

Patient Name SAMPLEREP,LDLRK	Patient ID 0000180999	Age 46	Gender F	Order # 0000180999
Ordering Phys				DOB 06/10/1966
Client Order # 0000180999	Account Information			Report Notes
Collected 03/14/2013 22:30	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 07/01/2013 11:17				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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LDLR Gene, Known Mutation
RECEIVED: 03/14/2013 22:30 **REPORTED:** 07/01/2013 11:13

LDLR Gene, Known Mutation

Result

MCR

One copy of the LDLR Exon 4, nucleotide c.501C>A, amino acid p.Cys167X (p.C167X) familial mutation was detected in this individual.

Interpretation

MCR

The LDLR p.Cys167X mutation was reportedly identified at an outside laboratory in a family member of this individual. The presence of this mutation, therefore, suggests that this individual is at risk for development and/or exacerbation of features of familial hypercholesterolemia (FH). Appropriate surveillance procedures and/or management strategies should be considered.

Since a mutation has been identified in the LDLR gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the on-line test catalog at www.mayomedicallaboratories.com for information about how to order the test for LDLR Gene, Known Mutation (89393). Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the LDLR gene (GenBank number NM_000527.3) which was previously identified in an affected family member of this individual.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available from the lab upon request.

CAUTIONS:

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Rare polymorphisms could interfere with test results. 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.

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP,LDLRK	Collection Date and Time 03/14/2013 22:30	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

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Reviewed By LDLR Known Mutation Sequencing Sequencing		Jennifer Herman Performed			MCR MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Page 2 of 2		** End of Report **

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