Ambry Genetics®

Clinician Management Resource for *CHEK2*

This overview of clinical management guidelines is based on this patient's positive test result for a *CHEK2* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1,2] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS[1,2]	AGE TO START	FREQUENCY
Female Breast Cancer[1]		
Breast Screening ▪ Mammography ▪ Consider breast MRI with and without contrast	Mammogram starting at age 40 years and consider breast MRI at age 30-35 years, or 5-10 years before the earliest known breast cancer in the family, whichever is earlier	Every 12 months
Evidence insufficient for risk-reducing mastectomy recommendation. Manage based on family history.	Individualized	N/A
Colorectal Cancer[2]		
Colonoscopy *For patients with colorectal cancer, please refer to the surveillance recommendations for post-colorectal cancer resection in the NCCN Guidelines for Colon Cancer and Rectal Cancer.[2]*	For probands without a personal history of colorectal cancer (CRC): High quality colonoscopy screening beginning at age 40, or 10 years prior to age of first-degree relative's CRC diagnosis	Every 5 years
Prostate Cancer		
Consider prostate cancer screening	Starting at age 40 years	Individualized

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V2.2024. © National Comprehensive Cancer Network, Inc. 2023. All rights reserved. Accessed September 27, 2023. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.
2. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal. V1.2023. © National Comprehensive Cancer Network, Inc. 2023. All rights reserved. Accessed June 1, 2023. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

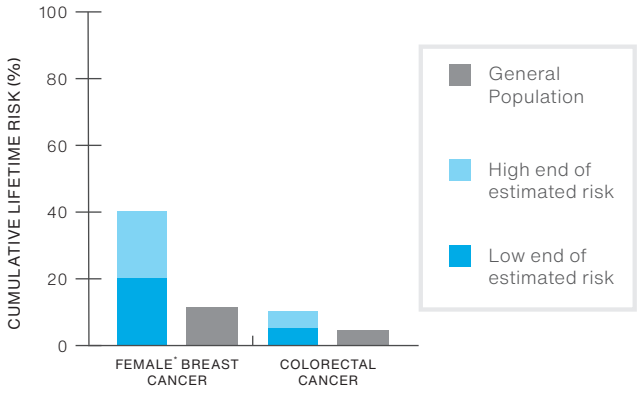
Understanding Your Positive *CHEK2* Genetic Test Result

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 and may have an increased chance to develop colorectal cancer.
3	What you can do	Risk management decisions are very personal. There are 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 works for you.
4	Family	Family members may also be at risk – they can be tested for the *CHEK2* mutation that was identified 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.

CHEK2 Mutation Lifetime Cancer Risks**



CHEK2 Mutations in the Family

There is a 50/50 random chance to pass on a *CHEK2* mutation to each of your children. The image below shows that everyone can carry and pass on these mutations, regardless of their sex at birth.

* Refers to sex assigned at birth
** Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history.

RESOURCES

- Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer
- American Cancer Society cancer.org
- Bright Pink brightpink.org
- FORCE facingourrisk.org
- ICARE Inherited Cancer Registry InheritedCancer.net
- Imerman Angels imermanangels.org
- Susan G. Komen Foundation komen.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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