Family Testing for Clotting Disorders

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A genetic thrombophilia, or an inherited predisposition to develop blood clots, affects both you and your family members

This document is to provide information about:

- Informing relatives of inherited risks to develop a blood clot.
- Educating family members and understanding their reactions.
- Advantages and disadvantages of genetic testing for the hereditary thrombophilias.

UNDERSTANDING GENETIC RISKS

Genetic changes (also called mutations) in clotting factors, like factor V and factor II (prothrombin), are the most common, inherited, predisposing factors for blood clots; 5%-7% of Caucasians have factor V Leiden (a common change in the clotting factor V) and 2%-3% have a prothrombin mutation (a change in the clotting factor II). Other inherited thrombophilias include protein C deficiency, protein S deficiency, and antithrombin III deficiency. All of these conditions are inherited in an "autosomal dominant" pattern. Autosomal dominant inheritance means that only one gene mutation is required to have an increased risk, and there is a 50/50 chance that the mutation will be present in first-degree relatives (parent(s), brother(s), sister(s), and children). Inheritance is not dependent on gender. In almost all cases, this means that if you have tested positive for an inherited blood clotting disorder, at least one of your parents will also be positive. Other relatives, including siblings, children, aunts, uncles, and cousins, may also be at risk. Our fact sheet, "The Genetics of Thrombophilia," contains further information about the inheritance of clotting genes.

Having a genetic predisposition does not mean that you will definitely have a blood clot or related condition (which may include recurrent pregnancy losses). Abnormal clotting of blood is a complex condition, in which genetic risks interact with non-genetic risks. Non-genetic risks may include smoking, using hormones, recent trauma or surgery, a sedentary lifestyle, or recent immobility. A blood clot will only happen if a combination of these factors is present. In fact, most people (up to 90%) with an inherited predisposition (like factor V Leiden and the prothrombin mutation) will never develop a blood clot. However, it is particularly important for those with genetic predispositions to avoid controllable risk factors that may cause them to develop a blood clot.

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Telling Your Relatives about Genetic Risks in the Family

Genetic risks, by nature, affect not only you, but your family members as well. Genetics professionals often recommend that information about genetic risks be shared with other at-risk relatives. This may worry you, especially if your family does not openly communicate about health conditions. On the other hand, you may be anxious to contact your family members about their risk. In either case, it is important to think about your approach when contacting relatives and consider how they may react to the information you are presenting.

Contacting Relatives

Talking with family members

You may decide to talk with family members, in person or over the phone, about your genetic test results. If you find it intimidating to explain information about genetics or blood clotting disorders, it may be helpful to have some of the information available from NBCA on hand to help you with your explanation. Just remember, you will likely know a lot more than your family members. It may take time for them to understand this information, and it may be helpful to provide only basic details in your first conversation.

A Family Letter

For some people, it is easier to contact relatives through a letter, email, or text message. An advantage of these contact methods is that they allow you time to formulate your thoughts, versus having a phone or in person conversation where you may feel put on the spot. Writing a letter or sending an email or text message is also a good way to reach several people at one time. Also, your relatives may find it helpful to have the information in writing, so that they may process it at their own rate and have it available for future reference.

A sample letter is available on the NBCA website, which you can use as a model.

Emotional Reactions to Genetic Risk

It is likely that your various family members will have differing reactions to the news you have to share. Some may be relieved that you have found an explanation as to why you had a blood clot or related disorder. They may want to know about their own risks and if others in the family need genetic testing.

Others in your family may wish to avoid the subject altogether. They may not want to know about their genetic risks. Some may think that testing positive means that they will get sick. Genetic concepts and inheritance may be difficult to understand. Others may feel guilty or deny that a genetic risk is in the family. Education can help alleviate a number of these misconceptions.

Your Parents' Reactions

For some parents, learning that a genetic factor is related to a family illness may lead to feelings of guilt or grief. A parent may deny that the risk factor is genetic. A parent may

also assume that he or she must not have the genetic risk factor if he or she has not had a blood clot or related problem. Some parents will feel reluctant to have genetic testing, since a positive test may make them feel responsible for passing a genetic mutation on to a child.

Other parents may have an opposite response. A parent may be intrigued by the information you have learned and may be interested in testing to see if he or she has the risk factor, too. Sometimes, learning more about genetics can help make sense of the family history. For example, it may shed light on why another relative had a blood clot, multiple pregnancy losses, or other related conditions.

There is little you can do to control your parents' response to the information. If your parent responds with feelings of guilt or grief, it is important to emphasize that genetic factors are just one small piece of the puzzle—a blood clot did not result from the genetic mutation alone. It is also important to emphasize that we all carry gene mutations that can lead to disease, and no one has control over what we pass on to our children. Parents may also benefit from speaking with others facing a similar situation. This type of support is available through NBCA. Lastly, if your parents are curious to learn more, they may find resources on the NBCA website to be of interest.

Your Children: Where to Go from Here?

If you have an inherited thrombophilia, you may wonder if your children have inherited it from you. You may be concerned and wonder if your children should be tested.

In general, medical experts recommend that children make an independent, informed decision about testing. It is not routine for children, who are otherwise healthy, to have testing, unless they have been fully informed

of the pros and cons and can make an independent choice. It is also important to consider whether or not testing will influence a child's medical management. For example, testing may be more strongly encouraged for females when they are at an age where they may consider taking birth control pills, since this medication adds additional risk to the inherited risk. Testing is not as strongly indicated for boys because test results would not affect decisions about oral contraceptives or pregnancy.

