**ARUP LABORATORIES | www.aruplab.com**
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories



Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

## Patient: Patient, Example

| | |
|---|---|
| **DOB** | 1/25/1995 |
| **Gender:** | Male |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Fragile X (FMR1) with Reflex to Methylation Analysis

ARUP test code 2009033

| | |
|---|---|
| FRAG X Specimen | Whole Blood |
| Fragile X Allele 1 | 47 CGG repeats |
| Fragile X Allele 2 | Not Applicable CGG repeats |
| Fragile X Methylation Pattern | Not Applicable |
| Fragile X Interpretation | See Note<br><br>Intermediate Allele: This individual has an allele in the intermediate range. Expansion resulting in affected offspring has not been reported from carriers of intermediate range alleles, yet his female offspring are at increased risk for inheriting a premutation allele. Genetic counseling is recommended.<br><br>This result has been reviewed and approved by Wei Shen, Ph.D. |

H=High, L=Low, *=Abnormal, C=Critical

Background Information for Fragile X (FMR1)
Characteristics: Fragile X syndrome, the most common heritable form of mental retardation, is characterized by moderate mental retardation in males and mild mental retardation in females, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders behavioral phenotype, and connective tissue anomalies. Adult males may have physical findings including: macroorchidism, a long narrow face, prominent ears and jaw, and a single palmar crease.
Incidence: 1 in 4,000 Caucasian males and 1 in 8,000 Caucasian females; unknown in other ethnicities.
Inheritance: X-linked dominant.
Penetrance: Reduced in females.
Cause: Expansion of the FMR1 gene CGG triplet repeat.
Full mutation: >200-230 CGG repeats (methylated)
Premutation: 55-200 CGG repeats (unmethylated)
Intermediate: 45-54 CGG repeats (unmethylated)
Normal: 5-44 CGG repeats (unmethylated)
Clinical Sensitivity: 99 percent.
Methodology: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis. Methylation-specific PCR analysis is performed for CGG repeat lengths of 55 or greater. Methylation analysis is used to distinguish between premutation and full mutation alleles.
Analytic Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations.

See Compliance Statement C: www.aruplab.com/CS

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| FRAG X Specimen | 19-081-110860 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Fragile X Allele 1 | 19-081-110860 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Fragile X Allele 2 | 19-081-110860 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Fragile X Methylation Pattern | 19-081-110860 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Fragile X Interpretation | 19-081-110860 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |

END OF CHART

**H=High, L=Low, *=Abnormal, C=Critical**