

1-800-533-1710

PATIENT NAME TESTING, QA89284		PATIENT NUMBER		AGE 33	SEX F	ACCESSION # G9134774
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 09/15/09 02:23 P DATE TIME	RECEIVED 09/15/09 02:23 P DATE TIME	REPORT PRINTED 11/11/09 01:29 P DATE TIME		SPECIMEN INFORMATION 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 *
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MECP2 Gene, Full Gene Analysis

Specimen	Blood	MCR
Specimen ID	1033341	MCR
Order Date	06 Oct 2009 09:23	MCR
Reason For Referral		MCR

Not Provided. Test for the presence of a mutation within the MECP2 gene.

Method		MCR
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Fluorescent DNA sequence analysis was performed to test for the presence of mutations in all 4 exons of the MECP2 gene. Additionally, gene dosage analysis (MLPA) is used to test for the presence of large deletions and duplications in this 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).

Result		MCR
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A mutation was NOT detected.

Interpretation		MCR
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This result reduces the likelihood, but does not rule out the diagnosis of an MECP2 related disorder (e.g. Rett syndrome, variant and atypical Rett syndrome) 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. mutations in the promoter, intron, etc.). A genetic consultation may be of benefit. A list of common polymorphisms identified for this patient is available upon request.

CAUTIONS:

Test results should be interpreted in context of clinical

* Perform Site Legend on last page of report

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<p>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.</p> <p>Extraction Performed? Yes MCR MLPA Performed? Yes. MCR See #89854, MECP2 Large Deletion/Duplication, MLPA, for billing information.</p> <p>Reviewed By Keri Jane Kruckeberg MCR Release Date 06 Oct 2009 09:24 MCR</p>			

* 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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