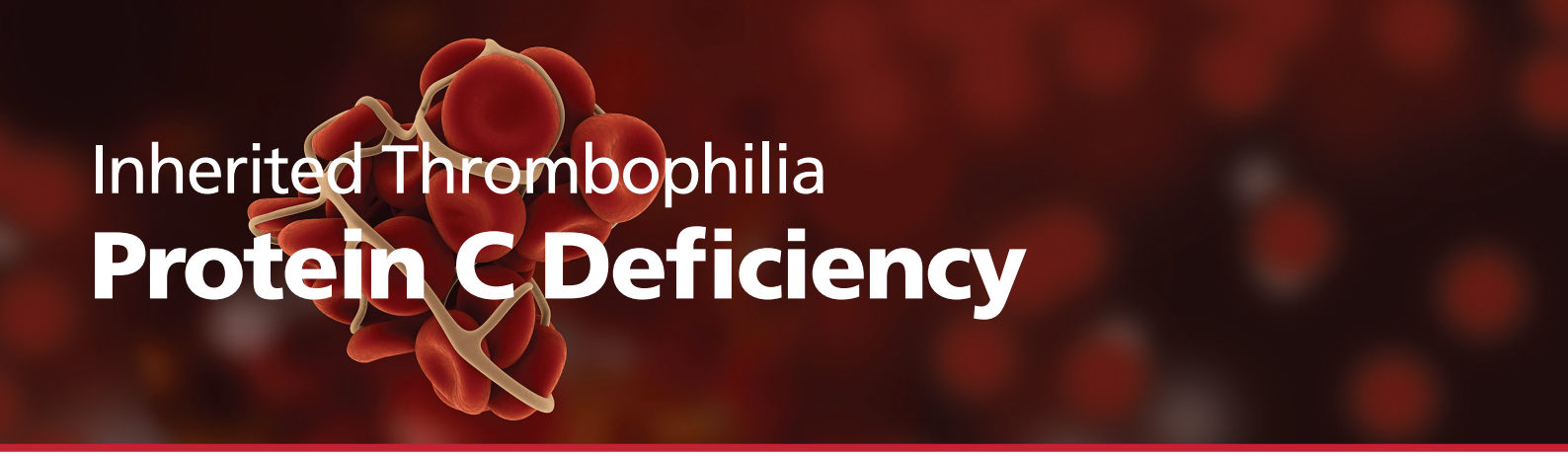

Inherited Thrombophilia
Protein C Deficiency

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.[1] 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 protein C deficiency?

Genes make proteins like protein C in your body. The function of protein C is to reduce blood clotting. People with the protein C deficiency gene mutation do not make enough protein C. This results in excessive clotting.

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

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

[1] 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 protein C deficiency?

People with protein C deficiency are at 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.

Most heterozygous people will not develop a blood clot.[2]

Warfarin-induced skin necrosis is a rare skin problem. It may occur when people with low protein C levels start taking high doses of warfarin. The rash is initially red and turns dark purple over hours to days. It is very painful. This condition requires urgent treatment.

Neonatal purpura fulminans is a very rare life-threatening condition that can occur in newborns who are born with very low levels of protein C. This is usually caused by homozygous protein C deficiency. Affected babies suffer from severe blood clotting and bleeding.

What do I do now that I know I have protein C deficiency?

Initial management of a blood clot in a person with protein C 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.

If warfarin is prescribed, the patient will be started on a low dose and the dose increased slowly. A person with protein C deficiency who develops a blood clot may be advised to stay on anticoagulant medication for a longer time. The treatment duration will also depend on why the clot formed and other risk factors.

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 protein C deficiency should take the following precautions.

- Depending on your other risk factors, it may be safer to avoid oestrogen-containing medications. Speak to your doctor about alternative contraceptive methods.
- Some people with protein C deficiency should have preventative anticoagulant medication during pregnancy, after birth, or both.
- Avoiding smoking and maintaining a healthy body weight will reduce your general risk of developing blood clots.
- If you have had a blood clot and are not taking anticoagulant medication, 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.
- Since inherited protein C 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.

[2] Sanson B., et al. The Incidence of Venous Thromboembolism in Asymptomatic Carriers of a Deficiency of Antithrombin, Protein C, or Protein S: A Prospective Cohort Study. Blood. 1999, 94(11): pp. 3702-3706.

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