

# Genetic testing for Inherited Thrombophilia

## What is thrombophilia?

Thrombophilia is a clotting condition that causes medical problems.

Medical problems due to thrombophilia include:

- **Deep vein thrombosis (DVT)** – A clot in the blood vessels in your muscles. DVTs usually happen in the legs, causing pain and swelling in that area.
- **Pulmonary embolism (PE)** - A clot that blocks blood flow to part of the lung. This can happen when a clot forms in another part of the body, then breaks off and moves to the lungs.
- **Arterial embolism** – A clot in a blood vessel that moves oxygen to the body. Clots in an artery can cause a heart attack or stroke.
- **Pregnancy complications:** Too much clotting can affect the placenta and lead to a higher chance for problems during pregnancy, like miscarriage.

## What causes clotting problems?

Clotting problems happen when the body forms blood clots too easily or breaks down clots too slowly. This may be caused by common risk factors, like obesity or diabetes. There are also some people who have clotting problems due to inherited (**genetic**) risk. Clotting events are more likely to happen in a person with multiple risk factors.

## What is inherited thrombophilia?

A clotting problem due to a genetic cause is called **inherited thrombophilia**. About 1 out of every 10 to 20 people has a genetic risk.

## What causes inherited thrombophilia?

Inherited thrombophilia is caused by a change (mutation) in a gene that affects clotting. There are different genes that can lead to clotting problems.

## How do I know if I have a genetic risk?

Your medical history and your family history can help identify if you have a genetic risk. DVT and PE are the most common clotting events in someone with genetic risk. Heart attacks, strokes, or complications during pregnancy are usually **not** due to inherited thrombophilia.

Family history may not always show a genetic risk. Most people with a genetic risk never have clots that need medical care. Some families with inherited thrombophilia have only one or two people with a serious clotting problem. Other families have many people with clotting problems.

### POSSIBLE CAUSES OF THROMBOPHILIA

#### Inherited thrombophilias

- Factor V Leiden (very common)
- Prothrombin 20210 (common)
- Protein S deficiency
- Protein C deficiency
- Antithrombin deficiency (rare)

#### Health problems that can cause clotting (Acquired thrombophilias)

- Obesity
- Diabetes
- Cancer
- Heart failure
- Organ transplantation
- Antiphospholipid syndrome

#### Other risk factors

- Travel (sitting for long trips)
- Smoking
- Pregnancy
- Oral contraceptives
- Hormone use (estrogen or testosterone)
- Getting older
- Surgery (especially orthopedic surgery)
- Major trauma (accident or injury)
- Central venous catheters

# Genetic testing for Inherited Thrombophilia

## How is inherited thrombophilia diagnosed?

Blood tests can be done to measure clotting proteins or look for mutations in thrombophilia genes. Testing is usually only done when the medical history suggests a genetic risk. Testing may also look for non-genetic health problems that can cause clotting problems. Your doctor can help determine whether or not testing is useful and which tests to order.

## What are the benefits and limits of genetic testing?

### Benefits

If testing finds a genetic risk for thrombophilia:

- You can make lifestyle choices that could lower your chance for a clotting event.
- Your doctor may recommend using birth control methods that do not include estrogen.
- Testing may be offered to your family members.

### Limits

Testing is not always helpful.

- Test results may not change your medical care. Most people with a genetic risk do not need any special treatment.
- We cannot change our genes, we can only change some risk factors like our diet and exercise.
- Genetic testing cannot tell you if you will have a clotting event.
- Finding a genetic risk may cause you to worry.

## Can I have testing during pregnancy?

Testing for mutations in thrombophilia genes can be done at any time, including during pregnancy. However, tests that measure clotting proteins are not reliable during pregnancy.

## Medical problems that suggest inherited thrombophilia

### You or someone in your family has had:

- A blood clot without any known risk factors
- More than one blood clot
- A blood clot in childhood
- A blood clot before 50 years old with only one risk factor (minor surgery, bone fracture, pregnancy).
- A close relative diagnosed with a genetic risk for thrombophilia

## What if all the test results are normal?

A normal result could mean that the clotting problems are not due to a genetic risk. But not all genetic risk can be found with the current tests. A strong family history of clotting problems may still be due to inherited thrombophilia, even when genetic testing is normal. It is important to talk with your doctor about the meaning of any test result you receive, even normal results.

## Will I develop a blood clot if I have a genetic risk for thrombophilia?

No test can tell you if or when you will develop a blood clot. Most people with a genetic risk for clotting never having a clotting event. Your chance to develop a serious blood clot depends on which gene is changed and whether or not you have other risk factors. The chance may also be higher if you carry changes in two or more genes.

## References:

GeneReviews – Factor V Leiden Thrombophilia (2018)  
GeneReviews – Prothrombin Thrombophilia (2021)

Up-To-Date - Overview of the causes of venous thrombosis (2021)