

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 12/7/1991

**Gender:** Female

**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 30 CGG repeats

Fragile X Allele 2 30 CGG repeats  
Presumed Homozygous

Fragile X Methylation Pattern Not Applicable

Fragile X Interpretation See Note

Negative: This individual has two normal alleles; therefore, she is not a Fragile X carrier. This test does not detect rare mutations in less than 1% of Fragile X cases.

This result has been reviewed and approved by Yuan Ji, 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](http://www.aruplab.com/CS)

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FRAG X Specimen	19-098-402264	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	19-098-402264	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	19-098-402264	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	19-098-402264	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation	19-098-402264	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