Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk
ARUP test code 2013337

**APOE Specimen**
Whole Blood

**APOE Cardiovascular Risk, Genotype**
e2/e2

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from all patients (or their legal guardians) prior to pursuing any diagnostic genetic testing or testing to assess carrier status. These forms must be kept on file by the ordering physician. Biochemical and DNA testing patient consent forms can be accessed from ARUP's web site: www.aruplab.com.

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

HOMOZYGOUS APO e2 (e2/e2): This genotype shows strong association with type III hyperlipoproteinemia (HLP III). Although it provides additional evidence for a diagnosis of HLP III in individuals with clinical symptoms, by itself, it is not diagnostic.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.
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.


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

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

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.

This test is performed pursuant to an agreement with Roche Molecular Systems, Inc.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS