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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/8/1988
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	See Note
Fragile X Allele 1	> 200
Fragile X Allele 2	Not Applicable CGG repeats
Fragile X Methylation Pattern	See Note *
Fragile X Interpretation, Fetal	See Note

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

Indication for Testing: Prenatal diagnosis for Fragile X syndrome.

Full Mutation Mosaic: This fetus has an allele with subpopulations of full mutations (>200 repeats) that are partially methylated, premutations (112 repeats) that are unmethylated, and a normal allele (30 repeats) that is unmethylated.

Interpretation: According to information provided to ARUP, the mother of this fetus is a carrier of a Fragile X premutation allele (105 CGG repeats), tested at an outside laboratory. This male fetus has one CGG repeat in the full mutation range, one repeat in the normal range, and one repeat in the premutation range detected by PCR on a CVS sample. The expanded allele shows both methylated and unmethylated patterns. If the expanded allele is later determined to be fully methylated, this fetus would be predicted to be affected with fragile X syndrome.

Recommendations: To further assess methylation status of the expanded allele, repeat testing on amniocytes is recommended (Fragile X with Reflex to Methylation Analysis, Fetal; ARUP test code 2009034). Genetic consultation is recommended. For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor (800-242-2787 ext. 2141) prior to specimen submission.

This result has been reviewed and approved by [redacted]

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 in males. Females are usually less severely affected than males.
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. 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).
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 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.

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

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

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.

Maternal Contam Study, Maternal Spec

Whole Blood

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Fragile X Fetal Specimen	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation, Fetal	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	20-160-120747	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	20-160-120747	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