Thrombosis UK

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Inherited thrombophilia

The term genetic or inherited thrombophilia covers a range of conditions that are inherited by someone at birth. This means that their blood is stickier than normal, which increases the risk of blood clots and thrombosis.

Factor V Leiden and Prothrombin 20210 are the most common thrombophilias among people of European origin. Other genetic thrombophilias include protein-C deficiency, protein-S deficiency and antithrombin deficiency.

Factor V Leiden

Factor V Leiden is by far the most common genetic thrombophilia. In the UK it is present in 1 in 20 individuals of European origin. It is rare in people of Afro-Caribbean or Asian origin.

Factor V Leiden is caused by a change in the gene for Factor V, which helps the blood to clot. To stop a clot spreading, a natural blood thinner, known as protein C, breaks down Factor V.

If you have Factor V Leiden, the Factor V molecule in the blood is more resistant to being broken down and the clotting process goes on for longer. This makes you more prone to blood clots, which can lead to venous thrombosis.

This risk of venous thrombosis is about eight times greater for people who have Factor V Leiden than for those who don't. Factor V Leiden does not increase the risk of arterial thrombosis, so there is no increased risk of heart attacks or strokes.

Inheriting Factor V Leiden

If you inherit Factor V Leiden from one parent, this is known as heterozygous Factor V Leiden. But as Factor V Leiden is relatively common, some individuals inherit it from both parents and this is known as homozygous Factor V Leiden. It affects 1 in 1600 people of European origin.

Factor V Leiden and venous thrombosis

Most people with Factor V Leiden have a low risk of venous thrombosis unless you have one or more other risk factors.

The risk of having a venous thrombosis is about eight times greater if you have heterozygous Factor V Leiden than for someone without Factor V Leiden. However, this risk is still relatively low and most people with the condition are not affected.

The risk of venous thrombosis is much higher if you have homozygous Factor V Leiden. It is about 80 times greater than for those without the condition.

Factor V Leiden and pregnancy

If you are pregnant and have Factor V Leiden, it is important that you discuss this with your midwife as you have an increased risk of venous thrombosis during pregnancy. Some evidence suggests that you may also have a slightly higher risk of placental problems.

Prothrombin 20210

Prothrombin is one of the blood-clotting factors. It circulates in the blood and, when activated, is converted to thrombin. Thrombin causes fibrinogen, another clotting factor, to convert fibrin strands, which make up part of a clot.

The condition known as Prothrombin 20210 is due to a mutation of the prothrombin gene. If you have the condition, you will tend to have slightly stickier blood due to higher prothrombin levels.

Prothrombin 20210 is present in 2 in 100 people of European origin. This means that the risk of having a venous thrombosis is twice that of someone without the condition.

Prothrombin 20210 does not increase the risk of arterial thrombosis, so there is no increased risk of heart attacks or strokes.

Inheriting Prothrombin 20210

Prothrombin 20210 is inherited in an 'autosomal dominant way'. This means that if one parent has it, there is a 50:50 chance that you will inherit it. If you do inherit it, is known as heterozygous Prothrombin 20210.

There are a few cases where both parents have it and so you would inherit homozygous Prothrombin 20210. However, this is extremely rare.

Prothrombin 20210 and venous thrombosis

If you have Prothrombin 20210, you have a low risk of venous thrombosis unless you have one or more extra risk factors.

The risk of venous thrombosis if you have hetero or homo Prothrombin 20210 is about twice that of someone without the condition. However, this risk is relatively low and most people with the condition are not affected.

However, if you have Prothrombin 20210 as well as Factor V Leiden or Protein S, the risk of thrombosis is substantially increased.

Prothrombin 20210 and pregnancy

If you have Prothrombin 20210 and are pregnant, you should discuss this with your midwife. As well as an increased risk of venous thrombosis, women with Prothrombin 20210 may have a slightly higher risk of placental problems during pregnancy.

Testing for Prothrombin 20210

As it is a common condition, Prothrombin 20210 is usually included as part of standard thrombophilia screening. It is detected through a genetic test - a blood sample is taken and the DNA is examined to look for the mutation (inherited defect) in the prothrombin gene.

Testing other family members

If you have Prothrombin 20210, it is recommended that close blood relatives are tested, including brothers and sisters and sons and daughters, as there is a 50:50 chance that they will inherit the condition.

Because people with genetic thrombophilias do not tend to have problems until they are adults, children are not usually tested until they are old enough to understand why the blood sample is being taken. This is usually after age 13.

