Open Access Case Report

Undiagnosed Hemophilia B in an Adult Male: A Case Report

Rafia Behzad
Tahira Zafar
Nadeem Ikram
Lubna Zafar

Hemophilia Patients´ Welfare Centre Rawalpindi

Address for Correspondence
Dr Umbreen Hashim
rafia.behzad@gmail.com

Abstract

We present a case of Haemophilia B patient, previously undiagnosed, who underwent coronary angiography with per cutaneous coronary intervention (PCI) complicated by prolonged post-procedural bleeding. Pressure maneuvers didn't control his bleeding and his condition got worsened which led to an investigation into a potential bleeding disorder.

This report aims to highlight the potential dangers of surgical complications in patients with undiagnosed coagulopathy emphasizing the crucial importance of a thorough diagnostic workup, including a detailed patient history to prevent these complications and ensure safe surgical outcomes.

Keywords: Hemophilia B, Coronary angiography, Post procedural bleeding, Patient history.

Introduction

Hemophilia B, also called Christmas disease, is an X-linked recessive inherited bleeding disorder that results from deficiency of clotting factor IX and is affecting one in every 30,000 males.[1] It accounts for approximately 15-20% of hemophilia cases worldwide.[2] The severity of Hemophilia is categorized based on plasma factor levels ranging from mild (5-40%) to moderate (1-5%) and severe (less than1%).[3] Joint and muscle bleeds are most common in hemophiliac patients. While the severe forms typically present early in their life with bleeding episodes, mild hemophilia cases often remain undiagnosed unless exposed to hemostatic challenge like trauma and surgery which poses them to high risk of bleeding during these provocative events.[3,4] This case of mild hemophilia B, diagnosed after coronary angiography underscores the importance of clinical vigilance. Routine assessments may miss subtle coagulopathies, so a thorough and careful bleeding history is important. Early diagnosis can prevent adverse surgical outcomes in such patients.[5]

Case Report

A 48 years old male presented to outpatient department of Rawalpindi institute of Cardiology (RIC) with complaint of bleeding from his angiography site (right radial artery) for the past 1 month. He was a diagnosed case of Triple Vessel Coronary Artery Disease (TVCAD). He had been doing pressure bandage over his bleeding site for the past 1 month but the pressure maneuvers didn't control his bleeding and his condition got worsened. Several laboratory tests were sent off. The investigations revealed a normal CBC with a hemoglobin of 12.6g/dl, a platelet count of 236,000/ul and normal liver function tests. Coagulation screen showed normal Prothrombin Time (PT) and International Normalized Ratio (INR) however, the Activated partial thromboplastin time (APTT) came out as elevated at 53 seconds (Normal<35 seconds). As only the APTT was affected, he was assayed for clotting factor specific to the intrinsic pathway and was determined to have factor IX deficiency. His factor IX levels were 9.5 IU/dL (normal range 50–150) leading to the diagnosis of 'mild' hemophilia B. The cardiologist planned for his radial artery exploration/ ligation. He was referred to Hemophilia Treatment Center (HTC) Rawalpindi for hematological assessment, control of bleeding and guidance for the surgical plan regarding radial artery exploration which was in plan by vascular surgeon.

On examination, the patient looked pale, alert and a bit hesitant. Aside from his angiography site wound, no sites of active bleeding or bruises were observed. Careful elicitation of history during his post procedure interview revealed bleeding episodes during his childhood for which fresh frozen plasma were transfused after which he remained asymptomatic for decades. He also underwent two to three dental extractions with no intervention needed to control bleeding. The patient had a notable medical history of high blood pressure and ischemic heart disease. His current medications were tablet metoprolol tartrate 50mg twice daily and tablet atorvastatin 20mg once daily, while his anti-platelet drugs were withheld

He was given long-acting factor IX concentrates (Alprolex) to control his active bleeding. The surgical plan for radial artery exploration/ligation was provided according to World Federation of Hemophilia (WFH) guidelines as follows: Preoperatively: factor IX concentrates as 40 IU/kg (30 to 60 minutes before operation) Postoperatively: 30 IU/kg on postoperative day (POD) 2 and 15 IU/kg on POD 4 and 6. The surgery was executed according to the given plan. In addition, ancillary treatment with Injection Transamine (500mg) was initiated a night before surgery and continued for 10 days postoperatively. He underwent his surgery as a high- risk case, a radial artery pseudoaneurysm was found. Clots were evacuated from pseudoaneurysm cavity, hemostasis secured and radial artery was ligated. The patient was counselled and was given guidance for future medical and dental procedures.

Discussion

Mild hemophilia B is a very manageable health condition which may not receive a diagnosis until such patients encounter trauma or undergo surgery, as their bodies produce sufficient factor IX to prevent noticeable bleeding in routine daily activities. However mild hemophilia as in our case, can sometimes complicate small injuries, medical procedures and trauma of surgery.[1] Mild hemophilia patients may incongruently face a higher risk of life-threatening blood loss following surgery or medical procedures due to their often undiagnosed and consequently untreated condition unlike more severe cases that typically receive prompt treatment and management.[6] It substantiates the importance of early diagnosis and management of hemophilia B with prophylactic F-IX infusions. This enables smooth and effective hemostatic control.[7]

Usually, a patient's thorough personal and family history is adequate to assess the bleeding risk making unselective pre-operative coagulation screening unnecessary and further investigations only warranted as needed. However, such coagulopathies remain under-diagnosed in cases where the history is not taken efficiently, as it is evident in this case. [8]

A comprehensive history, in patients with congenital and acquired bleeding disorders plays a vital role in decision-making and management of the patient.[9] But this is a task, which besides health care providers/surgeons depends significantly on patients' ability to accurately recall and report relevant information. This case suggests that the surgeons or health care providers should be vigilant and must have an awareness of the possibility of unexpected bleeding due to previously undiagnosed coagulopathy. Unexpected or prolonged post-operative/ post procedural bleeding may serve as an indication of previously undiagnosed coagulopathy.

Surgeons play a crucial role in the identifying and investigating potential bleeding disorders especially in pediatric patients who may experience their first bleeding episode during surgery, although this responsibility also extends to older patients who may have previously undiagnosed conditions.

It is imperative to ensure comprehensive measures are in place so that we can strive towards a future where patients with bleeding disorders can receive optimal care.[10] Pre-operative history forms be customized to be bleeding disorder-sensitive detailing the type and severity of bleeding disorder, previous bleeding episodes, and any specific precautions, which can help in mitigating any potential risks during the surgery .It is vital to raise awareness and proactive measures in fostering collaboration among healthcare providers, surgeons, anesthesiologists and hematologists in particular, on the significance of a detailed inquiry of any bleeding disorder. Patients' empowerment plays a crucial role in their healthcare journey through active communication, by sharing relevant information and carrying important documentation.

Conclusion

This case highlights the importance of heightened clinical vigilance and comprehensive history taking in surgical patients, especially when encountered with unexpected bleeding. Early diagnosis of mild hemophilia plays a vital role in improving outcomes through tailored perioperative management.

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