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Case Report

Congenital Factor X Deficiency: Diagnosis of a Rare Case in African Pediatric Practice

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Abstract

Introduction: Congenital factor X deficiency is a rare inherited bleeding disorder with autosomal recessive transmission, affecting fewer than one in 500,000 individuals. It affects both sexes equally and is more frequently encountered in populations with a high rate of consanguinity. We report the case of a male neonate hospitalized at the Children's Hospital of Diamniadio (Dakar).

Case Report: A male newborn born to a 20-year-old primigravida and primiparous mother with no notable medical history, and with first-degree parental consanguinity. The neonate was admitted on day 1 of life for a clinical hemorrhagic syndrome associated with severe biological hemostatic abnormalities. Investigations revealed an isolated deficiency of factor X activity (Stuart factor). The clinical course was complicated by severe bleeding events related to the unavailability of specific factor replacement therapy in our setting. The infant was therefore managed with a transfusion-based protocol, which helped limit these complications. Due to limited resources, genetic testing to identify the underlying etiology could not be performed, although a hereditary cause remains highly probable given the close parental consanguinity.

Conclusion: Congenital factor X deficiency is a rare disorder with non-specific clinical manifestations, often leading to diagnostic errors. Etiological diagnosis remains challenging in low-resource settings due to limited access to specialized laboratory and genetic investigations.

Keywords: Inherited Disorder; Factor X; Coagulation; Child

Introduction

Factor X, also known as Stuart factor, is a vitamin K–dependent coagulation factor. It is activated through both the extrinsic and intrinsic pathways of coagulation [1]. The gene encoding factor X is located on chromosome 13. Activated factor X (Xa) converts prothrombin into thrombin in the presence of calcium, phospholipids, and its cofactor factor V (Va) [2].

Congenital factor X deficiency, first described in 1956 by Telfer and colleagues and subsequently in 1957 by Hougie, is an extremely rare inherited bleeding disorder. This autosomal recessive condition affects fewer than one in 500,000 individuals and occurs more frequently in populations with a high prevalence of consanguinity. To date, only about sixty cases have been reported in the medical literature worldwide [3-5].

The deficiency is severe in homozygous individuals, whereas heterozygous carriers are generally asymptomatic. Clinically, the diagnosis is suggested by the presence of a hemorrhagic syndrome, while confirmation relies on the measurement of factor X activity. Other coagulation abnormalities may be observed and can provide diagnostic orientation, notably prolonged prothrombin time and elevated international normalized ratio (INR).

In sub-Saharan Africa, epidemiological data on congenital coagulation disorders such as factor X deficiency are scarce. The aim of this report is to describe a case observed at the National University Children's Hospital of Diamniadio (Dakar).

Case Report

Clinical presentation

A male newborn was born to a 20-year-old mother, primigravida and primipara, with no significant past medical history. There was a first-degree consanguinity between the parents. The pregnancy was well monitored, and delivery occurred at 39 weeks + 3 days of gestation. The newborn received vitamin K1 and routine essential neonatal care in the delivery room.

He was admitted on day 1 of life for umbilical bleeding. Clinical examination revealed:

- Hemorrhagic syndrome**,** including diffuse scalp hematomas, hematemesis, umbilical bleeding, and bleeding at puncture sites
- Non-hemolytic anemic syndrome, characterized by mucocutaneous pallor, tachycardia, and a functional cardiac murmur
- Neurological status: Clear consciousness (Blantyre score 5/5), slightly decreased neurological responsiveness, and a normal anterior fontanelle.

Paraclinical investigations

Laboratory findings

- Complete blood count showed normocytic normochromic anemia with hemoglobin level of 9.4 g/dL and a normal platelet count
- C-reactive protein (CRP): Elevated at 16 mg/L
- Fibrinogen level: normal
- Factor VIII and IX activities: Within normal ranges
- Coagulation profile:
- Prothrombin rate: 7.1%
- International Normalized Ratio (INR): 7.25
- Activated partial thromboplastin time (aPTT): 78.1 seconds (control: 29 seconds)
- Factor X activity assay: markedly reduced, <5%

Imaging

Transfontanellar ultrasound: Normal on day 1 of life.

