International Journal of Current Advanced Research
ISSN: O: 2319-6475, ISSN: P: 2319-6505, Impact Factor: SJIF: 5.995
Available Online at www.journalijcar.org
Volume 6; Issue 12; December 2017; Page No. 8302-8303
DOI: http://dx.doi.org/10.24327/ijcar.2017.8303.1331

Research Article

POST TRAUMATIC INTRAMUSCULAR HEMATOMA UNVEIL A RARE COAGULATION DISORDER: FACTOR X DEFICIENCY

Mahmoud EL Hussein., Shafiqa Saleh., Sura Al Doory., Mohamed Ali and Maysa Saleh*

Pediatric Department, Latifa Women and Children hospital, UAE

A R T I C L E I N F O

Article History:
Received 20th September, 2017
Received in revised form 5th October, 2017
Accepted 16th November, 2017
Published online 28th December, 2017

Key words:
Factor X; haemophilia; coagulant; autosomal recessive

A B S T R A C T

Factor X has an essential role in the fibrin formation pathway, as it is the first enzyme in this cascade. Factor X is one of the rarest bleeding disorder that can be inherited or acquired. This case report describes two members of a family who were found to have factor X deficiency. A 2 years 9 months old boy who presented to the ED with intra-muscular hematoma. Work up revealed factor X deficiency. The low incidence and rarity of this deficiency producing significant complication compelled us to report this case.

INTRODUCTION

Background

Factor (FX), which is also known as Stuart-Prower factor, is a vitamin K–dependent enzyme with a pivotal role in the coagulation cascade (1). It can be inherited or acquired. Inherited Factor X deficiency is one of the rarest bleeding disorders; which was estimated to occur in 1: 1000000 individualsto 1: 2000000 (2, 3). Its inheritance is autosomal recessive; the heterozygous inheritance is usually asymptomatic. On the other hand, the homozygous individuals usually experience episodes of bleeding which range from mild bruising to hematuria, soft soft-tissue hemorrhages, hemarthroses, recurrent epistaxis, and menorrhagia (4). The acquired factor x deficiency is a sequence of vitamin K deficiency, drugs, or severe liver disease (5).

Case presentation

A 2 years 9 months old boy admitted with one-week history of limping, right leg painand swelling post traumatic. On second day posttraumatic, he sought medical advice when he was reassured and discharged on conservative management. As the pain and swelling were not improving hence brought again to hospital. There was a history of similar episodes of limb swelling and pain after sustaining minor trauma. No history of prolonged bleeding after circumcision. No history of epistaxis. He was born full term to consanguineous parents, his elder sister is healthy, but his father and elder brother have history of recurrent epistaxis. On examination he was lying comfortably in bed. He is pinkish with pain score 0 and stable vital signs. Right calf was swollen with tenderness. No joint swelling and peripheral pulses were intact. Rest of examination was unremarkable.

Haematological investigations; FBC FBC: WBC Count 13.6 10^3/uL, HGB 10.4 g/dL, MCV 67.7 fL, MCH 20.9 pg , RDW 17.1 %, Platelet Count 515 10^3/uL.

PT >maximum. Rechecked with fresh sample and APTT :> maximum. Rechecked with fresh SAMPLE. Fibrinogen: 450 mg/dL (normal). Coagulation Factor Assays: were within normal except Factor 10 Assay 2 %. Ultrasound Lower Limb: Findings are in favour of intra-muscular hematoma. Doppler USS showed normal deep venous system.

His father and elder brother gave history of recurrent epistaxis. His brother coagulation profile was done which showed prolonged PT 174.6 Secs and PTT 162.3 with deficient factor X (2%) as well on the other hand, his father had normal PT and PTT with FACTOR 10 ASSAY48%.

Differential diagnosis

Since both PT and APTT were prolonged in our patient, the activity of the clotting factors that are involved in the common (both extrinsic and intrinsic) pathway of the coagulation cascade were investigated. Except for Factor X, other factors all came to be normal. As FX is synthesized in the liver, so any liver disease may result in acquired FX deficiency, along with the other liver-produced factors including prothrombin, factor V (FV), Factor VII (FVII), and Factor IX (FIX). Vitamin K

Corresponding author:* **Mahmoud EL Hussein
Pediatric Department, Latifa Women and Children hospital, UAE

deficiency and warfarin use also result in decreased levels of FX, FVII, and FIX.

Acquired FX deficiency occurs in up to 5% of patients with amyloidosis due to adsorption into splenic amyloid fibrils. There have been reports of acquired FX deficiency with cancer, myeloma, infection, and use of sodium valproate. Acquired inhibitors to FX have been identified in burns, respiratory infections, and exposure to topical thrombin.

