Case Report

DOI: 10.7860/JCDR/2022/57513.16864

Polyglobulia Masquerading as Polycythaemia Vera Presenting as Superior Mesenteric Vein Thrombosis: A Case Report

Internal Medicine Section

JANHAVI NITIN MAHAJAN[1], PRERNA VERMA[2], YOGESH KAKDE[3], SUNIL KUMAR[4], ANIL WANJARI[5]

ABSTRACT

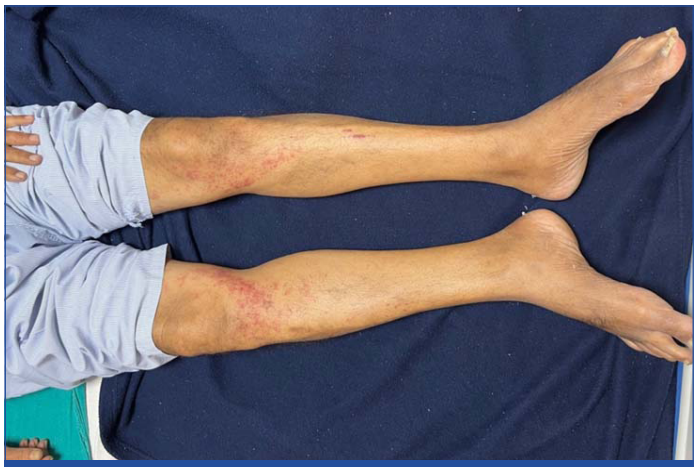
Polyglobulia is secondary polycythaemia commonly due to underlying non haematological diseases like Chronic Obstructive Pulmonary Disease (COPD), obstructive sleep apnoea and also sometimes in people living in hilly or forest areas. It can occur in any venous or atrial thrombosis of the vessels, but cardiac, cerebral, and mesenteric vessels are usually involved. One of the rare causes of abrupt severe abdominal discomfort is portal vein thrombosis, usually associated with liver cirrhosis and thrombophilia. In this case report, the authors have highlighted a case of a 36-year-old male residing in the hilly area of Maharashtra, India, who reported to hospital with severe abdominal pain due to superior mesenteric vein thrombosis. On investigation, he had increased haemoglobin with raised haematocrit diagnosed as polyglobulia and became part of polycythaemia with positive JAK2 V617F mutation.

Keywords: Haemoglobin, JAK2 mutation, Myeloproliferative disorder, Pain abdomen

CASE REPORT

A 36-year-old male welder presented to casualty with complaints of intermittent abdominal pain for 2 months associated with distension and both lower limb swelling. He had not consulted any physician for the same. He was a chronic smoker, who used to smoke 20 bidis per day since 10 years, residing in the hilly area of Maharashtra, India. He denied any history of taking any medication. There was no history of hypertension, diabetes mellitus or kidney disease.

On clinical examination, the patient had tender abdomen, and pitting oedema on both legs [Table/Fig-1]. The spleen was palpable and non tender. Other systemic examinations were within normal limits. His blood investigations are shown with normal reference range in [Table/Fig-2]. Viral markers were non reactive in view of raised haematocrit and haemoglobin, BCR-ABL was advised which came out be negative, JAK2 mutation was positive, and erythropoietin were markedly raised. Serial laboratory investigations of the patient from the day of admission till final diagnosis are presented in [Table/Fig-3].



[Table/Fig-1]: Oedema in both lower limbs with petechiae around the knee.

His Ultrasonogram (USG) of the abdomen and pelvis revealed narrowing of the portal vein and thrombosis of the superior mesenteric vein with multiple splenic infarcts with minimal ascites with distention of bowel loops. Contrast-Enhanced Computed Tomography (CECT) of the abdomen revealed chronic liver parenchyma disease with portal hypertension, multiple portosystemic collaterals, chronic portal vein, splenic vein, and proximal superior mesenteric vein thrombosis with gross ascites with splenomegaly with multiple infarcts with dilated small bowel loops with some of the jejunal loops showing mural enhancement [Table/Fig-4].

Investigation	Patient's test results	Reference range
Haemoglobin (g%)	19.7	12-15
Packed cell volume (%)	69	38-47
Red blood cell (million/cumm)	5.8	4.5-5.5
Total leucocyte count (cells/cumm)	9,600	4000-11000
Platelets (lacs/cumm)	4.1	1.5-4.5
Total bilirubin (mg/dL)	8	0.2-1.3
Direct bilirubin (mg/dL)	2.2	0.0-0.3
Serum total proteins (g/dL)	5.9	5.5-8
Serum albumin (g/dL)	2.8	3.5-5
Serum ferritin (ng/mL)	405	11.1-264
Serum LDH (U/L)	1063	120-246
Serum vitamin B12 (pg/mL)	1000	239-931
Serum procalcitonin (ng/mL)	9.42	More than 0.25
Prothrombin time and international normalized ratio	1.44	1
Serum uric acid (mg/dL)	186	3.5-8.5
Serum creatinine (mg/dL)	2.1	0.52-1.04
Antinuclear antibodies (IU/mL)	1.62	More than 1.1 positive

