PEDIATRIC ONCALL
CHILD HEALTH CARE

CASE REPORTS

PANCYTOPENIA WITH SPLENOMEGALY IN AN INFANT – A SILENT PRESENTATION OF BUDD CHIARI SYNDROME

Harshit Vachher[1], Dhruvi Shah[1], Darsita Jatakia[2], Vishrutha Poojari[1], Shivangi Tetarbe[1], Ira Shah[1].

[1]Bai Jerbai Wadia hospital for children, Parel, Mumbai, India,
[2]Seth GS medical college and KEM hospital, Parel, Mumbai, India.

ABSTRACT

Introduction: Budd-Chiari Syndrome (BCS) is a rare condition where the hepatic venous outflow is obstructed. While adults typically show signs like ascites or jaundice, infants are much harder to diagnose. In young children, the symptoms are often vague, leading to a delay in diagnosis and treatment.

Case Report: A 13 months old male child presented with a history of recurrent infections and massive splenomegaly with pancytopenia. The child had recurrent episodes of diarrhea and pneumonia requiring admission in the past. Initial workup, including whole exome sequencing and bone marrow aspiration were normal, except for positive cytomegalovirus and Parvo virus B19 status. On Doppler ultrasound, the truth emerged, showing severely narrowed hepatic veins. The child underwent digital angiography with successful venoplasty to open the blocked veins and was started on anticoagulation.

Conclusion: This case shows that BCS can present atypically as isolated hypersplenism and pancytopenia without signs of liver involvement. Doppler ultrasonography is an important tool for diagnosing such cases, and timely radiological intervention can prevent further liver damage and improve outcomes.

ARTICLE HISTORY

Received 25 March, 2026

Accepted 20 May, 2026

KEYWORDS

Pancytopenia, BCS, Rare presentation, Hypersplenism.

Introduction

Budd–Chiari syndrome (BCS) is an uncommon vascular disorder characterized by obstruction of hepatic venous outflow, leading to hepatic congestion, portal hypertension, and progressive liver dysfunction. In infants and young children, BCS is rare and often presents with non-specific symptoms, making timely diagnosis challenging.[1] Classical manifestations such as ascites, hepatomegaly, abdominal distension, and liver dysfunction may be subtle or absent early, particularly in children, and diagnosis is often delayed until complications develop.[2] We report an infant presenting with recurrent infections, pancytopenia, and splenomegaly who was diagnosed with BCS.

CASE REPORT

A 13-month-old male child, born from third degree consanguineous marriage, was referred in view of splenomegaly and pancytopenia. He was hospitalised at 1.5 months oage for 10 days for persistent diarrhoea. At 6 months, he had fever for 7 days with pus collection at the vaccination site, and splenomegaly. His blood investigations showed pancytopenia so he underwent bone marrow aspiration, which showed cellular marrow with trilineage haematopoiesis. At 7 months, he was hospitalised for viral pneumonia with cardiogenic shock and hepatosplenomegaly with pancytopenia. His haemoglobin electrophoresis and whole exome sequencing were normal. He was referred to us in November 2025 for further management. There is history of Celiac disease in his elder sister, so the parents never introduced gluten in his diet. On presentation, his weight was 9.2 kg [<-2SD according to World Health Organization (WHO)] and height was 75 cm (<-2SD according to WHO). Laboratory investigations are depicted in Table 1. Cytomegalovirus (CMV) showed log 3.1 and viral load 1425 IU/mL on Polymerase chain reaction (PCR) and Parvovirus B19 showed log of 4.8 and viral load 75,650 IU/mL. Intravenous immunoglobulin (IVIG) was given. Doppler ultrasonography (DSA) abdomen showed chronic liver parenchymal disease with left lobe hypertrophy, splenomegaly, intrahepatic portosystemic collaterals, and non-visualisation of right hepatic vein. Computed tomography angiography (CTA) showed moderate hepatomegaly, gross splenomegaly, with attenuated intrahepatic portal vein and non-opacification of hepatic veins. Digital angiography showed diffuse narrowing of middle hepatic vein (MHV), narrowing at ostia of right hepatic vein (RHV) and left hepatic vein (LHV), and narrowing in middle segment of RHV. Venoplasty of RHV and LHV was done and was kept on low molecular weight heparin (LMWH), and discharged. Upper gastrointestinal scopy was not done in view of thrombocytopenia. On follow up the boy was hemodynamically stable and had improvement in laboratory parameters, and DSA showed patent RHV and LHV.

