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Available online at: http://www.iajps.com *A Case Report*

PORTAL VEIN CAVERNOUS TRANSFORMATION IN A PATIENT WITH ESSENTIAL THROMBOCYTHEMIA: A RARE AND INTERESTING PRESENTATION

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Abstract:

Essential thrombocythemia is a myeloproliferative disorder characterized by an elevated platelet count, with symptoms such as vasospasm and a predisposition to bleeding and microvascular complications. While rare, one complication that can arise from chronic portal vein thrombosis is the cavernous transformation of the portal vein. In this case report, we explore a unique presentation of cavernous transformation of the portal vein (CTPV) in a patient diagnosed with essential thrombocythemia (ETC). This case report gives importance to considering myeloproliferative disorders like ETC as a potential cause of chronic portal vein thrombosis leading to the CTPV.This case report describes a 26-year-old male patient diagnosed with cavernous transformation of the portal vein (CTPV) during a workup for hematemesis and melena. The patient was found to have severe anemia, thrombocytosis, splenomegaly, and a positive JAK2 mutation, consistent with a diagnosis of essential thrombocythemia. Although thrombotic events in essential thrombocythemia patients are common, portal vein thrombosis and CTPV are rare complications. Cavernous transformation of the Portal vein is a condition that is seen in the setting of long standing portal vein thrombosis. There are diverse causes to be considered when chronic portal vein thrombosis leading to cavernous transformation is encountered clinically and radiologically. In this case report the predisposing condition was found to be essential thrombocythemia. The article emphasizes the importance of considering cavernous transformation of the portal vein (CTPV) as a complication in patients with essential thrombocythemia.

Keywords: *Essential thrombocythemia, Portal vein thrombosis, myeloproliferative neoplasms, Cavernous transformation of the portal vein, case report.*

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INTRODUCTION:

Essential thrombocythemia (ETC) is a chronic disorder characterized by increased platelet counts. Patients with ETC generally have a normal life expectancy. Management of ETC focuses on assessing the risks of thrombohemorrhagic events. High-risk patients undergo platelet reduction, while those at intermediate risk are managed based on the severity of other risk factors, particularly cardiovascular factors. Low-risk patients are typically observed without active intervention[1]. As per the World Health Organization (WHO) guidelines, Essential Thrombocythemia (ETC) can be diagnosed based on the following criteria: platelet counts equal to or greater than 450 × 10^9/L, presence of three driver mutations (JAK2, CALR, or MPL), or absence of other causes of thrombocytosis (reactive or clonal) along with bone marrow morphologic assessment[2].

Arterial or venous thrombosis is the primary cause of morbidity and mortality in (ETC). Thrombosis can be the initial symptom that leads to the diagnosis, with an estimated incidence of around 14% at 10 years and a prevalence of 10% to 35% at diagnosis. Microvascular complications such as erythromelalgia, migraine, and paresthesia may also occur. Paradoxical bleeding can occur in cases of extreme thrombocytosis. Treatment options include Aspirin for reducing thrombosis risk and providing symptomatic relief, as well as cytoreductive therapies like Hydroxycarbamide, Interferon-alfa, and Angrelide[3].

In the past, CTPV was considered rare and had limited literature, mostly consisting of clinical series and case reports. Common symptoms of CTPV include bleeding from gastro-esophageal varices, enlarged spleen, and portal biliopathy after a gradual and prolonged presentation.

We report a case of cavernous transformation of the portal vein in 26 years old male secondary to ETC who initially presented with complaints of fever, hematemesis, melena and abdominal pain. This paper signifies the importance of considering essential thrombocythemia, as a cause of non-cirrhotic portal hypertension due to chronic portal vein thrombosis, which can manifest radiologically as cavernous transformation of the portal vein.

CASE REPORT:

A 26-year-old male presented to the accident and emergency department of Khyber Teaching Hospital in Peshawar, Pakistan with complaints of fever, vomiting, and abdominal pain. According to his history of presenting illness, he had been experiencing hematemesis and melena intermittently for the past month. His symptoms worsened a week prior to presentation, with the onset of abdominal pain, nausea, vomiting, anorexia, and fever for the past two days. The patient reported a similar episode three years ago but had no other co morbidities or surgical history, and no family history of similar conditions. He denied any regular medication use and had no history of narcotics use. On general physical examination, he appeared lethargic, had white nails, palmar and conjunctival pallor, and mildly malnourished. Abdominal examination showed mild distension in the abdomen, epigastric tenderness, splenomegaly on palpation, dull percussion notes in the abdomen due to ascites and positive shifting dullness. Heart sounds were normal without any added sounds. Nervous system examination was normal. Respiratory system examination revealed equal air entry bilaterally, without any abnormal breath sounds.

