A YOUNG BOY WITH BUDD CHIARI SYNDROME SECONDARY HETEROZYGOUS TO FACTOR V LEIDEN – A CASE REPORT

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ABSTRACT

Budd Chiari syndrome (BCS) is a rare condition with exact incidence not known. It results from obstruction to the venous outflow of liver anywhere from the hepatic veins till the terminal inferior vena cava. Most cases are idiopathic; commonest cause being hypercoagulable state. Other causes include hepatic or metastatic malignancies, infections, inflammatory bowel disease, Behcet syndrome, aspergillosis and rarely, Inferior vena cava webs. Clinical presentation of BCS is variable. Most of the patients present insidiously with abdominal pain, hepatomegaly and ascites. On rare occasions, onset can be in the form of lethal upper gastrointestinal bleeding due to ruptured esophageal varices.

We report a case of BCS secondary to heterozygous factor V Leiden mutation in a young boy. Presentation in our patient was insidious, progressive ascites with abdominal pain.

KEY WORDS: Budd Chiari Syndrome, Factor V Leiden, Ascites, Portal Hypertension

INTRODUCTION

Budd Chiari syndrome (BCS) is a syndrome that results from the obstruction of hepatic venous outflow, from hepatic veins till termination of inferior vena cava, involving a variable segment of the outflow tract.1 Most caes of BCS are idiopathic. Known etiologies can be classified as prothrombotic conditions and non-thrombotic factors. Most common etiologies are linked to hereditary thrombophilias including Factor V Leiden, protein C and S deficiency, anti thrombin III deficiency and activating mutations of prothrombin. Less often, acquired hematological disorders as myeloproliferative disorders, anti phospholipid syndrome and paroxysmal nocturnal hemoglobinuria are also incriminated.2 Rare reported etiologies include tumor invasion (hepatocellular or renal cell carcinoma invasion)3 and inferior vena cava webs.4 The classical presentation of BCS is abdominal pain, hepatomegaly and ascites.1 Sometimes, patients present with bleeding from ruptured esophageal varices because of portal hypertension.5 Doppler ultrasound can establish the diagnosis with characteristic finding of non visualization of hepatic veins; or rarely a thrombus can be demonstrated in the lumen.7 Contrast CT scan, MRI and magnetic venography establishes diagnosis in most cases. Hepatic venography; although considered goldstandard, but is invasive and rarely necessary.9 BCS is managed in a step wise manner with anti coagulation therapy, angioplasty, followed by TIPS. Liver transplantation is the last resort if these measures fail.9 Factor V Leiden (FVL) also known as Activated protein C resistance, is a common thrombotic disorder affecting 1.3% Pakistani population10 and 5% US population.11 It is the most common cause of thrombophilia in Pakistan responsible for 14.2% cases with thromboembolic complications; while other studies have shown 10-60% frequency.11 Heterozygous FVL increases risk of thromboembolism by 5-7 times while homozygous for FVL increases the same by 80-100 times. Common complications are venous thrombo-embolism but rarely, arterial thrombosis can occur in homozygous patients. Treatment is with lifelong anticoagulation with target INR 2-3.12

CASE PRESENTATION

A 5 years boy was with complaints of insidious, progressive abdominal distension for 02 years and edema feet for 1 week. He also had generalized abdominal pain that gradually worsened. Child developed edema feet 1 week back which extended till knees in 4-5 days time. He was a fully vaccinated, developmentally normal child with no history of tuberculosis or other significant illness in the family.

At time of presentation he was conscious, cooperative boy with obvious abdominal distension and prominent veins over anterior abdominal wall. Abdomen was grossly distended with an everted umbilicus. Prominent tortuous veins with blood flow away from umbilicus; were visible on anterior abdominal wall. Abdominal girth was 64 cm. Liver was palpable 3 cm below the right costal margin with total span of 11 cm. Spleen was not palpable. Fluid thrill was present and bowel sounds audible. Hernial orifices were intact. Genitalia were normal male type with no genital edema.