Protein-C deficiency

Protein C is one of the natural anticoagulants found in theblood. If you do not have enough protein C, you are more likely to have a venous thrombosis. It is uncertain whether there may also be a very slight risk of arterial thrombosis.

Inheriting protein-C deficiency

Protein C is inherited in an 'autosomal dominant' way. This means that if one parent has it, there is a 50:50 chance that you will inherit it.

It is extremely rare for both parents to have it (less than one in a million). If a child does inherit it from both parents, this is known as homozygous protein-C deficiency. Babies born with this condition have skin thromboses soon after birth and need protein-C concentrates or other anticoagulants to keep them healthy and free from further thrombosis.

Other causes of protein-C deficiency

Occasionally, medical conditions, such as kidney disease, can cause low levels of protein C.

Protein-C deficiency and venous thrombosis

The risk of venous thrombosis is increased if you have protein-C deficiency, but the level of risk varies. This is because each family with the condition has a different mutation within the protein-C gene, which affects the level of risk.

One of the best guides to risk is to look at other members of your family who have protein-C deficiency.

If nobody in your family has had a venous thrombosis, the risk is probably low unless you have other risk factors.

If a number of family members with protein-C deficiency have had a venous thrombosis, the risk is greater. However, you can reduce this risk by taking preventive measures.

Protein-C deficiency and pregnancy

If you have protein-C deficiency and are pregnant, you should discuss this with your midwife. As well as an increased risk of venous thrombosis, you may have a slightly higher risk of placental problems during pregnancy such as a slightly smaller baby or pre-eclampsia.

Testing for protein-C deficiency

There are a number of tests to detect protein-C deficiency and these vary between different laboratories. Sometimes a hospital may send the test to a specialist laboratory.

Usually a test is carried out to measure the amount of protein C that is working in the blood. This is known as a functional protein C assay. Other tests may measure the total amount of protein C in the blood.

Testing other family members

Testing is recommended for close blood relatives of people with a protein-C deficiency, including brothers and sisters and sons and daughters, as there is a 50:50 chance that they will inherit the condition.

Because people with genetic thrombophilias do not tend to have problems until they are adults, children are not usually tested until they are old enough to understand why the blood sample is being taken. This is usually after age 13.

Protein-S deficiency

Protein S is one of the natural anticoagulants found in the blood. If you do not have enough protein S, you are more likely to have a venous thrombosis. It is uncertain whether there may also be a very slight risk of arterial thrombosis.

Inheriting protein-S deficiency

Protein S is inherited in an 'autosomal dominant' way. This means that if a parent has it, there is a 50:50 chance that a child will inherit it.

Other causes of protein-S deficiency

Protein-S levels in the blood fall naturally during pregnancy and when women used the combined oral contraceptive pill or certain types of hormone replacement therapy (HRT). This is a factor in the increased risk of venous thrombosis in pregnant women and those taking combined oral contraceptives and HRT.

Occasionally other medical conditions, such as kidney disease, can cause low levels of protein S.

Protein-S deficiency and venous thrombosis

The risk of venous thrombosis is increased if you have protein-S deficiency, but the level of risk varies. This is because each family with the condition has a different mutation within the protein-S gene, which affects the level of risk.

One of the best guides to risk is to look at other members of the family with protein-S deficiency. If nobody in your family has had a venous thrombosis, the risk is probably low unless you have other risk factors.

If a number of family members with protein-S deficiency have had a venous thrombosis, the risk is greater. You can reducethis risk by taking preventive measures.

Protein-S deficiency and pregnancy

If you have protein-S deficiency and are pregnant, you should discuss this with your midwife. As well as an increased risk of venous thrombosis, you may have a slightly higher risk of placental problems during pregnancy. These include having a smaller baby or pre-eclampsia.

Testing for protein-S deficiency

There are a number of tests to detect protein-S deficiency and these vary between different laboratories. Sometimes a hospital may send the test to a specialist laboratory.

Usually a test is carried out to measure the amount of protein S that is working in the blood. This is known as a functional protein S assay. Other tests may measure the total amount of protein S in the blood, or how much free protein S is present.

Testing other family members

Testing is recommended for close blood relatives of people with a protein-S deficiency, including brothers and sisters and sons and daughters, as there is a 50:50 chance that they will inherit the condition.

Because people with genetic thrombophilias do not tend to have problems until they are adults, children are not usually tested until they are old enough to understand why the blood sample is being taken. This is usually after age 13.

