

About *CHEK2* Gene Mutations

About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work right, they help keep cancerous cells from developing. If one copy has a mutation, it cannot act as it should. This raises the risk for certain cancers.

Having a *CHEK2* gene mutation raises the risk for breast, colorectal, and prostate cancers above the general population. The risks for other cancers can rise with *CHEK2* mutations, but ongoing research is providing more understanding.

Most of the information known about *CHEK2* is based on a mutation called 1100delC. About 1% of people with Northern and Eastern European origin have this mutation.

CHEK2 Mutations and Cancer Risk

BREAST CANCER

Most women have a 10% lifetime risk of getting breast cancer. That increases to 20-30% for women with a *CHEK2* mutation. For women with a *CHEK2* mutation and a family history of breast cancer, this risk may be even higher. Women with a *CHEK2* mutation who already had breast cancer are more at risk of getting a second, new breast cancer.

Male breast cancer is very rare—less than 1% of men will get it during their lifetime. Men with *CHEK2* mutations have a slightly higher lifetime risk of getting breast cancer.

COLON CANCER

People with *CHEK2* mutations may have a higher risk for colorectal cancer. This risk may be higher if the family has a history of colorectal cancer.

PROSTATE CANCER

The risk of prostate cancer in men with *CHEK2* mutations may be slightly higher.

Recommendations

WOMEN

Starting at age 40: Mammogram and breast MRI every year (scheduled 6 months apart)

Women may think about taking medicine, such as tamoxifen or raloxifene, to lower the risk of getting breast cancer.

MEN

Current screening recommendations for prostate or breast cancer in men with *CHEK2* mutations are the same as for the general population.

WOMEN AND MEN

Starting at age 40: Colonoscopy every 5 years

People who have parents or siblings with colorectal cancer under age 50 should start screening 10 years before their age at diagnosis.

Cancer screening recommendations for people with *CHEK2* mutations update regularly. Talk to your doctor or genetic counselor every few years for updates to cancer risks and screenings.

KIDS

Children of people with a *CHEK2* mutation have a 50% chance of also having the mutation. The recommendation is to get genetic testing and counseling after they turn 18.

FAMILY MEMBERS WHO TEST NEGATIVE

People who do not have the familial *CHEK2* mutation likely have the same level of risk for breast, prostate, and colorectal cancers as the general population. It depends on family history and other risk factors. Family members who test negative for the *CHEK2* mutation should talk with their doctor or genetic counselor about the right cancer screenings for them.

Resources

If you have a personal or family history of cancer, you might consider genetic testing and counseling. Our team can help create a plan if you or a family member has a gene mutation. Please call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: **801-587-9555**.