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Rare Case of Jak 2 Positive Polycythemia Vera Presenting As Transient Loss of Vision: A Case Report

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ABSTRACT

Background: Polycythemia vera (PV) is a lesser-known condition with high variability of disease presentation. This case report sheds some light on a rare diagnostic dilemma that was eventually diagnosed as an unreported complication of PV. Typically, a patient with PV is diagnosed incidentally or presents with splenomegaly. Blackening of both upper and lower limbs is common and often associated with pruritus.

Case Presentation: However, our patient, a 51-year-old female presented with hypertension and rapidly progressed to transient bilateral loss of vision (Amaurosis Fugax). It was difficult to form a clinical diagnosis but her blood examination raised a suspicion of PV and it was swiftly confirmed by her JAK2 positive status. What followed was a series of phlebotomies which eventually restored her vision and fixed her impaired blood parameters.

Conclusion: This case report enlightens the medical community about the various thromboembolic events that should warrant a search for PV. This report also appreciates the impact of newer medical remedies, namely JAK2 inhibitors in the light of PV.

KEYWORDS: Polycythemia vera, transient Loss of vision, Amaurosis Fugax, Phlebotomy, Case report, thromboembolic events in PV, JAK2.

I. INTRODUCTION

Polycythemia Vera is a hematopoietic stem cell disorder typically marked by erythropoiesis, granulopoiesis, and megakaryopoiesis without other abnormal cellular findings. The condition is associated with a mutation in JAK2 located on the short arm of chromosome 9 which replaces valine with phenylalanine (V617F) and causes the clinical spectrum of PV. It has a low prevalence rate of 44 per 100,000 population [5], making it among some of the rare diseases in the world. PV has a wide clinical spectrum ranging from hypertension and pruritus to life-threatening thromboembolic events. Helicobacter Pylori infestation is also commonly seen among PV patients.

Our patient, a 51-year-old female presented with unresolved hypertension, a darkening of limbs with pruritus, and acid reflux. Before we could investigate further she developed acute bilateral loss of vision which was spontaneously restored in 20 minutes. Ophthalmological and Neurological examination revealed no abnormalities but her complete blood count showed elevated RBCs, WBCs, and platelets. JAK2 positive status confirmed PV.

This case report is a journey of her successive phlebotomies which restored her blood picture in addition to preventing further thromboembolic events.

CASE PRESENTATION AND DIAGNOSTIC ASSESSMENT

A 51-year-old, Indian female presented to the clinic with non-resolving hypertension despite using Telmisartan 40mg for 6 months which was being prescribed by her family doctor. Upon examination of her vitals, a blood pressure of 180/100mmHg was noted. Her external examination revealed a darkening of her limbs with pruritus. The patient had complaints of acid reflux which was being controlled by Pantoprazole 40mg once a day before breakfast. Her abdominal examination revealed no splenomegaly. Before the patient could be taken for bloodwork, she developed acute, painless loss of vision which was restored after 20 minutes. Upon immediate ophthalmological evaluation, there were no significant findings and her visual acuity was also restored perfectly. Her CNS ocular examination for cranial nerves 3, 4, and 6 revealed no abnormalities. There was no gait ataxia or dysdiadochokinesia. The patient had a history of craniotomy following intra-cerebral haemorrhage due to a road traffic accident 2 years back. There were no signs of polycythemia in any previous records. There was no family history of PV or any myeloproliferative disorders.

Her bloodwork revealed Hemoglobin of 21.7%, a Leukocyte count of 32,840, platelets count of 391k, and haematocrit of 71.7%. JAK2

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was positive and this confirmed our suspicion of Polycythemia Vera.

THERAPEUTIC INTERVENTIONS

The primary treatment was successive phlebotomies with a target haematocrit of <45%. Therapeutic phlebotomy was started twice a week for one week, once a week for 2 weeks, and once every 15 days till the therapeutic goal was achieved and 300-350 ml blood was removed during each session. Below is a table of comparison of her blood work during her first week of treatment.

