

## SUNY Upstate Medical University

## Authorization for the Molecular Test for Factor V (Leiden and D2194G) and Prothrombin 20210G&gt;A

**1. What is Thrombosis?** Venous thrombosis is abnormal clotting of blood within the veins of the body. Normal clotting of blood occurs when blood components, called coagulation factors, are “turned on”. When adequate clotting has taken place, the coagulation factors must be “turned off”. Too much clotting can lead to the formation of clots (thrombosis) and blockage of blood vessels, and this in turn results in an increased risk of stroke or heart attack. Two of the coagulation factors that play an important role in normal regulation of blood clotting are Factor V and Prothrombin. At present two genetic variations at factor V and one at Prothrombin, that are associated with these coagulation factors, can be tested. They are the **Factor V Leiden and Factor V D2194G** mutations of factor V, and the **Prothrombin 20210G>A** gene mutation. These abnormal variants can fail to function normally to stop the blood from clotting thus resulting in an increased risk of venous thrombosis (presence of a blood clot in the circulatory system). In the case of the Factor V D2194G mutation, this increased risk primarily occurs in people who also carry a Factor V Leiden mutation. For this reason, we will only test for the Factor V D2194G mutation in individuals that are heterozygous for Factor V Leiden.

**2. What is the purpose of the test and what are its limitations?** There are two copies each of the factor V gene and the prothrombin gene in an individual’s cells. Approximately 1 in 10 people have one normal copy of the factor V gene and one copy of either of these genetic variants that can result in the production of abnormal factor V. Some individuals have two copies of the abnormal factor V Leiden gene. **Prothrombin 20210G>A** is less common, being found only in about 1% of the general population, and 18% of patients with a family history of venous thrombosis. It is possible to detect the presence or absence of these abnormal genes by molecular testing. The assays performed identify only the factor V Leiden, factor V D2194G and prothrombin 20210G>A abnormalities. Other abnormalities of the factor V gene, the prothrombin gene, or other risk factors associated with developing a thrombosis will not be detected with this testing. If these mutations are not found, it does not mean that the risk of developing venous thrombosis is not present. Venous thrombosis due to non-hereditary causes will not be detected by this testing. A positive result by itself should not be used as the sole criteria for diagnosis. Rare (less than 1% of the time) errors may occur, for example due to sample mix-ups, or due to technical errors such as rare genetic variants that mimic or mask the mutation being tested.

**3. What is required to perform the test?** You will be required to donate 10 ml blood. This is equal to about two teaspoons. In addition, you may be asked to provide information regarding your medical history. A correct history is critical for proper interpretation of the data.

**4. Is there a cost for the test?** This is a routine clinical laboratory test and the results from it may aid in diagnosis, so you or your health insurer will be billed for the procedure.

**5. What will happen to the DNA once the test is complete?** The only testing that will be performed on this sample is the test for factor V Leiden, factor V D2194G and/or prothrombin 20210G>A. Residual DNA may be stored indefinitely (this does not constitute DNA banking) to be used as a laboratory control, in which case all identifying information will be removed.

**6. How will I obtain results from the test?** The test result will be provided to your physician who will discuss it with you. Genetic counseling may also be appropriate as follow up. To the extent permitted by law, all of the records, findings, and results of this test are confidential and shall not be disclosed without your written consent specifically authorizing to whom such records, findings, and results are to be released.

If you have any questions about the test, which will be performed, you may wish to obtain genetic counseling prior to signing this form. You may also contact the Molecular Diagnostics Laboratory at ( 315) 464-6806.

\_\_\_\_\_  
Patient's name (**printed**)

\_\_\_\_\_  
Patient's Medical Record Number  
*(for office use only)*

**For the Patient:**

Please print the name, phone number, and address (if known) of all health care professionals, physicians (in addition to the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the Factor V and/or Prothrombin mutation test result. (Medical results cannot be sent to a patient, a patient's family member, or guardian.) Please print legibly.

Name and title	Address	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this testing. \_\_\_\_\_Factor V( Leiden and D2194G) \_\_\_\_\_Prothrombin G20210A

Date: \_\_\_\_\_

\_\_\_\_\_  
**Signature** of Patient

\_\_\_\_\_  
Name of Parent/Guardian

\_\_\_\_\_  
**Signature** of Parent/Guardian if patient is a minor

*As referring physician/health care professional, I understand the benefits and limitations of this clinical assay. I hereby attest to the fact that I have provided the patient or patient's guardian with the information contained above in compliance with the NYS Civil Rights Act, Section 79-L, have answered any questions fully, and have obtained a signed informed consent as appropriate. I request that the genetic test indicated above be performed.*

\_\_\_\_\_  
**Printed** name of Physician/ Health Care Professional

\_\_\_\_\_  
**Signature** of Physician/Health Care Professional