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Case Report

Haemophilia B- A coagulation disorder

Neena Sharma [1,*]

[1] *Dept. of Physiology, Acharya Shri Chander College of Medical Sciences, Jammu, Jammu and Kashmir, India*

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ABSTRACT

Background: Haemophilia is a group of inherited blood disorders in which blood does not clot properly. Bleeding disorders are due to defect in blood vessels, the coagulation mechanism or the blood platelets. When the coagulation factors are deficient, the blood does not clot properly and bleeding continues.
Haemophilia is a X-linked recessive disorder that affects males, while the females are protected by normal gene on X-chromosome. Haemophilia-A is the most common genetic defect due to deficiency of Factor VIII while Haemophilia-B is the second most common genetic defect due to deficiency of Factor IX also called as Christmas disease.
The present case report is of a 18-year-old male boy admitted in emergency wing of Government Medical College and Hospital, Jammu, who developed iliopsoas haematoma which after complete investigations revealed deficiency of clotting Factor IX.
Aim: A report of presentation of a case of haemophilia-B.
Objective: The present case report was done to demonstrate the sign and symptoms of haemophilia-B, complete haematological, bleeding and clotting test done for diagnosis and its treatment.
Results: The case of haemophilia-B presented with sudden inability to walk demonstrating right iliopsoas haematoma on ultrasonography. The patient was anaemic with haemoglobin – 7 gm%, TLC – 7000 mm^3 and platelets 2.5 lakh/μl. The prothrombin time was decreased (10 seconds), activated partial thromboplastin time was increased (45 seconds), Factor VIII levels were within norrmal range, while Factor IX was decreased. Finally, a diagnosis of haemophilia-B was made.

1. Introduction

Blood coagulation mechanism is a process which transforms blood from a liquid into a solid and involves different clotting factors that generates fibrin fibers through extrinsic or intrinsic coagulation pathway which along with platelet plug stops bleeding. When coagulation factors are deficient, the blood does not clot properly and bleeding continues. An affected individual may bleed spontaneously or for longer than a healthy person after injury or surgery.

Haemophilia is a X-linked recessive disorder that affects males, and females are protected by normal gene on other X-chromosome. Queen Victoria was a carrier of haemophilia and passed the mutation to her son Leopold and through her daughters to members of Royal families of Spain, Russia and Germany. [1]

Patients with haemophilia-A and haemophilia-B have genetic defect in deficiency of blood clotting factors. [2] Haemophilia-A is the most common genetic defect due to deficiency of Factor VIII – 70% of cases are inherited as X-linked recessive trait and 30% of cases have no family history and arise form spontaneous mutations. Haemophilia-B is the second most common genetic defect,

* *Corresponding author.*
E-mail address: neenasharma1404@gmail.com (N. Sharma).

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also called as Christmas disease after the name of Stephen Christmas who had the disease.[3] It is an X-linked recessive disorder caused in 70% of cases by mutation of Factor IX gene located on X-chromosome (Xq27.1 – q27.2) leading to deficiency of factor IX and in 30% of cases by spontaneous mutation.[4]

2. Case Report

An 18-year old male developed pain in the right leg and was unable to walk. The pain was so severe to the extent that he could not move from the bed. There was no history of trauma, fall or injury. The symptoms began instantaneously. With these complaints he was admitted to the Government Medical College, Jammu. General physical examination revealed pallor, pulse 62 beats/minute and hypertension. Examination of the right leg revealed tenderness in right thigh with swelling. Investigation profile revealed haemoglobin – 7 gm%, TLC – 8000 mm^3, DLC – N-64, L-25, M-5, E-2 and platelets 2.5 lakh/μl. Peripheral blood film showed microcytic hypochromic anemia.

Ultrasonography of right thigh showed 9.5×4.5 cm heterogenous area in right iliopsoas indicating right iliopsoas haematoma. The ultrasonographic report of haematoma in the iliopsoas muscle pointed towards bleeding disorder as there was no history of injury, fall or trauma. The platelet count was within normal range.

Other investigations conducted were prothrombin time, activated partial thromboplastin time (aPTT), Factor IX and Factor VIII levels. The prothrombin time was 10 seconds (normal – 11 seconds), aPTT 45 seconds (normal – 34 seconds), Factor IX 20 IU (normal 70-120 IU) and Factor VIII 80 IU (normal 60-150 IU).

