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A Case Report of Budd-Chiari Syndrome in Pregnancy Secondary to Paroxysmal Nocturnal Hemoglobinuria

Dr. Devathi Mounika[1], Dr. Bindu Vijaykumar[2]

[1]Senior Resident, Department of Obstetrics and Gynaecology, Institute of Maternal and Child Health (IMCH), Government Medical College, Kozhikode, Kerala, India
Corresponding Author Email: *mdevathi365[at]gmail.com*

[2]Professor, Department of Obstetrics and Gynaecology, Institute of Maternal and Child Health (IMCH), Government Medical College, Kozhikode, Kerala, India

Abstract: ***Aims: To present a rare and complex case of Budd-Chiari Syndrome (BCS) secondary to previously undiagnosed Paroxysmal Nocturnal Hemoglobinuria (PNH), presenting for the first time during pregnancy, and to highlight the successful management through a multidisciplinary approach. Presentation of Case: A 21-year-old primigravida at 19 weeks of gestation presented with right upper quadrant pain, nausea, and jaundice. Initial investigations revealed severe hemolytic anemia, thrombocytopenia, and deranged liver function. Imaging studies confirmed hepatosplenomegaly, ascites, and extensive thrombosis of the hepatic, portal, and splenic veins, diagnostic of Budd-Chiari Syndrome. Further hematological evaluation, including flow cytometry, identified a large PNH clone, establishing it as the underlying etiology. The patient was managed throughout her pregnancy with anticoagulation, multiple blood transfusions, and close monitoring by a team of obstetricians, hematologists, and gastroenterologists. An elective Caesarean section was performed at 36 weeks, resulting in the delivery of a healthy male infant and a favorable maternal outcome. Discussion: The co-occurrence of PNH and BCS is rare, and its initial presentation during pregnancy poses significant diagnostic and therapeutic challenges. The prothrombotic state of pregnancy likely unmasked the underlying PNH. Management required a delicate balance between preventing further thrombosis and mitigating the risk of hemorrhage, especially during delivery. This case underscores the necessity of considering rare hematological disorders in pregnant patients with atypical presentations of liver disease. Conclusion: Timely diagnosis and collaborative, multidisciplinary management are crucial for achieving successful maternal and fetal outcomes in pregnancies complicated by severe conditions like PNH-induced Budd-Chiari Syndrome.***

Keywords: Budd-Chiari Syndrome, Paroxysmal Nocturnal Hemoglobinuria, Pregnancy, Thrombosis, Anticoagulation, Multidisciplinary Care

1. Introduction

Budd-Chiari Syndrome (BCS) is a rare disorder characterized by the obstruction of hepatic venous outflow, leading to hepatic congestion, portal hypertension, and liver damage. While often idiopathic, it is frequently associated with underlying hypercoagulable states. (1) Paroxysmal Nocturnal Hemoglobinuria (PNH) is an acquired clonal stem cell disorder characterized by chronic intravascular hemolysis and a profound predisposition to thrombosis, making it a significant cause of BCS. (2)

The initial presentation of PNH as BCS during pregnancy is exceedingly rare. Pregnancy itself induces a physiological hypercoagulable state, which can exacerbate underlying prothrombotic disorders, leading to life-threatening complications for both mother and fetus.(3) Managing anticoagulation, anemia, and the hemodynamic changes of pregnancy in such a complex scenario presents a formidable challenge. We report the case of a young primigravida who presented with acute BCS, leading to the diagnosis of PNH, and was successfully managed through a multidisciplinary approach to achieve a favorable outcome.

2. Presentation of Case

A 21-year-old primigravida, with no known comorbidities, presented at 19 weeks of gestation with a three-week history of intermittent right hypochondrial pain, nausea, exertional dyspnea, and jaundice. She was initially evaluated at a local hospital where she was found to have severe anemia (Hemoglobin [Hb] 5.5 g/dL) and thrombocytopenia (Platelet count 75,000/µL). After receiving three units of Packed Red Blood Cells (PRBC), she was referred to a tertiary care center for further evaluation.

Upon admission, she was pale but hemodynamically stable. Physical examination revealed hepatosplenomegaly and abdominal distension due to ascites. Laboratory investigations confirmed severe anemia, thrombocytopenia, and features of hemolysis, including elevated total bilirubin (2.10 mg/dL), and markedly elevated Lactate Dehydrogenase (LDH) at 763 U/L. Her coagulation profile was normal.

An urgent ultrasound of the abdomen with Doppler study revealed hepatosplenomegaly, moderate ascites, and significantly altered hepatic blood flow. The middle and left hepatic veins were attenuated, and there was a reversal of flow in the right hepatic vein, findings consistent with Budd-Chiari Syndrome (Figure 1).

A subsequent Magnetic Resonance Imaging (MRI) of the abdomen confirmed these findings and further demonstrated extensive thrombosis involving the main portal vein, its branches, the splenic vein, and the superior mesenteric vein (Figure 2).

