

BENEFITS AND LIMITATIONS OF TESTING

COUNSELING WITH THE OPTION OF TESTING

If counseling with the option of testing is recommended, patients should be counseled about the [benefits and limitations of testing](#) and encouraged to make a decision regarding whether they would like to pursue testing. Individuals in this category may wish to pursue testing if it will affect their medical management. Examples may include women who are contemplating pregnancy or using oral contraception, individuals who are being treated for a malignancy, or people who require surgery.

[Benefits and limitations of testing](#)

The benefits and limitations of testing should be covered when counseling patients about the option of genetic testing. Patients should be made aware of the implications testing may have for themselves and other family members.

Con of testing include:

- 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. This may lead to undo psychological stress in patients who test positive for polymorphisms.
- 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.
- Individuals who test positive for a polymorphism could potentially experience higher life, disability and long-term care insurance premiums, loss of privacy, and/or the psychological stress of 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 members be pursued.

Pros include:

- Testing may encourage individuals who know they carry a polymorphism 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 may have an increased awareness of the signs and symptoms of thrombosis so they can seek treatment earlier.
- If a person tests negative for an inherited risk factor, it may relieve feelings of anxiety they might have experienced upon learning they are at risk to carry the polymorphism
- Testing may help explain the cause of VTE's or complications with pregnancy for an individual, which has both management and psychological benefits.
- Testing may be helpful to assess the risk for other family members.

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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.