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Case Report

Premature myocardial infarction as the initial manifestation of JAK2-positive polycythemia vera in a young adult: a case report

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ABSTRACT

Polycythemia vera is a clonal myeloproliferative neoplasm characterized by erythrocytosis and commonly associated with leukocytosis, thrombocytosis, and splenomegaly. Activating mutations in the JAK2 gene play a central role in its pathogenesis. Although typically a disease of older adults, polycythemia vera may present at a younger age with atypical and severe manifestations. We report the case of a 37-year-old Indian male who presented with massive splenomegaly and abnormal blood counts. Notably, the patient had a prior history of premature myocardial infarction requiring percutaneous coronary intervention at a young age, in the absence of conventional cardiovascular risk factors. Current evaluation revealed erythrocytosis, leukocytosis, thrombocytosis, and panmyelosis on bone marrow examination. Molecular testing confirmed the presence of a JAK2 mutation, establishing the diagnosis of polycythemia vera. The earlier myocardial infarction was retrospectively attributed to the underlying prothrombotic state associated with the disease. This case highlights that polycythemia vera may initially manifest as premature arterial thrombosis in young individuals. Awareness of such presentations is essential, as early diagnosis and appropriate management can prevent recurrent thrombotic complications and disease progression.

Keywords: Polycythemia vera, JAK2 mutation, Myocardial infarction, Premature thrombosis, Myeloproliferative neoplasm, Splenomegaly

INTRODUCTION

Polycythemia vera (PV) is a chronic clonal myeloproliferative neoplasm characterized by sustained erythrocytosis with variable leukocytosis, thrombocytosis, and splenomegaly, arising from constitutive activation of the JAK–STAT signalling pathway. The discovery of activating mutations in the Janus kinase 2 (JAK2) gene has provided a molecular basis for the disease and remains central to its diagnosis.[1,2]

Although PV is classically described in older adults, emerging data from India and other resource-limited settings indicate that younger patients frequently present with advanced disease, often due to delayed recognition.[3,7] In such patients, thrombotic events may precede overt hematological manifestations by several years.

Arterial thrombosis including myocardial infarction represents a particularly serious and sometimes under-recognized complication, driven by hyper viscosity, qualitative platelet dysfunction, leukocyte activation, and endothelial injury.[8,13]

Early identification of PV in young individuals presenting with unexplained arterial thrombosis is critical, as timely intervention can significantly reduce long-term morbidity and prevent recurrent vascular events.[10,15]

CASE REPORT

A 37-year-old Indian male, a farmer by occupation, presented with a gradually enlarging abdominal lump that he had noticed for approximately one year. Over the preceding two weeks, he developed persistent abdominal discomfort, which prompted him to seek medical attention. He denied fever, nausea, vomiting, weight loss, pruritus, headache, visual disturbances, bleeding manifestations, or any alteration in bowel or bladder habits.

The patient had a significant past history of ischemic heart disease. He had suffered an acute myocardial infarction five years prior, at the age of 32 years, for which he underwent percutaneous coronary intervention with angioplasty. The event occurred at a relatively young age and in the absence of conventional cardiovascular risk factors. At that time, an evaluation for secondary causes of thrombosis had been undertaken at the treating centre; however, no definitive hematological diagnosis was established.

There was no known history of chronic medical illness or hematological disease in the patient or his family. He reported long-standing alcohol consumption for nearly 17 years. He was a non-smoker and had no history of residence at high altitude or exposure to medications associated with secondary erythrocytosis.

On examination, the patient was alert and oriented, with stable vital signs and a body mass index of 21.58 kg/m². Abdominal examination revealed a markedly enlarged spleen extending well below the left costal margin, along with mild hepatomegaly. No other abnormal findings were noted on systemic examination.

Laboratory evaluation revealed elevated hemoglobin (16.8 g/dl) and hematocrit (54.9%), with a red blood cell count of 6.42 million/µl. The total leukocyte count was increased to 21.42 ×10⁹/l with neutrophilic predominance, and the platelet count was 537×10⁹/l. Peripheral blood smear examination showed predominantly normocytic, normochromic red blood cells with mild anisocytosis and occasional macrocytes. Neutrophils were increased in number and showed hyper segmentation. Platelets were mildly increased, with occasional giant forms. No hemoparasites were identified. Bone marrow evaluation demonstrated a hypercellular marrow with trilineage hematopoiesis, erythroid predominance, and increased megakaryocytes, findings consistent with a myeloproliferative neoplasm on clinical correlation (Figure 1).

Iron studies demonstrated reduced serum iron with normal ferritin levels, suggestive of functional iron deficiency in the setting of increased erythropoietic activity. Serum uric acid was mildly elevated. Renal function tests and serum electrolytes were within normal limits. Ultrasonography of the abdomen confirmed mild hepatomegaly and gross splenomegaly, with the spleen measuring 228 mm in its longest axis.

