

HOTLINE: Effective August 19, 2019

New Test	3001764	Pancreatitis (<i>SPINK1</i>) Deletion/Duplication	SPINK1 DD
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Patient History for Pancreatitis (*SPINK1*)
Del/Dup



Additional Technical Information

Methodology: Multiplex Ligation-dependent Probe Amplification
Performed: Varies
Reported: 12-14 days

Specimen Required: Collect: Lavender (K₂EDTA), Pink (K₂EDTA), 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

Reference Interval: By Report

Interpretive Data:

Background Information for Pancreatitis (*SPINK1*) Deletion/Duplication:

Characteristics: Hereditary pancreatitis typically presents with recurrent episodes of pancreatic inflammation, abdominal pain, nausea, and vomiting. Ultimately, this evolves into chronic pancreatitis resulting in permanent pancreatic damage.

Epidemiology: Incidence of chronic pancreatitis is 5-12 in 100,000 per year and prevalence is approximately 50 in 100,000.

Inheritance of SPINK1-related pancreatitis: Autosomal dominant.

Penetrance: Variable.

Cause: Pathogenic variants in *SPINK1*, *PRSS1*, *CFTR*, *CASR*, *CTRC*, *CPA1* and *CLDN2* genes are associated with pancreatitis.

Clinical Sensitivity: 6 percent of hereditary pancreatitis is caused by pathogenic *SPINK1* copy number variants.

Methodology: Multiplex ligation-dependent probe amplification (MLPA)

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. Single base pair substitutions, small deletions/duplications, regulatory region and deep intronic variants will not be detected. Deletion/duplication breakpoints will not be determined. Variants in genes other than *SPINK1* will not be detected.

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

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.