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Original Research Article

Unveiling the Uncommon: A Case Report of Atypical Budd-Chiari Syndrome Presentation

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Abstract:

Background: Budd-Chiari syndrome (BCS) is an uncommon liver disorder characterized by the obstruction of the hepatic venous outflow tract. It is characterized by an obstruction in either the hepatic veins or the inferior vena cava. We share a case study of a female patient who initially exhibited mild symptoms, like weakness, fatigue, with a history of menorrhagia, without any overt signs of chronic liver disease. Subsequent evaluation revealed that she was actually suffering from Chronic Budd-Chiari syndrome. This condition typically presents with a classical triad of symptoms: abdominal pain, hepatomegaly, and ascites. This case highlights the diagnostic challenge posed by such disorders and the need for a detailed evaluation in a case of unexplained pancytopenia. However, atypical presentations can complicate diagnosis and management. This case report highlights an uncommon presentation of BCS, detailing its clinical course, diagnostic challenges, and management strategies.

Case Report: A 24-year-old female presented with generalized weakness, fatigue, and menorrhagia. She had a history of blood transfusion a few years prior. Physical examination revealed pallor and hepatosplenomegaly, without signs of heart failure or liver disease. Laboratory workup indicated severe anemia with pancytopenia, consistent with iron deficiency anemia. Despite treatment, persistent thrombocytopenia prompted further investigation. Abdominal ultrasound confirmed hepatosplenomegaly, and a triple-phase CT scan suggested Budd-Chiari syndrome. Digital Subtraction Angiography (DSA) revealed a complete blockage of the suprahepatic inferior vena cava (IVC). Balloon venoplasty and stenting were performed to improve blood flow and reduce collateral circulation.

Result: Post-procedure, the patient showed marked clinical improvement, with a decrease in hepatosplenomegaly and resolution of anemia. However, thrombocytopenia persisted, necessitating ongoing monitoring. This case underscores the importance of considering atypical presentations in the diagnosis of Budd-Chiari syndrome and highlights the efficacy of advanced interventional radiology techniques in its management.

Conclusion: Budd-Chiari Syndrome can present with atypical symptoms, complicating its diagnosis. Early recognition and intervention are crucial for improving patient outcomes. Advanced imaging and interventional radiology techniques play a pivotal role in managing BCS, providing symptomatic relief and improving prognosis.

Recommendations: Clinicians should maintain a high index of suspicion for BCS in patients with unexplained hepatic abnormalities and pancytopenia. A multidisciplinary approach, involving hepatologists, radiologists, and hematologists, is essential for optimal management.

Regular follow-up and monitoring are recommended to manage potential complications and ensure sustained patient improvement.

Keywords: Budd-Chiari Syndrome, Atypical Presentation, Thrombocytopenia, Interventional Radiology, Hepatic Vein Obstruction.

Introduction

Budd-Chiari Syndrome (BCS) is a rare vascular disorder characterized by obstruction of the hepatic veins, leading to hepatic congestion and subsequent liver damage. This condition often presents with a classical triad of symptoms: abdominal pain, hepatomegaly, and ascites. However, atypical presentations can occur, complicating the diagnosis and management of this already challenging condition.

Budd-Chiari Syndrome was first described in the 19th century, and its understanding has evolved significantly over the years [1, 2]. Traditionally, BCS is associated with conditions that predispose to thrombosis, such as myeloproliferative disorders, hypercoagulable states, and chronic inflammatory diseases. The pathophysiology involves the obstruction of hepatic venous outflow, leading to increased sinusoidal pressure, hepatic congestion, and subsequent ischemic damage to the liver parenchyma [3]. Despite its well-established clinical picture, BCS can present atypically, making it a diagnostic enigma in certain cases.

Atypical presentations of BCS can include symptoms and signs that deviate from the classical triad. These may encompass nonspecific symptoms like mild abdominal discomfort, fatigue, and weight loss, or even atypical sites of venous thrombosis. For instance, there have been reports of BCS presenting with isolated inferior vena cava thrombosis without significant hepatic vein involvement, or as a secondary phenomenon in systemic diseases such as Behçet's disease or antiphospholipid syndrome [4]. Such atypical manifestations often lead to delays in diagnosis and appropriate management, emphasizing the need for heightened clinical vigilance and thorough investigative protocols.

