MAYO CLINIC LABORATORIES

1-800-533-1710

BWRS

BWS/RSS Molecular Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000922	SAMPLEREPDMGLM, VLD20150713A0183		2005-01-01	M	10
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00000922	SA00000922	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jul 2015 17:07			

BWS/RSS Molecular Analysis

Result Summary

MCR

NEGATIVE

Result

MCR

IC1 (H19): normal methylation
IC2 (LIT1): normal methylation

No deletions or duplications were identified.

Interpretation

[1] MCR

These results decrease the likelihood but do not rule out the diagnosis of Beckwith-Wiedemann syndrome (BWS). Some individuals with a clinical diagnosis of BWS do not have a detectable abnormality in the 11p15 critical regions. This assay does not assess for other causes of BWS, including point mutations in the CDKN1C gene. If not already performed, genetic testing of the CDKN1C gene (CDKZ/CDKN1C Gene, Full Gene Analysis) may provide additional diagnostic information.

Additionally, methylation abnormalities may not be detected in the presence of low level mosaicism.

A genetic consultation may be of 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 IC1 (H19) and IC2 (LIT1) critical regions on chromosome 11p15. 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 Beckwith Wiedemann syndrome (BWS). Analyze the IC1 (H19) and IC2 (LIT1) critical regions.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:25 **Reported:** 22 Jul 2015 10:53

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