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Clinical Case Reports

Eye as the First Clue: Behçet's Disease Unveiled by Retinal Vasculitis and Vein Occlusion

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Abstract

Summary: Behçet's disease is a chronic and recurrent multisystem vasculitis of unknown aetiology. Ocular inflammation is one of the most severe and vision-threatening complications. We present a case of severe ocular Behçet's disease with occlusive retinal vasculitis in a young man. A 20-year-old boy had gradually deteriorating vision in both eyes for three months before experiencing a sudden loss of vision in the left eye for two weeks. Ocular examination showed bilateral anterior uveitis with vitritis and perivasculitis. Branch retinal vein occlusion (BRVO) with macular oedema and a macular pseudohole was observed in the left eye, which was confirmed by fundus fluorescein angiography and optical coherence tomography. A positive pathergy test and repeated oral and vaginal ulcers confirmed the diagnosis of Behçet's syndrome with panuveitis and occlusive vasculitis. Adalimumab, azathioprine, and systemic corticosteroids were used to treat the patient, which led to partial recovery of vision and a noticeable reduction in inflammation. Early detection of Behçet's uveitis and timely initiation of immunomodulatory therapy are essential to prevent irreversible retinal damage and subsequent visual loss.

Keywords: Behçet's Disease, Retinal Vasculitis, Branch Retinal Vein Occlusion, Macular Edema, Adalimumab, Uveitis.

Introduction

Behçet's disease (BD) is a chronic, relapsing vasculitis of uncertain aetiology that affects many organs. The disease involves a triad of recurrent oral aphthous and genital ulcers, skin lesions, and ocular inflammation.[1] It is most prevalent along the ancient Silk Road region, with higher rates in Turkey, the Middle East, and East Asia.[1] The condition primarily affects young adults, mainly boys, and can cause considerable morbidity due to vision loss when ocular involvement develops.[1,2]

Behçet's uveitis is usually bilateral, recurring, and non-granulomatous, with a relapsing-remitting history.[3] Ocular symptoms appear in up to 70-80% of patients and frequently affect the disease outcome.[4] The most frequent ocular manifestations are pan-uveitis with retinal vasculitis, which can cause vascular occlusions along with macular oedema, which is cystoid in nature. Anterior uveitis in Behçet's is non-granulomatous with mobile hypopyon.[4,5] On fluorescein angiography, BD uveitis exhibits fern-pattern leakage, superficial haemorrhages, and occlusive periphlebitis, in contrast to sarcoid or tuberculosis-associated uveitis.[5,6]

The International Criteria for Behçet's Disease (ICBD) combine ocular signs with systemic features such as recurring oral mucosal and genital ulcers, erythema nodosum, papulopustular skin lesions, and a positive pathergy test, which serve as the basis for the predominantly clinical diagnosis.[1,7] For accurate diagnosis and treatment of non-granulomatous uveitis, other differentials need to be ruled out, such as sarcoidosis, reactive arthritis, systemic lupus erythematosus, inflammatory bowel disease, or herpes infections.[8]

Case Presentation

A 20-year-old man presented at OPD with gradual, painless blurring of vision in both eyes for three months, followed by an abrupt onset of vision loss in the left eye. He stated that for the past four years, he had suffered from excruciating recurrent genital and oral ulcers, each of which healed on its own but left scars. He had intermittent erythematous papulopustular skin lesions but no history of fever, joint pain, backache, cough, weight loss, or ocular injury.

On examination, the best-corrected visual acuity (BCVA) was 6/12 in the right eye and finger counting at 1 m in the left eye. The anterior chambers of both eyes showed 2+ cells with mild flare and bilateral vitritis (2+), with more pronounced haze in the left eye. Fundus examination revealed temporal mild disc pallor, perivasculitis, scattered haemorrhages in all quadrants, ghost vessels inferonasally, and a macular pseudohole in the left eye.

Systemic evaluation revealed a history of recurrent oral and genital ulcers and erythematous papulopustular skin lesions.

Investigations

Ocular Investigations

Optical coherence tomography (OCT) confirmed macular oedema in the left eye. Fundus Fluorescein Angiography (FFA) showed fern-pattern leakage and areas of capillary non-perfusion consistent with occlusive vasculitis and BRVO.

Dermatographia may be seen in some patients with Behçet's disease as a sign of skin hyperreactivity, but it is nonspecific. The pathergy test remains a relevant diagnostic skin test for Behçet's disease.

