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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 45 years Male

Specimen Collected: 08-Mar-22 10:55

PNPLA3 Genotype | **Received: 08-Mar-22 10:55** **Report/Verified: 14-Mar-22 13:55**

Procedure	Result	Units	Reference Interval
PNPLA3 Specimen	Whole Blood		
PNPLA3 Variant	Negative		
PNPLA3 Interpretation	See Note [f1 i1]		

Result Footnote

f1: PNPLA3 Interpretation

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 Patricia Slev, Ph.D.

Test Information

i1: PNPLA3 Interpretation

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 (PCR) and fluorescence monitoring.
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.

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: PNPLA3 Interpretation

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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