

Understanding Your Positive Pancreatitis Genetic Test Result

INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC**

Result	MUTATION	Your testing shows that you have a pathogenic mutation (disease-causing change in the gene, like a spelling mistake) or variant that is likely pathogenic in a gene that causes an increased risk for pancreatitis. Either result should be considered as positive.
Gene	DEFINITION	Everyone has two copies of each gene, which we randomly inherit from each of our parents. One mutation in this gene may slightly increase your risk to develop pancreatitis.
Pancreatitis	MAY BE PRESENT	Individuals who carry one mutation in this gene are at an increased risk to develop pancreatitis.
Cancer Risks	VARIES	Cancer risks associated with mutations in this gene are not well defined. Individuals with pancreatitis may be at an increased risk for pancreatic cancer.
Management Options	FOR PANCREATITIS	Lifestyle modifications to reduce the chance of pancreatitis may include: smoking cessation and low-fat diet. Talk to your doctor about what options may be right for you.
Management Options	FOR PANCREATIC CANCER	Options for screening and early detection may be available. There are no standard recommendations for pancreatic cancer screening currently available, so it is recommended that you talk to your doctor about what options may be right for you.
Risk Management	VARIES	Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than the general population and is often more frequently performed. It is important to discuss these options with your doctor.
Family Members	50/50 CHANCE	Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the mutation that you carry, and other family members (like your aunts, uncles, cousins) may also inherit it. Your relatives can be tested for this same mutation. Depending on the family history, those who DO NOT have it may NOT have an increased chance (above the general population) to develop pancreatitis. Talk to your healthcare provider about your family history to find out if other family members may benefit from genetic testing.
Next Steps	DISCUSS	It is recommended that you share this information with your family members so they can learn more and discuss this with their healthcare providers.
Reach Out	RESOURCES	<ul style="list-style-type: none"> National Society of Genetic Counselors nsgc.org Genetic Information Nondiscrimination Act (GINA) ginahelp.org

*Pancreatitis is characterized by recurrent attacks of inflammation (swelling) that can gradually cause damage to your pancreas and the surrounding tissue. Pancreatitis can be acute (occurs suddenly), acute-recurrent (occurs suddenly more than once in 6 months), or chronic (lasting longer than 6 months). The impact on your pancreas increases risks of diabetes and pancreatic cancer.

Pancreatitis in the Family

There is a 50/50 random chance to pass on your mutation to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.



Please discuss this information with your healthcare provider. The genetics field is constantly evolving, so updates related to your result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.