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Systemic Investigations

Parameter	Findings
ESR	**62 mm/hr.** (elevated)
CBC, LFTs, RFTs	Within normal limits
Serum ACE	13 U/L (normal 9–67 U/L)
Serum Calcium	9.9 mg/dL
Pathergy Test	**Positive** (pustule formation at 48 hours)
HLA-B51	**Positive**
Chest X-ray/HRCT	No hilar lymphadenopathy or pulmonary lesions
ANA, ANCA, Infectious panel (TB, syphilis, HSV)	Negative

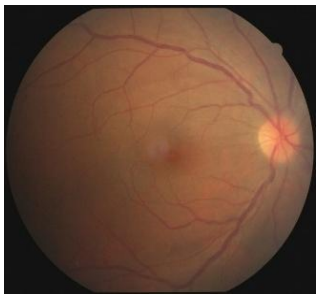

Figure 1: Fundus Photograph showing hazy fundal view in the Right eye and temporal disc pallor with occlusive perivasculitis, and a macular pseudohole in the left eye

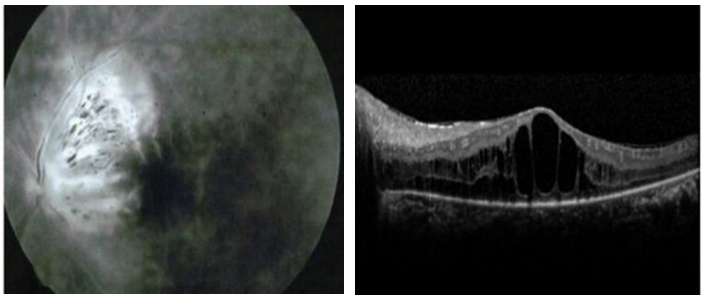

Figure 2: FFA image of the Late phase showing fern pattern vasculitis in the Left eye and OCT of the Left eye showing macular edema

ICBD Classification Criteria Fulfillment

Diagnosis was made clinically using the International Criteria for Behçet's Disease (ICBD) criteria.

According to the International Criteria for Behçet's Disease (ICBD), the patient met the diagnostic threshold:

- Recurrent oral ulcer history (2 points)
- Genital ulcer history (2 points)
- Ocular lesions (uveitis + retinal vasculitis) (2 points)
- Positive pathergy test (1 point)

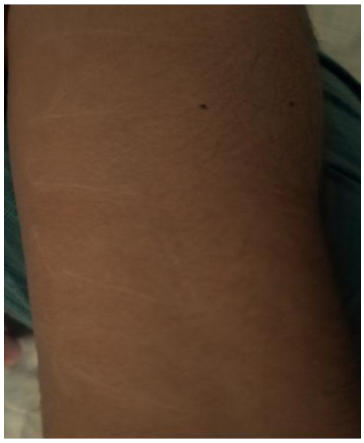

Figure 3: Dermatographia

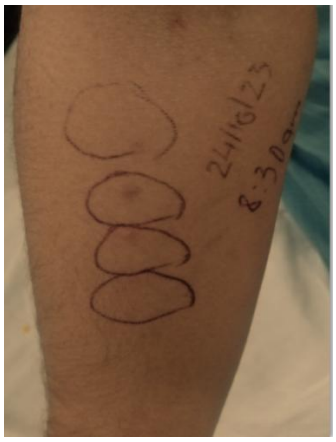

Figure 4: Positive Pathergy test

A total score of **7 points** exceeds the ≥4 requirement, confirming the diagnosis of Behçet's disease.

The uniqueness of this case lies in the unusually early and severe posterior segment involvement, including BRVO and macular pseudoholes, as the first major presenting ocular feature.

These results excluded other granulomatous and autoimmune diseases, including sarcoidosis, tuberculosis, systemic lupus erythematosus (SLE), and inflammatory bowel disease (IBD).

Differential Diagnosis

1. Inflammatory Bowel Disease (IBD)

IBD can present with oral ulcers and uveitis; however, this patient had no gastrointestinal symptoms or colonoscopic evidence of IBD. The ocular pattern—occlusive retinal periphlebitis with BRVO and macular oedema —is more typical of Behçet's disease, as IBD rarely causes occlusive posterior vasculitis.

