Outpatient anticoagulant service

Thrombophilia

This leaflet is for patients and relatives who may be having testing or an assessment to see if they have Thrombophilia.

What is Thrombophilia?

It is essential that blood remains fluid within the circulation but clots at the site of an injury. Blood should not usually clot within a blood vessel. Thrombosis is the name given to a blood clot within a blood vessel, and Thrombophilia is the name given for an increased tendency for blood to clot within a blood vessel.

Deep vein thrombosis (DVT) is the name given to blood clots forming within the deep veins. The most common site for a DVT is the deep veins in the legs. When a DVT forms, pieces of the clot can break away and pass through the circulation and lodge in the blood vessels in the lungs. These vessels are known as pulmonary arteries. This type of clot is termed a pulmonary embolus (PE).

Deep vein thrombosis (DVT) and pulmonary embolus (PE) are collectively known as venous thrombosis (VT) or venous thromboembolism (VTE).

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.

Thrombophilia testing should not be offered to patients who are continuing anticoagulation, or if their DVT or PE was provoked. For patients who have had a DVT or a PE which is unprovoked their doctor may suggest having blood tests after receiving treatment for the event.

Types of Thrombophilia blood tests

Some people may have an underlying genetic predisposition to DVT and PE. The purpose of these blood tests are to see if there is an imbalance in clotting chemicals, too much clotting factor or too little substance that prevents clots. If the results indicate that they have a form of thrombophilia it may partly explain why they had a blood clot. These tests are offered to evaluate the blood for its ability to clot, and to evaluate how likely the patient is to have a bleeding or clotting problem.

There are many different types of thrombophilia. Within your circulation you have natural anticoagulants and some people may have a reduced level of these natural anticoagulants, and this may increase their risk of a DVT or PE. The 3 main anticoagulants are;

- Antithrombin
- Protein C
- Protein S

A variation within an individual's genetic code known as a mutation is much more common than a deficiency of a natural anticoagulant. Some mutations are present in
about one in 30 people, and may also increase the risk of DVT and PE, but to a lesser
degree than a 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').

D-Dimer - This is a blood test to look for a substance called D-dimer. It is a fibrin
degradation product (or FDP) from the breakdown of a blood clot. It is used to help
further risk stratify a patient's risk of recurrence of DVT or PE, after they have received
their initial anticoagulant therapy.

Antiphospholipid antibodies (APS) are an autoimmune, hypercoagulable state caused by
Antiphospholipid antibodies. APS can provoke blood clots. After completing treatment for an
unprovoked VTE testing for Antiphospholipid antibodies may be recommended, as their
presence may favour restarting anticoagulation.

Anticardiolipin antibodies (ACA) are antibodies often directed against cardiolipin and
found in several diseases, including the Antiphospholipid syndrome. After an unprovoked
VTE testing for Antiphospholipid antibodies may be recommended as their presence may
favour restarting anticoagulation.

Specific actions if you have confirmed Thrombophilia

What special precautions

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.
- Pregnancy and the post-partum period.

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.
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.

Should children be tested?

As a general rule young children should not be tested. Children have special defences against forming blood clots. 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.

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 pill 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 is likely to 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 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. Inform your doctor as soon as you know you are pregnant.

References

- NICE clinical guideline 144. Venous thromboembolic diseases: the management of venous thromboembolic diseases and the role of thrombophilia testing. Issued June 2012.
If you have any questions, please ask the healthcare team who will be happy to discuss this with you. For further advice please contact:

**Addenbrooke’s outpatient anticoagulant service team**

**Addenbrooke’s Hospital / Box 217**  
**Hills Road**  
**Cambridge CB2 0QQ**  
**Tel 01223 217127**  
**Open: Monday to Thursday 0900 to 17.00**

We are now a smoke-free site: smoking will not be allowed anywhere on the hospital site. For advice and support in quitting, contact your GP or the free NHS stop smoking helpline on 0800 169 0 169.

**Other formats:**

If you would like this information in another language, large print or audio, please ask the department where you are being treated, to contact the patient information team: patient.information@addenbrookes.nhs.uk.

Please note: We do not currently hold many leaflets in other languages; written translation requests are funded and agreed by the department who has authored the leaflet.

**Document history**

Authors: Caroline Baglin – Consultant Nurse, Trevor Baglin – Consultant Haematologist  
Department: Cambridge University Hospitals NHS Foundation Trust, Hills Road, Cambridge, CB2 0QQ [www.cuh.org.uk](http://www.cuh.org.uk)  
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