Pathogenic (harmful) variants in the CHEK2 gene have been associated with a moderately increased risk for breast cancer and colon cancer. Some studies have also suggested associated risk for other cancers, although these are not as well established.

CHEK2 pathogenic variants are inherited in an autosomal dominant pattern, meaning that children of someone who carries a pathogenic variant each have a 50% risk to inherit the variant and associated cancer risks. Notably, women and men both have the CHEK2 gene and have the same chances to inherit and pass down variants in these genes. Therefore, both sides of the family are important when assessing inherited risk. However, the associated cancers and lifetime risks are different between men and women.

Cancer type	Risk in the general population	Risk in CHEK2 pathogenic variant carriers
Female breast	12%	28% – 37%
Colon cancer	5.5%	undefined

Genetic testing for CHEK2

Genetic testing for pathogenic variants in CHEK2 are currently available, but there are a few different ways to approach testing:

- *Single site analysis*: Testing specific to a known pathogenic variant in the family
- Full gene sequencing and rearrangement analysis: Comprehensive testing to search for all currently detectable variants in the gene
- Gene panels: Newer, more broadly based gene tests that would include not only the CHEK2 gene, but other genes known or suspected to be associated with increased cancer risks

Cancer Screening & Management for CHEK2

If you are tested and found to have a pathogenic variant in the CHEK2 gene, it is recommended to discuss your management plan with your healthcare team, and if available, to seek consultation through a specialized high-risk clinic. General recommendations are included here based on updated guidelines of the NCCN, but may be tailored to your specific medical and family history.

For women, breast cancer screening recommendations include starting annual mammograms beginning at age 40 (or 5-10 years earlier than the youngest diagnosis in the family), and also to consider breast MRI.

Colon cancer screening recommendations for CHEK2 include colonoscopy every 5 years beginning at age 40, or if there is family history of early colon cancer in a parent or sibling, beginning 10 years prior to their age at colon cancer diagnosis, whichever is earlier.

If there is a history of other cancers in the family, your doctor or healthcare provider may recommend additional, earlier, or more frequent screening for those other cancers based on the family history.

Click here to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.