Christmas Disease (Hemophilia –B) – A Case Report

Md. Rafiqul Alam[1], Mohammad Saiful Habib[2], Mohammad Arifur Rahman[2], Md. Mizanur Rahman Khan[2], M A Jalil Chowdhury [3], Taimur A K Mahmud[3]

[1]Assistant Professor, [2] Medical Officer,[3]Professor, Department of Medicine, Bangabandhu Sheikh Mujib Medical University, Shahbag, Dhaka-1000, Bangladesh

Abstract:

We report a 25 years old man developed Haemarthrosis of left hip joint with a history of recurrent swelling and pain in multiple joints and prolonged bleeding following minor trauma since childhood. Subsequent investigations revealed Christmas disease (Haemophilia B). Hemophilia B is an X-linked bleeding disorder. This case emphasises the importance of considering a diagnosis of haemophilia in a man with unexplained bleeding, even in the absence of a positive family history.

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Introduction:

Christmas disease (Hemophilia B, factor IX hemophilia) is a rare bleeding disorder due to deficiency of coagulation factor IX [1]. Most commonly factor IX is quantitatively reduced, but in one-third of cases an abnormally functioning molecule is immunologically present. Factor IX deficiency is one–seventh as common as factor VIII deficiency hemophilia but is otherwise clinically and genetically identical.

Factor IX deficiency or dysfunction occurs in 1 in 100,000 male births[2]. Accurate laboratory diagnosis is critical, since it is indistinguishable clinically from factor VIII deficiency (hemophilia A) but requires different treatment.

Case Report:

A 25 years old college student hailing from Mymenshing got himself admitted in BSMMU hospital on 2nd February, 2009 with complaints of pain and restricted movement of left hip joint for 10 days, recurrent swelling and pain in multiple joint for last 20 years and prolonged bleeding following minor trauma since childhood.

On examination patient is mildly anaemic, non icteric, pulse-92/min, BP- 110/70 mm of Hg, no edema or lymphadenopathy. On examination of musculoskeletal system, there was wasting of thigh and calf muscles, tone was normal, power was 4/5, movement was reduced in both knee & elbow joints, as well as left hip joint. Nervous system and other system examination reveled nothing abnormal.

Address of Correspondence: Dr. Md. Rafiqul Alam, Assistant Professor, Department of Medicine, Room No: 632, Block- C, BSMMU, Email: taimur@bsmmu.org

Investigation findings were Hb-15gm/dl, ESR-20mm in 1st hour, Platelet count-330000/cmm, PBF- Normal, BT- 2.30 sec, CT- 6.00 sec, PT- 14.5 sec (INR-1.2), APTT- 82.8 sec, Plasma factor VIII activity – 121% and Plasma factor IX activity – 3.9%. Considering all features he was diagnosed as a case of Christmas disease with Haemarthrosis of left hip joint and was treated with fresh frozen plasma only.

Discussion:

Christmas diseases is an X-linked recessive disorder. Only males are affected [3]. All daughters of diseased are obligate carriers and sisters have a 50% chance of being a carrier. If a carrier has a son, he has a 50% chance of having Christmas disease, and a daughter has a 50% chance of being a carrier .Female carriers of Christmas disease may suffer due to lyonisation. Here there is a low level of factor IX coagulant activity (Normal- 50-150%) [4]. Most common site of bleeding are joints (knee, ankles, elbows), muscles and from gastrointestinal tract. Patients with severe disease bleed spontaneously. A severely affected, may have one or two bleeds each week. Recurrent bleeding into joints lead to synovial hypertrophy, destruction of the cartilage and secondary osteoarthrosis. Muscle haematoma (calf and psoas muscles) are also characteristic of this disease. Untreated haematomas causes subsequent contraction and shortening. Intracranial hemorrhage also occur which is often fatal.

In Christmas disease APTT is prolonged. Prothrombin time, bleeding time, fibrinogen level and VWF are normal. Factor VIII activity is normal but factor IX activity is reduced[5]. Christmas disease is managed with factor IX concentrates. 80 units/ Kg is necessary to achieve a 100% level. Half-life of factor IX is 18 hours. During major surgery 80 units/Kg (6000 U) initially followed by 40 units/Kg (3000 U) every

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18 hours. Factor IX concentrates can be used prophylactically twice weekly[6]. DDAVP is not useful in this disorder. Patient should be cautioned to avoid aspirin. All patients should be registered at comprehensive care centers (CCC) for Medical, Social and Psychological Supports.

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