Factor V Leiden Thrombophilia

Factor V Leiden thrombophilia is an inherited disorder of blood clotting that results from a single point mutation in the factor V gene resulting in a mutant factor V, which is called factor V Leiden. This mutation causes factor V to be resistant to degradation by activated protein C leading to hypercoagulability. Patients have an increased risk for blood clots, recurrent deep vein thromboses (DVT) and thromboembolism.



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Pathophysiology

Most Common Genetic Clotting Disorder in Caucasians
#1 Foam-finger on Caucasians with Clogs
A mutation in Factor V Leiden is the most common cause of inherited thrombophilia in caucasians. Thrombophilia is defined as an increased likelihood of thrombosis (blood clots).

Genetic Point Mutation
Genes Mutated by Point of claw
This disorder results from a single point mutation at position 506 in the factor V gene.

Glutamine Replaces Arginine
Glued-armor Replaces Argyle
A single point mutation causes an amino acid change (arginine is replaced with glutamine) in the factor V protein produced.

Mutant Factor V
Mutated (5) Hand
The single point mutation results in a mutated factor V, termed factor V Leiden. The mutated factor V lacks the cleavage site for deactivation by proteins C and S.

Resistant to Degradation by Activated Protein C
Protein-ribbon Cat cannot Degrade
Normally, factor V is a procoagulant clotting factor that increases the production of thrombin, which converts fibrinogen to fibrin, ultimately leading to clot formation. Activated protein C (APC) is a natural anticoagulant serine protease in plasma that inactivates factors V and VIII, therefore limiting clotting. Since factor V Leiden lacks the cleavage site of APC, the molecule is less susceptible to inactivation, causing a hypercoagulable state.

Symptoms

Hypercoagulability
Hiker-clogs
Also called thrombophilia or a prothrombotic state, hypercoagulability is an abnormal coagulation of blood resulting in increased risk for thrombosis (formation of blood clots in blood vessels). In factor V Leiden, factor V cannot be inactivated because it lacks the APC cleavage site which causes increased clotting (hypercoagulability).

Recurrent DVT (Increased Risk of Thromboembolism)
DVT with redness and swelling
Due to their hypercoagulable state, patients can develop recurrent deep vein thromboses (DVT). DVT is typically characterized by unilateral limb swelling, tenderness and erythema, although some patients with DVT may be asymptomatic. With increased risk of DVT, inherently, there is a higher chance of thromboembolism and thromboembolic event in patients. Factor V Leiden mutation is also a risk factor for cerebral, mesenteric and portal vein thromboses.

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Considerations

Avoid Oral Contraceptives

Avoid-sign with OCP-case

Oral contraceptives increase the risk of developing venous thromboses. Having factor V Leiden and taking oral contraceptives greatly increases the risk of developing thromboses and therefore patients with a mutated factor V are advised against taking oral contraceptives. Those who wish to take oral contraceptives need to consult their hematologist.

Caution During Pregnancy

Caution-tape Pregnancy

Since pregnancy is associated with a prothrombotic state, pregnant patients with Factor V Leiden should be closely monitored. This disorder may play a role in cases of recurrent late pregnancy loss, possibly because of thrombosis of the placental vessels.

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