MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CDKMS	SA00049006	17	M	SA00049006
Ordering Phys				**DOB** 06/15/1995
Client Order # SA00049006	**Account Information**			**Report Notes**
Collected 09/14/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/05/2012 16:35				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CDKN1C Gene, Full Gene Analysis			REPORTED 09/21/2012 01:30		
Specimen		Blood			MCR
Specimen ID		1038796			MCR
Order Date		14 Sep 2012 09:13			MCR
Reason For Referral					MCR
Patient reported to have features suggestive of Beckwith-Wiedemann syndrome. Test for the presence of a mutation in the CDKN1C gene.					
Method					MCR
Bi-directional sequence analysis was used to test for the presence of a mutation in all coding regions and intron/exon boundaries of the CDKN1C gene (excluding c.481-c.595). Mutation nomenclature is based on GenBank accession number; NM_000076.2.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR

This result does not rule out the diagnosis of Beckwith-Wiedemann syndrome for this individual. We predict that a small percentage of individuals with Beckwith-Wiedemann syndrome caused by CDKN1C may have mutations that are not identified by the method described (e.g., mutations occurring at c.481-c.595, large deletions/duplications, promoter mutations, or deep intronic mutations).

Additionally, this test does not rule out other known causes of Beckwith-Wiedemann syndrome including chromosomal abnormalities, deletions, paternal uniparental disomy, or imprinting center defects. If not already performed, consider methylation and deletion/duplication analysis (Mayo test code 83278).

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.

CAUTIONS:
Test results should be interpreted in context of clinical

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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 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.					
Laboratory developed test.					
Reviewed By					MCR
Kandelaria Margarita Rumilla MD					
Release Date		21 Sep 2012 01:27			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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