ORIGINAL RESEARCH PAPER

General Medicine

A CASE REPORT OF POLYCYTHEMIA RUBRA VERA MANIFESTING AS INTRACARDIAC THROMBOSIS AND PULMONARY EMBOLISM

KEY WORDS: Polycythemia Vera (PV), Intracardiac Thrombosis, Pulmonary Embolism, JAK2

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ABSTRACT

Polycythemia vera (PV) is a myeloproliferative disorder, with significant risk for thrombotic complications. But intracardiac thrombosis and pulmonary embolism is a rare initial presentation of PV. We describe the case of female patient who presented with intracardiac thrombosis, pulmonary embolism and right sided heart failure. This case report presents a patient with very rare combination of polycythemia rubra vera and right heart thrombi along with pulmonary embolism who was successfully managed with anticoagulation and phlebotomy

INTRODUCTION

Polycythemia Vera (PV) is one of the chronic myeloproliferative disorders, characterized by erythrocytosis and resultant manifestations, predominantly thrombotic events and related complications. Even though thrombosis is a known complication, intracardiac thrombosis is rare initial manifestation of polycythemia rubra vera and is seen usually in patients who have other risk factors for intracardiac thrombosis, such as valve disease, prosthetic valve, device induced or cardiomyopathy (1).

Here we present a case of polycythemia vera who presented with intracardiac thrombosis and pulmonary embolism

Case presentation

A 64 years old female patient with no remarkable past history was admitted with complaints of sudden onset of retrosternal and left sided chest pain. she also had associated breathlessness was acute in onset, gradually progressive, NYHA IV, not associated with cough, no diurnal variation. No history of fever, no history of any addiction.

The patient presented with the above complaints to emergency department

On examination BP was 130/80, spo2 89 % room air, JVP was raised, conjunctival congestion present, no edema feet. On cardiovascular system examination tachycardia was present. Respiratory system examination revealed tachypnea and crepitation on left side. On abdominal examination right hypochondrial tenderness present. CNS examination was normal.

ECG was done as patient was having chest pain and it was showing S1Q3T3 pattern, sinus tachycardia, P pulmonale, which probed us to plan for 2D ECHO and CT pulmonary angiography.

2D-ECHO was suggestive of global LV Hypokinesia with large homogenous mass seen in RA measuring 30*35 mm and 20*35 mm and moderate LV dysfunction which gave us suspicion of thrombus so cardiac MRI was planned. With Cardiac MRI thrombus in RA was confirmed. Cardiac MRI also revealed chronic infarction of apical inferior, mid inferior, midanteroseptal, inferobasal, basal inferoseptal and basalanterioseptal region of left ventricle and hypokinesia of posteromedial wall.

CTPA revealed partial non contrast opacification / thrombotic occlusion seen involving left main descending pulmonary artery for length of 9 mm causing 70-80% luminal compromise.

For our surprise, CBC revealed a raised hemoglobin of 20.6 g/dL ang hematocrit of 66%, raised total leucocyte count (22,800 cells/ mm^3) and raised platelet count (9.87 lakh). So probable diagnosis of PV was kept and was confirmed by bone marrow aspiration and subnormal erythrolein level (based on WHO criteria as mentioned in Table 1). Patient didn't give consent for JAK 2 mutation testing.

Retrospectively, complaints specific to PV like itching after taking a shower, tinnitus, vertigo headache, chest pain, diplopia or blurry vision was asked but patient denied all of these. Among the other investigations showed raised D Dimer, CRP and CPKMB. USG abdomen plus pelvis and fundus examination was also normal. So, diagnosis of intracardiac thrombosis with pulmonary embolism due to PV was made.

