Budd-Chiari Syndrome: Diagnosis Achieved with High Clinical Suspicion Raised by EUS-Guided Portal Pressures and Liver Biopsy

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Introduction

Budd Chiari syndrome (BCS) is a rare syndrome due to hepatic venous obstruction in the absence of cardiac cause, mostly secondary to thrombosis. As high as 80%of patients present with ascites. The diagnosis should be suspected in patient with acute or chronic liver disease without identified cause. Venous obstruction can be seen on Doppler ultrasound, CT, or MRI of the hepatic veins and inferior vena cava. In some cases, hepatic venogram is needed for its diagnostic and therapeutic benefits. We report a 50-year-old woman with BCS who underwent successful wire recanalization, angioplasty and thrombolysis of the right hepatic vein.

Case presentation and course

50-year-old woman with no significant past medical history presented with abdominal distension and bilateral lower extremity swelling of two weeks duration. On exam, abdomen was distended with positive signs of fluid collection with +3 bilateral lower extremity edema. Labs revealed total bilirubin 3.3 direct bilirubin 1.4, ALP 477, ALT 38, AST 96, PT 14.4, INR 1.35

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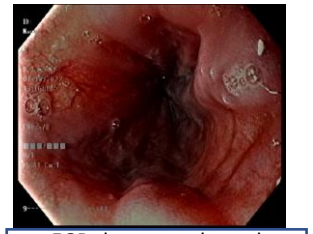
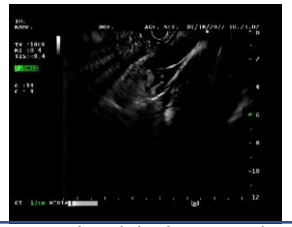
Ultrasound showed Heterogeneous appearance of liver and large volume ascites. Doppler study showed normal hepatic and portal venous flow.
Abdominal MRI showed hepatic steatosis and caudate lobe hypertrophy. Large volume paracentesis was done with analysis showing SAAG of 3.1, WBC 364, Neutrophils 8.

EGD revealed large esophageal varices. Viral hepatitis panel, ceruloplasmin, alpha-1antitripsin, ASMA, AMA antibodies were negative. EUS guided liver biopsy and portal pressure measurement was done showing venous pressure gradient of 13 consistent with portal hypertension. Liver biopsy showed grade 2 fibrosis and zone 3 congestion. Hepatic venogram showed chronically occluded right hepatic vein on which successful wire recanalization was done.

Pt was worked up for hypercoagulable state and was found to have JAK 2 mutation and Protein C deficiency. Apixaban, Beta blocker and diuretics were started. Patient showed significant improvement with ascites and MELD score.



EGD: large esophageal varices



EUS: Caudate lobe hypertrophy and compression of IVC

Liver biopsy: Zone 3 congestion

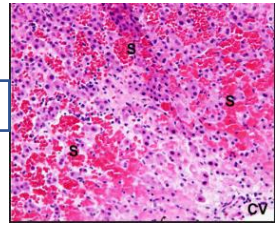


Table: prothrombotic risk factors

A. Acquired thrombophilia	B. Inherited thrombophilia	C. Systemic factors	D. Hormonal factors
• Myeloproliferative disorder • Polycythemia vera • Essential thrombocytosis • Idiopathic myelofibrosis • JAK 2 mutation • PNH • Hyperhomocysteinemia	• Factor V Leiden • Prothrombin gene G20210A mutation • MTHFR C677T mutation • Thalassemia • Protein C deficiency • Protein S deficiency • Antithrombin deficiency	-Sarcoidosis -Vasculitis -Behcet's disease -Connective tissue disease -Inflammatory bowel diseases	-Recent OCP use -Pregnancy

Discussion

First line investigation for diagnosis of BCS is doppler ultrasound. Hepatic venous obstruction is present in around 80% of patients investigated by ultrasound alone. Contrast-enhanced computed tomography (CT) or magnetic resonance imaging (MRI) can be used to confirm the diagnosis. Doppler US and MRI venography didn't reveal the hepatic venous thrombosis in our patient. However, high index of suspicion, finding of elevated IVC pressure on EUS, and caudate lobe hypertrophy found on imaging as well as congestion liver biopsy findings led us to further investigate with hepatic venogram which confirmed the diagnosis and provided therapeutic benefits.
An underlying hypercoagulable condition can be identified in over 80% of patients with Budd-Chiari syndrome and more than one risk factor is identified in about a quarter of patients.
According to a large population based in the US, the most common risk factors were (7.75%) myeloproliferative disorder (essential thrombocythemia, polycythemia vera, myelofibrosis, chronic myeloid leukemia) followed (7.32%) by a hypercoagulable state (primary thrombophilia, protein C deficiency, factor V Leiden mutation, antiphospholipid antibody syndrome or prothrombin gene mutation)
In our patient, Workup for hypercoagulable state was positive for JAK2 mutation and protein C deficiency.

References

1. van Wettere M, Bruno O, Rautou PE, Vilgrain V, Ronot M. Diagnosis of Budd–Chiari syndrome. Abdominal Radiology. 2018 Aug 1;43(8):1896–907.
2. Alukal JJ, Zhang T, Thuluvath PJ. A Nationwide Analysis of Budd–Chiari Syndrome in the United States. J Clin Exp Hepatol. 2021 Mar 1;11(2):181–7.
3. Simonetto DA, Singal AK, Garcia-Tsao G, Caldwell SH, Ahn J, Kamath PS. ACG Clinical Guideline: Disorders of the Hepatic and Mesenteric Circulation. Vol. 115, American Journal of Gastroenterology. Wolters Kluwer Health; 2020. p. 18–40.
4. Darwish Murad S, Plessier ; Aurelie, Manuel Hernandez-Guerra ;, Fabris F, Chundamannil ;, Eapen E, et al. Etiology, Management, and Outcome of the Budd-Chiari Syndrome [Internet]. 2009. Available from: www.annals.org