Hemophilia B with Spontaneous Intraperitoneal Bleeding: A Case Report

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Abstract:

Hemophilia B is a congenital bleeding disorder due to the decrease of the clotting factor IX activity. Its common manifestation in severe form is spontaneous hemarthrosisparticularly at the weight-bearing joints. Herein we report an 18-year old Thai man who was diagnosed as severe hemophilia B since two months of age and he continually has had one or two episodes of hemarthroses a year. He repeatedly has an isolated prolongation of aPTT and his factor IX activity at the steady state is around 0.8% while factor VIII activity is normal. At this admission, he spontaneously develops acute abdominal pain for one day due to the intraperitoneal bleeding,the unusual site of spontaneous bleeding manifestation of hemophilia B. He has no previous abdominal trauma. The computerized tomography of the abdomen confirms lots of hemoperitoneum and the thickening of bowel wall that is supposed to be the original site of bleeding whereas the liver and spleen appear normal. His hematocrit is found to be 18% and he is treated with many units of packed red blood cell transfusion as well as factor IX concentrate 5,000 unit every day. His abdominal symptom is gradually improved and he can be discharged within seven days without any operation.

Key Words: Hemophilia B, Spontaneous Intraperitoneal Bleeding

บทคัดย่อ: ฮีโมฟีเลีย บี ที่เกิดภาวะเลือดตกในช่องท้องได้เอง: รายงานผู้ป่วย 1 ราย

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ฮีโมฟีเลีย บี เป็นความผิดปกติแต่กำเนิดที่มีอาการเลือดออกง่าย หยุดยาก เนื่องจากกิจกรรมของปัจจัยที่ IX ของการแข็งตัวของเลือดลดลง อาการแสดงที่พบบ่อยในผู้ที่มีอาการรุนแรง คือ ภาวะเลือดออกเองในข้อ โดยเฉพาะข้อที่ต้องรับน้ำหนักในรายงานนี้เป็นผู้ป่วยชายไทย อายุ 18 ปี ซึ่งได้รับการวินิจฉัยว่าเป็น ฮีโมฟีเลีย บี ชนิดรุนแรง ตั้งแต่อายุได้ 2 เดือน หลังจากนั้นผู้ป่วยจะมีเลือดออกในข้อประมาณ 1-2 ครั้งต่อปี ตรวจพบเพียง aPTT ยาวนานกว่าปกติหลายครั้ง ระดับกิจกรรมของปัจจัยที่ IX ช่วงที่ไม่มีอาการอยู่ประมาณร้อยละ 0.8 ในขณะที่ระดับกิจกรรมของปัจจัยที่ VIII ปกติในครั้งนี้ผู้ป่วยมีอาการปวดท้องกะทันหันใน 1 วัน เนื่องจากตกเลือดในช่องท้อง ซึ่งถือว่าเป็นตำแหน่งเลือดออกที่พบไม่บ่อยในโรค ฮีโมฟีเลีย บี ผู้ป่วยไม่เคยมีภยันตรายใด ๆ ที่หน้าท้องมาก่อน ตรวจเอกซเรย์คอมพิวเตอร์ช่องท้อง พบเลือดจำนวนมากในช่องท้อง ผนังลำไส้บวมหนาซึ่งเชื่อว่าเป็นตำแหน่งที่เลือดออก ส่วนตับและม้ามปกติตรวจเลือดพบhematocrit 18% ผู้ป่วยได้รับการรักษาด้วยการเติมเลือดหลายหน่วยร่วมกับปัจจัยที่ IX เข้มข้น 5,000 หน่วยทุกวันอาการทางหน้าท้องค่อยดีขึ้นตามลำดับ และได้รับอนุญาตให้กลับบ้านได้ในวันที่ 7 โดยไม่ต้องผ่าตัด

คำสำคัญ: ฮีโมฟีเลีย บี, ภาวะตกเลือดเองในช่องท้อง

Introduction

Hemophilia B is the congenital bleeding diathesis due to deficiency of factor IX clotting activity. It is genetically transmitted as sex-linked recessive as hemophilia A but it is rarer. For the severe case which is defined if the factor activity is less than 1%[1], the main manifestation is the spontaneous bleeding symptom whereas the common sites of such bleeding area joint, so-called the hemar-throsisespecially the weight-bearing joints with possible chronic arthropathy as a late complication[2]. The hemarthrosis accounts for 80 % of all bleedingmani-festations, it can suddenly produce pain and swelling in mainly one joint at a time[3]. Whereasthe ankle is the most frequently affected joint in the children, the knee, elbow and ankle are the most frequent affected joints in adults[4]. The less common sites include themuscleparticularly the quadriceps, iliopsoas and the forearm leading to hematoma formation, the central nervous system especially the spontaneous intracerebral hemorrhage, the gastrointestinal tract, and the genitourinary tract[3].

