**CHEK2**

The **CHEK2** gene is a tumor suppressor gene. Tumor suppressor genes slow down cell division, repair DNA mistakes, or tell cells when to die. When they don't work properly, cells can grow out of control, which can lead to cancer. The primary role of **CHEK2** is to pause cell division in order to make the critical decision of whether to repair damaged DNA or instruct the cell to die by a process known as apoptosis. The death of cells with significant DNA damage helps to prevent these cells from replicating out of control and forming a tumor.

Like most genes, each person has two copies of the **CHEK2** gene: one inherited from each parent. A mutation in a single **CHEK2** gene inherited from either parent is known to increase risk of certain cancers over a lifetime, including breast, colorectal, prostate, and possibly others.

Lifetime breast cancer risk estimates for women with **CHEK2** mutations range from 20% for those with no relatives with breast cancer to 44% for those with strong family history (defined as more than one close relative affected with breast cancer). Further research is needed to understand the interactions of **CHEK2** and family history on lifetime breast cancer risk.

If an individual inherits two **CHEK2** mutations (one from each parent), they may have a significantly increased risk for cancer, particularly female breast cancer as an adult.

**How common are mutations in the **CHEK2** gene?**
Mutations in the **CHEK2** gene are rare—one mutation is found in approximately 3-7 out of 1,000 (0.3-0.7%) people of Dutch descent. Studies to establish how common **CHEK2** mutations are in other populations are ongoing.

**How mutations in this gene impact risk**

**Women**

If a woman has a mutation in the **CHEK2** gene, her chances of developing breast and colorectal cancer are greater than that of the average US woman. This does not mean that she has a diagnosis of cancer or that she will definitely develop cancer in her lifetime.

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Cancer by age 70 | Average US woman\(^6\) | With CHEK2 mutation
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Breast | 7.1% | 20-44%\(^1,2\)
Colorectal | 1.6% | Elevated (2-5\(^{\%}\))\(^7\)

Elevated: Risk is increased, but further research may clarify the exact risk figure.

Men
If a man has a mutation in the CHEK2 gene, his chances of developing prostate and colorectal cancers are greater than that of the average US man. This does not mean that he has a diagnosis of cancer or that he will definitely develop cancer in his lifetime.

Cancer by age 70 | Average US man\(^6\) | With CHEK2 mutation
--- | --- | ---
Colorectal | 2.0% | Elevated (3-6\(^{\%}\))\(^7\)
Prostate | 7.2% | Elevated\(^8\)

Elevated: Risk is increased, but further research may clarify the exact risk figure.

Additional information
CHEK2 studies have focused on one specific mutation.
The majority of studies related to CHEK2 are for individuals with one specific mutation called 1100delC. This mutation is more commonly reported in those of Dutch ancestry.

Screening guidelines
Below is a summary of screening guidelines from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) established by experts at the National Comprehensive Cancer Network (NCCN).\(^9\) They are for individuals with a mutation in the CHEK2 gene. If you have a

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\(^9\) Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial

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mutation in this gene, your healthcare provider may use these NCCN Guidelines® to help create a customized screening plan for you.

Women
Breast cancer

- Starting at age 40: Your provider may discuss mammogram and breast MRI with contrast every year.

Colorectal cancer

- **Beginning at age 40 or 10 years younger than the earliest diagnosis of colorectal cancer in a parent, sibling, or child (whichever is earlier):** Colonoscopy every 5 years.
- These recommendations may change if you have polyps, colorectal cancer, inflammatory bowel disease (IBD), or family history of colorectal cancer.

Men
Colorectal cancer

- **Beginning at age 40 or 10 years younger than the earliest diagnosis of colorectal cancer in a parent, sibling, or child (whichever is earlier):** Colonoscopy every 5 years.
- These recommendations may change if you have polyps, colorectal cancer, inflammatory bowel disease (IBD), or family history of colorectal cancer.

Prostate cancer

- Currently, there are no prostate cancer screening guidelines from the National Comprehensive Cancer Network (NCCN) specific to CHEK2 mutation carriers. Your provider may discuss earlier or more frequent screening or referral to a specialist.

Useful resources

**FORCE**
Providing support, education, research, and resources for survivors and people at increased risk of cancer due to an inherited mutation or family history of cancer.

[www.facingourrisk.org](http://www.facingourrisk.org)
Kintalk
An educational and family communication site for individuals and their families with hereditary cancer conditions.
www.kintalk.org

Susan G. Komen
Dedicated to reducing deaths from breast cancer by funding breast cancer research, ensuring access to care through community programs worldwide and supporting public health policies that help people facing breast cancer.
www.komen.org/

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