Patient ID	Patient Name		Birth Date	Gender	Age
SA00066670	SAMPLEREPORT, ADHP		1966-06-10	F	47
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00066670	SA00066670	Client, Client			
Account Information		Collected			
C7028846 DLMP Rochester		02 Apr 2014 00:00			

FH/ADH Genetic Reflex Panel

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).

ADDITIONAL INFORMATION

A three-tiered testing approach was used to identify variants within the APOB (GenBank number NM_000384.2; build hg19) and LDLR (GenBank number NM_000527.4; build hg19) genes that are associated with autosomal dominant hypercholesterolemia [familial defective apoB-100 (FDB) and familial hypercholesterolemia (FH), respectively]:

Tier 1 consists of direct analysis for the common APOB pathogenic variants p.R3500W and p.R3500Q (HGVS nomenclature: c.10579C>T, p.R3527W and c.10580G>A, p.R3527Q) following polymerase chain reaction (PCR) amplification and allele-specific primer extension.

Tier 2 consists of LDLR gene sequencing via fluorescent DNA sequence analysis of the promoter and all 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 pathogenic or likely pathogenic variant was identified in a tier, then testing was stopped and subsequent tier(s) were not performed. If a pathogenic or likely pathogenic variant 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 benign variants identified for this patient is available from the lab upon request. These variants are not expected to impact this patient's clinical presentation or management.

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

CHRISTINE THOE

Performing Site Legend

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

Patient ID	Patient Name		Birth Date	Gender	Age
SA00066670	SAMPLEREPORT, ADHP		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00066670	SA00066670	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		02 Apr 2014 00:00			

APOB Genotype

APOB Genotype (Result)

MCR

Neither the p.R3500Q nor p.R3500W mutation was detected in APOB.

ADDITIONAL INFORMATION

Laboratory developed test.

Received: 03 Apr 2014 13:13 **Reported:** 03 Apr 2014 13:29

Performing Site Legend

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