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Bernard-Soulier Syndrome in Pregnancy: A Case Report

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ABSTRACT

Background & aim: Bernard-Soulier syndrome (BSS) is known as a rare congenital bleeding disorder, characterized by thrombocytopenia and the presence of giant platelets (PLTs). The treatment process is debatable. Data on the clinical course and outcome of pregnancy in women with Bernard Soulier syndrome is scattered in individual case reports. From this perspective, this case report aimed to present pregnancy in this disorder.

Case report: A 23-year-old primigravid woman by clinical manifestations of frequent episodes of epistaxis and thrombocytopenia hospitalized in the 38th weeks of pregnancy. She was undergoing frequent PLT transfusions, had a vaginal delivery with episiotomy through which a baby girl was born with an Apgar score of 8-9 and a normal birth weight. The patient was then discharged after six days in good condition. Due to the rarity of the disease, it may not appear foremost on the clinician's differential. Affected individuals tend to bleed excessively.

Conclusion: Pregnancy in such cases is unpredictable, so its management is still controversial and merely based on multi-specialty consultation and practice. In this line, PLT transfusion, recombinant activated factor VII (rFVIIa) administration, and desmopressin use should be considered to shorten bleeding time, but anti-PLT medications must be avoided.

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Introduction

Bernard-Soulier syndrome (BSS) has been documented as a rare congenital bleeding disorder, characterized by thrombocytopenia and the presence of giant platelets (PLTs). This illness comes from a quantitative or qualitative defect in the glycoprotein (GP) receptor complex, located in the PLT membrane, which is the main receptor for von Willebrand factor (vWF), and transmitted as an autosomal recessive trait (1, 3). Furthermore, women exposed to fetal PLTs carrying this GP can develop antibodies against this fetal GPIb/IX antigen to induce fetal and neonatal alloimmune thrombocytopenia (FNAIT). As an example, the prevalence rate of early postpartum hemorrhage (EPH) had been reported by 33% during 30 pregnancies in 18 women investigated in one study, and half of them had undergone blood transfusions. As well, there were six cases of FNIAT and two perinatal deaths. In view of that, careful monitoring during pregnancy and six weeks after delivery is thus necessary to prevent this potentially life-threatening hemorrhage (4), which was first

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discovered by two French physicians, Jean-Bernard and Jean-Pierre Soulier in 1948, and named after them. In this disorder, the survival of PLTs is also reduced, probably being the cause of mild thrombocytopenia (5). The clinical manifestations of this syndrome, which often affects women and is rarely resolved by itself, are bruising, bleeding gums, menorrhagia, epistaxis, postpartum hemorrhage (PPH), gastrointestinal (GI) bleeding, and post-traumatic hemorrhage (PTH) (1, 2, 5, 6).

Case presentation

A 23-year-old pregnant woman with the gestational age of 38 weeks and 6 days was admitted to a tertiary referral hospital, in Mashhad, Iran, for the termination of pregnancy in December 2022. The patient was a known case of BSS since childhood (three months old), with a history of frequent PLT transfusions during pregnancy and hospitalizations due to epistaxis. Upon consultations with the hematologist, prednisolone (50 mg), four times a day, had been started in the prior week of her referral. The patient's vital signs at admission were also normal, and there were no uterine contractions or vaginal bleeding. The case had no history of using alcohol, tobacco, and other substances. Besides, the fundal height was at term, and the cephalic presentation and fetal heart rate (FHR) were normal (140-150 beats per minute). The case had impaired glucose tolerance (IGT) with diet control. At the onset of hospitalization, PLT count was 50,000, the hemoglobin (Hb) level was equal to 13.9 g/dL, and the hematocrit (Hct) level was 40.2%. Positive macrocytes and giant PLTs as well as a low PLT count were further reported during peripheral blood assessments. In U/A, the white blood cell (WBC) count was 20-25, and other criteria were negative. Fasting and two-hour postprandial BG was also normal. PLTs (10 units), packed cells (4 units), and recombinant activated factor VII (rFVIIa) (5 units) were further reserved in the patient's bedside refrigerator.

