

Laboratory Service Report

1-800-533-1710

| Patient Name SAMPLEREPORT,ME2KM A | Patient ID SA00058760 | Age 46 | Gender F | Order # SA00058760 |
|--------------------------------------|--|-----------|-------------|---------------------------|
| Ordering Phys CLIENT,CLIENT | | | • | DOB 06/10/1966 |
| Client Order # SA00058760 | Account Information | | | Report Notes |
| Collected 06/04/2013 00:00 | C7028846-DLMP Rochester 3050 Superior Drive | | | |
| Printed 06/11/2013 15:23 | Rochester, MN 55901 | | | |

| | | | | Reference | Perform |
|--|-----------------------|-------------------|-------|-----------|---------|
| Test | Flag | Results | Unit | Value | Site* |
| MECP2 Gene, Known Mutation | | | | | |
| RECEIVED: 06/04/2013 17:37 REPOR | red: 06/10/201 | l3 15:06 | | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1062042 | | | MCR |
| Order Date | | 05 Jun 2013 08: | 43 | | MCR |
| Reason For Referral | | | | | MCR |
| Family history of Rett syn | ndrome or othe | er MECP2-related | | | |
| disorder. Test for the pr | resence of a m | mutation in the M | MECP2 | | |
| gene. | | | | | |
| Method | | | | | MCR |
| Fluorescent DNA sequence a | analysis was u | used to test for | the | | |
| presence of the p.T158M (| c.473C>T) alte | eration in exon 4 | 4 of | | |
| the MECP2 gene. GenBank a | accession numb | per NM_004992.3 | | | |
| (MECP2A) will be utilized | | | | | |
| except those identified in | | | | | |
| be reported using GenBank | | | | | |
| (MECP2B). Testing was peri | | _ | | | |
| because it was previously | | n an affected fam | nily | | |
| member of this individual | | | | | |
| Result | | | | | MCR |
| The following heterozygous | s sequence cha | ange was detected | : £ | | |
| Exon: 4 | | | | | |
| DNA change: c.473C>T | | | | | |
| Amino Acid change: p.T158 | , | | | | |
| Classification: DELETERIOU | JS | | | | |
| Interpretation | | | | | MCR |
| This alteration is a known | n deleterious | mutation. | | | |
| | | | | | |

This result is consistent with positive carrier status of an MECP2 mutation for this individual. This interpretation assumes this individual is healthy and not clinically affected with an MECP2-related disorder. MECP2 mutations have variable clinical significance (i.e. asymptomatic carriers, variant or atypical Rett syndrome, or classic Rett syndrome). This individual is at risk to have a child affected with an MECP2-related disorder.

Since a mutation has been identified, genetic testing of other at-risk family members is possible.

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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|---|--------------------------|------------------------------|--|--|
| Patient Name | Collection Date and Time | Report Status | | |
| SAMPLEREPORT,ME2KM A | 06/04/2013 00:00 | Final | | |
| Page 1 of 2 | | >> Continued on Next Page >> | | |

^{*} Report times for Mayo performed tests are CST/CDT



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Test Flag Results Unit Value Site*

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.

Yes.

Laboratory developed test.

Extraction Performed?
Reviewed By

Emily Christine Lauer

Release Date 10 Jun 2013 15:05

MCR MCR

MCR

* Performing Site:

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

| Patient Name | Collection Date and Time | Report Status | |
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| SAMPLEREPORT,ME2KM A | 06/04/2013 00:00 | Final | |
| Page 2 of 2 | | ** End of Report ** | |

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