Case Report

Cholelithiasis as the presenting feature of polycythemia vera: An extremely rare case report

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ABSTRACT

Polycythemia vera (PV) is a JAK2-mutated myeloproliferative neoplasm that typically presents with erythrocytosis-related symptoms or thrombotic events. Usually, it presents at the age of 40–60 years, and the majority are in females. Gallstone disease, though common in older women, is an exceedingly uncommon initial manifestation of PV. We describe a 63-year-old postmenopausal female with congenital talipes equinovarus who presented with intermittent right upper quadrant abdominal pain and was found to have cholelithiasis and hepatosplenomegaly on ultrasonography. Routine laboratory tests revealed persistently elevated hemoglobin and hematocrit with suppressed serum erythropoietin (EPO), and subsequent JAK2 V617F testing confirmed PV. This case illustrates how a common gastrointestinal complaint may unmask an extremely rare occurrence of an underlying hematological malignancy, PV, which was otherwise silent, emphasizing the need for thorough evaluation of unexplained erythrocytosis in patients presenting with otherwise typical biliary pathology.

Key words: Cholelithiasis, Hyperviscosity, Phlebotomy, Polycythemia vera

Polycythemia encompasses primary and secondary forms. Primary polycythemia vera (PV) is a clonal myeloproliferative neoplasm driven by JAK2 mutations, resulting in erythrocytosis, which may or may not be accompanied by leukocytosis and thrombocytosis. Secondary polycythemia arises from elevated erythropoietin (EPO) due to chronic hypoxia, smoking, obstructive sleep apnea, high-altitude exposure, renal artery stenosis, or EPO-secreting tumors [1,2]. Gaisböck syndrome represents relative polycythemia caused by plasma volume contraction without true erythrocytosis [3]. Diagnostic criteria for PV include elevated hemoglobin or hematocrit, a hypercellular bone marrow with trilineage proliferation, JAK2 V617F or exon 12 mutation, and low serum EPO [2,4]. PV may predispose to bilirubin gallstones via increased hemoglobin turnover, hyperviscosity-induced biliary stasis, and hyperuricemia [1,2,4]. Standard management includes phlebotomy, aspirin, and cytoreductive therapy, with prognosis generally favorable despite risks of thrombosis and disease progression [2,4].

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CASE REPORT

A 63-year-old postmenopausal female with a known history of bilateral congenital talipes equinovarus (CTEV) presented with intermittent upper abdominal pain for the past 3 years.

On general examination, pulse and blood pressure were within normal limits. She had CTEV (Fig. 1), and besides that, the other finding on physical systemic examination was mild hepatosplenomegaly on per abdomen.

An initial ultrasound of the abdomen performed during evaluation confirmed mild hepatosplenomegaly and revealed cholelithiasis, measuring 15 mm in size. She denied headache, vertigo, warmth, or burning sensations in the feet, helping to exclude erythromelalgia, which may occur in myeloproliferative neoplasms. She subsequently underwent routine laboratory tests, which showed elevated hemoglobin and hematocrit on two separate complete blood count reports. Other investigations are shown in Table 1. Serum EPO levels were found to be low, and in the absence of identifiable risk factors for secondary polycythemia (such as smoking, chronic hypoxia, renal disease, or high-altitude exposure), a provisional diagnosis of PV was considered. This was confirmed when the JAK2 mutation test

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Table 1: Investigations of the case

Laboratory parameter	At presentation	Reference range
Hemoglobin (g/dL)	19.4	12–17
Hematocrit (%)	60.8%	38–45
TLC (cells/mm^3)	6700	4000–11000
DLC: Granulocytes/Lymphocytes (%)	76/17	40–80/20–40
Platelet count (lakh cells/mm^3)	3.22	1.5–4
Sodium/potassium/ionized calcium (mmol/L) (serum)	140.2/5.07/1.16	135–150/3.5–5.3/1.13–1.32
SGOT/SGPT/ALP (U/L)	51/45/307	<40/<45/<270
S. Protein/albumin/globulin (g/dl)	8.1/4.2/3.9	6.4–8.3/3.5–5.2/2–3.5
S. urea/creatinine (mg/dL)	18.5/0.47	13–43/0.8–1.3
S. Bilirubin (total/direct/indirect) (mg/dL)	0.89/0.16/0.73	<1.2/<0.2
S. Uric acid (mg/dL)	9.3	2.7-7.3
S. EPO (mIU/mL)	2.23	4.3–29
JAK2 V617F mutation	Detected Positive	
USG W/A	Hepatomegaly (18.7 cm) with increased echotexture, splenomegaly(15.5 cm). Cholelithiasis with multiple calculi, the largest being 20 mm	
HIV	Non reactive	
2D Echo	Normal study (LVEF=60%)	
Pulmonary function test	Within normal limits	

TLC: Total leukocyte count, DLC: Differential leukocyte count, SGOT: Serum glutamic oxaloacetic transaminase, SGPT: Serum glutamic pyruvic transaminase, ALP: Alkaline phosphatase, USG W/A: Ultrasound whole abdomen, S: Serum, EPO: Erythropoietin, JAK: Janus Kinase, LVEF: Left ventricular ejection fraction

