

CASE REPORT

POLYCYTHAEMIA VERA PRESENTING AS A PORTA HEPATIS MASS

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Polycythaemia Vera (PV) is a myeloproliferative disorder in which bone marrow has increased production of red blood cells, white blood cells and platelets. The hallmarks of the disease are veno-occlusive events, secondary to increased blood viscosity. Polycythaemia Vera rarely presents with portal vein thrombosis below age of 55 years especially in absence of any chronic liver disease. We report a case of 30-years-old South Asian male presenting with abdominal pain, weight loss and vomiting for 3 months. On evaluation, he was found to have oesophageal varices. Furthermore, CT scan showed infiltration at porta-hepatis and portal venous thrombosis. Polycythaemia Vera was diagnosed with a positive JAK2 mutation and increased haemoglobin. Laparoscopy was done to perform biopsy of the porta-hepatis mass. Biopsy showed engorged vessels with no sign of malignancy. Patient underwent repeated sessions of upper GI endoscopy for band ligation and multiple sessions of venous phlebotomy which drastically improved his blood indices. He was started on lifelong aspirin and was advised regular follow-ups. With early recognition and prompt management patients can be prevented from potential complications which can prove to be detrimental.

Keywords: Polycythaemia Vera; Portal vein thrombosis; Varices; Splenomegaly

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INTRODUCTION

Polycythaemia Vera (PV) is a chronic myeloproliferative neoplasm (MPN) defined by clonal proliferation of myeloid cell lines with variable morphologic maturity.¹ Polycythaemia Vera is differentiated from other MPNs by the presence of an increased red blood cell mass independent of erythropoietin levels. We report a patient who was managed by a multidisciplinary team of Gastroenterologists, Haematologists and Radiologists after presenting with an unusual set of symptoms for PV.

CASE REPORT

A 30 years old gentleman, ex-smoker with prior no known co morbid presented with history of abdominal pain, decreased appetite, 7 kg weight loss, one episode of hematemesis in the past three months. On physical examination he showed signs of wasting and dehydration. On abdominal examination he had an enlarged spleen. The haematological workup showed Haemoglobin of 19.5g/dl, RBC count of $7.63 \times 10^{12}/L$ and Normal Erythropoietin of 3.29 mIU/ml. Rest of the blood work up was normal. Upper-gastrointestinal endoscopy showed large oesophageal varices and severe congestive portal gastropathy.

CT scan showed ill-defined soft tissue mass like infiltration surrounding the porta-hepatis. There was massive splenomegaly, portal vein thrombosis with cavernous transformation and extensive varices at gastro-oesophageal region. No

evidence of liver cirrhosis was appreciated. The Ultrasound guided biopsy showed fragments of mature adipose tissue with thick walled blood vessels and numerous capillary sized engorged vascular channels. No evidence of malignancy was seen. On further testing he was found to be positive for JAK2V617F gene mutation. Bone marrow biopsy was refused by the patient.

He underwent repeated sessions of upper GI endoscopy for band ligation along with venous phlebotomy after which his haematocrit went down below 40%. He was started on aspirin and advised to continue regular follow-ups. Ethical approval was obtained from the patient for use of details for research purposes.



Figure-1: Gross presentation showing hyper vascularization at porta hepatis



Figure-2: CT-scan showing mass at porta hepatis and portal vein thrombosis

DISCUSSION

Polycythaemia Vera rarely presents in thirties and can have a wide range of clinical symptoms including erythromelalgia, pruritis, fatigue, myocardial infarction, stroke and thrombosis. Patients with PV have an increased risk of thrombosis (e.g., stroke, myocardial infarction, deep vein thrombosis, pulmonary embolus) and hemorrhage². Acute thrombosis can be asymptomatic or can result in bleeding of varices and shock. Chronic thrombosis can present with splenomegaly, ascites, gastro oesophageal varices, and chronic gastrointestinal bleeding.³ High index of clinical suspicion is required to consider Polycythaemia Vera as a possible cause of non-cirrhotic portal hypertension which can present as upper GI bleeding.

It is associated with gain of function mutation of JAK2 (Janus Kinase) V617F gene and it is found to be positive in more than 95% of PV cases as agreeing to our case result.⁴ A porta-hepatis mass was seen on CT scan which raised suspicion of an extra medullary hematopoietic site. Liver and spleen are the two most common sites involved. Other porta hepatis masses can include Portal vein cavernous,

portal vein aneurysm, hepatic artery thrombosis and stenosis, hilar cholangiocarcinoma, choledochal cyst and schwannoma of liver to name a few.⁵ Phlebotomy and Anticoagulants are the mainstay of treatment to reduce blood viscosity and the risk of thromboembolism. Phlebotomy-induced maintenance of target haematocrit levels of less than 45% are associated with significantly lower cardiovascular and major thrombotic diseases morbidities.⁶ There are no true guidelines concerning the optimal haematocrit level in patients with PV. Other treatment options include Hydroxyurea (HU) a cytoreductive drug and beta blockers.⁷ In conclusion, abdominal mass/thrombosis can be the initial manifestation of myeloproliferative disease and clinicians should keep a high index of suspicion for PV to reduce morbidity and mortality in such cases.

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