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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 43 years Female

Specimen Collected: 08-Mar-22 10:52

Angelman / Prader-Willi Syndromes, Fetal | **Received: 08-Mar-22 10:52** **Report/Verified: 10-Mar-22 14:05**

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

Result Footnote

f1: Angelman and Prader-Willi Result

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 Rong Mao, M.D.

f2: Maternal Contamination Study Fetal Spec

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

Test Information

i1: Angelman and Prader-Willi Result

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.

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

Test Information

i1: Angelman and Prader-Willi Result

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.

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

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

Test Information

i2: Maternal Contam Study, Maternal Spec
submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

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