



Patient ID SA00062772	Patient Name TESTINGRNV, LDLM AB	Birth Date 1980-01-21	Gender F	Age 33
Order Number SA00062772	Client Order Number SA00062772	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Sep 2013 09:00		

LDLR Large Del/Dup

Interp

MCR

This result is consistent with a diagnosis of heterozygous familial hypercholesterolemia.

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 mayomedicallaboratories.com for information about how to order the test for LDLR Gene, Large Del/Dup (89073).

ADDITIONAL INFORMATION

Large deletion/duplication analysis of a portion of the promoter and all 18 exons of the LDLR gene (GenBank number NM_000527.3) was performed via multiplex ligation-dependent probe amplification (a PCR-based method).

A genetic consultation may be of benefit.

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.

Reviewed By

MCR

Jamie Brufat

Result

MCR

One copy of a large genomic deletion of Exon 10 in LDLR was detected (del Exon 10).

Received: 25 Sep 2013 14:01

Reported: 25 Sep 2013 14:53

Performing Site Legend

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