

44. Strange inheritance patterns

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Q: "I have factor V Leiden. It is my understanding that factor V Leiden is passed down from one, if not both of my parents. I am confident that they are both my biological parents, but they both tested negative for factor V Leiden. This is confusing."

A: This, indeed, appears confusing. There are several possibilities for such discrepant test results:

1. The test results are not correct (i.e. false-negative or false-positive test). No test system is perfect. Reasons for false-positive or false-negative tests are: (a) the patient's blood sample was confused with somebody else's sample; accidental mislabeling and mishandling of samples and clerical errors in the health care system are well known causes of incorrect results; (b) genetic variations in other parts of the factor V gene may lead to false-negative results.\
2. The physician misread or misinterpreted the test report;
3. The patient had an abnormal APC resistance test and the physician falsely assumed that abnormal APC resistance is identical to factor V Leiden. However, an abnormal APC-resistance can be due to other reasons than factor V Leiden, which are not inherited (see also [Q/A 31](#));
4. One or both of the patient's parents is/are not the biological parent(s);
5. The patient developed factor V Leiden as a spontaneous, new mutation (de novo). This is very unlikely, since it is exceedingly rare. It only occurs in approximately 1 in 1 billion people (see also [Q/A 12](#)).

In a situation of discrepant test results a patient should ask for a copy of his/her factor V Leiden original test report and check whether he/she really has what the doctor said he/she has. Copies of the parent's test reports should also be obtained and reviewed. The patient should make sure that a genetic test was performed and not an APC-resistance test. If, after review of the genetic test reports, a discrepancy remains, it may be worthwhile to repeat the genetic testing to confirm the results and rule-out false-negative or false-positive previous testing. The hematologist may also need to speak to the Genetics Lab director to discuss the discrepant findings and see whether there was a problem with the test.