International Journal of Research in Medical Sciences
Bharmal U et al. Int J Res Med Sci. 2023 Jul;11(7):2715-2718
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20232129

Case Report

A rare presentation of chronic Budd Chiari syndrome in a 13 years old female

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Received: 25 April 2023
Revised: 17 May 2023
Accepted: 18 May 2023

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ABSTRACT

These days pathologies related to liver are increasing due to one or other factors. Here, we review a case of a girl with Budd Chiari syndrome (BCS), which is due to obstruction of hepatic veins, in turn causing portal hypertension. The pathology of the syndrome includes hepatic venous outflow tract obstruction due to thrombosis along the vein. This in turn leads to portal hypertension and increased pressure in the inferior vena cava (IVC). Clinically, it affects the heart, liver, esophagus, rectum, superficial skin. Early diagnosis can be done by Doppler and liver biopsy to prevent chronic complications of hepatic fibrosis and cirrhosis. The patient was a 13-year-old girl who had abdominal distension, pedal edema, discoloured and decreased urine and fever. On examining the patient, we found out stunted linear and intellectual growth as per her age, low IQ and communication deficits. On ultrasonography, a nodular liver surface with rounded edges and hypoechoic nodules within the parenchyma were seen. Biochemical investigations showed increase in liver enzymes. Also, there was free fluid in abdomen, few anechoic channels in periumbilical and perisplenic regions showing color flow on Doppler. Ascitic tap showed a high total cell count with a lymphocytic predominance. Based on history by the patient, radiological findings, Ascitic tap and biochemical investigations we have come to the conclusion that this is a case of BCS. After being admitted, we gave antibiotics, diuretic, folic acid, albumin, lactulose. Patient was discharged on propanolol as prophylaxis for portal hypertension.

Keywords: Portal hypertension, Ascitic tap, Doppler, Cirrhosis, Inferior vena cava

INTRODUCTION

Budd-Chiari syndrome (BCS) is an infrequent occurrence, predominantly found in south-eastern Asia with an incidence of one per one million across the world annually.[1,2]

Primary BCS is associated with hepatic venous outflow tract obstruction due to thrombosis along the course of hepatic veins to the junction of inferior vena cava (IVC) with right atrium, clinically presenting acutely with ascites, hepatomegaly and right upper quadrant pain, and chronically with symptoms of portal hypertension and liver failure, which is more common. Studies have reported genetic predispositions causing thrombophilia in such patients.[1,2]

Early diagnosis can be done by a Doppler study and liver biopsy to prevent chronic complications of portal hypertension, and progression to hepatic fibrosis and cirrhosis.

Management is spearheaded by using non-invasive methods like oral anticoagulants, followed by radiological interventions (stenting/transjugular intrahepatic

portosystemic shunt) and hepatic transplant for refractory cases.[3]

CASE REPORT

A 13-year-old girl presented to the emergency department with complaints of generalized abdominal distension which had an insidious onset, progressing gradually, accompanied with pedal edema till the groin since the past 7 days. There was decreased urinary output with discolored urine and fever lasting for 2 days. She had no history of jaundice, vomiting, hematemesis, black stools, bleeding disorders, hypertension, tuberculosis, chest pain, breathlessness, altered sensorium, convulsions, or similar complaints in the past.

Table 1: Lab investigations pre-treatment.

Parameter	Patient value	Biological reference range
Haemogram		
Hemoglobin (gm/dl)	12.0	11-15
Total leucocyte count/cmm	9000	4000-10000
Platelets/cmm	1,22,000	1,50,000-41,000
Coagulation profile		
PT (s)	14	11-14
INR	1.16	
PTT (s)	30.6	28-36
Biochemistry		
S. Ferritin (ng/ml)	1073	10-120
S. Cholesterol (mg/dl)	102	15-220
S. Triglyceride (mg/dl)	105	0-150
S. HDLD (mg/dl)	35	40-70
S. LDLC (mg/dl)	46	60-130
S. VLDL (mg/dl)	21	12-30
HIV	Negative	
HCV	Negative	
HBsAg	Negative	
S. total protein (mg/dl)	7.10	6-8
S. albumin (mg/dl)	2.60	3.2-5
S. globulin (mg/dl)	4.50	2.3-3.6
A/G ratio	0.58	1-2
S. ALT (IU/l)	108	0-40
S. AST (IU/l)	203	0-37
S. ALP (IU/l)	36	28-382
S. Urea (mg/dl)	16	14-40
S. Creatinine (mg/dl)	0.65	0.6-1.2
S. Sodium (mEq/l)	138	135-145
S. Potassium (mEq/l)	4.90	3.5-5.1
S. Total bilirubin (mg/dl)	1.00	0.1-1.2
S. Direct bilirubin (mg/dl)	0.40	0-0.4
S. Indirect bilirubin (mg/dl)	0.60	0.1-0.8
S. Ceruplasmin (mg/dl)	27.14	14-40

General examination revealed pitting pedal edema. She had stunted linear and intellectual growth as per her age, with stunted height, low IQ and communication deficits. All vitals were normal.