Prothrombin time explores the defect in extrinsic coagulant pathway. The prothrombin time was recorded to be normal. The activated partial thromboplastin time takes into account any effect of deficiency in intrinsic coagulation pathway. It is a screening test for disorder of intrinsic pathway, mainly Factor VIII, Factor IX and Contact Factor, thus permitting the diagnosis of haemophilia. The aPTT was increased in the subject which pointed towards the inner of intrinsic pathway. Factor VIII levels were determined which was within normal range but Factor IX levels were decreased. Finally, a diagnosis of haemophilia-B was made in context of increased aPTT and Factor IX levels.

The subject was administered replacement therapy with slow intravenous infusion of concentrate of clotting Factor IX 4000 U on day 1 and day 2 and the dose was tapered to 2400 U on day 3 and finally 1800 U on day 4 and day 5. In addition to the replacement therapy with clotting factor, symptomatic treatment for pain, and antifibronolytic drugs were given and the patient was discharged on day 6.

3. Discussion

Haemophilia-B is less common than haemophilia-A, affecting 1 in 25,000 males. Sometimes a baby is born with haemophilia-B when there is no family history i.e., the gene may be hidden or passes through several generations of female carriers without affecting male members or change in X-chromosome is new due to spontaneous mutation.

The normal level of Factor IX range from 50-150% and the severity of haemophilia depend on the amount of clotting factor in blood. On the basis of amount of clotting factor in the blood, there are three levels of haemophilia:

1. **Mild haemophilia:** There is 5-50% of normal clotting factor in blood. The presentation of the patient with mild haemophilia-B is such that the post-bleeding episode may not occur till adulthood, unless an injury, surgery or tooth extraction results in prolonged bleeding. Women with mild haemophilia present with menorrhagia, heavy menstrual periods and haemorrhage after child birth.
2. **Moderate haemophilia:** There is 1-5% of normal clotting factor in blood. The patients have bleeding episodes after injury or without any cause called spontaneous bleeding episodes.
3. **Severe haemophilia:** There is less than 1% of normal clotting factor in blood. The patients have frequent spontaneous bleeding episodes often into joints and muscles.

The mainstay of treatment for haemophilia is replacement therapy with concentrates of clotting Factor VIII for haemophilia-A and clotting Factor IX for haemophilia-B given slowly intravenously. Home treatment for haemophilia is advocated as the clotting factors are easy to store, mix and use at home. But home treatment can lead to formation of antibodies or inhibitors in 20-30% of people with severe haemophilia-a and 2-5% of people with haemophilia-B. These antibodies can destroy the clotting factor before it has chance to work.

A known complication of replacement therapy is that the clotting factors made from human blood can carry the viruses that cause AIDS and hepatitis. However, this can be prevented by careful screening of blood donors, testing of donated blood products, heat treatment of donated blood products and vaccinating the people who have haemophilia for hepatitis-A and B.

The delay in treatment of haemophilia can lead to the damage to joints, muscles or other parts of body. In addition to the replacement therapy with clotting factors, antifibrinolytic drugs like tranexemic acid and epsilon amino caproic acid is used to prevent the clot from breaking down. Research studies are being undertaken by researchers to correct faulty genes that cause haemophilia.

4. Conclusion

A case of haemophilia-B after diagnosis needs replacement therapy with slow intravenous infusion of concentrate of clotting Factor IX, symptomatic treatment for pain and antifibrinolytic drugs.

5. Source of Funding

None.

6. Conflict of Interest

The authors declare no conflict of interest.

References

1. Price M. Case closed: famous Royals suffered from haemophilia; 2009.
2. Biggs R, Douglas AS, Macfarlene RG, Dacie JV, Pitney WR, Marskey C, et al. Disease: a condition previously mistaken for haemophilia. *Br Med J*. 1952;2(4799):1378–82. doi:10.1136/bmj.2.4799.1378.
3. Christmas Disease". Who named it? . Available from: www.whonamedit.com/synd.cfm/2321.html.
4. Rogaer EI, Grigorenko AP, Faskhutdinova G, Kittler ELW, Moliaka YK. Genotype analysis identifies the cause of the 'Royal Disease. *Science*. 2009;326(5954):817.

Author biography

Neena Sharma, Associate Professor https://orcid.org/0000-0003-4096-5117

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