

Factor V

The gene for factor V is involved in the blood clotting system. The blood clotting system is regulated by a balance between pro-coagulant (blood clot promoting) and anti-coagulant (blood clot inhibiting) mechanisms. Unless we are injured, this balance favors blood flowing freely through our vessels. If this balance is disturbed, a predisposition to bleeding or clotting may exist. Factor V is a clotting factor that is normally controlled by an anti-coagulant (called activated protein C), which limits the development of blood clots. A particular change in the in factor V gene, known as Factor V Leiden, prevents activated protein C from working well as an anti-coagulant. The result is an increased chance for blood clots in the veins (venous thrombosis). This may cause a condition known as deep vein thrombosis (DVT), which most commonly occurs in the leg or arm. Rarely, blood clots may also travel to the lung – these are known as pulmonary emboli.

Factor V Leiden is very common among Caucasians, being present in about 1 in 20, or 5% of the population. It is estimated that individuals who carry the factor V Leiden are at approximately a 5 to 10 fold increased risk for venous thrombosis. However, if you have never had a blood clot, your risk of developing one remains low, since factor V Leiden actually has a very small role in increasing the risk of clotting. Please see the table below for a comparison of risks for DVT in the general population and for those with factor V Leiden.

Table 1. Incidence of Deep Vein Thrombosis (DVT) in men and women. (Silverstein *et al.*, 1998)

	Age group (years)			
	20-39	40-59	60-79	80+
Incidence in Population				
DVT in men	0.02%	0.05%	0.17%	0.24%
DVT in women	0.04%	0.05%	0.13	0.24%
Incidence in factor V Leiden carriers				
DVT in men (5-10x)	0.1 - 0.2%	0.3 - 0.5%	0.9 - 1.7%	1.2 - 2.4%
DVT in women (5-10x)	0.2 - 0.4%	0.3 - 0.5%	0.6 - 1.3%	1.2 - 2.4%

** Note that factor V Leiden does not increase the risk of heart attacks or aneurysms.

Both hereditary and environmental factors are involved in the formation of a blood clot. Environmental factors include aging, major surgery, cancer, sources of inflammation (e.g. infections), pregnancy, immobilization (e.g. sitting still on a long journey, being confined to a wheelchair), oral contraceptives, hormone replacement therapy, smoking, and obesity. Several different hereditary factors are also involved in blood clotting. In most cases, a combination of these factors (hereditary and/or environmental) causes a clot to form. A person with an inherited tendency to clot will not always develop a blood clot in their lifetime; their chances are just greater compared to people without the genetic risk factor.

The common signs and symptoms of the formation of a blood clot in a vein are swelling and/or pain in the legs or arms associated with redness, numbness, and tingling in these areas. A blood clot in the lungs is associated with shortness of breath and/or sudden pain the lungs. If you experience any of these symptoms, you should consult your doctor immediately.

Genetics of Factor V Leiden

All of the cells of the body, except for the eggs and sperm, have 46 chromosomes (23 pairs). These chromosomes are like packages of information and in them, like beads on a string, are the genes that influence everything about us. Genes, like chromosomes, come in pairs. The eggs and sperm go through a process of division so that they have only one from each chromosome pair, or 23 chromosomes. This means that we inherit half of our genetic information from each of our parents.

Factor V Leiden is an inherited condition caused by a change in the gene for Factor V. An individual who carries Factor V Leiden has one copy of the gene for Factor V that works properly and one copy of the gene for Factor V (the “Leiden” copy) that does not work properly. Sometimes we refer to carriers as “heterozygotes” for factor V Leiden. In each pregnancy, a carrier of Factor V Leiden has a 50% chance to pass on the working factor V gene and a 50% chance to pass on the non-working factor V gene. If both parents are carriers, there is a 1 in 4 (25%) chance that a child may receive a non-working Factor V gene from *both* parents. In this case, the child is “homozygous” for factor V Leiden and would be at further increased risk for venous blood clots. When both parents are carriers, there is also a 50% chance to have a child who is a carrier, and a 25% chance to have a child with two normal copies of factor V. **Because most people with Factor V Leiden will never develop a clot, we do not recommend routine testing of relatives of an individual with Factor V Leiden.** However, if one of your relatives has a blood clot, their doctor should consider testing for Factor V Leiden and other conditions predisposing to clotting; their doctor will decide on the appropriate testing according to their medical history and family history. Sometimes, testing for Factor V Leiden may also help inform the choice of a contraceptive method and the management of pregnancy; your relatives can discuss this directly with their health care provider.

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