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A CASE OF ANEMIA WITH A PARADOX

Medicine

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ABSTRACT

Polycythemia vera is a clonal disorder involving a multipotent haematopoietic progenitor cell in which phenotypically normal red cells, granulocytes and platelets accumulate in the absence of a recognizable physiologic stimulus. It is one of the common form of chronic myeloproliferative diseases. A few patients present with its actual clinical manifestations. We present a case where polycythemia vera presenting as a paradox in the form of anemia.

On admission, the patient presented with giddiness which was of acute onset and black colored stools since one week. OGD was normal. Esophageal ultrasonography showed duodenal and peripancreatic collaterals. CT Abdomen showed chronic portal vein and superior mesentric vein thrombosis with mesentric, peripancreatic, paraduodenal collaterals. Excluding causes of thrombosis, erythropoietin levels was low, and polycythemia was then suspected. JAK2 mutation was positive, and the patient was diagnosed with polycythemia vera.

KEYWORDS

Polycythemia vera, Thrombosis, Anemia

Introduction:

Polycythemia vera (PV) is a chronic myeloproliferative disorder characterized by increased red cell mass with normal hemoglobin oxygen saturation, and may have an elevated white cell and platelet count. Low erythropoietin levels and JAK2 mutation are highly specific for PV. Here we present a case of a 40-year-old male who had anemia due to gastrointestinal bleeding was diagnosed with PV.

Case report:

A 40 year old male presented to our hospital with giddiness which was of acute onset and black colored stools since one week. He denied any history of hematemesis, abdominal distension, lower limb swelling, pruritus, dizziness, vasomotor symptoms, fatigue, and paresthesia. Past medical history was significant for diabetes and hypertension. He had a history volunteering for blood donation, 12 times in the last 5 years and on last blood donation he had Hb of 18.2 g/dl. He is on oral hypoglycaemic agents and antihypertensives since 3 years. There was no history of smoking, alcohol or illicit drug use.

On physical examination: Pulse-110/minute, feeble. BP- 90/60mmHg. Severe pallor was present. No stigmata of chronic liver disease. Cardiovascular, abdominal, and neurological examinations were normal. Laboratory data on admission are mentioned in table 1.

Table 1. Laboratory data revealed

Complete blood count	Result	Reference range
Hemoglobin (g/dL)	4.0	11.5–16.0
Hematocrit (%)	16	40.0–54.0
MCV (fL)	85.2	75.0–102.0
MCH (pg)	27.1	26.0–34.0
MCHC (g/dL)	31.9	30.5–36.0
WBC ($\times 10^9$/L)	4.60	3.40–11.00
Platelets	455×10^9/L	$130–400 \times 10^9$/L
ESR(mm/hr)	35	0-22
RBS (mg/dL)	132	<140

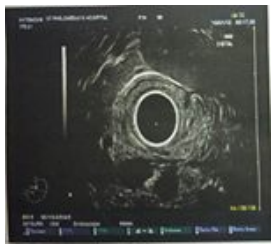
Note: MCH = mean corpuscular hemoglobin; MCHC = mean corpuscular hemoglobin concentration; MCV = mean corpuscular volume; RBC = red blood cell; WBC = white blood cell.

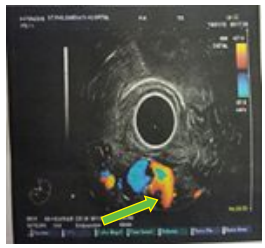
Renal function tests, liver function tests, coagulation profile was within normal limits. However stool for occult blood was positive.

His arterial blood gas (ABG) showed the following: pH, 7.37 (7.35–7.45); pCO2, 45 mmHg (30–44 mmHg); pO2, 98 mmHg (90–108 mmHg).

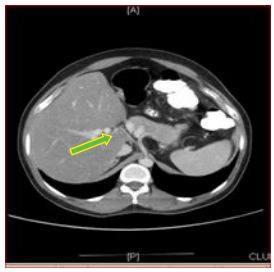
The patient was suspected to have upper gastrointestinal bleed. The plan of action was to treat anemia and to diagnose the cause of GI bleeding which was thought of.

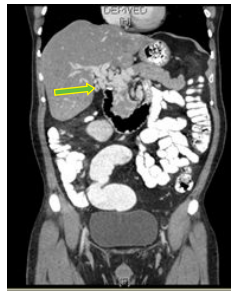
The upper GI endoscopy was normal, however the esophageal ultrasonography showed duodenal and peripancreatic collaterals.





CT abdomen showed Chronic portal vein and superior mesentric vein thrombosis with cavernoma formation and mesentric, peripancreatic, paraduodenal collaterals with few duodenal submucosal varices.





The acquired causes of thrombosis were excluded, Antiphospholipid antibodies titre were not detectable. However erythropoietin levels were reduced 3 U/L (NORMAL 10 -25 U/L). The reduced levels of erythropoietin were suggestive of polycythaemia rubra Vera. To confirm this, JAK -2 gene mutation study was done. And on PCR qualitative analysis JAK-2 mutation was detected.

Discussion:

The incidence of PV is 2.3/100,000 with a slightly higher rate of incidence among men than in women (1.2:1)[1]. The symptoms include aquagenic pruritus, headache, paraesthesia, visual disturbance, bleeding, and peptic ulcer disease[2]. The vascular complications in PV due to increased blood viscosity typically thrombocythemia including erythromelalgia, peripheral ischemia, myocardial infarction, atypical cerebral ischemic attacks, and major arterial and venous thrombotic events[3]. Polycythemia should be suspected when there is an raised hemoglobin or hematocrit on normal oxygen saturation. However, as in the case discussed above, the finding can be masked in severe anemia. Splenomegaly is the first presenting sign in PV but most often it is recognized by high haemoglobin and haematocrit levels on first presentation. Other findings also include an elevated white blood cell

count, thrombocytosis, elevated lactate dehydrogenase levels, and splenomegaly. Serum erythropoietin is to be assessed to differentiate between primary and secondary polycythemia. Low erythropoietin is suggestive of PV, which can be furthur confirmed with a positive JAK2 mutation[4].

As per 2016 revised WHO guidelines, diagnostic criteria for PV include two major and three minor criteria:[5]

1. Hemoglobin >16.5 g/dL in men and >16 g/dL in women, or hematocrit >49% in men and >48% in women, or red cell mass >25% above mean normal predicted value.
2. Bone marrow biopsy showing hypercellularity for age with trilineage growth (panmyelosis) including prominent erythroid, granulocytic, and megakaryocytic proliferation with pleomorphic, mature megakaryocytes (differences in size).
3. Presence of JAK2V617F or JAK2 exon 12 mutation.

The minor WHO criterion is as follows:
Serum erythropoietin level below the reference range for normal.

Since there is no proven drug to completely cure PV, the mainstay of treatment is phlebotomy, to keep the hematocrit level below 45%. Aspirin – 81 mg daily, Cytoreductive therapy for patients at high risk for thrombosis. Splenectomy in patients with painful splenomegaly or repeated episodes of splenic infarction. Hydroxyurea is the most commonly used cytoreductive agent. If hydroxyurea is not effective or not tolerated, alternatives include the Interferon alfa, Busulfan – (in patients older than 65 years) Ruxolitinib[6].

Very few patients present with actual symptoms. In our patient, the peculiarity was the hemoglobin was 4g/dl on initial presentation which was secondary to upper GI bleed. There was no splenomegaly both clinically and radiologically. Our case presented with unusual site of presentation of thrombosis. Polycythemia presenting as upper GI bleed is very rare.

Conflicts of interest: None.

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