On per-abdomen examination tender abdominal distension was noted. She did not allow further examination due to discomfort.

Ultrasonography (USG) findings revealed a nodular liver surface with rounded edges and hypoechoic nodules within the liver parenchyma. Free fluid in the abdomen consistent with gross ascites and internal septations were seen. Few anechoic channels were seen in perisplenic and periumbilical regions showing colour flow on doppler, persistent with collateral formation.

Contrast enhanced computed tomography (CECT) was performed which showed enlarged caudate and left lobes of the liver, reducing the calibre of the intrahepatic inferior vena cava (IVC). There was relative atrophy of the right lobe with surface nodularity. Only the left hepatic vein was seen patent on a contrast study, the rest had filling defects. Gross ascites, several collaterals, dilated main portal vein, and splenomegaly were indicative of portal hypertension.

Table 2: Lab investigations post treatment.

Parameter	Patient value	Biological reference range
S. total protein (mg/dl)	7.29	6-8
S. albumin (mg/dl)	3.60	3.2-5
S. globulin (mg/dl)	3.69	2.3-3.6
A/G ratio	0.98	0.9-2
S. ALT (IU/l)	26.5	0-40
S. AST (IU/l)	64.10	0-37
S. ALP (IU/l)	97	28-382
S. total bilirubin (mg/dl)	3.45	0.1-1.2
S. direct bilirubin (mg/dl)	2.56	0-0.4
S. indirect bilirubin (mg/dl)	0.89	0.1-0.8

Developmental dysplasia of the hip was seen.

Electrocardiography (ECG) revealed sinus tachycardia.

Cytology of ascitic fluid showed a high total cell count of 8200/ul with a lymphocytic predominance (44%) and mesothelial cells in clusters.

The patient was hospitalised and managed conservatively with cefotaxime, metronidazole, rifaximine 200 mg tds, furosemide, pantoprazole, optineuron, albumin, aldectone, folic acid, lactulose and propranolol 20 mg BD.

4 days after admission and continued treatment and monitoring, ALT and AST levels had fallen, albumin and globulin levels approached towards normal ranges, and the

A: G ratio had normalised, however bilirubin (direct) was elevated (Table 2).

The patient was discharged on propranolol 20 mg BD as prophylaxis for portal hypertension, and was advised to return for paracentesis when required.

DISCUSSION

BCS is mostly encountered in the adult population and considered relatively uncommon in children.

Within the paediatric age group, maximum cases have been reported from the Indian subcontinent with male predominance.[4]

One way of classifying BCS etiologically is according to the presence of prothrombotic and anti-thrombotic factors. Generally, one or more thrombogenic disorders and a local factor is necessary for hepatic vein thrombosis. In most cases, the local factor cannot be determined. However, the predisposing factor can be recognized in more than 90% of cases.[5]

The most common etiologies include hereditary thrombophilic disorders like protein C and S deficiency, factor V Leiden mutation, and antithrombin III (ATIII) deficiency. Acquired disorders include myeloproliferative disorders, antiphospholipid antibody syndrome, paroxysmal nocturnal hemoglobinuria (PNH). Some systemic disorders which may predispose to thrombosis include Celiac disease, Behcet's disease, sarcoidosis, immunoallergic vasculitis, granulomatosis, and ulcerative colitis. Hepatic vein obstruction may also follow as a consequence of amoebic liver abscess, hydatid disease, or benign or malignant tumours, compression by adjacent organs, polycystic kidney disease. Oral contraceptives, especially high-oestrogen content pills are also involved in thrombus formation.

BCS can also be classified as primary or secondary depending on the origin of the obstructive lesion. Primary BCS occurs due an intraluminal obstruction of the hepatic vein whereas secondary BCS involves compression due to external factors like invasive tumour.

Pathogenesis of BCS includes obstruction of hepatic veins. This leads to hemodynamic changes including sinusoidal dilation and eventually liver congestion which gives rise to symptoms like right upper quadrant pain, portal hypertension induced ascites, pedal edema. Hypoxic damage to hepatocytes (hepatocellular necrosis) occurs due to decreased perfusion of liver via portal venous system. Inflammatory changes ensue and ultimately within a few weeks, fibrotic changes may occur in the form of nodular regeneration/reactive hyperplasia. Ultimately cirrhosis may be seen. Often the caudate lobe may undergo hypertrophy due to IVC obstruction.

