New Test 2014599  Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping

Available Now

Additional Technical Information

Methodology: Polymerase Chain Reaction/Fluorescence Monitoring
Performed: Sun-Sat
Reported: 7-10 days

Specimen Required: Collect: Lavender (EDTA).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin
Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: unacceptable

Interpretive Data:
Background Information for 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.

See Compliance Statement C: www.aruplab.com/CS

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.