

FACTOR V LEIDEN

This is by far the commonest genetic thrombophilia.

What is Factor V Leiden?

It is due to a change in the gene for Factor V which makes individuals with this profile more prone to venous thrombosis. The risk of venous thrombosis is probably eight-fold greater than those without the variant.

Why is it called Factor V Leiden?

It is named after the town of Leiden, in Holland, where it was originally described.

How is Factor V Leiden inherited?

Factor V Leiden is inherited in an autosomal dominant way. This means that if one parent has it, there is a 50:50 chance (1 in 2 chance) that any child will inherit it. One dose of Factor V Leiden is known as heterozygous Factor V Leiden.

How does Factor V Leiden make blood more prone to venous thrombosis?

Factor V helps in promoting blood clotting. In order to stop blood clotting a natural blood thinner known as Protein C breaks down factor V to stop a clot spreading. If an individual has Factor V Leiden, the factor V molecule is relatively resistant to being broken down, thus the clotting process goes on for longer.

Does Factor V Leiden increase the risk of arterial thrombosis such as heart attacks of strokes?

There has been a lot of work done in this area. Factor V Leiden does not increase the risk of arterial thrombosis. Thus there is no increased risk of heart attacks or strokes.

When can thrombosis of the veins occur?

Thrombosis of the veins in individuals with Factor V Leiden does not occur spontaneously. It usually occurs when other risk factors for thrombosis are present, and more than one other are usually required.

One of the risk factors is increasing age. As we age our blood gets stickier, thus it is very rare for children to have blood clots but quite common in old age.

Other factors are:

- Immobility
- Having an operation especially surgery on the legs, hips, or pelvis
- Pregnancy and the 6 weeks after pregnancy
- Severe illnesses such as inflammatory bowel disease or cancer
- After an accident
- Being overweight
- The use of the combined oral contraceptive pill
- The use of hormone replacement therapy
- The presence of other thrombophilias

What is the risk of having a venous thrombosis with factor V Leiden?

Research suggests the risk is eight times greater than an individual without this defect. In fact to give a sense of proportion, the risk of having a venous thrombosis is very small, and most

individuals with Factor V Leiden do NOT have a venous thrombosis (otherwise 5% of the population would have had one!) Nevertheless it seems sensible that at times of increased risk such as after surgery, that adequate preventative measures are taken.

Are there any advantages in having Factor V Leiden?

This has been the subject of a lot of research recently. For a genetic change to be as common as appearing in one in 20 of the population, then there should be a survival advantage. It seems that the advantage of having Factor V Leiden is that individuals bleed less when they have a wound. Up to less than 100 years ago women often died from bleeding after childbirth. It seems that having Factor V Leiden, women will bleed less in this situation and thus stand a better chance of survival than those without. With modern obstetric care, it is very rare for women to die from bleeding in childbirth and so this is no longer an issue.

Having found out that I have Factor V Leiden, what shall I do?

Firstly you need to remember you are not ill or abnormal. All that has happened is that your increased risk of venous thrombosis has been highlighted.

You can decrease the future risks of venous clots in the future by following the advice below:

- Lead a physically active life
- Eat healthily and avoid becoming overweight
- Avoid prolonged immobility when ill or travelling
- Women should get medical advice about contraception and hormone replacement therapy
- Women should mention they have Factor V Leiden to their obstetrician when they are pregnant
- Liaise with your doctor if you are entering a high risk period for thrombosis such as major surgery, immobility or pregnancy.
- Avoid cigarette smoking

Should other members of the family be tested?

This is recommended for close blood relative (brothers and sisters, sons and daughters) as there is a 50:50 chance that each one may be affected. Because individuals with Factor V Leiden do not tend to have problems until they are adults, it is not usual to test children until they can have blood taken without distress to them and they are old enough to understand why the blood sample is being taken (usually after the age of 13).

What tests are performed to detect Factor V Leiden?

In the laboratory there are two tests. The first is known as *ACTIVATED PROTEIN C RESISTANCE (APCR)*. This is to test for the effect of Factor V Leiden on the clotting system. The "ultimate" test is to look for the change of Factor V Leiden in the genes - a genetic test. APCR is usually performed first as a screening test.

This document is intended to provide general information only and is not intended to provide personal advice or as a substitute for advice covering a specific situation. Readers should seek appropriate advice before taking or refraining from taking any action in response to information contained in this document.

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