The intraperitoneal bleeding in case of hemophilia B hasbeen very rarely reported(5). Herein we report a case of spontaneous intraperitoneal bleeding in a known case of hemophilia B.

Case Report

An 18-year old Thai man presented with acute abdominal pain and fullness at the epigastrium after drinking heavy alcohol. He vomited for many times and most vomitus was food debris without blood. His abdominal pain was continued and gradually increased through a day. He had no fever/bowel habit change/ ecchymosis/abdominal trauma. His physical examination revealed only generalized slightly distended abdomen with mild tenderness and nohepatosplenomegaly or no joint deformity was found. In the past, he was diagnosed as severe hemophilia B since two months of age because of the spontaneous swelling of one knee joint, and low factor IX activity. And his elder brother was already diagnosed as severe hemophilia B before he had hemarthrosis. After the diagnosis of

hemophilia B was established, he continually had the averaged attack of the hemarthrosis one or two times a year and was treated with factor IX concentrate and no residual joint deformity was left.

Laboratory tests: Hb5.7 g%, Hct18.0%, WBC 18,600/mm^3, platelet 313,000/mm^3,NRBC 1/100 WBC, N 94%, L 6%, normal liver and kidney function test, amylase 40 U/L (normal <100 U/L), lipase 8.0 U/L (normal <60), urine amylase 550 U/L (normal <450). Coagulo-gram: PT 13.4 sec (control 8.9-14.6), aPTT 41.8 sec (control 21.9-34.2), TT 19.8 sec (control 16.8-22.7), INR 1.11, factor VIII activity 96.5% (control 50-150), factor IX activity 0.8% (control 73-109),factor VIII inhibitor<1 BU, factor IX inhibitor-negative

Anti-HIV, HBsAg, anti-HCV were negative.

The computerized tomography of the abdomen showed lots of intra-abdominal free fluid, maybe ascites or hemoperitoneum; bulging inhomogeneous thickened wall of a bowel loop at posterior mid and left sided abdomen which showed streak arterial enhancing, maybe bleeding from bowel wall with internal intra-muscular hematoma, and suspected extension of hematoma into the peritoneum; unremarkable liver, gall bladder, pancreas, spleen, kidneys, bladder and adrenal glands.

He was diagnosed asspontaneous hemoperito -neumpossibly due to bleeding from the bowel wall with the underlying severehemophilia B and treated with packed red blood cells transfusion and concentrate factor IX injection of 5,000 units a day. His abdominal manifestations were gradually improved without abdominal tapping or surgery and he could be discharged onthe 7th day of admission.

Discussion

Our patient is formerly diagnosed ashemophilia B based on the isolated prolongation of aPTT, markedly decreased activity of factor IX (0.8%), normal factor VIII activity, no factor VIII and IX inhibitors and the family back ground of hemophilia B in his elder bother. And he usually responds well to concentrated factor IX transfusion.

Actually hemophilia is the most common congenital bleeding disorder in Thailand. In the national survey of congenital bleeding disorders, 1,325 from 1,450 patients (91.4%) are hemophilia A and hemophilia B[6]. In general, the prevalence of hemophilia B isaveraged to be 0.17±0.12 per 100,000 males[7] and it is found about 5 times less common than hemophilia A[8].

While the joint symptom is the most common manifestations of hemophilia, the mucosal bleeding is considered uncommon such as epistaxis, hematuria, melena, hemoptysis, conjunctival hemorrhage, intracerebral hemorrhage, hemothorax, hemoperitoneum that is found in one from 14 cases of hemophilia B[8].

Some causes which can be identified in cases of the spontaneous intraperitoneal bleeding in hemophilia, are the rupture of the spleen in hemophilia B that may occur spontaneously or from minute trauma[5], the spontaneous rupture of the left gastroepiploic artery in a moderate hemophilia A[9], and the ruptured intramural intestinal hematoma in severe hemophilia A[10]. But the spleen in our case is demonstrated unremarkable on the CT study. On contrary, the bowel wall is thickened and it is supposed to be the original source of severe bleeding into the peritoneum. And it

does not need the surgical intervention.

The intraperitoneal bleeding has high attenuation on the computerized tomography, so it can be differentiated from simple ascites that will have low attenuation[11]. Although the intraperitonealbleeding in hemophilia is uncommon, its recognition is important because it can mimic other intra-abdominal processes. Therefore, its diagnosis may be delayed leading to the improper management and fatal outcome[12].

Conclusion

An 18-year old Thai man presents with acute abdominal pain for one day due to the spontaneous intraperitoneal bleeding with the underlying severe hemophilia B. It is an uncommon manifestation of hemophilia but it is important because its delayed diagnosis can result in the fatal outcome.

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