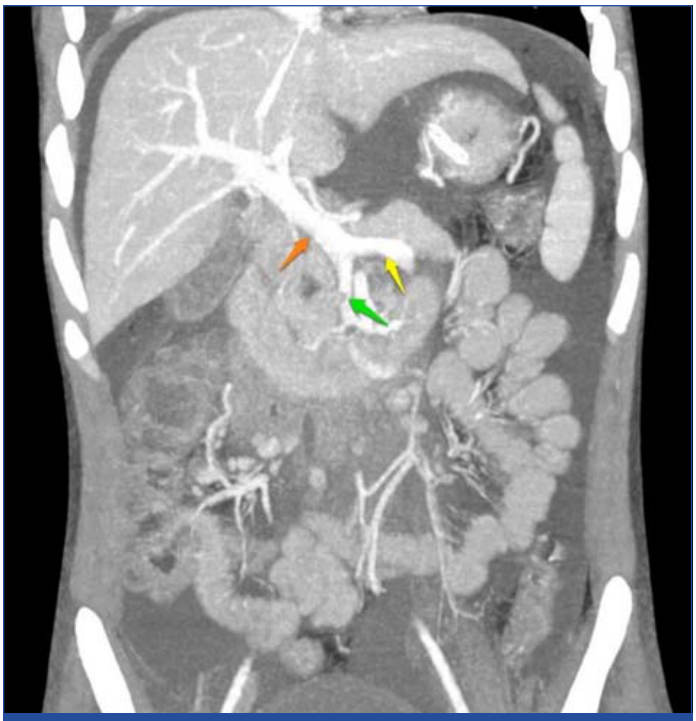
[Table/Fig-2]: Investigations with their normal range.

Bone marrow aspiration indicates hypercellular marrow with megaloblastic hyperplasia. It also shows that there is increased megakaryocytosis, which confirms myeloproliferative disorder, possibly polycythaemia vera.

The patient was started on hydroxyurea and aspirin 75 mg per day. Phlebotomy was done multiple times (every alternate day for one week). His general condition improved and he was discharged, and regular follow-up was advised. The first two months of follow-up were uneventful.

Investigation date	Total RBC count (million/cumm)	Total WBC count (cells/cumm)	Haemoglobin (%)	MCHC (g/dL)	MCV (µm3)	MCH (pg/cells)	Total platelet count (lacs/cumm)	HCT (%)	RDW (%)
Day 1	8.83	9600	19.7	32.1	69.5	22.3	4.6	61.3	23.1
Day 2	7.11	7300	16.4	33.2	69.6	23.1	4.23	49.5	22.6
Day 3	7.25	4700	16.7	33.1	69.6	23.1	4.4	50.5	22
Day 4	7.29	8700	16.7	32.5	70.8	23	3.83	51.6	23.3
Day 5	7.24	9100	16.7	33.1	69.8	23.1	3.08	50.5	22.5
Day 7	7.56	8100	17.3	32.3	71	22.9	2.84	53.6	22.6
Day 8	6.94	3900	16.2	33.3	70.3	23.4	2.68	48.8	21.6
Day 11	6.81	6100	15.1	32.5	68.5	22.2	2.62	46.6	21.4
Day 12	5.86	5600	13.9	33.5	70.7	23.7	2.38	41.4	21.9
Day 14	5.74	3500	13.3	32.9	70.3	23.1	1.8	40.3	21.5

[Table/Fig-3]: Serial investigations.
RBC: Red blood cell; WBC: White blood cell; MCHC: Mean corpuscular haemoglobin concentration; MCV: mean corpuscular volume; MCH: Mean corpuscular haemoglobin; HCT: Haematocrit; RDW: Red blood cell distribution width



[Table/Fig-4]: Image shows portal vein and its branches (orange arrow), splenic vein from splenic hilum (yellow arrow) to the portal vein confluence and superior mesenteric vein suggestive of superior mesenteric vein thrombosis (green arrow).

DISCUSSION

Clinically, Polycythaemia Vera (PV) is distinguishable from other myeloproliferative disorders by increasing RBC mass. Furthermore, patients with aberrant haematocrit readings before the JAK2 V617F period may have had occult or latent JAK2 V617F [1].

Patients suffering from secondary polycythaemia as a consequence of physiologically significant reaction, requires high RBC mass than typical for adequate oxygen supply to tissues. Hypoxia stimulating factors that increase erythropoietin's gene expression are revitalised by increased hypoxia [2].

Thrombosis of the mesenteric, hepatoportal and splenic vein, bruising, peptic ulcer disease, gastrointestinal bleeding and digital ischaemia can occur due to uncontrolled erythrocytosis [2].

Another complication of thrombocytosis in PV is erythromelalgia, which causes erythema, burning, and pain in the limbs due to increased platelet stickiness. Hyperuricaemia with secondary gout, uric acid stones, and hypermetabolic symptoms might result from the rapid turnover of haematopoietic cells [3,4].

