ARUP LABORATORIES
800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/18/1983
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal

ARUP test code 2012232

Angelman and Prader-Willi Result

PraderPositive *

Methylation pattern: Abnormal

Interpretation: According to information provided to ARUP, a genomic microarray performed for this fetus at an outside laboratory was positive for either Angelman syndrome or Prader Willi syndrome. Methylation analysis was requested to distinguish between these possibilites. Only the maternally contributed Angelman Syndrome (AS)/Prader-Willi Syndrome (PWS) critical region is present in this prenatal sample; therefore, this fetus is predicted to be affected with PWS.

Recommendations: Genetic consultation is recommended. To provide accurate information regarding recurrence risk, determination of the precise molecular mechanism involved is necessary. This result should be correlated with the fetal genomic microarray result.

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: Angelman Syndrome and Prader-Willi Syndrome by Methylation

CHARACTERISTICS OF ANGELMAN SYNDROME (AS): Developmental delays by 6-12 months of age, seizures, microcephaly, movement or balance disorder, minimal or absent speech, and a distinctive behavioral phenotype, which includes a happy demeanor with frequent laughter, hand flapping, and excitability.
PREVALENCE: 1 in 15,000.
INHERITANCE: Varies, depending on the molecular genetic mechanism.
CAUSE: Absence of maternal expression of the UBE3A gene.
MOLECULAR GENETIC MECHANISMS: Microdeletions in the AS/PWS critical region (68 percent), UBE3A mutations (11 percent), paternal uniparental disomy of chromosome 15 (7 percent), imprinting center defects (3 percent), unbalanced chromosome translocation (less than 1 percent), and unknown (10 percent).
Clinical Sensitivity: 78 percent.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
METHODOLOGY: Bisulfite conversion and PCR amplification to detect methylation using melting curve analysis.
LIMITATIONS: Molecular mechanisms not affecting methylation patterns that may result in AS will not be assessed. Diagnostic errors can occur due to rare sequence variations.

CHARACTERISTICS OF PRADER-WILLI SYNDROME (PWS): Neonatal hypotonia, hyperphagia, obesity, global developmental delay, mild intellectual disability, hypogonadism, and a distinctive behavioral phenotype, which includes temper tantrums, stubbornness, manipulative behavior, and obsessive-compulsive behavior.
PREVALENCE: 1 in 15,000.
INHERITANCE: Varies, depending on the molecular genetic mechanism.
CAUSE: Absence of the paternally contributed PWS/AS critical region of chromosome 15q11.2-q13.
MOLECULAR GENETIC MECHANISMS: Microdeletions in the PWS/AS critical region (70-75 percent), maternal uniparental disomy of chromosome 15 (25-29 percent), imprinting center defect or balanced chromosome translocation (less than 1 percent).
CLINICAL SENSITIVITY: Over 99 percent.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
METHODOLOGY: Bisulfite conversion and PCR amplification to detect methylation using melting curve analysis.
LIMITATIONS: Molecular mechanisms not affecting methylation patterns that may result in PWS will not be assessed. Diagnostic errors can occur due to rare sequence variations.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

Angelman and Prader-Willi Fetal Specimen	Amniocytes
Maternal Contamination Study Fetal Spec	Unknown Origin

Single genotype. A maternal specimen was not submitted for correlation. The fetal sample was tested using STR markers to rule out maternal cell contamination. Only a single genotype was detected. Testing a maternal sample can confirm that this genotype is from the fetus.

INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen
Please refer to fetal report for interpretation.

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Maternal Contam Study, Maternal Spec	Not Received 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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Angelman and Prader-Willi Result	19-262-401626	8/30/2019 9:15:00 AM	9/19/2019 1:14:12 PM	9/25/2019 10:49:00 AM
Angelman and Prader-Willi Fetal Specimen	19-262-401626	8/30/2019 9:15:00 AM	9/19/2019 1:14:12 PM	9/25/2019 10:49:00 AM
Maternal Contamination Study Fetal Spec	19-262-401626	8/30/2019 9:15:00 AM	9/19/2019 1:14:12 PM	9/24/2019 3:54:00 PM
Maternal Contam Study, Maternal Spec	19-262-401626	8/30/2019 9:15:00 AM	9/19/2019 1:14:12 PM	9/24/2019 3:54:00 PM

END OF CHART

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