

Polycythemia Vera in a Young Adult: A Rare Case Report

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Abstract: Polycythemia vera is a rare clonal disorder originating in a single aberrant hematopoietic precursor cell (stem cell) in the bone marrow. Characteristic features are increased erythrocyte mass, splenomegaly, increased platelet count, and neutrophilia. PV, essentially a disease of older age (60-80), usually presents with non-specific complaints like headache, tiredness, vertigo, visual disturbances, epigastric burning or thrombotic phenomenon. We report a case of 38 year old male who presented to us with pain abdomen, pruritis, fever, headache, melena, swelling in the left upper abdomen and raised haematocrit who fulfilled the diagnostic criteria for Polycythemia Vera. Treatment strongly affects survival. Hence high index of suspicion, even in a young male, is essential in spite of the nonspecific presentation.

Keywords: Polycythemia Vera, Haematocrit, Myelo Proliferative Disorders

INTRODUCTION

Polycythemia Vera (PV), also called Polycythemia Rubra Vera, is a chronic clonal myeloproliferative disorder characterized by a striking absolute increase in the number of red blood corpuscles and in the total blood volume, and usually by leukocytosis, thrombocytosis, and splenomegaly [1]. It has been previously estimated that between 2.3 and 2.8 per 100,000 persons per year are affected [2]. It usually presents with non-specific complaints like headache, tiredness, vertigo, visual disturbances, epigastric burning or thrombotic phenomena. Pruritis is common especially after a warm bath [3]. A hallmark of polycythemia is an elevated hematocrit, with hematocrit > 55% seen in 83% of cases [4]. PV tends to be a disease of older individual, with peak incidence observed at 60 to 80 years of age [5]. Very few cases of PV have been reported in patients younger than the age of 40 years [6]. Herewith we report a case of PV in a young adult presented to us with complaints of pain abdomen, pruritis, fever, headache, melena, swelling in the left upper abdomen and raised hematocrit.

CASE REPORT

A 38 years old male presented with complaints of pain abdomen, pruritis, fever, headache, melena and swelling in the left upper abdomen for the past 2 months. There was no history of alcohol or exposure to drugs and hepatotoxic chemicals. Physical examination revealed congested conjunctiva and splenomegaly upto the level of umbilicus. Other physical examination findings were unremarkable.

The laboratory findings were as follows: Hemoglobin 23 gm% (N -13 to 17 gm%), Hematocrit

69 vol % (N -40 to 50 %) , RBC count 7.6 million/cumm (N - 4.5 to 5.5 million/cumm), Total leucocyte count 18600 cells/cumm (N -4000 to 11,000/cumm), Differential count was Neutrophils-72%, Lymphocytes -22%, Monocytes - 01%, Eosinophils - 02%, Basophils- 03%, Platelet count 5.0 lakhs/cumm (N -1.5 to 4.1 lakhs/cumm) and Reticulocyte count 0.5%(N -0.5 to 2.5%). Blood picture was Normocytic Normochromic with neutrophilic leucocytosis and thrombocytosis. RBC showed increased rouleaux formation and no hemoparasites were found. Bone marrow aspiration was done and it revealed hypercellular marrow, Erythroid:Myeloid ratio - 1:1. Erythropoiesis was markedly increased with increase in the basophilic precursors, Myelopoiesis and Megakaryopoiesis were also markedly increased.

Other investigations included Ultrasound abdomen which showed Splenomegaly, Vitamin B 12 levels 683 pg/ml (N -160 to 925 pg/ml), Oxygen saturation 98%, Serum Erythropoietin levels 18 m U/ml (N - 4 to 24 mU/ml), LAP score 84 (N - 20 to 100), LDH 515 U / L (N - 140 to 280 U/L), Serum Uric acid levels 3.9 mg/dl (N - 3.4 to 7.0 mg/dl) and Upper Gastro Intestinal endoscopy showed Gastro Esophageal Reflux Disease with Esophageal varices? Portal hypertension.

Based on the hematological findings, which fulfilled the WHO criteria [7] of Polycythemia Vera, this patient was diagnosed as PV (Table 1). The major criteria found in this patient were increased hemoglobin concentration and the two minor criteria present were bone marrow findings of panmyelosis and

erythropoietin level which was not raised. This patient was given treatment for PV which included serial phlebotomies, hydroxyurea, betablocker and

ecosprin. His symptoms and signs subsided after treatment.

Table 1: WHO criteria (2009) for Polycythemia vera

Major criteria <ol style="list-style-type: none">1. Hb > 18.5g/100ml in men or 16.5g/100ml in women or Hb > 17g/100ml in men or 15g/100ml in women if associated with 2g/100ml increase from baseline that is not attributed to correction of iron deficiency anemia. Hb or Hct > 99 percentile of reference range for age /sex/altitude or red cell mass > 25% above mean normal predicted value.2. Presence of JAK2 V617F or JAK2 exon 12 mutation.
Minor criteria <ol style="list-style-type: none">1. BM biopsy hypercellular for age with panmyelosis with prominent erythroid, granulocytic and megakaryocytic proliferation.2. Serum EPO level below the reference range for normal.3. Endogenous erythroid colony formation in vitro.

For diagnosis of polycythemia vera requires presence of both major criteria or one minor criteria or the presence of first major criteria and two minor criteria.



