MILD HEMOPHILIA A CARRIER WITH CONCOMITANT STORAGE POOL DISEASE: DIAGNOSTIC CHALLENGES IN A SYMPTOMATIC FEMALE PATIENT

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Background. Women who are carriers of hemophilia A may present with significant bleeding symptoms even in the presence of mildly reduced factor VIII (FVIII) levels. In these cases, additional inherited bleeding disorders should be considered, particularly when the hemorrhagic phenotype appears disproportionate to coagulation factor levels.

Case Report. A 71-year-old woman (born in 1954), obligate carrier of mild hemophilia A was evaluated for a lifelong bleeding tendency. Baseline FVIII activity was 44% by one-stage assay and 36% by chromogenic assay. Family history was significant for mild hemophilia A affecting her father, son and nephew. The patient showed a clinically relevant hemorrhagic phenotype with an ISTH-BAT score of 13, including epistaxis, bleeding from minor wounds, severe menorrhagia, excessive bleeding after dental extractions, and post-tonsillectomy hemorrhage. She also experienced major bleeding complications after cesarean section, with development of a large surgical-site hematoma associated with severe anemia requiring blood transfusions. Although FVIII levels were compatible with mild hemophilia, the severity of bleeding manifestations appeared disproportionate to the degree of factor deficiency. Platelet function analyzer (PFA) testing showed prolonged closure times with ADP and epinephrine cartridges (132 and 185 seconds, respectively), prompting further investigation with platelet lumiaggregometry. Extended platelet studies demonstrated an associated storage pool disease, consistent with a combined inherited hemostatic disorder. A desmopressin (DDAVP) challenge test (0.3 μg/kg) showed an adequate laboratory response. In 2012, the patient successfully underwent adnexectomy after prophylactic desmopressin (Emosint®), without hemorrhagic complications. In 2023, the patient underwent appendectomy at another hospital where perioperative management was not coordinated with the specialized hemophilia center and recommended prophylactic measures were not implemented. The postoperative course was complicated by a massive 24-cm subhepatic hematoma with slow radiological resolution, further confirming the patient's high hemorrhagic risk. The patient was subsequently treated with fresh frozen plasma (FFP).

Conclusions. This case highlights the diagnostic complexity of symptomatic hemophilia A carriers and the importance of investigating associated platelet function disorders when the bleeding phenotype is not fully explained by FVIII levels alone. The coexistence of mild hemophilia A and storage pool disease may markedly increase hemorrhagic risk and influence perioperative management. The case also underlines the crucial importance of communication with specialized hemophilia centers to ensure appropriate perioperative prophylaxis and prevent severe bleeding complications.