Recent advancements in diagnostic imaging and interventional radiology have significantly improved the detection and management of BCS. Doppler ultrasound, computed tomography (CT), and magnetic resonance imaging (MRI) are crucial in identifying the extent and nature of venous obstruction [5]. Furthermore, interventional procedures such as trans jugular intrahepatic portosystemic shunt (TIPS) and endovascular stenting have emerged as effective therapeutic options, offering symptomatic relief and improved prognosis for patients with BCS.

Case Presentation

A 24-year-old female patient presented with complaints of generalized weakness, fatigue, and menorrhagia. She had a prior history of blood transfusion a few years back. Physical examination revealed pallor and hepatosplenomegaly, with no signs of heart failure or stigmata of liver disease.

Initial Laboratory Workup and Findings

- Laboratory Results: Severe anemia with pancytopenia.

- Diagnosis: Iron deficiency anemia confirmed by subsequent investigations.

- Peripheral Blood Smear (PBS): Consistent with Iron Deficiency Anemia. Total leukocyte count (TLC) was normal, but thrombocytopenia persisted.

Table 1:

Parameters	Values
Hb	6.6 g/dl
TLC	3.10 thou/mm^3
TPC	5600 thousand/mm^3
Vitamin B_{12}	363
LDH	153 U/L
Iron	25 µg/dl
TIBC	384 µg/dl
Tsat	6.51%
Ferritin	1.53 pg/MI
ICT/DCT	Negative
Ret Count	2.44%

Immediate Management

The patient received one unit of packed red blood cells (PRBC) transfusion and was initiated on intravenous iron replacement therapy. A repeat complete blood count (CBC) showed improvement in hemoglobin and TLC levels; however, thrombocytopenia remained unexplained.

Further Investigations

The patient experienced a single episode of fever, prompting additional tests including:

- NS1 Ag
- Dengue serology
- Rapid malarial antigen test (RMAT)
- Widal test
- Blood culture and sensitivity (C/S)

All these tests returned negative.

Table 2:

Parameters	Values
RMAT	Negative
NS1 Ag	Negative
S procalcitonin	<0.05
WIDAL	Negative
Blood c/s	No growth
Urine c/s	No growth

Table 3:

Parameters	Values
PNH (By flow cytometry)	Negative
ACL IgM	Negative
ACL IgG	Negative
Protein C	Negative
Protein S	Negative
Antithrombin 3	Negative
Jak2v617f mutation	Negative
Lupus anticoagulant	Negative

Imaging and Bone Marrow Biopsy

- Abdominal Ultrasound (USG): Confirmed hepatosplenomegaly.
- Bone Marrow Biopsy: Indicated a normocellular marrow with erythroid hyperplasia.

Due to evidence of hypersplenism and persistent thrombocytopenia, the patient was scheduled for a triple-phase CT scan.

CT Scan Results

The CT scan revealed:

- Mottled appearance of the liver.

- Non-visualization of hepatic veins.

- Significant narrowing of the inferior vena cava (IVC) at the junction of intrahepatic IVC with the right atrium.

- Splenomegaly, suggestive of Budd-Chiari syndrome.

Further Assessment and Interventional Radiology

A battery of tests ruled out all hematological and prothrombotic states, chronic infections, and tumors. Upon reviewing the CT scan, the hepatic veins were noted to be absent, and a web-like structure was visualized near the IVC.

The patient was referred to the interventional radiology department, where Digital Subtraction Angiography (DSA) was performed. DSA revealed:

- Complete blockage of the suprahepatic IVC.

