

## Application for Heritable Thrombophilia Testing

### Drawing of samples

Please do not draw any bloods until the testing has been approved.

### Indications for heritable thrombophilia testing

Purpura fulminans in neonates or children – test for Protein C and S deficiency.

Young patients (<40 years) with unprovoked venous thromboembolic event (VTE) and/or with a strong family history (1<sup>st</sup> degree relatives) of **unprovoked** VTE.

Pregnant women, or women planning a pregnancy, who have a first degree relative with unprovoked or oestrogen-associated VTE.

Splanchnic vein thrombosis but only if discontinuation of anticoagulation after the initial short-term anticoagulant treatment is being considered.

### Not indicated

1. Provoked VTE.
2. Arterial thrombosis – lupus anticoagulant and anticardiolipin may be indicated depending on clinical context.
3. Assisted conception, ovarian hyperstimulation syndrome, pregnancy morbidity, recurrent miscarriages.
4. Prevention of thrombosis in women considering oestrogen-containing hormone oral contraception.
5. Asymptomatic relative of patients with low-risk thrombophilic disorders (Factor V Leiden and Prothrombin gene variant).
6. Retinal vein thrombosis.

### Timing of sample collection

Samples are not to be collected during acute VTE episode, while on anticoagulation or within two weeks of stopping warfarin or within 3 days of stopping a non-warfarin anticoagulant.

### Genetic testing

Note that testing may reveal genetic variants that can have implications for family members – therefore pretest counselling is important.

### Once approved

The patient will require a laboratory request form for approved tests.

Note: Lupus anticoagulant, anti-cardiolipin, anti-beta-2-microglobulin, JAK2 mutation and PNH testing may be appropriate in some patients. If sufficient clinical information is provided, the laboratory haematologist may provide advice about non-heritable thrombophilia testing.

**Application for Heritable Thrombophilia Testing (for venous thromboembolism)**

 Please email the request form to the haematology laboratory: [dnhaem@awanuilabs.co.nz](mailto:dnhaem@awanuilabs.co.nz)

<b>Date of application:</b>	<b>NHI:</b>
<b>Patient's surname:</b>	<b>First name:</b>
<b>DOB:</b>	
<b>Requesting doctor's name and practice:</b>	
<b>The patient has been counselled about the genetic and clinical implications of testing:</b>	
	Yes / No
<b>Reason for referral; i.e., how will the result alter patient management?</b>	
<b>Personal history:</b> Has the patient ever had a deep vein thrombosis (DVT) pulmonary embolus (PE) or other VTE?	
	Yes / No
If yes, how many, and at what ages did the VTE occur?	
Did the VTE occur during pregnancy, while on the combined contraceptive pill, after surgery, trauma, or following a period of immobility? If yes, please give details.	
	Yes / No
Please list medications they are currently taking, especially anticoagulants and oral contraceptive pill.	

**Family history:** If any first-degree relative was ever diagnosed with a DVT or PE, please provide details (relationship to applicant, associated surgery, immobility or other risk factors, site of VTE, age).

Did the family members' DVT or PE occur during pregnancy, oestrogen use? Provide details.

Is there an inherited genetic variant in their family that causes thrombosis? If yes, which gene is affected (Protein C, Protein S, Antithrombin, Factor V Leiden, or Prothrombin)?

**LABORATORY HAEMATOLOGIST USE ONLY:**
**Approved / Declined – \_\_\_\_\_ Date: \_\_\_\_\_**

Protein C, Protein S, Antithrombin, Factor V Leiden, Prothrombin variant.

Note: if appropriate the laboratory haematologist may provide advice about non-heritable thrombophilia testing.