MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BWSRS	SA00049890	26	F	SA00049890
Ordering Phys				**DOB** 06/10/1986
Client Order # SA00049890	**Account Information**			**Report Notes**
Collected 10/17/2012 14:07	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/17/2012 14:38				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Amniotic Fluid Culture/Genetic Test			REPORTED 10/17/2012 14:12		
Specimen		Amniotic Fluid			MCR
Specimen ID		1059264			MCR
Order Date		17 Oct 2012 14:08			MCR
Reason For Referral		not provided			MCR
Method		Amniocyte culture			MCR
Interpretation					MCR
We have established a cell culture on this specimen for possible molecular or biochemical genetic testing. The cell culture ordered does not include cryopreservation and we cannot maintain this specimen indefinitely. Please call (800) 533-1710 ext. 42950 to inform the Cytogenetics Laboratory of your intentions. This specimen will be stored for six months from the date it was received in the Cytogenetics Laboratory (10/17/2012).					
Consultant		Sheila Marie Wilson			MCR
Report Date		17 Oct 2012 14:09			MCR
BWS/RSS Molecular Analysis			REPORTED 10/17/2012 14:11		
Specimen		Amniotic Fluid			MCR
Specimen ID		1038915			MCR
Order Date		17 Oct 2012 14:08			MCR
Reason For Referral					MCR
Suspected diagnosis of Beckwith Wiedemann syndrome (BWS).					
Method					MCR
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.					
Result					MCR
IC1 (H19): hypermethylation IC2 (LIT1): hypomethylation No deletions or duplications were detected.					
Interpretation					MCR
Results are consistent with a diagnosis of Beckwith Wiedemann syndrome (BWS). This patient is at risk for developing clinical manifestations associated with BWS and should be monitored accordingly. In particular, abnormal methylation of H19 is associated with an increased risk for embryonal tumors. Appropriate screening procedures should be considered.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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.

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.

For research use only.

Reviewed By	Sheila Marie Wilson	MCR
Release Date	17 Oct 2012 14:10	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.