

Client: Example Client ABC123  
123 Test Drive  
Salt Lake City, UT 84108  
UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 5/9/1965  
**Gender:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk**

ARUP test code 2013337

APOE Specimen whole Blood

APOE Cardiovascular Risk, Genotype

**e2/e2 \***

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

**H=High, L=Low, \*=Abnormal, C=Critical**

**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.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
APOE Specimen	22-189-103074	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
APOE Cardiovascular Risk, Genotype	22-189-103074	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, \*=Abnormal, C=Critical

Unless otherwise indicated, testing performed at: