MATHEWS

Case Report

Mathews Journal of Pediatrics

Factor 10 Deficiency: A Case Report

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ABSTRACT

Factor X deficiency is a rare coagulation disorder characterized by early onset, severity and spontaneous hemorrhagic manifestations. We report the case of a male neonate from a consanguineous marriage admitted with intense pallor following bleeding at the site of a blood sample taken as part of the preoperative work-up for active hydrocephalus due to Dandy Walker malformation. The work-up confirmed the presence of factor X deficiency.

Keywords: Hemorrhagic Syndromes, Intra-Ventricular Hemorrhage, Factor X Deficiency.

INTRODUCTION

Factor X deficiency is an extremely rare blood coagulation disorder, which can lead to a variety of complications depending on the severity of the disease [1]. It accounts for around 8% of all rare coagulation disorders, affecting both men and women, regardless of race or ethnic origin. This deficiency is associated with a more severe hemorrhagic tendency than that observed in other coagulation factor deficiencies, notably by the occurrence of intracranial hemorrhage in the absence of any prior trauma.

Our aim is to emphasize the value of factor assays in hemorrhagic syndromes, and to highlight the impact of genetic counseling. We report a case of factor X deficiency revealed in the neonatal period.

OBSERVATION

This was a male newborn, born to a 22-year-old primigravida primiparous mother, from a presumed pregnancy at 41 weeks' amenorrhea, delivered by caesarean section because of suspected hydrocephalus. The pregnancy was poorly monitored, with first-degree consanguinity, no similar cases in the family, and good adaptation to extrauterine life. Clinical examination at birth revealed a newborn with an isolated macrocrania measuring 46cm (+2DS), for which a cranial CT scan was performed, showing a Dandy-Walker malformation associated with major triventricular hydrocephalus, indicating surgical treatment. As part of the pre-operative work-up, a blood sample was taken, and during monitoring, significant bleeding was noted at the blood sampling site, associated with generalized pallor. The rest of the examination was unremarkable.

Vol No: 10, Issue: 01

Received Date: December 01, 2024

Published Date: January 02, 2025

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Citation: Tihamy S, et al. (2025). Evolution of Pediatric Feeding Disorders. Mathews J Pediatr. 10(1):37.

The initial management consisted of an emergency workup and transfusion of packed red blood cells at a rate of 20cc/kg/3hours, and administration of vitamin K at a dose of 2mg/kg. The workup revealed anemia at 9g/dlHt at 26. 8% white blood cells at 13700mm3without hyper or leukopenia, neutrophils at 9810mm3 with normal platelets at 261000mm3, the haemostasis work-up showed a low prothrombin rate at 8%, activated partial thromboplastin time at 131. 9 seconds, with a fibrinogen level of 3.07g/l, indicating a coagulation disorder involving the common pathway. Factor X deficiency was found in the coagulation factor assay, with a level of <1%, while the other coagulation factors were normal. The patient was transfused with human prothrombin complex.

In a family investigation, both parents were asymptomatic, and the factor X assay showed a decreased level of 60% (70-140) in the mother, and 55% in the father.

The picture was complicated by an increase in head circumference with the reappearance of pallor, a distended wooden abdomen associated with rectorrhagia, and a follow-up CT scan of the brain showing active intracranial bleeding with active tri-ventricular hydrocephalus and hemoperitoneum on abdominal ultrasound with Hb: 11. 5g/dl, ht: 35.5% WBC:5700mm3, PT: 29.5%, APTT 74.1sec.

The evolution was marked by a progressive deterioration despite the administration of human prothrombin complex, culminating in the death of the newborn following a fulminant hemorrhage. The parents were referred for genetic counselling.

DISCUSSION

Factor X (FX) is a vitamin K-dependent factor synthesized by the liver. It is a plasma glucoprotein involved in the formation of the prothrombinase complex. It is activated by the extrinsic and intrinsic coagulation pathways and enables conversion of prothrombin to thrombin.

FX deficiency is a rare hemorrhagic disorder, constituting one of the most serious coagulation anomalies of autosomal recessive inheritance, with an estimated prevalence of around 1 per 500,000 births [1]. The incidence is particularly high in populations where consanguineous marriages are frequent.

The gene responsible is located on chromosome 13 at 13q34-ter. It may be isolated or associated with a deficiency of other FVII factors or more complex FII, VII, IX and X factors [2].

The clinical manifestations of FX deficiency are varied. Heterozygous forms are generally asymptomatic, often diagnosed on the basis of family history or abnormal preoperative laboratory tests [3].

Homozygous forms, on the other hand, are prone to life-threatening spontaneous hemorrhages, notably intracranial hemorrhages (9% to 26% of patients according to various studies [4,5], which often occur within the first month of life. Severe hemorrhages can include umbilical cord bleeding, spontaneous intra-abdominal hemorrhage, gastrointestinal hemorrhage, pulmonary hemorrhage, and post-vaccination hemarthrosis and intramuscular hematoma.

The diagnosis of FX deficiency is suspected by a low prothrombin level and prolonged activated partial thromboplastin time, and confirmed by a low factor X level.

FX deficiency is classified according to the residual proportion of the factor:

- Mild deficiency (>40%),
- Moderate deficiency between 10-40%
- Severe deficiency (<10%).

In our case it was a severe deficiency with factor 10 <10%

Initial treatment of hemorrhage associated with congenital FX deficiency is based on transfusion of fresh frozen plasma (FFP) at a dose of 15-20 ml/kg, used in our case on 2 occasions.

Another therapeutic option is the use of prothrombin complex concentrates, containing FX and four other vitamin K-dependent factors, although this treatment is associated with a risk of thromboembolic complications due to the high concentrations of FII, FVII, and FIX [6]. The dosage and duration of treatment vary according to the severity of the deficiency, the location of the bleed, the type of treatment (preventive or curative), and the patient's clinical and biological condition. In general, a loading dose of 15-25 ml/kg is administered intravenously. Because of the relatively long half-life of factor X, which is 20 to 40 hours and varies according to the individual and the dose administered, this loading dose may be followed by maintenance doses of 3-6 ml/kg intravenously every 12 to 24 hours, enabling factor X to be maintained at a level of 10% to 20% [7].

Vitamin K is ineffective in correcting this constitutional factor X deficiency.

Each case must be discussed by a multidisciplinary team, depending on the risk of bleeding, the severity of bleeding and the factor X concentration.

Fresh frozen plasma (FFP) transfusion and FX replacement therapy have reduced mortality and prevented major sequelae in these patients. Prognosis depends on the etiology and severity of the disease; patients with very low levels of factor X are more prone to severe bleeding.

CONCLUSION

Factor X deficiency, although rare, is one of the most serious coagulation anomalies, requiring rigorous, individualized management. Transfusion of fresh frozen plasma and administration of human prothrombin complex have reduced mortality and prevented major sequelae in these patients. However, the prognosis depends on the severity of the disease; patients with very low levels of factor X are particularly at risk of severe bleeding. Multidisciplinary management tailored to each individual case remains essential to improve the quality of life and clinical outcome of patients with this deficiency. Genetic counseling plays a crucial role in prevention and early detection.

ACKNOWLEDGMENTS

None.

CONFLICTS OF INTEREST

Authors declare that there are no conflicts of interest.

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