

Patient Name SAMPLEREP,ME2MS N	Patient ID SA00058690	Age 46	Gender F	Order # SA00058690
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00058690	Account Information C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			Report Notes
Collected 06/02/2013 00:00				
Printed 06/05/2013 08:55				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MECP2 Gene, Full Gene Analysis					
RECEIVED: 06/03/2013 15:51 REPORTED: 06/04/2013 15:03					
Specimen		Blood			MCR
Specimen ID		1062020			MCR
Order Date		04 Jun 2013 11:29			MCR
Reason For Referral		Possible diagnosis of Rett syndrome or other MECP2-related disorder. Test for the presence of a mutation in the MECP2 gene.			MCR
Method		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).			MCR
Result		A mutation was NOT detected.			MCR
Interpretation		This result reduces the likelihood, but does not rule out the diagnosis of an MECP2-related disorder (e.g. Rett syndrome, variant or atypical Rett syndrome, severe neonatal encephalopathy) for this individual. We predict that some individuals who have a diagnosis of an MECP2-related disorder may have a mutation that is not identified by the methods described above (e.g. promoter mutations, deep intronic mutations, etc.).			MCR
		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

Patient Name SAMPLEREP,ME2MS N	Collection Date and Time 06/02/2013 00:00	Report Status Final
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* 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
MLPA Performed?	Yes.		MCR
Reviewed By			MCR
Emily Christine Lauer			
Release Date	04 Jun 2013 15:01		MCR

* Performing Site:

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