

Bernard-Soulier Syndrome : Peripartum management of a rare platelet disorder in pregnancy

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Introduction

Bernard-Soulier syndrome (BSS) is a rare inherited disorder of clotting characterized by large platelets, thrombocytopenia and prolonged bleeding time. It is an autosomal recessive disorder caused by a deficiency of glycoprotein 1b (GpIb), the receptor for von Willebrand factor leading to qualitative and quantitative platelet disorder. The incidence is less than 1 in a million. However, in our small region with a population of approximately 1.8 million, there is a much higher incidence with approximately 7-8 cases.

Case report

A 26-year-old primigravida was diagnosed with Bernard-Soulier Syndrome (BSS) after the onset of menstruation as a teenager. Her usual platelet count maintained at 20-25 x10 /L. She had multiple platelet and blood transfusions as a result of this disorder. Menstruation was managed with oral contraceptive pill (OCP) and Tranexamic acid. She was referred to our unit as requiring specialist Haematological care in her second trimester. However, she presented at 34 weeks with a retinal detachment, which required an uneventful operative repair under general anaesthetic. Subsequently, planned delivery by elective caesarean section was indicated.

A multidisciplinary meeting led by the Consultant Haematologist with Obstetric and Anaesthetic teams was organized to plan for delivery. The plan for management of delivery via caesarean section was documented and disseminated. Regional anaesthesia was contra-indicated.

On the day of surgery, patient's haemoglobin was 120g/L and platelets 22x10 /L.

As planned by the Haematology Consultant, who was present in theatre suite throughout, the patient received two units of platelets immediately preoperatively.

General anaesthesia was induced with a modified rapid sequence and maintenance with total intravenous anaesthesia using Remifentanil and Propofol to minimize uterine atony and decrease risk of postpartum haemorrhage.

Recombinant activated factor VII (Novoseven) 7 mg was given just before knife to skin.
Cell salvage was used during caesarean section. At delivery, Tranexamic acid 1 g bolus was given followed by 1 g infusion over 8 h. After delivery, a bolus of Oxytocin was followed by prophylactic IV Oxytocin infusion and rectal Misoprostol 1 g. A further 2 units of Platelets were transfused immediately postoperatively.

Outcome

Blood loss was approximately 500 mL The patient remained on labour ward for post operative care and monitoring for the next 24 hr and received Morphine PCA with regular IV Paracetamol. NSAIDs and LMWH were omitted.

The patient received 1 unit of Platelets twice daily for the next 5 days and Tranexamic acid TID for 14 days. Intravenous Iron was given before discharge empirically.
The patient was commenced on the combined OCP on day 15.
Baby's Platelet count was >250 x10 /L at birth.

Discussion

This was successful management of a rare but life-threatening bleeding disorder which the Obstetric and Anaesthetic team had no previous experience. Direction and support by the Consultant Haematologist was vital throughout.

Pregnancy in BSS can result in primary or secondary haemorrhage and emergency hysterectomy[1]. Multiple platelet transfusions can result in antibody formation to further platelet transfusion adding further risk to future pregnancies. Neonates may develop allo-aimmune thrombocytopenia leading to intrauterine or neonatal death.

Recombinant Factor VII enhances Thrombin formation and is employed in disorders characterised by impaired Thrombin generation such as platelet disorders like BSS[2].TXA inhibits Fibrin degradation and is also thought to have other pro-coagulant effects. The Woman Trial showed that TXA reduces death due to bleeding in PPH. Pro-coagulant effects by TXA in a sub-study within the WOMAN trial showed no statistical significance, but patient numbers were small[3]. TIVA was chosen as GA technique to minimise uterine atony.

References

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3. Dallaku Ket al. No effect of Tranexamic acid on platelet function and thrombin generation (ETAPiaT) in postpartum haemorrhage: a randomised placebo controlled trial. Welcome Open Res 2019, 4:21