

HOTLINE: Effective August 19, 2019

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<b>New Test</b>	<b>3001760</b>	<b>Pancreatitis (<i>PRSSI</i>) Deletion/Duplication</b>	<b>PRSSI DD</b>
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Patient History for Pancreatitis (*PRSSI*)  
Del/Dup



Additional Technical Information

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

**Specimen Required:** Collect: Lavender (K<sub>2</sub>EDTA), Pink (K<sub>2</sub>EDTA), 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 (*PRSSI*) Deletion/Duplication:**

**Characteristics:** Hereditary pancreatitis typically presents in late childhood 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 recessive and possibly digenic.

**Penetrance:** Variable.

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

**Clinical Sensitivity:** Unknown.

**Methodology:** Multiplex ligation-dependent probe amplification (MLPA) of the *PRSSI* gene.

**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 *PRSSI* will not be detected.

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

New York DOH approval pending. Call for status update.

**CPT Code(s):** 81479

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