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**Reporting Title:** Fragile X Syndrome, Mol. Analysis**Performing Location:** Rochester**Ordering Guidance:**

**Due to the complexity of prenatal testing, consultation with the laboratory is required.** To speak with a genetic counselor about this testing option, call 800-533-1710.

**FMR1-methylation status cannot be assessed on chorionic villus specimens.** Contact a molecular genetic counselor/consultant at 800-533-1710 to discuss the limitations of testing prior to sending a chorionic villus specimen for fragile X analysis.

**Additional Testing Requirements:**

**All prenatal specimens must be accompanied by a maternal blood specimen.** Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen. **This must be a different order number than the prenatal specimen.**

**Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

**Specimen Requirements:**

**Patient Preparation:** A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood

**Container/Tube:**

**Preferred:** Lavender top (EDTA) or yellow top (ACD)

**Acceptable:** Any anticoagulant

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**

**Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated

**Prenatal Specimens**

**Due to its complexity, consultation with the laboratory is required for all prenatal testing;** call 800-533-1710 to speak to a genetic counselor.

**Specimen Type:** Amniotic fluid

**Container/Tube:** Amniotic fluid container

**Specimen Volume:** 20 mL

**Specimen Stability Information:** Refrigerated (preferred)/Ambient

**Additional information:**

1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur.

2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

**Specimen Type:** Chorionic villi  
**Container/Tube:** 15-mL tube containing 15 mL of transport media  
**Specimen Volume:** 20 mg  
**Specimen Stability Information:** Refrigerated  
**Additional Information:**

1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 2 to 3 weeks is required to culture chorionic villi before genetic testing can occur.
2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

**Specimen Type:** Confluent cultured cells  
**Container/Tube:** T-25 flask  
**Specimen Volume:** 2 Flasks  
**Collection Instructions:** Submit confluent cultured cells from another laboratory.  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated  
**Additional Information:** All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

- Forms:**
1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:  
[-Informed Consent for Genetic Testing](#) (T576)  
[-Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Molecular Genetics: Congenital Inherited Diseases Patient Information](#) (T521)
3. If not ordering electronically, complete, print, and send a [Neurology Specialty Testing Client Test Request](#) (T732) with the specimen.

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies        | Varies      |      |                   |

Result Codes:

| Result ID | Reporting Name      | Type         | Unit | LOINC®  |
|-----------|---------------------|--------------|------|---------|
| 52870     | Result Summary      | Alphanumeric |      | 50397-9 |
| 52871     | Result              | Alphanumeric |      | 81856-7 |
| 52872     | Interpretation      | Alphanumeric |      | 69047-9 |
| 52873     | Reason for Referral | Alphanumeric |      | 42349-1 |
| 52874     | Specimen            | Alphanumeric |      | 31208-2 |
| 52875     | Source              | Alphanumeric |      | 31208-2 |
| 52876     | Method              | Alphanumeric |      | 85069-3 |
| 52877     | Released By         | Alphanumeric |      | 18771-6 |

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

- 81243
- 88233 (if appropriate)
- 88240 (if appropriate)
- 88235 (if appropriate)
- 81265 (if appropriate)
- 81244 (if appropriate)

Reflex Tests:

| Test Id | Reporting Name                        | CPT Units | CPT Code | Always Performed | Available Separately |
|---------|---------------------------------------|-----------|----------|------------------|----------------------|
| CULFB   | Fibroblast Culture for Genetic Test   | 1         | 88233    | No               | Yes                  |
| CULAF   | Amniotic Fluid Culture/Genetic Test   | 1         | 88235    | No               | Yes                  |
| MATCC   | Maternal Cell Contamination, B        | 1         | 81265    | No               | Yes                  |
| FUFXS   | Fragile X, Follow up Analysis         | 1         | 81244    | No               | No                   |
| _STR1   | Comp Analysis using STR (Bill only)   | 1         | 81265    | No               | No, (Bill only)      |
| _STR2   | Add'l comp analysis w/STR (Bill Only) | 1         | 81266    | No               | No, (Bill only)      |

Result Codes for Reflex Tests:

| Test ID | Result ID | Reporting Name         | Type         | Unit | LOINC®  |
|---------|-----------|------------------------|--------------|------|---------|
| CULAF   | 52304     | Result Summary         | Alphanumeric |      | 50397-9 |
| CULAF   | 52306     | Interpretation         | Alphanumeric |      | 69965-2 |
| CULAF   | 52305     | Result                 | Alphanumeric |      | 82939-0 |
| CULAF   | CG767     | Reason for Referral    | Alphanumeric |      | 42349-1 |
| CULAF   | 52307     | Specimen               | Alphanumeric |      | 31208-2 |
| CULAF   | 52308     | Source                 | Alphanumeric |      | 31208-2 |
| CULAF   | 52309     | Method                 | Alphanumeric |      | 85069-3 |
| CULAF   | 54641     | Additional Information | Alphanumeric |      | 48767-8 |
| CULAF   | 52310     | Released By            | Alphanumeric |      | 18771-6 |
| CULFB   | 52327     | Result Summary         | Alphanumeric |      | 50397-9 |
| CULFB   | 52329     | Interpretation         | Alphanumeric |      | 69965-2 |
| CULFB   | 52328     | Result                 | Alphanumeric |      | 82939-0 |
| CULFB   | CG770     | Reason for Referral    | Alphanumeric |      | 42349-1 |
| CULFB   | CG899     | Specimen               | Alphanumeric |      | 31208-2 |
| CULFB   | 52331     | Source                 | Alphanumeric |      | 31208-2 |
| CULFB   | 52332     | Method                 | Alphanumeric |      | 85069-3 |

|       |       |                        |              |  |         |
|-------|-------|------------------------|--------------|--|---------|
| CULFB | 54625 | Additional Information | Alphanumeric |  | 48767-8 |
| CULFB | 52333 | Released By            | Alphanumeric |  | 18771-6 |
| MATCC | 53285 | Result Summary         | Alphanumeric |  | 50397-9 |
| MATCC | 53286 | Result                 | Alphanumeric |  | 40704-9 |
| MATCC | 53287 | Interpretation         | Alphanumeric |  | 69047-9 |
| MATCC | 53288 | Reason for referral    | Alphanumeric |  | 42349-1 |
| MATCC | 53289 | Specimen               | Alphanumeric |  | 31208-2 |
| MATCC | 53290 | Source                 | Alphanumeric |  | 31208-2 |
| MATCC | 53291 | Released By            | Alphanumeric |  | 18771-6 |
| MATCC | 55150 | Method                 | Alphanumeric |  | 85069-3 |
| FUFXS | 52421 | Comment                | Alphanumeric |  | 48767-8 |
| FUFXS | 52422 | Specimen               | Alphanumeric |  | 31208-2 |
| FUFXS | 52423 | Source                 | Alphanumeric |  | 31208-2 |
| FUFXS | 52424 | Released By            | Alphanumeric |  | 18771-6 |

Reference Values:

Normal alleles: 5-44 CGG repeats  
Intermediate (grey zone) alleles: 45-54 CGG repeats  
Premutation alleles: 55-200 CGG repeats  
Full mutation alleles: >200 CGG repeats  
An interpretive report will be provided.

Methylation status:  
Unmethylated: < or =20%  
Partially methylated: 21-69%  
Fully methylated: > or =70%