Thrombophilia-of-unknown-origin-induced Budd-Chiari syndrome

Ioannis Gavalas, MD[1]; Nikoleta Kakoulidou, MD[1]; Orestis Karatzis, MD[1]; Dimos Foutsitsakis, MD[1]; Zacharo Sachla, MD[1]
[1]Department of Internal Medicine, General Hospital G. Papanikolaou, Thessaloniki, Greece

ABSTRACT

Budd–Chiari syndrome is a rare disorder characterized by hepatic venous outflow obstruction, with an estimated annual incidence of 1–2 cases per million, most commonly associated with underlying prothrombotic conditions such as myeloproliferative neoplasms, antiphospholipid syndrome, and paroxysmal nocturnal hemoglobinuria. We present the case of a 65-year-old woman with a history of recurrent venous thromboembolism, including deep vein thrombosis and recent cerebral venous sinus thrombosis, who presented with a presyncopal episode and right upper quadrant pain. Initial laboratory tests revealed deranged liver function, and abdominal ultrasound demonstrated subacute thrombosis of the splenic–portal axis. Further imaging with triplex ultrasound, CT, and MRA/MRV confirmed extensive thrombosis involving the hepatic veins, portal vein and its intrahepatic branches, as well as the splenic and superior mesenteric veins, raising strong suspicion for an underlying yet unidentified prothrombotic disorder despite negative thrombophilia testing. The patient's clinical course rapidly deteriorated, with progressive liver dysfunction, development of hepatorenal syndrome, anuria, and hemodynamic instability requiring vasopressor support, necessitating transfer to a specialized center for consideration of direct intrahepatic portosystemic shunt (DIPS). This case highlights the importance of considering Budd–Chiari syndrome in patients with unexplained hepatic dysfunction and a history of thrombosis, as early recognition and timely intervention are critical to prevent acute liver failure and improve clinical outcomes.

CONTACT

Ioannis G. Gavalas
General Hospital G. Papanikolaou
Email: john.g.gavalas@gmail.com
Phone: 6974747326

INTRODUCTION

Budd-Chiari syndrome is a rare clinical condition characterized by occlusion or obstruction of the hepatic veins, with an estimated annual incidence 1-2 cases per million. In most cases a pro-thrombotic cause is identified, which may be myeloproliferative neoplasm, antiphospholipid syndrome, Paroxysmal Nocturnal Hemoglobinuria (PNH) and other acquired or inherited thrombophilic disorders. Herein we present a case of a woman with Budd-Chiari syndrome and a history of possible thrombophilia.

CASE DESCRIPTION

A 65-year-old woman with a history of two episodes of deep vein thrombosis, recent cerebral venous sinus thrombosis and negative thrombophilia tests attended the emergency department with presyncopal episode. The physical examination revealed Right Upper Quadrant (RUQ) pain. Due to deranged Liver Function Tests (LFTs) an abdominal ultrasound was performed, which demonstrated thrombosis of the splenic–portal axis of subacute onset.

CLINICAL HYPOTHESIS

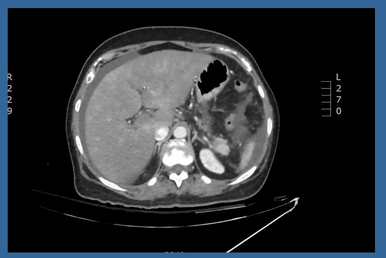
The findings were highly suggestive of an underlying, yet unidentified, pro-thrombotic condition predisposing the patient to recurrent venous thromboses.

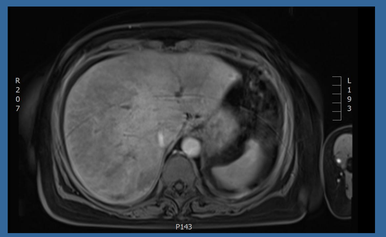
DIAGNOSTIC PATHWAYS

An abdominal Ultrasound (US) Triplex, Computer Tomography (CT) and Magnetic Resonance Angiography-Magnetic Resonance Venography (MRA-MRV) were conducted, revealing thrombosis of hepatic veins, portal vein and its intrahepatic branches, splenic and superior mesenteric vein. During hospitalization, the patient's condition was constantly deteriorating with rapid increase of liver enzymes, appearance of hepatorenal syndrome, anuria and hemodynamic instability with need of vasopressors. The patient was admitted to specialized center for DIPS procedure.

DISCUSSION AND LEARNING POINTS

Budd–Chiari syndrome should be suspected in patients with unexplained hepatic dysfunction and a history of thrombosis. Early diagnosis and timely intervention are vital to prevent liver failure and improve outcomes.





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