

Thrombophilia

Information for patients and their relatives

Introduction

Blood circulates around the body delivering oxygen and removing waste products. If the blood clots in the circulation this is known as a **Thrombosis**.

When blood clots in a deep vein it is known as a **Deep Vein Thrombosis** or **DVT**. When a DVT occurs, pieces of clot can break off and travel through the circulation and lodge in the lungs. This is known as a **Pulmonary Embolus** or **PE**. Sometimes a small clot can form in a vein under the skin and the vein becomes hard, hot and tender, this is known as **Superficial Thrombophlebitis**. It is not the same as DVT and PE.

Risk factors for DVT and PE are

- surgery
- prolonged immobility
- major trauma

There is also a risk in women using oestrogen-progestogen combined oral contraceptive pills or hormone replacement therapy but the risk is very small, typically less than 1 in 3000. Pregnancy is also a risk factor (*see below under Specific Actions*).

In addition to these risks individuals may have an underlying genetic predisposition to DVT and PE- this is known as **Thrombophilia**.

(Blood can also clot in an artery and this can cause a stroke or a heart attack. Individuals with thrombophilia are not typically predisposed to **Arterial Thrombosis**. Smoking, high blood pressure and high blood lipid (fat) levels are risk factors for arterial thrombosis).

Blood tests can be done after a DVT or a PE to test for thrombophilia

If you have had a DVT or a PE your doctor may choose to do blood tests after you have recovered, this is usually after you have completed anticoagulant treatment. The purpose of the tests is to try and determine if you have **thrombophilia**. If the test results indicate that you have a form of thrombophilia this would partly explain why you suffered a blood clot. The risk of another blood clot is not significantly greater in most individuals with thrombophilia than it is in individuals without thrombophilia. Therefore, your treatment, for example your anticoagulant therapy, will probably be no different from that given to other patients with DVT or PE.

Occasionally, your doctor may consider you for prolonged treatment. This decision will be based not only on whether you have thrombophilia but more importantly on the circumstances that led to your thrombosis, for example whether or not the clot followed an operation and if you have a strong family history of DVT or PE.

What tests can be done?

A variety of tests can be done and your doctor or thrombophilia nurse specialist will determine these. Some tests are blood clotting tests which actually measure clotting factors and some are genetic tests that determine if you have a variation in your genetic code that is associated with a predisposition to DVT or PE. The results of these tests should be confidential and only available to you and your health care team.

What types of thrombophilia are there?

There are many different types of thrombophilia and new types are being discovered and recognised all the time. For example within your circulation there are a variety of **Natural Anticoagulants**, which are naturally present and control blood coagulation in healthy individuals. About 1 in 3000 people have a reduced level of one of these natural anticoagulants and this may increase their risk of DVT and PE. The three main anticoagulants are:

- Antithrombin
- Protein C
- Protein S

Much more common than deficiency of a natural anticoagulant is a variation within an individual's genetic code known as a mutation. Some mutations are present in about 1 in 30 people and also increase the risk of DVT and PE, but to a lesser degree than deficiency of a natural anticoagulant. The two main mutations that do this are:

- Factor V Leiden (*V is pronounced 'five'*)
- The prothrombin gene mutation, also known as factor II Leiden (*II is pronounced 'two'*).

Finally, an increase in some clotting factors can increase the risk of blood clots. An example of this type of thrombophilia is:

- High factor VIII level

What special precautions should I take if I have thrombophilia?

The precautions you should take are the same as for anyone who has suffered a DVT or a PE. If you are overweight you should lose weight. If you are a smoker you should stop smoking. You should adopt a healthy lifestyle with a sensible diet and take regular exercise.

There are certain situations in which you may be at significantly increased risk of another DVT or PE and you should consult your doctor or nurse specialist for advice. If you are admitted to hospital before you have time to get advice you must inform the medical staff that are looking after you that you have thrombophilia and that you have had a previous DVT or PE. Situations that are associated with a potentially increased risk of another DVT or PE are:

- Surgical operations (only major surgery is associated with a risk, for example abdominal surgery under general anaesthetic or an orthopaedic operation. Heparin injections may be given to reduce thrombosis risk. Minor surgery, such as dental surgery or biopsies under local anaesthetic are not high risk situations)
- Prolonged immobility or plaster casts
- Use of Combined Oral Contraceptive pills containing oestrogens
- Use of Hormone Replacement Therapy

Remember to seek advice if any of these apply to you at any time. It may well be that a simple precaution can avoid problems.

