

Understanding Your Positive Pancreatitis Genetic Test Result

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

RESULT	The testing found a variant that is either pathogenic (known to be disease-causing) or likely pathogenic (likely to be disease-causing) in a gene known to cause an increased risk for pancreatitis. Either type of variant should be treated as a positive result.
GENES	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. Variants (changes) in certain genes can cause pancreatitis. These gene changes may be passed down in families or be brand new for the person who has pancreatitis. Even if there is no history of pancreatitis in your family, it can still be caused by a change in a gene.
PANCREATITIS	Individuals who carry one pathogenic or likely pathogenic variant in this gene are at an increased risk to develop pancreatitis. 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.
CANCER RISK	Individuals with pancreatitis may be at an increased risk for pancreatic cancer, however specific cancer risk numbers are not well defined for this gene.
WHAT YOU CAN DO	Risk management decisions are very personal and depend on many factors. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
FAMILY	Family members may also be at risk – they can be tested for the same gene mutation that was found in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.
RESOURCES	<ul style="list-style-type: none"> National Society of Genetic Counselors nsgc.org Canadian Association of Genetic Counsellors cagc-accg.ca

Pancreatitis in the Family

There is a 50/50 random chance to pass on your mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth



Please discuss this information with you 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.