ARUP LABORATORIES | aruplab.com
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Julio C. Delgado, MD, MS, Chief Medical Officer

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108-
USA

Provider: 10082429.0 -TEST,

Patient: **FHNGS, 3**
DOB: 20-Aug-20
Gender: Female
Patient Identifiers: 21160
Visit Number (FIN): 21391
Client Supplied ID:

Specimen Collected: 21-Aug-20 11:27

Familial Hypercholesterolemia by NGS | **Received: 21-Aug-20 11:36** | **Report/Verified: 21-Aug-20 12:09**

	Result	Units	Reference Interval
Familial Hypercholosterolemia Specimen	See Note		
Familial Hypercholesterolemia Interp	Indeterminate [i1]		

Test Information

i1: Familial Hypercholesterolemia Interp

BACKGROUND INFORMATION: Familial Hypercholesterolemia Panel, Sequencing

CHARACTERISTICS: Familial hypercholesterolemia (FH) is the most common inherited cardiovascular disease. It is characterized by markedly elevated low-density lipoprotein cholesterol (LDL-C) and premature atherosclerotic cardiovascular disease (ASCVD). Manifestations include coronary artery disease (CAD), cardiovascular disease (CVD), angina, myocardial infarction, xanthomas, and corneal arcus. Homozygous FH (HoFH) is a less common disorder, resulting from biallelic variants in a dominant FH-associated gene. HoFH is characterized by severe early-onset CAD, aortic stenosis, and high rate of coronary bypass surgery or death by teenage years.

EPIDEMIOLOGY: FH 1/250, HoFH 1/200,000 in the general population.

CAUSE: Pathogenic germline variants in genes associated with FH.

INHERITANCE: Autosomal dominant for LDLR, APOB and PCSK9-associated FH. Autosomal recessive for LDLRAP1-associated FH. HoFH results from biallelic variants in an autosomal dominant FH gene.

PENETRANCE: Estimated at 73-90% in individuals with molecularly confirmed FH; influenced by gene, variant, and non-genetic factors.

CLINICAL SENSITIVITY: Up to 85% for FH.

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Julio Delgado, MD, MS

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Test Information

i1: Familial Hypercholesterolemia Interp

GENES TESTED: APOB, LDLR, LDLRAP1, PCSK9.

METHODOLOGY: Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99% for single nucleotide variants (SNVs) and greater than 93% for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of FH. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants, deep intronic variants, and large deletions/duplications/inversions will not be identified. Deletions/duplications/insertions of any size may not be detected by massive parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Non-coding transcripts were not analyzed.

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