In our patient, Liver function test was within normal, no history of any drugs intake, no past history of significance. Furthermore, the positive family history proven that this an inherited deficiency.

Treatment

Patient received daily fresh frozen plasma 10ml/kg, the calf swelling was progressively improving; he was discharged in good condition after 5 days.

Outcome and follow up

Patient was planned for regular follow up in paediatric haematology clinic. An emergency card was given to the parents, describing his and his sibling condition. Advice was given to parents to avoid trauma, and to attend emergency to receive fresh frozen plasma if needed.

Parents were counselled appropriately.

The child was readmitted three times with shoulder swelling, or elbow swelling, with remarkable improvement after receiving fresh frozen plasma.

DISCUSSION

FX-deficient is an extremely rare inherited coagulation disorder. Reports have greatlyexpanded our knowledge about clinical phenotype (1).Peyvandi *et al*., 1998, classified FX deficiency severity based on its activity; FX: C to severe with FX: C measurement of <1%, moderate with measurement 1-5% and mild with FX: C measurement of 6-10% (4).

Factor X deficiency can manifest at any age with different presentations depending on the severity of the deficiency. It can present as early as during neonatal period with umbilical stump bleeding, bleeding during circumcision, intracranial haemorrhage or gastrointestinal bleeding especially in severe deficiency. On the other hand, moderately deficient FX may be diagnosedafter haemostatic challenge, such as surgery, trauma or menses. Moreover, Mild FX deficiency may be recognizedonly during routine screening or because of a positive family history.

Our presented patient and his affected sibling have 2% of normal factor X activity indicating moderate disease.

The diagnosis of FX is usually suspected when both the prothrombin time (PT) and activated partial thromboplastin time (APTT) are abnormal and correct with a 1:1 mix with normal plasma. FX functional activity (FX:C) is quantified by performing serial dilutions with FX-deficient plasma. PT reagents may vary in sensitivity to FX deficiency and congenital variants have been identified in which both the PT and PTT are normal (1).

Factor X levels are low at birth and should be compared with age- and gestational age-matched normal ranges before a deficiency is diagnosed in the neonate. FX levels in healthy full-term infants average 0.40 (SD, 0.14) IU mL)(1), and do not approximate adult values until after 6 months of age . Because FX is synthesized in the liver, liver disease will result in low levels of FX, along with the other liver-produced factors prothrombin, FV, FVII and FIX. Vitamin K deficiency and warfarin use also result in low levels of FX, FVII and FIX. Acquired FX deficiency occurs in up to 5% of patients with amyloidosis as a result of adsorption into amyloid fibrils in the spleen. There have been reports of acquired FX deficiency with cancer, myeloma, infection and use of sodium valproate (5).

Acquired inhibitors to FX have been identified in burns, respiratory infections and exposure to topical thrombin.

There is no specific FX replacement product yet readily available, but fresh frozen plasma and prothrombin complex concentrates can be used for treatment of bleeding symptoms and preparation for surgery.For minor bleeding symptoms, topical therapies and antifibrinolytic agents may be sufficient treatment.Aminocaproic acid is alsoreported to be effective in the treatment of idiopathicmenorrhagia and is used with generally good resultsin women with bleeding disorders(5).

Factor X is more common in communities with higher rate of consanguinity, thus it will be prudent to investigate patients with bleeding diathesis for Factor X deficiency that are not diagnosed as Factor VIII/IX deficiency.

References

1. Brown DL and Kouides PA. Diagnosis and treatment of inherited factor X deficiency. *Haemophilia* 2008; 14 (6): 1176-82.
2. Uprichard J, Perry DJ. Factor X deficiency. *Blood Rev*. 2002 Jun. 16(2):97-110
3. Barik S, Budhraja A, Bhalla M, Diwan S. Inherited Factor X deficiency in two brothers. *Indian J Pathol Microbial* 1997; 40:63-65.
4. Peyvandi, F., Mannucci, PM., Lak, M., *et al*. "Congenital Factor X Deficiency: Spectrum of Bleeding Symptoms in 32 Iranian Patients," *Brit J Haematol* 1998;102:626-8.
5. Dalal A, Pradhan M, Agarwal S. Genetics of Bleeding Disorders. *Int J Hum Genet* 2006; 6(1): 27-32.

How to cite this article:

Mahmoud EL Hussein *et al* (2017) 'Post Traumatic Intramuscular Hematoma Unveil A Rare Coagulation Disorder: Factor X Deficiency', *International Journal of Current Advanced Research,* 06(12), pp. 8302-8303.
DOI: http://dx.doi.org/10.24327/ijcar.2017.8303.1331

****