

Guidelines for Thrombophilia Testing

Indications for Testing

In most occasions of thrombosis, thrombophilia screening will not be indicated (these tests are poor at determining risk of recurrence of VTE and do not alter clinical management).

Generally, if anti-coagulant or clinical management will change because of the test result from the thrombophilia screen, then testing may well be indicated.

A limited screen for acquired thrombophilia is more likely to have impact on management when tested in certain circumstances below (this includes PNH – paroxysmal nocturnal haemoglobinuria, MPN panel – myeloproliferative neoplasm and APL – anti-phospholipid testing)

- *Thrombosis at unusual sites*
 - *Splanchnic or CSVT – MPN, APL*
 - *Abnormal FBC – PNH, MPN*
- *Unprovoked VTE – APL*
- *Recurrent thrombosis on anticoagulation – PNH, APL, MPN*
- *Selective testing of 1st degree relatives with protein C, S and Anti-thrombin – [see guideline](#)*
- *Retinal vein occlusion with no risk factors – APL*
- *Arterial thrombosis*
 - *With no risk factors – APL*
 - *With abnormal FBC – MPN, PNH*
- *CVA*
 - *Less than 50 with no risk factors – APL*
 - *With abnormal FBC – PNH, MPN*
- *Purpura fulminans or warfarin induced skin necrosis – protein C, S*
- *Neonates with multiple unexplained thrombosis – APL, heritable thrombophilia screen*
- *Pregnancy associated thrombosis*
 - *Anti-thrombin testing if family history of anti-thrombin deficiency or heparin resistance*
 - *APL, test when not pregnant*
 - *Recurrent or late pregnancy loss - APL*

When to test?

- *Avoid testing in the acute post-thrombotic period - delay testing for 3 months*
Wherever possible avoid testing while on anticoagulant drugs (Warfarin, UFH, DOAC) - delay testing.

PNH, MPN panel, anti-cardiolipin and anti B2-glycoprotein may be tested on anticoagulation where possible in primary care

Avoid testing during pregnancy, delay testing for 2 months.

If in doubt please discuss requests for thrombophilia screening with a Consultant Haematologist or Senior BMS.

References

NICE Guidelines

<http://www.nice.org.uk/guidance/cg144>

RCOG Clinical Guidelines

https://www.rcog.org.uk/globalassets/documents/guidelines/gtg_17.pdf

<https://www.rcog.org.uk/globalassets/documents/guidelines/gtg-37a.pdf>

Clinical Guidelines for Testing for Heritable Thrombophilia (BJH, 2010)

<http://onlinelibrary.wiley.com/doi/10.1111/j.1365-2141.2009.08022.x/pdf>

[Thrombophilia testing: A British Society for Haematology guideline 2022](#)