



RECURRENT CENTRAL RETINAL VEIN THROMBOSIS IN A PATIENT WITH MILD HEMOPHILIA A AND HETEROZYGOSITY FOR THE PROTHROMBIN G20210A VARIANT

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Background. Hemophilia A is an X-linked inherited bleeding disorder caused by a deficiency of coagulation factor VIII (FVIII), leading to impaired hemostasis. It is classified as severe (<1% FVIII activity), moderate (1-5%), or mild (>5-40%). In mild forms, bleeding is often limited to trauma or invasive procedures, resulting in delayed diagnosis. Treatment consists of FVIII replacement therapy, administered on demand or prophylactically in severe cases. Despite its hemorrhagic nature, rare thrombotic events have been described in hemophilia A, particularly in the presence of additional risk factors or hemostatic treatments, suggesting a complex balance between bleeding and thrombosis.

Case Report. We report the case of a 62-year-old man with mild hemophilia A and a history of significant post-traumatic and post-surgical bleeding. He had a previously treated and eradicated hepatitis C virus infection. The patient was managed with on-demand recombinant FVIII (Beriate). At the time of the first event (2022), he was not receiving FVIII therapy. He presented to the Emergency Department with acute visual loss in the left eye. Laboratory testing excluded acquired thrombophilia; however, genetic analysis revealed heterozygosity for the prothrombin G20210A variant. Fluorescein angiography showed retinal hemorrhages with hypofluorescent areas consistent with central retinal vein occlusion (CRVO). Due to the bleeding risk, anticoagulation

was not initiated. The patient was treated with intravitreal anti-VEGF injections, achieving complete visual recovery. In March 2024, he returned with blurred vision in the same eye. Fluorescein angiography confirmed a recurrent CRVO, similar to the previous episode. He again received intravitreal anti-VEGF therapy and low-dose antiplatelet treatment. Further evaluation showed mildly elevated LDL cholesterol (66 mg/dL; target <55 mg/dL), and ezetimibe therapy was started. Thrombophilia screening revealed Protein S 93%, Antithrombin 89%, APC resistance ratio 0.95, Factor V 97%, and FVIII 13.7%. Carotid Doppler ultrasound showed normal findings, and coronary CT angiography excluded significant coronary artery disease.

Conclusions. This case describes recurrent central retinal vein thrombosis in a patient with mild hemophilia A, occurring in the absence of replacement therapy. The coexistence of a congenital bleeding disorder and a prothrombotic genetic variant (prothrombin G20210A) highlights the complexity of hemostatic balance. Although hemophilia is primarily a bleeding disorder, thrombotic events may occur when additional risk factors are present. Improved life expectancy in hemophilia patients has led to an increasing overlap with age-related cardiovascular risk profiles. The traditional view that hypocoagulability protects against thrombosis is no longer universally valid, emphasizing the need for individualized risk assessment