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

Physician: Doctor, Example

Patient: Patient, Example

DOB	8/21/1991
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, Fetal

ARUP test code 2009034

Fragile X Fetal Specimen	Cultured Amnio
Fragile X Allele 1	> 200
Fragile X Allele 2	Not Applicable CGG repeats
Fragile X Methylation Pattern	**Full** *
Fragile X Interpretation, Fetal	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.

According to information provided to ARUP, the mother of this fetus is reported to be a carrier of a Fragile X premutation allele with 97 CGG repeats. This male fetus has a loss-of-function FMR1 allele (typically greater than 200 CGG repeats that is fully methylated), thus, is affected with fragile X syndrome. Genetic consultation is recommended.

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, Fetal
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 approx 200 CGG repeats (unmethylated).
Intermediate: 45-54 CGG repeats (unmethylated).
Normal: 5-44 CGG repeats (unmethylated).
CLNICAL 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 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: Methylation patterns may not be fully established in early gestation; thus, diagnostic testing on chorionic villus samples is not recommended. Diagnostic errors can occur due to rare sequence variations. Rare FMR1 variants unrelated to 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.

Maternal Contamination Study Fetal Spec	Fetal Cells

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

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Maternal Contam Study, Maternal Spec	Whole Blood

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Fragile X Fetal Specimen	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation, Fetal	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	24-235-100954	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	24-235-100954	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