MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,ME2KM	SA00047690	46	M	SA00047690
Ordering Phys				**DOB** 06/14/1966
Client Order # SA00047690	**Account Information**			**Report Notes**
Collected 07/13/2012 13:47	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/13/2012 14:48				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MECP2 Gene, Known Mutation			REPORTED 07/13/2012 14:28		
Specimen		Blood			MCR
Specimen ID		1038374			MCR
Order Date		13 Jul 2012 14:17			MCR
Reason For Referral					MCR

Family history of Rett syndrome or other MECP2-related disorder. Test for the presence of a mutation in the MECP2 gene.

Method MCR

Fluorescent DNA sequence analysis was used to test for the presence of the p.P379S (c.1135C>T) alteration in exon 4 of the MECP2 gene. GenBank accession number NM_004992 (MECP2A) will be utilized in reporting all alterations except those identified in exon 1. Exon 1 alterations will be reported using GenBank accession number NM_001110792 (MECP2B). Testing was performed for this specific alteration because it was previously identified in an affected family member of this individual.

Result MCR

The p.P379S alteration was NOT detected.

Interpretation MCR

Absence of the mutation previously identified for an affected family member indicates that this individual's risk to be a carrier of, or to develop symptoms associated with an MECP2-related disorder is less than 1%.

This assay does not rule out the presence of other mutations within this gene. Errors in the diagnosis or pedigree provided to us may lead to an erroneous interpretation of test results.

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 findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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.					
Extraction Performed?		Yes			MCR
Reviewed By		Heather Lynn Owen			MCR
Release Date		13 Jul 2012 14:25			MCR

* Performing Site:

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

TEST SITE