

Testing for Inherited Hypercoagulable States

TEST RECOMMENDATIONS

Since the polymorphisms that predispose to thrombophilia are fairly common, it is possible that an individual will have more than one. For this reason, The Genomedical Connection™ recommends that an **initial workup consist of a panel of tests** for the six most common inherited risk factors to rule out the possibility of double heterozygosity.

INFORMED CONSENT

The College of American Pathologists recommends obtaining **oral consent** when testing for inherited thrombophilia with the appropriate notation in the patient's medical record. Specifically, patients should be informed:

- About the limitations of the tests
- That testing may have implications for family members

NAME OF DISORDER	TYPE OF TEST
Factor V Leiden	PCR-based DNA analysis OR "Second generation" APC resistance functional assay
Prothrombin G20210A	DNA analysis of the <i>F2</i> gene
Antithrombin III deficiency	Antithrombin amidolytic assay
Protein C deficiency	Protein C amidolytic assay
Protein S deficiency	Functional or immunoassay for free protein S
Hyperhomocysteinemia*	Plasma homocysteine level

*HYPERHOMOCYSTEINEMIA

Testing for the common polymorphism, **MTHFR C677T**, is **NOT** recommended as part of the work-up for inherited thrombophilia.

- Approximately 34%-37% of U.S. Caucasians are heterozygous for this polymorphism
- Approximately 12% of the this same population is homozygous for MTHFR C667T
- The best indicator of thrombotic risk is the level of homocysteine in the blood.

FOLLOW-UP FOR A NEGATIVE WORKUP

For individuals with a history of thromboses and a negative thrombophilia workup, a referral to a hematologist may be warranted to rule out rare causes of inherited thrombophilia.

DO NOT TEST

The American College of Medical Genetics and the College of American Pathologists have guidelines stating **thrombophilia testing should not be used for the following:**

- general population screening
- routine screening test during pregnancy
- routine screening test prior to the use of oral contraceptives
- prenatal or newborn testing
- testing of asymptomatic children
- routine initial testing in adults with an arterial thrombosis