The patient was treated with aspirin 150 mg/day, and enoxaparin 100 mg subcutaneously twice daily and was overlapped with warfarin 5mg od. After stabilizing the patient was given 2 sittings of phlebotomy (250 ml of blood was removed in each sitting) and tab hydroxyurea 500 mg BD daily. In addition, she was also started on metoprolol 25 mg/day and atorvastatin 40 mg/ day to lower his risk of future adverse cardiac events. The patients general condition improved and no other complications were noted during her stay

DISCUSSION

PV is a chronic myeloproliferative disease which occurs in all population and all age group including adulthood, increasing with age, median age of diagnosis 60 year, with more prevalence in female rather in male(ratio ranges between 1.2:1-2.2:1), correlating with age and sex of our patient in this case report(2). The diagnosis of PV in our patient was made using WHO criteria (2016) mentioned in Table 1 in which our patient satisfied 2 major and 1 minor criteria '(3). The specific and characteristic symptom of aquagenic pruritus, which is described in nearly 50% of PV patients (as per literature) was absent in our patient (4)

Splenomegaly is estimated to affect 30% to 40% of PV patients and is usually associated with an advanced disease which was absent in our case.

PV is associated with an increased risk of venous or arterial thrombosis which occurs in 20-50 % cases (5). In a study conducted by Tefferi et al. in 1545 patients with PV, arterial

thrombosis was present in 16 percent and Venous thrombosis was present in 7 percent (6). As per study conducted in 587 patients by Cerquozzi et al ., acute coronary syndrome was the most common arterial event (45%) that occurred before or at the time of diagnosis, whereas splanchnic vein thrombosis (45%) was the most common venous event(7). Patients with PV have a high incidence of coronary artery disease and myocardial infarction and proposed mechanisms include coronary thrombosis and occlusive intimal proliferation(8).

Table 1 showing WHO diagnostic criteria for Polycythemia Rubra Vera

2016 World Health Organization diagnostic criteria for polycythemia vera

Major criteria	Minor criteria
• Hemoglobin > 16.5 g/dL(men)Hemoglobin > 16.0 g/dL (women)or Hematocrit > 49% (men) Hematocrit > 48% (women) or increased red cell mass (RCM)c • BM biopsy showing hypercellularity for age with trilineage growth (panmyelosis) including prominent erythroid, granulocytic and megakaryocytic proliferation with pleomorphic, mature megakaryocytes (differences in size) • Presence of JAK2 or JAK2 exon 12 mutation	Subnormal serum erythropoietin level

Diagnosis of PV requires meeting of all 3 major criteria or 2 major criteria and 1 minor criteria

The factors which cause thrombosis in PV include —increased hematocrit, thrombocytosis, leukocytosis, impaired fibrinolytic activity, platelet activation, leucocyte activation, endothelial damage, interactions between platelets and endothelium, increased whole-blood viscosity and various modalities of therapy(9).

Right heart thrombi are commonly described in patients with atrial fibrillation/flutter, prolonged central venous catheters, or transvenous pacing leads(10). Although our patient did not have any such risk factors, which might have contributed to the formation of thrombus in the setting of PV. But intracardiac thrombosis and subsequent pulmonary embolism is rarely described as initial manifestation of PV. The exact incidence and prevalence of PV causing intracardiac thrombosis ad subsequent pulmonary embolism is not available in literature.

Our patient's hypercoagulable state from PV, predisposed to the intracardiac thrombus formation and thereby pulmonary embolization, which like in other case reports was treated with anticoagulation, phlebotomy and hydroxyurea

While a variety of thrombotic events can occur in patients with PV, intracardiac thrombosis has rarely been reported and is, in our opinion, likely under estimated. Routine 2DECHO should be done in patients with PV to establish the prevalence of, risk factors and treatment, intracardiac thrombosis in patients with PV.

CONCLUSION

This case report is a reminder to keep PV as a possible etiology in middle aged female patient with right atrial thrombosis and subsequent pulmonary embolism. This case report is also reminding us the need for doing 2D ECHO in patients with PV to detect any risk factors that is likely to cause intracardiac thrombosis and there by pulmonary embolism.

Funding

The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors

Conflict of interest

Authors declare there is no conflict of interest in publishing the article.

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