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Case Report

Massive Post Partum Heamorrhage due to Rare Genetic Platelet Disorder

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Abstract

Bernard-Soulier syndrome (BSS), also called hemorrhagiparous thrombocytic dystrophy [1] is a rare autosomal recessive bleeding disorder that causes a deficiency of glycoprotein Ib (GpIb), the receptor for von Willebrand factor [2]. It is characterised by qualitative and quantitative defects of the platelet membrane glycoprotein (GP) Ib-IX-V complex.

The incidence of BSS is estimated to be less than 1 case per million persons.

Data on the clinical course and outcome of pregnancy in women with Bernard Soulier syndrome is scattered in individual case reports.

A case report of forty-two years old female, known case of BSS since childhood and had splenectomy 1992, para two previous caesarean sections underwent caesarean section at thirty-three weeks for fetal distress of a baby with congenital heart disease on 16/10/2018 in private Hospital, was transferred to WWRC, HDU in unstable condition and successfully managed conservatively with good outcome.

Keywords: *Bernard Soulier Syndrome; Pregnancy*

Abbreviations

WWRC: Women Wellness Research Center; HDU: High Dependancy Unit; BSS: Bernard Soulier Syndrome

Introduction

Bernard-Soulier syndrome (BSS), also called hemorrhagiparous thrombocytic dystrophy [1] is a rare autosomal recessive bleeding disorder that causes a deficiency of glycoprotein Ib (GpIb), the receptor for von Willebrand factor [2]. It is characterised by qualitative and quantitative defects of the platelet membrane glycoprotein (GP) Ib-IX-V complex.

The incidence of BSS is estimated to be less than 1 case per million persons.

Data on the clinical course and outcome of pregnancy in women with Bernard Soulier syndrome is scattered in individual case reports.

BSS is a giant platelet disorder, it is characterized by abnormally large platelets.

- Clinical presentation of bleeding tendency varies, morbidity tends to be more in females than males' due to menstrual bleeding and childbirth. Diagnosis depends on clinical history and laboratory investigation including, full blood count and picture that will show large platelets the bleeding time (a standardized test of the time it takes for a small cut to stop bleeding) is longer than normal. This test may be difficult to perform in young children.
- The closure time (a test that measures the time it takes for a platelet plug to form in a sample of blood) is longer than normal.

- Platelets appear larger than normal under a microscope.
- There are usually fewer platelets than normal.
- Platelets do not clump together normally in the presence of ristocetin (a substance that normally promotes platelet aggregation).
- GPIb/IX/V is not detectable in blood samples (using a test called flow cytometry).

Treatment

In general, no medications are needed in Bernard-Soulier syndrome (BSS).in case of bleeding some medications can be used

- Ant fibrinolytic drugs.
- Recombinant factor VIIa.
- Desmopressin.
- Fibrin sealants.
- Hormonal contraceptives (to control excessive menstrual bleeding).
- Iron replacement (if necessary to treat anaemia caused by excessive or prolonged bleeding).
- Platelet transfusions (only if bleeding is severe).

Data on the clinical course and outcome of pregnancy in women with Bernard Soulier syndrome is scattered in individual case reports.

Case Report

A forty two years old female, known case of BSS since childhood and had splenectomy 1992, para two previous caesarean sections underwent caesarean section at thirty three weeks for fetal distress of a baby with congenital heart disease on 16/10/2018 in private Hospital, Doha, Qatar, Intraoperative course was complicated with sever adhesions and bleeding, estimated blood loss is 2 litres that needed blood transfusion of 6 units of packed RBC and 6 units platelets, Patient was shifted to high dependency unit where she stayed for 24 hours but condition deteriorated and developed acute renal insult and was shifted to WWRC, HDU for intensive care.

On 17/10/2018, in WWRC, High dependency unit, patient was having tachycardia, tachypnoea, low blood pressure and anuria with abdominal distention and drop of haemoglobin. Was managed by intensive care team together with obstetric team.

Ultrasound sound report was inconclusive initially, But the condition worsened.

