Clotting disorders

Blood is meant to clot. If you nick yourself, blood seeps out and then clots.

But it is only meant to clot outside the body. If it clots inside the body, there is a problem.

If blood clots in a vein in the calf, for example, then the person affected might get a sore swollen leg. This is known as a deep venous thrombosis or DVT.

If the clot stays where it is, it can feel uncomfortable. But if parts of the clot break off, they can travel through the bloodstream to other parts of the body, such as the lung or the brain. This condition, known as venous thromboembolism, can be quite dangerous.

Most people with DVT and/or venous thromboembolism do not have a genetic alteration that caused it.

But some do. Some have an altered gene which affects normal clotting. People with this gene alteration are said to have hereditary thrombophilia.

There are many different types of hereditary thrombophilia, all caused by different gene alterations, all of which affect people in slightly different ways.

Hereditary thrombophilias are passed on through an autosomal dominant pattern of inheritance (see fact sheet on '*How do genetic conditions occur?*'). This means they are likely to be present in other members of the family.

It also means that any child of an affected person has a 1 in 2 chance of having the gene alteration and a 1 in 2 chance of not having it.

In general, these gene alterations don't cause blood clots on their own, but give people a high likelihood of getting blood clots.

Other indicators

People with hereditary thrombophilia are more likely to develop clots if they:

- Are overweight
- Stay in bed for 10 days or more
- Have surgery or a serious injury
- Are pregnant or have just given birth
- Smoke
- Take the oral contraceptive pill or hormone replacement therapy
- Have cancer

People with hereditary thrombophilia are more likely to develop clots as they get older.

Contraception and pregnancy

Oral contraceptive pills containing oestrogen increase the risk that a woman with hereditary thrombophilia will develop a clot. Women with hereditary thrombophilia should talk to their doctor about whether or not other forms of contraception may be better.

Any woman with hereditary thrombophilia who becomes pregnant is at higher risk than usual of developing a clot.

Any woman with hereditary thrombophilia also has a slightly higher than usual risk of having problems with the pregnancy, such as miscarriage, poor growth of the baby and problems with the placenta. However, most women with hereditary thrombophilia who become pregnant still have normal pregnancies.

Women with hereditary thrombophilia who become pregnant should see a specialist obstetrician for advice.

What to do?

People should be assessed to see whether they may have a hereditary thrombophilia if they have a venous thrombosis or thromboembolism:

- Before the age of 50
- With nothing obvious that could have caused it
- And others in the family have had the same problem
- In an unusual position, such as the brain, the abdomen or the arm.

People with a close relative who has been shown to have a hereditary thrombophilia, or who has had a thrombosis or thromboembolism, should ask their doctor about testing for a hereditary thrombophilia.

Testing

Tests are available to detect the common forms of hereditary thrombophilia.

Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Australasian Genetic Alliance at **http://www.australasiangeneticalliance.org.au**
- Better Health Channel at **http://www.betterhealth.vic.gov.au**
- The Centre for Genetics Education at **http://www.genetics.edu.au**
- HealthInsite at **http://www.healthinsite.com**
- MedicineNet at **http://www.medicinenet.com**
- MyDr at **http://www.mydr.com.au**
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email **gtis-australia@unimelb.edu.au** or visit Biotechnology Australia's website at **http://www.biotechnology.gov.au**