Thrombophilia is when your body forms blood clots too easily or breaks down clots too slowly.

- You have proteins to form clots and stop you from bleeding.
- You also have proteins that break down clots when they are no longer needed.

Thrombophilia (clotting problems) can cause many different medical problems, including:

- **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** - 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 thrombophilia?

There are many common risk factors for thrombophilia. You are more likely to have a clotting problem when you:

- Are overweight
- Smoke
- Are pregnant
- Use birth control pills
- Have surgery
- Get older

But there are also people who are more likely to clot because of inherited (genetic) 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 that can affect clotting.

What causes inherited thrombophilia?

Inherited thrombophilia is caused by a mutation (genetic change) in one of the genes needed for clotting. There are several genes in your body that make clotting proteins. When one of the clotting proteins is made in the wrong amount or doesn’t work right, it can affect the clotting process.

How do I know if I have an inherited risk for thrombophilia?

Inherited thrombophilia may be suspected based on your medical history and your family history. Someone with an inherited risk for thrombophilia has a higher risk of getting blood clots, especially in the leg or the lungs. However, there are many steps in making and breaking down clots, and most people with an inherited risk for thrombophilia never have a clotting problem. Some families with inherited thrombophilia have just one or two people with a serious clotting problem. Other families have had many people with clotting problems. DVT is the most common clotting problem in these families.

Thrombophilia can sometimes lead to heart attacks, strokes, or complications during pregnancy. However, these type of medical problems are usually not related to inherited thrombophilia genes.
How is inherited thrombophilia diagnosed?
When inherited thrombophilia is suspected, blood tests may be done to measure clotting proteins or to look for mutations in thrombophilia genes. Testing is most useful when there is a strong history of clotting problems. 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 an inherited risk for thrombophilia:
- You can make lifestyle choices that could lower your chance for a clotting event.
- Testing may be offered to your family members in some situations.

Limits
Testing is not always helpful. Genetic testing:
- May not change your medical care. For example, decisions about using blood thinners (anticoagulants) are not usually based on genetic test results.
- Does not tell you if you will have a clotting event.
- May cause unneeded anxiety.

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 the amount of clotting proteins in your blood are not reliable during pregnancy.

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 causes 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 an inherited 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 live their entire lives without ever having a clotting event. Your chance to develop a serious blood clot depends on which protein is not working normally and whether or not you have other risk factors.

POSSIBLE CAUSES OF THROMBOPHILIA

Inherited thrombophilias
- Factor V Leiden (common)
- Prothrombin 20210
- 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 contraceptive use
- Hormone replacement therapy (HRT)
- Getting older
- Surgery (especially orthopedic surgery)
- Central venous catheters

References:
GeneReviews – Prothrombin-Related Thrombophilia (2014)
Up-To-Date - Overview of the causes of venous thrombosis (2016)