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

Physician: Doctor, Example

## Patient: Patient, Example

| | |
|---|---|
| **DOB** | 12/31/1752 |
| **Gender:** | Unknown |
| **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

Negative

Methylation pattern: Normal

Interpretation: Both the maternally and paternally contributed Angelman Syndrome (AS)/Prader-Willi Syndrome (PWS) critical regions are present in this prenatal sample. This result reduces, but does not exclude, a diagnosis of AS. Approximately 20 percent of individuals with AS will have normal methylation patterns. Within that group, approximately half will have UBE3A causative mutations, 1 percent will have a cytogenetically visible chromosomal rearrangement and the remainder (approximately 10 percent) will have an unidentified genetic mechanism. This result greatly reduces the chance for PWS, since 99 percent of individuals with PWS have abnormal methylation patterns.

Recommendations: Genetic consultation is recommended. 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

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|---|---|
| Angelman and Prader-Willi Fetal Specimen | Amniocytes |
| Maternal Contamination Study Fetal Spec | Fetal Cells<br><br>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 |
|---|---|---|---|---|
| Angelman and Prader-Willi Result | 20-042-100999 | 2/11/2020 6:05:00 AM | 2/11/2020 6:05:30 AM | 2/11/2020 10 00:00 AM |
| Angelman and Prader-Willi Fetal Specimen | 20-042-100999 | 2/11/2020 6:05:00 AM | 2/11/2020 6:05:30 AM | 2/11/2020 10 00:00 AM |
| Maternal Contamination Study Fetal Spec | 20-042-100999 | 2/11/2020 6:05:00 AM | 2/11/2020 6:05:30 AM | 2/11/2020 10 00:00 AM |
| Maternal Contam Study, Maternal Spec | 20-042-100999 | 2/11/2020 6:05:00 AM | 2/11/2020 6:05:30 AM | 2/11/2020 10 00:00 AM |

END OF CHART

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