Case Report

Bernard–Soulier Syndrome (BSS) & tuberculosis: A case report

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ABSTRACT

Background: Bernard–Soulier Syndrome (BSS) is a rare congenital bleeding disorder characterized by thrombocytopenia. BSS causes bruising, epistaxis, gingival bleeding hemorrhage, post-partum bleeding, gastrointestinal bleeding and post-traumatic hemorrhage, but there is no report of hemoptysis in BSS.

Case report: A 14-year-old girl was referred to this center due to massive hemoptysis. Her chest X-ray revealed complete collapse of the left lung. Rigid bronchoscopy was performed and the intrabronchial clots were removed. Smear and culture of direct sputum was positive for Mycobacterium tuberculosis. She received anti-tuberculosis treatment. During treatment, she developed massive vaginal bleeding, because of drug interaction between rifampin and low dose contraceptive (LD), which she had been taking for control of massive menstrual bleeding. Her vaginal bleeding was controlled by platelet infusion and recombinant factor 7 infusion. After two months of anti-TB treatment, the sputum smear and culture examination became negative. One year after treatment, the pulmonary tuberculosis was completely cured and no hemoptysis occurred.

Conclusion: When hemoptysis occurs in patients with BSS, other differential diagnoses should also be considered.

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Introduction

Bernard–Soulier Syndrome (BSS) is a rare congenital bleeding disorder characterized by thrombocytopenia with giant platelets [1,2]. This syndrome is caused by a quantitative or qualitative defect in the receptor complex of 1b/1X glycoprotein-located on the platelet membrane as a main recipient for Von Willebrand factor (vWF) [3–5]. The clinical manifestations of this syndrome are: bruising, epistaxis, gingival bleeding hemorrhage, post-partum bleeding, gastrointestinal bleeding and post-traumatic hemorrhage [5]. Occurrence of spontaneous bleeding is unusual in BSS, and there is no report of hemoptysis in this syndrome [6].

Case report

A 14-year-old girl was referred to this center with massive hemoptysis which was diagnosed as BSS. She suffered from cough and hemoptysis for longer than 48-h. The patient’s main complaint was pain in the left side of her chest, and she did not present with any other types of pain. Upon physical check-up, no breath sound was heard in the left
hemi-thorax. The period of bleeding was long, and the platelet count was 10,000 per ml. The other clinical parameters such as white cells count, hemoglobin, liver function test, urine test and ECG were normal. The X-ray film and CT scan of chest showed infiltration and bronchiectasis in the left lower lobe with cardiac and mediastinal shift being observed (Figs. 1–3). To remove the clot, rigid bronchoscopy under general anesthesia was performed. Then, due to patient’s resistance to platelet infusion, the platelet infusion was converted to Novoseven infusion. For definitive diagnosis, more complementary examinations were performed, i.e., sputum samples were investigated for the presence of acid-fast bacilli (AFB). Based on the positivity report of smear and culture, the patient received Isoniazid (INH), Rifampin (RF), Pyrazinamid (PZA) and Ethambutol (ETB) for anti-TB treatment. However, as the level of liver enzymes increased and hepatotoxicity were observed, the drug regimen was changed to INH, ETB and Ciprofloxacin (Cip). After taking this regimen, the liver enzyme levels returned to normal status. The patient received this regimen for 12 months. After one year of treatment, the patient was healthy and her monthly check-up showed no sign of diseases.

**Discussion**

BSS, also called hemorrhagiparous thrombocytoc dystrophy, is a rare autosomal recessive coagulopathy (bleeding disorder) that causes a deficiency of glycoprotein lb (GpIb), the receptor for vWF, an important glycoprotein involved in hemostasis [2,7]. In childhood, BSS presents from moderate to severe bleeding. The syndrome’s symptoms consist of: bruising, epistaxis, gingival bleeding hemorrhage, post-partum bleeding, gastrointestinal bleeding and post-traumatic hemorrhage; while spontaneous bleeding is absolutely unusual [6,7]. In this study, hemoptysis was shown as a clinical manifestation of BSS. Indeed, hemoptysis is an awareness sign and should be considered as a symptom of different disease that needs more detailed investigations. The most common causes of hemoptysis are: bronchiectasis, pulmonary tuberculosis and lung cancer. Generally, radiologic manifestations of disease, history, clinical findings upon physical check up and demographic factors guide us toward the evaluation of patients with minor hemoptysis. To the contrary, massive hemoptysis is a life-threatening process and it may cause hospitalization in the intensive care unit (ICU) as well. Immediate examination of the patient by emergency bronchoscopy is necessary for the localization of the bleeding site. Intrabronchial and bronchial artery embolization techniques should be considered as an effective treatment for massive bleeding control and to reduce the requirement of emergency surgical therapy [9]. Additionally, in such cases, the protective treatment which consists of parents or patients’ education play important roles; this consists of training individuals how to handle different types of bleeding, to avoid of minor trauma, to avoid the use of anti-platelet drugs, to handle iron therapy and to avoid the use of Halothane as an anesthetic drug due to a disorder in the platelet’s normal activity. Last, but not the least, prescribing Novoseven factor for patients with severe bleeding. NovoSeven® is a recombinant human coagulation FactorVIIa (rFVIIa), and is a vitamin K-dependent glycoprotein consisting of 406 amino acid residues (MW 50 KDa) [8,10]. NovoSeven in patients with thrombocytopenia or in
congenital or acquired platelet defects, may cause excess thrombin on a surface of activated platelets for homeostasis actions [10].

Conflict of interest

We have no conflict of interest to declare.

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REFERENCES


