



Inherited Thrombophilia Factor V (five) Leiden Gene Mutation

What is inherited thrombophilia?

“Inherited thrombophilia” is a condition that can cause blood clots in veins. Inherited thrombophilia is a genetic condition you were born with.

There are five common inherited thrombophilia types. They are:

- Factor V Leiden.
- Prothrombin gene mutation.
- Protein S deficiency.
- Protein C deficiency.
- Antithrombin deficiency.

About 35% of people with blood clots in veins have an inherited thrombophilia.¹ Blood clots can be caused by many things, like being immobile.

Not everyone with an inherited thrombophilia will get a blood clot.

How did I get an inherited thrombophilia?

Inherited thrombophilia is a gene mutation you were born with. The gene mutation affects coagulation, or blood clotting. The gene mutation can come from one or both of your parents. Sometimes the gene mutation occurs for the first time in patients soon after conception.

You have two copies of every gene (except your gender genes). One copy is inherited from your mother and one copy from your father. Patients can have one copy or

two copies of the gene mutation. If the gene mutation is in:

- Two copies: the patient is **homozygous**.
- One copy: the patient is **heterozygous**.

If other family members suffered blood clots, you are more likely to have inherited thrombophilia.

The gene mutation can be passed on to your children.

How do I find out if I have an inherited thrombophilia?

Blood tests are performed to find inherited thrombophilia.

The blood tests can either:

- Look at your genes (this is DNA testing).
- Measure protein levels.

What is factor V Leiden?

Genes make proteins in your body. The “Factor V Leiden” gene mutation changes the gene that makes a protein called “Factor V”. Factor V helps make your blood clot. Factor V is switched off by a protein called “Activated protein C.”

The Factor V Leiden gene mutation means that Factor V cannot be easily switched off by activated protein C. This can cause excessive blood clotting.

Factor V Leiden is the most common inherited thrombophilia. About 1 in 20 Caucasian people have this gene mutation.²

¹ Weingarz L., et al. Prevalence of thrombophilia according to age at the first manifestation of venous thromboembolism: results from the MAISTHRO registry. *BJH*. 2013, 163(5): pp. 655-665.

² Ridker P.M., et al. Ethnic distribution of factor V Leiden in 4047 men and women. Implications for venous thromboembolism screening. *JAMA*. 1997, 277(16): pp. 1305-1307.



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What happens if you have factor V Leiden?

People with the factor V Leiden gene mutation are at increased risk of developing blood clots such as:

- Deep vein thrombosis (DVT) - blood clots in deep veins of the body.
- Pulmonary embolism (PE) - blood clots in lungs.

Most heterozygous people (with one gene mutation) will not develop a blood clot.³ Homozygous people (with two gene mutations) have a high risk of developing a blood clot.

What do I do now that I know I have factor V Leiden?

Initial treatment of a blood clot in a person with factor V Leiden is no different to that of someone without an inherited thrombophilia. Blood clots are treated with anticoagulant (anti-clotting) medication. Anticoagulant medication helps the body dissolve blood clots and prevents new blood clots forming.

Most people with homozygous Factor V Leiden who develop a blood clot will be advised to stay on anticoagulant medication indefinitely. This is because the chances of the person developing more clots are very high.

See your doctor immediately if you have symptoms of a blood clot or DVT in an arm or leg. Symptoms include:

- Swelling.
- Pain.
- Warmth.
- Redness.

See your doctor immediately if you have symptoms of a Pulmonary Embolism. Symptoms include:

- Shortness of breath.
- Chest pain.
- Rapid heart beat.

To reduce the risk of developing blood clots, people with factor V Leiden should do the following:

- If you have had a blood clot and are not taking anticoagulant medication, you should have preventative anticoagulant medication during all high risk periods. For example:
 - After surgery.
 - Flights over 4 hours.
 - If you are immobile for any reason.
- Avoid oestrogen-containing medications. Speak to your doctor about alternative contraceptive methods.
- Some people with factor V Leiden may need preventative anticoagulant medication during pregnancy, after birth, or both.
- If you are homozygous for factor V Leiden, during periods of high risk, you should take anticoagulant medication.
- Since factor V Leiden gene mutation is a genetic condition, you may wish to tell your family members (e.g. siblings and children) of your diagnosis. This is so they can seek medical advice about their own situation.
- Avoiding smoking and maintaining a healthy body weight will reduce your general risk of developing blood clots.

³ Middeldorp S., et al. A prospective study of asymptomatic carriers of the factor V Leiden mutation to determine the incidence of venous thromboembolism. *Ann Intern Med.* 2001, 135(5): pp. 322-327.

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