

SUNY Upstate Medical University Authorization for the Molecular Test for Fragile X Syndrome

1. What is fragile X syndrome? Fragile X syndrome is the most common inherited form of mental retardation. Individuals affected with fragile X have varying degrees of mental retardation, and most affected males have characteristic physical and behavioral features. The gene that causes this syndrome, known as FMR-1, is located on the X chromosome and is inherited in an 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 FMR-1 gene mutation (a carrier of the disorder) is at risk of having a child affected with fragile X syndrome.

2. What is the purpose of the test and what are its limitations? Fragile X syndrome nearly always results from an increase in size of a repeated DNA segment in the fragile X gene. In individuals affected with fragile X syndrome, the DNA region shows expansion to greater than 200 repeats. In normal individuals, the DNA region repeats itself only 6 to approximately 40 times. In asymptomatic male and female carriers of fragile X, the DNA region has moderate expansions of 60 to approximately 200 repeats. Although asymptomatic carriers do not demonstrate symptoms, these individuals are at risk for having descendants with repeat sizes in the affected range. A "gray zone" also exists (40 – 60 repeat range) within which the risk for subsequent expansion in offspring is difficult to predict. The Molecular Diagnostics Laboratory can perform an analysis that allows differentiation between large, moderate, and normal size genes for the diagnosis of an affected individual or for the detection of a carrier who is at risk of passing the gene on to a child. The accuracy of the test for detection of fragile X syndrome is >99%. Very rarely (<1%), other abnormalities of the fragile X gene have been found to give rise to the fragile X syndrome, and these causes would not be detectable by the use of this assay. 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? A 10 ml blood sample is necessary. This is equal to about two teaspoons. For prenatal diagnosis, 10-20 ml (2-4 teaspoons) of amniotic fluid collected by amniocentesis is necessary, and submission of a maternal blood sample is also required. 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 fragile X syndrome. The 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 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 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 fragile X 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 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 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