

Using Genetic Testing for Diagnosis and Risk-Assessment for Chronic Pancreatitis (CP)

Indications:

- Unexplained pancreatitis episode in children
- Recurrent unexplained attacks of acute pancreatitis or chronic pancreatitis, in presence of a positive family history
- Unexplained chronic pancreatitis without a positive family history, after exclusion of other causes such as anatomic anomalies, metabolic disorders, trauma, and inflammatory bowel disease

Benefits:

Genetic testing for CP

- Can confirm a clinical diagnosis of CP and identify the familial mutation(s)
- Can differentiate genetic from alcohol-induced CP, indicating increased risk of pancreatic cancer
- May, in some cases, inform the prognosis based on the nature of the familial mutation
- Can identify carriers of known familial mutations in relatives of patients, alerting them to their increased susceptibility for CP

Background:

- Chronic pancreatitis (CP) leads to irreversible morphological changes that can result in impaired exocrine and/or endocrine pancreatic function and an increased risk of pancreatic cancer.¹
- About 75-80% of CP is estimated to be alcohol induced, about 1-3% is familial, and the remainder idiopathic.²
- About 50% of familial and idiopathic forms of CP have been associated with mutations in the genes *PRSS1*, *SPINK1*, or *CFTR* (2-4).²
- Genetic CP shows earlier age of onset than alcohol-induced CP and higher risk of pancreatic cancer.¹
- Genetic CP typically follows an autosomal dominant inheritance pattern, but penetrance can vary from 80% to less than 1% depending on the exact underlying genetic cause.¹⁻³
 - Gain-of-function variants in *PRSS1* and rare loss-of-function mutations in *SPINK1* can cause CP with high penetrance (30-75%).¹⁻³
 - The common *SPINK1* variant N34S and presence of non-classic cystic fibrosis increase risk of CP by 20 to 40 fold if occurring separately and by up to 900fold if occurring together.

References: 1. Rosendahl J et al (2007) Orphanet Journal of Rare Diseases 2:1-10.
2. Keim V (2008) World J Gastroenterol 14:1011-5. 3. Chen J-M, Ferec C (2009) Annu Rev Genom Human Genet 10:63-87.

Ordering Information: Please see other side.

Ordering Information for Chronic Pancreatitis Testing

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Ordering Information for Single Gene Tests

Gene(s)	Test Code
<i>PRSS1</i>	252773
<i>SPINK1</i>	252780
<i>CFTR</i>	252787

Ordering Information for Multi-Gene Panel

<i>PRSS1, SPINK1, CFTR</i>	252794
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Family Testing (single amplicon)

Family Testing is available for all genes. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.