Clinically, a suspicion of BCS should be raised when there is abdominal distension secondary to ascites, fever, bleeding secondary to hematemesis/melena, jaundice, dehydration due to diarrhoea/vomiting.

Laboratory investigations including hemogram which may show decreased platelet count and monocyte count, coagulation profile, biochemistry panel including S. ferritin (elevated), S. cholesterol (reduced), S. triglyceride, S. HDLD (reduced), S. LDLC (reduced) should be performed (Table 1). Rapid HIV, HCV and HBsAg should be done to rule out the same. Ascitic fluid analysis reveals elevated total cell count with increased polymorphs and lymphocytes and mesothelial cells in clusters.

Low S. albumin and elevated S. globulin along with liver function tests including SGPT and SGOT may be elevated. Alkaline phosphatase may be reduced. Total and direct bilirubin may appear elevated (Table 1).

Ultrasonography of the liver should be the investigation of choice when a suspicion of BCS is raised.[5]

Nodular liver surface with rounded edges and hypoechoic nodules in the liver parenchyma may be seen.

Non-invasive procedures like contrast enhanced computed tomography (CECT) of abdomen and pelvis along with portovenography may be performed. Enlarged caudate and liver lobes due to compensatory mechanisms may be seen along with surface nodularity suggestive of cirrhosis. Significant narrowing of the intrahepatic IVC may be seen due to compression secondary to caudate lobe hypertrophy. Hepatic veins may appear unopacified with contrast suggestive of chronic thrombosis. Superior mesenteric vein and portal vein may appear dilated with multiple collaterals including periumbilical, perigastric, periesophageal, perisplenic, ileo-renal, anterior abdominal wall, anorectal suggestive of portal hypertension. Splenomegaly may be seen.

MRI can be performed as an alternative imaging technique but it is unsuitable for non-affording patients hence not frequently used. Liver biopsy and hepatic venography can also be performed.[5]

In case of this patient additional findings include the bilateral femoral head appearing dislocated posterosuperiorly with shallow acetabulum suggestive of developmental dysplasia of the hip and severe intellectual disability. No association between BCS, Intellectual disability and dysplasia of the hip could be found as a result of lack of gene analysis studies due to financial constraints.

Due to lack of availability of data regarding paediatric BCS management, the adult treatment is followed by convention. Sequential use of pharmacotherapeutics like anticoagulants and diuretics are first line, followed by

invasive thrombolysis, angioplasty, TIPS, and finally, liver transplant.[6]

With only medical management, there is often a failure of improvement, necessitating radiological interventions; there is also a substantial risk of fatal complications with anticoagulants like intracranial haemorrhage.[3]

Angioplasty and thrombolysis may often require further treatment with TIPS or transplant.[6] Percutaneous recanalization has been shown to have a high success rate in BCS with short length stenosis.[7]

The main indicators for TIPS include refractory ascites, hepatic failure, and portocaval hypertension induced bleeding; generally reserved for when all three veins are totally blocked.[8] This procedure has an overall good outcome, allays the requirement of transplant, and can also be used as rescue therapy.[9] Radiological intervention supersedes in terms of feasibility in children as compared to shunt surgery due to the probably insufficient caliber of portal vein.[8]

It is of special note with regard to Asian context that without thorough confirmatory investigations, the patients should not be started on antituberculous therapy, which also often presents with ascites with a lymphocytic predominance.[8]

CONCLUSION

BCS is a disease complex associated with hepatic vein outflow obstruction, rarely seen in paediatric patients. Such patients may present with abdominal distension secondary to ascites, pedal edema, fever as seen in this case. Appropriate diagnostic modalities should be used to make a diagnosis of BCS as soon as possible, so as to prevent complications and eventually mortality. Treatment involves use of anticoagulants and diuretics as pharmacotherapy. Due to high chances of failure of improvement seen with pharmacotherapy, invasive methods like angioplasty or thrombolysis can be used, which although have a better outcome, might cause more complications. TIPS and eventually liver transplant can be done in refractory cases.

ACKNOWLEDGEMENTS

The authors would like to thank the patient and her family for cooperating during the research process.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Bharmal U, Mital R, Sharma B, Bhatt V. A rare presentation of chronic Budd Chiari syndrome in a 13 years old female. Int J Res Med Sci 2023;11:2715-8.