To terminate pregnancy, an Extra-amniotic Foley's catheter was fixed, which expelled spontaneously a few hours later. The patient received 6 doses of misoprostol (25 µg) and one unit of PLTs in total, and was then under continuous FHR monitoring. The next day, the PLTs decreased (32,000). In consultation with the hematologist and the oncologist, the CBC was also checked daily, and prednisolone administration continued for the patient.

Following a vaginal delivery with episiotomy, a baby girl was born with an Apgar score of 8-9 and a normal weight 3200 grams. The 3-cm tear in the vagina was then repaired. Due to excessive PH, oxytocin (80 units) was infused. As well, PLTs (6 units), apheresis PLTs (2 units), fresh frozen plasma (FFP, 2 units), and packed cells (3 units) were transferred to the operating room for further investigations, bleeding control, and vaginal examination. Under general anesthesia and ultrasound guidance in the lithotomy position, the pelvic, cervix, and vagina were also examined, and the vaginal tear was sutured by the gynecologist. As reported, the Hb level was 9 g/dL, the PLT count was 20,000, the BUN range was 22 mg/dL, the Cr level was 0.6 mg/dL, and her liver enzymes and vital signs were normal. The patient was then transferred to the intensive care unit (ICU). Her Glasgow Coma Scale (GCS) value was 15 and the uterus was contracted. She was alert and answered the questions. Afterward, rFVIIa (5 units), PLTs (9 units), and packed cells (1 unit), along with ampicillin, tranexamic acid ampoule, hydrocortisone, vitamin C, and clindamycin were prescribed. The Hb level was then equal to 7.6 g/dL, the Hct level was 22.5%, and the PLT count was 21,000. Abdominal and pelvic ultrasound was also normal for hematoma. The patient had tachycardia (with 10,000 PLTs). After receiving packed cell (1 unit) and PLTs (5 units), the PLT count reached 20,000. According to the consultation with the cardiologist, a chest ultrasound and the Doppler sonography of the lower extremities were requested. The echocardiography (Echo) results were also normal and the ejection fraction (EF) was by 55%. Of note, the auscultation of the heart and the lungs was normal, without rales. The hypothesis of anemia was then justified for the patient's tachycardia. After consultation with the hematologist, the case was discharged after six days in a good condition.

Discussion

As a rare hereditary bleeding disorder, BSS characterized by thrombocytopenia and the presence of giant PLTs (7, 8), with the

prevalence rate of one case in one million pregnancies (9). The disease progress is different in each pregnant woman and even in the same patient during different pregnancies. The most common incident is hemorrhage during and after delivery, which may need emergency hysterectomy (10, 11) that can be potentially life-threatening. There is insufficient evidence for the superiority of the mode of delivery (8), so each case must be managed individually (7). For the fetus, the placental transfer of maternal anti-PLT antibodies entails the likelihood of fetal and neonatal alloimmune thrombocytopenia (FNAIT), which requires treatment. The newborn is also at the increased risk of severe bleeding, such as intracranial hemorrhage (ICH) (9).

Based on this case report, severe thrombocytopenia occurred in this pregnancy and there was a need for blood transfusions. Although there was no agreement on the superiority of the mode of delivery with reference to the available evidence(9), the trial of labor was planned for this patient to reduce the risks associated with PLT depletion and bleeding complications in Cesarean-section (C-section), as well as hematomas associated with hemorrhage in the site of regional anesthesia. Hospitalization in the tertiary referral hospital in order to provide multi-specialty consultation and practice, access to blood products and the neonatal ICU, and the postpartum monitoring of the mother and her baby also paved the ground for favorable maternal and neonatal outcomes in this pregnancy.