Management and outcome

Emergency treatment consisted of fresh frozen plasma (FFP) administration at a dose of 20 mL/kg every 12 hours for 3 days, due to the unavailability of specific factor X concentrate.

Follow-up and evolution

At 45 days of life, the patient was readmitted for oral bleeding. Clinical examination revealed:

- Severe hemorrhagic syndrome with profuse gingival bleeding and diffuse scalp hematoma associated with an increase in head circumference
- Altered level of consciousness (Glasgow Coma Scale: 10/15)
- Focal tonic-clonic seizures involving the left side
- Severe anemic syndrome

Laboratory investigations showed:

- Severe anemia with hemoglobin level of 4.4 g/dL
- Coagulation tests revealing incoagulable prothrombin time and incoagulable aPTT

Imaging findings

- Cranial ultrasound: Enlargement of subarachnoid spaces with evidence of subarachnoid hemorrhage
- Brain computed tomography (CT scan): Findings illustrated in Figures 1, 2, and 3.

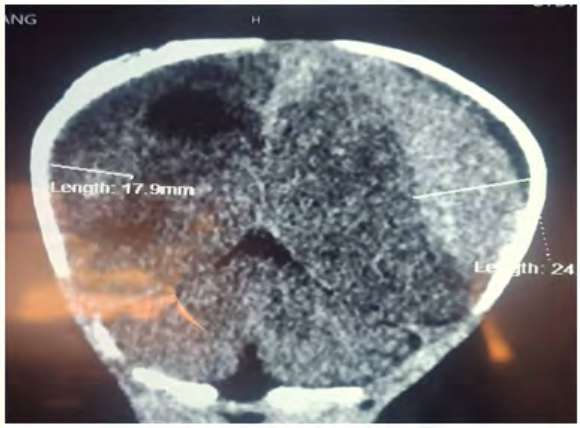

Figure 1: Bilateral hemispheric subdural hematomas of different ages. Right parieto-occipital porencephalic cavity as a sequela.

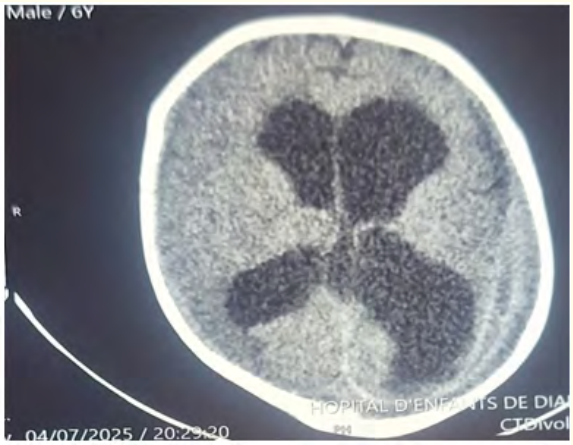

Figure 2: Extradural hematoma with associated triventricular hydrocephalus.

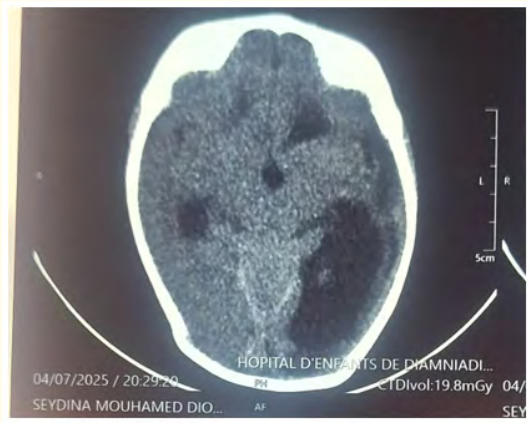

Figure 3: Chronic subdural hematoma.

Emergency management

Emergency treatment included:

- Resuscitative measures
- Fresh frozen plasma (FFP) transfusion protocol: 20 mL/kg every 12 hours for 3 days
- Corticotherapy with intravenous dexamethasone at a dose of 0.3 mg/kg every 6 hours for 3 days
- Packed red blood cell transfusions at 15 mL/kg, administered twice, 24 hours apart

Outcome

The patient's clinical course was favorable, with achievement of hemodynamic stability and recovery of clear consciousness. However, at 8 months of age, a mild psychomotor developmental delay was noted, while somatic growth remained appropriate for age.

