

Patient Name SAMPLEREP,ADHP N	Patient ID SA00059527	Age 47	Gender F	Order # SA00059527
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00059527	Account Information			Report Notes
Collected 06/27/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 09/16/2013 08:16				

Test	Flag Results	Unit	Reference Value	Perform Site*
------	--------------	------	-----------------	---------------

FH/ADH Genetic Reflex Panel
RECEIVED: 07/03/2013 13:30 **REPORTED:** 09/04/2013 09:14

FH/ADH Genetic Interpretation

MCR

These results do not rule out the diagnosis of Familial Hypercholesterolemia (FH) or Familial Defective ApoB-100 (FDB) in this patient. Some individuals who have features of FH or FDB and involvement of LDLR or APOB may have a mutation that is not identified by the methods used here. In addition, some individuals with features of FH or FDB may have involvement of a gene other than LDLR or APOB (e.g. PCSK9).

A three-tiered testing approach was used to identify mutations within the APOB (GenBank number NM_000384.2) and LDLR (GenBank number NM_000527.3) genes that are associated with autosomal dominant hypercholesterolemia [familial defective apoB-100 (FDB) and familial hypercholesterolemia (FH), respectively]: Tier 1 consists of direct mutation analysis for the common APOB mutations p.R3500W and p.R3500Q following polymerase chain reaction (PCR) amplification and allele-specific primer extension. Tier 2 consists of LDLR full gene sequencing via fluorescent DNA sequence analysis of the promoter, 18 exons, and exon/intron boundaries of the LDLR gene. Tier 3 consists of large deletion/duplication analysis of the promoter and all 18 exons of the LDLR gene via multiplex ligation-dependent probe amplification (a PCR-based method).

The tiered testing was performed sequentially. If a mutation was identified in a tier, then testing was stopped and subsequent tier(s) were not performed. If a mutation was not identified in a tier, then testing was continued to include the next tier.

A genetic consultation may be of benefit.

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

CAUTIONS:

Rare polymorphisms exist 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

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP,ADHP N	Collection Date and Time 06/27/2013 00:00	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

Patient Name SAMPLEREP,ADHP N	Patient ID SA00059527	Age 47	Gender F	Order # SA00059527
Ordering Phys CLIENT,CLIENT				DOB 06/10/1966
Client Order # SA00059527	Account Information		Report Notes	
Collected 06/27/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 09/16/2013 08:16				

Test	Flag Results	Unit	Reference Value	Perform Site*
<p>clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.</p> <p>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.</p> <p>Laboratory developed test.</p>				
Reviewed By	Jamie Bruflat			MCR
APOB Genotype				MCR
APOB Genotype (Result)	Neither the p.R3500Q nor p.R3500W mutation was detected in APOB.			
	Laboratory developed test.			

LDLR Large Del/Dup
RECEIVED: 07/03/2013 13:30 **REPORTED:** 09/04/2013 09:14

Result

 A mutation was not detected in LDLR.
 For research use only.

MCR

LDLR, Full Gene Sequence
RECEIVED: 07/03/2013 13:30 **REPORTED:** 09/04/2013 09:12

Result

 A mutation was not detected in LDLR.
 Laboratory developed test.

MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
-----	-----------------------------------------------------------------------------------------	---------------

Patient Name SAMPLEREP,ADHP N	Collection Date and Time 06/27/2013 00:00	Report Status Final
Page 2 of 2		** End of Report **

* Report times for Mayo performed tests are CST/CDT