

Patient information

Factor V Leiden Mutation – Homozygous

What is Factor V Leiden?

Factor V Leiden is an inherited disorder that makes blood more likely to clot. If you have this disorder you are at risk of developing blood clots, especially in your leg veins, but most people with this disorder have no problems.

Why does blood clot?

Blood has a very important role. It carries nutrients and waste products around the body and carries oxygen from the lungs to all the other tissues. In order to do this blood must remain a liquid but at the same time must be able to clot following injury. Our bodies have developed a complex system of clotting factors in the blood to prevent serious blood loss after minor injuries.

How does blood clot?

When there is blood loss, eg from a cut finger, the clotting process is set in motion in order to prevent prolonged bleeding. The damaged blood vessels trigger a process that leads to a “chain reaction” in which one clotting factor activates another, which in turn activates another and so on, until finally a clot is formed.

Where does Factor V fit in?

When blood clots, clotting Factor V is converted to an active form called Factor Va, which helps to speed up clot formation. When bleeding stops however, clotting needs to be turned off.

Factor V Leiden is an abnormal form of Factor V. It works perfectly in the clotting pathway and speeds up clotting as expected, but it cannot be turned off easily. Therefore people with Factor V Leiden have an increased risk of developing blood clots.

What is the cause of Factor V Leiden?

Genes are the inherited DNA sequences that are the body's means of storing the information needed to produce chemical substances within the body, such as clotting Factor V. You have 2 copies of most genes, one copy being inherited from each of your parents. **Homozygous** means that *both* copies of a gene are the same; in your case both of your Factor V genes code for Factor V Leiden. When this happens there is a greater risk of a blood clot than if only one gene codes for Factor V Leiden.

Factor V Leiden is due to an error in the DNA sequence of the Factor V gene. Approximately 1 in 25 people are born with one Factor V Leiden gene, and around 1 in 2,500 are born with two.

If I have Factor V Leiden...

What problems might I have?

You have an increased risk of developing blood clots in the circulation. These usually occur in veins, especially in the legs. A blood clot in the deep veins of the leg is called a deep vein thrombosis or DVT for short. When a DVT develops blood cannot drain out of the leg properly and the leg becomes swollen and painful and sometimes hot and inflamed. Very occasionally blood clots can form in the veins around the shoulder causing a swollen, painful arm. The main concern about a DVT is that part of the clot can break off and pass through the heart to the main artery of the lungs. This is called a pulmonary embolus or PE for short. A pulmonary embolus blocks the blood supply to the lungs and can cause chest pain and breathlessness.

When am I likely to have problems?

It is important to realise that not everybody with Factor V Leiden will develop a blood clot. In fact, many cases are found during family studies in otherwise healthy people. It is very unusual for clots to develop in childhood, but by the age of twenty a few affected people will have suffered at least one blood clot. The incidence increases with age. Clots occur more frequently after operations, during pregnancy and following immobility, such as long-haul flights, or when dehydrated. Women on the combined contraceptive pill also have a higher risk of thrombosis than normal. In some cases the thrombosis occurs without any obvious cause.

How will I be treated?

If you have a DVT or PE you will receive treatment with anticoagulant medication for a standard length of time, i.e. 3-6 months.

Present advice is that you do not require anticoagulant treatment if you have never had a blood clot.

What is the risk of developing a blood clot (thrombosis)?

The risk in individuals with Factor V Leiden inherited from both parents (homozygous) is approximately 80 times higher than normal. An additional environmental or genetic risk factor for thrombosis is usually present at the time of a thrombosis.

What if I have had a blood clot?

You will be treated with anticoagulant medication for a standard length of time (i.e. 3-6 months) and should follow the advice below to minimise the risk of further blood clots. Anticoagulant medication has, by its nature, a risk of bleeding, which your doctor will need to assess when starting treatment.

Whether you should be treated with anticoagulant medication for longer than the standard length of time, should be discussed with your doctor. The decision to treat for longer will depend on a number of issues, (such as provoking factors and where the clot was), which have to be carefully balanced to minimise the risk of further blood clots and the risk of bleeding on anticoagulants.

How can I reduce the risk of blood clots?

- Stop smoking. Smoking increases the blood's ability to clot.
- Achieve your ideal weight. Obesity increases the risk of clotting.
- Inform your doctor about your condition if you require surgery. You will require heparin treatment or a similar medication around the time of surgical procedures. Compression stockings may also be used.
- Avoid long periods of immobility. For a long-haul flight you may be recommended to take a preventative dose of heparin before the flight (and possibly after the flight). You should also walk around the plane when you can, avoid alcohol and drink plenty of water. You may be advised to wear compression stockings or 'flight socks' for journeys longer than three hours. A haematologist can provide the latest advice with regard to travel.
- Avoid dehydration.

Information for women

What if I want to take the contraceptive pill?

In general, women taking the combined (oestrogen and progestogen) contraceptive pill are **4** times more likely to develop blood clots than women not on the combined contraceptive pill. The presence of the Factor V Leiden mutation increases this risk further. Although the figures sound alarming it should be remembered that blood clots occur very rarely in young women on the pill.

It is recommended that women with Factor V Leiden use an alternative form of contraceptive if possible, especially if they have already had a blood clot, or there are first-degree blood relatives with a history of blood clots. You should discuss alternatives with your doctor.

What if I become pregnant?

You should inform your GP and midwife of the Factor V Leiden disorder as soon as you know you are pregnant, but ideally beforehand.

If you have already had a blood clot: You will need anticoagulant medication during pregnancy in the form of heparin injections. The highest risk period is usually during the late stages of pregnancy (third trimester) and the six weeks after delivery. You will need to continue heparin for six weeks after delivery and wear graduated compression stockings during this period. Your obstetrician or a haematologist can advise.

If you have never had a blood clot: Anticoagulant medication may not be necessary during pregnancy. However you will be recommended to have preventative doses of heparin for at least seven days after delivery and wear graduated compression stockings.

If you develop symptoms of a DVT or PE during pregnancy you should seek urgent medical attention.

What if I want to take hormone replacement therapy (HRT)?

HRT approximately doubles your chances of developing a blood clot. In real terms this is a very small risk. For many women the benefits of HRT outweigh the risk. There is some suggestion that HRT patches may have a slightly lower risk for DVT and PE than HRT tablets. You are advised to discuss this with your doctor.

Can I pass it on to my children?

If you have two copies of the Factor V Leiden gene, one copy will be passed on from you to each of your children; they will carry therefore at least one Factor V Leiden gene. It is important that your children are aware of the risk for blood clots and try to reduce them as far as possible; however, children have a lower risk of developing blood clots than adults.

If you have any questions concerning the information given in this leaflet, please contact the Department of Haematology via the West Suffolk Hospital switchboard 01284 713000.

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