Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,ME2MS A	SA00058691	46	F	SA00058691
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058691	**Account Information**			**Report Notes**
Collected 06/02/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/05/2013 08:56				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MECP2 Gene, Full Gene Analysis					
RECEIVED: 06/03/2013 15:51 **REPORTED:** 06/04/2013 15:07					
Specimen		Blood			MCR
Specimen ID		1062021			MCR
Order Date		04 Jun 2013 11:29			MCR
Reason For Referral					MCR
Possible diagnosis of Rett syndrome or other MECP2-related disorder. Test for the presence of a mutation in the MECP2 gene.					
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the MECP2 gene. Additionally, gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in this gene. GenBank accession number NM_004992.3 (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_001110792.1 (MECP2B).					
Result					MCR
The following heterozygous sequence change was detected: Exon: 4 DNA change: c.473C>T Amino Acid change: p.T158M (Thr158Met) Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known deleterious mutation. This result is consistent with a diagnosis of an MECP2-related disorder (e.g. Rett syndrome,variant or atypical Rett syndrome, severe neonatal encephalopathy) for this individual. Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for an MECP2-related disorder is available at Mayo Medical Laboratories by ordering ME2KM/89285 MECP2 Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test. 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					

Performing Site Legend on Last Page of Report

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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. 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
MLPA Performed?		Yes.			MCR
Reviewed By Emily Christine Lauer					MCR
Release Date		04 Jun 2013 15:05			MCR

* Performing Site:

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