When to offer genetic testing for factor V Leiden (FVL)

Testing **is appropriate** in the following circumstances: [1,2]	Other clinical circumstances in which testing **may be appropriate** include the following: [1,2]	FVL testing is **not** routinely recommended: [1,2]
A first unprovoked VTE at any age (especially age <50 years)	Female smokers < 50 years with a myocardial infarction or stroke	• For the general population
A history of recurrent VTE	Women with recurrent unexplained first-trimester pregnancy losses, or an unexplained fetal loss after 10 weeks gestation, or stillbirth	• During routine pregnancy screening
Venous thrombosis at unusual sites (e.g., cerebral, mesenteric, hepatic, or portal veins)	Selected women with unexplained severe preeclampsia, placental abruption, or a fetus with severe intrauterine growth restriction	• Before the use of estrogen contraception hormone replacement or SERMs
VTE and a strong family history of thrombotic disease	A first VTE related to the use of tamoxifen or other selective estrogen receptor modulators (SERMs)	• For prenatal testing and screening of asymptomatic newborns, neonates, and children
VTE during pregnancy or the puerperium	Neonates and children with non-catheter-related idiopathic VTE or stroke	
VTE associated with the use of estrogen contraception or hormone replacement therapy (HRT)	Asymptomatic adult family members of individuals with a known FVL mutation, especially those with a strong family history of VTE at a young age (<50y), when that knowledge may influence pregnancy management, consideration of estrogen contraception use or pregnancy*	• For patients with a personal or family history of arterial thrombosis (acute coronary syndrome or stroke), unless unexplained in an individual under age 50
A first VTE and a first-degree family member with VTE < 50 years		

*Because thrombosis rarely occurs before young adulthood, asymptomatic relatives younger than 18 years are not usually tested, even relatives of homozygotes.[2]

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Management recommendations for asymptomatic FVL carriers

Education	Additional testing	During high risk situations
Carriers should be educated about: ✓ Circumstances that might increase the likelihood of VTE (obesity, age, surgery, reduced mobility due to injury or travel, use of oral contraceptives, HRT, or SERMs, and pregnancy) ✓ The signs and symptoms of VTE that require immediate medical attention ✓ The potential need for prophylactic anticoagulation in high-risk circumstances (e.g. postpartum)[4]	FVL is often seen with other inherited and/or acquired disorders. An individual with FVL should be tested for other thrombophilia disorders to better assess the absolute risk of thrombosis[1,2]. Consider:[1] ✓ Genetic testing for prothrombin 20210G>A variant ✓ Serologic assays for anticardiolipin antibodies and antibeta2glycoprotein 1 antibodies ✓ Multiple phospholipid-dependent coagulation assays for a lupus inhibitor	During high-risk clinical situations (e.g. surgery, pregnancy) prophylactic anticoagulation may prevent some VTE episodes. However, there is no evidence confirming the benefit of primary prophylaxis for asymptomatic FVL heterozygotes. Decisions regarding prophylactic anticoagulation should be based on a risk/benefit assessment in each individual case. [1,3] Consultation with a specialist may be considered.

For more information on FVL see the GEC-KO *on the run* or the more comprehensive GEC-KO Messenger at www.geneticseducation.ca in Educational Resources.

[1] Kujovich JL. Factor V Leiden thrombophilia. *Genet Med* 2011; 13(1): 1-13

[2] Grody WW, Griffin JH, Taylor AK, Korf BR, Heit JA, ACMG Factor V. Leiden Working Group. American College of Medical Genetics consensus statement on factor V Leiden mutation testing. *Genet Med* 2001; 3(2):139-48

[3] Geerts WH, Bergqvist D, Pineo GF, Heit JA, Samama CM, Lassen MR, Colwell CW; American College of Chest Physicians. Prevention of venous thromboembolism: American College of Chest Physicians Evidence-Based Clinical Practice Guidelines (8th Edition). *Chest* 2008; 133(6 Suppl):381S-453S.

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