Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,ADHP A	SA00059528	47	F	SA00059528
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00059528	**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 09:18					
FH/ADH Genetic Interpretation					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).
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.

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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.					
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.					
Laboratory developed test.					
Reviewed By		Jamie Bruflat			MCR
APOB Genotype					
APOB Genotype (Result)					MCR
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 09:17

Result					MCR
One copy of a large genomic deletion of Exon 10 in LDLR was detected (del Exon 10).					
For research use only.					

LDLR, Full Gene Sequence
RECEIVED: 07/03/2013 13:30 **REPORTED:** 09/04/2013 09:16

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

* Performing Site:

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