PATIENT & CAREGIVER EDUCATION

About Mutations in the CHEK2 Gene

This information explains how having a mutation in the CHEK2 gene may affect you and your family.

In this resource, the word “family” means family members related to you by blood. They are not related to you through marriage or adoption.

Your CHEK2 gene normally helps prevent cancers. A mutation in this gene causes it to stop working like it should. This increases your risk for certain types of cancers.

What is my cancer risk if I have a CHEK2 mutation?

A CHEK2 gene mutation increases your risk for certain types of cancers. Your cancer risk may be different depending on the specific CHEK2 mutation you have. Most CHEK2 mutations increase your risk for breast cancer.

Some of the most common CHEK2 mutations slightly
increase your risk for colorectal (colon and rectal) cancer.

While a *CHEK2* gene mutation means you have a somewhat higher risk of developing cancer than the average person, it may not fully explain why your blood relatives had cancer.

As we learn more about these mutations, we may learn they increase the risk for other types of cancers. Your genetic counselor will give you more information about your cancer risk if you have a mutation.

For more information, read *Hereditary Breast Cancer* - www.mskcc.org/genetics/breast-cancer

**What can I do about my cancer risk if I have a *CHEK2* mutation?**

If you have a mutation, your genetic counselor will review your results and your personal and family history of cancer and give you cancer screening recommendations.

They may recommend you start having cancer screenings at a younger age, have them more often than most people, or get specialized screenings to help find cancer as early as possible.
Some examples of these cancer screenings include:

- Having breast magnetic resonance imaging (MRI) scans and mammograms (x-rays of your breast).
- Having colonoscopies starting at an earlier age and more often than most people.

Your genetic counselor will also talk with you about whether there are any other screening or prevention options that may be right for you.

**What happens if I don’t have a CHEK2 mutation?**

If you don’t have a mutation, your genetic counselor will review your personal and family history and talk with you about the general cancer screening guidelines you should follow.

**What does a CHEK2 mutation mean for my blood relatives?**

If you have a mutation, your biological parents, siblings, and children each have a 50% chance of having the same mutation. This means there’s an equal chance they will or won’t have the mutation. Your distant family members may also be at risk for having the same mutation.
Males and females have an equal chance of passing down a mutation in their family. You only need to inherit a mutation from one parent to have an increased risk for cancer.

Your genetic counselor will review your family history and talk with you about whether they recommend genetic testing for your blood relatives.

**Contact Information**

If you have any questions or concerns, talk with a genetic counselor in the Clinical Genetics Service. You can reach them Monday through Friday from 9:00 a.m. to 5:00 pm at 646-888-4050.

For more resources, visit [www.mskcc.org/pe](http://www.mskcc.org/pe) to search our virtual library.

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