In view of the patient's history of premature myocardial infarction and the current findings of erythrocytosis, leukocytosis, thrombocytosis, and massive splenomegaly, a myeloproliferative neoplasm was strongly suspected. Bone marrow aspiration revealed a hypercellular marrow with proliferation of erythroid, myeloid, and megakaryocytic lineages, consistent with panmyelosis. Molecular analysis detected a JAK2 mutation, confirming the clonal nature of the disorder. Taken together, these findings supported a diagnosis of polycythemia vera in accordance with World Health Organization criteria.[5]

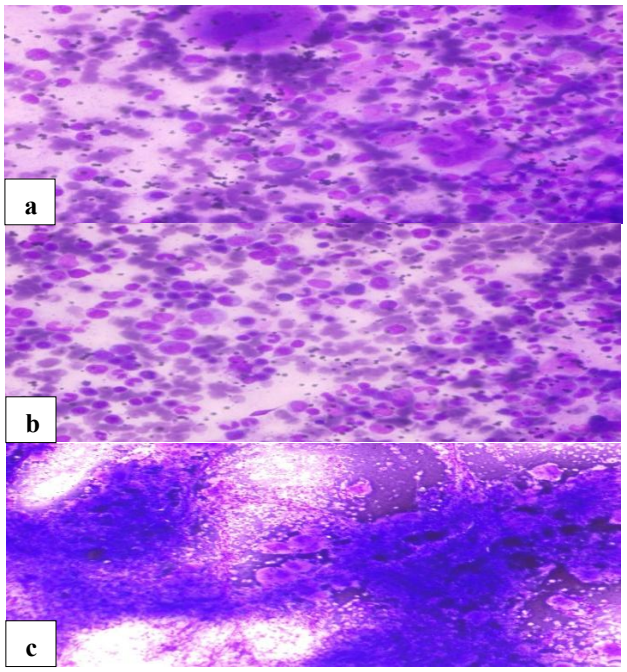

Figure 1: (a) Bone marrow aspirate smear showing a hypercellular marrow with trilineage hematopoiesis, including prominent erythroid hyperplasia and increased myeloid lineage cells, consistent with a myeloproliferative neoplasm on clinical correlation (Leishman stain, ×400); (b) high-power view of bone marrow aspirate revealing increased erythroid precursors and granulocytic series without excess blasts, supporting the diagnosis of polycythemia vera in the appropriate clinical and molecular context (Leishman stain, ×400); and (c) low-power view of bone marrow aspirate demonstrating marked hypercellularity with clustering of hematopoietic cells and increased megakaryocytes, suggestive of a myeloproliferative neoplasm (Leishman stain, ×100).

The patient was managed with therapeutic phlebotomy, with removal of 350 ml of blood, and was initiated on antiplatelet therapy. Supportive supplementation was provided, and he was counselled regarding alcohol cessation. Regular follow-up was advised to monitor

hematological parameters and assess ongoing thrombotic risk.

DISCUSSION

Thrombosis remains the most clinically significant complication of polycythemia vera and is a major determinant of morbidity and mortality. Both arterial and venous thrombotic events are well documented, with arterial events including myocardial infarction often occurring early in the disease course.[8,13] These complications may precede the formal diagnosis by several years, particularly in younger patients who lack traditional cardiovascular risk factors.[12]

The pathogenesis of thrombosis in PV is multifactorial. Increased red cell mass leads to hyper viscosity and impaired microcirculatory flow, while qualitative platelet abnormalities and leukocyte-mediated endothelial injury further promote a prothrombotic milieu.[1,4,14] JAK2 mutation burden has also been shown to correlate with thrombotic risk, underscoring the molecular contribution to vascular events.[16]

In the present case, the patient experienced an acute myocardial infarction five years prior to the diagnosis of polycythemia vera, which likely represented an early arterial manifestation of the underlying myeloproliferative disorder. The absence of conventional cardiovascular risk factors, followed by the later emergence of classical hematological features and massive splenomegaly, supports a causal association rather than coincidence. Similar presentations have been described in younger PV cohorts, particularly in the Indian population, where delayed diagnosis remains a challenge.[3,7,11]

This case highlights the importance of maintaining a high index of suspicion for occult myeloproliferative neoplasms in young patients presenting with unexplained arterial thrombosis. Recognition of such atypical timelines allows for appropriate risk stratification and initiation of therapy aimed at preventing recurrent thrombotic events.[9,15]

CONCLUSION

This case illustrates that polycythemia vera can present in young adults with premature arterial thrombosis, preceding the diagnosis by several years. A myocardial infarction occurring at an early age, particularly in the absence of conventional risk factors, should prompt consideration of underlying myeloproliferative neoplasms. Early diagnosis supported by molecular testing enables timely intervention and may prevent recurrent, potentially life-threatening thrombotic complications.

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