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A Rare Case Report Of Bernard-Soulier Syndrome In Pregnancy

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Abstract:
Bernard-Soulier syndrome (BSS) is an autosomal recessive bleeding disorder characterized by thrombocytopenia and the presence of giant platelets. In this rare case report, we describe the course of pregnancy and delivery in a 25 year old Primigravida who is a known case of Bernard-Soulier syndrome, at a gestational age of 38 weeks + 1 day with fetus in Breech presentation. The patient had bleeding manifestations and underwent multiple platelet transfusions before undergoing Emergency Lower Segment Cesarean Section under General Anaesthesia. The placental transfer of maternal anti- platelet antibodies entails the likelihood of Fetal and Neonatal Alloimmune Thrombocytopenia (FNAIT). As Bernard-Soulier syndrome may complicate pregnancy; maternal and neonatal outcomes can be promising if delivery is planned through a multidisciplinary approach.

Key Words: *Bernard-Soulier syndrome, Neonatal Alloimmune Thrombocytopenia, Platelet transfusion.*

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I. Introduction

Bernard-Soulier syndrome (BSS) is an autosomal recessive congenital bleeding disorder characterized by thrombocytopenia and the presence of giant platelets. This is as a result of a quantitative or qualitative defect in the glycoprotein [GP] Ib-IX-V receptor complex located in the platelet membrane which is the primary receptor for von Willebrand factor (vWF). More than 30 genetic mutations have been identified, determining various severities of bleeding. The clinical manifestations of this syndrome are bruising, bleeding gums, menorrhagia, epistaxis and gastrointestinal bleeding. Bernard-Soulier syndrome is associated with a significant risk of primary and secondary Post Partum Haemorrhage (PPH) and wound haematoma. Management of delivery requires careful planning with a multidisciplinary team. Peripartum haemostatic management should be guided by assessment of the individual phenotype. The fetus of an affected mother will be heterozygous provided the father is not a carrier. Furthermore, women exposed to fetal platelets carrying this glycoprotein can develop antibodies against this fetal glycoprotein Ib-IX-V receptor antigen to induce Fetal and Neonatal Alloimmune Thrombocytopenia (FNAIT).

II. Case Description

A 25 year old female, known case of Bernard-Soulier syndrome, Primigravida, with a gestational age of 38 weeks + 1 day, was referred to the emergency department with complaints of gingival bleeding and epistaxis. The patient was a known case of Bernard-Soulier syndrome which was diagnosed at 3 months of life. She had history of menorrhagia, epistaxis and easy bruising during her adolescence which was managed conservatively. During her pregnancy, she had history of frequent platelet transfusions and hospitalizations due to epistaxis. She was already on Tablet Prednisone 40 mg twice daily as per her hematologist's instructions. She presented with 5 episodes of epistaxis and gingival bleeding. The patient was moderately built and nourished.

At the time of admission, the patient's vitals were as follows : Height = 153 cm, Weight = 65 kg, Blood pressure = 110/60 mmHg, Pulse rate = 88 beats per minute. Cardiovascular system (CVS) = S1S2 heard, no murmurs noted; Respiratory system (RS) = Bilateral air entry present, normal vesicular breath sounds heard; Central nervous system (CNS) = no focal neurological deficit. Obstetric examination showed uterus corresponding to term size with fetus in breech presentation, fetal heart rate was good. Ultrasound assessment further confirmed breech presentation with Amniotic Fluid Index (AFI) of 10 cm. Blood investigations showed Hb = 12 gm/dl, TC = 7860 cells/cumm, Plt = 0.09 lakhs/cumm and peripheral smear showed the presence of thrombocytopenia with giant platelets.

Hematologist's opinion was obtained and was advised to continue oral steroids and to give stress dose of intravenous Methyl prednisolone prior to surgery. As per instructions, 4 units of Single Donor Platelets (SDP) was transfused and repeat Plt count showed 0.46 lakhs/cumm. On day 3 of admission, patient had complaints of pain abdomen. She had no complaints of bleeding p/v or leaking p/v and perceived fetal movements well.

Cardiotocography (CTG) monitoring was done and was reassuring. Patient had spontaneous progression of labour. Per vaginal examination done showed cervix : soft, midposition, 1 cm long, 2 cm dilated and presenting part (fetal buttock) at -3 station.

