

## Factor V (Leiden) GenotypR™ INFORMED CONSENT

1. **What is Factor V Genotyping?** Factor V (Leiden) genotyping assay detects a mutation associated with more than 50% of all patients with familial or recurring venous thrombosis. Factor V (Leiden) genotyping is recommended for patients for whom a hypercoagulable disorder is suspected, including: family history for venous thrombosis, individual history of recurrent thrombosis, thrombosis during pregnancy or oral contraceptive use, thrombosis appearing at an atypical site in an otherwise healthy adult and thrombosis at age less than 45 years or pediatric stroke.

2. **What is the purpose of this test and what are its limitations?** Everyone has two copies of the Factor V (Leiden) gene. An individual may have two normal copies, two abnormal copies, or one normal and one abnormal. This test detects the presence of a specific genetic change (mutation) which has been shown to predispose people to develop deep vein thrombosis. Two abnormal copies have been found in 20% of all Caucasians with venous thrombosis. If this mutation is not found by the testing procedure, it does not mean that the risk of carrying or developing deep vein thrombosis is not present. It simply means that this specific mutation has not been found, although other mutations may be present. It is also possible that such a patient may have secondary deep vein thrombosis due to non-genetic causes that would not be detected by this test. The relative risk for a person with one or two copies of the mutation to develop deep vein thrombosis is 7.9 and 91, respectively.

3. **What is required to perform this test?** You will be asked to provide 5 mL of blood, which is equal to about one tablespoon. DNA will be extracted from this blood sample and tested. The only discomfort that you may feel is the stick of the needle in your arm. You may also experience a small bruise at the site of the needle puncture. You will also be asked to provide information regarding your medical history, which is necessary for proper interpretation of your test result. In the unlikely event that you should be injured in the course of being tested, your physician will provide any necessary medical care. However, you would be expected to bear the cost of such medical care. Compensation will not be provided in the event of any injury.

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

5. **What will happen to the DNA once the test is complete?** The original blood sample will be discarded at the end of the testing process or stored not more than 60 days. The DNA will be retained for a minimum of 6 months. In some circumstances, a patient's DNA may be used anonymously as a negative or positive control sample in future testing, but, in this circumstance, all identifiers will be removed prior to re-testing and the DNA sample and results obtained will remain anonymous.

I understand and agree that my DNA remaining after testing may be stored for up to 6 months should additional testing be required. *Please initial.*

6. **How will I obtain results from this test?** DNA testing and interpretation of results are complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results. The laboratory will not provide results directly to patients. Your physician may suggest genetic counseling prior to performing this test or if your results are abnormal. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

### Patient Attestation of Informed Consent:

My signature below indicates that I have received information about this test, **Factor V GenotypR™**, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

\_\_\_\_\_  
Patient Signature

\_\_\_\_\_  
Date

\_\_\_\_\_  
Signature of Parent/Guardian if Patient is a minor

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

### For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

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

\_\_\_\_\_  
Print Name of Physician/Health Care Professional