Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping
ARUP test code 2014599

PNPLA3 Specimen: Whole Blood
PNPLA3 Variant: Homozygous *
PNPLA3 Interpretation: See Note

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: 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.

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.

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).
METHODODOLOGY: 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.

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

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