

Patient Name SAMPLEREPDMGLM,VLD20150713A0191	Patient ID SA00000930	Age 10	Gender M	Order # SA00000930
Ordering Phys CLIENT,CLIENT				DOB 01/01/2005
Client Order # SA00000930	Account Information C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			Report Notes
Collected 07/12/2015 17:16				
Printed 08/03/2015 11:56				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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CDKN1C Gene, Full Gene Analysis

Result Summary	NEGATIVE	MCR
Result	See Below	MCR

Result: No reportable variants were identified.

Interpretation		MCR
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This result decreases the likelihood but does not rule out the diagnosis of Beckwith-Wiedemann syndrome (BWS). We predict that there are individuals with a diagnosis of BWS who have a pathogenic mutation that is not detectable by the method described (e.g. mutations occurring at c.481 to c.595, large deletions/duplications, promoter mutations, or deep intronic mutations). Additionally, this assay does not rule out other known causes of BWS, including chromosomal abnormalities, deletions, paternal uniparental disomy, or imprinting center defects.

This result should be interpreted in the context of clinical findings and family history. If not already performed, consider methylation and deletion/duplication analysis (BWRS / BWS/RSS Molecular Analysis).

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

-----ADDITIONAL INFORMATION-----

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries (excluding c.481-c.595) of the CDKN1C gene (GenBank accession number NM_000076; build GRCh37 (hg19)).

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.

Performing Site Legend on Last Page of Report

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Page 1 of 2		>> Continued on Next Page >>

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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 Medical 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.

Laboratory developed test.

PDF Report available at:

<https://test.mmlaccess.com/Reports/C7028846-qIIIVDWMuZ.ashx>

Specimen WB Whole Blood
Released By EMILY LAUER

MCR
MCR

RECEIVED: 07/13/2015 20:25 **REPORTED:** 07/31/2015 16:44

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: William G. Morice, II, M.D., Ph.D.
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Page 2 of 2		** End of Report **

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