BC CANCER HEREDITARY CANCER
Provincial Health Services Authority

CHEK2 Cancer Risks and Management

Overview

People with a pathogenic loss-of-function variant in the *CHEK2* gene have a higher chance of developing certain types of cancer compared to the general population. Cancers may happen at a younger age for *CHEK2* carriers.

This document summarizes the cancer risks and management recommendations for individuals with a confirmed *CHEK2* pathogenic loss-of-function variant.

Cancer risks associated with *CHEK2*

Cancer Type	*CHEK2* (Lifetime Risk)	General Population
Breast cancer (female)	25%*	12.5%
Breast cancer (male)	1%	Less than 1%
Prostate cancer	Moderate increase	12.5%

* Average lifetime risk, with risk ranging up to 40% depending on other genetic and environmental risk modifiers.

Contralateral breast cancer (females) – 13% at 10 years after a premenopausal breast cancer, 4% at 10 years after a post-menopausal breast cancer.

Other cancers (e.g. thyroid, kidney cancer) have been reported at increased frequency in some families. Further studies are needed to confirm an association.

Cancer Screening and Risk Reduction

Female Breast Cancer

Female Breast Cancer Screening:

- Starting at age 18, females should become familiar with the normal look and feel of their breast tissue and to report any changes to their primary care provider promptly. Regular and consistent breast self-exams can support breast self-awareness and are often most effective when done at the end of menstruation.
- **Annual clinical exam** of the breast and regional nodes from **age 30**
- **Annual mammograms** beginning **at age 40** and continue as long as clinically indicated.
- **Annual breast MRI** beginning at **age 35** until age 70.

Female Breast Cancer Prevention:

- Discussion of **risk reducing medication** options and review of potential benefits and side effects is recommended. Medications such as tamoxifen, raloxifene, anastrozole and exemestane may reduce the risk of developing a hormone-receptor positive breast cancer.
- **Risk reducing bilateral mastectomy (RRBM)** can reduce the risk of breast cancer by over 90%. The decision to have RRBM is complex and most suited to women known to be at high risk of developing breast cancer. People may wish to consider RRBM if they have a *CHEK2* pathogenic variant in addition to a strong history of breast cancer. The decision to have RRBM requires consideration of the benefits and risks of the surgery in the context of a person's general health, life expectancy and personal health beliefs. Routine breast imaging (mammogram and/or breast MRI) is not required after bilateral mastectomy.

Male Breast Cancer

- Starting at age 35, males should become familiar with the normal look and feel of their breast tissue and to report any changes to their primary care provider promptly. Regular and consistent breast self-exams can support breast self-awareness.
- **Annual clinical exam** of the breast and regional nodes from age 35.

Prostate Cancer

Consider annual **digital rectal examination (DRE)** and/or serum **prostate specific antigen test (PSA)** testing as early as 40-45 years of age or 5-10 years before the youngest diagnosis of prostate cancer in the family (whichever is earlier)

The Canadian Urological Association recommends healthcare providers engage in shared decision-making with their patients to come to an individualized screening decision following a thorough discussion on the potential risks and benefits of the PSA test. In BC, PSA testing in asymptomatic men is not an insured benefit.

Note: In the information above, male/female refers to sex assigned at birth.

High Risk Clinic

Individuals with breast tissue who carry a pathogenic variant in the *CHEK2* gene, or are at 50% risk of having inherited one, can be referred to the Hereditary Cancer Program's High Risk Screening Clinic for ongoing cancer risk management and decision support. Read more about the High Risk Clinic.

Family and Reproductive Considerations

Inheritance

Each child of someone with a *CHEK2* pathogenic variant has a 50% chance of inheriting the variant.

Family members are encouraged to contact their local genetics clinic to learn more about whether genetic testing or cancer screening may be helpful for them. Family members who live in British Columbia or the Yukon can contact our program directly at hereditarycancer@bccancer.bc.ca. In BC/Yukon, genetic testing is generally available starting at age 19.