Anaesthesiologist's opinion was obtained and decision for Emergency Lower Segment Cesarean Section (LSCS) made (Indication : Breech presentation in labour) under General Anaesthesia (GA). As per anaesthetist's orders, 2 units SDP was transfused pre-operatively and repeat Plt = 0.53 lakhs/cumm. Intraoperatively, bicornuate uterus noted with pregnancy in right horn. A girl baby weighing 3.154 kg was delivered as breech presentation by breech extraction. Baby cried immediately after birth, delayed cord clamping done and cord blood sample taken. Placenta and membranes were delivered in toto and uterus closed in two layers with continuous interlocking sutures. Mild atonicity was noted and hence was managed medically with intravenous Oxytocin, Methylergometrine and Tranexamic acid. Intraoperative blood loss was 850 ml and hemostasis was secured.

On post operative day 1, patient was kept nil oral and foley's catheter was retained. Vitals monitoring was done and was within normal limits and patient had no bleeding manifestations. On post operative day 1, repeat Hb = 10.1 gm/dl, TC = 8756 cells/cumm and Plt = 0.36 lakhs/cumm. As per Hematologist's orders, intravenous steroids were tapered, oral steroids were continued, 4 units Random Donor Platelets (RDP) and 1 unit SDP were transfused. Patient resumed normal bowel and bladder habits. On post operative day 3, dressing was removed and wound appeared healthy. Serial platelet monitoring was done. Complete evaluation was done for the baby and Fetal Neonatal Alloimmune Thrombocytopenia (FNAIT) was ruled out as platelet counts were within normal limits. Patient was discharged with oral steroids on post operative day 7 after suture removal with a platelet count of 0.92 lakhs/cumm. Patient was followed up postnatally and was on serial platelet monitoring.

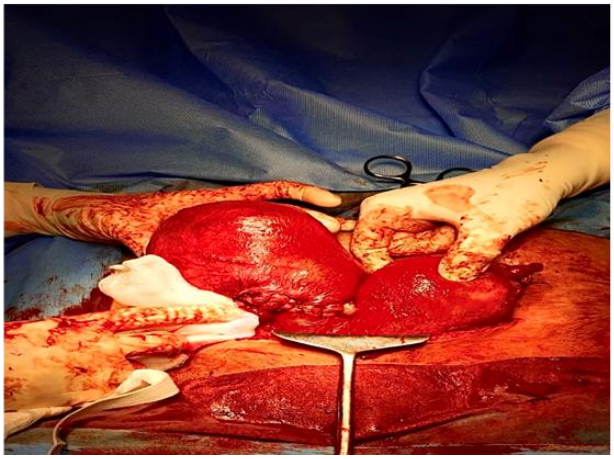

Figure 1: Intraoperative image showing bicornuate uterus with pregnancy in right horn following suturing of uterine incision.

III. Discussion

Bernard-Soulier Syndrome (BSS) results from either a quantitative or qualitative deficiency in the glycoprotein Ib-IX-V complex on the platelet membrane, which disrupts normal platelet adhesion. Over 30 genetic mutations have been identified, contributing to a spectrum of bleeding severity. Bleeding typically begins during childhood, often presenting as gingival bleeding or epistaxis; however, some individuals are not diagnosed until pregnancy. Heavy menstrual bleeding (menorrhagia) is also a frequent symptom and may be difficult to control. Bernard-Soulier Syndrome (BSS) is extremely rare in pregnancy, with an estimated occurrence of approximately one case per million pregnancies and published data on this topic remains scarce. As BSS is an autosomal recessive disorder, in areas of high consanguinity, it may be prudent to test the spouse using platelet flow cytometry for glycoprotein (GP) - Ib surface density. Pregnancy and childbirth presents significant hemorrhagic risks and requires careful management. Hence, it is mandatory that women are informed of their risk before becoming pregnant.

Royal College of Obstetricians and Gynaecologists (RCOG) guidelines recommends that peripartum haemostatic management should be guided by assessment of the individual phenotype in BSS. It also recommends that patients with a bleeding history should be given a platelet transfusion (HLA matched) prophylactically at delivery or before caesarean section, in combination with tranexamic acid. Tranexamic acid may be given at the onset of labour and continued regularly through the postpartum period.

Central neuraxial anesthesia should be avoided whenever possible and the overall risk of bleeding and opportunity for corrective treatment should be discussed with the patient by anaesthesiologists. Further, birth should be planned in an obstetric-led unit with a neonatal unit that routinely provides high-dependency care. Maternal platelet count should be monitored weekly from 36 weeks onwards. If platelets count is < 0.50 lakhs/

cumm, a plan for intrapartum care with a multidisciplinary team, that includes a hematologist should be discussed. Usage of steroids or intravenous immunoglobulins to raise the maternal platelet count is also recommended.

At the time of delivery, fetal blood sampling should not be carried out and fetal scalp electrodes should be used with caution, Usage of ventouse for delivery of fetal head is not routinely recommended, whereas mid-cavity or rotational forceps may be used, but with caution. Routine measurement of neonatal platelet count in the umbilical cord blood at the time birth is strongly recommended. Women with bleeding disorders are at increased risk of primary and secondary postpartum haemorrhage. Hence, active management of third stage of labour, rather than physiological management should be offered to these patients and uterotonics should never be given through the intramuscular route.

