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A RARE CASE OF BUDD CHIARI SYNDROME SECONDARY TO ANTI PHOSPHOLIPID ANTIBODY SYNDROME AND INHERITED THROMBOPHILIA

Saikrishna Katepally

Gandhi Hospital, Hyderabad, India

Background: Budd Chiari Syndrome is a heterogenous condition characterised by hepatic venous outflow obstruction at the level of hepatic veins and/or inferior vena cava.
Case Report: A 20 years old, non-alcoholic male presented with progressive abdominal distension, hematemesis and jaundice since 4 weeks with H/O similar complaints twice in the past 1 year for which he was hospitalised and managed conservatively with medications and blood transfusions On examination pallor+, icterus+, clubbing+, Absent pubic and axillary hair and testicular atrophy noted. Abdomen distended, ascites present. Engorged veins noted over anterior abdominal wall and flanks with direction of flow of blood below upwards and away from umbilicus. Hepatosplenomegaly: present patient was evaluated for the cause of cirrhosis of liver and screened for viral markers, Wilson's disease, autoimmune hepatitis are found to be negative. Color Doppler of hepatic veins revealed non-visualization of hepatic veins. MRI abdomen with MR venogram suggested: CLD with cirrhosis of liver, portal hypertension with ascites, caudate lobe hypertrophy causing retrohepatic IVC narrowing, non-visualisation of hepatic veins S/O Budd Chiari Syndrome. Coagulation profile: serum APLA IgG: 18.24 μ/ml (<10), Factor V functional:19% (70–120%), anti-thrombin III activity: 35% (80–120%), serum homocysteine: 5.77 μmoles/l (5.46–16.40), protein S functional: 41% (60–140%).
Conclusion: This patient is classified as having antiphospholipid antibody syndrome as per presence of 1 lab and 1 clinical criteria. This patient is also having inherited thrombophilia along with APLA syndrome leading to the development of Budd Chiari Syndrome with cirrhosis of liver.

CONFLICTS OF INTEREST

The author has none to declare.

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ADAMTS13 MUTATIONS ASSOCIATED WITH DEFECTIVE ADAMTS13 SECRETION IN A PATIENT WITH NON-CIRRHOTIC PORTAL HYPERTENSION

Ashish Goel [1], V. Raghupathy [1], G.J. Amirtharaj [1], Aaron Chapla [1], Aparna Ramachandran [1], Saravanan Sureshkumar [1], K.A. Balasubramanian [1], Anup Ramachandran [1], B. Ramakrishna [1], Nihal Thomas [1], Ian Mackie [2], C.E. Eapen [1], Elwyn Elias [1,3]

[1]*Christian Medical College, Vellore, India,* [2]*University College London, London, UK,* [3]*University Hospital Birmingham, Birmingham, UK*

Background and Aim: ADAMTS13 (a disintegrin like and metalloproteinase with thrombospondin type 1 motifs—13)—vWF (von-Willebrand factor) imbalance in non-cirrhotic intrahepatic portal hypertension (NCIPH) raises possibility of a mutation in CUB domain (exons 26, 27) of ADAMTS13 gene. We looked for such ADAMTS13 mutations in NCIPH.
Methods: Plasma vWF, p-selectin (which anchors vWF multimers) levels and ADAMTS13 activity (by in-house collagen binding assay (CBA)) were assayed in NCIPH patients and healthy controls. Exons 26/27 of ADAMTS13 were sequenced in 17 NCIPH patients and 3 healthy controls. In the NCIPH patient with ADAMTS13 mutation, ADAMTS13 immunostatin (indirect immunofluorescence) of liver biopsy and next-generation sequencing (NGS) (Illumina platform) of entire ADAMTS13 gene were done. Restriction fragment length polymorphism (RFLP) targeting the detected mutation was done in other subjects.
Results: Significantly lower ADAMTS13 activity; higher vWF and p-selectin concentrations were seen in 24 (liver biopsy confirmed) NCIPH patients (20 males; age: 37, 17–66 years; median, range) compared to 22 controls (17 males; age: 33, 28–61 years). CUB domain sequencing showed novel heterozygous mutation in 1 NCIPH patient at rs140450669 (c.3829 C>T) leading to amino acid change (p. R1277W). NGS detected another heterozygous mutation at rs2301612 (exon 12, c.1342 C>G), in cysteine-rich domain, leading to amino acid change (p. Q448E). This 21 year old male had splenorenal shunt surgery for variceal bleed and developed glomerulonephritis 1 year later. He had severe ADAMTS13 deficiency (CBA: 10%) and high vWF (533 IU/dl). Anti-ADAMTS13 antibody immunostaining of his liver biopsy showed punctate diffuse staining (low power) and globules of ADAMTS13 (high power) in stellate cells, unlike normal and cirrhotic controls.

