MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| SAMPLEREPORT,CDKKM | SA00049005 | 23 | M | SA00049005 |
| **Ordering Phys** | | | | **DOB** 06/15/1989 |
| **Client Order #** SA00049005 | **Account Information** | | | **Report Notes** |
| **Collected** 09/14/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 10/05/2012 16:45 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **CDKN1C Gene, Known Mutation** | | | REPORTED 09/21/2012 01:32 | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1038794 | | | MCR |
| Order Date | | 14 Sep 2012 09:12 | | | MCR |
| Reason For Referral | | | | | MCR |
| Family history of Beckwith-Wiedemann syndrome. Test for the presence of the familial alteration in the CDKN1C gene. | | | | | |
| Method | | | | | MCR |
| DNA sequence analysis was used to test for the presence of the c.139C>T (p.Q47X) alteration in exon 1 of the CDKN1C gene. Testing was performed for this specific alteration because it was previously identified in a family member. Mutation nomenclature is based on GenBank accession number; NM_000076.2. | | | | | |
| Result | | | | | MCR |
| The following heterozygous sequence change was detected:<br>Exon: 1<br>DNA change: c.139C>T<br>Amino acid change: p.Q47X (Gln47X)<br>Classification: DELETERIOUS | | | | | |
| Interpretation | | | | | MCR |
| The p.Q47X alteration is a known deleterious mutation. | | | | | |

This result indicates that this individual is a carrier of Beckwith-Wiedemann syndrome. This interpretation assumes that this individual is healthy and is not clinically affected with Beckwith-Wiedemann syndrome. Offspring of a male carrier have a 50% chance to be carriers for Beckwith-Wiedemann syndrome. Therefore, successive generations may be at increased risk for having children with Beckwith-Wiedemann syndrome.

Since a mutation has been identified, testing of other at risk family members is possible.

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.

***Performing Site Legend on Last Page of Report***



* Report times for Mayo performed tests are CST/CDT

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CAUTIONS:
Test results should be interpreted in 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 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 01:29 | | | MCR |

* Performing Site:

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



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