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FACTOR VII DEFICIENCY: A CASE REPORT

Paediatrics

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ABSTRACT

Factor VII deficiency is a rare inherited autosomal recessive bleeding disorder. It plays a role in initiating the clotting process by activating factor X, which ultimately leads to the formation of stable blood clot. It is caused by a genetic mutation that leads to a deficiency in the clotting factor VII which is essential for the formation of blood clot. Most cases remain asymptomatic and cases with severe clinical presentation are rarely reported.

KEYWORDS

Factor VII Deficiency, Coagulation Disorder, Intracranial Hemorrhage, Prothrombin Complex Deficiency, Rare Bleeding Disorder.

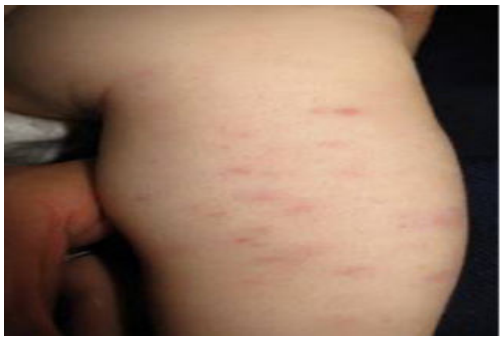
INTRODUCTION

Factor VII is synthesized in liver and found to be one of the vitamin K dependent coagulation factors. It has short circulating half-life of 2-4hour. Factor VII deficiency was first described by Alexander et al[1]. it is a rare autosomal bleeding disorder with prevalence of 1: 500,000 that is usually detected only in the homozygous state[2]. Factor VII gene is located on chromosome 13[3]. Factor VII deficiency may be inherited as autosomal recessive disorder or may be acquired. Two types have been described. Type 1 deficiency is due to decreased biosynthesis and type 2 deficiency is due to dysfunctional molecule[3]. Here, other factor assays in blood are found to be normal. Acquired factor VII deficiency arises due to vitamin K deficiency, liver diseases, vitamin K antagonist therapy. But in these cases there will be reduced levels of other vitamin k dependent factors. Acquired factor VII deficiency is more common than inherited deficiency.

Case Presentation

A 6 month 4 days old male child was brought to the hospital by his parents with complaints of fever since 2 days, refusal to feed since 2 days , rashes over left thigh since 2 days , bleeding from nose 2 episode since 1 day, difficulty in breathing since 1 day and one episode of abnormal jerky movement involving all 4 limbs with uprolling of eyes lasting for 2 mins. Baby was delivered at home by normal vaginal delivery, birth weight of 2.8kg, baby cried immediately after birth and no history of NICU admission. Baby was brought to our center with above mentioned complaints. The vitals at time of admission was PR: 126bpm, PV: good, RR: 30/min with mild subcostal retraction, SpO2: 97% on Npo2. Child was lethargic and on respiratory system examination, bilateral crackles were heard.

The patient was connected to CPAP, injection ceftriaxone was started in view of sepsis , infusion Paracetamol, asthalin nebulization, saline nasal drops and injection vitamin K was given. On anthropometrical examination child was found to have severe acute malnutrition. PT-22.1 sec and APTT- 28.7 sec and INR 2.1. factor 7 assay came to be 11%, so diagnosis of factor VII deficiency was made. Initially the patient was treated with FFP and recombinant factor VII was planned but they could not afford it.



Investigation:

Complete blood count: Hemoglobin: 8.8 g/dl, Total count: 24,000, platelet: one lakh.

KFT: urea: 11.08 mg/dl, creatinine: 0.27mg/dl
Serum electrolytes: sodium: 115.5mmol/l, potassium: 4.76mmol/l, chloride 93.61mmol/l.
CRP: <0.5mg/dl.
Prothrombin time: 22.1 sec, APTT- 28.7sec, INR: 2.1.
Liver function test: total protein: 5.51 gm/dl, albumin: 3.01, globulin: 2.50gm/dl.
Neonatal bilirubin unconjugated :0.20mg/dl, conjugated: 0.00, total: 0.20mg/dl
AST: 131 U/L
ALT: 58 U/L
ALP: 129 U/L.
FACTOR VII assay: 11%

Management:

The child was initially connected to CPAP, antibiotic injection ceftriaxone, infusion paracetamol asthalin nebulization and saline nasal drops were started. Injection vitamin k was given. 3 units FFP tranfusions were done. Child condition gradually improved and PEM supplements were started and recombinant factor VII was arranged, but the parents could not afford it and left against medical advice.

DISCUSSION:

Factor VII deficiency is a rare autosomal bleeding disorder that is usually detected only in the homozygous state. It is more prevalent in communities practicing consanguineous marriages[6]. Among the rare bleeding disorders, the prevalence of factor VII deficiency is described as the highest (35-40%). Severity of bleeding varies from mild to severe with hemarthroses, spontaneous intracranial hemorrhage and mucocutaneous bleeding, especially epistaxis (nosebleed) and menorrhagia. The bleeding severity usually does not correlate with the factor VII level. Patients with this deficiency have greatly prolonged PT but normal PTT. Factor VII assays show a marked reduction in factor VII. Because the plasma half – life of factor VII is 2 – 4 hour, therapy with FFP is difficult and is often complicated by fluid overload. A commercial concentrate of recombinant factor VIIa is effective in treating patients with factor VII deficiency[4,5].

Recombinant factor VII is produced through genetic engineering techniques. It is created by inserting the factor VII gene into host cells, which then produce the factor VII protein. This process allows for the production of a purified and consistent form of the clotting factor. Recombinant factor VIIa works by directly activating factor X, and promotes the formation of blood clots at the site of bleeding.

CONCLUSION:

Factor VII deficiency should be suspected in patients presenting with bleeding and in patients with deranged PT but normal aPTT and platelet count. Efforts should be made to make factor VII available

atleast in tertiary hospitals. Preventive measures such as avoiding certain medications and activities that increase the risk of bleeding, as well as regular monitoring and medical management recommended. Overall, it is a challenging disorder to manage, but with early diagnosis and timely intervention we can prevent the serious complications.

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