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Congenital Factor VII Deficiency Revealed in the Neonatal Period: A Case Report at CHNEAR

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Abstract

Congenital factor VII deficiency is a rare, autosomal recessive disorder with an estimated prevalence of 1 in 1,000,000. We report the case of a female newborn, born to second-degree consanguineous parents, admitted at 22 days of age for management of a subarachnoid hemorrhage. Hemostasis testing revealed a prothrombin time (PT) of 16%, with a preserved activated partial thromboplastin time (aPTT). Coagulation testing demonstrated an isolated factor VII deficiency. Through this case, we highlight the clinical, biological, and therapeutic characteristics of this anomaly during the neonatal period.

Keywords

Factor VII, Neonatal Hemorrhage, Congenital, Autosomal Recessive, Hemostasis

1. Introduction

Coagulation factor VII, also known as proconvertin, is a blood glycoprotein synthesized by the liver and dependent on vitamin K. It plays an essential role in initiating coagulation. Congenital factor VII deficiency is a very rare condition, with an estimated prevalence of approximately 1/500,000 to 1/1,000,000, resulting in a hemorrhagic syndrome with extremely variable clinical presentation [1]-[3]. Manifestations can range from moderate mucocutaneous or post-surgical bleeding to severe intracranial hemorrhages or hemarthrosis [4] [5]. Neonatal diagnosis is

rare but crucial, as the severity of bleeding can be life-threatening [6] [7].

We report here a case of factor VII deficiency revealed in the neonatal period, illustrating the clinical, biological, and diagnostic features of this rare condition [4] [8].

2. Clinical Observation

This is a female newborn, 22 days old at admission, born to second-degree consanguineous parents. The mother, 24 years old, blood type O Rh+, was in her third pregnancy with two previous unexplained neonatal deaths. The pregnancy was carried to term, and the vaginal delivery was uncomplicated (birth weight: 3100 g, Apgar score 8/10). Vitamin K was administered at birth and again upon admission to rule out any risk of late bleeding due to vitamin K deficiency before factor VII testing.

The infant experienced minor umbilical cord bleeding on the fifth day of life, followed by ear bleeding and epistaxis on the tenth day. At 22 days, refusal to feed and irritability prompted hospitalization. The initial lumbar puncture revealed hemorrhagic cerebrospinal fluid. An emergency ETF followed by a CT scan revealed a left temporoparietal hematoma with intraventricular hemorrhage complicated by communicating hydrocephalus.

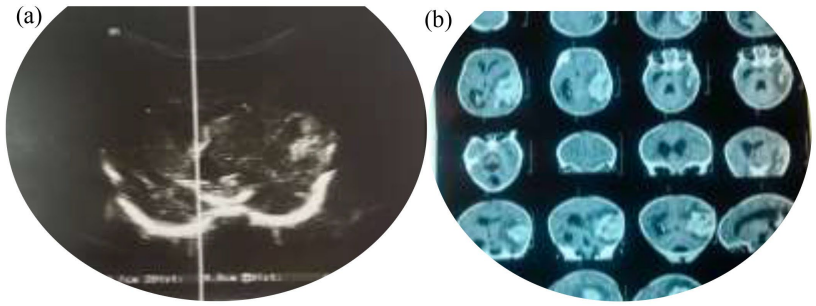

Figure 1. Neonatal brain ultrasound showing intracranial hemorrhage in the lateral ventricles.

Given this clinical presentation, a congenital hemostasis disorder was strongly suspected, leading to a coagulation workup which showed a prothrombin time (PT) of 16% and a normal activated partial thromboplastin time (aPTT). A hematological consultation was requested, and a transfer to a specialized facility was scheduled. Upon transfer to the CHNEAR (National Hospital Center of the North of the East) at 48 days, the examination revealed a pale, hypotonic newborn with a bulging fontanelle and persistent bleeding at the puncture sites. The complete blood count showed anemia with a hemoglobin level of 8.2 g/dL and thrombocytosis of 639,000/mm^3. Coagulation factor testing revealed a factor VII level < 1%, confirming a severe congenital deficiency. Infectious disease was diagnosed through blood culture, which isolated *Streptococcus* spp. Management consisted of transfusions of fresh frozen plasma and packed red blood cells, and administration of recombinant activated factor VII (rFVIIa, 90 µg/kg IV bolus every 8 hours). The

difficulty in our patient lay in managing episodes of intracranial hypertension and sepsis. Intraventricular hemorrhages necessitated paracentesis, which was managed with factor VII administration or fresh frozen plasma transfusions, despite the risk of bleeding. The multidrug-resistant *Streptococcus* spp. infection was maintaining this intracranial hypertension. After several lines of antibiotic therapy, the patient's condition improved with a fourth-generation cephalosporin (C4G). The episodes of intracranial hypertension resolved, and the neurological status stabilized, with no indication for surgery after CT scan.

