

<b>Patient Name</b> SAMPLEREPORT,BWSRS N	<b>Patient ID</b> SA00058923	<b>Age</b> 47	<b>Gender</b> F	<b>Order #</b> SA00058923
<b>Ordering Phys</b> CLIENT,CLIENT			<b>DOB</b> 06/10/1966	
<b>Client Order #</b> SA00058923	<b>Account Information</b> C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			<b>Report Notes</b>
<b>Collected</b> 06/10/2013 00:00				
<b>Printed</b> 06/12/2013 10:11				

Test	Flag	Results	Unit	Reference Value	Perform Site*
<b>BWS/RSS Molecular Analysis</b>					
<b>RECEIVED:</b> 06/11/2013 12:59 <b>REPORTED:</b> 06/11/2013 17:10					
Specimen		Blood			MCR
Specimen ID		1062174			MCR
Order Date		11 Jun 2013 13:14			MCR
Reason for Referral		Suspected diagnosis of Beckwith Wiedemann syndrome (BWS).			MCR
Method		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.			MCR
Result		IC1 (H19): Normal methylation IC2 (LIT1): Normal methylation			MCR
		No deletions or duplications were detected.			
Interpretation		These results decrease the likelihood but do not rule out the diagnosis of Beckwith Wiedemann syndrome (BWS). Some patients 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 (CDKMS/60228 CDKN1C Gene, Full Gene Analysis) may provide additional diagnostic information.			MCR
		Additionally, methylation abnormalities may not be detected in the presence of low level mosaicism.			
		A genetic consultation may be of benefit.			
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.			

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

<b>Patient Name</b> SAMPLEREPORT,BWSRS N	<b>Collection Date and Time</b> 06/10/2013 00:00	<b>Report Status</b> Final
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\* Report times for Mayo performed tests are CST/CDT

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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  
Release Date

Benjamin Robert Kipp  
11 Jun 2013 17:09

MCR  
MCR

\* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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