International Journal of Research in Medical Sciences
Kanuga KH et al. Int J Res Med Sci. 2025 Nov;13(11):5008-5010
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20253642

Case Report

An aggressive clinical course of polycythemia vera with coexisting complications: a case report

Kshitij H. Kanuga[1]*, Anoushka Arora[1], Yashraj Dhruva[2]

[1]Smt. N.H.L Municipal Medical College, Ahmedabad, Gujarat, India
[2]SVPIMR

Received: 24 September 2025
Accepted: 18 October 2025

***Correspondence:**
Dr. Kshitij H. Kanuga,
E-mail: kshitijkanuga@gmail.com

ABSTRACT

Polycythemia vera (PV) is a myeloproliferative disorder characterized by rapid proliferation of red blood cells. It may present with symptoms of hyper viscosity. Complication of thrombosis is well recognized, but its rapid progress and coexistence of involvement of various organ sites simultaneously are not seen as often. We present a case of a 40-year-old female who was incidentally diagnosed with PV due to her presentation with extrahepatic portal venous obstruction (EHPVO). Her elevated platelet count prompted a bone marrow biopsy, leading to the finding of Janus kinase-2 (JAK2)-positive PV. Her condition rapidly progressed to decompensated chronic liver disease, likely triggered by the portal hypertension due to EHPVO. This was further complicated by her PV progressing to myelofibrosis, a rare complication of PV. This progression typically occurs after 10-15 years of diagnosis, but in this case, it was seen within 3 years, demonstrating the rapid course of her disease. This case is noteworthy for how differently PV can present. It highlights the importance of investigating patients for underlying blood malignancies when they present with clots without any identifiable cause. It also sheds light on how fast this disease can progress and shows the importance of early diagnosis and appropriate management to prevent several complications associated.

Keywords: Polycythemia vera, Hyper viscosity, EPO

INTRODUCTION

Polycythemia can be defined as an increase in absolute red blood cell (RBC) mass in the body.[1] This translates to the laboratory values as increased hemoglobin levels or increased hematocrit levels. The normal hemoglobin levels are 12.3-16.4 g/dL for males and 11.0-14.4 g/dL for females.[2] Similarly, normal hematocrit levels are 40 to 54% for men and 36 to 48% for women.[3]

Polycythemia can be spurious or true. Spurious polycythemia is due to volume contraction rather than increased absolute RBC mass. This can be seen in cases of dehydration. True polycythemia can be divided into primary polycythemia or secondary polycythemia based on serum erythropoietin (EPO) levels and underlying cause. Primary polycythemia has decreased EPO levels. It is caused by uncontrolled RBC production in the bone marrow due to dysfunction. The most common cause of primary polycythemia is PV. PV is a myeloproliferative neoplastic disorder that causes uncontrolled RBC production with a concurrent stimulation of myeloid and megakaryocytic lineages, leading to increased white blood cell and platelet production.[4] The most common pathophysiology causing PV is a mutation in JAK2 gene.[5] A valine to phenylalanine substitution at position 617 of the JAK2 gene results in active cytokine receptors, and this mutation is observed in over 95% of patients with PV.[6]

The excessive RBC production leads to complications due to hyperviscosity, which leads to impaired oxygen delivery. Additionally, hyperviscosity leads to increased complications due to thrombus formation. Some presentations include pulmonary embolism, stroke, arterial

thrombosis, splanchnic vein thrombosis, deep vein thrombosis, and Budd-Chiari syndrome (BCS). BCS is a rare and challenging complication. It is caused by the obstruction of venous outflow system.[7] Myeloproliferative disorders are the leading cause of BCS.[8] Another rare and challenging complication of PV is myelofibrosis.

Myelofibrosis is another myeloproliferative neoplasm associated with JAK2, CALR, or MPL mutation.[9] The risk of progression of PV to myelofibrosis is 4.9% and 9.4% at 10 and 15 years from diagnosis, respectively.[10] It has been hypothesized that myelofibrosis is due to the replacement of blood-forming cells with reticulin fibres caused by JAK-STAT-induced inflammation and acquired mutation in hemopoietic stem cells.[11]

CASE REPORT

A 40-year-old female presented to the hospital in March 2019 with abdominal pain and diarrhea. The patient was vitally stable. On physical examination, an abdominal lump/swelling was palpated in the left upper quadrant that crossed midline. To further investigate, an ultrasonography (USG) was done, which showed massive splenomegaly. Additionally, to find the root cause of the massive splenomegaly, a Doppler was performed, which showed absent flow in the splenic vein, alluding to a splenic vein thrombosis. Thus, EPVO was diagnosed.

Since this was a case of unprovoked EPVO, further tests were conducted to find the root cause, which included complete blood count (CBC) and coagulation profile. Her coagulation profile was normal, but her CBC revealed a high platelet count of 6,33,000. For further investigation, a bone marrow (BM) biopsy was conducted, which showed “Mildly hypercellular marrow with panmyeloid hyperplasia consistent with CMPD-PV”. This led to a diagnosis of PV. A cytogenetic study was conducted to delineate the cause, which revealed a JAK2 V617F mutation. Her final diagnosis is EHPVO with PV. To manage the thrombocytosis, she was started on 500 mg hydroxyurea (HU) once daily.

Post diagnosis, she had multiple hospitalisations for hemoptysis, which was caused by esophageal varices. These varices resulted from the elevated portal pressure due to the EPVO. She underwent multiple esophagoscopy and banding procedures to manage these varices from 2019 to 2020.

