The Silent Culprit: Factor V Leiden's Covert Role in Recurrent Miscarriages and First-Time Thromboembolism

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Introduction:

Favor V Leiden (FVL) mutation is a leading cause of inherited thrombophilia affecting 1-5% of White populations, often manifesting as venous thromboembolism. Rarely does FVL result in arterial occlusions or miscarriage. We present a case of FVL associated with high-burden venous and arterial clots in a post-menopausal white female.

Case Description:

A 66-year-old-female with twelve prior spontaneous abortions and extensive recent vehicle travel without history of malignancy, prior thromboembolism, smoking, or hormone replacement therapy presented with 1 week of persistent shortness of breath and acute left upper extremity pain and numbness, right upper and lower extremity numbness, and bilateral lower extremity swelling.

Examination revealed no tachycardia, tachypnea, or added lung sounds but there was left arm pallor and coolness to the touch with diminished pulses. A 12-lead ECG Showed normal sinus rhythm with nonspecific T-wave changes. Laboratory evaluation demonstrated a BNP of 258 pg/mL (0-100), troponin of 0.08 ng/mL (<0.06), and D-dimer, performed given low Wells Score, of >= 20.00 ug/mL (<0.5). Chest CTA revealed moderate-to-large volume bilateral pulmonary emboli with evidence of right heart strain. Venous ultrasounds showed occlusive bilateral lower extremity deep venous thrombi without any occlusion in the upper extremities. Left upper extremity arterial duplex showed occlusion of the left axillary and proximal brachial arteries.

Thrombophilia evaluation did not show Lupus anticoagulant or cardiolipin IgM/IgG or beta-2 glycoprotein IgM/IgG antibodies, but revealed a heterozygous FVL mutation, R506Q variant. For intermediate-risk PE, treatment began with LMWH at 1 mg/kg twice daily with transition to apixaban 5 mg twice daily in consultation with Hematology/Oncology and Pulmonology.

Discussion:

Inherited thrombophilia is a rare disorder with FVL being the most prevalent. For first-time thromboembolism with a provoking factor, hypercoagulability studies are typically not pursued. With no clear guidelines for thrombophilia evaluation, this case describes the importance of considering it in those with extensive clot burden or arterial thrombi, even with identified

provoking factor. Complete evaluation includes antiphospholipid antibodies: cardiolipin, beta-2 glycoprotein, and Lupus anticoagulant, which if performed during the event, should be confirmed after resolution. Without any special indications for alternative therapy, DOACs are preferred for anticoagulation.