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

Patient Report

Patient Age/Sex: 41 years Male

Specimen Collected: 08-Mar-22 10:49

Angelman and Prader-Willi Syndrome | **Received: 08-Mar-22 10:49** **Report/Verified: 10-Mar-22 14:03**

Procedure	Result	Units	Reference Interval
Angelman and Prader-Willi Specimen	Whole Blood		
Angelman and Prader-Willi Result	Negative [f1 i1]		

Result Footnote

f1: Angelman and Prader-Willi Result

Methylation pattern: Normal

Both the maternally and paternally contributed Angelman Syndrome (AS)/Prader-Willi Syndrome (PWS) critical regions are present in this 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: Medical screening and management should rely on clinical finding and family history. A genetics consultation is recommended.

This result has been reviewed and approved by Rong Mao, M.D.

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.
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.

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i1: Angelman and Prader-Willi Result

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.

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Tracy I. George, MD