There can be three types of test result:

- no change (negative result),
- mutation occurs in one of the two copies of the gene (positive results)
- Leiden mutation is detected on both copies of the gene (positive results)

This result is for the life and does not need to be repeated. The so called APC resistance phenomenon detectable by laboratory testing of coagulation is caused mainly by Leiden mutation.

Indications for testing may be:

- venous thrombosis occurred during pregnancy or use of contraceptive pills
- prior to taking contraceptive pills – history of thrombosis or family history of thromboses
- some cases of repeated abortion
- unexpected obstetrical complications (pregnancy toxaemia, placental abruption, foetal growth retardation or mortality)
- venous thrombosis below 50 years of age
- thrombosis at an unusual site or recurrent thromboses
- venous thrombosis and expressed family history
- abnormal APC resistance test result
- family member confirmed to be Leiden carrier

You should not be frightened even when the result is positive.

Women who carry this genetic disorder may only start taking hormonal contraception after careful consideration of the advantages and disadvantages and only under close control.

In some cases your doctor may recommend prophylactic anticoagulant treatment, change in your lifestyle, frequent vein monitoring and inform you in the early symptoms of thrombosis. There will be opportunity to test the close relatives since the mutation is inherited from our parents and may be transferred to our children.

Assess your Thrombosis risk!

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Deep vein thrombosis (blood clots in the veins) is a common disorder, one in ten persons is affected. Clots form primarily in the large veins of the lower limbs, and the clot may block the path of the blood flow. If a clot breaks away and is carried by the bloodstream, it may lodge in the lungs, the brain or other organs (embolism), which have severe consequences.

Who are at risk?

Particularly, the women of childbearing potential are at risk since the use of contraceptive pills, pregnancy and delivery significantly increase the risk of thrombosis. This is associated with the anticoagulant effect of female sex hormones. This effect is also seen in case of treatment with female sex hormones to promote conception, prevent breast cancer or alleviate menopausal symptoms.

In addition, the development of deep vein thrombosis is facilitated by prolonged bedrest, major operations, trauma, malignant tumours or smoking.

How much is the risk of deep vein thrombosis?

What could be the reason that some people have this disease and others do not? It is not well known that hereditary factors have important role in the tendency to thrombosis. These hereditary factors make the person more prone to thrombosis irrespectively of the mentioned conditions, and can increase the risk by several times if combined with such conditions.

Among the predisposing factors, the most important is genetic variant termed **Leiden mutation**. A mutation in the coagulation **factor II (prothrombin)**, and a variation in the **MTHFR gene** also represent mild risk factors for deep vein thrombosis.

Leiden mutation is present in 20% to 30% of the patients with thrombosis. About 80% of the hereditary predisposition to thrombosis can be attributed to this disorder.

This susceptibility can be inherited both from the father and the mother. If it is inherited only from one parent, we carry 8-fold risk; if from both parents, we carry 80-fold risk to deep vein thrombosis compared to the average population, even when there are no additional risk factors.

Due to the thrombogenic effect of female sex hormones women taking contraceptive pills have deep vein thrombosis 4 times more frequently than those not taking contraceptive pills irrespectively of any susceptibility factors.

If in addition to the use of contraceptive pills a person is prone to have thrombosis (carries Leiden mutation), this risk increases by 30 times compared to the normal population! Literature shows that Leiden mutation may have a role in the development of repeated abortion or some obstetrical complications.

How can it be detected?

An oral mucosal swab taking several seconds without any discomforts is appropriate or the sample taken during gynaecological test.