The vital signs were recorded and monitored. His blood pressure was 100/60 mmHg, pulse was 104 beats per minute, temperature was 101°F, and oxygen saturation was 96% on room air. Lab results showed severe anemia with a hemoglobin level of 5.3 g/dL and MCV of 61 fL, a white blood cell count of 8.89, and an elevated platelet count of 1.076 million. His viral profile, including HIV, was negative, and liver function tests showed a total bilirubin of 0.77 mg/dL

and ALT of 20.5 U/L. Renal function tests showed a creatinine level of 0.76 mg/dL. His serum electrolyte levels showed serum sodium of 144 mmol/L and potassium of 5.1 mmol/L. The coagulation profile revealed a prothrombin time (PT) of 16 and an International Normalized Ratio (INR) of 1.4.

The patient was admitted for a workup of hematemesis, severe anemia, and thrombocytosis. After initial resuscitation with blood transfusion and intravenous fluids, an ultrasound was performed, which showed, splenomegaly and ascites. However, the portal vein was not visualized, and instead, a few venous channels with increased echogenicity, consistent with cavernous transformation of portal vein and an enlarged hepatic artery were seen in the porta hepatis. Further imaging included ordering a CT scan that revealed splenomgaly.

For the workup of the hematemesis, the patient underwent upper GI endoscopy, which revealed bleeding esophageal varices. Band ligation was applied to treat the varices. Additionally, for the workup of the elevated platelet count (thrombocytosis), a bone marrow biopsy was performed, which showed a cellular marrow with markedly increased megakaryocytes. PCR for JAK2 mutation and CARL and MPL were recommended and the PCR for JAK2 mutation came back positive. The possibility of iron deficiency anemia (IDA) leading to reactive thromobcytosis was therefore, ruled out as IDA as a primary diagnosis, is not associated with the presence of JAK2 mutation. The patient was diagnosed as a case of essential thrombocythemia (ETC).

Over the course of the patient's admission, the patient was stabilized, and the anemia was corrected with two pints of blood transfusion. Furthermore, to reduce the risk of thrombosis, the patient was started on Tab. Aspirin 75mg one tablet daily, Tab. Clopidogrel 75mg one tablet daily and Tab. Hydra 500mg 1 tablet, two times daily. The patient was then referred to a hematologist for further care.

