UPSTATE
UNIVERSITY HOSPITAL

AUTHORIZATION FOR FRAGILE X SYNDROME GENETIC TESTING

Patient Name: _____ MR#: _____

Account #: _____ DOB: _____ Date: _____

Fragile X syndrome is the most common inherited form of intellectual disability. Individuals affected with fragile X have varying degrees of intellectual disability, and most affected males have characteristic physical and behavioral features. The *FMR1* gene that causes the syndrome is located on the X chromosome and is inherited in a X-linked pattern. Therefore, males with the gene abnormality are usually more severely affected than females. This also means that a woman who has the *FMR1* gene mutation is a carrier of the disorder and at risk of having an affected child. Fragile X syndrome nearly always results from an increase in the size of a repeated DNA segment in the *FMR1* gene. Whereby, individuals affected with fragile X syndrome, show a DNA expansion of greater than 200 repeats. In normal individuals, the DNA region repeats itself from 6 to 44 times. In individuals termed "premutation carriers" of fragile X, the DNA region has moderate expansions of about 55 to approximately 200 repeats.

Molecular test for Fragile X syndrome You will be required to donate 10 mL of blood, which is 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. 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.

Significance of the results The Molecular Diagnostics Laboratory performs an analysis that differentiates between normal, moderate and large size expansions of the *FMR1* gene for the diagnosis of an affected individual or for the detection of a premutation carrier. The significance of the results will depend on the patient's gender, as well as the size and methylation status of the repeated segment in the fragile X gene. A gray zone exists (of 45-54 repeats) within which the risk for subsequent expansion in offspring is difficult to predict. Premutation carriers do not generally exhibit the characteristic features of fragile X syndrome; however, there is an increased risk for developing fragile X-associated tremor ataxia syndrome (FXTAS), especially for males, and females have an additional increased risk to develop primary ovarian insufficiency (FXPOI). Premutation carriers are also at risk for having descendents with expanded repeat sizes in the fragile X affected range. A normal or moderate expanded repeat result does not eliminate the risk of being affected by fragile X as there may be other contributing factors not tested for by this assay. **A positive result by itself should not be used as the only reason for diagnosis.** Rare (less than 1% of the time) errors may occur, for example sample mix-ups, or technical errors such as rare genetic variants that may mask the mutation being tested. **To understand your results, you should consult your physician and may wish to consider further independent testing or pursue genetic counseling.**

Limitations The accuracy of the test for detection of fragile X syndrome is >99%. Very rarely (<1%), other abnormalities of the *FMR1* gene have been found to give rise to fragile X syndrome that would not be detected by this assay.

Results from the test The test result will be provided to health care professionals directly involved with your care. 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. In accordance with NYS law, any remaining DNA will be discarded at the end of testing or not more than sixty (60) day after the sample was taken unless it is retained as a laboratory control, in which case the sample will be de-identified whereby all specific information identifying you will be removed.

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I have read this entire document and understand its contents. In addition, I understand that I am free to withdraw any portion of my consent by crossing off and initialing unacceptable statements. Please note that crossing off the laboratory's option to bill for the testing will result in the testing being cancelled.

If you have any questions about the test to be performed, you may also wish to obtain genetic counseling prior to signing this form. You may also contact the Upstate 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 (other than the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the fragile X syndrome 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 fragile X syndrome testing. **I hereby authorize SUNY Upstate Medical University to furnish my designated insurance carrier the information on this form if necessary for reimbursement. I also authorize benefits to be payable to SUNY Upstate Medical University. I understand that I am responsible for any amount not paid by insurance for reasons including, but not limited to, non-covered and non-authorized services. I permit a copy of this authorization to be used in place of the original.**

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 above indicated genetic test be performed.

Signature of Physician/Health Care Professional: _____

Printed Name/Stamp/Title: _____ Date/Time: _____