

40. Family testing for genetic abnormalities

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Q1: "Is it important for all family members to be tested once you have discovered factor V Leiden in one member?"

A1: For some family members it may be prudent to get tested, for others not. There is no best approach whom to test and whom not to test. Individual decisions need to be made (see discussion below)".

Q2: "I was diagnosed with factor V Leiden about 4 years ago. I have tried very hard to get the rest of my family to get tested, with varying success. My father, then 74, asked his doctor to test him. The doctor replied, "if you haven't clotted by now, you never will", and refused to test him. Dad died this summer. Pulmonary embolism. I don't know that testing would have saved him, but maybe it would have given him a better chance."

A2: To be able to make a comment on whether factor V Leiden testing would have "given the patient a better chance" the circumstances of the clot would need to be known. Sometimes it is helpful in getting optimal DVT prophylaxis and prompt diagnostic evaluation for DVT or PE if the patient tells the physician that he/she has a thrombophilia, such as factor V Leiden. It serves as a red flag to the physician. However, optimal DVT prophylaxis and prompt diagnostic evaluation should, in the optimal world, be given and done, no matter whether a thrombophilia is present or not. However, my suspicion is that testing would unlikely have given the patient above a better chance or saved him (see discussion below).

Q3: "Personally, I have found testing gives peace of mind."

A3: Peace of mind of the patient is, in my opinion, a good reason for testing. In private health insurance systems (like the United States), in which the patient pays the insurance premium or the bill for the test, I think that he/she should get the test that he/she wants. However, in a nationalized health care system, testing at present has to be limited due to the cost of the tests and the limitation of financial resources.

Q4: "I have factor V Leiden. Should I get my children tested? If I do and when they get older, will the fact that they have factor V Leiden prevent them from getting health or life insurance? Maybe I shouldn't get my children tested if this could be a possibility."

A4: There is no right or wrong whether one should get them tested or not and, if one gets them tested, when. In most instances I do not recommend testing of children; I recommend considering testing girls when they are in their teenage years and consider taking oral contraceptives; they can then make an informed decision as to whether they want to take estrogen-containing oral contraceptives, progestin-only contraceptives, or use non-hormonal methods.. For boys I usually do not see a reason to test them. It always needs to be considered that, in the U.S. insurance system, individuals who have been found to have an inherited thrombophilic abnormality may have to (a) pay higher health insurance premiums if on an individual insurance plan, and (b) pay higher life insurance premiums or get denied life insurance. For details see [Q/A 41](#).

Q5: "I am hetero FVL plus hetero factor II 20210. Should I have my baby tested? What about insurance issues? Will she have preexisting condition issues in the future?"

A5: There is probably no medical need to test the baby at this point. She has a chance of 50 % to also be heterozygous for factor V Leiden, 50 % to be heterozygous for II 20210, and 25 % to be heterozygous for both (for inheritance patterns see [Q/A 12](#)). Knowing exactly whether she has one or both of the mutations would not make a treatment difference at this point. With our present knowledge about factor V Leiden and the prothrombin 20210 mutation, I would wait until the teenage years to consider testing a child if it is a girl. For insurance issues see A4 and [Q/A 41](#).

Q6: "I am 22 and had a DVT when I took the contraceptive pill. I was found to have heterozygous factor V Leiden and also heterozygous prothrombin 20210. Our coach now does not let me play on the college soccer team any more, even though I am not on coumadin® [=warfarin] any more. He says my risk of blood clots is too high. I wish I had never been tested"

A6: For some people genetic testing leads to the opposite of "peace of mind". Individual decisions as to whom to test are obviously necessary.

If a patient has been found to have factor V Leiden or another inherited clotting (= thrombophilic) disorder, there is no right or wrong answer as to which family members of the patient should also be tested. Since most people who have factor V Leiden or the prothrombin 20210 mutation will never develop a blood clot, and since coumadin can cause

serious and life-threatening bleeding, one hardly ever puts individuals who have not had a clot on prophylactic coumadin. Also, Aspirin does not seem to play any significant role in the prevention of venous thrombosis. Thus, even if an individual knows that he or she has an inherited clotting abnormality, no therapy for prevention of an unlikely future clot is available. However, knowledge of the genetic status may have other benefits.

Reasons to get tested

- A woman who has never had a blood clot, but who has been found to have factor V Leiden, may decide against estrogen-containing contraceptives, because she assesses a 1:700 risk of a blood clot per year as too high and therefore unacceptable. For this woman the genetic testing was important. Another woman may assess a 1:500 risk per year as very low and may decide to take estrogen-containing contraceptives. For this woman genetic testing is not important.
- Knowing that one has a thrombophilic abnormality, may encourage a person to try harder to make life-style changes, that may decrease the risk for venous thrombosis, i.e. loose weight, stop smoking, lead a less sedentary lifestyle.
- Being able to tell a physician or surgeon that one has a thrombophilic abnormality may (a) prompt the physician to take symptoms that could be caused by a clot more serious and (b) prompt a surgeon to give more thorough DVT prophylaxis in case of surgery. Mentioning the abnormality may serve as a "red flag" to the doctors.
- Knowing whether one has a thrombophilic abnormality or not may give a person "peace of mind". A recent study of individuals with factor V Leiden showed that the majority of individuals (88 %) were glad they knew their genetic status; 5 % were indifferent. However, it also noteworthy that 7 %, for whatever reasons, wished they did not know their genetic status. 1

Reasons to decide against getting tested

- Finding an abnormal test may not have any health care consequences.
- Physicians may give patients with abnormal results incorrect or questionable medical advice (for example: "if you have factor V Leiden you should never get pregnant again", or "if you have factor V Leiden you should not take contraceptives."
- Fear of having to pay higher health or life insurance premiums, or of having difficulties getting insurance.
- Cost of the test.
- If the testing reveals a genetic pattern that can not be explained by inheritance (for example: father is found to be homozygous for factor V Leiden and child does not have factor V Leiden; see [Q/A 12](#)), paternity may be questioned and emotional problems in the family may arise.

What I often do

As always, individual decisions need to be made. However, I often recommend that the following family members get tested:

- The female family members (daughters, sisters, and mother) of a patient with factor V Leiden or prothrombin 20210 mutation. Knowledge of their genetic status allows them to know their risk of venous thrombosis with use of contraceptives or hormone replacement therapy and allows them to make an informed decision. I recommend to wait testing girls until they get into their teenage years.
- If a patient is homozygous for factor V Leiden, I recommend that his/her sisters get tested, because postpartum DVT prophylaxis should be considered if they are homozygous for factor V Leiden.

I often tell patients that I do not see any strong reasons to test the following individuals:

- Male family members of patients with factor V Leiden or prothrombin 20210.

Reference:

- Hellman EA et al. Knowledge and educational needs of individuals with factor V Leiden mutation. J Thromb Haemost 2003;1:2335-9.