Follow-up

The patient has since been maintained on a regular follow-up protocol consisting of:

- Weekly fresh frozen plasma (FFP) transfusions at a dose of 20 mL/kg
- Weekly monitoring of coagulation parameters (Table I)
- Biweekly complete blood counts
- Monthly screening for:
- Irregular (allo-) antibodies

- Hepatitis B virus infection (HBsAg)
- Human immunodeficiency virus (HIV).

Age (days)	PT (%)	INR	aPTT		Ratio
			Patient	Control	
1	7.1	7.25	78.1	29	2.6
4	14.3	3.63	58.4	24	2.40
8	7.9	6.55	101.6	28	3.56
45	8.0	6.3	90	29	3.1
60	68.4	1.32	38.5	30	1.2
90	<5	>10	>120	29	—
240	70	1.2	35	28	1.25

Table I: Evolution of the patient's coagulation profile during follow-up.

PT: Prothrombin Time; INR: International Normalized Ratio; aPTT: Activated Partial Thromboplastin Time; Dashes (—) indicate values not calculable due to incoagulable results.

Discussion

Congenital factor X deficiency is an extremely rare genetic disorder and is considered the rarest among congenital coagulation factor deficiencies. It is an autosomal recessive condition requiring both parents to carry the defective gene. Transmission affects males and females equally, and the disorder is more prevalent in populations where consanguineous marriages are common.

Several genetic abnormalities have been described. These result from mutations affecting either the phospholipid-binding domain due to incomplete γ-carboxylation, the catalytic domain, or the signal peptide cleavage site, leading to impaired secretion of the molecule. Partial deletion of exons 7 and 8, which encode the catalytic domain of factor X, is associated with severe deficiency. In our case, genetic testing would have helped identify the underlying mutation; however, this investigation could not be performed due to limited resources, despite the presence of first-degree parental consanguinity.

Three clinical forms of factor X deficiency are classically described:

- Severe deficiency, with factor X activity <1%;
- Moderate deficiency, with activity between 1% and 5%;
- Mild deficiency, with activity >5%.

In our patient, the activity level was reported as <5%, without precise quantification, making a severe form highly probable. Hemorrhagic manifestations represent the main clinical presentation, and their severity correlates closely with factor X activity levels. In our case, hematemesis was the primary reason for emergency consultation.

Other clinical manifestations, such as hematomas—particularly diffuse scalp hematomas, as observed in our patient—have been reported in the literature and may mimic hemophilia. This resemblance led us, especially given the male sex of the patient, to initially investigate hemophilia A and B. However, factor VIII and IX activity levels were normal.

Intracranial hemorrhage remains a rare but life-threatening complication of congenital factor X deficiency. Our patient presented with multiple hematomas of varying ages, later complicated by hydrocephalus, although no frank intracranial hemorrhage was initially detected.

Biological diagnosis is suggested by a markedly prolonged prothrombin time (PT), prolonged activated partial thromboplastin time (aPTT), and prolonged Quick time, with normal platelet counts. Definitive diagnosis relies on coagulation factor assays demonstrating an isolated reduction in factor X activity, while immunological assays are often normal. In our case, the initial

biological profile (low PT, prolonged aPTT, normal platelet count) led us to suspect hemorrhagic disease of the newborn, a common condition related to vitamin K deficiency or ineffective prophylaxis at birth. Following partial correction of PT and aPTT, extended coagulation factor assays were performed, revealing an isolated decrease in factor X activity.

Treatment is based on replacement therapy using prothrombin complex concentrates (PCCs) when available, or fresh frozen plasma (FFP) as an alternative. Dosage and duration depend on clinical severity and biological parameters. In cases of severe bleeding or surgical procedures, treatment aims to maintain factor X activity at or above 25%. In our context, the patient received FFP transfusions for 72 hours, resulting in clinical improvement, followed by a weekly FFP transfusion protocol during follow-up.

Conclusion

Congenital factor X deficiency is a rare disorder with non-specific clinical manifestations, which may lead to diagnostic errors. A thorough clinical history, particularly exploring familial and consanguinity backgrounds, is essential to prompt investigation of coagulation factor abnormalities. Early identification of the specific factor deficiency is crucial to guide appropriate replacement therapy, especially in resource-limited settings where access to specific concentrates remains a challenge.

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