International Clinical and Medical Case Reports Journal
Case Report (ISSN: 2832-5788)

A 21-Year-Old Male diagnosed with Bernard-Soulier Syndrome-Case Report

Muhammad Usama Bin Shabbir[1], Tehreem Fatima[2], Silla Naeem[2], Sundus Ali[3], Faiza Fatima[4], Ahila Ali[5*], Rukshar Thapa[6]

[1]Department of Internal Medicine, Pakistan Institute of Medical Sciences, Islamabad, Pakistan

[2]Department of Internal Medicine, King Edward Medical University, Lahore, Pakistan

[3]Pathology Department Pakistan Institute of Medical Sciences, (PIMS), Islamabad

[4]Services Institute Medical Sciences, Lahore, Pakistan

[5]Department of Internal Medicine, Dow Medical College, Karachi, Pakistan

[6]Lankenau Medical Center, Main line health, PA

Citation: *Muhammad Usama Bin Shabbir, Tehreem Fatima, Silla Naeem, Sundus Ali, Faiza Fatima, Ahila Ali*, Rukshar Thapa. A 21-Year-Old Male diagnosed with Bernard-Soulier Syndrome-Case Report. Int Clinc Med Case Rep Jour. 2025;4(5):1-5.*

Received Date: 20 May 2025**; Accepted Date:** 22 May 2025**; Published Date:** 26 May, 2024

***Corresponding author:** Ahila Ali, Department of Internal Medicine, Dow Medical College, Karachi, Pakistan

ABSTRACT

Bernard-Soulier Syndrome (BSS) is a rare autosomal recessive disorder characterized by the presence of large platelets and faulty platelet aggregation. Through this case report, we emphasize the various obstacles a male patient could face when diagnosed with BSS. Limited awareness and resources are among the most formidable challenges. Laboratory tests were carried out for the patient. They revealed severe thrombocytopenia (platelet count 8000/µL) and anemia (hemoglobin 9.7 g/dL). The clotting profile showed that prothrombin time and activated partial thromboplastin time were normal. It is crucial to recognize and manage this disease early, as inappropriate treatment will jeopardize the health of a patient who is suffering from recurrent mucocutaneous bleeding episodes. Financial barriers appear to be the most troublesome hindrances in conducting diagnostic tests and impeding the diagnosis. To reduce the rates of BSS and its effective management, heightened awareness among healthcare providers and the general population-particularly in communities with elevated rates of consanguinity is imperative.

INTRODUCTION

In 1948, it was a male patient who was first diagnosed with Bernard-Soulier syndrome and died due to recurrent episodes of bleeding. Multiple genetic mutations result in deficiency of the GPIb-IX-V complex and the emergence of BSS [1]. Although BSS presents as an autosomal recessive disorder, however, some autosomal dominant inheritance patterns have also been found in the past [2]. To achieve adequate hemostasis, the GPIb-IX-V complex performs a vital function by allowing platelet adhesion to von Willebrand Factor (VWF) on damaged vascular surfaces .Owing to the paucity of this complex, defective platelet adhesion occurs, prompting the patient towards increased bleeding tendencies. BSS can emerge with a wide array of symptoms such as nosebleeding,

ecchymosis, cutaneous and gum bleeding; it can also cause menometrorrhagia and gastrointestinal bleeding. Episodes of heavy bleeding occur in the case of patients' injury or surgery [3].

Findings found on laboratory tests are sometimes sufficient to establish the diagnosis of BSS. However, it is tragic that on various occasions it is often misdiagnosed as idiopathic thrombocytopenic purpura (ITP) without further clinical investigation. To confirm BSS, tests should demonstrate either absent ristocetin-induced platelet aggregation or a near-absent GPIb-IX-V complex [4]. Flow cytometry can also be utilized to confirm defects in the GPIb-IX-V complex

It can be confidently stated that platelet and blood transfusions are the most effective therapeutic treatments for confronting this disease and its complications [5]. Two children with stem cell transplantation have been treated proficiently, although this treatment should be considered only in severe cases [6]. As healthcare resources are very limited in developing countries, diagnosing and treating BSS is challenging [7]. We present the case of a twenty-one-year-old male diagnosed with BSS, to investigate how the disease was identified and managed, and the challenges faced during its management.

CASE PRESENTATION

The 21-year-old male patient was diagnosed with Bernard-Soulier Syndrome (BSS) at age of 5 months. He was admitted to the hospital due to multiple episodes of nasal and gum bleeding. His laboratory tests showed anemia and a platelet count below 20,000/µL, while a peripheral smear revealed large, giant platelets. Initially, a bone marrow biopsy could not be performed due to the low platelet count, but it was later performed, confirming the diagnosis. Tranexamic acid injections and platelet transfusions were the cornerstone of treatment. With the help of ristocetin aggregation tests, the diagnosis was confirmed, although flow cytometry was not conducted due to financial issues.

