Accurate data on the risk of recurrence in patients with hereditary thrombophilia is unavailable (HTTF and BCSH, 2001), for the wider population, it is around 15-20% within two years (Prandoni et al, 1996).

There is no evidence to support the permanent use of anticoagulants in hereditary thrombophilia, as the associated risk of haemorrhage is too high (HTTF and BCSH, 2001). However, patients should be warned about the risk of thrombosis recurring and the signs and symptoms to look out for, and be advised to seek medical attention if they suspect they have a blood clot (HTTF and BCSH, 2001).

**Side-effects**

Treatment with anticoagulants can have a number of side-effects, including haemorrhaging. This can occur more frequently when patients first start taking anticoagulants, and has been linked to the fact that dosages are still being determined (Pal-areti et al, 1996).

Evidence suggests that side-effects may lead to patients not adhering to treatment regimens, so it is important to check that they are concordant with treatment before entering high-risk situations such as having surgery.

**Prophylaxis**

Guidance on reducing the risk of thromboembolism says patients admitted to hospital who have an increased risk of thrombosis should be offered prophylactic treatment. This includes those with thrombophilia.

The guidance also recommends encouraging patients to move as soon as possible after surgery to reduce the risk of thrombosis developing (NICE, 2010).

**Patient understanding**

Saukkko et al (2007) found that some thrombophilia patients tested for the factor V Leiden mutation were unaware they had undergone a genetic test. The researchers also found that some of those who tested positive for the mutation were only given the test result by telephone, with no further “advice and clarification”.

It is therefore important not to assume patients are aware that a genetic test to look for mutations linked to hereditary thrombophilia has been carried out. They may have only received limited information about their condition and will rely on healthcare professionals to help reduce their risk.

Patients with hereditary thrombophilia should have their increased risk of developing thrombosis explained, as well as the lifestyle factors that can increase risk, such as oral contraceptive use, being overweight and taking long flights or sitting for long periods. Guidelines recommend movement on long journeys to reduce the risk of thrombosis, and those taking journeys of more than eight hours should wear compression stockings (Watson and Baglin, 2010). Patients who have more than one risk factor for thrombosis should wear compression stockings for journeys of three hours or more (Watson and Baglin, 2010). Those with cancer should be offered anticoagulation treatment (NICE, 2010).

As the condition is inherited, patients may also need support in telling relatives about their diagnosis.

**Conclusion**

Hereditary thrombophilia is a common cause of thrombosis and is diagnosed by genetic testing. It can be caused by one of a number of genetic mutations which can change the proteins involved in the blood coagulation cascade. This leads to an increased risk of inappropriate blood clotting and thrombophilia.

Risk factors include surgery and/or trauma, immobility, advancing age, pregnancy and the use of combined oral contraceptives.

Management of thrombosis involves the use of anticoagulants, but these are time-limited to reduce the risk of inappropriate bleeding.

Evidence suggests patients may not be aware or understand that they have undergone a genetic test for the condition, and may have received limited information about hereditary thrombophilia.

Nurses may see patients who have been admitted to hospital as a result of thrombosis, or after their diagnosis of thrombophilia when treatment becomes preventative rather than life-saving.

Nurses may assess whether patients should be offered a referral for testing based on their own or their family’s history of thrombosis.

Patients may be unsure what the diagnosis of thrombophilia means and therefore need advice and reassurance about the condition. They may need help to understand the need for treatment – particularly if they experience unpleasant side-effects, or information on lifestyle factors that could increase their risk of thrombosis. They may also have ongoing treatment needs, for example, learning about their options for thromboprophylaxis in high-risk situations such as long haul flights or immobility after surgery.

**Patients may also need support in working out who in their family may be affected by hereditary thrombophilia. Nurses can therefore play an important role in providing advice and support to patients.**

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<tr>
<th>Table 1. Increase in risk of hereditary thrombophilia from gene mutations</th>
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<tbody>
<tr>
<td>Protein</td>
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<tr>
<td>Factor V Leiden</td>
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<tr>
<td>80% for homozygote</td>
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<tr>
<td>PTG2020A</td>
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<tr>
<td>Antithrombin</td>
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**References**


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