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Laboratory data showed a normal blood picture (Hb 13.0g/dL, Platelets 160,000/mm³, TLC 13100/mm³) and ESR (14mm/hour). He had slightly elevated liver transaminase and alkaline phosphatase (ALT 63, Alk. Phosphatase 468, Bilirubin 0.98mg/dL), serum albumin and total protein were normal (Serum Albumin 3.2g/dL, Total Protein 5.1 g/dL). There was no proteinuria or glycosuria and hepatitis B and C viral serology was negative. Coagulation screening was also unremarkable (PT 13/13 seconds, APTT 34/34 seconds). Ascitic fluid was transudative (Protein 0.4g/dL, cell count 32/uL, 98% lymphocytes, LDH 42iu/ml).

Abdominal ultrasound showed normal sized Liver and spleen, moderate ascites, on color Doppler, direction portal blood flow was normal with adequate velocity. No blood flow was seen in hepatic veins on Color Doppler Ultrasound. Triphasic CT Scan Abdomen was done which showed hepatomegaly with caudate lobe hypertrophy. Splenic size was normal with moderate ascites. Hepatic veins could not be visualized and hepatic segment of Inferior Vena Cava was compressed. Upper gastrointestinal endoscopy was performed which revealed normal esophagus and stomach with no evidence of esophageal varices. Factor V Leiden mutation analysis was done and patient was heterozygous for Factor V Leiden.

Patient started on oral diuretic (furosemide) and anti coagulation therapy. Subcutaneous enoxaprin given for 5 days with warfarin overlap starting after 02 days. Pedal Edema settled in 6-7 days and abdominal girth started to decrease. Repeat color Doppler showed partial recanalization of middle and left hepatic veins. Patient was discharged with OPD follow up.

**DISCUSSION**

BCS is a disorder resulting from hepatic venous outflow obstruction. Common presenting features of BCS include insidious onset with gradually progressive abdominal pain, hepatomegaly and ascites. Our patient presented with typical clinical features of gradually progressive abdominal pain and abdominal distension due to ascites. In patients presenting with atypical presentations, such as isolated upper gastrointestinal bleeding; high index of suspicion is mandatory to establish the diagnosis. Close differential diagnoses including Chronic liver disease, Nephrotic syndrome and Tuberculous ascites were easily ruled out on the basis of clinical and laboratory parameters including absent stigmata of Chronic liver disease, negative serological markers for hepatitis B and C virus, absent proteinuria and transudative, lymphocytic ascites. Doppler
Ultrasonographic finding and contrast CT scan confirmed the diagnosis of BCS in our patient obviating the need for further imaging studies.

Known etiological factors for BCS are diverse but thrombophilias are commonest. Common thrombophilias incriminated in the pathogenesis of BCS are FVL, protein C and S deficiency and anti thrombin III deficiency. Myeloproliferative disorders are also important etiological factors for BCS. In our case the BCS was due to the thrombotic obstruction of all the three hepatic veins. Pedal edema was caused by the non occlusive compression of hepatic segment of inferior vena cava because of preferential enlargement of caudate lobe. Thrombosis in our patient was because of hypercoagulable condition elicited by heterozygous state for FVL. Other etiologies include tumor invasion (hepatocellular carcinoma or renal cell carcinoma), extrinsic compression of the hepatic venous outflow and inferior vena cava webs. These diagnostic possibilities were effectively ruled out by imaging studies; color Doppler ultrasound and contrast CT scan in our case; allowing focused investigation of the prothrombotic state. Etiology in our patient was established by FVL mutation analysis. Indefinite anticoagulant therapy is the mainstay of treatment in patients with primary Budd-Chiari, including the case presented. TIPS and liver transplantation are viable options for resistant cases.

CONCLUSION

Budd Chiari Syndrome is a rare condition and a diagnostic challenge. High index of suspicion is required to timely diagnose the condition. Once diagnosed; all effort must be made to establish the etiology.

REFERENCES