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Case Report

A rare autoimmune disorder – Behçet's disease

Bhandari Gurbir Singh, Jasleen Kaur

Departments of [1]Medicine, [2]Microbiology, Adesh Institute of Medical Sciences and Research, Bathinda, Punjab, India.

***Corresponding author:**
Dr. Bhandari Gurbir Singh,
Room No 2., Superspeciality block, Adesh Hospital,
Bathinda, Punjab, India.

bhandari.gurbir.singh@gmail.com

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ABSTRACT

Behçet's disease (BD) is a rare autoimmune disorder also classified as a variable vessel vasculitis which is characterized by recurrent oral and genital ulcerations, eye involvement, musculoskeletal symptoms, and other systemic features. Clinical presentation may vary from simple mucocutaneous manifestations to life-threatening pulmonary artery aneurysms and central nervous system involvement. The disease is much more severe in males as compared to females in contrast to all other autoimmune diseases where vice versa is true. Diagnosis is predominantly made by clinical presentation according to the International Criteria for BD. Early initiation of treatment is important as it can lead to significant morbidity and mortality if not treated. Glucocorticoids along with other immunosuppressants are the mainstay of treatment.

Keywords: Variable vessel vasculitis, Oral aphthae, Genital ulcerations and hypopyon uveitis, Immunosuppression

INTRODUCTION

Behçet's disease (BD) defined as the syndrome of the tri-symptom complex of oral aphthae, genital ulcerations, and hypopyon uveitis is one of the rarest autoimmune disorders.[1] Although it can affect every age group, the usual onset is in the third decade of life. It is characterized by the presence of

- Mucocutaneous manifestations (painful oral ulcers, genital ulcers, and erythema nodosum such as skin lesions and papulopustular lesions)
- Ophthalmological findings (hypopyon uveitis, retinal vasculitis, and visual loss)
- Central nervous system (parenchymal and vascular)
- Musculoskeletal system (inflammatory oligoarthritis)
- Vascular system (arterial and venous thrombosis)
- Respiratory system (pulmonary artery aneurysms).

BD has a strong genetic association with HLA-B 51 gene.[2]

The diagnosis is usually clinical along with other supportive investigations.

Treatment depends on the site of involvement with immunosuppressants being the mainstay of treatment.[3]

CASE REPORT

A 37-year-old male patient presented to us with complaints of painful recurrent erythematous, circular, and slightly raised areas evolving into oval or round ulcers within 48 h on the mucous membranes of the lips, gingiva, cheeks, and tongue which healed in about 15 days without

scarring. He also complained of genital ulcers on and around scrotum which healed with scars [Figure 1]. Mucosal ulcers were accompanied by inflammatory polyarthralgias involving shoulders, elbows, and knees with marked early morning stiffness.

On examination, the patient had oral ulcers and scrotal scars. Synovitis was present in the right knee joint. Ophthalmological examination was normal. Inflammatory markers (erythrocyte sedimentation rate and C-reactive protein) were raised. His HLA-B 51 was positive, CXR was normal, and X-ray knee joints were s/o secondary OA changes in the right knee joint. Pathergy test was positive.

The patient was managed with low-dose oral glucocorticoids, azathioprine, colchicine, and other supportive treatments.

DISCUSSION

BD is usually described as a systemic vasculitis that affects many organs. Skin and mucosal lesions, uveitis, major vessel disease, and musculoskeletal, neurological, and gastrointestinal manifestations are seen in varying combinations.[4] The diagnosis is clinical and a course characterized by remissions and exacerbations is typical. The intensity of the attacks decreases with the passage of time.

Painful oral ulcers are seen in 98% of patients and are often the first disease manifestation.

They resemble ordinary aphthae but are more frequent and multiple. Genital ulcers are usually located on the scrotum in men and on the major and minor labiae in women. Less common locations are the shaft and glans penis and the vaginal and cervical areas. Compared with oral ulcers, they are larger, deeper, less recurrent, and more resistant to healing. The pathergy phenomenon is a non-specific hyperreactivity in response to minor trauma. Although it has a moderate sensitivity (≈50%), it is highly specific for BD (>95%). It is usually used as a diagnostic test and is performed by inserting a 20 gauge needle into the dermis of the forearm of the patient. The presence of a papule or pustule at 48 h is considered positive.

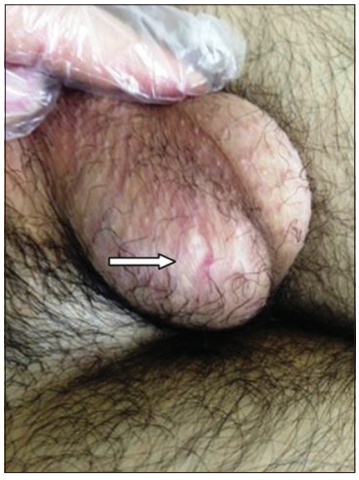

Figure 1: Depicting scrotal scar.

Eye involvement in BD causes serious morbidity and is a common cause of blindness in the Mediterranean Basin, the Middle East, and the Far East. It is found in 50% of patients, but the frequency reaches 70% in young men aged <25 years. It is usually seen during the first 2 years of the syndrome and is bilateral in 70–80% of patients. The most frequent pattern is a panuveitis not infrequently accompanied by a retinal vasculitis.

Neurological involvement is seen in 5% of patients with BD. It usually occurs after the first 5 years of disease and almost never as a presenting feature. Two main patterns of involvement are seen: Vascular and parenchymal. Parenchymal disease leads to inflammatory lesions in the brain stem, diencephalon, basal ganglia, and, less frequently, the spinal cord and cerebellum. The second pattern of neurological involvement is dural sinus thrombosis, mainly characterized by headache and papilledema. It is usually associated with deep vein thrombi in other areas and has a better prognosis than parenchymal involvement.[5]

BD affects both arterial and venous large vessels. Major vessel involvement is more frequently seen in men and overall is found in up to 40–50% of patients; 60–80% of these lesions are venous thromboses of the deep veins of the lower extremities. Systemic artery involvement may be seen in 1.5–7.5% of patients. The typical lesions are aneurysmal, or uncommonly, occlusive. The abdominal aorta is the most frequently affected site followed by the iliac, femoral, popliteal, carotid, and subclavian vessels. Pulmonary arterial aneurysms are the most devastating arterial involvement in BD.[6] They are usually seen in the young male patient, who often has thrombosis of the leg veins or the vena cava, and are manifested by recurrent hemoptysis, dyspnea, and pleuritic chest pain.

Management depends on the system involved. Mucocutaneous manifestations respond well to topical glucocorticoids and colchicine and, in refractory cases, require immunosuppression. In most patients with BD, arthritis can be managed with colchicine. If the patient has severe eye disease defined as >2 lines of drop in visual acuity on a 10/10 scale and/or retinal disease (retinal vasculitis or macular involvement), it is recommended that either cyclosporine A or infliximab be used in combination with azathioprine and glucocorticoids; alternatively, interferon alpha with glucocorticoids may be used. For the management of acute deep vein thrombosis in BD, immunosuppressive agents such as glucocorticoids, azathioprine, cyclophosphamide (CYC), or cyclosporine A are recommended. For the management of pulmonary and peripheral arterial aneurysms, CYC and glucocorticoids are recommended.[7,8]

CONCLUSION

BD is a very rare disease with varied clinical presentations which is most often misdiagnosed. Prompt diagnosis and early initiation of immunosuppressants is very important.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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