

Patient ID SA00059530	Patient Name SAMPLEREPOR, APOB A	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00059530	Client Order Number SA00059530	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jun 2013 00:00		

APOB Genotype

Reason for Referral

MCR

Patient has clinical features consistent with familial defective apoB-100. Test for the presence of the R3500Q and R3500W mutations within the APOB gene.

Method

MCR

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

Result

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One copy of the p.R3500Q mutation was detected.

Interpretation

MCR

This result is consistent with a diagnosis of heterozygous familial defective apoB-100.

Since a mutation has been identified in the APOB gene in this individual, genetic testing of other family members is recommended.

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 09:47

Performing Site Legend

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