

Factor V (Five) Leiden Mutation

What is Factor V Leiden mutation?

Factor V Leiden is a common change in a gene that controls a protein called Factor V. Factor V is involved in blood clotting, and the Factor V Leiden gene change (mutation) is linked to an increased risk of blood clots.

How did I get Factor V Leiden mutation?

Factor V Leiden mutation is an inherited condition. We inherit one copy of each gene from each of our parents. One (or both) of your parents will have passed the mutation on to you, and you may pass it on to your children.

How common is the Factor V Leiden mutation?

Factor V Leiden mutation is common — it occurs in about 1 in every 20–25 people in Australia, more common than many other mutations. Because it's so common, it has been suggested that Factor V Leiden may confer some benefit, though this has never been confirmed.

What are the main problems with having the Factor V Leiden mutation?

The main problem is an increased risk of developing blood clots, which can sometimes travel to different parts of the body including the lungs. If you have one copy of the mutation (heterozygote), you are at around 4–8 times more risk of developing a blood clot compared to someone without it. If you have two copies (homozygote), the risk is substantially higher again.

Importantly, the great majority of people with the Factor V Leiden mutation — including most people with two copies — never actually develop a blood clot in their lifetime. Carrying the mutation is a risk factor, not a diagnosis of a future clot.

Am I going to get a blood clot because I have this mutation?

Most people with one copy of the Factor V Leiden mutation do not develop blood clots. Clots usually only develop when there are other “risk factors” present — for example, after long flights, after surgery, or in people with other serious medical conditions such as cancer or heart failure.

What about pregnancy and the use of hormones / oral contraceptive pills?

Both pregnancy and the use of the pill or hormone replacement therapy (HRT) after menopause can increase the risk of developing a blood clot. Women on the pill have around a 3–4 fold increased risk compared to those not on the pill; women on HRT have around a 2–3 fold increased risk. If a woman also has one copy of the Factor V Leiden mutation, these risks increase further. These figures sound high, but clots remain relatively uncommon in the community.

Current international guidelines do not recommend routinely testing every woman for Factor V Leiden before starting the pill — testing is instead generally reserved for women with a personal or strong family history of blood clots. The risks and suitability of the pill or HRT for you individually should be discussed with your doctor.

Are there any benefits of having Factor V Leiden mutation?

Because blood clots more easily in people with Factor V Leiden, it has been suggested this may be beneficial during bleeding episodes (e.g. menstruation or childbirth). This is only a theory and has not been confirmed by studies.

Is there any treatment for Factor V Leiden?

No treatment to change genes is currently available. Most people with Factor V Leiden mutation do not need any ongoing treatment, but should be careful at times when clot risk may be increased (e.g. after surgery, during long flights — see information sheet on Clots and Flights). Some people may need blood thinning medication for a period of time, depending on personal or family history of clots and the specific situation (e.g. pregnancy, major surgery).

Is there anything else I should do to protect myself against getting blood clots?

Maintaining a healthy weight, stopping smoking, staying active, and keeping other medical conditions under control should also help protect against blood clots. Tell your doctor or surgeon that you have the Factor V Leiden mutation before any operations or prolonged periods of bed rest.

Should my family be tested for Factor V Leiden?

This is now less straightforward than it might seem, and current thinking has shifted. Current international haematology guidelines generally recommend against routinely testing relatives who have no personal history of a blood clot and no other significant risk factors — because knowing the result rarely changes what a doctor would actually recommend, and most carriers never develop a clot regardless.

Testing may still be worthwhile in specific situations — for example, a woman planning a pregnancy with a strong family history of clots, or family members with their own history of blood clots at a young age. This is best discussed individually with your treating haematologist rather than assumed to be a routine test for the whole family.

On the insurance question: new Australian legislation has now banned life insurers from using genetic test results in their decisions altogether (the Treasury Laws Amendment (Genetic Testing Protections in Life Insurance and Other Measures) Act 2026, taking effect from October 2026), which removes much of the historical concern about genetic testing affecting insurance. Clots in children remain very uncommon, so testing is still generally deferred until children are older and can be involved in the decision themselves, unless there is a specific medical reason to test earlier.

When should I seek urgent medical advice?

See a doctor promptly, or go to your nearest emergency department, if you develop swelling or pain in a leg (particularly if one-sided), or symptoms such as chest pain, shortness of breath, or coughing up blood — these can indicate a blood clot in the leg or lungs and need urgent assessment.

References

1. Middeldorp S, Nieuwlaat R, Baumann Kreuziger L, et al. American Society of Hematology 2023 guidelines for management of venous thromboembolism: thrombophilia testing. *Blood Advances*. 2023;7(22):7101-7138.
2. Factor V Leiden Thrombophilia. GeneReviews, National Center for Biotechnology Information. Updated 2024.
3. Treasury Laws Amendment (Genetic Testing Protections in Life Insurance and Other Measures) Act 2026 (Cth).

Further Questions?

The information presented in this fact sheet is intended as a general guide only.

Patients should seek further advice and information about **Factor V (Five) Leiden Mutation** and their individual condition from their treating haematologist or doctor.

For additional information about blood disorders and their treatment, or to contact one of our specialist haematologists, visit the Melbourne Haematology website:

www.melbournehaematology.com.au