Factor-X deficiency; a rare disorder to be looked for in cases of congenital bleeding tendency

by Fatima Ayaz, Saeed Bin Ayaz, Muhammad Furrukh

Submission date: 16-Mar-2018 11:16AM (UTC+0500)
Submission ID: 931238311
File name: 18270-72480-1-SP.docx (26.49K)
Word count: 1181
Character count: 6837

Title

Factor-X deficiency; a rare disorder to be looked for in cases of congenital bleeding tendency

Key words

Bleeding disorder, factor-X deficiency, inherited

Dear Editor;

Factor X deficiency is a rare coagulation disorder with autosomal recessive inheritance pattern. People with this disorder present with a myriad of early life bleeding complications. We report here a case who had presented to the physicians with many bleeding complications at different times of his life but was not diagnosed earlier.

A 23-years-old male presented to the medical emergency unit of our hospital with complaints of hematuria for five months, black stools for ten days, and bleeding from nose for four days. He had a history of recurrent bleeding from different sites, starting from the time of his circumcision on seventh day of his life. The circumcision wound bled so severely that his wound had to be sutured to achieve hemostasis. At the age of 11 years, he developed severe epistaxis, for which he remained admitted in the hospital for 15 days and received transfusion of a standard pack of red cell concentrate (RCC). At 12 years of age, he was re-admitted for a massive hematoma in right leg after getting injection from a quake that resulted in weakness of his right leg due to nerve compression. After seven years, he was operated upon for the drainage of psoas abscess. During operation, he developed severe bleeding, and had to be transfused with 18 pints of fresh frozen plasma (FFP), and one pack of RCC. He remained admitted in the hospital for almost seven months when his wound healed completely after daily wound wash, and aseptic dressings. The patient did not report fever, chills, night sweats, or weight loss. He denied tobacco, alcohol, or drug abuse.

He was born to consanguineous parents. One of his elder sister had complaints of heavy menstrual bleeding since menarche but was not yet investigated. He was not taking any medications affecting coagulation function.

On examination, he had marked palmar and conjunctival pallor. His vital signs were normal. Examination of the oral cavity did not show any petechiae or gingival anomalies. Abdominal examination showed scar mark of drainage of psoas abscess and associated incisional hernia. There was no lymphadenopathy nor abdominal organomegaly. Rest of the systemic examination was unremarkable.

His laboratory evaluation showed a hemoglobin of 11.2 g/dL (normal range: 12.9–16.1 g/dL)[2], and a normal platelet and total leucocyte count. The prothrombin time (PT) was 30 s (normal: 15 s), and activated partial thromboplastin time (aPTT) was 62 s (normal: 33 s). His D-dimer values were >250 ng/mL D-Dimer Units (normal: ≤250 ng/mL D-Dimer Units) but the fibrinogen level was not decreased, excluding the possibility of disseminated intravascular coagulation. The total protein levels were in the normal range. PT and aPTT mixing studies corrected with a 1:1 mix with normal plasma (1). The assessment for serum complement levels, antinuclear antibody, and antinuclear cytoplasmic antibody revealed normal values. Serologic tests for hepatitis B and C and rheumatoid factor were non-reactive. The levels of different coagulation factor levels were tested and only factor X levels were decreased i.e. 13% (normal: 50-150%)

He was managed with 6 pints of FFPs because factor X concentrates were not available. (Figure-1A) He was discharged after ten days following complete resolution of bleeding. Prior to discharge, he was properly counseled about the disease and management options, and was advised to seek immediate medical care in case of bleeding from any site.

Seven months after discharge, he presented again to the hospital with non-healing wound, and purulent discharge from the site where psoas abscess was surgically drained. After diagnostic imaging, the patient was diagnosed to have enterocutaneous fistula that was excised surgically,

and right hemicolectomy with ilio-colic anastomosis had to be done. The wound of the surgical incision did not heal, and purulent discharge persisted. The patient was again operated upon for exploration. The surgeon found multiple adhesions of descending colon and small-gut loops and small-gut tears along with many long fistulous communications in right hepatic flexure. The adhesions were broken, affected portion of the small gut and descending colon were removed, and tube colostomy was done in the right hypochondrium while ileostomy was constructed in the left hypochondrium. (Figure-1B). The wounds for ileostomy and colostomy kept on bleeding for three months before coming to an arrest, and he had to be transfused with 4-5 pints of FFPs every week. The patient is now getting food orally, and stable on home medications and colostomy.

