The 5 th Biopharm Scientific Annual Meeting
BSAM5, Alger le 28 juin 2025

Case Study of Hemophilia C

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Abstract

Hemophilia C (Rosenthal syndrome) is a rare bleeding disorder caused by factor XI deficiency, often diagnosed after trauma or surgery due to variable bleeding severity. This case involves a woman diagnosed after prolonged aPTT was found during preoperative testing, with management involving fresh frozen plasma before surgery.

Key-words :*Hemophilia, Factor XI, deficiency, bleeding.*

1-INTRODUCTION: Hemophilia C, also known as Rosenthal syndrome, is a rare congenital disorder first described in 1953. It is characterized by a deficiency in factor XI and is inherited in an autosomal recessive manner with variable penetrance. The condition is usually diagnosed in adulthood and differs from classic hemophilia by the rarity of spontaneous bleeding, which typically occurs only after trauma or surgery. Clinical manifestations vary significantly from person to other, and even in the same individual, ranging from minimal bleeding to severe hemorrhages, particularly after trauma or surgical interventions. The diagnosis is based on specific laboratory tests, including plasma factor XI activity levels and genetic testing. Treatment primarily focuses on preventive measures, such as avoiding contact sports, intramuscular injections, and drugs that interfere with hemostasis. Factor XI concentrates or fresh frozen plasma may be administered in case of bleeding, and prophylactic approaches may be necessary in high-risk situations.

2-MATERIALS AND METHODS: We report a case of a patient with prolonged aPTT, whose etiological workup revealed a factor XI deficiency.

3-OBSERVATION: A 35-year-old woman, mother of three delivered by cesarean section and with a history of severe postpartum hemorrhage treated by hemostatic hysterectomy, was referred for evaluation after preoperative tests for hernia repair revealed a markedly prolonged activated partial thromboplastin time (aPTT) of 113 seconds (control 29 seconds). Further laboratory investigations, including coagulation factor assays, confirmed a severe deficiency of factor XI at 2.7%, leading to the diagnosis of Rosenthal syndrome. The patient had no other clinical signs of bleeding, and her prothrombin time, fibrinogen level, and platelet count were normal. Given her stable condition, no immediate treatment was administered; however, a perioperative plan was established to correct her coagulation defect with fresh frozen plasma transfusion prior to surgery, allowing for a safe operative procedure and uncomplicated postoperative recovery.

4-CONCLUSION: Factor XI deficiency is an uncommon hereditary bleeding disorder that must be included in the differential diagnosis of patients presenting with unexplained prolonged bleeding times and coagulation abnormalities. Optimal management of this condition requires a multidisciplinary approach involving hematologists, surgeons, and anesthesiologists to tailor individualized care plans. Preventive strategies, including perioperative hemostatic support and avoidance of trauma or medications that may exacerbate bleeding, are critical to minimizing hemorrhagic complications in at-risk patients. Early identification and appropriate prophylactic interventions are essential to improve clinical outcomes and ensure patient safety during invasive procedures