HOTLINE: Effective August 19, 2019

New Test **3001768** **Pancreatitis (*PRSS1*) Sequencing and Deletion/Duplication** **PRSS1 FGA**

Patient History for Pancreatitis Testing

Additional Technical Information

Out of Pocket Estimator

Methodology: Polymerase Chain Reaction/Sequencing and Multiplex Ligation Dependent Probe Amplification
Performed: Varies
Reported: 2-3 weeks

Specimen Required: Collect: Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Interpretive Data:
Background Information for Pancreatitis (*PRSS1*) Sequencing and Deletion/Duplication
Characteristics: Characteristics of *PRSS1*-related hereditary pancreatitis: Recurrent episodes of pancreatic inflammation that typically begin to present in late childhood, often with signs and symptoms including abdominal pain, nausea, and vomiting. Ultimately, these recurrent episodes of acute pancreatitis progress to permanent damage of the pancreas.
Epidemiology: Incidence of chronic pancreatitis: 5-12 in 100,000 per year
Prevalence of chronic pancreatitis: approximately 50 in 100,000
Inheritance: Autosomal dominant.
Penetrance: Varies geographically; estimated at 80 percent in the US.
Cause: Pathogenic variants in the cationic trypsinogen (*PRSS1*) gene.
Clinical Sensitivity: 15 percent of hereditary pancreatitis is caused by pathogenic *PRSS1* copy number variants or sequence variants.
Methodology: Bidirectional sequencing of *PRSS1* coding regions and intron/exon boundaries and multiplex ligation-dependent probe amplification (MLPA) of the *PRSS1* gene.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants and deep intronic variants will not be detected. The breakpoints of large deletions/duplications will not be determined. Variants in genes other than *PRSS1* are not evaluated.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com
See Compliance Statement C: www.aruplab.com/CS

New York DOH approval pending. Call for status update.

CPT Code(s): 81404, 81479

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