

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 7/18/1974
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 **Homozygous ***

PNPLA3 Interpretation

See Note

Indication for testing: screening for genetic susceptibility to non-alcoholic fatty liver disease.

Positive: Two copies of the PNPLA3 c.444C>G; p.I148M variant were detected. This individual has a moderately increased genetic risk for the development and progression of non-alcoholic fatty liver disease. This genotype is also associated with a moderately increased risk for cirrhosis among individuals with alcoholic liver disease. Other genetic or environmental risk factors not detected by this assay may be present.

This result has been reviewed and approved by [REDACTED]

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

Unless otherwise indicated, testing performed at:

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	20-139-400979	5/14/2020 11:19:00 AM	5/18/2020 12:29:36 PM	5/20/2020 5:35:00 PM
PNPLA3 Variant	20-139-400979	5/14/2020 11:19:00 AM	5/18/2020 12:29:36 PM	5/20/2020 5:35:00 PM
PNPLA3 Interpretation	20-139-400979	5/14/2020 11:19:00 AM	5/18/2020 12:29:36 PM	5/20/2020 5:35:00 PM

END OF CHART

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 20-139-400979
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Page 2 of 2 | Printed: 1/29/2021 6:29:54 AM
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