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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/28/1966
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping

ARUP test code 2014599

PNPLA3 Specimen	Whole Blood
PNPLA3 Variant	Negative
PNPLA3 Interpretation	See Note Indication for testing: screening for genetic susceptibility to non-alcoholic fatty liver disease. Negative: This sample is negative for the PNPLA3 c.444C>G; p.I148M variant. This result predicts that this individual is not at increased genetic risk for non-alcoholic fatty liver disease; however, other genetic or environmental factors not detected by this assay may be present. This genotype is also associated with a lower risk for cirrhosis among individuals with alcoholic liver disease. This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping

CHARACTERISTICS: Fatty liver disease is the accumulation of excessive triglycerides in the liver that may cause an inflammatory response, which can progress to fibrosis, cirrhosis, and liver cancer. The c.444C>G; p.I148M variant in the PNPLA3 gene confers an increased risk for the onset and progression of non-alcoholic fatty liver disease (NAFLD). This allele also confers an increased risk for the onset and progression of cirrhosis among individuals with alcoholic liver disease.
INCIDENCE: NAFLD occurs in approximately 20-30 percent of individuals in the US.
G ALLELE FREQUENCY: Varies by ethnicity; Latino 0.57, East Asian 0.38, European 0.23, South Asian 0.22, Africans 0.14.
CAUSE: Risk factors for non-alcoholic fatty liver disease include obesity, diabetes, insulin resistance and genetic risk factors including PNPLA3 c.444C>G; p.I148M.
INHERITANCE: Multifactorial.
CLINICAL SENSITIVITY: Unknown.
VARIANT TESTED: PNPLA3 c.444C>G; p.I148M (rs738409).
METHODOLOGY: Polymerase chain reaction followed by high-resolution melt analysis.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Only the c.444C>G; p.I148M variant in the PNPLA3 gene will be targeted. Diagnostic errors can occur due to rare sequence variations.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
PNPLA3 Specimen	20-156-401995	6/2/2020 1:19:00 PM	6/4/2020 12:49:53 PM	6/7/2020 3:59:00 PM
PNPLA3 Variant	20-156-401995	6/2/2020 1:19:00 PM	6/4/2020 12:49:53 PM	6/7/2020 3:59:00 PM
PNPLA3 Interpretation	20-156-401995	6/2/2020 1:19:00 PM	6/4/2020 12:49:53 PM	6/7/2020 3:59:00 PM

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