Case Report

Volume-8 | Issue-11 | November-2018 | PRINT ISSN No 2249-555X

Immunohematology

NOT ONLY A ROYAL DISEASE: HEMOPHILIA B (A CASE REPORT)

Dr Rounak Dubey Senior Resident, Department of Transfusion Medicine, AIIMS, Raipur

Dr Amit Biswas Assistant Professor, Department of IH & BT, AFMC, Pune

Dr Amit Pawar* Armed Forces Transfusion Centre (AFTC, New Delhi) *Corresponding Author

ABSTRACT Hemophilia B accounts for approximately 1 in 25000 births worldwide. Clinically Hemophilia A and B are indistinguishable. Both result in bleeding which lasts for a longer duration. Hemophilia is a group of X-linked recessive disorders wherein only the males are affected and females are carriers. Protracted bleeding after an invasive procedure may be the only manifestation in less severe forms. We present a case of moderate Hemophilia B diagnosed at a later age due to scarce diagnostic modalities, deficient family history of bleeding and lack of adequate awareness of the disease.

KEYWORDS : Hemophilia, Coagulation disorder, hemostasis, Mixing Studies

INTRODUCTION:

Male descendants of Queen Victoria were cursed with poor health. A number of them bled to death owing to the Royal inheritance of a disease preventing blood from clotting(1). Known as the Christmas disease (Hemophilia B) after a boy named Christmas who was found to be suffering from it, the disease has an X-linked recessive inheritance. Rosemary Biggs et al published a case series in the British Medical Journal in 1952 giving the similarity in presentation of all 7 cases to Hemophilia(2). Hemophilia B however, is a rarer subtype of Hemophilia accounting for approximately 1 in 25000 births worldwide(3) compared to hemophilia A which occurs 1 in 5000 births. Here we present a case of early onset joint pain without any positive family history of bleeding and diagnosed as a case of Hemophilia B only in adulthood.

CASE REPORT:

A 32-year-old native of rural India presented with chief complaints of swelling and pain of the elbow and shoulder joint on and off since birth. He had no history suggestive of any mucocutaneous or frank bleed. Family history was also not contributory. The patient was managed empirically with pain killers. He was operated for a lipoma over left forearm which was followed by unstoppable bleeding. Bleeding was eventually arrested after giving fresh frozen plasma.

Initial coagulation work-up revealed PT – 12.3 sec, APTT – 53.6 sec, INR – 1.00, TT – 15.7 sec, Fibrinogen – 297 mg/dl, Platelet count – 313000/cmm, Fibrin degradation products - negative and D-Dimer – 0.22 microgram/ml. In view of isolated prolonged APTT, mixing study was done. On incubating normal pooled plasma with patients' platelet poor plasma for 2 hours the APTT got corrected to 38.4 sec, while without incubation APTT of the mixture was 38.2 sec. This indicated a factor deficiency which could have been responsible for the bleeding. Subsequently, plasma Factor VIII and IX activity (Normal range – 50 to 160%) were found to be 100% and 12% respectively. The results of the investigations are summarized in Table 1. Considering the presenting features and the abnormally low Factor IX levels a diagnosis of moderate Hemophilia B was made. The patient was then enrolled in the Hemophilia Society of India (Pune Chapter) for requirement of Factor IX concentrate.

TABLE 1 : Result Of The Investigations Performed On The Patient

TEST PERFORMED	VALUE	REFERENCE RANGE
PT	12.3 sec	12-15 sec
APTT	53.6 sec	34-38 sec
FIBRINOGEN	297 mg/dl	150-350 mg/dl
PLATELET COUNT	3,13,000/cmm	1.5 - 4.5 lakhs/cmm
FDP	Negative	-
CORRECTED APTT (AFTER INCUBATION)	38.4 sec	-
CORRECTED APTT (WITHOUT INCUBATION)	38.2 sec	-
Plasma Factor VIII activity	100%	60 – 100 %
Plasma Factor IX activity	12 %	60 – 100 %

DISCUSSION:

Deficiencies of factors VIII, IX and XI result in Hemophilia A, B and C respectively. Hemophilia A is the most common followed by Hemophilia B (Christmas disease), while Hemophilia C is a rarer entity with a prevalence of 1 in 100000(4). Hemophilia is a group of X-linked recessive disorder wherein only the males are affected and females are carriers. The female carriers may exhibit the disease rarely due to lyonization. There is a 50% chance of male progeny of a female carrier being affected and 50% chance of her female progeny being born a carrier for the disease(5).

Clinically Hemophilia A and B are indistinguishable. Both result in bleeding which lasts for a longer duration. Common sites of bleeding are the joints, muscle/soft tissues, skin/subcutaneous tissues, gum and nasal mucosa. Protracted bleeding after an invasive procedure may be the only manifestation in less severe forms(6). It can also result in rapidly fatal internal organ hemorrhages depending on the severity of the condition. Classically, depending on the plasma factor activity it can be categorized into mild (Factor activity – 5 to 40%), moderate (Factor activity – 2 to 5%) and severe (Factor activity – less than 1%). This traditional classification predicts not only the onset and severity of bleeding and also guides the management of such cases(7).

This patient had a moderate deficiency of Factor IX, which could be the reason for the diagnosis at such later age and only after an invasive procedure. The patient belonged to a rural background where these costly and highly specialized tests are not available. Lack of family history of bleeding could have further precluded the diagnosis. Only 16000 hemophiliacs are presently registered with the Hemophilia Society of India(8). This represents just the tip of the iceberg. Being an X-linked recessive disease, a basic coagulation screening of the males before marriage could detect the milder cases and prevent the birth of a hemophiliac who requires a massive medical, social and psychological support. However easier said than done, a colossal effort needs to be put in to increase the awareness.

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