SUNY UPSTATE MEDICAL UNIVERSITY
Authorization for Genetic Test for Factor XI deficiency

1. **What is "Factor XI deficiency"?** Individuals with factor XI deficiency have increased risk of bleeding, particularly after surgery. This disorder is rare except among individuals of Ashkenazi ancestry. One study found that one in 190 Ashkenazi Jews living in Israel had two defective factor XI genes (i.e., were homozygous), and so about one in 7 to 8 would be expected to carry one defective factor XI gene (i.e., be heterozygous). Most homozygotes and 20-50% of heterozygotes have increased bleeding tendencies and occasionally significant bleeding.

2. **What is the purpose of the test and what are its limitations?** Everyone has two copies of the factor XI (F11) gene. An individual may have two normal copies, two abnormal copies, or one normal plus one abnormal copy. This test is designed to detect the presence of either the E117X or F283L mutations within the factor XI gene that have been found to account for 96% of the abnormal alleles seen in Ashkenazi Jews. This assay is not appropriate for investigation of Factor XI deficiency ourside of the Askenazi population. If these mutations are not found by the testing procedure, it does not mean that the risk of carrying or developing the above disorders is no longer present. It simply means that <u>these particular</u> mutations have not been found although other mutations could be present. There is no good correlation between which mutations are found and the severity of this disorder. This test can be used to identify whether or not an individual is a carrier of either of these mutations.

3. **What is required to perform the test?** You will be required to donate 10 ml of blood, which 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 to test for factor XI genetic variation. Residual DNA may be stored indefinitely (this does <u>not</u> 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 questions about the test that 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 useonly)*

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 XI test result. (Medical results cannot be sent to a patient, a patient's family member, or guardian.) Please print legibly.

Name of licensed physician	Address (if known)	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this factor XI testing.

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