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 properly, they help keep cancerous cells from developing. If one copy of a gene has a mutation, it cannot function as it should. This increases the risk for certain cancers.

Having a mutation in the CHEK2 gene increases the risk for breast, colorectal, and prostate cancers above the general population. The risks for other cancers may be increased with CHEK2 mutations, but research is still being done to understand more.

Most of the information known about CHEK2 is based on a mutation called 1100delC. This mutation is found in about 1% of people with Northern and Eastern European ancestry.

**CHEK2 Mutations and Cancer Risk**

**BREAST CANCER**
Women have a 10% average lifetime risk of developing breast cancer. The risk increases to approximately 20–30% lifetime risk for breast cancer in 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 have an increased risk to develop a second, new breast cancer.

Male breast cancer is very rare in the general population—less than 1% of men will develop it during their lifetime. Men with CHEK2 mutations have a slightly higher lifetime risk of breast cancer than the general population.

**COLON CANCER**
People with CHEK2 mutations may have an increased 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 increased.

**Recommendations**

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

Women may consider taking medicine such as tamoxifen or raloxifene to reduce the risk of developing 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*

For people with a first-degree relative (parents or siblings) diagnosed with colorectal cancer under age 50, start screening 10 years before the relative's age at diagnosis.

Cancer screening recommendations for people with CHEK2 mutations are developing continually. Follow up with 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. Genetic testing and counseling are recommended after age 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, depending 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.

**Resources**

If you have a personal or family history of cancer, you may be eligible for genetic testing and counseling. If you know you or a family member has a CHEK2 or other gene mutation, our team of doctors and genetic counselors can help create a management plan. Please call Huntsman Cancer Institute’s Family Cancer Assessment Clinic to learn more: 801-587-9555.