CHEK2 Gene Information

The CHEK2 gene plays a role in preventing people from developing tumors or cancer. Everyone has two copies of CHEK2 – one copy of the gene inherited from each parent. If you inherit one copy of a CHEK2 gene with an alteration in the genetic code that prevents the gene from working properly, this leads to increased chances to develop cancer. Medically significant alterations in the genetic code are called “mutations” or “pathogenic variants.” Not everyone who is born with a CHEK2 mutation will develop cancer – many will not.

Cancer Risks

- **Breast cancer:** Inherited mutations in the CHEK2 gene confer an increased risk for female breast cancer which is estimated to be 20%-40% over a female’s lifetime. The general population risk for female breast cancer is about 13%.
- **Colorectal cancer:** Prior studies have demonstrated an increased risk for colorectal cancer associated with inherited mutations in the CHEK2 gene; however, this association has not been consistently seen in current studies.
- **Other cancers:** A recent study showed an increased risk for kidney cancer and thyroid cancer among individuals with inherited mutations in the CHEK2 gene.

It is possible that the information about cancer risks associated with CHEK2 could change over time as our knowledge about this gene expands.

Medical Recommendations

The recommendations below are meant as general guidelines and may change based on other personal risk factors or family history. To make an appointment to discuss personal recommendations with a Cancer Genetics Physician at Dana-Farber, please call **617-632-2178**.

- **Breast cancer** screening is recommended to begin at age 30-35 with annual breast MRI with contrast. Annual mammogram is recommended to begin at age 40. Currently, there is no information to support risk-reducing bilateral mastectomies.

- **Colorectal cancer** screening is recommended to begin at age 40 with colonoscopy. If there is a family history of a first-degree relative with colorectal cancer, then screening with colonoscopy should begin ten years before the relative’s age of diagnosis. Colonoscopy screening is recommended every five years unless personal or family history suggests otherwise.
• Recommendations for kidney cancer and thyroid cancer screening may be made on an individual basis and may include ultrasound of the kidney and/or thyroid every 1-2 years.

These recommendations are based on our current understanding of the CHEK2 gene and may change over time. Recommendations are also influenced by other factors, including but not limited to: cancer treatments, family history of cancer, personal preference, and gender affirming care. You should discuss personalized recommendations with your physician.

Inheritance and Meaning for Your Family

It is important that you share your test results with your biological (blood-related) relatives. Your close relatives, including parents, siblings, and children, have a 1 in 2, or 50%, chance to have inherited the CHEK2 mutation, as well. All individuals in the family can inherit and pass along CHEK2 mutations to their children. Mutations in CHEK2 do not skip a generation. If someone in your family did not inherit the CHEK2 mutation, they cannot pass it along to children.

It is recommended that young adults wait until they are at least 18 years old before seeking genetic testing. However, people may wait to do testing until the age their medical care would change if they carried the CHEK2 mutation, and/or they feel emotionally ready to learn this information.

© 2023 Dana-Farber Cancer Institute