

BENEFITS AND LIMITATIONS OF TESTING

COUNSELING WITH THE OPTION OF TESTING

Screening asymptomatic family members for an inherited thrombophilic disorder is controversial. Asymptomatic individuals should clearly understand the advantages and disadvantages of testing prior to making decisions about whether to pursue testing.

TESTING IS RECOMMENDED

In those instances where testing is recommended The College of American Pathologists also advises physicians to review the pros and cons associated with testing and to obtain oral consent prior to ordering the test.

BENEFITS AND LIMITATIONS OF TESTING

The benefits and limitations of testing for patients and their family members include:

Benefits

- Positive test result may encourage individuals to take steps in high-risk situations to lower their risk for a thrombotic event (i.e. avoid oral contraceptives with estrogen and stop smoking).
- Individuals can be monitored more closely, or potentially receive thromboprophylaxis when they are exposed to risk factors such as surgery or pregnancy.
- Individuals at increased risk can learn the signs and symptoms of thrombosis so they can seek treatment earlier.
- Negative test results may relieve feelings of anxiety that a patient might experience upon learning they are at risk to carry the polymorphism
- Testing may explain the cause of idiopathic VTE's or complications in pregnancy. This has both management and psychological benefits.
- Testing may be helpful in assessing the risk for other family members.

Limitations

- Inherited polymorphisms are low-penetrance risk factors for venous thrombosis. In other words, only a relatively low percentage of individuals with a predisposition to thrombophilia will develop clinical manifestations within their lifetime. Patients who test positive for polymorphisms may undergo undue psychological stress.
- There is a lack of standard treatment guidelines, even in high-risk situations.
- Other than avoiding environmental risk factors, there is nothing that asymptomatic individuals with an inherited predisposition for thrombophilia can do to reduce their risk for thrombosis.
- An individual who tests positive for a polymorphism could experience higher premiums for life, disability and long-term care insurance, loss of privacy, and/or psychological stress from worrying about these issues.
- Individuals who test negative for a polymorphism may have a false sense of reassurance. They may feel that their chance of developing a VTE has been eliminated and not recognize that their risk has simply been reduced to that of the general population.
- Non-paternity may be identified should the testing of family members be pursued.