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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 49 years Male

Specimen Collected: 08-Mar-22 11:02

Apolipoprotein E (APOE) Genotyping, CR | **Received: 08-Mar-22 11:02** **Report/Verified: 10-Mar-22 14:17**

Procedure	Result	Units	Reference Interval
APOE Specimen	Whole Blood		
APOE Cardiovascular Risk,Genotype	e2/e3 * f1 i1		

Result Footnote

f1: APOE Cardiovascular Risk, Genotype

Indication for testing: Assess genetic risk for type III hyperlipoproteinemia.

HETEROZYGOUS APO e2/e3: This genotype is not significantly associated with an increased risk for type III hyperlipoproteinemia

This result has been reviewed and approved by Rong Mao, M.D.

Test Information

i1: APOE Cardiovascular Risk, Genotype

BACKGROUND INFORMATION: Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk

Characteristics: Hyperlipoproteinemia III (HPL III) is characterized by increased cholesterol and triglyceride levels, presence of B-VLDL, xanthomas, and premature vascular disease including coronary heart disease (CHD) and peripheral artery disease.

Incidence of HPL III: Approximately 1 in 5,000.

Inheritance of HPL III: Multifactorial; greater than 90 percent of affected individuals are homozygous for the e2 allele but other factors such as diabetes and hypothyroidism also play a large role in development of disease.

Penetrance: 1 to 5 percent of individuals homozygous for the e2 will develop HPL III.

Cause: 2 copies of the e2 allele provides supporting evidence for a diagnosis of HPL III in a symptomatic individual but e2 homozygosity is neither necessary nor sufficient for HPL III.

Variants Tested: APOE gene alleles, e2 (c.388T, p.130Cys and c.526C>T, p.Arg176Cys), e3 (c.388T, p.130Cys and c.526C, p.176Arg), e4 (c.388T>C, p.Cys130Arg and c.526C, p.176Arg).

Clinical Sensitivity: 90 percent of individuals with HPL III are homozygous for the e2 variant.

Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Only the e2, e3 and e4 variants will be detected. Rare isoforms of APOE will not be detected. If rare alleles are suspected, phenotyping by isoelectric focusing may be indicated. Diagnostic errors can occur due to rare sequence variations.

Test Information

i1: APOE Cardiovascular Risk, Genotype

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.