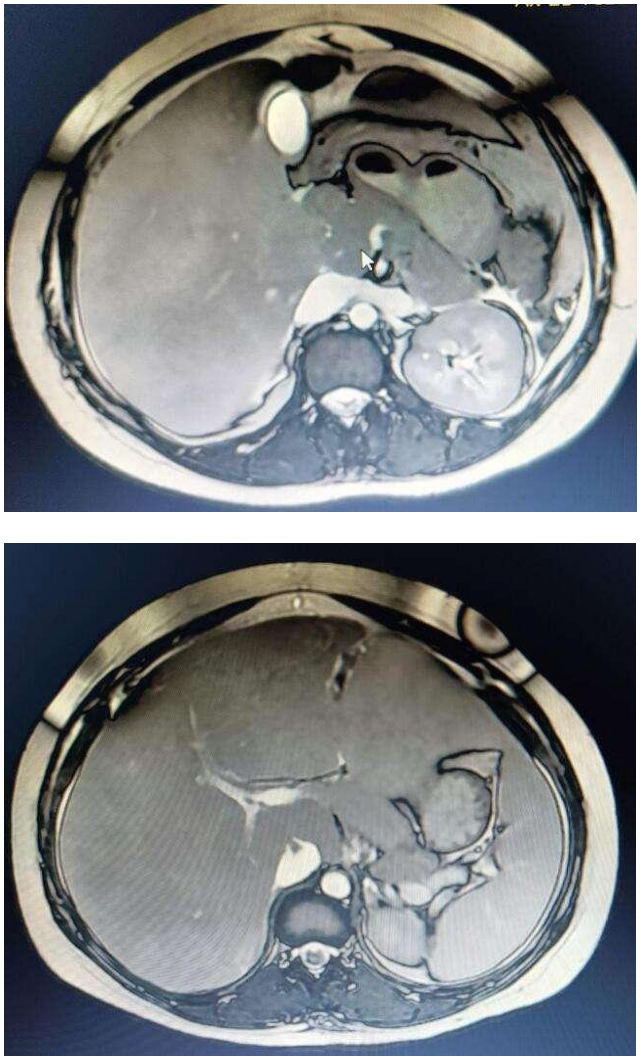

Figure 1: Axial T2-weighted MRI showing hepatomegaly, ascites, and altered signal within portal structures suggestive of thrombosis

Given the extensive thrombosis in a young patient, an underlying prothrombotic state was suspected. A comprehensive hematological workup was initiated. Autoimmune screens, including Antinuclear Antibody (ANA) and Antiphospholipid Antibody (APLA) profiles, were negative. The definitive diagnosis was established through flow cytometry, which revealed a large PNH clone within neutrophils (90.62%) and monocytes (90.07%).

A multidisciplinary team comprising obstetricians, hematologists, and gastroenterologists was convened. The patient was started on therapeutic anticoagulation with low-molecular-weight heparin (Enoxaparin 80mg twice daily). Her severe anemia required multiple PRBC transfusions throughout the pregnancy, with a target Hb level above 9 g/dL. She also developed Gestational Diabetes Mellitus (GDM), which was managed with diet control and insulin.

The patient was monitored with serial ultrasounds to assess fetal growth and doppler parameters, which remained normal. Regular blood counts and liver function tests were performed. A plan for delivery was formulated at 34 weeks of gestation. Given the high risk of variceal bleeding during labor and the need for controlled anticoagulation management, the team recommended an elective Lower Segment Cesarean Section (LSCS).

At 36 weeks and 2 days of gestation, after stopping anticoagulation 24 hours prior, she underwent an elective LSCS under spinal anesthesia. A healthy male infant weighing 2.66 kg was delivered with good APGAR scores. The surgery was uneventful with minimal blood loss. The infant was transferred to the NICU for observation due to maternal comorbidities and had an uneventful course. Postoperatively, anticoagulation was restarted once hemostasis was secured. The mother's recovery was smooth, and she was discharged in stable condition on continued anticoagulation and oral iron supplementation.

3. Discussion

This case highlights a rare but life-threatening convergence of PNH, Budd-Chiari Syndrome, and pregnancy. PNH is a primary driver of thrombosis, particularly in unusual sites like hepatic and portal veins, and pregnancy acts as a potent secondary trigger. (2, 4) The diagnosis in our patient was challenging, as initial symptoms could mimic other pregnancy-related liver disorders like HELLP syndrome. However, the profound hemolytic anemia and imaging findings of extensive thrombosis guided the workup toward a primary thrombotic etiology.

The management strategy was centered on a multidisciplinary approach. Therapeutic anticoagulation is the cornerstone of treatment for BCS to prevent thrombus extension and promote recanalization. (1) LMWH was the agent of choice due to its safety profile in pregnancy. Managing anemia with transfusions was critical to ensure adequate oxygen delivery to both mother and fetus.

The timing and mode of delivery are crucial decisions in such high-risk pregnancies. Vaginal delivery was considered high-risk due to the potential for bleeding from undetected esophageal varices during straining. An elective LSCS allowed for a controlled environment, including planned cessation and reinitiation of anticoagulation, minimizing both thrombotic and hemorrhagic risks. (5)

4. Conclusion

The presentation of Budd-Chiari syndrome during pregnancy should prompt an immediate and thorough investigation for underlying prothrombotic disorders, including rare conditions like PNH. Despite the grave prognosis often associated with this condition, our case demonstrates that with a timely diagnosis, aggressive management of thrombosis and anemia, and a coordinated, multidisciplinary team approach, a successful outcome for both mother and child is achievable.

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