CASE REPORT

Central retinal artery occlusion as a presenting manifestation of polycythaemia vera

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SUMMARY

A 60-year-old woman with no premorbidities presented with symptoms of sudden painless vision loss in the left eye (LE). Best-corrected visual acuity in the LE was counting fingers close to face. A relative afferent pupillary defect was observed in the LE. Ocular fundus examination of LE was suggestive of central retinal artery occlusion. Systemic evaluation revealed splenomegaly and normal cardiac and carotid arteries. Haematological investigations revealed increased haemoglobin, haematocrit, platelet count and leucocytosis with low erythrocyte sedimentation rate (ESR). Features of myeloproliferative neoplasm were noted on bone marrow aspiration. An assay for *JAK2* mutation was positive. Since erythropoietin levels were normal, a diagnosis of primary polycythaemia vera was made and treated with aspirin and phlebotomy twice weekly until the target haematocrit of under 45% was achieved.

BACKGROUND

Polycythaemia vera (PV) is a clonal disorder characterised by erythrocytosis, leucocytosis and thrombocytosis. Patients with PV are prone to haemorrhagic and thrombotic events which contribute to morbidity and mortality from the disease. Ocular involvement is rarely seen in the form of bilateral retinal haemorrhages, peripheral non-perfusion, retinal vein occlusion, retinal artery occlusion and visual defects secondary to cerebral non-perfusion. Increased adhesiveness of the erythrocytes to the vascular endothelium causes thrombus formation and arterial occlusion. PV should be considered to be a possible aetiology in the evaluation of a patient presenting with CRAO.

CASE PRESENTATION

A female patient aged 60 years presented to us with sudden onset of painless vision loss of 1 day duration in the left eye (LE). The best-corrected visual acuity was 6/9, N10 in the right eye (RE) and counting finger close to face (CFCF), <N36 in LE. Slit lamp biomicroscopy and intraocular pressure were within normal limits except for the presence of relative afferent pupillary defect (RAPD) in the LE. Left fundus examination of LE showed retinal whitening, cherry red spot at the macula and narrowed arterioles (figure 1) consistent with a diagnosis of CRAO. RE fundus was normal.

Systemic examination revealed palmar erythema and a palpable spleen (5 cm below the left costal margin). Cardiovascular system and carotid arterial examinations were normal.

INVESTIGATIONS

Fundus fluorescein angiogram showed delayed arterial filling consistent with the diagnosis of CRAO.

Haematological investigations were significant for the following features: the total leucocyte count 45 000/mm³, haemoglobin 15.9 g% with a haematocrit of 55%; total platelet count was 75 700/mm³. Peripheral smear showed an increased number of erythrocytes, neutrophils and giant platelets, suggestive of a myeloproliferative disorder (figure 2).

Bone marrow trephine biopsy showed erythroid hyperplasia (colonies) in the marrow with mild dysplasia of myeloid elements (figure 3) and dysplastic megakaryocytes with clustering and paratrabecular localisation (figure 4) suggestive of PV.

The hybrid transcript for *BCR-ABL* was not detected in the lymphocytes of the blood specimen but was positive for *JAK2* mutation. Serum erythropoietin levels were within normal limits.

Ultrasound of the abdomen and pelvis showed mild hepatomegaly and splenomegaly.

On the basis of the diagnostic workup, laboratory evaluation and consultation with haematologist, the diagnosis of primary PV with central retinal artery occlusion (CRAO) was made.

TREATMENT

Immediate treatment with low dose aspirin was initiated, and regular phlebotomies were performed twice weekly until the haematocrit dropped to 45%.

The poor prognosis for visual recovery in the LE was explained to the patient. She was counselled regarding regular follow-up for the RE.

Figure 1 Fundus photo of the LE showing retinal whitening, cherry red spot at the macula and narrowed arterioles. LE, left eye.

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Figure 2 Peripheral smear showing crowded erythrocytes (Leishman; ×100).

Figure 3 Bone marrow trephine biopsy showing erythroid hyperplasia (colonies) in the marrow with mild dysplasia of myeloid elements (Leishman; ×200).

OUTCOME AND FOLLOW-UP

The patient was followed up at regular intervals as the chance of developing CRAO in the RE is high. Her haematocrit persisted to be below the target level of 45% throughout the follow-up.

DISCUSSION

PV, also known as polycythaemia rubra vera, a chronic clonal disorder is generally characterised by an increase in the red cell mass along with normal arterial oxygen saturation, thrombocytosis, leucocytosis and splenomegaly.¹ The incidence of PV is reported to be 0.8–2.6 in 100 000 persons.² Higher whole blood viscosity, resulting in reduced red cell deformability and abnormal red cell adhesion, especially in the *JAK2* mutation patients, is responsible for vascular occlusion in patients with PV.³ Venous thrombosis occurs in about 40% of the individuals (involving mesenteric or hepatic vessels) and are more fatal than arterial thromboses.⁴ Bleeding, usually gastrointestinal, though seen, is less common than thrombosis.

Ocular symptoms of PV are either due to hyperviscosity or thrombosis.⁵ Reported ocular manifestations include bilateral simultaneous central retinal vein occlusion, dilated prominent conjunctival vessels,⁶ transient vaso-occlusive disease, retinal

Figure 4 Bone marrow trephine biopsy showing dysplastic megakaryocytes with clustering and paratrabecular localisation (Leishman; ×400).

haemorrhages and monocular blindness.^{6 7} A case of patient with *JAK2* mutation who presented with CRAO and ipsilateral middle cerebral artery stroke has also been reported.⁸

When untreated, polycythaemia can be life threatening due to cardiovascular thrombotic events, or transformation of PV to myelofibrosis, leukaemia or myelodysplastic syndromes. Management of PV is aimed at prevention of thrombotic complications and maintaining normal haematocrit level by periodic phlebotomies. Thrombocytosis can be controlled by hydroxyurea, interferon- α , anagrelide or busulfan.¹

In summary, patients presenting with features of CRAO require a thorough systemic evaluation and complete haematological workup. As seen in our patient, uncommon conditions such as PV may rarely present to the ophthalmologist first, with ocular events preceding the systemic manifestations. In the present case, potential life-threatening systemic complications of PV were averted due to the prompt diagnosis and treatment. This case highlights the importance of thorough systemic and haematological workup in patients with CRAO.

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