Erythropoietin assay differentiates between primary and secondary polycythaemia, which is elevated later. In addition, when a higher red cell mass is discovered, a JAK2 mutation must be evaluated to rule out polycythaemia vera. The JAK2 V617F mutation is PV susceptible (97% sensitivity) and nearly 100% specific in increasing haematocrit. The risk of a misleading laboratory result is successfully mitigated by evaluating serum erythropoietin levels, which are below normal in more than 85% of PV patients [4,5]. In our case dilemma was raised erythropoietin with positive JAK2 mutation, whether to think secondary polycythaemia or polycythaemia vera.

The management aspect of secondary polycythaemia depends on the causes that are involved. It needs the correction of precipitating factors used as a correction for haematologic abnormality [5,6]. One of the causes of portal vein thrombosis is myeloproliferative diseases, which have recently emerged as a prominent systemic cause of portal vein thrombosis, though it has also been reported in sickle cell disease [7]. Those patients who smoke are advised to quit smoking and recommend the appropriate supportive therapy and psychological and pharmacological intervention.

Administration of low flow oxygen helps correct hypoxia and secondary PV, especially in the case of a patient with Chronic Obstructive Pulmonary Disease (COPD), where obesity, hypoventilation syndrome, and weight loss can be treated through lifestyle modification, bariatric surgery, and pharmacological therapy [4,8].

An approach for the management of secondary polycythaemia varies along with the complications. Low-dose aspirin helps to treat and prevent thromboembolic episodes. In polycythaemia vera, venesection is associated with a lower risk of cardiovascular mortality and thrombosis; extrapolation of these findings to secondary polycythaemia, as in our instance, is common. In contrast to individuals with secondary polycythaemia caused by physiologically acceptable erythrocytosis, phlebotomy is therapeutic for maintaining haematocrit values of 42-46% [6,8].

CONCLUSION(S)

In conclusion, this case report is a rare case of polycythaemia vera presented with the portal vein and splenic vein thrombosis. The underlying haematological condition is essential in diagnosing and treating this scenario because transformation to Chronic Myeloid Leukaemia (CML) or Myelodysplastic Syndrome (MDS) is a primary cause of death in polycythaemia vera.

REFERENCES

[1] Ahmed S, Irfan O, Siddique S, Abid S. Polycythaemia vera presenting as a porta hepatis mass. J Ayub Med Coll Abbottabad. 2019;31(4):627-28.
[2] Dhiman P, Saxena P. JAK2V617F: Is it sufficient as a single player in splanchnic venous thrombosis? Case Rep Hematol. 2015; 2015:373490.
[3] Erdinc B, Ramachandran P, Boris A. Polycythemia vera presenting with normal hemoglobin and hematocrit: A rare variant. Cureus. 2020;12(6):e8404.
[4] Tevet M, Ionescu R, Dragan C, Lupu AR. Influence of the JAK2 V617F mutation and inherited thrombophilia on the thrombotic risk among patients with myeloproliferative disorders. Maedica (Bucur). 2015;10(1):27-32.
[5] Hasan T, El Makki E. Unusual involvement of the portal vein in polycythemia vera-Genetic perspective. Acta Medica Int. 2015;2(2):182-85.

[6] Kumar N, Sharma S, Binota J, Ahluwalia J, Varma N, Naseem S, et al. JAK2V617F mutation in patient with splanchnic vein thrombosis. Indian J Hematol Blood Transfus. 2020;36(4):700-04.
[7] Kumar S, Joshi R, Jain AP. Portal hypertension associated with sickle cell disease. Indian J Gastroenterol. 2007;26(2):94.
[8] Thomas A, Robert A, Chemplavil DB. Unusual presentation of JAK2-negative polycythemia. Indian J Case Rep. 2018;4(3):176-78.

PARTICULARS OF CONTRIBUTORS:
1. Junior Resident, Department of Medicine, Datta Meghe Institute of Health Sciences, Wardha, Maharashtra, India.
2. Junior Resident, Department of Medicine, Datta Meghe Institute of Health Sciences, Wardha, Maharashtra, India.
3. Junior Resident, Department of Medicine, Datta Meghe Institute of Health Sciences, Wardha, Maharashtra, India.
4. Professor, Department of Medicine, Datta Meghe Institute of Health Sciences, Wardha, Maharashtra, India.
5. Professor, Department of Medicine, Datta Meghe Institute of Health Sciences, Wardha, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Janhavi Nitin Mahajan,
Department of Medicine, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences (Deemed to be University), Wardha, Maharashtra, India.
E-mail: drjanhavimahajan96@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: May 07, 2022
- Manual Googling: May 28, 2022
- iThenticate Software: Sep 08, 2022 (5%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **May 02, 2022**
Date of Peer Review: **Jun 01, 2022**
Date of Acceptance: **Jun 26, 2022**
Date of Publishing: **Oct 01, 2022**