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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/27/1995
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	Amniotic fluid
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

Test results should be interpreted with caution. Assay was performed at client's request on a sub-optimal specimen.

According to information provided to ARUP, the mother of this fetus has fragile X carrier screening at an outside laboratory, which detected a premutation allele (104 repeats) and a normal allele (30 repeats) in the FMR1 gene. This male fetus has one expanded FMR1 allele. The allele has a loss-of-function cell line (methylated with greater than 200 CGG repeats) and a functional cell line (unmethylated and 104 CGG repeats). This result is consistent with a diagnosis of fragile X syndrome in this fetus. Nearly all males with size mosaicism for an expanded allele have intellectual disability, but these individuals may be higher functioning than those with only a nonfunctional cell line. 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.
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 White males and 1 in 8,000 White 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.

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.

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Fragile X Fetal Specimen	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation, Fetal	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	23-320-403245	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	23-320-403245	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