Antithrombin deficiency

Antithrombin (previously known as antithrombin III) is one of the natural anticoagulants found in the blood. If you do not have enough antithrombin, you are more likely to have a venous thrombosis. It is uncertain whether there may also be a very slight risk of arterial thrombosis.

Inheriting antithrombin deficiency

Antithrombin deficiency is inherited in an 'autosomal dominant' way.

This means that if a parent has it, there is a 50:50 chance that a child will inherit it.

If it is inherited from one parent, this is known as heterozygous antithrombin deficiency. It is extremely rare for both parents, to have it and if a foetus inherits it, it is known as homozygous antithrombin deficiency. In this situation, the foetus does not survive.

Other causes of antithrombin deficiency

Occasionally, medical conditions, such as kidney disease, can cause low levels of antithrombin.

Antithrombin deficiency and venous thrombosis

Of all the genetic thrombophilias, heterozygous antithrombin deficiency is associated with the greatest risk of venous thrombosis. As many as 1 in 2 people with antithrombin deficiency will have a venous thrombosis before the age of 50 if they do not follow advice to reduce the risks.

During pregnancy, up to half of all women with antithrombin deficiency may have a venous thrombosis if they do not receive thromboprophylaxis to reduce the risk. This involves giving small amounts of anticoagulant drugs to prevent blood clots.

The level of risk varies between individuals with antithrombin deficiency. This is because each family with the condition has a different mutation within the antithrombin gene, which affects the level of risk.

One of the best guides to risk is to look at other members of your family with antithrombin deficiency. If nobody in your family has had a venous thrombosis, the risk is probably low unless you have other risk factors.

If a number of family members with antithrombin deficiency have had a venous thrombosis, the risk is greater. You can reduce this risk by taking preventive measures.

Antithrombin deficiency and pregnancy

If you have antithrombin deficiency and are pregnant, you should discuss this with your midwife. As well as an increased risk of venous thrombosis, you may have a slightly higher risk of placental problems during pregnancy. These include having a smaller baby or pre-eclampsia.

Testing for antithrombin deficiency

There are a number of tests to detect antithrombin deficiency and these vary between different laboratories. Sometimes a hospital may send the test to a specialist laboratory.

Usually a test is carried out to measure the amount of antithrombin that is working in the blood. This is known as a functional antithrombin assay. Other tests may measure the total amount of antithrombin present in the blood.

Testing other family members

Testing is recommended for close blood relatives of people with antithrombin deficiency, including brothers and sisters and sons and daughters, as there is a 50:50 chance that they will inherit the condition.

Because people with genetic thrombophilias do not tend to have problems until they are adults, children are not usually tested until they are old enough to understand why the blood sample is being taken. This is usually after age 13.

Other genetic thrombophilias

There are a few other rare conditions that are associated with venous thrombosis. These include dysfibrinogenaemia.

Dysfibrinogenaemia

People with dysfibrinogenaemia have an abnormal fibrinogen molecule in the blood. Some people with this condition have a tendency to bleed more, others have a tendency to thrombosis, and some have both.

Inheriting dysfibrinogenaemia

Dysfibrinogenaemia is inherited in an 'autosomal dominant' way. This means that if a parent has it, there is a 50:50 chance that you will inherit it.

Dysfibrinogenaemia and venous thrombosis

The risk of thrombosis depends on the type of dysfibrinogenaemia.

The risk of venous thrombosis increases if you have one or more other risk factors. You can reduce this risk by taking preventive measures.

Dysfibrinogenaemia and pregnancy

If you have dysfibrinogenaemia and are pregnant, you should discuss this with your midwife as you are at increased risk of venous thrombosis during pregnancy.

Preventing venous thrombosis

Anyone who has a genetic thrombophilia can reduce the risk of venous thrombosis by making lifestyle changes like:

- leading a physically active life;
- eating a healthy, balanced diet;
- avoiding becoming overweight or obese;
- avoiding long periods of not moving around during
- illness or when travelling; and
- stopping smoking.

If you have genetic thrombophilia, you should also get medical advice before major surgery or when you are likely to be unable to move around for a long period, as this increases the risk of venous thrombosis.

if you are a woman, you should also get medical advice before taking oral combined contraception or hormone replacement therapy, and when pregnant or planning to become pregnant.

For more information

If you have any more questions about DVT or PE, speak to your doctor.

Thrombosis UK can also offer you more information and be contacted by the following means:

Thrombosis UK,
PO Box 58, Llanwrda,
Carmarthenshire
SA19 0AD.

Phone: 0300 772 9603,
email: admin@thrombosisuk.org
or visit our website at www.thrombosisuk.org

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