Healthcare providers should thus consider patient education for seeking medical assistance, preventing traumas, not taking aspirin and other anti-PLT medications, refraining from traumatic sports, and devoting much attention to oral and dental health to avoid gingivitis (8, 12). A close observation of postpartum period is recommended due to high risk of hemorrhage and tranexamic acid to cover this period may be used to reduce this risk. Due to high rate of C.S in our country, should be attention to contraindication of regional analgesia/anaesthesia in this cases.

General coagulation studies may not be helpful, and the paucity of data may obscure the diagnosis. Acumen, supplemented by suspicion, may be the only way to approach this disorder. The disease is familial, and the diagnosis of one patient, the proband, necessitates the consideration of family members as well.

This activity reviews the evaluation and treatment of Bernard-Soulier syndrome and highlights the role of the interprofessional healthcare team in evaluating and managing patients with this condition. By delving into the intricacies of this rare blood clotting disorder, healthcare professionals will enhance their acumen and suspicion skills, crucial for accurate identification and subsequent multidisciplinary treatment.

Conclusion

Although BSS is a rare congenital blood disorder that may complicate pregnancy, maternal and neonatal outcomes can be promising under multi-specialty consultation and practice. The necessity of delivery in a tertiary referral hospital should be thus included in the content of education and counseling of patients with this syndrome. Report of individual experience and management of these pregnancies based on further cases will be helpful to obtain better knowledge and it's necessary to access an international registry for BSS in pregnancy.

Declarations

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Conflicts of interest

The authors declared no conflicts of interest.

Ethical considerations

Informed consent was obtained from the patient to report her illness without mentioning her personal identity.

Use of Artificial Intelligence (AI)

We have not used any AI tools or technologies to prepare this manuscript**.**

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Authors' contribution

The first author participated in idea generation, data collection and article writing, the second author participated in monitoring the patient's treatment and disease progression. The third author was responsible in idea generation and article writing. All authors have read and approved the present study.

References

1. Michael CB, Robert KA. Bernard-Soulier syndrome. Haematologica. 2011; 96(3): 355-359.
2. Longo DL. Atlas of Hematology. In: Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J, editors. Harrison's Principles of Internal Medicine, 20 th ed. New York, NY: McGraw-Hill Education; 2018.
3. Cunningham FG, Leveno KJ, Dashe JS, Hoffman BL, Spong CY, Casey BM. Editors. Williams Obstetrics, 26 th ed. New York, NY: McGraw Hill; 2022.
4. Almomani MH, Mangla A. Bernard-Soulier Syndrome. InStatPearls [Internet] 2024 Jan 11. StatPearls Publishing.
5. Pham A, Wang J. Bernard-Soulier syndrome: An inherited platelet disorder. Archives of Pathology & Laboratory Medicine. 2007; 131(12): 1834-1836.
6. Mitrović M, Elezović I, Miljić P, Antić D. Obstetric and gynecological intervention in women with Bernard-Soulier syndrome: report of two cases. Srpski Arhiv Za Celokupno Lekarstvo. 2014; 142(5-6): 351-355.
7. Macêdo MB, Brito JdMM, Macêdo PdS, Brito JA. Primigravida with Bernard-Soulier Syndrome: A case report. BMC Research Notes. 2015; 8(1): 178.
8. Durai V, Subburaj S, Subbaiah M. Pregnancy Outcome in Bernard–Soulier Syndrome. The Journal of Obstetrics and Gynecology of India. 2023; 73(5): 445-450.
9. Peitsidis P, Datta T, Pafilis I, Otomewo O, Tuddenham EG, Kadir RA. Bernard-Soulier syndrome in pregnancy: A systematic review. Haemophilia. The Official Journal of the World Federation of Hemophilia. 2010; 16(4): 584-591.
10. Macêdo MB, Brito Jde M, Macêdo Pda S, Brito JA. Primigravida with Bernard-Soulier Syndrome: A case report. BMC Res Notes. 2015; 8: 178.