



Patient ID 0000180999	Patient Name SAMPLEREPOR, LDLRK	Birth Date 1966-06-10	Gender F	Age 46
Order Number 0000180999	Client Order Number 0000180999	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Mar 2013 22:30		

LDLR Gene, Known Mutation

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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.

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.

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.

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.

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).

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA. Laboratory developed test.

Reviewed By

MCR

Jennifer Herman

ADDITIONAL INFORMATION

Fluorescent DNA sequence analysis was used to test for the presence of a specific mutation in the LDLR gene (GenBank

LDLR Known Mutation Sequencing

Sequencing

MCR

Performed

Received: 14 Mar 2013 22:30

Reported: 01 Jul 2013 11:13

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905