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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/14/2004
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	107 CGG repeats
Fragile X Allele 2	35 CGG repeats
Fragile X Methylation Pattern	Unmethylated
Fragile X Interpretation	See Note

This individual has one FMR1 allele in the normal range and one allele in the premutation range. Although she is not affected with fragile X syndrome, she is at risk for FMR1-related disorders such as primary ovarian insufficiency and fragile X-associated tremor/ataxia syndrome (FXTAS) (Sherman, S. AJMG 2000; 97; 189-194). She also has a risk for transmitting an expanded allele to her offspring and having a child affected with fragile X syndrome. The risk for affected offspring corresponds with the premutation CGG repeat size as well as the sex of the offspring. Genetic consultation is recommended.

The expanded allele is unmethylated and predicted to be functional.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Fragile X (FMR1) with Reflex to Methylation Analysis
CHARACTERISTICS OF FRAGILE X SYNDROME (FXS): Affected males have moderate intellectual disability, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders and connective tissue anomalies in males. Females are usually less severely affected than males. FXS is caused by FMR1 full mutations.
CHARACTERISTICS OF FRAGILE X TREMOR ATAXIA SYNDROME (FXTAS): Onset of progressive ataxia and intention tremor typically after the fourth decade of life. Females also have a 21 percent risk for primary ovarian insufficiency. FXTAS is caused by FMR1 premutations.
Incidence of FXS: 1 in 4,000 Caucasian males and 1 in 8,000 Caucasian females.
INHERITANCE: X-linked.
PENETRANCE OF FXS: Complete in males; 50 percent in females.
PENETRANCE OF FXTAS: 47 percent in males and 17 percent in females >50 years of age.
CAUSE: Expansion of the FMR1 gene CGG triplet repeat.
  Full mutation: typically >200 CGG repeats (methylated).
  Premutation: 55 to approx 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 >100 to distinguish between premutation and full mutation alleles.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent; estimated precision of sizing for intermediate and premutation alleles is within 2-3 CGG repeats.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Rare FMR1 variants unrelated to trinucleotide expansion will not be detected. A specific CGG repeat size estimate is not provided for full mutation alleles. AGG trinucleotide interruptions within the FMR1 CGG repeat tract are not assessed.

PHENOTYPE	NUMBER OF CGG REPEATS
Unaffected	<45
Intermediate	45-54
Premutation	55-200
Affected	>200

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FRAG X Specimen	24-040-148127	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	24-040-148127	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	24-040-148127	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	24-040-148127	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation	24-040-148127	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