2. Systemic Lupus Erythematosus (SLE)

SLE may cause retinal vasculitis and oral ulcers, but this patient had no systemic features (photosensitivity, rash, serositis, renal disease) and a negative ANA test. The presence of genital ulcers and a positive pathergy test further favoured a diagnosis of Behçet's disease over lupus.

3. Seronegative / Reactive Arthritis
can present with mucocutaneous lesions and uveitis, but posterior occlusive vasculitis and BRVO are rare. The absence of arthritis, prior infection, and the presence of recurrent oral/genital ulcers with positive pathergy ruled out reactive arthritis.
4. Herpetic Infections (HSV/VZV)
Herpetic retinitis is usually unilateral, necrotising, and rapidly progressive and is characterised by retinal necrosis, haemorrhage, and retinal detachment. Bilateral occlusive periphlebitis without necrosis and the absence of corneal or serological evidence excluded a viral aetiology.
5. Sarcoidosis
Sarcoidosis causes granulomatous uveitis with hilar lymphadenopathy and elevated ACE levels. This patient had non-granulomatous uveitis, normal imaging findings, and normal ACE levels, making sarcoidosis unlikely.
6. Other Systemic Vasculitides (e.g., Granulomatosis with Polyangiitis, Polyarteritis Nodosa)
typically cause systemic involvement (renal, ENT, or neurologic) with positive serology, such as ANCA. The absence of multisystem disease and mucocutaneous ulceration pointed toward Behçet's disease.
7. Relapsing Polychondritis
Characterized by auricular and nasal cartilage inflammation with audio vestibular symptoms, which were not present in this case.
8. Multiple Sclerosis (MS)
MS can present with intermediate uveitis but lacks oral/genital ulcers, pathergy positivity, or occlusive retinal vasculitis. Normal neuroimaging findings further excluded MS.

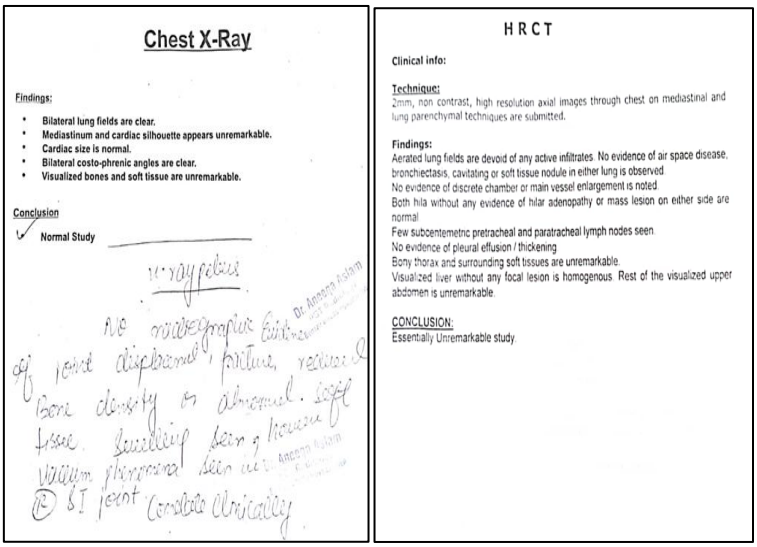

Figure 5: Normal chest X-ray and HRCT

Diagnosis
Based on the clinical findings and systemic manifestations, a diagnosis of Behçet's disease with severe ocular involvement (pan-uveitis with occlusive vasculitis) was made.
Treatment
The patient was started on oral prednisolone (1 mg/kg/day) for three weeks, followed by gradual tapering. Azathioprine 100 mg twice daily was initiated as a steroid-sparing agent.[4,9] Owing to severe posterior segment inflammation, adalimumab was added (loading dose 80 mg subcutaneously, followed by 40 mg every other week).[1,10] Supportive therapy included omeprazole, folic acid, and calcium with vitamin D3 supplementation.
Outcome And Follow-Up
At one month, ocular inflammation had markedly subsided, with clearing of the vitreous haze. At three months, OCT demonstrated the resolution of macular oedema. The final BCVA was 6/6 and 6/18 in the right and left eyes, respectively. The patient remains under close follow-up on maintenance immunomodulatory therapy with adalimumab 40 mg administered subcutaneously every two weeks, planned for 18–24 months to maintain remission and prevent ocular relapses.[9,10]