In 2021, the patient had complaints of abdominal pain, distention, nausea, and vomiting for 10 days. Further investigation with FibroScan revealed the development of liver cirrhosis due to persistently elevated portal vein pressure. In May of 2022, a Fibroscan was done to confirm the diagnosis of decompensated chronic liver disease (DCLD).

Since the diagnosis of PV, she has been required to do regular blood counts as part of her follow-up. During a follow-up in 2023, her hemoglobin was reported to be 7.8. This led to a subsequent bone marrow biopsy, which showed transformation from PV to Secondary Myelofibrosis. Ever since then, the patient has been transfusion dependent and has had multiple episodes of blood transfusions in the last 2 years.

DISCUSSION

Our case demonstrates a highly complex presentation of PV complicated by EHPVO, subsequent liver cirrhosis, and eventual progression to myelofibrosis.

Studies show that 40% of patients with non-cirrhotic non-solid tumor-associated splanchnic vein thrombosis have an underlying myeloproliferative neoplasm. In our case, the first manifestation was splanchnic vein thrombosis, causing splenomegaly. Here, the consequent presentation of splenomegaly and thrombocytosis prompted the need for a bone marrow biopsy, which confirmed PV with a JAK2 V617F mutation.

Classically, PV presents with symptoms of hyperviscosity, elevated hemoglobin and hematocrit, plethora, pruritus after coming in contact with hot water, and weight loss. Our patient, on the other hand, had no such presenting symptoms. This unique presentation makes the diagnosis challenging.

Following diagnosis, the patient developed portal hypertension, possibly due to EHPVO, which manifested as recurrent hemoptysis due to bleeding esophageal varices. This was treated by repeated endoscopic band ligations. She progressed to have decompensated chronic liver disease in an unusually short period of 3 years.[12]

Another interesting feature of our case was the transformation of PV to myelofibrosis in a short time period. The progression of polycythemia to myelofibrosis in a period of 3 years does not have the presence of supporting literature, and is less than 10% in a period of less than 10 years.[13] This accelerated evolution suggests an aggressive disease phenotype and highlights the heterogeneity in PV progression.

CONCLUSION

This case underscores the coexistence of two rare complications of PV, EHPVO with portal hypertension and early progression to myelofibrosis. Both occurred at a much faster rate than typically described in the literature. It emphasizes the importance of maintaining a high index of suspicion for atypical presentations of PV and recognizing complications early to optimize patient outcomes.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Pillai AA, Fazal S, Mukkamalla SKR, Babiker HM. Polycythemia. In: StatPearls. StatPearls Publishing. 2025.
2. Ashavaid TF, Todur SP, Dherai AJ. Establishment of reference intervals in Indian population. Indian J Clin Biochem. 2005;20(2):110-8.
3. Billett HH. Hemoglobin and Hematocrit. In: Walker HK, Hall WD, Hurst JW, eds. Clinical Methods: The History, Physical, and Laboratory Examinations. 3rd ed. Butterworths. 1990.
4. Lu X, Chang R. Polycythemia Vera. In: StatPearls. StatPearls Publishing. 2025.
5. James C, Ugo V, Le Couédic J-P, Judith S, François D, Catherine L, et al. A unique clonal JAK2 mutation leading to constitutive signalling causes polycythaemia vera. Nature. 2005;434(7037):1144-8.
6. Spivak JL, Silver RT. The revised World Health Organization diagnostic criteria for polycythemia vera, essential thrombocytosis, and primary myelofibrosis: an alternative proposal. Blood. 2008;112(2):231-9.
7. Metzger PB, Costa KR, Silva SLE, Valter RDS, Vinicius N, Murilo QBF, et al. Budd-Chiari syndrome due to hepatic venous web outflow obstruction: percutaneous treatment with balloon angioplasty. J vasc bras. 2021;20:e20200133.
8. Smalberg JH, Arends LR, Valla DC, Kiladjian J-J, Janssen HLA, Leebeek FWG. Myeloproliferative neoplasms in Budd-Chiari syndrome and portal vein thrombosis: a meta-analysis. Blood. 2012;120(25):4921-8.
9. Tefferi A. Primary myelofibrosis: 2023 update on diagnosis, risk-stratification, and management. Am J Hematol. 2023;98(5):801-21.
10. Kiladjian J-J, Gardin C, Renoux M, Bruno F, Bernard J-F. Long-term outcomes of polycythemia vera patients treated with pipobroman as initial therapy. Hematol J. 2003;4(3):198-207.
11. Gleitz HFE, Benabid A, Schneider RK. Still a burning question: the interplay between inflammation and fibrosis in myeloproliferative neoplasms. Curr Opin Hematol. 2021;28(5):364-71.
12. Beleva EA. Splanchnic Vein Thrombosis in Myelofibrosis-An Underappreciated Hallmark of Disease Phenotype. Int J Mol Sci. 2023;24(21):15717.
13. Cerquozzi S, Tefferi A. Blast transformation and fibrotic progression in polycythemia vera and essential thrombocythemia: a literature review of incidence and risk factors. Blood Cancer J. 2015;5(11):e366.

Cite this article as: Kanuga KH, Arora A, Dhruva Y. An aggressive clinical course of polycythemia vera with coexisting complications: a case report. Int J Res Med Sci 2025;13:5008-10.