Factor V Leiden (FVL): the most common genetic risk factor for Venous Thromboembolism (VTE)

Choosing an appropriate anticoagulant regimen and preventing VTE in FVL is quite challenging.

Inadequate anticoagulation has led to life threatening complications in patients homozygous for FVL.

Case Presentation

83-year-old female, homozygous for FVL mutation, with a history of multiple occurrences of DVT and PE, presented with shortness of breath and cyanosis.

No VTE events while on warfarin for 10 years; switched to rivaroxaban for the convenience of monitoring, found to be in cardiogenic shock: Echocardiogram revealed Right Ventricle thrombus.

Treated with heparin drip and then warfarin; however, remained critically ill and expired four weeks later from acute on chronic respiratory failure.

Discussion

FVL - point mutation that eliminates a critical cleavage site in factor V and factor Va → Increased VTE risk.

The choice of anticoagulation is based on severity of thrombosis, patient preference and adherence to therapy.

Hematologists’ preference: Direct Oral Anticoagulation (DOAC) for typical VTE, and warfarin for poor adherence/who benefit from maintaining an INR in the high end of the therapeutic range (e.g., sub-massive/massive PE or DVT with proximal clot burden).

Conclusion

The individual patient factors should be considered when choosing the appropriate anticoagulant regimen in patients homozygous for FVL.

As inadequate anticoagulation may lead to devastating complications, clinicians should obtain a thorough history of thromboembolic events prior to choosing anticoagulation strategy.