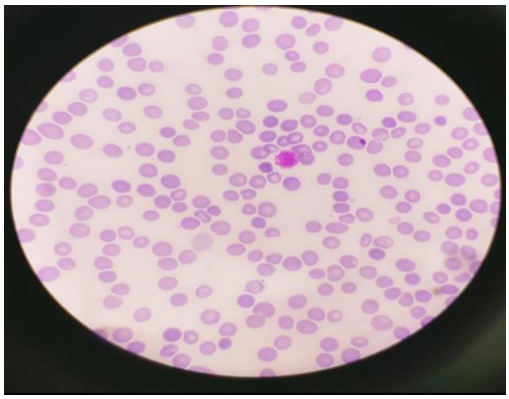

Figure 1: Giants platelets on peripheral blood smear.

The patient is currently managed as an outpatient, with controlled bleeding episodes. His general physical examination was not significant. Furthermore, his systemic examination was also unexceptional. Current laboratory reports indicate ongoing management of his condition, with regular monitoring and supportive care. The lab investigations revealed anemia (hemoglobin 9.7 g/dL) and severe thrombocytopenia (8,000/µL). A peripheral blood smear revealed hypochromic microcytic red blood cells, anisocytosis, and large platelets. The bleeding time of the patient appeared to be high. Prothrombin time, activated partial thromboplastin time, and international normalized ratio were normal. The born marrow biopsy has also been done. It revealed some hyper

cellular bone marrow segments with increase in megakaryocytic series cell. He mostly experiences nasal bleeding, which tends to occur at night and is managed with medication. Occasionally, he requires hospitalization for platelet transfusions. He has no history of trauma and leads an active life without significant hindrance. He has received factor VII infusions 5 to 6 times only. The patient's family history includes a consanguineous marriage; he has three siblings, all of whom have also been diagnosed with BSS. As a result of their medical condition, bone marrow transplantation has been discussed but not pursued at this time.

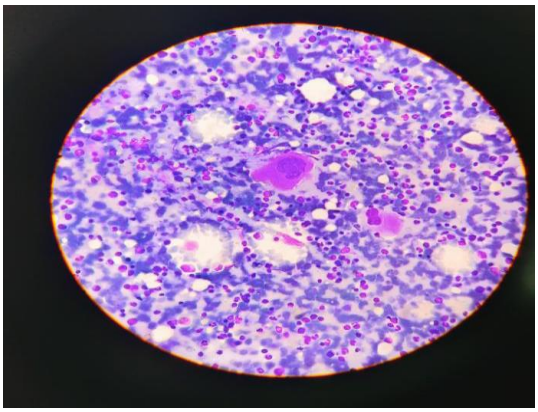

Figure 2: Bone marrow aspirate showing presence of megakaryocytes.

To control acute bleeding episodes, tranexamic acid–soaked gauzes were supplied, while in cases of uncontrolled bleeding episodes, intravenous tranexamic acid and platelet transfusions were given. This case is unique not only due to the rarity of BSS, particularly in a male patient, but also because it highlights the significant challenges faced in managing the condition within a resource-limited setting.

Table 1: Peripheral film showing anemia and thrombocytopenia.

Test	Patient 's Value	Reference Valve
Hemoglobin	9.7 gm/dl	13-17 mg/dl
MCV	72.9 fl	76-96 fl
R.B.C.s	4.32 million ul	4.5-5.5 millions /ul
Platelet count	8000/ul	150,000-4,10,000/ul

Table 2: Clotting profile revealing normal PT, APTT and Fib.

Test	Patient 's Value	Reference Valve
Prothrombin time	10.8 sec	11-16 sec
A.P.T.T	26.1 sec	26-40 sec
Fibrinogen level	204.2 mg/dl	180-360 mg/dl

CASE DISCUSSION

Bernard-Soulier Syndrome (BSS) is a rare autosomal recessive disorder. It is characterized by the presence of large platelets and faulty platelet function [8]. The symptoms of BSS can vary, often leading to serious mucocutaneous bleeding and potentially life-threatening hemorrhages during trauma, surgery, menstruation, or childbirth. Historically, there have been few reported cases in Pakistan, but the prevalence is rising, with many cases going unreported

To achieve the goal of a decline in the cases of BSS, the notion of avoidance of consanguineous marriages should be supported, or at least genetic testing should be done before consanguineous marriages [9]. Mutations

responsible for the existence of BSS result in a deficiency of one of the four glycoproteins on the platelet surface [10].

Laboratory investigations are essential for diagnosing BSS, but in Pakistan, financial barriers can impede testing. Definitive diagnosis is typically confirmed through flow cytometry. Through this test, we can help identify the deficiency of the GP Ib/IX/V complex on the platelet surface. A reduced expression of GP Ib confirms BSS [11]. In our case, BSS was identified by a ristocetin test, and a peripheral smear.

