Patient Education

BAPTIST MD Anderson Cancer Center
1.844.632.2278

CHEK2 Mutation

The *CHEK2* gene normally works to protect the body from cancer. A genetic change (mutation) in the *CHEK2* gene causes the gene to stop working normally. When the *CHEK2* gene stops working normally, cancer risk increases. People with *CHEK2* gene mutation have an increased risk of developing certain cancers.

Breast Cancer

Women with a *CHEK2* gene mutation have an increased risk of developing breast cancer. The average woman has a 10%-20% risk of developing breast cancer in her lifetime. For women with a *CHEK2* gene mutation, the risk is increased to about 24% to 36%. The chance to develop a new or second breast cancer may be increased, but the risk is unknown.

Men with a *CHEK2* gene mutation may have an increased risk of developing breast cancer, but the risk is unknown. More research is needed.

The type of *CHEK2* gene mutation you have may affect you risk for breast cancer. Certain types of mutations in the *CHEK2* gene may lead to lower risk than other mutations. Talk with your doctor or genetic counselor about your risk and the care options that are best for you.

Colon Cancer

Men and women with a *CHEK2* gene mutation have an increased risk of developing colon cancer. The average person has a 3% to 4% risk of developing colon cancer in their lifetime. For people with a *CHEK2* gene mutation, the risk is increased to 6% to 11%.

Other Cancers

CHEK2 gene mutations may increase risks for other cancers including prostate, thyroid, ovarian, and kidney cancer. Exact risks are unknown at this time.

Screening and Prevention

Screening helps detect cancers as early as possible when it may be easier to treat. People with *CHEK2* gene mutation may benefit from additional screenings. Screening recommendations depend on age, personal history, and family history. Your doctor's recommendations may include:

● Annual mammograms and breast magnetic resonance imaging (MRI) at age 40. Every 6 months, alternate mammograms with breast MRIs. If you have family history of breast cancer,

screening may start 10 years before the age of the youngest breast cancer diagnosis in your family.

- Consider preventative breast surgery based on family history.
- Colonoscopy every 5 years starting at age 40. If you have family history of colon cancer, screening may start 10 years before the age of the youngest colon cancer diagnosis in your family.

Recommendations may change as researchers learn more about how *CHEK2* mutations relate to cancer risks. Talk with your doctor about your care options.

Family Members

People with a *CHEK2* gene mutation inherited the mutation from either their father or mother. A person with the *CHEK2* gene mutation has a 50% (1 in 2) chance to pass the *CHEK2* mutation to each child. Men and women have equal chances to pass on the mutation.

Family members may benefit from genetic testing when a person in the family has a genetic disorder. Predictive genetic testing is for family members who shoe no features or symptoms of the *CHEK2* gene mutation at the time of testing. Testing can help decide which family members should consider additional cancer screenings and preventative care.

Depending on family history, family members without the *CHEK2* gene mutation may still benefit from additional screenings.

Resources

FORCE
www.facingyourrisk.org
FORCE is a nonprofit organization with resources for families with an increased risk for developing breast and ovarian cancer. FORCE has education resources and in-person and virtual support programs.

Bright Pink
www.brightpink.org
Bright Pink is a nonprofit organization that provides support and education to women ages 18 to 45 who have a high risk for developing breast and ovarian cancer.

National Cancer Institute-Colorectal Cancer
www.cancer.gov/types/colorectal
The National Cancer Institute provides information about colon cancer screening, prevention, and treatment.

The Genetic Information Nondiscrimination Act (GINA)
www.ginahelp.org
This website provides information about the laws protecting against discrimination based on genetic information. It includes information about employers and health insurance, and answers commonly asked questions about the Genetic Information Nondiscrimination Act (GINA).

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