



# Testing for Thrombophilia



## What it covers

- Recommendations to guide testing for hereditary and/or acquired thrombophilia (a condition characterized as a tendency to form blood clots).
- The guidelines, developed using a case-based approach and modeling, are founded on three potential treatment decisions based on the outcome of testing:

Whether to continue or discontinue treatment for secondary prevention of VTE.

Whether to provide anticoagulant prophylaxis for prevention of VTE in patients with risk factors.

Whether to avoid hormonal treatments, such as oral contraceptive pills (OCP) or hormone replacement therapy (HRT).



## Why it matters

- Hereditary and acquired thrombophilia are blood coagulation disorders that increase the risk for venous thromboembolism (VTE) and affect approximately 10% of the population.
- Thrombophilia testing is costly and whether testing helps in guiding treatment decisions is controversial.
- However, lifelong anticoagulation is expensive as well, and testing to withhold treatment might be cost effective.



## Who it affects

- Clinicians and health care professionals including hematologists, internists, primary care physicians, obstetricians and gynecologists, clinical laboratory physicians, and emergency care physicians.
- Patients with VTE and individuals with a family history of thrombophilia and/or VTE.
- Women considering using combined oral contraceptives or hormone replacement therapy, women who are planning pregnancy, and patients with cancer who are classified to be at low or moderate risk of VTE.



## What are the highlights

- The ASH panel suggests limiting testing for thrombophilia to specific situations:

Patients with VTE provoked by non-surgical risk factors, including pregnancy, postpartum, or use of oral contraceptives.

Patients with VTE at unusual sites within the body, if the standard of care is to treat patients for three to six months.

Individuals with a family history of VTE and high-risk thrombophilia (antithrombin, protein C, or protein S deficiency) in order to determine if pharmacological thrombosis prophylaxis during transient thrombosis risk factors is necessary.

Women with a family history of VTE and high-risk thrombophilia (antithrombin, protein C, or protein S deficiency), in order to avoid use of hormone treatments, such as birth control pills.

Women with a family history of VTE and high-risk thrombophilia (antithrombin, protein C, or protein S deficiency), in order to determine if postpartum and/or antepartum prophylaxis is necessary.

Ambulatory cancer patients with a family history of VTE and who are otherwise at low or intermediate risk for VTE, to guide decisions on use of pharmacological thromboprophylaxis.

- In all other instances, the panel suggests not testing for thrombophilia.
- Nearly all recommendations are based on very low certainty in the evidence resulting in only one strong recommendation (against testing the general population before starting combined oral contraceptives).

**Total number of panel recommendations: 23**

### REFERENCE

Middeldorp S, Nieuwlaar R, Baumann Kreuziger L, et al. American Society of Hematology 2023 Guidelines for Management of Venous Thromboembolism: Thrombophilia Testing [published online ahead of print 17 May 2023]. *Blood Advances*. doi: <https://doi.org/10.1182/bloodadvances.2023010177>.

For more information on the ASH Clinical Practice Guidelines on Venous Thromboembolism, visit [www.hematology.org/VTEguidelines](http://www.hematology.org/VTEguidelines)

ASH guidelines are reviewed annually by expert work groups convened by ASH. Resources derived from guidelines that require updating are removed from the ASH website.

The American Society of Hematology (ASH) ([www.hematology.org](http://www.hematology.org)) is the world's largest professional society of hematologists dedicated to furthering the understanding, diagnosis, treatment, and prevention of disorders affecting the blood. For more than 60 years, the Society has led the development of hematology as a discipline by promoting research, patient care, education, training, and advocacy in hematology.

