MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| TESTINGRNV,GSNMS | SA00048172 | 17 | M | SA00048172 |
| **Ordering Phys** | | | | **DOB** 06/15/1995 |
| **Client Order #** SA00048172 | **Account Information** | | | **Report Notes** |
| **Collected** 08/01/2012 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 08/30/2012 15:18 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **GSN Gene, Full Gene Analysis** | | REPORTED 08/08/2012 08:24 | | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1038507 | | | MCR |
| Order Date | | 01 Aug 2012 09:37 | | | MCR |
| Reason For Referral | | | | | MCR |
| Possible diagnosis of amyloidosis V. Test for the presence of a mutation in the gelsolin (GSN) 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 GSN gene. Mutation nomenclature is based on GenBank accession number; NM_000177.4. | | | | | |
| Result | | | | | MCR |
| A mutation was NOT detected. | | | | | |
| Interpretation | | | | | MCR |

This result does not rule out the diagnosis of familial amyloidosis since disease causing mutations in other genes (i.e. transthyretin, apolipoprotein AI, apolipoprotein AII, fibrinogen alpha chain, lysozyme, or cystatin C) have been described. This assay does not rule out the presence of disease causing mutations in other genes that are associated with familial amyloidosis.

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:
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

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| 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 08:20 | | | 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. |



* Report times for Mayo performed tests are CST/CDT