Inherited Thrombophilia

Prothrombin Gene Mutation

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. 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 prothrombin gene mutation?

Genes make proteins in your body. Prothrombin is a very important protein involved in blood clotting. Prothrombin is turned into thrombin, which helps blood clot. People with the prothrombin gene mutation produce more prothrombin than they need. This can cause excessive clotting.

Prothrombin gene mutation is the second most common inherited thrombophilia after Factor V Leiden. Some patients may have a prothrombin gene mutation as well as a Factor V Leiden mutation. These patients have a higher risk of clotting.

What happens if you have prothrombin gene mutation?

People with prothrombin gene mutation 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.
- Cerebral vein thrombosis - blood clots in veins of the brain.

Most heterozygous people will not develop a blood clot.

What do I do now that I know I have prothrombin gene mutation?

Initial treatment of a blood clot in a person with prothrombin gene mutation 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.

A person with prothrombin gene mutation 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 prothrombin gene mutation 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 prothrombin gene mutation 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 prothrombin gene mutation 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.

---