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SPONTANEOUS INTRAVENTRICULAR HEMORRHAGE IN A YOUNG MALE WITH COMBINED PROTHROMBIN COMPLEX AND FACTOR X DEFICIENCY: A RARE CASE REPORT

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ABSTRACT Combined deficiencies of coagulation factors II (prothrombin) and X are exceedingly rare autosomal recessive disorders, usually presenting with mild mucocutaneous bleeding. However, life-threatening hemorrhagic events, including intracranial hemorrhage (ICH), may occur in severely deficient individuals. Isolated spontaneous intraventricular hemorrhage (IVH) is a particularly rare manifestation. We report the case of a 24-year-old male with congenital combined prothrombin and factor X deficiency who presented with acute headache and was diagnosed with isolated IVH. Laboratory studies revealed markedly prolonged PT and APTT, corrected with mixing studies. Specific assays confirmed severely reduced factor X and moderately reduced factor II levels. The patient was successfully managed with fresh frozen plasma (FFP), antiepileptics, osmotherapy, and close neurocritical care monitoring. This case underscores the need for heightened clinical vigilance and early neuroimaging in patients with known bleeding disorders presenting with neurological symptoms, and highlights the challenges of managing such rare disorders in resource-limited settings.

KEYWORDS : Intraventricular hemorrhage, Factor X deficiency, Prothrombin deficiency, Rare bleeding disorder, Neurocritical care

INTRODUCTION

Congenital bleeding disorders involving the common coagulation pathway are exceedingly rare. Factor X deficiency has an estimated prevalence of 1:500,000 to 1:1,000,000, while prothrombin deficiency is even rarer. Combined deficiencies of both factors are typically inherited in an autosomal recessive pattern and are often associated with a spectrum of bleeding symptoms, ranging from epistaxis and ecchymoses to life-threatening events like intracranial hemorrhage (ICH). Among the various forms of ICH, spontaneous isolated intraventricular hemorrhage (IVH) is exceptionally rare in adult patients with inherited bleeding disorders. We present a case of a young adult male with known combined factor II and X deficiency who developed spontaneous isolated IVH. The patient was managed conservatively with excellent neurological outcome, despite the absence of specific factor concentrates. This case emphasizes the diagnostic and therapeutic challenges of rare bleeding disorders, particularly in low-resource settings.

Case Report

A 24-year-old male presented to the emergency department with a 2-day history of severe holocranial headache of sudden onset, associated with nausea and photophobia. There was no history of vomiting, seizures, trauma, or loss of consciousness. He had no prior history of hypertension or diabetes. The patient had a known diagnosis of combined factor II and X deficiency, confirmed in early childhood. His previous bleeding episodes included recurrent epistaxis and spontaneous bruising, but no prior central nervous system (CNS) bleeds. He had received FFP in the past for minor procedures. On examination, he was conscious and oriented (GCS 15/15), afebrile, and hemodynamically stable. There were no signs of meningismus or focal neurological deficits. Fundus examination showed no papilledema.

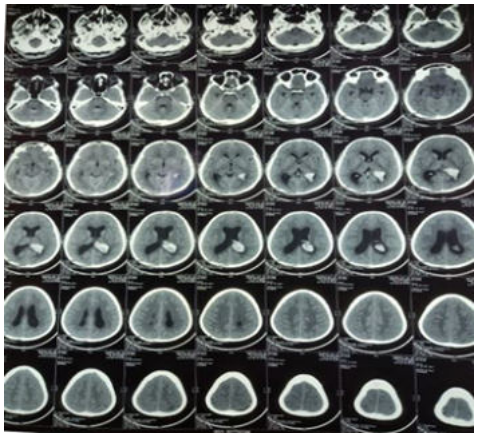

Figure 1- Showing Intraventricular Hemorrhage Localized To Left Lateral Ventricle

Routine blood tests revealed hemoglobin 13.7 g/dL, total leukocyte count 14,800/mm³, and platelet count 2.85 lakhs/mm³. Coagulation studies revealed a PT of 18.6 seconds (control: 13 sec), INR of 1.73, and an APTT greater than 170 seconds. Mixing studies with normal plasma corrected both PT and APTT, confirming a factor deficiency rather than an inhibitor. Specific factor assays revealed a factor X level <1% and factor II level at 15%. Neuroimaging with non-contrast CT brain revealed an isolated intraventricular hemorrhage localized to the left lateral ventricle, without associated hydrocephalus or intraparenchymal extension as shown in Figure 1.

Management: The patient was admitted to the intensive care unit. Hemostatic therapy was initiated with fresh frozen plasma (FFP) at 15 mL/kg every 12 hours for 3 days, then once daily for 2 days (total: 22 units), targeting a factor X activity >20%. Osmotherapy with 20% mannitol (100 mL IV every 8 hours) was given for 72 hours. Seizure prophylaxis with intravenous levetiracetam 500 mg twice daily was initiated. Oral nimodipine (30 mg every 6 hours) was administered to prevent vasospasm. Over the next 5 days, the patient remained neurologically stable with no deterioration. Repeat CT on day 5 showed resolving hemorrhage. He was discharged on day 10 with no neurological deficits and was advised regular follow-up in the hematology clinic. At 6-month review, he remained clinically well with no recurrence.

DISCUSSION

Factor X (FX) and prothrombin (factor II) are vitamin K–dependent clotting proteins synthesized in the liver and crucial for the conversion of prothrombin to thrombin, a pivotal step in fibrin clot formation. Deficiency of FX is classified as severe (<1%), moderate (1–5%), or mild (>5%) based on factor levels. Patients with levels <1% are at increased risk of spontaneous CNS hemorrhage, hemarthrosis, and gastrointestinal bleeding. Combined deficiency of FX and FII is extremely rare and may arise from contiguous gene deletions or mutations affecting shared transcriptional regulation on chromosome 13q34. The clinical presentation depends on the severity of deficiency and the availability of therapeutic options.

Intraventricular hemorrhage (IVH) in adults is most commonly associated with trauma, aneurysm rupture, arteriovenous malformations, or hypertension. Spontaneous isolated IVH in a young adult with a bleeding disorder is exceedingly rare and must prompt a high index of suspicion in predisposed individuals. The diagnosis of factor deficiency is confirmed by prolonged PT and/or APTT, with correction in mixing studies. Specific factor assays are essential but may be unavailable in low-resource settings. Neuroimaging is critical for diagnosing and monitoring ICH. CT brain remains the investigation of choice for acute bleeds.

Management includes factor replacement to maintain hemostasis. While prothrombin complex concentrates (PCCs) and specific factor concentrates are ideal, they are often unavailable in resource-

constrained settings. FFP remains a practical and effective alternative. Recombinant factor X (rFX) is available in some high-income countries but remains inaccessible in many parts of the world. Our patient responded well to FFP-based management. The absence of neurological deterioration and resolution of bleed without surgical intervention highlights the value of timely diagnosis, appropriate supportive care, and interdisciplinary collaboration in such rare presentations.

CONCLUSION

Spontaneous IVH is a rare but serious manifestation of congenital combined factor II and X deficiency. Early recognition, neuroimaging, correction of coagulopathy with available hemostatic agents, and supportive care are critical in ensuring favorable outcomes. This case underscores the need for increased awareness of rare coagulation disorders and improved access to diagnostic and therapeutic resources in developing countries.

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