

Factor V Leiden Thrombophilia Educational Resource (F5 gene)



Why was this tested?

Certain changes in the *F5* gene impact a person's risk of thrombosis. Knowing about an increased risk of thrombosis may impact medication recommendations. These potential medication impacts are why this gene was included on this test.

What are the risks?

Possible increased risk of thrombophilia (clotting). People with one copy of the gene change (heterozygotes) have a slightly higher risk (5-7 fold higher) than the general population. People with two copies of the gene change (homozygotes) have a much higher risk of clotting (25-50 fold higher). There is not a known significant association between Factor V Leiden and pregnancy loss.

Current management recommendations for people with one copy of the gene change (heterozygotes)

- With a personal history of thrombosis:
 - Avoid estrogen-containing contraception and HRT
 - Short-course prophylactic anticoagulation when circumstantial risk factors are present
 - Consider long-term prophylactic anticoagulation
- Without a personal history of thrombosis:
 - Consider alternative forms of contraception and control of menopausal symptoms:
 - Oral contraceptives: avoid third generation and other progestins with higher clot risk
 - Short-term HRT for severe menopausal symptoms should avoid oral formulations
 - Consider short-course prophylactic anticoagulation when circumstantial risk factors are present
 - Long-term prophylactic anticoagulation is not routinely recommended

Current management recommendations for people with two copies of the gene change (homozygotes)

- Avoid estrogen-containing contraception and HRT
- Short-course prophylactic anticoagulation when circumstantial risk factors are present
- Consider long-term prophylactic anticoagulation

General management recommendations

There are many other factors that go into whether someone develops thrombosis, including other genetic and environmental factors. The following are some practical ways to lower clotting risk:

- Maintain ideal body weight, stay active and get regular exercise
- Avoid prolonged periods of immobility
- Don't smoke
- Control other chronic medical conditions such as diabetes, high cholesterol, or congestive heart failure
- Let doctors know that you have Factor V Leiden

What does this mean for family members?

Factor V Leiden thrombophilia is inherited in a dominant pattern, meaning a person who has an F5 gene change has a 50% chance of passing that gene change (and clotting risk) to a child. First degree relatives (parents, siblings, children) of a person with Factor V have a 50% chance of also having the genetic risk factor. At-risk relatives can have genetic testing to determine their status.

References and Resources:

1. GeneReviews: Factor V Leiden Thrombophilia (<https://www.ncbi.nlm.nih.gov/books/NBK1368/>)
2. Ornstein DL & Cushman M. Factor V Leiden. *Circulation*. 2003. 107(15) (<https://www.ahajournals.org/doi/full/10.1161/01.cir.0000068167.08920.f1>)
3. National Blood Clot Alliance (<https://www.stoptheclot.org/>)

This summary information is provided as an educational resource for physicians and other practitioners with the understanding that it is not a substitute for the treating practitioner's independent clinical judgment. The treating practitioner must clinically evaluate each patient considering many relevant factors to determine the final diagnosis and plan of care. Last update July 23, 2020.