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

Physician: Doctor, Example

Patient: Patient, Example

DOB	8/21/1990
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	31 CGG repeats
Fragile X Allele 2	29 CGG repeats
Fragile X Methylation Pattern	Not Applicable
Fragile X Interpretation	See Note

Section 79-L of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

This individual has two FMR1 alleles with CGG sizes in the normal range; therefore, she is predicted to be neither affected with, nor a carrier of, fragile X syndrome (FXS). This test does not detect rare FMR1 variants causing less than 1% of FXS.

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. Females are usually less severely affected than males. FXS is caused by FMR1 full mutations.
CHARACTERISTICS OF FRAGILE X-ASSOCIATED TREMOR ATAXIA SYNDROME (FXTAS):
Onset of progressive ataxia and intention tremor typically after the fifth decade of life. Cognitive impairment and behavioral features may also develop. FXTAS is caused by FMR1 premutations.
CHARACTERISTICS of FRAGILE X-ASSOCIATED PRIMARY OVARIAN INSUFFICIENCY (FXPOI): Primary ovarian insufficiency or hypergonadotropic hypogonadism before 40 years of age. FXPOI is associated with FMR1 premutations.
CHARACTERISTICS of FRAGILE X-ASSOCIATED NEUROPSYCHIATRIC DISORDERS (FXAND): Symptoms may include anxiety, depression, adult ADHD, or addictive behavior. FXAND is associated with FMR1 premutations.
PREVALENCE OF FXS: Approximately 1 in 4,000-7,000 males and 1 in 8,000-11,000 females.
PREVALENCE OF PREMUTATION ALLELE: Approximately 1 in 150-300 females and 1 in 300-800 males.
INHERITANCE: X-linked.
PENETRANCE OF FXS: Complete in males; 50 percent in females.
PENETRANCE OF FXTAS: For individuals greater than 50 years of age, approximately 40 percent in males and 16-20 percent in females.
PENETRANCE OF FXPOI: Approximately 20 percent in females.
CAUSE: Expansion of the FMR1 gene CGG triplet repeat.
Full mutation: Typically greater than 200 CGG repeats (methylated). Premutation: 55 to approximately 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 greater than 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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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	24-235-100956	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	24-235-100956	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	24-235-100956	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	24-235-100956	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation	24-235-100956	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