

Patient ID 0000180994	Patient Name SAMPLEREPOR, LDLRK	Birth Date 1966-06-10	Gender F	Age 46
Order Number 0000180994	Client Order Number 0000180994	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Mar 2013 22:31		

LDLR Gene, Known Mutation

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Result

MCR

The LDLR Exon 4, nucleotide c.501C>A, amino acid p.Cys167X (p.C167X) familial mutation was not detected in this individual.

Interpretation

MCR

The LDLR p.Cys167X (p.C167X) mutation was previously identified in a family member with features of a familial hypercholesterolemia (FH). Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having FH.

ADDITIONAL INFORMATION

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.

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

LDLR Known Mutation Sequencing

Sequencing

MCR

Performed

Received: 14 Mar 2013 22:31

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