Scarborough, Hull and York Pathology Service

Guidelines for Thrombophilia Testing

Indications for Testing

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

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 in most cases).

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
- Routine testing of 1st degree relatives is not indicated, however in very select cases it may be indicated where it would alter future management, for example for women of childbearing age considering pregnancy, oral contraception, or HRT.
- 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
  - 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 can be tested on anticoagulation.

- Avoid testing during pregnancy or during the post-natal period - delay testing for 2 months

Laboratory considerations

- Repeat any abnormal or borderline test: at least 2 abnormal results before issuing final diagnosis of proven thrombophilia – *could now consider genetic test if there is valid indication for test in first place i.e. will it affect clinical management*

- Referrals to a consultant Haematologist or incomplete clinical details: ensure sufficient aliquots of plasma are frozen and retain the primary sample tube(s) for testing if subsequently indicated

1. Rejected Samples

- Request a PROBHA set in Telepath and add the coded comments TBR1 – TBR4, i.e.

"Inappropriate for full thrombophilia screen. Happy to discuss *Dr. A Bachh*"

"Samples will be stored for 6 weeks - please contact lab with full clinical details if testing required"

References

Thrombophilia testing: A British Society for Haematology guideline (BJH, 2022)
Thrombophilia testing: A British Society for Haematology guideline (wiley.com)