Factor X is the first enzyme in the coagulation pathway to fibrin formation. Factor X deficiency has an estimated prevalence of 1 in 5 -10 x 10^5 in the general population (1), more common in populations in which consanguineous marriage is traditional (1). Pakistan has a reported incidence of 3.5%, 6%, and 26.1% in three different reports about patients with bleeding disorders (2-4). Patients with congenital factor X deficiency can present at any age. Generally, patients with more severe disease present during infancy. Heterozygotes are often clinically asymptomatic. Homozygous individuals experience symptoms such as easy bruising, hematuria, soft-tissue hemorrhages, hemarthroses, epistaxis, and menorrhagia (5). Differential diagnoses of Factor X deficiency include von-Willebrand disease, Waterhouse-Friderichsen syndrome, Cryoglobulinemia, Cushing Syndrome, Disseminated Intravascular Coagulation, Dysfibrinogenemia, Factor II, IX, V, VII,VIII, XI deficiency, Hemolytic-Uremic Syndrome, Immune Thrombocytopenic Purpura, Osler-Weber-Rendu Syndrome, Scurvy, Thrombotic Thrombocytopenic Purpura, and Vitamin K Deficiency.

Laboratory findings include prolonged PT and prolonged aPTT. For the treatment of factor X deficiency, restoring circulating factor X levels to 10-40% of normal is usually adequate. Therapeutic measures may include infusion of FFP, and prothrombin complex concentrates. The prognosis for patients with factor X deficiency depends on the severity of the disease. In general, patients with very low levels of functional factor X have a greater tendency to bleed, and face a greater risk of life-threatening complications (6).

In conclusion, factor X deficiency, though rare, is a life-threatening disease. Its knowledge is important to differentiate it from other disorders of coagulation. Timely diagnosis of the condition will save the patient from unnecessary interventions (like Factor VIII concentrate injections), and will lead to adequate management.

References

1. Brown DL, Kouides PA. Diagnosis and treatment of inherited factor X deficiency. Haemophilia 2008; 14(6):1176-82.
2. Anwar M, Hamdani SN, Ayyub M, Ali W. Factor X deficiency in North Pakistan. J Ayub Med Coll Abbottabad 2004;16(3):1-4.
3. Borhany M, Shamsi T, Fatima N, Fatima H, Naz A, Patel H. Rare Bleeding Disorders are not so Rare in Pakistan. J Hematol Thromb Dis 2013; 2:122.
4. Khalid S, Bilwani F, Adil SN, Khurshid M. Frequency and clinical spectrum of rare inherited coagulopathies--a tricenter study. J Pak Med Assoc 2008;58(8):441-4.
5. Uprichard J, Perry DJ. Factor X deficiency. Blood Rev 2002;16 (2):97-110.
6. Coucke L, Trenson S, Deeren D, Van Haute I, Devreese K. Life-threatening bleeding tendency provoked by an acquired isolated factor X deficiency associated with respiratory infection. Ann Hematol 2013; 92(10):1437-8.

Figure legends

Figure-1: 1A : Figure showing the patient being transfused with fresh frozen plasma at initial presentation. 1B: Figure showing the patient with right tube colostomy and left ileostomy

Factor-X deficiency; a rare disorder to be looked for in cases of congenital bleeding tendency

ORIGINALITY REPORT

23%	14%	19%	%
SIMILARITY INDEX	INTERNET SOURCES	PUBLICATIONS	STUDENT PAPERS

PRIMARY SOURCES

1	Manikkan, Ajaydas T.. "Factor X deficiency: An uncommon presentation of AL amyloidosis", Upsala Journal of Medical Sciences, 2012. Publication	5%
2	emedicine.com Internet Source	5%
3	misc.medscape.com Internet Source	4%
4	onlinelibrary.wiley.com Internet Source	2%
5	M. Sinan Beksaç. "Severe factor X deficiency in a twin pregnancy", Archives of Gynecology and Obstetrics, 04/26/2009 Publication	2%
6	Vinod, K. Hitha, B. Kaaviya, R. Dutta, T.. "Near fatal spontaneous intraperitoneal bleeding: A rare manifestation in a congenital factor X defic", Indian Journal of Critical Care Medicine, March 2015 Issue	1%

Publication

7	Gandhi, Saumil Kalantar-Zadeh, Kamyar Do. "Thin-glomerular-basement-membrane nephropathy: is it a benign cause of isolated hematuria?", Southern Medical Journal, July 2002 Issue Publication	1%
8	The Coagulation Consult, 2014. Publication	1%
9	mayomedicallaboratories.cws.net Internet Source	1%
10	"ABSTRACTS OF SUBMISSIONS ACCEPTED FOR PRESENTATIOIN", Journal of General Internal Medicine, 4/2005 Publication	1%
11	Deng, Xingqi, Yanyan Li, Ling Zhou, Chunyan Liu, Mei Liu, Nianchang Ding, and Jinyan Shao. "Gender Differences in the Symptoms, Signs, Disease History, Lesion Position and Pathophysiology in Patients with Pulmonary Embolism", PLoS ONE, 2015. Publication	1%

Exclude quotes	On	Exclude matches	Off
Exclude bibliography	On		