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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: 33 years Female

Specimen Collected: 14-Aug-25 12:55

Angelman and Prader-Willi Synd MLPA, FE **|Received: 14-Aug-25 12:55** **Report/Verified: 15-Aug-25 11:44**

Procedure	Result	Units	Reference Interval
Maternal Contamination Study Fetal Spec	Fetal Cells [f1]		
Maternal Contam Study,Maternal Spec	Whole Blood		
AS-PWS Fetal Specimen	Cultured Amnio		
AS-PWS Fetal Interpretation	**AS Deletion** * [f2] [i1]		

Result Footnote

f1: 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.

f2: AS-PWS Fetal Interpretation

Methylation Pattern: Abnormal maternal methylation pattern
Copy Number Analysis: Deletion detected

According to information provided to ARUP, prenatal cell-free DNA (cfDNA) screening performed on this pregnancy identified increased risk for Angelman syndrome (AS). Only the paternally contributed AS/Prader-Willi Syndrome (PWS) critical region is present in this fetal sample. Copy number analysis of this region detected a deletion. This result is consistent with a diagnosis of AS in this fetus due to a deletion in AS/PWS critical region.

Recommendations: Genetic consultation is indicated.

Please send any postnatal outcomes concerning this prenatal result to ARUP Genetic Counseling fax: 801-584-5236.

This result has been reviewed and approved by [redacted]

Test Information

i1: AS-PWS Fetal Interpretation

BACKGROUND INFORMATION: Angelman Syndrome and Prader-Willi Syndrome by Methylation-Specific MLPA, Fetal

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.

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.

Test Information

i1: AS-PWS Fetal Interpretation

Prevalence: 1 in 15,000 for AS; 1 in 15,000 for PWS.

Inheritance: Varies, depending on the molecular genetic mechanism.

Cause: AS: Absence of maternal expression of the UBE3A gene.
PWS: Absence of the paternally contributed PWS/AS critical region of chromosome 15q11.2-q13.

Molecular Genetic Mechanisms: AS: 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).
PWS: 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: PWS: Over 99 percent. AS: 80 percent.

Methodology: Methylation-specific multiplex ligation probe 15q11.2-q13.

Analytical Sensitivity and Specificity: 99 percent for AS and PWS.

Limitations: Prenatal specimens with maternal cell contamination may give false-negative results. Disease mechanisms causing AS that do not alter methylation patterns will not be detected. Diagnostic errors can occur due to rare sequence variations. This assay is not validated to detect increased copy number of 15q11.2-q13 nor determine parent of origin for duplications. This assay cannot distinguish between UPD or an imprinting defect for PWS or AS.
AS and PWS mosaicism will not be assessed by this assay.
Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.
Methylation patterns may not be fully established in early gestation; thus, diagnostic testing on chorionic villus samples is not recommended.

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.

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