What symptoms or signs should I look out for that might indicate another DVT or PE?

After a DVT it is common for an individual to suffer pain and swelling in the leg. This is due to damage to the valves in the veins and the symptoms are known as the **Post Phlebotic Syndrome**. The symptoms are usually worse at the end of the day, or after prolonged standing, and are better in the morning. The important thing is to recognise a new blood clot. Therefore if you develop a new pattern of pain or swelling, for example if the pain or swelling are progressive and don't ease after a nights rest or you develop very severe pain and swelling that you don't usually have, then you should contact your doctor.

The symptoms of a PE are chest pain, typically very sharp and worse on breathing and coughing. Sharp chest pain lasting a few seconds is very common in everybody. However, sharp chest pain that is persistent, for example lasting more than one hour is unusual and you should contact your doctor. Occasionally, difficulty breathing without any chest pain is the first sign of a PE. Therefore, if you develop a new problem with breathing, which is persistent, you should contact your doctor.

The important things to remember are

- Ask for advice concerning high risk situations, and
- Do not delay seeking help if you think you have another DVT or PE

Should other members of my family be tested for thrombophilia?

Screening the general population for blood clotting abnormalities is unhelpful. However, if someone who has suffered a DVT or a PE is found to have evidence of thrombophilia it may then be possible to search for the same thrombophilia abnormality in other family members. The reason for doing this would be to identify other family

members who might be at risk of DVT or PE in high-risk situations. This is known as **Case-Finding**. The benefit of case-finding has not yet been proven and any close relative of someone who has suffered a DVT or a PE should inform their medical attendants of this family history of DVT or PE regardless of whether or not they have had thrombophilia tests.

Should children be tested?

As a general rule young children should not be tested. Children have special defences against forming blood clots and it is not until they reach puberty that their risk of blood clots due to thrombophilia begins to increase. Teenage daughters of patients with thrombophilia can be considered for testing if the results would influence decisions relating to contraceptive use.

SPECIFIC ACTIONS

What should I do if I need an operation?

If you are to have a planned operation tell the doctor that you have thrombophilia when you are reviewed in clinic before the operation. Tell the doctors and nurses again when you are admitted for your operation.

What if I need to take a long journey?

Keep well hydrated and exercise your legs during the journey. There is some evidence that support stockings may be beneficial. Aspirin may be recommended by your doctor. Very occasionally, some individuals at very high risk may be advised to take an injection of heparin, or some other medication. Ask your doctor for advice.

FEMALES - Are Oral Contraceptive Pills safe?

Oral contraceptive pills containing oestrogen are associated with a very small risk of DVT and PE. Each year about 20 to 30 women in every 100,000 pills users will suffer a DVT or a PE. If you have had a DVT or a PE your risk of another blood clot if you take one of these pills will probably be much higher. Therefore, you should not take one of these pills unless you have discussed the risks and the reasons for taking it in detail with your doctor. There are other types of pills, which appear safer from this point of view, for example progestogen only pills (*known as the mini-pill*).

If a female has a close family relative who has suffered a DVT or a PE they should also discuss the risk of taking an oestrogen containing pill with their doctor.

FEMALES- Is Hormone Replacement Therapy Safe?

The same type of precautions should be taken as for oral contraceptive pills (see above). The important thing is to discuss the issue with your doctor.

FEMALES- Is pregnancy safe?

Pregnancy is safe in the majority of women with thrombophilia. Ask your doctor for advice and an explanation of what is likely to be required before you become pregnant and inform your doctor as soon as you know you are pregnant. Remember that you should avoid becoming pregnant if you are taking warfarin. If you do become pregnant whilst taking warfarin inform your doctor immediately.