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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/29/1981
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	Amniocytes
Fragile X Allele 1	66 CGG repeats
Fragile X Allele 2	Not Applicable CGG repeats
Fragile X Methylation Pattern	Normal
Fragile X Interpretation, Fetal	See Note

According to information provided to ARUP, the mother of this fetus had fragile X carrier testing performed by an outside laboratory, which detected a premutation allele (64 repeats) and a normal allele (26 repeats) in the FMR1 gene. This male fetus has one FMR1 allele in the premutation range with a normal methylation pattern, and thus, is predicted to be unaffected with fragile X syndrome. However, this fetus is at risk for developing fragile X associated tremor/ataxia syndrome (FXTAS); approximately 40-45% of males with a premutation develop FXTAS after age 50 (Am J Hum Genet. 2004; 74:805-816). Genetic consultation is recommended.

Please note, estimated precision of sizing for premutation alleles is within 2-3 repeats. 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 at (800) 242-2787 extension 2141 prior to specimen submission.

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 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	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation, Fetal	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	20-083-126135	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	20-083-126135	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