

TriCore Reference Laboratories offers fragile X testing for diagnosis or carrier screening for this disorder.

Fragile X syndrome (FXS) is the most common cause of intellectual disability. It affects 1 in 4000 males and 1 in 5000-8000 females. The disease is caused by the reduction of protein encoded by fragile X mental retardation 1 (*FMR1*) gene located on the X chromosome (Xq27.3) due to expansion of CGG repeats in the 5' untranslated region of *FMR1* gene associated with hypermethylation and inactivation (transcriptional silencing) of the gene.

The fragile X mental retardation protein (FMRP) is normally expressed in high concentration in the brain and neural tissue, essential for normal cognitive development. Diagnostic testing for fragile X is recommended by the American College of Medical Genetics (ACMG) in symptomatic patients and in women with ovarian dysfunction, as well as in individuals with tremor/ataxia syndrome.

For additional information, contact TriCore's molecular diagnostics laboratory at 505-938-8684.

For consultation, please contact:

Dr. Mohammad Vasef                      mvasef@salud.unm.edu  
 Dr. David Czuchlewski                  dczuchlewski@salud.unm.edu

## FRAGILE X (FMR1) WITH REFLEX TO METHYLATION ANALYSIS

|   |  |
|---|--|
| Test Code.....FRAGXS  | Testing Performed.....Thursday, Friday |
| CPT Code.....81243; if reflexed add 81244                             | Reported.....Within 3-14 days          |
| Methodology.....Polymerase Chain Reaction (PCR) and Fragment Analysis |  |

### COLLECTION

|                      |   |
|----------------------|---|
| Specimen             | Preferred: Whole Blood<br>Acceptable: Bone Marrow                     |
| Collection Container | Preferred: EDTA - Lavender<br>Acceptable: EDTA - Pink<br>ACD - Yellow |
| Collection Amount    | Preferred.....5.0 mL<br>Minimum.....1.5 mL                            |
| Rejection Criteria   | Frozen samples are unacceptable.                                      |

### PROCESSING

|                                  |  |
|----------------------------------|--|
| Specimen Processing Instructions | CRITICAL: Do not centrifuge. Do not open or share tube.<br>Do not centrifuge whole blood sample or separate; specimen must be kept as whole blood in original container. |
|----------------------------------|--|

### SHIPPING/TRANSPORT AND STORAGE

|  |  |                    |                  |         |        |              |        |        |              |
|--|--|--------------------|------------------|---------|--------|--------------|--------|--------|--------------|
| Stabilities/Storage<br>(Collection to initiation of testing) | <table border="0"> <tr> <td><b>Temperature</b></td> <td><b>Stability</b></td> </tr> <tr> <td>Ambient</td> <td>5 days</td> </tr> <tr> <td>Refrigerated</td> <td>5 days</td> </tr> <tr> <td>Frozen</td> <td>Unacceptable</td> </tr> </table> | <b>Temperature</b> | <b>Stability</b> | Ambient | 5 days | Refrigerated | 5 days | Frozen | Unacceptable |
| <b>Temperature</b>   | <b>Stability</b>   |                    |                  |         |        |              |        |        |              |
| Ambient  | 5 days   |                    |                  |         |        |              |        |        |              |
| Refrigerated   | 5 days   |                    |                  |         |        |              |        |        |              |
| Frozen   | Unacceptable   |                    |                  |         |        |              |        |        |              |
| Shipping Instructions  | Ship refrigerated.   |                    |                  |         |        |              |        |        |              |