GUIDELINE FOR HERITABLE THROMBOPHILIA TESTING[1]

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REVISION DESCRIPTION
Section 5.0 Laboratory requests – Amendment of link reference for Thrombophilia screen/Lupus anticoagulant request and Patient Consent Form, for in-house requestors.

1.0 Introduction

Heritable thrombophilia describes an inherited tendency for venous thrombosis (deep vein thrombosis (DVT) with or without associated pulmonary embolus (PE)). Testing for heritable thrombophilia typically does not predict likelihood of recurrence in unselected patients with symptomatic venous thrombosis.

Testing for heritable Thrombophilia **should only be performed** if the results are likely to influence management.

Testing **should not be performed** in the acute setting of a thromboembolic event or while on anticoagulant therapy.

2.0 Purpose/Scope

The aim of this guideline is to provide recommendations to clinicians in relation to testing for heritable Thrombophilia in context of clinical management of venous thrombosis and pregnancy morbidity.

3.0 Definitions

Heritable thrombophilia testing may include the following assays:

- Prothrombin Time
- Activated partial thromboplastin time
- Antithrombin
- Protein C
- Free Protein S
- Fibrinogen
- Activated Protein C Resistance
- Thrombin Time
- Genetic testing for Factor V Leiden gene mutation
- Genetic testing for the Prothrombin gene mutation

4.0 Most frequent indications for heritable thrombophilia testing and situations in which testing should not be performed

4.1 Testing for heritable thrombophilias in the following situations:

- Testing for heritable thrombophilias may be considered in **selected patients**, such as those with a strong family history of unprovoked recurrent thrombosis
- Case finding of asymptomatic relatives with high risk thrombophilia, such as deficiency of antithrombin, protein C or protein S, should only be considered in selected thrombosis-prone families. If testing is performed, the risks, benefits and limitations of testing should be discussed
- It is suggested that adults who develop skin necrosis in association with oral vitamin K antagonists (VKAs) are tested for protein C and S deficiency after VKA treatment is withdrawn.

4.2 Testing for heritable thrombophilias is <u>NOT INDICATED</u> in the following:

- Unselected patients presenting with a first episode of venous thrombosis.
- Patients with arterial thrombosis
- Patients with retinal vein occlusion.
- Case finding of asymptomatic relatives with low risk thrombophilia, such as Factor V Leiden or the prothrombin gene mutation.
- Asymptomatic women before assisted conception and those with ovarian hyperstimulation syndrome.
- Unselected patients with upper limb venous thrombosis
- Patients with central venous catheter (CVC)-related thrombosis

Please note:

For women considering hormonal therapy if a first-degree relative with venous thrombosis has been tested and the result is positive then suggest consideration of an alternative contraceptive or transdermal HRT before offering testing as a negative test result does not exclude an increased risk of venous thrombosis. Testing for heritable thrombophilia may assist counselling of selected women particularly if a high risk thrombophilia has been identified in the symptomatic relative.

4.3 Specific pregnancy-related consideration:

- Women should be assessed for risk of pregnancy-associated venous thrombosis primarily in relation to clinical risk factors. Referral to a haematologist with expertise in management of obstetric patients is recommended
- In the asymptomatic pregnant woman with a family history of venous thrombosis, testing is not required if the clinical risks alone are sufficient to result in thromboprophylaxis. Testing may be considered in selected cases, for example where there is a family history of high risk thrombophilia or multiple family members with unprovoked recurrent events. Referral to a haematologist with expertise in management of obstetric patients is recommended.
- Antithrombotic therapy should not be given to pregnant women with a history of pregnancy complications based on testing for heritable thrombophilia.

If uncertainty exists in relation to whether a patient should be tested, please contact the haematology team.

5.0 Laboratory Requests

All Thrombophilia screen requests **MUST** be accompanied by a **FULLY COMPLETED** "Thrombophilia screen/Lupus anticoagulant request and patient consent form" which can be found:

For in house requestors:

https://maternet.mmuh.ie/departments-and-offices/pathology/Thrombophilia-screen-Lupus-anticoagulant-request.pdf

For external requestors:

http://www.mater.ie/healthcare-professionals/gpreferrals/Guideline_for_heritable_thrombophilia_testing.pdf

This form **MUST** contain the following information:

- Patient Name, MRN (where applicable), DOB, Requesting source.
- Clinical indication for testing
- Details of any anticoagulant therapy
- Name (in BLOCK capitals) & signature of requesting Doctor.
- Indication that written consent has been obtained for genetic testing.

A "Patient Information Leaflet for Genetic Testing for Thrombophilia is also available:

For in house requestors:
https://maternet.mmuh.ie/departments-and-offices/pathology/Patient-information-Genetic-testing-for-Thrombophilia.pdf

For external requestors:
http://www.mater.ie/healthcare-professionals/gp-referrals/Genetic-testing-for-Thrombophilia_information-leaflet.pdf

In the event that the above criteria are not met the requesting clinician will be notified and samples will be held for a maximum of 8 weeks pending additional data.

6.0 References:

Clinical guidelines for testing for heritable Thrombophilia: Baglin et al; British Journal of Haematology (2010), 149, 209-220. www.BCSHguidelines.com