RFLP analysis did not detect R1277W mutation in 23 other NCIPH patients and 22 controls.
Conclusion: ADAMTS13 mutations may affect ADAMTS13 secretion in some NCIPH patients.

CONFLICTS OF INTEREST

The authors have none to declare.

Corresponding author: .
E-mail: drashishgoel@cmcvellore.ac.in

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HEPATIC SARCOIDOSIS

Anil Pal, Amey Sonawane, Deepak Amarapurkar

Bombay Hospital and Medical Research Centre, Mumbai, India

Background and Aim: Sarcoidosis is a multisystemic granulomatous disorder of unknown etiology. It primarily affects the lungs and hilar lymph nodes. Hepatic sarcoidosis (HS) is an infrequent extrapulmonary site of disease. The aim of our study was to assess the clinical features, disease characteristics and treatment response of patients with HS.
Methods: This study included 22 patients with HS who attended the gastroenterology clinic between 2000 and 2016. We analysed the patient demographics, co-morbidities, presenting symptoms, laboratory parameters, radiologic features, extent of disease, liver biopsy and response to treatment. Other causes of granulomatous liver disorders were thoroughly ruled out prior to establishing the diagnosis of HS.
Results: 22 patients (15 males, 7 females) were diagnosed with HS. The mean age of the cohort was 47.36 $\pm$ 14.87 years. The associated co-morbidities were diabetes mellitus (36.4%) and hypertension (27.3%). 40.9% had received anti-tuberculous therapy in the past. Two patients had family history of sarcoidosis. The commonest presenting symptoms were generalized weakness (68.2%), weight loss (59.1%), fever (50%) and loss of appetite (40.9%). Hepatomegaly (90.9%), splenomegaly (77.3%), icterus (50%) and pallor (31.8%) were common presenting signs. Cirrhosis of liver was evident in 22.7% of the patients. Imaging revealed mediastinal, abdominal and retroperitoneal lymphadenopathy in 50% of cases. Concurrent lung involvement was observed in 40.9% of the cases. The commonest liver function abnormalities were hyperbilirubinemia, elevated transaminase levels and serum alkaline phosphatase levels (ALP). 36.4% had elevated angiotensin converting enzyme levels. All patients underwent a liver biopsy which revealed granulomatous liver disease. 63.6% were treated with oral corticosteroids. Response to steroids was brisk with normalization of serum bilirubin, transaminases and ALP levels in most of the cases.
Conclusion: The diagnosis of HS must be made on clinico-laboratory findings combined with assessment of other organ affection and histological evidence of non-caseating granulomas.

CONFLICTS OF INTEREST

The authors have none to declare.

Corresponding author: Anil Pal.
E-mail: anilpal605@gmail.com

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THROMBOELASTOGRAPHY ANALYSIS IN PATIENTS WITH CIRRHOSIS AND ITS CORRELATION WITH CONVENTIONAL COAGULATION PARAMETERS

Rachit Agarwal, Piyush Ranjan

Sir Gangaram Hospital, New Delhi, India

Background and Aim: Conventional parameters of coagulation such as prothrombin time (PT) do not accurately assess the coagulation profile in cirrhosis because of reduction in both anticoagulant and procoagulant factors. Thromboelastography (TEG) is a rapid viscoelastic test of coagulation that assesses clot formation in whole blood, including plasmatic and cellular components. The aim of this study was to see correlation of TEG with conventional coagulation tests and severity of liver disease (CTP score).
Methods: Consecutive patients with cirrhosis admitted to the Gastroenterology Department were included. They were investigated with TEG parameters [*r* time, *k* time, alpha angle (AA), maximum amplitude (MA)] and conventional coagulation parameters namely platelet count, PT, aPTT, fibrinogen and d-dimer. Statistical analysis was done with Spearman's correlation coefficient tests.
Results: 113 patients with cirrhosis were included. The etiology was alcohol 42%, cryptogenic 40%, and viral 18%. *k* time, AA and MA showed significant correlation with PT, aPTT, fibrinogen and platelet counts ($P < 0.05$). There was significant correlation of