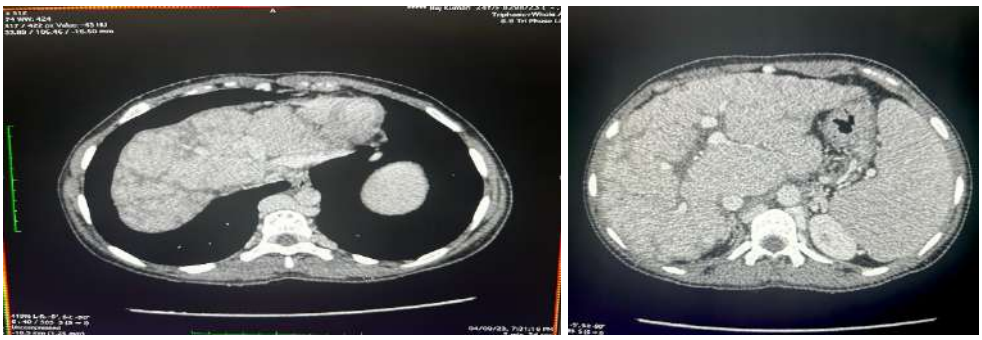

Figure 1:

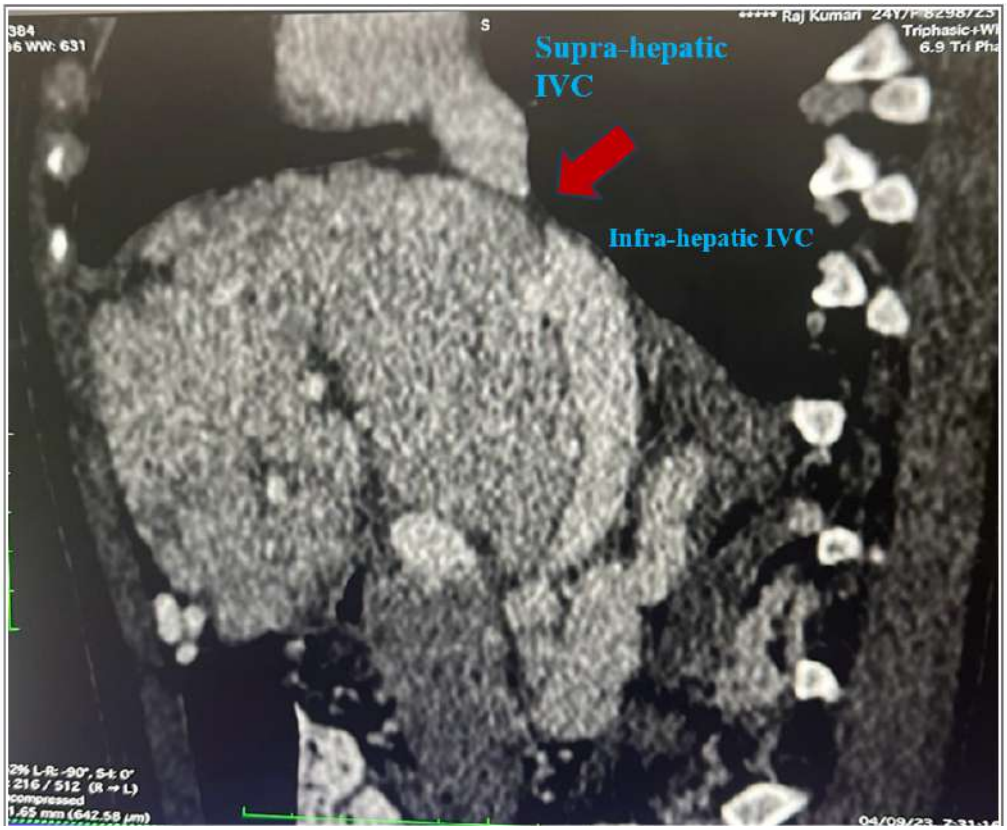

Figure 2:

