MAYO CLINIC LABORATORIES

1-800-533-1710

PWAS

Prader-Willi/Angelman Syndrome, Molecular Analysis, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00001027	SAMPLEREPDMGLM, VLD20150713A0288		1981-01-01	M	34
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00001027	SA00001027	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		12 Jul 2015 19:19			

Prader Willi/Angelman Mol Analysis

Result Summary MCR

NEGATIVE

Result MCR

MLPA demonstrated a normal methylation pattern. No deletions or duplications were identified.

Interpretation [1] MCR

These results decrease the likelihood but do not rule out a diagnosis of Prader-Willi syndrome.

The diagnosis of Prader-Willi syndrome is not excluded because a small number of individuals with a clinical diagnosis of Prader-Willi syndrome (approximately 2%) have point mutations or deletions which are not detected by this assay.

Because some chromosome abnormalities may have overlapping clinical features with Prader-Willi syndrome, a chromosomal microarray is recommended if not already performed.

A genetic consultation may be a benefit.

ADDITIONAL INFORMATION

Methylation-sensitive multiplex ligation-dependent probe amplification (MLPA) was used to test for the presence of large deletions, duplications, and/or methylation defects in the Prader-Willi/Angelman syndrome (PW/AS) critical region. An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Reason for Referral MCR

Evaluation for a diagnosis of Prader-Willi (PW) Syndrome. Analyze the PW critical region for alterations in the DNA methylation pattern.

Specimen MCR

WB Whole Blood

Released By MCR

EMILY LAUER

Received: 13 Jul 2015 20:29 **Reported:** 27 Jul 2015 10:28

Test Environment EHR Template

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292