Management of BSS involves several key aspects, with preventive care being paramount. Educating patients regarding their disease, challenges, encouragement, and measures for adaptation in case of bleeding episodes forms the crux of management. Healthcare specialists should be aware of the adversities faced by BSS patients in Pakistan and should try to eliminate them by providing awareness, social support, and better healthcare. Detailed information should be provided to them regarding the medication they can take, its dosage, and the regular monitoring of their condition. In case of blood transfusion, they should be assured regarding its usage by eliminating the fear of it and the health improvement achieved through it.

Through this case, we highlight the cultural and social barriers faced in Pakistan. Widespread diagnostic facilities are not available for it. Even where they are available in a few places, they come at a high price. Patients belonging to low socioeconomic rural areas have limited access to healthcare and social support, both causing hindrances in acquiring effective treatment. There is always stigma regarding bleeding disorders-not treating them as a disease. Also, women face severe social disparity regarding this matter. They face restrictions on their daily activities and marriage. Furthermore, patients know the exact treatment but still prefer traditional treatments and end up with major complications

Various treatments are available for Bernard-Soulier Syndrome (BSS); however, platelet transfusions are the main treatment for bleeding episodes [11]. A lot of local interventions, such as compression by sponges and nasal packing, are utilized. Tranexamic acid, an antifibrinolytic agent, has proven its efficacy as it helps to stabilize clots. It is being used to diminish the episodes of mucocutaneous bleeding and menorrhagia. In our case, the patient uses tranexamic acid and receives platelet transfusions during major bleeding episodes. Work is being done on the effectiveness of other treatment options, such as eltrombopag, allogeneic stem cell transplantation, and gene therapy for BSS patients.

CONCLUSION

Bernard-Soulier Syndrome (BSS) is a disease with multiple challenges. Such challenges amplify in severity and number in developing countries like Pakistan. In order to ensure early diagnosis and management in a resource-limited setting, steps should be taken to increase awareness among healthcare providers and the public about BSS. In addition, we should try to overcome the financial barriers encountered by patients during diagnosis and treatment by providing them with appropriate care and facilities. Furthermore, advanced research is needed to develop novel therapies and management of BSS. By ensuring the implementation of all these steps, we can significantly improve outcomes and the quality of life for patients with BSS.

REFERENCES

1. Almomani MH, Mangla A. Bernard-Soulier syndrome. In: StatPearls [Internet]. StatPearls Publishing; 2024.
2. Silvia Vettore, Raffaella Scandellari, Stefano Moro, Anna Maria Lombardi, Margherita Scapin, Maria Luigia Randi, et al. Novel point mutation in a leucine-rich repeat of the GPIbα chain of the platelet von Willebrand factor receptor, GPIb/IX/V, resulting in an inherited dominant form of Bernard-Soulier syndrome affecting two unrelated families: the N41H variant. Haematologica. 2008;93(11):1743-1747.
3. Pham A, Wang J. Bernard-Soulier syndrome: an inherited platelet disorder. Arch Pathol Lab Med. 2007;131(12):1834-6.
4. Berndt MC, Andrews RK. Bernard-Soulier syndrome. Haematologica. 2011;96(3):355.
5. Fisher M, Chapman JR, Ting A, Morris PJ. Alloimmunization to HLA antigens following transfusion with leukocyte-poor and purified platelet suspensions. Vox Sang. 1985;45:823.
6. Balduini CL, Iolascon A, Savoia A. Inherited thrombocytopenias: from genes to therapy. Hematologica. 2002;87(8):860-880.
7. Iqbal S, Zakar R, Fischer F, Zakar MZ. Consanguineous marriages and their association with women's reproductive health and fertility behavior in Pakistan: secondary data analysis from demographic and health surveys, 1990-2018. BMC Womens Health. 2022; 22(1): 118.
8. Boeckelmann D, Hengartner H, Greinacher A, et al. Patients with Bernard-Soulier syndrome and different severity of the bleeding phenotype. Blood Cells Mol Dis. 2017;67:69-74.
9. Boeckelmann D, Hengartner H, Greinacher A, et al. Patients with Bernard-Soulier syndrome and different severity of the bleeding phenotype. Blood Cells Mol Dis. 2017;67:69-74.
10. Anna Savoia, Annalisa Pastore, Daniela De Rocco, Elisa Civaschi, Mariateresa Di Stazio, Roberta Bottega, et al. Clinical and genetic aspects of Bernard-Soulier syndrome: searching for genotype/phenotype correlations. Haematologica. 2011;96(3):417-423.
11. Almomani MH, Mangla A. Bernard-Soulier syndrome. In: StatPearls. 2022;30.