

**AUTHORIZATION FOR THE  
MOLECULAR TEST FOR  
FRAGILE X SYNDROME**

Patient Name: \_\_\_\_\_ MR#: \_\_\_\_\_

Account #: \_\_\_\_\_ DOB: \_\_\_\_\_ Date: \_\_\_\_\_

**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 the syndrome, known as FMR1, 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 (a carrier of the disorder) is at risk of having a child affected with fragile X syndrome. Fragile X syndrome nearly always results from an increase in the 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 individuals termed "premutation carriers" of fragile X, the DNA region has moderate expansions of 60 to approximately 200 repeats.

**Molecular test for Fragile X syndrome** You will be required to donate 10 mL of blood, which 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. 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 allows differentiation between large, moderate, and normal size genes 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 (40-60 repeat range) within which the risk for subsequent expansion in offspring is difficult to predict. Premutation carriers are generally not mentally impaired but are at increased risk of premature ovarian failure (POF) if they are female or fragile X-associated ataxia syndrome (FXTAS) if they are male. Premutation carriers are also at risk for having descendents with repeat sizes in the fragile X affected range. **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. **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 fragile X gene have been found to give rise to fragile X syndrome and these causes 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 unless it is retained as a laboratory control, in which case all information identifying the DNA to you will be removed.

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Patient's Name: \_\_\_\_\_ Account #: \_\_\_\_\_ MR#: \_\_\_\_\_

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 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.

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: \_\_\_\_\_