

Eyes-A Window to Systemic Disease: A Case Report of an Atypical Ocular Presentation of Chronic Myeloid Leukemia

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Abstract

Ocular manifestations may serve as the primary indicator of underlying hematological malignancies. We report the case of a 23-year-old male who presented with sudden visual loss and was subsequently diagnosed with Chronic Myeloid Leukemia (CML) following characteristic fundus findings. This case emphasizes the crucial role of the ophthalmologist in detecting life-threatening systemic disease and demonstrates how retinal findings can mirror systemic disease activity and response to treatment.

Keywords: Chronic Myeloid Leukemia (CML); Leukemic retinopathy; Roth spots; Flame-shaped hemorrhages; BCR-ABL1

Introduction

Leukemia can involve the eye either through direct leukemic infiltration or secondary hematological abnormalities such as anemia, thrombocytopenia, and hyperviscosity. Common retinal manifestations include hemorrhages, Roth spots, cotton wool spots, and venous tortuosity. In several cases, ocular signs may precede systemic symptoms, making fundus examination a critical diagnostic window to systemic disease [1]. We describe a case in which bilateral retinal findings led to the diagnosis of CML.

Case Presentation

A 23-year-old male smoker presented with sudden, painless diminution of vision in both eyes. There was no significant past medical history.

Visual Acuity: BCVA was 6/9 in the right eye and 6/36 in the left eye.

Anterior Segment: Unremarkable bilaterally.

Fundus Examination:

Both eyes showed markedly dilated and tortuous retinal veins with pink-colored blood columns suggestive of leukostasis. Multiple flame-shaped hemorrhages and Roth spots were scattered over the posterior pole. The left eye additionally demonstrated a prominent sub-internal limiting membrane (sub-ILM) hemorrhage at the macula, correlating with reduced visual acuity [2].

Diagnostic workup

Based on the characteristic appearance of leukemic retinopathy, an urgent systemic evaluation was advised. Complete blood count revealed a grossly deranged profile with marked leukocytosis (Figure 1). The patient was referred to hematology services. Molecular testing using Sanger sequencing detected a BCR-ABL1 gene mutation, confirming the diagnosis of Chronic Myeloid Leukemia [3].

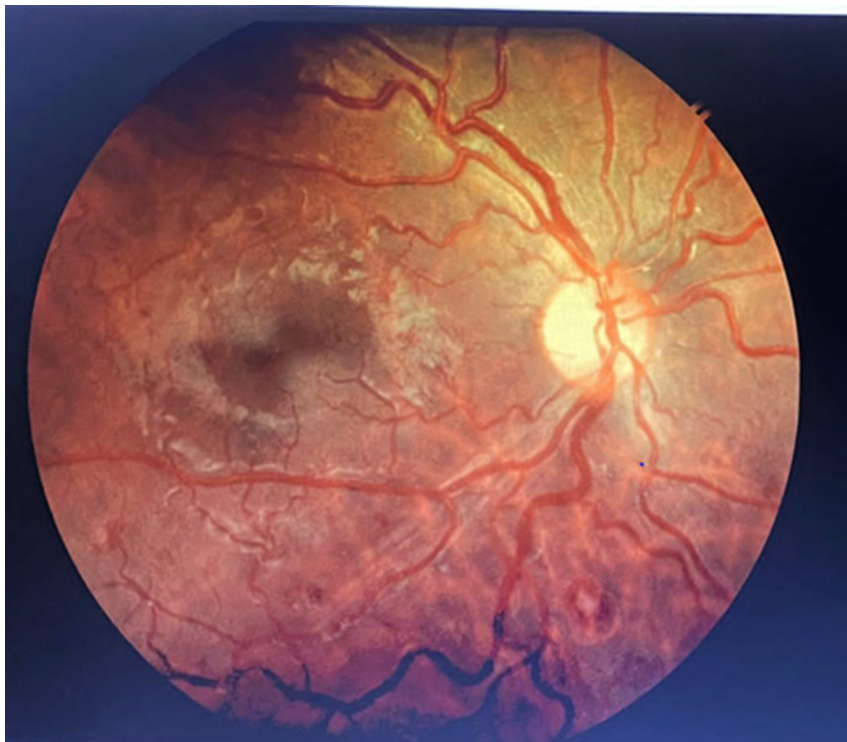


Figure 1: Right eye fundus photograph at six weeks showing significant resolution of flame-shaped hemorrhages and Roth spots.

