Factor V Leiden and its Association with Vascular Disease and Treatment in a Latino patient

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INTRODUCTION

Factor V Leiden (FVL) is the most common hereditary thrombophilia, and a single amino acid mutation renders Factor V resistant to inactivation by Activated Protein C resulting in a prothrombotic state. The association between FVL and vascular disease has been reported and debated. We present a case of a patient with FVL and its repercussion on medical treatment.

CASE DESCRIPTION

A 64-year old Hispanic man with a past medical history of coronary artery bypass grafting, Type 2 diabetes mellitus, hypertension and severe peripheral vascular disease with history of left above the knee amputation presented to the emergency department with severe, sharp right upper back pain that radiated to his chest for one day. It was not associated with fever, shortness of breath, or palpitations. He reported taking apixaban for the last three years however he was unsure of the indication. He denied smoking, prolonged inactivity or travel.

Vital signs included a temperature of 97.7^{o} F, heart rate of 97 bpm, respiratory rate of 17 breaths/min, blood pressure of 140/86 mmHg, and BMI of 28. Physical exam was remarkable for right paraspinal tenderness of the upper back upon palpation. Coagulation profile showed PT 15.8, INR 1.33, PTT 37.0. Patient was at moderate risk for a pulmonary embolism (PE) using the Wells' Score. A PE was noted on CT angiography and bilateral non-occlusive proximal deep vein thromboses (DVTs) of the right deep femoral vein, the left common and superficial femoral veins were found on venous doppler. The cardiac echocardiography showed no evidence of right heart strain and troponins were negative. He was initially started on a heparin drip and two liters of oxygen nasal cannula in the emergency room and was transitioned to enoxaparin after admission. On genetic analysis, it was found that the patient was heterozygous for the FVL (R506Q) variant in the Factor V gene. Protein C and Protein S levels were normal. Given failed anticoagulation therapy, an IVC filter was placed. Apixaban was changed to rivaroxaban and the patient was discharged home with close follow up**.**

CONCLUSION

Our patient with an extensive history of CAD and PAD was found to be heterozygous for FVL after presenting with a PE and bilateral DVTs despite anticoagulation. The EINSTEIN EXT trial found that there was an 82% relative risk reduction in recurrent DVT in patients who received rivaroxaban compared to placebo and acetylsalicylic acid. The AMPLIFY-EXT trial found a 64% relative risk reduction for recurrent VTE in patients who received apixaban compared to placebo.

Previous studies have shown that FVL is associated with increased severity of CAD and PAD however the pathophysiology is unclear. It has been hypothesized that long-term anticoagulation may prevent severe progression of PAD and CAD in patients with FVL however it is unknown whether anticoagulation would have prevented progression of vascular disease in our patient. In patients with CAD and PAD with venous thromboembolism despite anticoagulation, screening for FVL may be warranted for appropriate anticoagulation management.

References

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