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 Rong Mao M.D.
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).

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

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