

Patient ID SA00076157	Patient Name TESTING, REPORTS	Birth Date 1986-01-01	Gender F	Age 30
Order Number SA00076157	Client Order Number SA00076157	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Jun 2016 00:00		

Apolipoprotein E Genotyping, B

Result Summary

COMPLEX (SEE RESULT AND INTERPRETATION)

Result

Genotype: e4/e4

Interpretation

The e4 allele leads to down regulation of the LDL receptor in the liver and is associated with increased total cholesterol, triglycerides, and LDL. In a large meta-analysis, the presence of the e3/e4 or e4/e4 genotype was reported to confer a 42% increased risk of cardiovascular disease (1).

The APOE gene is also a known susceptibility gene for Alzheimer disease (AD). The e4 allele is associated with an increased risk for AD, particularly late-onset disease, in a dose dependent manner (2). This risk is also influenced by other factors. It is estimated that individuals with the APOE e3/e4 genotype have a four-fold relative risk for Alzheimer disease, while homozygotes for e4 allele have a 12-fold relative risk. Several studies have suggested a protective effect of the APOE e2 allele.

The APOE e4 allele, however, is neither sufficient nor necessary for the development of Alzheimer disease. Approximately 50% of individuals with Alzheimer disease carry an e4 allele and many individuals who have an e4 allele will never develop Alzheimer disease. The use of APOE analysis for predictive testing for Alzheimer disease is not currently recommended by the American College of Medical Genetics due to limited clinical utility and poor predictive value (3, 4).

Less common APOE variants that do not alter a restriction site for HhaI will not be detected by this assay.

REFERENCES

1. Ann Int Med. 2004; 141:137-47 (PMID 15262670)
2. JAMA 1997; 278:1349-1356 (PMID 9343467)
3. Genet Med 2011; 13:597-605 (PMID 21577118)
4. American College of Medical Genetics and Genomics 2015 July 10. Retrieved from www.choosingwisely.org/societies/american-college-of-medical-genetics-and-genomics/

MCR

MCR

1 MCR

ADDITIONAL INFORMATION

A PCR-based assay, including Hha I digestion of the amplified product, was utilized to identify the three common APOE alleles (epsilon2, epsilon3, and epsilon4). An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Reason for Referral

Test for the presence of the e2, e3, and e4 alleles in the APOE gene.

Specimen

WB Whole Blood

Released By

EMILY LAUER

Received: 09 Jun 2016 14:41

Reported: 10 Jun 2016 16:07

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



Patient ID SA00076157	Patient Name TESTING, REPORTS	Birth Date 1986-01-01	Gender F	Age 30
Order Number SA00076157	Client Order Number SA00076157	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Jun 2016 00:00		

Test
Environment
ETBM Template

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292