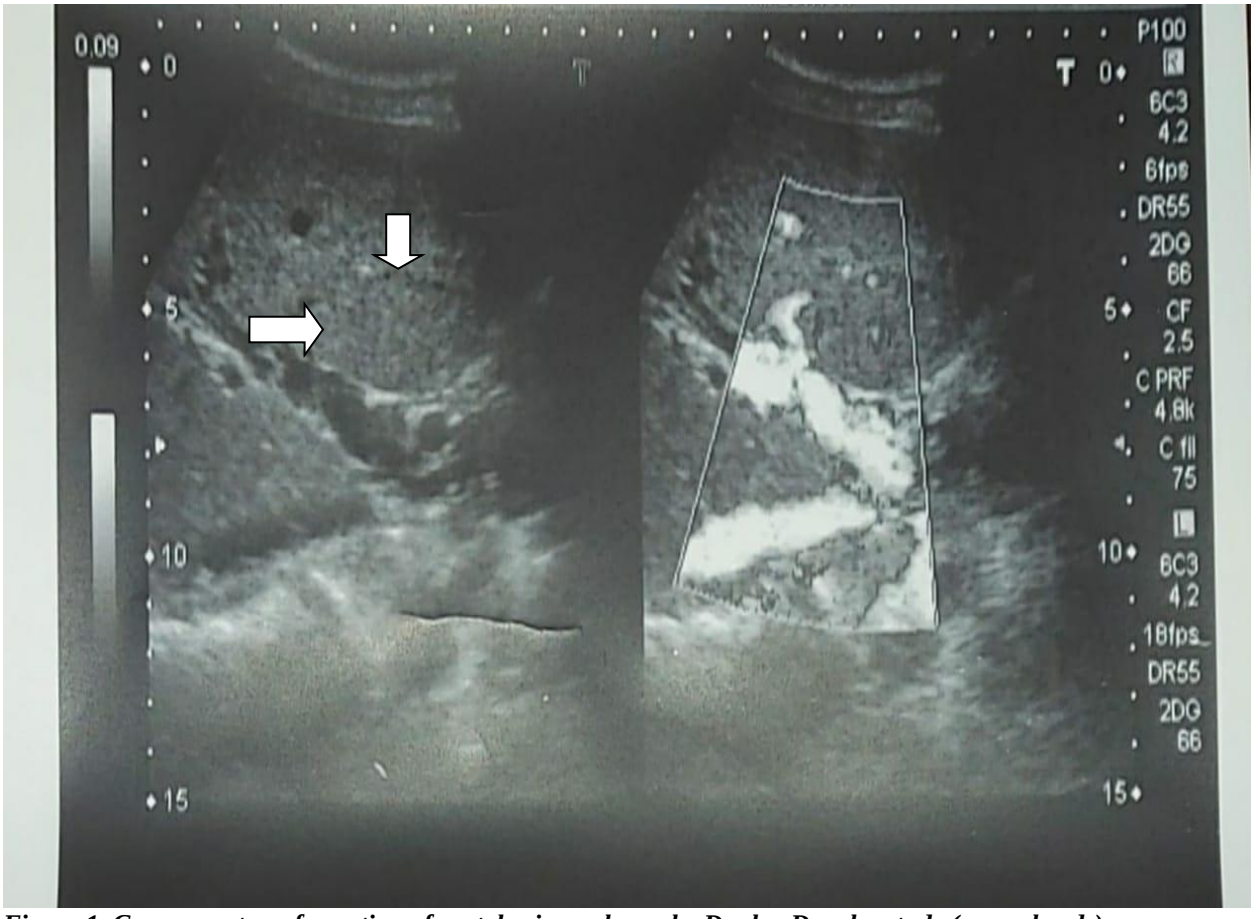

Figure 1. Cavernous transformation of portal vein as shown by Duplex Doppler study (arrow heads)

DISCUSSION:

Cavernous transformation of the portal vein (CTPV), a rare condition, has multiple causes and presents in diverse ways. It arises from prolonged portal vein thrombosis (PVT), resulting in portal hypertension and blockage of the portal vein. This leads to the formation and expansion of numerous small vessels around the recanalizing primary portal vein. CTPV is frequently observed in patients with non-cirrhotic, non-tumoral PVT and healthy liver[4]. Cavernous transformation of the portal vein rarely causes symptomatic biliary obstruction, making it an exceptionally uncommon condition that can be managed endoscopically[5]. In cases of complete extrahepatic portal vein obstruction (EHPVO), fibroblasts transform the clot into a solid, collagenous plug, leading to the development of twisted venous channels[6]. Confirmation of diagnosis depends upon abdominal ultrasonography, color Doppler ultrasound (US), computed tomography (CT) angiography, and magnetic resonance imaging (MRI)[7].

The patient in our study presented with symptoms that normally suggest cirrhotic changes in the liver leading to increased portal venous pressure and the development of complications such as bleeding esophageal varices. However, ultrasound studies of the liver showed no fibrotic or cirrhotic features. Instead, Doppler studies confirmed the presence of cavernous changes in the portal vein, leading to a diagnostic dilemma. This dilemma was ultimately resolved upon reaching a final unifying diagnosis of essential thrombocythemia (ETC), which possibly gave rise to a chronic portal vein thrombus, causing the resulting cavernous transformation.

A recurring somatic activating mutation in the JAK2 tyrosine kinase is found in hematopoietic cells of a significant number of patients with ETC. Blocking the JAK2V617F kinase with a small molecule inhibitor leads to the suppression of hematopoietic cell proliferation, indicating that targeting the JAK2 tyrosine kinase could be a promising approach for pharmacological intervention in patients with ETC[8].

Non-cirrhotic portal vein thrombosis is a condition in which thrombosis occurs in the portal veins without any evidence of cirrhosis. This condition has been reported in conjunction with various genetic and medical disorders such as malignancies, prothrombotic states, and infections. Multiple risk factors have been linked to this condition, including the use of oral contraceptive pills, previous abdominal surgeries or trauma, and acquired or congenital disorders of the clotting system. Furthermore, the condition can also occur without any identifiable cause[9].

Studies exploring this relationship are few and far between. A study by Jun Murakami did shed some light on the matter. It discovered an association ETC and liver abnormalities. It turns out that patients with ETC have a higher prevalence of liver abnormalities[10].

Another case report published by Xiao-Yan Cai, has also shown essential thrombocythemia causing chronic portal vein thrombosis leading to CTPV. For individuals with portal cavernoma, if typical reasons like cirrhosis and neoplasm are not identified, it is advisable to explore the possibility of an unusual cause of myeloproliferative disorder (MPD) through bone marrow biopsy and coagulation profile assessment. It's crucial to understand that the characteristics of (ETC) in blood cell counts may be concealed due to hypersplenism[11]. The case we presented here was different and unique, as there were no features of hypersplenism and the platelet count was appropriately high.

The approaches for managing CTPV consist of various methods such as medication, endoscopic interventions like sclerotherapy and band ligation, surgical procedures such as splenectomy, surgical shunt, and liver transplantation, as well as TIPS (Transjugular Intrahepatic Portosystemic Shunt)[12].

Portal vein thrombosis, a captivating condition that can occur in both cirrhotic and non-cirrhotic settings, is a medical enigma that has puzzled researchers and continues to fascinate the scientific community. While the connection between ETC and PVT remains a bit murky, findings from the International Journal of Hepatology hint at a potential link.

CONCLUSION:

The development of venous channels around or within a thrombosed portal vein is termed as, cavernous transformation of the portal vein (CTPV). Many different causes can increase the likelihood of thrombus formation in the portal vein. One of these conditions is essential thrombocythemia (ETC). Appropriate workup thus becomes very important to rule out ETC as a potential cause of the portal vein thrombosis leading to the CTPV.

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Consent: Informed consent was obtained from the respondents before filling out the questionnaire.

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