MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| TESTINGRNV,APO1S | SA00048251 | 45 | M | SA00048251 |
| **Ordering Phys** | | | | **DOB** 06/15/1967 |
| **Client Order #** SA00048251 | **Account Information** | | | **Report Notes** |
| **Collected** 08/07/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 08/31/2012 14:44 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **APOA1 Gene, Full Gene Analysis** | | | REPORTED 08/08/2012 08:07 | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1038543 | | | MCR |
| Order Date | | 07 Aug 2012 08:33 | | | MCR |
| Reason For Referral | | | | | MCR |

Possible diagnosis of apolipoprotein A-I (APOA1) associated familial amyloidosis. Test for the presence of a mutation in the APOA1 gene.

Method MCR

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the APOA1 gene. Mutation nomenclature is based on GenBank accession number; NM_000039.1.

Result MCR

The following sequence change was detected:
Exon: 2
DNA change: c.148G>C
Amino acid change: p.G50R (Gly50Arg)
Legacy nomenclature: p.G26R (Gly26Arg)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of APOA1 associated familial amyloidosis.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for APOA1 associated familial amyloidosis is available at Mayo Medical Laboratories by ordering APO1K/60724 APOA1 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 in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:

***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. | | | | | |
| 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. | | | | | |
| 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. | | | | | |
| Reviewed By<br>W Edward Highsmith Jr., PhD | | | | | MCR |
| Release Date | | 08 Aug 2012 08:05 | | | MCR |

* Performing Site:

| | | |
|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus<br>200 First St SW Rochester, MN 55905 | Lab Director: Franklin R. Cockerill, III, M.D. |



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