Although the prognosis was lifted, after three months of hospitalization the infant presented with neurological sequelae (**Figure 1**). He is currently being followed by pediatric neurology and undergoing physical therapy. Vaccination began at six months of age and is supported by short-stay transfusions of fresh frozen plasma (FFP). Since his discharge, the patient has not experienced any further spontaneous external bleeding. He is followed by a pediatric hematologist every 6 to 8 weeks due to the unreliable availability of Factor 7. Genetic counseling was given to the couple for a future pregnancy (**Figure 2**).

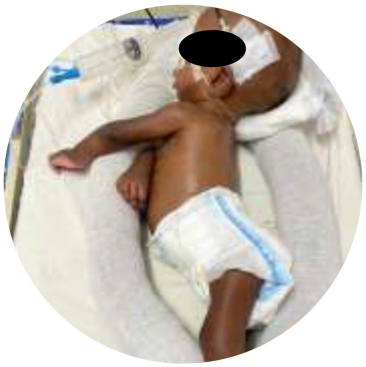

Figure 2. Newborn during hospitalization showing hydrocephalus, characterized by an increase in head circumference and bulging fontanelles.

3. Discussion

Congenital factor VII deficiency was first described in 1951 by Alexander [1]. Its prevalence is estimated at 1 in 1,000,000 and it is inherited in an autosomal recessive manner, which explains its frequency in consanguineous marriages [2] [3].

The gene responsible is located on chromosome 13 [3] [4]. Only homozygous or compound heterozygous patients present with a hemorrhagic syndrome, while simple heterozygotes generally remain asymptomatic [4] [5]. No clear link has been established between genotype or FVII:C levels and clinical severity [4] [5]. The family history, with two unexplained neonatal deaths, suggests a possible link to a hereditary coagulation disorder, such as the factor VII deficiency observed in our patient. This underscores the importance of a thorough family assessment to identify at-risk individuals and guide preventive and neonatal care.

The deficiency can manifest in the neonatal period with umbilical cord or in-

tracranial hemorrhages, or later with bleeding during trauma, surgery, or heavy menstrual bleeding [6] [7]. Central nervous system hemorrhages occur in approximately 4% of patients and are the leading cause of neonatal mortality [7] [8]. Three levels of severity are distinguished according to the factor VII level: <5%, 5% - 20%, and >20%, with severe bleeding generally associated with the <5% level [4] [8].

Diagnosis is based on hemostasis testing: prolonged PT, normal aPTT, and specific factor VII assay [4] [8] [9]. Family screening is essential to identify heterozygous carriers and plan monitoring of at-risk individuals [8]. In our case, an invasive procedure was performed before the family investigation, probably leading to the episodes of intracranial hemorrhage responsible for the sequelae in our patient.

Options include fresh frozen plasma (FFP), factor VII concentrates, and recombinant activated factor VII (rFVIIa). FFP can be effective but requires large volumes, while specific concentrates and rFVIIa allow for faster and more targeted correction [3] [5] [6] [10] [11]. Prophylactic treatments are indicated before surgical procedures and in cases of severe recurrent bleeding [3] [5] [6] [10] [11].

In our case, the severe deficiency (FVII < 1%) manifested in the neonatal period, associated with intracranial hemorrhages. The heterozygous parents were asymptomatic, confirming autosomal recessive inheritance. Treatment combined transfusions, appropriate antibiotic therapy, and rFVIIa administration, which controlled the bleeding episodes despite the limited availability of the latter [6] [9]-[14].

4. Conclusion

Congenital factor VII deficiency, a rare condition, can manifest in the neonatal period with severe hemorrhages, particularly intracranial hemorrhages, which can be life-threatening [1]-[14]. A low prothrombin time (PT) with a normal activated partial thromboplastin time (aPTT) should raise suspicion of this deficiency and warrant specific testing before any transfusion. Management, often complex due to the limited availability of recombinant activated factor VII, relies on a multidisciplinary approach to prevent hemorrhagic complications.

Ethics Statement

This study was approved by the CHNEAR Ethics Committee. Written informed consent was obtained from the parents for the publication of anonymized patient information.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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