Disease, our patient had BD diagnosis based on this findings [3]. BD, young adults between the second and fourth decades of life are mainly affected and it is equivalent between the sexes or more common in women in Western Europe and the USA. BD may influence the gastrointestinal tract. The involvement of the gastrointestinal tract is widely variable in different populations, being more common in Japan (50%–60%) and less common in the Mediterranean region, including Turkey (0%–5%). Intestinal symptoms appear 4-6 years after oral aphthous ulcer occurs. All patients develop mouth lesion and the disease may involve any part of the gastrointestinal tract from the mounth to the rectum. Esophageal involvement is quite uncommon and influence the middle part of the esophagus [7]. The most common colonoscopic findings are localized single or multiple ulcers in the ileocecal region, with only 4% having a diffuse distribution of lesions [5]. Tunc et al. [8] reported that gastrointestinal disease in BD is rare in Turkey (3%). Multiple superficial ulcers located predominantly in terminal ileum in Turkish patients differed from the single, large, deep ulcers with distinch borders described in the Far East [9]. Gastrointestinal tract effects of BD should be distinguished from the ulcerative colitis (UC) and Crohn's disease (CD). However, this may represent two diseases that coexist in the same patient. Behçet disease similar to Crohn's disease, BD manifests as discrete ulcers and discontinuous bowel involvement with relative sparing of the rectum. Unlike CD, a vasculitis of the small veins and venules with deep ulcerations characterizes BD, generally with no granulomas and cobblestoning, less inflammation surrounding the ulcer. Unlike UC, colonic BD consists of multiple aphthous ulcers with preservation of haustra and involvement primarily of the proximal colon and terminal ileum [5]. In our patient's oropharynx and all colonic segment were observed aphthous lesions with no granulomas and cobblestoning. She had BD diagnosis based on this findings.

In conclusion, CVID is a rare primary immunodeficiency disorder. Autoimmune disorders ocur with a higher incidence in CVID patients than in the general population. Also, in the CVID patients with gastrointestinal involvement should be considered Behcet's disease.

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