Address for Correspondance: Harshit Vachher, Bai Jerbai Wadia hospital for children, Parel, Mumbai-400012, India.
Email: drharshitvachher@gmail.com

Table 1: Serial laboratory investigations

Parameters	Day of admission	Day of discharge
Hemoglobin (gm/dl)	8.5	9
Packed Cell Volume (%)	27.5	27
Total Leukocyte Count (cumm)	2100	3900
Platelet count (cumm)	44,000	68,000
Neutrophils (%)	35.9	23
Lymphocytes (%)	49.4	73
Monocytes (%)	13.5	2
Eosinophils (%)	1	2
Prothrombin Time (seconds)	17	13.2
International Normalized Ratio (INR)	1.41	1.09
IgM (mg/dL)	155	-
IgA (mg/dL)	83	-
IgG (mg/dL)	117	-
IgE (mg/dL)	42	-

DISCUSSION

BCS is classified as primary when obstruction arises from intrinsic venous lesions, most commonly thrombosis, and secondary when due to external compression. Thrombosis accounts for most cases, with inherited or acquired prothrombotic states identified in up to 75% of patients. Myeloproliferative disorders are the most frequent cause, followed by Factor V Leiden and Factor II mutations. Acquired risks include paroxysmal nocturnal hemoglobinuria, oral contraceptive use, pregnancy, and deficiencies of natural anticoagulants, although liver dysfunction may obscure diagnosis. With systematic evaluation, predisposing factors can be identified in over 90% of cases.[3]

Celiac disease (CD) is an autoimmune disorder with well-recognized extraintestinal manifestations, including hepatic involvement ranging from transaminitis to autoimmune hepatitis and biliary cirrhosis. An association between CD and BCS has been reported, suggesting that progressive CD may induce biological or nutritional changes leading to secondary autoimmunity, vasculitis, and increased vascular risk.[4]

Our patient demonstrated an atypical presentation, with insidious pancytopenia and splenomegaly without classical features of liver disease. While pediatric BCS often presents acutely, chronic disease may manifest primarily with portal hypertension.[5] Isolated pancytopenia with massive splenomegaly is rare and can misdirect evaluation toward primary hematological disorders.[6] Normal liver enzymes and preserved synthetic function further delayed suspicion of hepatic pathology.

Recurrent viral infections such as CMV and parvovirus B19 may contribute to endothelial injury and prothrombotic states, potentially triggering vascular obstruction prior to overt liver disease.[7] Imaging is crucial for diagnosis, with Doppler ultrasonography serving as an effective first-line modality. Cross-sectional venography aids confirmation, while catheter venography remains the gold standard, especially for therapeutic planning.[7,8] Early detection enables timely intervention, preventing progression to portal hypertension and cirrhosis.[9]

The prognosis of BCS depends on the extent of venous obstruction, underlying prothrombotic conditions, and prompt intervention. Untreated, mortality is high, but advances in medical therapy, interventional radiology, and liver transplantation have significantly improved outcomes.[10] This case highlights that chronic BCS may progress silently, with preserved hepatic function masking significant portal hypertension and hypersplenism for prolonged periods.

Compliance with Ethical Standards

Funding: None

Conflict of Interest: None

References:

1. Menon KV, Shah V, Kamath PS. The Budd-Chiari syndrome. N Engl J Med. 2004 Feb 5;350(6):578-585.
2. Pinto RB, Schneider AC, da Silveira TR. Cirrhosis in children and adolescents: An overview. World journal of hepatology. 2015 Mar 27;7(3):392.
3. Aydinli M, Bayraktar Y. Budd-Chiari syndrome: etiology, pathogenesis and diagnosis. World journal of gastroenterology: WJG. 2007 May 21;13(19):2693.
4. Kochhar R, Masoodi I, Dutta U, Singhal M, Miglani A, Singh P, Singh K. Celiac disease and Budd Chiari syndrome: report of a case with review of literature. European journal of gastroenterology & hepatology. 2009 Sep 1;21(9):1092-4.
5. Huggins J, Hill S. Budd-Chiari Syndrome in Children: A Review of Clinical Presentation and Management. J Pediatr Gastroenterol Nutr. 2021;72(2):185-92.
6. Sharma S, Patel N. Atypical Presentation of Hepatic Vein Thrombosis: Isolated Pancytopenia and Splenomegaly. Case Rep Hepatol. 2019;2019:8745912.
7. Crill CM, Crosby S, Baskin M. Cytomegalovirus-associated thrombosis in infants and children. Pediatr Infect Dis J. 2006;25(10):952-955.
8. Plessier A, Rautou PE, Valla D. Management of Budd–Chiari syndrome. Hepatology. 2012;57(1):1962-1972.
9. Darwish Murad S, Plessier A, Hernandez-Guerra M, et al. Etiology, management, and outcome of the Budd–Chiari syndrome. Ann Intern Med. 2009;151(3):167-175.
10. Elshaer M, Hafez MM, Ramadan AG, Shedeed K, Tawheed A. Budd-Chiari syndrome: Prognostic scores, special populations, and management challenges. World Journal of Gastroenterology. 2025 Oct 21;31(39):111300.