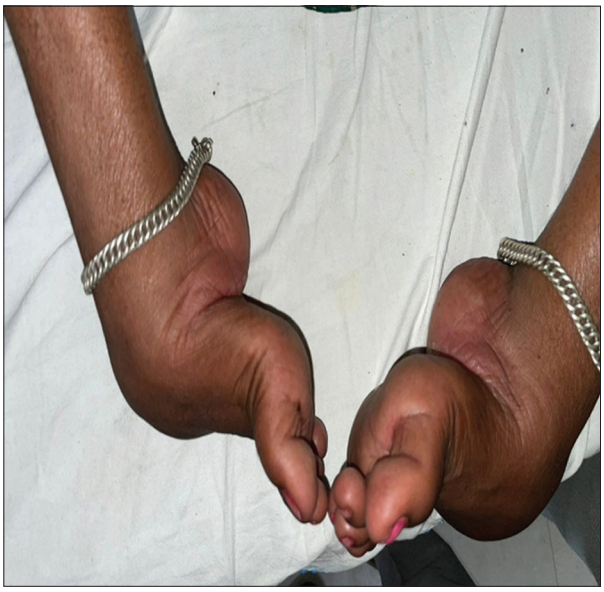

Figure 1: Showing bilateral congenital talipes equinovarus

returned positive. Computed tomography angiography was done to rule out the possibility of abdominal pain due to ischemic enteritis, which can be a common entity in patients with PV.

The patient was started on aspirin 75 mg once daily and allopurinol 300 mg once daily, and phlebotomy was done as part of standard management for PV. The patient improved gradually.

DISCUSSION

Gallstones are frequently observed in elderly females, with the majority in the age group of 41–60 years [5]; however, their emergence as an initial clinical indicator of PV is distinctly uncommon. In this patient, the diagnosis of PV was supported by persistently elevated hemoglobin, hematocrit, and red cell counts, alongside low serum EPO and JAK2 V617F positivity, fulfilling the World Health Organization major and minor criteria.

Although PV is known to predispose to pigment stone formation, gallstones seldom constitute the presenting feature.

Multiple mechanisms contribute to this association. Increased red cell turnover leads to excess bilirubin production, promoting black pigment stone formation [1]. Hyperviscosity, a hallmark of PV, may further impair biliary flow and contribute to stasis [6]. In addition, hyperuricemia, common in PV due to accelerated cell turnover, may alter bile composition and facilitate stone formation [7]. Our patient demonstrated a normal platelet count at presentation, although thrombocytosis is frequently observed in PV. The disease is characterized by an elevated cell mass leading to hyperviscosity, hypertension, and a high risk of venous or arterial thrombosis, particularly hepatic or portal vein thrombosis. Severe thrombocytosis may cause bleeding, digital ischemia, or erythromelalgia. Hyperuricemia, gout, and metabolic disturbances also arise due to increased cellular turnover [8].

PV patients presenting with the sole chief complaint of gallstones are a rare entity, with medical literature having scarce data on the prevalence. There is, however, a case report of an 11-week-old polycythemic infant who developed cholestasis from gallstones, highlighting the unusual link between excessive erythrocyte turnover and early-onset cholelithiasis in infancy [9]. Our case has bilateral CTEV. No medical literature to date has shown any association of CTEV with gallbladder stones or primary PV. Till date there was only one study in medical literature which showed some relation of CTEV with hepatobiliary system; Gowda M et al have documented through a descriptive study that CTEV, as a fetal primary skeletal anomaly, was seen in just

1 case out of total 13 cases with congenital hepatobiliary system abnormalities [10].

Therapeutic management options of polycythemia in general include aspirin, cytoreductive therapy for those at high risk for thrombosis, splenectomy for patients with painful splenomegaly or repeated episodes of splenic infarction, and hydroxyurea, which is the most commonly used cytoreductive agent. If hydroxyurea is not effective or not tolerated, alternatives include ropeginterferon alfa-2b, busulfan, in those who are older than 65 years, ruxolitinib, and fedratinib [11].

Despite this case being postmenopausal for two decades, an interval during which thrombotic risk typically rises, the patient never had any history of thrombotic events, and polycythemia was identified only when incidental abnormalities were detected on routine blood testing. Hydroxyurea is a commonly prescribed drug in patients with PV. This patient, however, was not started on hydroxyurea, keeping in mind the side effects, including bone marrow suppression, which may lead to neutropenia and thrombocytopenia. Moreover, it has not been found to be protective against venous thrombosis and rather has a leukemogenic effect [8]. The patient showed symptomatic relief, and hemoglobin was maintained at the desired levels after multiple sessions of phlebotomy. The patient did not give consent for a cholecystectomy after relief of abdominal pain. This reinstated the need for individualizing treatment therapy for patients.

CONCLUSION

This case highlights an unusual presentation of PV, where gallstone detection served as the initial clinical clue, underscoring the rarity of such an association. Early recognition of atypical manifestations is crucial, as delayed diagnosis may increase the risk of thrombotic and metabolic complications inherent to PV. The patient fulfilled the key diagnostic criteria, emphasizing the importance of systematic evaluation of unexplained erythrocytosis. This case reinforces the need for heightened clinical suspicion, particularly when common conditions such as gallstones occur alongside abnormal hematological parameters. Prompt identification and timely intervention remain essential for optimal outcomes in patients with PV.

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