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Case Report

Case Report on Factor VII Deficiency: A Rare Coagulopathy

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ABSTRACT

Factor VII deficiency is a rare inherited bleeding disorder affecting 1/500,000 individuals with an autosomal recessive pattern of inheritance. The range of clinical manifestations is wide, ranging from no symptoms to severe bleeding that may be fatal. Deficiency of factor VII leads to a very diverse group of patients. It is uncommon for patients with a factor VII: C level of at least 10-15% of normal to report bleeding issues. Bleeding is frequently of mucocutaneous type, but the whole array of haemophilic bleeding may also occur. Bleeding can occur from factor VII deficiency, especially if factor VII is very low. However, there are some cases where factor VII function is completely or partially absent, and these patients may not have a history of bleeding. This case report presents a detailed account of a patient with factor 7 deficiency, a rare bleeding disorder. It includes the patient's clinical presentation, diagnostic workup, management, and follow-up. The aim is to highlight the challenges and considerations in diagnosing and treating this condition. We report the case of a 57-year-old man with severe deficiency of factor VII (FVII), probably genetic in nature, with no history of bleeding in infancy and young age.

Although there is no known family history of bleeding, our patient's uncle experiences recurring episodes of hemarthrosis and epistaxis.

Keywords: Factor VII deficiency; Vitamin K; Coagulation; Central nervous system

Introduction

Factor VII deficiency is a rare inherited bleeding disorder. The liver produces coagulation factorVII (FVII), a serine protease that is dependent on plasma vitamin K[1]. Factor VII is a unique coagulation factor that contains just a small fraction (1%–3%) of free circulating activated form(FVIIa). Plasma levels range around 0.35 to 0.60 mg/L (for a normal coagulant activity comprising between 70% and 140%). Factor VIIa by itself is not sufficient to initiate coagulation; it requires the presence of tissue factor (TF). After an injury, the arterial lumen isexposed to the integral membrane protein TF, which can bind to the free-circulating FVIIa to initiate coagulation. Stable fibrin clot formation is the outcome of a burst of activated factors IX(FIXa) and X (FXa) produced by the FVIIa-TF complex. Minimal levels of FVII:c is able to protect from bleeding in different clinical scenarios are also not well defined[2], ranging from 8%to 15%-20%. The catalytic domain, the gamma-carboxyglutamic acid domain, and the two epidermal growth factor (EGF)-like domains of FVIIa are all implicated in the interaction between TF and FVIIa. FVIIa concentrations must be very low to cause coagulation. A broad range of clinical manifestations, including gastrointestinal (GI) and central nervous system (CNS) bleeding, to severe life-threatening bleedings, even in homozygous people, are indicative of inherited FactorVII deficiency. We report a case of a 57 year old man with recurrent epistaxis.

Case Report

A 57 year old man presented to us with the history of recurrent nose bleeding for 1 year, on and off which stopped when he raised his head and applied pressure. The time between bleeding episodes was initially about 4 months and duration of bleeding was 2-5 minutes; the time between episodes was getting shorter and duration of bleeding increased, subsequently lasting for more than 30 minutes at the time of presentation at our emergency department. His medical history was unremarkable. He is also a known smoker with a history of 30 packs a year. Upon examination, the patient was not pale and the mucous membrane of the nose showed no sign of inflammation and no signs of trauma. On abdominal examination, there was no organomegaly. Ultrasound abdomen was done which was unremarkable. Hepatitis A and B serology were negative. Initial laboratory investigations revealed a very prolonged prothrombin-time (PT) > 120 second and normal activated partial thromboplastin time (aPTT) i.e 26.2 seconds. Factor assay from Agha khan university hospital was done which showed less than 6% of Factor VII activity.

The patient was admitted and vitamin K and FFPs were transfused. After confirmation of diagnosis, the patient was registered with the hemophilia society of Pakistan and transamin was given. Bleeding was then stopped. Patient was discharged on tablet folic acid once daily for 30 days and capsule transamin in caseof bleeding for 5 days. In case of uncontrolled bleeding, rFVIIa is recommended. He was educated on the signs of bleeding and the importance of seeking prompt medical attention ifsymptoms recurred. Regular follow-up visits were scheduled to monitor his condition and adjustment treatment as necessary.

Discussion

In 1951[3], published the first description of FVII deficiencies in the medical literature under the name prothrombin conversion accelerator deficiency. One in 500,000 peopleare thought to have factor VII deficiency, a rare autosomal recessive condition. Factor FVII (FVII), also known as “serum prothrombin conversion accelerator” or “proconvertin,” was discovered in 1951. It has been demonstrated that FVII interacts with chemicals derived from tissues to improve the conversion of prothrombin into thrombin. Congenital FVII deficiency as ableeding disorder is characterized by a prolonged prothrombin time (PT), which is corrected by the administration of plasma. Factor VII also known as proconvertin is a clotting factor of the human coagulation system. This free circulating glycoprotein belongs to the serine protease family. It is a Vitamin K dependent protein synthesized exclusively by the liver. FVII was shownto have a half life of 3-4 hours. Factor VII is the only clotting factor that has a small proportion (1%-3%) of free circulating activated form (FVIIa) in the absence of coagulation activation.

[4]The FVII gene is made up of nine exons that are preceded by a promoter region and is located on the long arm of chromosome 13. FVIII congenital deficiency is characterized by a substantialdecrease in the total FVII mass and, consequently, in very low levels of circulating FVIIa which is virtually absent when measured with classical assays. This results in a variety of bleeding symptoms of different severity because (i) the minimal FVII levels needed to avoid bleeding in different clinical situations are still unknown and (ii) elements that are still uncharacterized areinfluencing the clinical picture.

Functional testing is a key to the routine diagnosis of FVII deficiency. It is based on the results of routine assays (prothrombin time [PT] and activated partial thromboplastin time [aPTT]) and the specific measurement of FVII coagulant activity. Factor VII deficiency can be easily suspected when a haemostatic screening reveals an isolated prolongation of the PT with a normal activatedpartial thromboplastin time. Molecular diagnosis is based on the conventional PCR techniques. Several therapeutic options can be offered to patients regarding severe bleeding symptoms[5-8].

Recombinant FVIIa is the most widely accepted replacement therapy in patients with acute bleeding symptoms. Plasma-derived FVII concentrates may provide an alternative to rFVIIa. Fresh frozen plasma (FFP) is a cheap and easily available option, but its effectiveness is limiteddue to the high volumes that have to be administered. Fluid overload may be a serious complication of FFP transfusion. The antifibrinolytic tranexamic acid could be used incombination with replacement therapy or alone in minor bleeding symptoms.

Conclusion

Factor VII deficiency is a rare defect with heterogeneous laboratory findings, genetic defects and clinical manifestation. This case of Factor VII deficiency in a 57 year old male underscores the variability in the presentation of this rare disorder and the need for heightened clinical awarenessto ensure timely diagnosis and management. FVII plasma determination is required for a patient’s characterization, one should keep in mind that FVII plasma level does not correlate well with the hemorrhagic diathesis. This makes both prophylaxis and bleeding management difficultfor the clinician.

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