

Patient Name TESTINGRNV,LYZKM	Patient ID SA00048317	Age 13	Gender M	Order # SA00048317
Ordering Phys			DOB 06/15/1999	
Client Order # SA00048317	Account Information			Report Notes
Collected 08/09/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/30/2012 15:27				

Test	Flag	Results	Unit	Reference Value	Perform Site*
LYZ Gene, Known Mutation			REPORTED	08/09/2012 13:49	
Specimen		Blood			MCR
Specimen ID		1038573			MCR
Order Date		09 Aug 2012 13:47			MCR
Reason For Referral		Family history of LYZ-related familial visceral amyloidosis. Test for the presence of a mutation in the LYZ gene.			MCR
Method		DNA sequence analysis was used to test for the presence of the c.388T>C (p.W130R) mutation in exon 4 of the LYZ gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number; NM.000239.			MCR
Result		The following sequence change was detected: Exon: 4 DNA change: c. 388T>C Amino acid change: p.W130R (Trp130Arg) Legacy nomenclature: p.W112R (Trp112Arg) Classification: DELETERIOUS			MCR
Interpretation		This alteration is a known deleterious mutation. This result is consistent with a diagnosis of LYZ-related familial visceral amyloidosis. Since a mutation has been identified, testing of 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 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.			MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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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

Melody Elizabeth Kimball

MCR

Release Date

09 Aug 2012 13:48

MCR

* Performing Site:

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