

Case Report

A young male with liver cirrhosis and portal hypertension found to have chronic idiopathic Budd Chiari syndrome

Dhulashiha Jegavanthan¹, Keerthi Kularatne¹, Indika Wickramatunga¹, Dineesha Kumarathunga¹, Suthendran Roeshanathan¹

¹Teaching Hospital Kandy, Sri Lanka

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Corresponding Author: Dhulashiha Jegavanthan, E-mail: dhulashi22@gmail.com>  <http://orcid.org/0000-0001-6671-4215>

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Introduction

Budd Chiari syndrome is an uncommon condition associated with hepatic out flow obstruction leading to hepatic congestion and microvascular ischaemia resulting in hepatocellular injury. Typically, it presents in the third and fourth decades of life and it is rare in children and in the elderly. Although a thrombotic diathesis is the cause in one third of patients, in most cases it is idiopathic. Budd Chiari syndrome classically presents with the triad of ascites, abdominal pain and hepatomegaly.

We present a case of a young man diagnosed with idiopathic Budd-Chiari syndrome, who went on to develop cirrhosis of the liver and portal hypertension.

Case presentation

A 20-year-old Sri Lankan male presented with progressive abdominal distension and abdominal pain of two months duration and loss of weight over three weeks despite a good appetite. His systemic inquiry was unremarkable. There was no significant past medical, surgical or allergic history. He is a tailor by profession and never married; He has two siblings who are healthy.

On examination he was dark in complexion and appeared well. He was not pale, or icteric. There was no clubbing or peripheral stigmata of chronic liver cell disease. There were no hepatic flaps or dependent oedema. Abdomen was distended with moderate, firm hepatomegaly and ascites. There was no hepatic bruit or splenomegaly. His cardiovascular, respiratory and neurological examinations were normal.

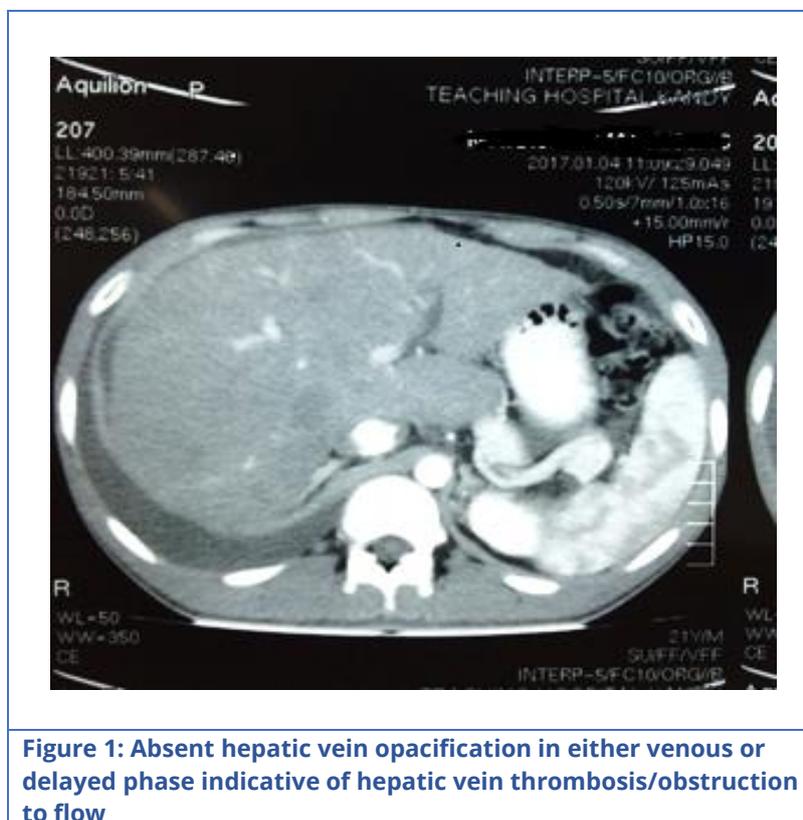
Routine investigations revealed a haemoglobin of 15.9 g/dL with a MCV of 83 fL. The total leucocyte count was $10.06 \times 10^9/L$ with normal differentials and platelet count of $313,000/mm^3$. Blood film showed acanthocytes and target cells which was suggestive of a liver pathology. Liver function tests showed an elevated alkaline phosphatase level of 319 U/L with normal alanine and the aspartate aminotransferases (AST= 21 IU/L, ALT=26 IU/L) and a normal serum bilirubin concentration. The total serum protein was 6.5g/dL, with albumin levels of 3.3g/dL. The Gamma GT level was mildly elevated (58.5 U/L).

