MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, BWS	L3MRNG9164795	34	F	G9164795

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
02/16/11 11:27 A	02/16/11 11:27 A	03/10/11 02:23 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

Beckwith-Wiedemann Syndrome, B — **REPORTED: 02/16/11 11:32 A**

Specimen	**Blood**	**MCR**
Specimen ID	**1035195**	**MCR**
Order Date	**16 Feb 2011 11:28**	**MCR**

Reason For Referral — **MCR**

Suspected diagnosis of Beckwith-Wiedemann syndrome (BWS).
Analyze the BWS critical region for abnormal methylation.

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 Beckwith-Wiedemann syndrome (BWS) critical region.

Result — **MCR**

LIT1: normal methylation
H19: normal methylation

Interpretation — **MCR**

These results decrease the likelihood but do not rule out the diagnosis of Beckwith-Wiedemann syndrome (BWS). Up to 38% of patients with a clinical diagnosis of BWS do not have a detectable LIT1 (KCNQ1OT1) or H19 methylation abnormality. 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.

** Perform Site Legend on last page of report*

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TESTING, BWS	Final	02/16/11 11:27 A

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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.
This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Reviewed By MCR
Michelle Dawn Fjerstad
Release Date 16 Feb 2011 11:29 MCR

* PERFORMING SITE

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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TESTING, BWS	Final	02/16/11 11:27 A