Testing of other relatives.

There is no right or wrong answer about who should have testing in a family. Individual decisions should be made based on a person's situation and opinions.

There are general advantages and disadvantages of testing that may be considered (below). Your family members may want to consult this list when discussing testing with a health care provider, such as a genetic counselor.

Genetic testing may be an advantage in the following circumstances:

Testing may help women weigh the risks of estrogen-containing contraceptives.
Women who have an inherited blood clotting disorder have a greater risk of developing a blood clot while using estrogen (a hormone normally produced by the body, which may also be supplemented). Use of estrogen-containing birth control pills increases the chance that any woman will develop a blood clot by about 5-fold. For women with a thrombophilia, like factor V Leiden, the risk to develop a blood clot is about 32 times higher if they are taking estrogen-containing birth control pills. Clearly, thrombophilia interacts with estrogen in some way. Each woman's exact risk to develop a blood clot will vary. On average, approximately 1 out of 700 women who has factor V Leiden and uses estrogen-containing contraceptives will develop a blood clot each year. Genetic testing might be helpful for women who would choose not to use oral contraceptives due to this level of risk.

Testing may inform individuals about personal risk so they can make decisions about lifestyle changes that might lower risk.
Knowing that one has an inherited thrombophilia may encourage a person to make lifestyle changes to decrease his or her risk for a blood clot. These changes might include losing weight, exercising, quitting smoking, or leading a more active lifestyle.

Testing may encourage a relative, or his or her doctor, to take symptoms of a blood clot more seriously.
Blood clots are frequently misdiagnosed, as the symptoms are like those seen with other conditions (i.e., leg pain may be mistaken for a muscle strain). Awareness of increased risk for clotting may help your relative, and his or her doctor, to take these symptoms more seriously. This could help ensure early diagnosis and treatment of a blood clot.

Testing may affect health care management during pregnancy.
All women have an increased chance to develop a blood clot while they are pregnant. However, women with an inherited thrombophilia are at greater risk. Women with an inherited thrombophilia may also be more likely to have a pregnancy loss, stillbirth, or certain complications of pregnancy. If there is a history of thrombophilia in the family, testing may be beneficial so that a woman can make informed decisions about using blood thinning medication during her pregnancy.

Before surgery, knowing that a person has a genetic thrombophilia could influence whether a doctor prescribes blood thinning medication after surgery.

Also, the dose or length of blood thinning treatment may be influenced by a person's thrombophilia status.

Testing may provide peace of mind.

For family members at risk, knowing for certain whether they have inherited a thrombophilia may provide reassurance.

People may choose not to get testing for several reasons. These might include:

Genetic testing may not change medical management.

Many people who have hereditary thrombophilia will never develop a blood clot. Since use of blood thinners can cause serious and life-threatening bleeding, physicians very rarely put individuals who have not had a blood clot on blood thinning medication. Thus, even if an individual knows that he or she has an inherited clotting abnormality, no therapy for prevention is currently available. Because genetic testing often does not result in a change of medical management, some people feel that testing is not very necessary or helpful.

Testing positive may lead to feelings of guilt

Some parents may feel guilty that they may have passed on a hereditary thrombophilia to their children, and therefore may prefer not to have testing since this would reveal the affected parent. Other relatives may not want testing because they would feel guilty if they did not have a hereditary thrombophilia, particularly if their relative has been negatively affected as a result of having a genetic predisposition (this is called survivor guilt).

Concerns about insurance discrimination

There are federal laws to help protect people from health insurance and job discrimination. However, these laws have limitations, and it is possible that a person with hereditary thrombophilia may have to (a) pay higher health insurance premiums if on an individual insurance plan, and/or (b) pay higher life or disability insurance premiums, and/or (c) get denied life or disability insurance if they test positive.

Testing may be costly

Testing may or may not be covered by insurance, depending on the reason for testing and the individual's insurance plan.

Testing may raise questions about paternity

Testing could reveal a genetic pattern that cannot be explained by inheritance (for example: a person who has one copy of the prothrombin mutation could discover that neither parent is positive.) This may lead to questions about paternity.

Individuals may be concerned about labeling, infringement of privacy, and autonomy of children

There is some concern that testing children would lead to labeling. This could result in treating a child differently, due to concerns that he or she is at high-risk for illness. Making the choice to test a child who has never had a health problem may also raise concerns about privacy and autonomy. Testing a child may be an invasion of privacy and interferes with autonomy, since the child is not able to make an independent decision about testing.

RIGHT FOR SOME, NOT FOR OTHERS

There is no definite right or wrong answer when it comes to genetic testing. There are many complexities— understanding who is at risk, dealing with emotional responses to this information, and determining if there is benefit to genetic evaluation of relatives. It is important to discuss these issues with a knowledgeable health care provider, who can assist you in identifying at-risk relatives and make recommendations regarding thrombophilia testing in your family. Genetic counselors are health care professionals who have experience and expertise in helping families with these issues. To find a genetic counselor in your area, visit https://findageneticcounselor.nsgc.org/, which allows you to search for genetic counselors by city, state, or hospital.