INVESTIGATIONS AND TIMELINE

	DAY 1	DAY 3	DAY 4	DAY 6	DAY 8
HEMOGLOBIN (g%)	21.7	21.7	21.9	21.0	20.5
TOTAL COUNT	32,840	26,300	29,900	27,690	28,730
DIFFERENTIAL COUNT	N88/L7/M4/E1	N89/L5/M4/E2	N89/L4/M5/E2	N92/L31/M3/E2	N90/L4/M3/E3
PLATELETS (LAKHS)	3.91	3.14	3.52	3.40	3.88
RBC COUNT	11.70	11.57	11.66	11.15	10.89
HEMATOCRIT (%)	70.7	70.5	71.9	68.4	67.8

1ST PHLEBOTOMY ON DAY 4
2ND PHLEBOTOMY ON DAY 8

Telmisartan 40 mg OD was continued as antihypertensive of choice and her blood pressure reverted within physiological range following a month of treatment. Aspirin 150mg OD to prevent further thromboembolic events was also added. Allegra 180mg OD was the preferred antihistaminic of choice for pruritus.

FOLLOW-UP AND OUTCOME

A series of phlebotomies was continued as mentioned above for a period of one month which saw her hematocrit levels reduce to 43%. There were no thromboembolic events apart from a solitary episode of Amaurosis Fugax. The patient is advised for monthly follow-ups and her hematocrit is being constantly monitored. She still requires phlebotomies but at monthly intervals to keep her haematocrit stable around 40%. She has not developed any adverse effects to the treatment yet.

II. DISCUSSION

Polycythemia Vera should always be considered in cases of high haemoglobin and haematocrit with normal oxygen saturation. The thromboembolic events following such high haematocrit should not be limited to the Hepatic vein (Budd Chiari syndrome), splenic vein (splenic infarction), or renal vein thrombosis, but any thromboembolic event in the right patient setting should raise a suspicion of PV. Prevention of further thromboembolic events should be of the highest priority as it is the most serious complication. [3]

Splenomegaly is another finding frequently implicated in these conditions and splenectomy was almost routine until the advent of JAK1/2 inhibitor, Ruxolitinib. It is a non-specific JAK 2 inhibitor and has been shown to reduce the size of the spleen, cure typical symptoms like pruritus,and even reduce the dependence on phlebotomies. It remains the second line to Hydroxyurea. [3]

Iron deficiency is often present in such patients, particularly females. Correction of serum ferritin should not be done before haematocrit control as it can lead to the flaring of erythropoiesis.

Hyperuricemia is also frequently associated with this condition and treatment with Allopurinol should be considered when the levels are >10mg/dl. [4]

If left untreated, the disease may progress into Acute Myeloid Leukemia (AML). This is usually seen in 3% of patients if left untreated for 10 years. Myelodysplastic syndromes and Myelofibrosis are also rare forms of disease progression and may occur prior to the disease developing into AML.[1]

III. CONCLUSION

Thromboembolic events following Polycythemia Vera could be seen in retinal vasculature causing transient loss of vision. Amaurosis Fugax in patients of PV should be treated as an emergency because if the blood circulation is impaired for a longer duration it might lead to irreversible repercussions. A vast majority of asymptomatic patients can be managed by successive therapeutic phlebotomies. Upon facing complications with venous access, Ruxolitinib can be administered. [3]

LIST OF ABBREVIATIONS

PV - Polycythemia Vera
RBC – Red Blood Cells
WBC – White Blood Cells
AML – Acute Myeloid Leukemia

DECLARATIONS

PATIENT CONSENT

- The patient has given her consent to publish the case

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

-Not applicable

CONSENT FOR PUBLICATION

- Not applicable

AVAILABILITY OF DATA AND MATERIALS

- Not Applicable

COMPETING INTERESTS

- The authors declare that they have no competing interests

FUNDING

- Not applicable

AUTHORS CONTRIBUTION

AMK compiled all the reports and compared them with other similar studies, SM did the literature research, and AHK reviewed and edited the final documents.

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Not Applicable

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