Follow-up and outcome

The patient was initiated on Imatinib therapy. At 10-week follow-up, BCVA improved to 6/6 in both eyes with near-complete resolution of retinal hemorrhages and Roth spots. At four months, the patient reported recurrent blurring of vision (BCVA

6/9). Fundus examination revealed reappearance of Roth spots. Concurrent hematological evaluation demonstrated worsening blood parameters along with gastrointestinal symptoms, indicating systemic disease flare and reaffirming the correlation between ocular and systemic disease activity (Figure 2).

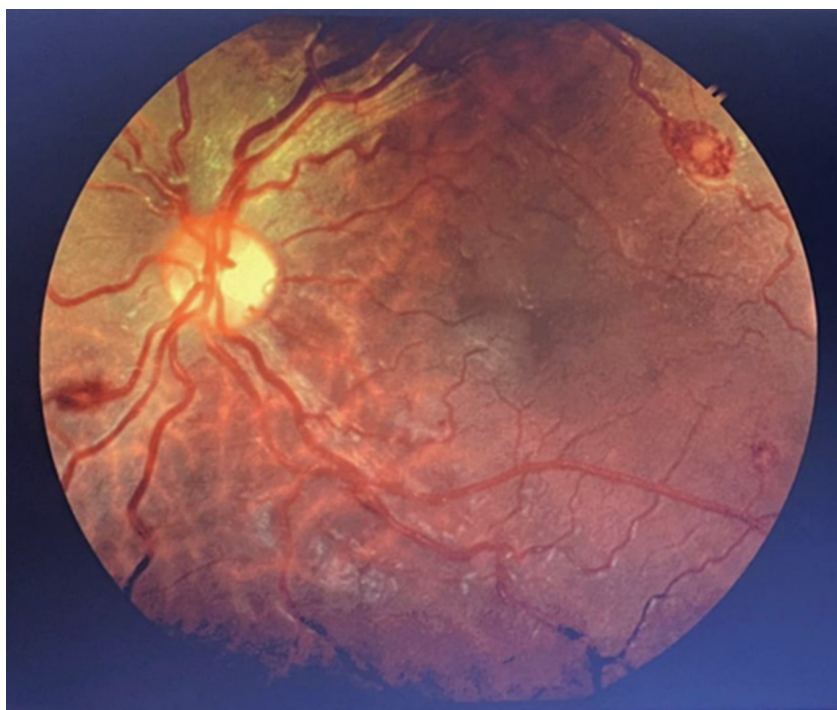


Figure 2: Left eye fundus photograph at six weeks showing resolution of sub-ILM hemorrhage and retinal lesions.

Discussion

Leukemic retinopathy reflects underlying hematological abnormalities and is commonly associated with high disease burden. While Roth spots are not pathognomonic, their presence alongside venous tortuosity and abnormal blood column coloration strongly suggests blood dyscrasia. This case is noteworthy because ocular symptoms were the sole reason the patient sought medical attention, leading to early diagnosis of a life-threatening malignancy. Furthermore, recurrence of retinal findings during follow-up acted as a visual biomarker of systemic disease activity. Although the presentation appeared atypical due to sudden ocular onset, molecular confirmation of BCR-ABL1 established a definitive diagnosis of CML.

Conclusion

Meticulous fundus examination can facilitate early detection of serious systemic disease. In patients with leukemia, periodic ophthalmic evaluation not only aids in preserving vision but may also provide valuable insight into systemic disease status and treatment response.

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