Discussion

Behçet's disease is a systemic vasculitis that affects arteries and veins of all calibres, with ocular involvement being the most serious and vision-threatening component.[1,2] Ocular Behçet's typically presents as bilateral, recurrent, non-granulomatous uveitis, most often involving the posterior segment.[3,4] Retinal vasculitis, particularly occlusive periphlebitis, is the hallmark feature observed in up to 85% of cases.[6] Recurrent inflammatory episodes can cause retinal ischaemia, macular oedema, and optic atrophy, and the disease has a relapsing-remitting course.[1]
Our patient demonstrated the characteristic ocular features of vitritis and occlusive retinal vasculitis with perivenular sheathing, along with recurrent oral and genital ulcers and a positive pathergy test. The presence of branch retinal vein occlusion (BRVO) and macular pseudoholes indicates severe vascular inflammation and ischaemic damage. A diagnostic gap exists because early ocular Behçet's disease can mimic several inflammatory and infectious conditions. Retinal periphlebitis and BRVO may be misattributed to other vasculitides, delaying the initiation of immunomodulatory treatment. This case reinforces the need to recognise ocular clues that point toward Behçet's disease before systemic manifestations become prominent.[3-6]

Differential diagnoses include infectious and non-infectious uveitic entities such as sarcoidosis, SLE, systemic lupus erythematosus, multiple sclerosis, and inflammatory bowel disease.[8] However, these were excluded based on the clinical presentation and relevant investigations. Behçet's disease remains a clinical diagnosis supported by systemic manifestations and the exclusion of conditions that mimic it. [1,7]

The management of Behçet's uveitis depends on the severity of the disease. Acute attacks require prompt high-dose corticosteroids, either oral or intravenous methylprednisolone (1 g/day for 3 days in severe inflammation), to control acute inflammation.[9,10] To prevent relapses and minimise corticosteroid dependency, long-term immunosuppressive agents such as azathioprine, cyclosporine, and methotrexate are recommended.[2,10] Biologic medicines, especially anti-tumour necrosis factor (TNF) treatments such as infliximab and adalimumab, have shown notable effectiveness in managing severe or resistant posterior segment illness. These agents have demonstrated efficacy in reducing inflammation, clearing macular oedema, and lowering the likelihood of relapse.[4,9]

Up to 25% of patients with ocular Behçet's disease may suffer from serious sight loss despite vigorous therapy, particularly young boys with early onset posterior illness.[3,5,6] Immunomodulatory treatment must be initiated as soon as possible to avoid irreparable vascular blockage and optic nerve injury. Long-term disease activity tracking and treatment response, along with therapy-associated side effects, are crucial. When prompt and appropriate therapy is initiated, approximately 60% of patients experience remission after the first several years.

Unlike typical Behçet's disease, which usually begins with recurrent oral/genital ulcers followed by anterior uveitis, this case is noteworthy because Behçet's disease rarely presents initially with severe occlusive vasculitis and branch retinal vein occlusion (BRVO) in young adults. The presence of a macular pseudohole further reflects significant ischaemic damage at first presentation, a combination infrequently reported in the literature.[3,10] Early detection of this aggressive ocular phenotype is clinically important because it necessitates rapid escalation to biological therapy. This case is significant because it illustrates the importance of considering Behçet's disease in young patients presenting with sudden retinal vein occlusion and occlusive vasculitis. Early recognition allows for the timely initiation of immunosuppressive and biological therapy, which is critical for preventing irreversible visual loss.

Young individuals presenting with recurrent oral/genital ulcers and ocular inflammation should be suspected to have Behçet's disease. Occlusive retinal vasculitis, macular oedema, and recurrent uveitis are the hallmarks of Ocular Behçet's disease, a unique and potentially blinding condition. To salvage vision, systemic corticosteroids and immunomodulatory medications must be initiated as soon as possible. Biologics may be used when these treatments are ineffective. Routine ocular and systemic follow-up is required to track disease activity and treatment-related side effects.

Conclusion

Behçet's uveitis primarily affects the posterior segment of the eye and is bilateral, recurrent, and non-granulomatous. Retinal vasculitis and vein occlusion are two common disorders that can result in severe vision loss. Biological treatment and early immunosuppression are essential for maintaining vision. Before establishing BD, it is important to rule out mimics, including lupus, sarcoidosis, and tuberculosis. The best results are guaranteed by multidisciplinary management, which includes dermatology, rheumatology, and ophthalmology management. Regular OCT and FFA monitoring helps in the early identification of recurrences.

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