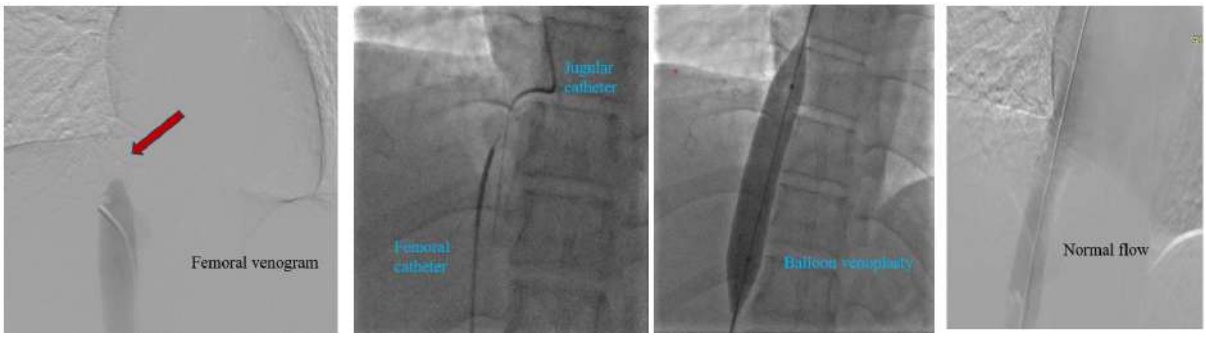

Figure 3: A close look-up of radiograph

Intervention

Balloon venoplasty and stenting were performed, leading to:

- Improved blood flow across the IVC walls.

- Significant reduction in collaterals.

A delayed venogram indicated partial recoil of the IVC web and persistence of collaterals, necessitating the stenting procedure.

Outcome and Follow-Up

Post-procedure, the patient showed marked improvement:

- Decrease in hepatosplenomegaly.

- Resolution of anemia.

The persistence of thrombocytopenia is being monitored, with plans for follow-up to assess long-term outcomes and manage any potential complications arising from Budd-Chiari syndrome.

Discussion

This case of a 24-year-old female with atypical BCS highlights several important aspects of the diagnosis and management of this rare condition. The patient's presentation with generalized weakness, fatigue, menorrhagia, and persistent thrombocytopenia complicated the diagnostic process, emphasizing the need for thorough and systematic evaluation in such complex cases.

Budd-Chiari Syndrome often presents with the classic triad of abdominal pain, hepatomegaly, and ascites. However, this patient's presentation lacked these hallmark features, instead showing symptoms more commonly associated with hematologic conditions, such as severe anemia and pancytopenia. This underscores the variability in BCS presentations and the importance of considering BCS in patients with unexplained hepatic abnormalities and thrombocytopenia.

Imaging studies played a crucial role in this case. The abdominal ultrasound initially confirmed hepatosplenomegaly, but it was the triple-phase CT scan that provided critical diagnostic insights. The CT scan revealed a mottled liver appearance, non-visualization of hepatic veins, significant narrowing of the IVC, and splenomegaly, which were suggestive of BCS. Recent literature emphasizes the importance of advanced imaging techniques, such as CT and MRI, in identifying the extent and nature of venous obstructions in BCS .

The patient underwent Digital Subtraction Angiography (DSA), which confirmed a complete blockage of the suprahepatic IVC. The use of balloon venoplasty and stenting significantly improved blood flow and reduced collateral circulation. This case highlights the critical role of interventional radiology in the management of BCS, with studies indicating that procedures such as TIPS (transjugular intrahepatic portosystemic shunt) and endovascular stenting can provide effective symptomatic relief and improve prognosis.

Despite initial improvement, the persistence of thrombocytopenia suggests the need for ongoing monitoring and

possibly further interventions. Long-term follow-up is essential in BCS cases to manage potential complications, such as stent recoil or re-thrombosis. Current guidelines recommend regular imaging and hematologic assessments to ensure sustained patency of stented vessels and to monitor for signs of disease progression.

Recent research on atypical presentations of BCS has highlighted several key findings. A 25-year-old female with BCS showed that early diagnosis and anticoagulant therapy are crucial for effective management [6]. Another study documented a 36-year-old female whose BCS was caused by PAI-1 4G/5G heterozygous polymorphism, demonstrating that inherited thrombophilia can lead to this condition [7].

Additionally, a 13-year-old boy with BCS complicated by hepatopulmonary syndrome underscored the need for early liver transplantation when hypoxemia persists despite symptomatic treatment [8]. In a unique case, a 32-year-old female developed BCS as the initial manifestation of systemic lupus erythematosus (SLE), later complicated by antiphospholipid syndrome, emphasizing the importance of considering BCS in patients with unexplained hepatic outflow obstruction [9].

Management of BCS in pregnancy was also explored, with a successful outcome achieved through hepatic vein angioplasty and stenting, highlighting the benefits of a multidisciplinary approach [10]. Advances in diagnostic techniques and endovascular interventions have significantly improved survival rates for BCS, with myeloproliferative neoplasms identified as common etiologies [11]. A case involving a young child with BCS revealed that early symptomatic treatment can improve conditions, though a liver transplant may still be necessary as a last resort [12].

