



# Inherited Thrombophilia

# Antithrombin Deficiency

## What is inherited thrombophilia?

“Inherited thrombophilia” is a condition that can cause blood clots in veins. Inherited thrombophilia is a genetic condition you were born with.

There are five common inherited thrombophilia types. They are:

- Factor V Leiden.
- Prothrombin gene mutation.
- Protein S deficiency.
- Protein C deficiency.
- Antithrombin deficiency.

About 35% of people with blood clots in veins have an inherited thrombophilia.<sup>1</sup> Blood clots can be caused by many things, like being immobile.

Not everyone with an inherited thrombophilia will get a blood clot.

## How did I get an inherited thrombophilia?

Inherited thrombophilia is a gene mutation you were born with. The gene mutation affects coagulation, or blood clotting. The gene mutation can come from one or both of your parents. Sometimes the gene mutation occurs for the first time in patients soon after conception.

You have two copies of every gene (except your gender genes). One copy is inherited from your mother and one copy from your father. Patients can have one copy or two copies of the gene mutation.

If the gene mutation is in:

- Two copies, the patient is **homozygous**.
- One copy, the patient is **heterozygous**.

If other family members suffered blood clots, you are more likely to have inherited thrombophilia.

The gene mutation can be passed on to your children.

## How do I find out if I have an inherited thrombophilia?

Blood tests are performed to find inherited thrombophilia.

The blood tests can either:

- Look at your genes (this is DNA testing).
- Measure protein levels.

## What is antithrombin deficiency?

Genes make proteins in your body. Antithrombin is a protein in our body that prevents blood clotting. People with the antithrombin deficiency gene mutation do not make enough antithrombin protein. This causes excessive blood clotting.

Sometimes, people produce enough antithrombin, but the mutation they have makes antithrombin protein that does not work properly.

Some people may develop low antithrombin levels due to conditions such as liver disease or severe infection. This is called acquired antithrombin deficiency. This is not a genetic problem.

<sup>1</sup> Weingarz L., et al. Prevalence of thrombophilia according to age at the first manifestation of venous thromboembolism: results from the MAISTHRO registry. BJH. 2013, 163(5): pp. 655-665.



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## What happens if you have antithrombin deficiency?

People with antithrombin deficiency are at significantly increased risk of developing blood clots such as:

- Deep vein thrombosis (DVT) - blood clots in deep veins of the body.
- Pulmonary embolism (PE) - blood clots in lungs.
- Mesenteric venous thrombosis - blood clots in veins of the intestines.

## What do I do now that I know I have antithrombin deficiency?

Initial treatment of a blood clot in a person with antithrombin deficiency is no different to that of someone without an inherited thrombophilia. Blood clots are treated with anticoagulant (anti-clotting) medication. Anticoagulant medication helps the body dissolve blood clots and prevents new blood clots forming.

Most people with antithrombin deficiency who develop a blood clot will be advised to stay on anticoagulant medication indefinitely. This is because the chances of the person developing more clots are very high.

In rare cases a patient will require antithrombin concentrate. This is prepared from blood donations.

**See your doctor immediately if you have symptoms of a blood clot or DVT in an arm or leg. Symptoms include:**

- Swelling.
- Pain.
- Warmth.
- Redness.

**See your doctor immediately if you have symptoms of a Pulmonary Embolism. Symptoms include:**

- Shortness of breath.
- Chest pain.
- Rapid heart beat.

**People with antithrombin deficiency who have never had a blood clot should take the following precautions:**

- Avoid oestrogen-containing medications. This includes the combined oral contraceptive pill and hormone replacement therapy. Speak to your doctor about alternative contraceptive methods.
- Preventative anticoagulant medication in the form of enoxaparin/Clexane® should be injected daily during pregnancy and after birth.
- Avoiding smoking and maintaining a healthy body weight will reduce your general risk of developing blood clots.
- You should have preventative anticoagulant medication during all high risk periods.  
For example:
  - After surgery.
  - Flights over 4 hours.
  - If you are immobile for any reason.
- Inherited antithrombin deficiency is a genetic condition, you may wish to tell your family members (e.g. siblings and children) of your diagnosis. This is so they can seek medical advice about their own situation.

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