ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108-
USA

Provider: .108 -TEST,

Patient: **AS PWS FE, POSITIVE**
DOB:
Sex: Female
Patient Identifiers: 40656
Visit Number (FIN): 40981
Client Supplied ID:

Specimen Collected: 19-Sep-22 16:26

Angelman / Prader-Willi Syndromes, Fetal | **Received: 19-Sep-22 16:38** **Report/Verified: 19-Sep-22 17:00**

Procedure	Result	Units	Reference Interval
Angelman and Prader-Willi Result	**Angel Positive** * f1 i1		
Angelman and Prader-Willi Fetal Specimen	Amniotic fluid		
Maternal Contamination Study Fetal Spec	**Maternal Cells** * f2		
Maternal Contam Study,Maternal Spec	Whole Blood i2		

Result Footnote

f1: Angelman and Prader-Willi Result

Methylation pattern: Abnormal

Interpretation: Only the paternally 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 AS.

Recommendations: Genetic consultation is recommended. To provide accurate information regarding recurrence risk, additional testing for AS is recommended to determine the precise molecular mechanism involved.

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 Rong Mao, M.D.

f2: Maternal Contamination Study Fetal Spec

Single maternal genotype; no fetal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination. Results were identical to the maternal genotype indicating that no fetal cells were present in the sample. Please request an additional sample.

Test Information

i1: Angelman and Prader-Willi Result

BACKGROUND INFORMATION: Angelman Syndrome and Prader-Willi Syndrome by Methylation

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Patient: **AS PWS FE, POSITIVE**
DOB:
Patient Identifiers: 40656

Test Information

i1: Angelman and Prader-Willi Result

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: Methylation Sensitive Polymerase Chain Reaction/Fluorescence Monitoring.
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: Methylation Sensitive Polymerase Chain Reaction/Fluorescence Monitoring.
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.

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Test Information

i1: Angelman and Prader-Willi Result

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.

i2: Maternal Contam Study, Maternal Spec

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