Conclusion

In conclusion, Budd-Chiari syndrome is a rare and potentially life-threatening condition that can sometimes manifest with seemingly trivial or vague symptoms, thereby complicating its diagnosis. This case emphasizes the importance of considering rare hematologic and congenital disorders in the differential diagnosis of patients with pancytopenia. Early recognition and management of Budd-Chiari syndrome can significantly improve outcomes and reduce the risk of further complications. Additionally, this case underscores the importance of considering atypical presentations in the diagnosis of Budd-Chiari syndrome. It highlights the role of advanced interventional radiology techniques in managing this complex condition, ultimately leading to significant clinical improvement for the patient.

References

1. Ding PX, Han XW, Liu C, Zhang Y, Cheng AL, Wu Y, Liang XX, Gao XM, Lee EW. Long-term outcomes of individualized treatment strategy in treatment of type I Budd-Chiari syndrome in 456 patients. Liver International. 2019 Aug;39(8):1577-86.
2. Jadallah KA, Sarsak EW, Khazaleh YM, Barakat RM. Budd-Chiari syndrome associated with coeliac disease: case report and literature review. Gastroenterology Report. 2018 Nov;6(4):308-12.
3. Sharma A, Keshava SN, Eapen A, Elias E, Eapen CE. An update on the management of Budd–Chiari syndrome. Digestive diseases and sciences. 2021 Jun;66:1780-90.
4. Khan F, Armstrong MJ, Mehrzad H, Chen F, Neil D, Brown R, Cain O, Tripathi D. a multidisciplinary approach to the diagnosis and management of Budd-Chiari syndrome. Alimentary pharmacology & therapeutics. 2019 Apr;49(7):840-63.
5. Gavriilidis P, Marangoni G, Ahmad J, Azoulay D. State of the art, current

perspectives, and controversies of Budd-Chiari syndrome: a review. Journal of Clinical Medicine Research. 2022 Apr;14(4):147.
6. Singh S, Wawre S, Pohek r S, Raut A, Gujar S, Dhengare A. Case Report: Budd Chiari Syndrome. J Pharm Res Int [Internet]. 2021 [cited 2024 Jun 23].
7. Sousa D, Silva SA, Jorge C, Fernandes RM, Rodrigues AI, Coelho MV, et al. Case Report: A case of PAI-1 4G/5G heterozygosity causing Budd-Chiari Syndrome. F1000Res [Internet]. 2019 [cited 2024 Jun 23];8:1568.
8. Tian F, Dong X, Hou XH, Yuan RY, Pan YW, Zhang D. Budd-Chiari syndrome with hepatopulmonary syndrome: a case report and literature review. Zhonghua Er Ke Za Zhi [Internet]. 2024 [cited 2024 Jun 23];62(1):71-75.
9. Solela G, Daba M. Budd-Chiari Syndrome as an Initial Presentation of Systemic Lupus Erythematosus Associated with Antiphospholipid Syndrome: A Case Report with Review of the Literature. Open Access Rheumatol [Internet]. 2023 [cited 2024 Jun 23];15:139-143.
10. Mishra V, Aggarwal R, Solanki SB, Mahalingam M. Management of Pregnancy in a Rare Case of Budd–Chiari Syndrome: A Case Report. J South Asian Feder Obst Gynae [Internet]. 2022 [cited 2024 Jun 23].
11. Sharma A, Keshava S, Eapen A, Elias E, Eapen C. An Update on the Management of Budd–Chiari Syndrome. Dig Dis Sci [Internet]. 2020 [cited 2024 Jun 23];66:1780-1790.
12. Timsaal Y, Ali S, Malik F, Chawla A, Ahmed J. Rare Case of Budd-Chiari Syndrome in a Young Child: A Diagnostic Conundrum. Cureus [Internet]. 2021 [cited 2024 Jun 23];13. Available from: https://consensus.app/papers/case-buddchiari-syndrome-young-child-diagnostic-tim-saal/9ff3be30c82059dba45e74ffec518d0b/?utm_source=chatgpt