**ARUP LABORATORIES | www.aruplab.com**
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories



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

Physician: Doctor, Example

## Patient: Patient, Example

| | |
|---|---|
| **DOB** | Unknown |
| **Gender:** | Unknown |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Apolipoprotein B (APOB) Mutation Detection

ARUP test code 0055654

| | |
|---|---|
| Apolipoprotein B Specimen | Whole Blood |
| Apolipoprotein B Mutation Detection | Homo RW |

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Indication for testing: Diagnostic testing for APOB-associated familial hypercholesterolemia.

R3500Q Negative: The patient is negative for the ApoB Arg3500Gln (G9775A) mutation.

R3500W Homozygous: The patient is homozygous for the Arg3500Trp (C9774T) mutation. This is associated with hypercholesterolemia in familial defective ApoB-100 and an increased risk of coronary artery disease.

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

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

BACKGROUND INFORMATION: Apolipoprotein B (APOB) Mutation Detection

CHARACTERISTICS: Familial defective APOB-100 (FDB) is a disorder which increases an individual's risk for coronary artery disease. FDB is caused by mutations in the APOB gene that prevents the APOB-LDL complex from binding to the LDL receptor.
INCIDENCE: 1.5 percent of familial hypercholesterolemia is due to an APOB gene mutation. The most common mutation, Arg3500Gln, has a frequency of 1/500-1/700 in Caucasian populations of North America and Europe. The rare Arg3500Trp mutation originally described in the Scottish population has also been identified in FDB of Asian descent (approximately 2 percent of FDB patients).
INHERITANCE: Autosomal dominant
PENETRANCE: Heterozygous carriers of an APOB mutation have an increased risk for coronary artery disease, although with a reduced penetrance. 40 percent of males and 20 percent of females heterozygous for these mutations develop coronary artery disease. Homozygotes or compound heterozygotes for R3500Q/R3500W are at greater risk for disease.
CAUSE: Ligand-defective apolipoprotein B-100.
METHODOLOGY: Patient DNA is assayed for Arg3500Gln and Arg3500Trp mutations in the APOB gene by polymerase chain reaction (PCR) and fluorescence monitoring using hybridization probes.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Sensitivity and specificity for detection of this mutation are 99.9 percent.
LIMITATIONS: Mutations in other genes or other mutations in the APOB gene that may cause familial hypercholesterolemia or increased risk for coronary artery disease are not ruled out. Diagnostic errors can occur due to rare sequence variations.

This test is not recommended for nonsymptomatic patients under 18 years of age.

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

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| Apolipoprotein B Specimen | 19-064-115822 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Apolipoprotein B Mutation Detection | 19-064-115822 | 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**