Prothrombin time was 16.3 seconds with an INR of 1.43. Serum electrolytes and renal function tests were within normal limits. The inflammatory markers were normal. Peritoneal fluid examination showed 40 cells/mm³ and a very high protein level of 8g/dL with negative acid-fast bacilli (AFB). Peritoneal fluid cytology was negative.

Ultrasonography of the abdomen revealed a hepatomegaly of 18.2cm with an enlarged caudate lobe and an architecture characteristic of chronic parenchymal liver disease. There was moderate ascites and bilateral pleural effusions. There was evidence of early portal hypertension. The initial colour doppler studies failed to show any abnormalities. Liver biopsy revealed effaced architecture with the liver parenchyma was separated into nodules by thick fibrous septae. Hepatocytes showed ballooning and degeneration. There was no steatosis, bile stasis, necro-inflammation or interphase hepatitis. These findings were compatible with established liver cirrhosis.

He had negative hepatitis B and C serology. Retroviral screening, Mantoux test and sputum AFB were negative. His serum iron studies, ferritin, ceruloplasmin level and 24hour urinary copper excretion were all within normal limits. Kayser Fleischer (KF) ring was not found on slit lamp examination of his eyes. Anti-nuclear factor was negative. His ECG and chest X-ray were normal. Echocardiogram showed a thin rim of pericardial effusion but was otherwise normal.

Contrast enhanced CT abdomen confirmed the radiological diagnosis of Budd Chiari syndrome with evidence of caudate lobe enlargement and hepatic vein thrombosis without involvement of the splenic vein, inferior vena cava or right atrium (Figure 1).



Upper gastrointestinal endoscopy showed the presence of oesophageal varices and therapeutic bands were applied. Anticoagulation with an INR target of 2 to 3 was started. Simultaneously an exhaustive thrombophilic screening was performed and all possible thrombophilic conditions including JAK-2 mutation and paroxysmal nocturnal haemoglobinuria (PNH) were excluded. Malignancy was negative by imaging and serological studies and chronic infectious and inflammatory causes were excluded. A diagnosis of chronic idiopathic Budd-Chiari syndrome was made.

Although ultrasound guided hepatic vein stenting was performed, the hepatic flow was not fully established (Figure 2).

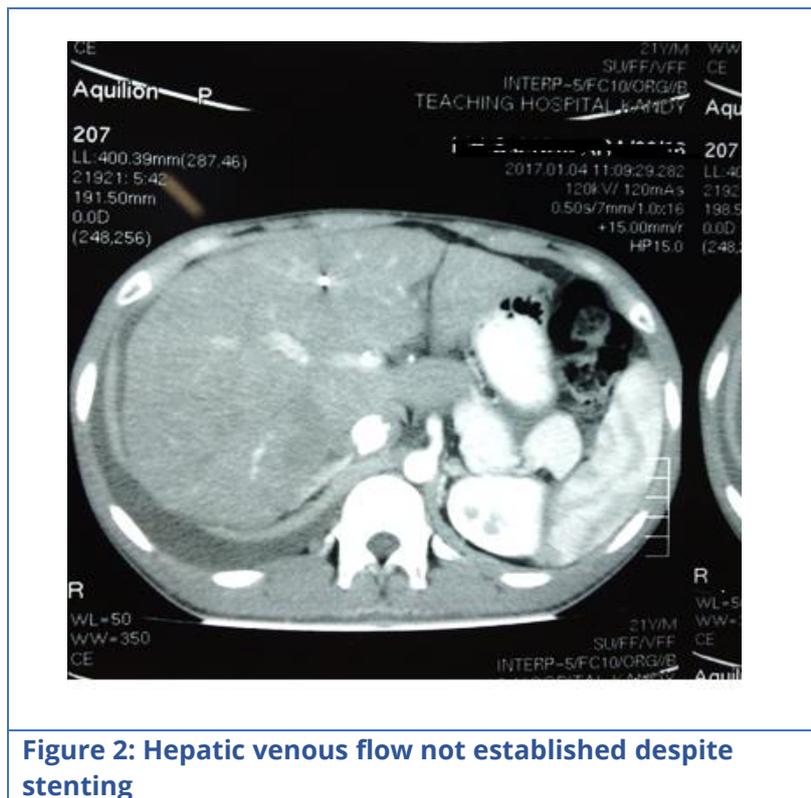


Figure 2: Hepatic venous flow not established despite stenting

Thereafter, on the patient's request, he was continued to be treated medically with sodium restriction, diuretics to control ascites, anticoagulation and general symptomatic management.

Discussion

Budd-Chiari syndrome is characterised by hepatic venous outflow obstruction. It is a potentially life-threatening disorder caused by obstruction of the hepatic outflow tract at any level between the junction of the inferior vena cava with the right atrium and the small hepatic veins, thrombosis or its fibrous sequelae [1]. Hepatic veno-occlusive disease and cardiac disorders are excluded from this definition. George Budd, a British internist, described three cases of hepatic vein thrombosis due to abscess-induced phlebitis in 1845, and Hans Chiari, an Austrian pathologist, added the first pathologic description of a liver with "obliterating endophlebitis of the hepatic veins" in 1899 [2]. This rare disease

affects mainly young adults [3] and it is estimated that 1.4 per million people are affected.

Presentation depends on the extent and rapidity of hepatic vein occlusion [4]. It is caused by both thrombotic and non-thrombotic conditions. Hepatic venous outflow obstruction involving the large hepatic veins is usually thrombotic and isolated obstruction of the inferior vena cava or of the small hepatic veins is usually non-thrombotic [5].

A hypercoagulable state is identified in 75% of patients. More than one aetiological factor may play a role in 25% of patients. Primary myeloproliferative diseases are the leading cause of this condition and is found in almost 50% of patients [6]. The recent discovery of the JAK2 mutation has significantly contributed to the increased diagnosis of myeloproliferative disorders [7]. However, it is now recognized that almost half the patients have multiple underlying prothrombotic risk factors [1].

Budd Chiari syndrome presents with heterogeneous clinical manifestations. Presentations range from acute liver failure to completely asymptomatic patients. The classic triad of abdominal pain, ascites and hepatomegaly is commonly present in patients, with abdominal pain presenting in 61%, ascites in 83% and hepatomegaly in 67% [1]. Other clinical features include fever, pedal oedema and dilated truncal veins. Less common clinical manifestations include oesophageal bleeding (5%) and hepatic encephalopathy (9%) [3]. Around 20% of patients are completely asymptomatic.

Doppler ultrasonography is usually sufficient to confirm the diagnosis, although tomographic imaging (CT) or magnetic resonance imaging (MRI) is often necessary for further [8]. Myeloproliferative neoplasms should be actively screened for even when a clear causative factor has been identified [4].

The prognosis is poor in patients with Budd-Chiari syndrome who remain untreated, with death resulting from progressive liver failure in 3 months to 3 years from the time of the diagnosis [9]. The management needs an individualized multidisciplinary approach. A stepwise therapeutic strategy has been proposed where anticoagulation, correction of risk factors, diuretics and prophylaxis for portal hypertension are used first. Angioplasty is used for short-length venous stenosis. Trans jugular intrahepatic portosystemic shunt (TIPS) is indicated in a subgroup of patients with progressive liver disease. It is safe, feasible and improves survival. Final treatment options include liver transplantation. Treatment progression is dictated by the response to previous therapy. This strategy has achieved 5-year survival rates approaching 90%. Medium-term prognosis depends on the severity of liver disease [3].

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