Due to the elevated risk of hemorrhage in individuals with Bernard-Soulier Syndrome, preventive strategies may include the use of tranexamic acid, desmopressin (DDAVP), recombinant activated factor VII (rFVIIa), and platelet transfusions. When delivery by caesarean section is planned or if significant bleeding occurs during vaginal birth, transfusion with HLA-matched platelets in combination with tranexamic acid is considered first-line treatment. Additional therapies such as intravenous immunoglobulin (IVIG), corticosteroids, and plasmapheresis have been employed to enhance the effectiveness of platelet transfusions and reduce the risk of neonatal thrombocytopenia. Given the ongoing debate over the safest approach to labor in these cases, the choice of delivery method is typically guided by the obstetrician, who must take into account the patient's individual and family history, and this decision should be made in close collaboration with a multidisciplinary team.

Fetal and Neonatal Alloimmune Thrombocytopenia (FNAIT) is a rare but potentially life-threatening disorder that can impact both the fetus and the newborn. It is characterized by a maternal immune response in which antibodies are formed against specific antigens found on fetal platelets. These platelet antigens are inherited from the father or, in cases of assisted reproduction, from the gamete donor, and are not present on the mother's own platelets. As a result, the maternal immune system identifies them as foreign. The antibodies produced cross the placenta and target the fetal platelets, leading to their destruction. This immune-mediated process can result in severe thrombocytopenia, increasing the risk of bleeding complications, including neonatal/ fetal intracranial hemorrhage.

Thrombopoietin (TPO) receptor agonists such as Romiplostim and Eltrombopag are newer drugs which have emerged as potential therapeutic options in BSS, although their use is still controversial with limited evidence primarily from case reports or small studies. These agents stimulate the thrombopoietin receptor (c-MPL) on megakaryocytes and hematopoietic stem cells, promoting increased platelet production. However, further studies are required to assess the safety and efficacy of these drugs in pregnancy.

IV. Conclusion

Management of Bernard-Soulier Syndrome during pregnancy requires a highly individualized, multidisciplinary approach due to the increased risk of bleeding and limited evidence-based guidelines. Early involvement of a team including obstetricians, hematologists, and anesthesiologists, ideally within a tertiary care center is essential for planning and monitoring. Mode of delivery should be tailored based on the patient's bleeding history, platelet count and fetal considerations. Bleeding prophylaxis may involve antifibrinolytics, recombinant factor VIIa, and selective use of HLA-matched platelet transfusions. Close antepartum, intrapartum and postpartum monitoring is critical along with neonatal evaluation.

References

[1]. Fogerty AE. Thrombocytopenia In Pregnancy: Approach To Diagnosis And Management. Semin Thromb Hemost 2020 Apr; 46: 256–263.
[2]. Mitrović M, Elezović I, Miljić Pet Al. Et Al. Obstetric And Gynecological Intervention In Women With Bernard-Soulier Syndrome: Report Of Two Cases. Srp Arh Celok Lek 2014; 142: 351–355.
[3]. Macêdo MB, Brito JMM, Macêdo PS, Et Al. Primigravida With Bernard-Soulier Syndrome: A Case Report. BMC Res Notes 2015; 8: 178.
[4]. Smaoui M, Elleuch S, Abidi S, Et Al. Bernard-Soulier Syndrome And Pregnancy: A Case Report. Ann Biol Clin (Paris). Nov Dec 2015; 73: 737–740.
[5]. Perez AV, Filho CMO, Pazinato TC, Et Al. Bernard-Soulier Syndrome In Pregnancy: A Case Report. OJOG 2019; 9: 838–844.
[6]. 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):584-91.
[7]. Amanda Vilaverde Perez, Cilomar Martins De Oliveira Filho, Bernard-Soulier Syndrome In Pregnancy: A Case Report, Open Journal Of Obstetrics And Gynecology. 2019, 838-844.
[8]. Rahimi G, Rellecke S, Mallmann P, Nawroth F. Course Of Pregnancy And Birth In A Patient With Bernard-Soulier Syndrome – A Case Report. J Perinat Med. 2005; 33:264–6.
[9]. Prabu P, Parapia LA. Bernard-Soulier Syndrome In Pregnancy. Clin Lab Haem. 2006;28:198–201.
[10]. Huq FY, Kadir RA. Management Of Pregnancy, Labour And Delivery In Women With Inherited Bleeding Disorders. Haemophilia. 2011;17:20–30.