Visual Disturbance as Presenting Feature and Macular Hole as Complication of Chronic Myeloid Leukemia

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Abstract

A 42-year old male presented with diminution of vision in left eye, weakness, and lethargy. On fundus examination (with +90 D lens), there was paramacular thickening with central macular and peripheral hemorrhages in left eye. Macular thickening was confirmed by Optical Coherence Tomography (OCT). Blood film demonstrated leukocytes are with the presence of precursor cells of the myeloid lineage. Fluorescence in Situ Hybridization (FISH) revealed the presence of Philadelphia chromosome in 48% of inter phase cells studied. Patient was put on treatment with Imatinib tablet 400mg twice daily and treatment monitored. In the treatment phase, lamellar macular hole developed in the left eye at nine months. This case is being reported because of its unusual presentation and complication (Lamellar Macular Hole).

Keywords: Chronic myeloid leukemia; Lamellar macular hole; Visual disturbance

Introduction

Chronic Myelogenous Leukemia (CML) in blast phase rarely involve extramedullary sites like the skin and eyes [1,2]. Retina is the commonly involved ocular tissue with hemorrhages, sheathing, vascular tortuosity, and cotton wool spots in the chronic phase; however, visual disturbance may rarely be the presenting feature of CML [3,4].

Case Presentation

A lamellar macular hole complicating leukemic infiltration of sub-retinal tissues has never been reported. A 42-year old male presented to the out-patient department with painless and gradual diminution of vision, weakness, and fatigue for 2 months. His Corrected Distance Visual Acuity (CDVA) in right eye was 6/9 and in the left eye was 1/60. Slit lamp examination with a +90D lens showed flame-shaped hemorrhages, periphlebitis and few cotton wool spots were seen in right eye (Figure 1a). In the left eye, there was a central macular hemorrhage surrounded by sub-retinal leukemic infiltration, thickening along with scattered dot and blot hemorrhages (Figure 1b). Increased macular thickness in the left eye was confirmed on optical coherence tonography (Figure 2a). The white blood cell count was 1.4 x 10^9/ mm^3 and hemoglobin was 9.4 mg/dl. The blood film demonstrated the presence of precursor cells of the myeloid lineage (Figure 2c). Chromosomal analysis with karyotyping of peripheral blood specimen was performed; all 20 metaphases analyzed were abnormal and showed t(9;22) which were consistent with CML (Figure 2d). Specimen was again studied for Philadelphia chromosome (t (9; 22) q34; q11) by Fluorescence in Situ Hybridization (FISH); 200 cells were studied using dual fusion DNA probes. Philadelphia chromosome was present in 48% of inter phase cells studied. The patient was referred to the regional cancer institute and was put on treatment with Imatinib tablets 400mg twice daily and treatment monitored. In the treatment phase, lamellar macular hole developed in the left eye at nine months. This case is being reported because of its unusual presentation and complication (Lamellar Macular Hole).

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Discussion

Approximately, 40% patients with CML are asymptomatic, diagnosed on the basis of abnormal counts and most patients present in the chronic phase (range 85-97%). The median age at presentation in India is a decade younger as compared to western countries (range 32-42 years) [5,6]. Splenomegaly is the most commonly reported symptom, followed by hepatomegaly, weakness and fatigue; in the present study, sudden diminution of vision was the presenting symptom, due to macular hemorrhage. The treatment response to Imatinib is variable due to population heterogenicity and about 91-97% patients achieve Complete Hematological Response (CHR) and 20-93% Complete Cytogenetic Response (CCyR) [6]. Skin pigmentation, anemia and thrombocytopenia have been the commonly reported complications secondary to Imatinib therapy. Lamellar macular hole as a complication of CML has not been reported. The probable cause was vitreo-macular traction on the macula by the fibrous band in the vitreous; these bands often form due to fibrovascular activity in the retina following hemorrhages [7].

References