Ambry Genetics®
A Konica Minolta Company

Understanding Your Positive *CHEK2* Genetic Test Result

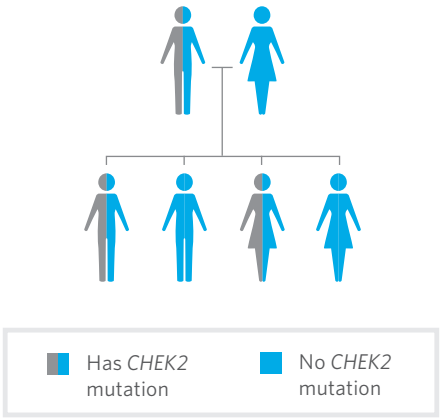
INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT, LIKELY PATHOGENIC**

4 Things To Know

1	*CHEK2* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *CHEK2* gene.
2	Cancer risks	You have an increased chance to develop female breast cancer, colorectal cancer, and possibly other cancers.
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 *CHEK2* mutation that was identified in you.

CHEK2 Mutations in the Family

There is a 50/50 random chance to pass on an *CHEK2* mutation 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 *CHEK2* gene. Both of these results should be considered positive.
Gene	***CHEK2***	Everyone has two copies of the *CHEK2* gene, which we randomly inherit from each of our parents. Mutations in one copy of the *CHEK2* 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 (around twice as high as the average woman), colorectal cancer, and possibly other cancers such as male breast cancer, prostate, thyroid, ovarian, or kidney.
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 early detection and prevention for both women and men depend on your family history of cancer and may include colonoscopies, comprehensive physical exams, or other screening options. 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, and children) have a 50/50 random chance of inheriting the *CHEK2* 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**	• Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer • American Cancer Society cancer.org • FORCE facingourrisk.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org • National Society of Genetic Counselors nsgc.org • Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *CHEK2* 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.

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