



Patient ID 0000181003	Patient Name SAMPLEREPOR, LDLRS	Birth Date 1966-06-10	Gender F	Age 46
Order Number 0000181003	Client Order Number 0000181003	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Mar 2013 22:33		

LDLR, Full Gene Sequence

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Interpretation

MCR

This result does not rule out the diagnosis of familial hypercholesterolemia (FH) in this patient. Some individuals who have features of FH may have a mutation in LDLR or another gene (e.g. APOB, PCSK9) that is not identified by the methods described above. Approximately 10–15% of LDLR mutations are due to large genomic rearrangements which generally cannot be detected via the sequencing methodology utilized here. If applicable, 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 Familial Hypercholesterolemia, LDLR Large Deletion/Duplication (89073) and/or Apolipoprotein B-100 Molecular Analysis, R3500Q and R3500W (89097).

ADDITIONAL INFORMATION

LDLR full gene sequencing was performed via fluorescent DNA sequence analysis of a portion of the promoter, 18 exons, and exon/intron boundaries of the LDLR gene (GenBank number NM_000527.3; build hg19).

A genetic consultation may be of benefit.

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

CAUTIONS:

Rare variants may be present 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 the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing. Laboratory developed test.

Reviewed By

MCR

Jennifer Herman

Result

MCR

A mutation was not detected in LDLR.

LDLR Gene Sequencing

Sequencing

MCR

Performed

Received: 14 Mar 2013 22:33

Reported: 01 Jul 2013 11:14

Performing Site Legend

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