MAYO CLINIC
Mayo Medical Laboratories

1-800-533-1710

APOB

Apolipoprotein B-100 Molecular Analysis, R3500Q and R3500W

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059529	SAMPLEREPORT, APOB N		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059529	SA00059529	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		27 Jun 2013 00:00			

APOB Genotype

Reason for Referral

MCR

Reason for referral was not provided by the requesting physician.

Method

MCR

A PCR-based method was used to test for the presence of the R3500Q and R3500W mutations within the APOB gene.

Result

MCR

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

Interpretation

MCR

This result does not rule out the diagnosis of familial defective apoB-100 (FDB). While the R3500Q and R3500W APOB mutations are the most frequently observed mutations associated with FDB, some individuals who have a diagnosis of FDB may have APOB mutations that are not identified by the methods described above.

Because of phenotypic overlap with familial hypercholesterolemia, genetic testing of the LDLR gene may be important to consider in this individual. Please contact the laboratory at 1–800–533–1710 or the on-line test catalog at mayomedicallaboratories.com for information regarding LDLR genetic testing. For future reference, APOB and LDLR genetic analysis can be ordered as a reflexive panel (FH/ADH Genetic Reflex Panel, test 83375).

ADDITIONAL INFORMATION

Direct analysis for the common pathogenic APOB variants p.R3500W and p.R3500Q (HGVS nomenclature: c.10579C>T, p.R3527W and c.10580G>A, p.R3527Q using transcript NM_000384; build hg19) was performed following polymerase chain reaction (PCR) amplification and allele specific primer extension.

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. Laboratory developed test.

Reviewed By

MCR

Jamie Bruflat

Received: 03 Jul 2013 13:31 **Reported:** 04 Sep 2013 09:45

Performing Site Legend

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