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	10/11/1991
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	Amniocytes
Fragile X Allele 1	71 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 harbors one FMR1 gene allele in the normal range (30 CGG repeats) and one allele in the premutation range (67 CGG repeats). This male fetus has one FMR1 allele in the premutation range (71 CGG repeats) with a normal methylation pattern; 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.

Methylation pattern is normal for gender.

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	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation, Fetal	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	23-346-120294	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	23-346-120294	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