Fig 1: Photograph showing splenomegaly

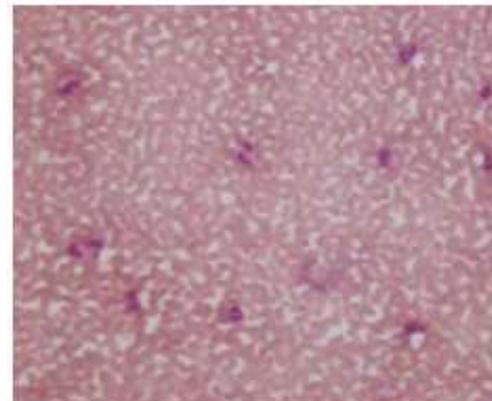


Fig. 3: Peripheral blood smear showing erythrocytosis (10X, Leishman's stain)



Fig. 2: Wintrobe tube showing raised hematocrit

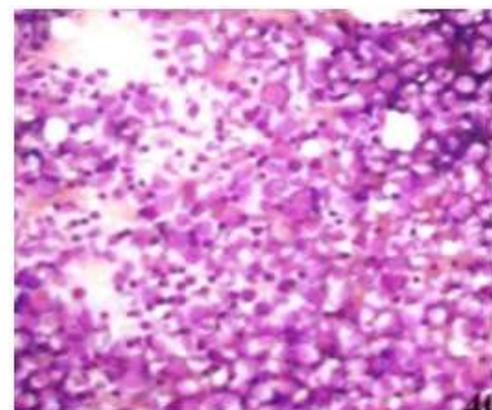


Fig. 4: Bone marrow aspirate (10X) showing hypercellular marrow with increase in all lineages

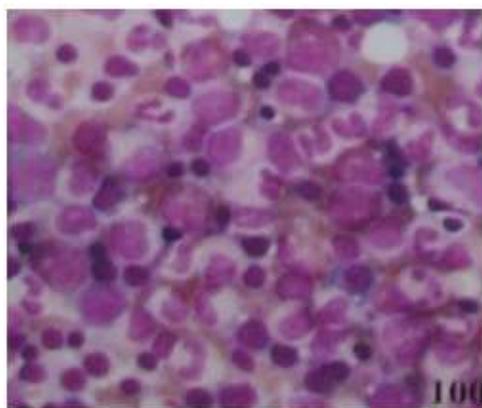


Fig. 5: Bone marrow aspirate (100X) showing hypercellular marrow

DISCUSSION

PV leads to excessive proliferation of erythroid, myeloid, and megakaryocytic elements within the bone marrow. WHO has set up criteria [7] to diagnose PV which includes, increased Hemoglobin level, JAK2 mutation, low serum erythropoietin levels, bone marrow findings of panmyelosis and in vitro formation of erythroid colony formation. A guanine to thymine mutation which results in a substitution of valine to phenylalanine at codon 617 within the pseudokinase domain (JH2) of JAK2 (JAK2V617F) is found to be the cause of PV [8].

PV differs from many other hematological malignancies, in that prolonged survival is enjoyed by most patients if the excessive production of red blood cells and platelets can be controlled [9]. Untreated patients are at particularly high risk of thrombotic and hemorrhagic events [10]. The median survival of untreated patients is reported to be 18 months [11]. Serial phlebotomies and chemotherapy are important in treatment [12].

This case was diagnosed early and treated with serial phlebotomies and hydroxyurea, which lead to marked improvement in the patients condition and also prevented development of complications pertaining to PV which signifies the importance of early diagnosis and treatment. Even though PV is more common in elderly, any patient irrespective of age when presents with symptoms and signs of increased viscosity of blood should be evaluated for myeloproliferative

disorders so that early diagnosis and treatment can be initiated for better prognosis.

REFERENCES

1. Adamson JW, Fialkow PJ, Murphy S, Prcha JFI, Steinmann L; Polycythemia vera: stem cell and probable clonal origin of the disease. *N Engl J Med.*, 1976; 295(17): 913-916.
2. Osgood EE; Polycythemia vera: Age relationship and survival. *Blood*, 1965; 26(3): 243-256.
3. Streiff MB, Smith B, Spivak JL; The diagnosis and management of polycythemia vera in the era since the Polycythemia Vera Study Group: a survey of American Society of Hematology members practice pattern. *Blood*, 2002; 99(4): 1144-1149.
4. Wallach J; Interpretation of Diagnostic Test, 7th edition, Lippincott Williams & Wilkins.
5. Modan B; An epidemiological study of polycythemia vera. *Blood*, 1965; 26(5): 657-667.
6. Najean Y, Mugnier P, Dresch C, Rain JD; Polycythemia vera in young people: an analysis of 58 cases diagnosed before 40 years. *Br J Haematol.*, 1987; 67(3): 285-291.
7. Tefferi A, Thiele J, Orazi A, Kvasnicka HM, Barbui T, Hanson CA *et al.*; Proposals for revision of the World Health Organization diagnostic criteria for polycythemia vera, essential thrombocythemia, and primary myelofibrosis: From an ad hoc international expert panel. *Blood*, 2007; 110(4):1092-1097.
8. James CI, Ugo V, Le Couédic JP, Staerk J, Delhommeau F, Lacout C *et al.*; A unique clonal JAK2 mutation leading to constitutive signalling causes polycythaemia vera. *Nature*, 2005; 434(7037):1144-1148.
9. Erf LA; Radioactive phosphorus in the treatment of primary polycythemia (vera). *Prog Hematol.*, 1956; 1:153.
10. Elliott MA, Tefferi A; Thrombosis and haemorrhage in polycythaemia vera and essential thrombocythemia. *Br J Haematol.*, 2005; 128(3): 275.
11. Chievitz E, Thiede T; Complications and causes of death in polycythemia vera. *Acta Med Scand.*, 1962; 172(5): 513.
12. Spivak JL; Polycythemia vera; myths, mechanisms, and management. *Blood*, 2002; 100(13): 4272-4290.