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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	11/2/2012
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Fragile X (FMR1) with Reflex to Methylation Analysis

ARUP test code 2009033

FRAG X Specimen	Whole Blood
Fragile X Allele 1	> 200
Fragile X Allele 2	Not Applicable CGG repeats
Fragile X Methylation Pattern	See Note *
Fragile X Interpretation	See Note Allele 1: >200 Allele 2: NA Methylation: See Note This individual has an expanded allele in the full mutation range (>200 CGG repeats); therefore he is predicted to be affected with Fragile X syndrome. Methylation PCR analysis also detected a mosaic pattern with subpopulations of full mutation allele (>200 CGG repeats) that are unmethylated. It is not possible to predict the severity of the full mutation from the size of the CGG repeat or the degree of associated methylation. Genetic consultation is recommended. This result has been reviewed and approved by Yuan Ji, Ph.D.

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

Background Information for Fragile X (FMR1)
Characteristics: Fragile X syndrome, the most common heritable form of mental retardation, is characterized by moderate mental retardation in males and mild mental retardation in females, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders behavioral phenotype, and connective tissue anomalies. Adult males may have physical findings including: macroorchidism, a long narrow face, prominent ears and jaw, and a single palmar crease.
Incidence: 1 in 4,000 Caucasian males and 1 in 8,000 Caucasian females; unknown in other ethnicities.
Inheritance: X-linked dominant.
Penetrance: Reduced in females.
Cause: Expansion of the FMR1 gene CGG triplet repeat.
Full mutation: >200-230 CGG repeats (methylated)
Premutation: 55-200 CGG repeats (unmethylated)
Intermediate: 45-54 CGG repeats (unmethylated)
Normal: 5-44 CGG repeats (unmethylated)
Clinical 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. Methylation analysis is used to distinguish between premutation and full mutation alleles.
Analytic Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FRAG X Specimen	19-063-401977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 1	19-063-401977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Allele 2	19-063-401977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Methylation Pattern	19-063-401977	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fragile X Interpretation	19-063-401977	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