About *CHEK2* I157T 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 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 two most common *CHEK2* mutations in European populations are p.I157T and c.1100delC.

The *CHEK2* I157T mutation has a lower risk for breast cancer and other cancers compared to the *CHEK2* c.1100delC mutation. The lower breast cancer risk for those with a *CHEK2* I157T mutation means that recommendations for screening may be different from others who have mutations in *CHEK2*. The risks for other cancers may be increased with all *CHEK2* mutations, but research is still being done to understand more.

CHEK2 I157T Mutation and Cancer Risks

BREAST CANCER

About 10 in 100 women will get breast cancer in their lifetime. For women with the *CHEK2* I157T mutation the risk increases to approximately 16-19 in 100 who will get breast cancer. For women with a *CHEK2* I157T mutation and a family history of breast cancer, this risk may be even higher.

COLON CANCER

About 5 in 100 people will get colon cancer in their lifetimes. People with *CHEK2* I157T mutations may have an increased risk for colorectal cancer. 6-9 in 100 people with *CHEK2* I157T may get colon 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* I157T mutations may be slightly increased.

Recommendations

WOMEN

Women should practice breast awareness, which involves being familiar with their breasts and reporting any changes to their healthcare provider.

Starting by age 35: Clinical breast exam by a doctor yearly.

Starting at age 40: Mammogram every year.

Without a family history of breast cancer, the *CHEK2* I157T mutation is not associated with a sufficient breast cancer risk to warrant breast MRI screening. Those with a family history of breast cancer in addition to *CHEK2* I157T should discuss their recommendations with a genetic counselor or doctor.

MEN

Current screening recommendations for prostate 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* I157T mutation have a 50% chance of also having the mutation. Genetic testing and counseling may be recommended after age 18.

FAMILY MEMBERS WHO TEST NEGATIVE

People who do not have the familial *CHEK2* I157T mutation may 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* I157T mutation should talk with their doctor or genetic counselor about the right cancer screenings.

Resources

If you have the *CHEK2* I157T mutation remember that your risks may be different from other *CHEK2* carriers. If you read online about *CHEK2,* keep this difference in mind. If 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.

Questions about your risk for cancer? We can help with a screening plan that is best for you. Contact the Family Cancer Assessment Clinic: 801-587-9555. Huntsmancancer.org/fcac