Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,ADHP	SA00066671	47	F	SA00066671
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00066671	**Account Information**			**Report Notes**
Collected 04/02/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 04/04/2014 14:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
FH/ADH Genetic Reflex Panel					
RECEIVED: 04/03/2014 13:14 **REPORTED:** 04/03/2014 13:36					
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). Please refer to family number <New Edit Field> when ordering testing on family members of this individual.
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

Performing Site Legend on Last Page of Report

TEST SITE

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Test	Flag	Results	Unit	Reference Value	Perform Site*

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.

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 CHRISTINE THOE 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: 04/03/2014 13:14 **REPORTED:** 04/03/2014 13:40

Interp 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).
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:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Rare polymorphisms could interfere with test 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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Client Order # SA00066671	**Account Information**			**Report Notes**
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Printed 04/04/2014 14:02				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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.					
Reviewed By		CHRISTINE THOE			MCR
Result					MCR
A mutation was not detected in FBN1.					

LDLR, Full Gene Sequence
RECEIVED: 04/03/2014 13:14 **REPORTED:** 04/03/2014 13:40

LDLR, Full Gene Sequence

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, Known Mutation (81183). Please refer to family number <New Edit Field> when ordering testing on family members of this individual. 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).

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. If results obtained do not match the clinical findings, additional testing should be considered.

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.

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Reviewed By		CHRISTINE THOE			MCR
Result					MCR
One copy of the following mutation was detected in LDLR: , nucleotide , amino acid .					
LDLR Gene Sequencing					
Sequencing		Performed			MCR

* Performing Site:

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

TEST SITE