Thrombophilia Screening in Venous Thrombosis-Is It Worth It?

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Objectives

After attending this presentation the participants shall be able to:

- discuss why thrombophilia testing does not alter the clinical management of the majority of patients with DVT or PE
- list three special clinical scenarios in which thrombophilia testing may alter the clinical management of patients
Classic Indications for Thrombophilia Screening for Venous Thrombosis

- **Strongly thrombophilic**
  - Idiopathic thrombosis before age 50 OR
  - History of recurrent thrombosis OR
  - First degree relatives with documented thromboembolism before age 50 OR
  - Thrombosis in an unusual site

- **Weakly thrombophilic**
  - Idiopathic thrombosis after age 50 AND
  - Negative family history of thromboembolism
Thrombophilia Screening

- Protein C
- Protein S
- Antithrombin
- Factor V Leiden
- Prothrombin G20210A
- Lupus Anticoagulant
- Phospholipid Antibodies
- Resistance to activated protein C
- Factor VIII
- (homocysteine)
Does Thrombophilia Testing Change Clinical Management of Patients?

In most cases the answer to this question is no.

- **Idiopathic first time venous thrombosis** - minimum of 6 months anticoagulation - consider long-term anticoagulation regardless of thrombophilia status.

- **Recurrent venous thrombosis** - patients should receive long-term anticoagulation.

- **Venous thrombosis with a transient risk factor** - anticoagulation for 3 to 6 months.
Remember Venous Thrombosis Is a Multifactorial Disorder

- Good history and physical assessment is important
  - Transient risk factors, family history
- A number of systemic medical conditions can also contribute to risk for thrombosis
- Remember that the strongest risk factor for recurrent thrombosis is a prior history of thrombosis
Medical Conditions To Consider

- Malignancy
- Hyperviscosity
- Hyperleukocytosis
- Sickle Cell Disease
- Myeloproliferative disorders
- Paroxysmal nocturnal hemoglobinuria
- Nephrotic syndrome
- Inflammatory Bowel Disease
- Drugs that induce phospholipid antibodies
Other Considerations in Thrombophilia Screening

**Cost of screening**

- Is the cost worth it if patient management doesn’t change?
- Evidence that thrombophilia screening did not change patient management in the vast majority of cases.
Other Considerations in Thrombophilia Screening - cont’d

- Other risk factors may better predict recurrence rates than those that are routinely tested in a thrombophilia workup
  - D-dimer$^2$
  - Evidence that hereditary thrombophilia does not predict rate of recurrence well$^3$

- Over-labeling/Psychological Impact
Other Considerations in Thrombophilia Screening- cont’d

- Timing of the thrombophilia testing impacts the quality of the results
  - One study in a tertiary care center demonstrates that the majority of testing occurs in the setting of acute thrombosis or pregnancy which can alter results:\(^1\)
    - 200 patients reviewed- 124 panels done during acute thrombotic event
    - 46 panels had low Protein C and S results- most attributable to warfarin and the remainder to pregnancy
    - 46 panels had a significant abnormality (23 lupus anticoagulant)
When Does Thrombophilia Screening Impact Patient Management?

- Those with a strong family history of DVT/PE
  - Identification of female relatives at risk
    - Pregnancy, OCP
- Testing for phospholipid antibodies in patients with venous/arterial thrombosis and/or defined pregnancy complications
- Those with thrombosis in an unusual site
  - But remember the list of medical conditions!
Conclusions

- Thrombophilia screening should be offered to a select group of patients, based on whether the results will impact patient management.
- Select patients may only need select laboratory tests.
- Need to consider other medical causes and rule them out.
- The need to know is not enough of a reason to do thrombophilia testing.
References

2. Palareti G et al. Thrombosis and Haemostasis 2002; 87: 7-12