Hereditary Pancreatitis Panel

Pancreatitis is a painful condition that occurs suddenly when the pancreas is inflamed. It causes digestive problems and intense abdominal pain. There are three types of pancreatitis:

Acute:	Occurs unexpectedly and usually lasts days to weeks
Recurrent Acute:	More than one episode of acute pancreatitis
Chronic:	Continuous pancreatitis-associated inflammation that weakens the pancreas function, often leading to chronic pain and digestive problems; Chronic pancreatitis can also increase the risk of diabetes (uncontrolled blood sugar) and pancreatic cancer.

Hereditary pancreatitis is more likely to be related to genetic risk factors; however, inheritance may be complex. The inherited risk may be the result of one or more pathogenic variants or risk alleles in a pancreatitis-associated gene(s). Environmental and lifestyle factors increase the risk of pancreatitis. These risk factors include, but are not limited to, smoking, alcohol use, and a diet high in fats.

Features of Hereditary Pancreatitis

Genetic testing with the Hereditary Pancreatitis Panel may be appropriate if you have a personal and/or family history of:

- Unexplained acute pancreatitis in early childhood or teenage years
- Recurrent acute pancreatitis of unknown cause, especially before the age of 20
- Chronic pancreatitis, especially before the age of 20

Genes Included on the Hereditary Pancreatitis Panel

PRSS1, CFTR, SPINK1, CTRC, CASR*,* and *CPA1*

Hereditary Pancreatitis Panel Results

Pathogenic variants in the *PRSS1* gene are associated with the highest risk of hereditary pancreatitis, and one pathogenic variant is enough to increase the risk. However, some variants identified by testing with the Hereditary Pancreatitis Panel are considered pancreatitis risk alleles. This means that these variants may increase the risk of pancreatitis but may not be enough to cause pancreatitis alone. When a person has two or more risk alleles, the combination may be consistent with an increased risk of pancreatitis. The inheritance of pancreatitis can be complex and although hereditary pancreatitis may be related to one or more genetic variants, environmental factors still play a role in pancreatitis development. Your test report will provide detailed information about the specific variants identified and their effect on pancreatitis risk.

Some Example Results are Provided in the Table Below

Result	May Increase Pancreatitis Risk (dependent on other risk factors)	Consistent with an Increased Risk of Pancreatitis
1 risk allele in *SPINK1*	✓	
2 risk alleles in *SPINK1*		✓
2 risk alleles: 1 in *SPINK1* and 1 in *CTRC*		✓
1 pathogenic variant in *PRSS1*		✓

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If you have one or more pathogenic variants or risk alleles in a pancreatitis-associated gene(s), your test report will include additional information about the specific variant(s) and the effect these variants may have on your risk for pancreatitis.

If you do not have a pathogenic variant or risk allele in a pancreatitis-associated gene(s), your risk for pancreatitis may still be increased based upon your personal and family histories and other risk factors. This result does not explain a history of pancreatitis but does not completely rule out hereditary pancreatitis. It remains possible that genetic factors influenced your history of pancreatitis but were not detectable by this test. A discussion with your healthcare provider is recommended to determine the most appropriate medical management options.

Medical Management Based on Genetic Test Results

Individuals at risk for pancreatitis should discuss surveillance and risk-reducing options with their healthcare providers.

Recommendations may include:

- Blood analysis
- Imaging exams such as CT imaging, endoscopic ultrasound, ERCP or MRCP
- Risk-reducing medications
- Consideration of pancreatectomy with islet cell auto-transplantation
- Lifestyle changes, such as a low fat diet, increased hydration (water intake), use of vitamins/antioxidants and avoidance of alcohol and tobacco

Regardless of the test result, you may wish to share the information with your family so they may discuss the results with their healthcare providers. If you have one or more variants, family members are at risk to have the same variant(s) and may consider genetic testing to better understand their chance of developing pancreatitis.

**Variants in this gene may be associated with conditions other than pancreatitis. Certain variants in the CASR gene are associated with autosomal dominant hypocalcemia (ADH), familial hypocalcuric hypercalcemia type 1 (FHH) and neonatal severe primary hyperparathyroidism (NSHPT). Certain variants in the CFTR gene are associated with cystic fibrosis (CF) and other CFTR-related disorders (such as chronic pulmonary conditions, sinusitis, and congenital absence of the vas deferens). If you have a risk allele or pathogenic variant in one of these genes, your test report will include additional information.*

Resources

General

American Cancer Society
www.cancer.org

GeneDx
www.genedx.com/oncology

National Cancer Institute
www.cancer.gov

Pancreatitis Resources

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)
www.niddk.nih.gov/health-information/digestive-diseases/pancreatitis

National Pancreas Foundation
www.pancreasfoundation.org

University of Pittsburgh Pancreatitis Research
www.pancreas.org

Find a Genetic Counselor

Canadian Association of Genetic Counsellors
www.cagc-accg.ca

National Society of Genetic Counselors
www.nsgc.org

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