
Lab Dept: **Anatomic Pathology**

Test Name: **FRAGILE X MOLECULAR ANALYSIS (MAYO)**

General Information

Lab Order Codes: FXMA

Synonyms: Fragile X Syndrome; Martin-Bell Syndrome; DNA Probe Analysis Fragile X; Carrier Detection Fragile X; FMR1

CPT Codes: 81243 – FMR1 gene analysis, evaluation to detect abnormal alleles
81244 – FMR1 gene analysis, characterization of alleles (if appropriate)

Test Includes: An interpretive report of the molecular analysis findings. If appropriate, Fragile X Follow Up Analysis testing will be performed and charged dependent upon the size of the CAG repeat found by PCR analysis.

Logistics

Test Indications: Determination of carrier status for individuals with a family history of Fragile X Syndrome or X-linked mental retardation. Confirmation of a diagnosis of fragile X syndrome, fragile X tremor/ataxia syndrome, or premature ovarian failure caused by expansions in the FMR1 gene.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Mayo Medical Laboratories (MML Test: FXS), possible reflex to FXFU.

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 14 – 21 days; Specimens received Monday through Thursday will be processed for the run starting on Monday. Specimens received on Friday and Saturday will be set up a week from the following Monday.

Special Instructions: A Molecular Genetics – [Congenital Inherited Diseases Patient Information Sheet \(Supply T521\)](#) is required with the submission of the specimen. Please contact the laboratory for this form or print from the link above. Specimens must arrive at MML within 96 hours of collection. Submit specimens promptly to the laboratory.

An [“Informed Consent Form for DNA Testing”](#) (Supply T576) is available. Contact the laboratory for this form or print from the link above.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube or Yellow top (ACD) tube
Draw Volume:	2.5 mL (Minimum: 0.5 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture, gently invert the specimen to mix
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should remain in original collection container and should be kept at room temperature. Submit the Congenital Inherited Diseases Patient Information Sheet along with the specimen. The specimen must be received within 96 hours of collection. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Mislabeled/unlabeled specimens at Children's. Note: At Mayo, no specimen should be rejected and communication should occur with Mayo if the specimen is collected in the wrong anticoagulant or at the wrong temperature.

Interpretive

Reference Range: An interpretive report will be issued.

Critical Values: N/A

Limitations: For predictive testing, it is important to first document the presence of a CGG-repeat amplification in the FMR1 gene in an affected family member to confirm that molecular expansion is the underlying mechanism of disease in the family.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Methylation status cannot be assessed on chorionic villus specimens.

A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo for instructions for testing patients who have received a bone marrow transplant.

Less than 1% of individuals clinically diagnosed with fragile X syndrome do not have the CGG amplification-type mutation. These individuals may have a different type of mutation within the FMR1 gene (eg, deletion or point mutation) or a mutation in another gene.

Due to incomplete penetrance and variable expression of the FMR1

expansion, this test is not reliable for prenatal testing for fragile X syndrome.

The absence of an expansion in the FMR1 gene does not eliminate the diagnosis of other inherited disorders that have overlapping clinical features with fragile X syndrome, fragile X tremor/ataxia syndrome, or premature ovarian failure.

Methodology:

Polymerase Chain Reaction (PCR)-Based Assays
(PCR is utilized pursuant to a license agreement with Roche Molecular Systems, Inc.)

Direct Mutation Analysis – Southern blot analysis utilizes a DNA probe of the FMR1 gene (STB 12.3) along with a double digest (EcoRI and Nru I). This analysis allows for the simultaneous detection of a (CGG)(n)- repeat amplification and abnormal methylation. Additionally, the (CGG)(n)- repeat number of premutations is determined by analyzing a Polymerase Chain Reaction-amplified product by capillary electrophoresis (Celera).

References:

[Mayo Medical Laboratories Web Page](#) August 2015

Updates:

1/13/2009: CPT updates

5/21/2009: CPT updates, method change

2/5/2013: CPT update

4/1/2013: Inclusion of possible Follow-up reflex testing