Differential Diagnosis of acute abdomen, sepsis, intraabdominal bleeding, paralytic ileus or acute renal insult were made.

The condition was challenging as to go for surgical intervention or expectant management. Here came the value of Multidisciplinary team and effective communication.

Detailed expert ultra sound scanning showed that there is bleeding from the uterine incision but looks most likely venous bleeder. Decision was made to go for conservative management and observe the condition, patient received 9 units of PRBC, platelets and tranexamic acid.

On 20/10/2018 patient condition improved without surgical intervention, abdominal distension was resolved, on 25/10/2018 patient was discharged from hospital in good condition.

Discussion

Platelet and/or blood transfusions remain the best therapeutic measure for uncontrolled bleeding and prophylaxis to control bleeding during surgery. The benefits of receiving the transfusions must be weighed against the risks of exposure. Repeated exposure to

blood products raises concern for alloimmunization and platelet refractoriness. The use of leukoreduced blood components has been shown to decrease alloimmune platelet refractoriness [3]. Although some authors have suggested that patients should receive platelets from human leukocyte antigen-matched donors in order to avoid alloimmunization [4], currently this is not a widely accepted strategy. Activated factor VIIa (FVIIa) has been reported to reduce bleeding times in patients with BSS. However, FVIIa is an experimental drug in treatment of inherited thrombocytopenia, and adverse reactions have been reported [4]. Desmopressin, a synthetic analog of antidiuretic hormone, may transiently increase factor VIII and von Willebrand factor by causing their release into blood. It is used for treatment of mild hemophilia A and von Willebrand disease. Desmopressin has been reported to shorten bleeding episodes for some patients, but a test dose is recommended to determine those patients who will benefit [4]. Stem cell transplantation has been successfully used to treat 2 children with BSS who had severe, life-threatening bleeding episodes; however, based on the study results, the use of transplantation should only be considered in severe disorders and after patients have developed antiplatelet antibodies [4]. Splenectomy, often performed when immune thrombocytopenia is mistakenly diagnosed, does not improve the platelet count or function in patients with BSS should be counselled about the importance of preventing even minor trauma as well as avoiding aspirin-containing medications and other platelet antagonists [4].

Bernard-Soulier syndrome is one of several inherited giant platelet disorders distinguished by a functional abnormality of the GPIb-IX-V platelet GP receptor complex. The disease is highly variable with bleeding tendencies that can range from mild to severe and life-threatening. Platelet aggregation studies and, more definitively, flow cytometry can provide an accurate diagnosis of this rare disease and allow for adequate therapeutic management.

Conclusion

Management of these cases nictitate multidisciplinary approach during pregnancy and delivery. Anti-platelet antigens and HLA antibodies to be monitored monthly throughout the pregnancy to assess the risk of neonatal alloimmune thrombocytopenia. Clear plans should be made in conjunction with haematology to manage antenatal admissions. Prophylactic cover with recombinant Factor VIIa and tranexamic acid may be needed during labour and delivery. Regional anaesthetic to be avoided.

The availability and utilization of resources in tertiary level medical care settings play tremendous role in the management of rare and challenging cases such as BSS.

In some cases, weighing the risk of intervention versus expectant management is vital as it will change the outcome.

Undertaking

This paper has not been submitted for a concurrent publication, and has not been published before in any other journal.

Acknowledgement

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Bibliography

1. Mahfouz RA., *et al*. "Novel mutation in the glycoprotein Ibß in a patient with Bernard-Soulier syndrome: possibility of distant parental consanguinity". *Blood Coagulation and Fibrinolysis* 23.4 (2012): 335-337.

2. Feng S., *et al*. "The glycoprotein Ib/IX complex regulates cell proliferation". *Blood* 93.12 (1999): 4256-4263.

3. Fisher M., *et al*. "Alloimmunization to HLA antigens following transfusion with leukocyte-poor and purified platelet suspensions". *Vox Sanguinis* 49.5 (1985): 331-335.

4. Balduini CL., *et al*. "Inherited thrombocytopenias: from genes to therapy". *Hematologica* 87.8 (2002): 860-880.

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