



### **Case Report:**

## **Combined Factor VII and X Deficiency**

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### **Abstract:**

Factor VII deficiency and factor X deficiency and very rare disorders individually. Combined Factor VII and X is a rare congenital blood disorder with very few cases reported in the literature. We report a case of 7 years old male child who presented to us as a diagnosed case of factor 7 deficiency with recurrent epistaxis.

**Key Words:** Combined factor VII and X deficiency

### **Introduction:**

Incidence of isolated factor 7 deficiency is 1: 5,00,000 children<sup>1</sup> and Homozygous factor X deficiency has an incidence of 1:1,000,000 in the general population.<sup>2</sup> Only 50 cases of factor X deficiency have been reported worldwide.<sup>3</sup> We report a case of 7 year old male child who was diagnosed as a case of combined factor VII and X deficiency.

### **Case Report:**

We present the case of a 7 years old male child, who came to us with epistaxis since 2 days. Bleeding started without any antecedent cause like nose picking or any trivial trauma. Child had persistent bleeding since past 12 hours with no relief from nose packing and ice packs. History of painless hematuria was present since one day.

Parents give history of fall from the stairs in the past about 3 years back when child was playing. Since then child is having recurrent episodes of bleeding from the nose on and off without any aggravating factor. Also there is history of multiple purpuras on the body and oral mucosa. There were episodes of blood in the stools and urine at times.

Patient walks with a limp since 1 month, no history of trauma is present. On examination hemarthrosis of the left hip joint with limitation of movement was present. Multiple ecchymotic and purpuric patches present all over the body. History of repeated admissions in the past is present for management of bleeding diathesis and fresh frozen plasma transfusions since 2 years of age.

Child was born by a non consanguineous marriage to a second gravida mother by normal vaginal delivery. At birth, there was purple patch on the ear and cheek on the left side, which resolved itself within 15 to 20 days of life. Elder sibling is a 9

year female child with no history of similar illness in her or any other family member.

Initially child was investigated at a tertiary hospital and labeled as congenital factor 7 deficiency

Investigations revealed Hb-6.5gm%, TLCaP-65%, L-32%, Platelet count- 3,70,000 cumm. Bleeding time was normal but clotting time was prolonged to 10 minutes 25seconds. Peripheral blood smear showed microcytic hypochromic picture. Renal function tests were normal. Patients Prothrombin time (PT) was 21 seconds (control -13 seconds), Activated Partial Thromboplastin time (APTT) was 52.70 seconds [(control- 34.89 secs), (Ref range- 28.65-41.1sec)]. Both these parameters were prolonged.

This prompted us to get factor VII, VIII and X levels. Factor VIII assay was normal 108% (normal 60-150%), activity of both factor VII and X were less than 1 percent of the normal. Factor II and IX activity were in the normal range.

Patient was diagnosed and managed conservatively by transfusing Fresh frozen plasma concentrates.

### **Discussion:**

Factor II, VII, IX and X are one of the vitamin K-dependent serine proteases. Congenital bleeding disorders of the vitamin K-dependent coagulation factors represent only about 15-20% of all congenital bleeding disorders.<sup>4</sup> Both factor VII and X deficiency individually have an autosomal recessive transmission.<sup>5</sup> The presence of more than one congenital clotting defect in a given patient is a rare event but not an exceptional one. Combined defects of factor X (FX) are very rare because congenital isolated FX deficiency is by itself very rare. This occurs because of abnormalities in chromosome 13 involving both FX and FVII genes. These genes are known to be very close and located on the long arm of chromosome 13,13q34. Isolated FX deficiency and, more frequently, combined FX + FVII deficiency appear also associated with coagulation-unrelated abnormalities (carotid body tumours, mitral valve prolapse, atrial septal defect, ventricular septal defect, thrombocytopenia absent radius (TAR) syndrome, mental retardation, microcephaly and cleft palate).<sup>6</sup> Diagnosis of a combined clotting defect could be difficult on the basis of global tests. As both isolated FX deficiency and combined FX + FVII deficiency yield a pro-

longation of basal PTT and PT. Only specific assays could allow one to reach the correct diagnosis.

In our patient the diagnosis was based initially on prolonged PT and depressed levels of Factor VII but abnormal aPTT values raised doubt regarding the diagnosis and levels other factors were subsequently done.

These deficiencies may be asymptomatic, only discovered on investigations. They may also be revealed by intracranial bleeding and other severe hemorrhages.<sup>8</sup>

Treatment consists of symptomatic management. Transfusion of Fresh frozen plasma (FFP) or Prothrombin complex concentrate (PCC ) during the episodes of bleeding diathesis. When administered at frequent intervals or over a prolonged period, PCCs may produce hypercoagulable complications, such as DIC, venous thromboembolism, stroke or myocardial infarction particularly in patients with liver dysfunction.<sup>9</sup> There are reports on the use of rFVIIa for congenital factor VII deficiency in a dose of 20µg/kg with FFP- two hourly till hemostasis is achieved.<sup>10</sup>

This case highlights the importance of keeping in mind the possibility of combined congenital clotting factor deficiency while dealing with a patient of hemophilias.

#### References:

1. Peyvandi F, Mannucci PM. Rare coagulation disorders. *Thromb Haemost* 1999;82(4):1207-1214.
2. Uprichard J, Perry DJ. Factor X deficiency. *Blood Rev* 2002;16(2):97-110.
3. Roberts HR, Escobar MA. Inherited disorders of prothrombin conversion, In: Colman RW, Marder VJ, Clowes AW, Geroge JN, Goldhaber SZ, eds. Hemostasis and thrombosis – Basic principles and clinical practice. 5th edition. Philadelphia: Lippincott Williams & Wilkins, 2006; 923-37.
4. Girolami A, Scandellari R, Scapin M, Vettore S. Congenital bleeding disorder of the vitamin K-dependent clotting factors. *Vitram Horm* 2008;78:281-374.
5. Mannucci PM, Duga S, Peyvandi F. Recessively inherited coagulation disorders. *Blood* 2004;104:1243-1252.
6. Girolami A, Ruzzon E, Tezza F, Scandellari R, Scapin M, Scarparo P Congenital FX deficiency combined with other clotting defects or with other abnormalities: a critical evaluation of the literature. *Haemophilia* 2008;14(2):323-328.
7. Boxus G, Slacmeudler M, Ninane J. Combined hereditary deficiency in factors VII and X revealed by a prolonged partial thromboplastin time. *Arch Pediatr* 1997;4(1):44-47.
8. Gupta PK, Kumar H, Kumar S. Hereditary Factor X (Stuart-Power Factor) Deficiency. *MJAFI* 2008;64:286-287.
9. Kinra P, Kumar H. Recombinant Factor VIIA. *MJAFI* 2009;65:59-61
10. Franchini M. Recombinant Factor VIIa: A Review on its Clinical use. *International Journal of Hematology*. 2006;83(2):126-138.