

Understanding Your Positive *PALB2* Genetic Test Result

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

4 THINGS TO KNOW

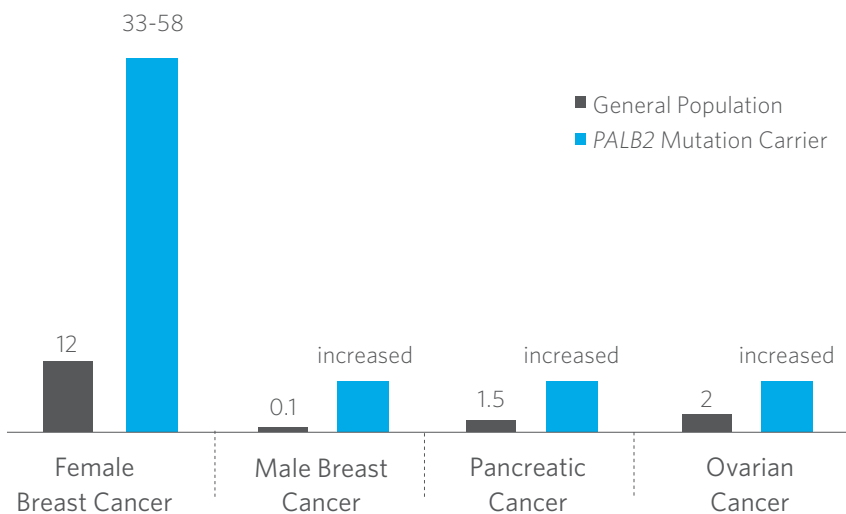
- 1 ***PALB2* mutation** Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *PALB2* gene.

- 2 **Cancer risks** You have an increased chance to develop female breast cancer, pancreatic cancer, ovarian cancer, and possibly other types of cancer like male breast cancer and prostate cancer.

- 3 **What you can do** There are risk management options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your doctor, and decide on a plan that best manages cancer risks.

- 4 **Family** Family members may also be at risk – they can be tested for the *PALB2* mutation that was identified in you.

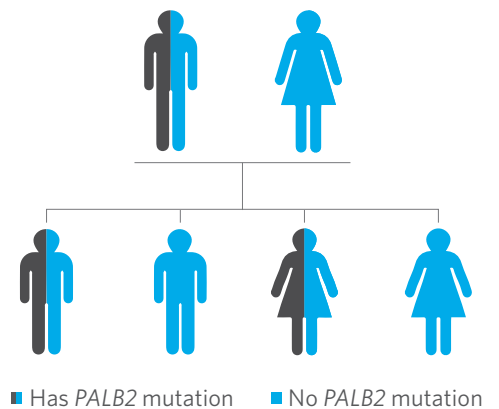
PALB2 MUTATION LIFETIME CANCER RISKS (%)*



*The above cancer risks represent the typical range for individuals with a mutation in this gene. If available, cancer risks specific to the mutation found in you will be provided in your results report.



PALB2 MUTATIONS IN THE FAMILY

There is a 50/50 random chance to pass on a mutation in *PALB2* to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.



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RESULT	MUTATION	Your testing shows that you have a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or a variant that is likely pathogenic in the <i>PALB2</i> gene. Both of these results should be considered positive.
GENE	<i>PALB2</i>	Everyone has two copies of the <i>PALB2</i> gene, which we randomly inherit from each of our parents. Mutations in one copy of the <i>PALB2</i> gene can increase the chance for you to develop certain types of cancer in your lifetime.
CANCER RISKS	INCREASED	You have an increased chance to develop female breast cancer, pancreatic cancer, ovarian cancer, and possibly other types of cancer like male breast cancer and prostate cancer. The risk to develop breast cancer can vary based on how many close relatives also have breast cancer. Your healthcare provider can better estimate your exact risk.
OTHER MEDICAL CONCERNS	MAY BE PRESENT	Individuals with <i>PALB2</i> mutations may have an increased risk (25%) to have a child with Fanconi anemia N (FA-N), but only if their partner <u>also</u> carries a mutation in the <i>PALB2</i> gene. Fanconi anemia is a rare condition that can cause specific physical characteristics, bone marrow failure, and an increased risk of certain cancers.
MANAGEMENT OPTIONS	 FOR WOMEN	Options for early detection and prevention for women depend on your family history of cancer and may include: breast exam, mammogram, breast MRI, and options for preventive surgery. Talk to your doctor about what options may be right for you.
MANAGEMENT OPTIONS	 FOR MEN & WOMEN	Options for screening and early detection depend on your family history of cancer, and may include pancreatic or other types of cancer screening. 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 <i>PALB2</i> 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 cancer.
NEXT STEPS	DISCUSS	It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.
REACH OUT	RESOURCES	<ul style="list-style-type: none"> • Ambry's hereditary cancer site for families patients.ambrygen.com/cancer • FORCE facingourrisk.org • Susan G. Komen Foundation komen.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • <i>PALB2</i> and Breast Cancer: A study to better understand breast cancer treatment among women with a <i>PALB2</i> mutation inheritedcancer.net/palb2-study/

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *PALB2* 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.



WATCH THE VIDEO

Scan or visit our website to watch a short video with additional information to help you understand your results.

ambrygen.com/cancer/uyr/palb2