Facts for people and families with a faulty CHEK2 gene

ID: 3966 v.1 **Endorsed**

This fact sheet contains general information. Each person should be referred to a genetic service for further information and advice about what a faulty CHEK2 gene means for them.

Key Points

- Women with a faulty CHEK2 gene have an increased chance of developing breast cancer.
- Both men and women can carry CHEK2 gene faults and may pass the faulty gene on to their children.
- Men and women can be referred to a genetic service where experts can provide information, advice and support about their chance of developing cancer and the option of genetic testing.

What is a faulty CHEK2 gene?

CHEK2 is a 'cancer protection' gene that helps to protect against breast cancer.

Everyone has two CHEK2 genes (one from their mother, and one from their father). If one of the genes is not working, this is known as having a *faulty CHEK2 gene*, or having a *CHEK2 mutation*.

What is the risk of cancer for people with a faulty CHEK2 gene?

- Most women with a faulty CHEK2 gene have between 20% and 30% chance of developing breast cancer over their lifetime. The chance of developing breast cancer may be lower than 20% or higher than 30% for some women, if they have close relatives with breast cancer.
- **Not everyone who has a faulty CHEK2 gene will develop cancer.**

How can this increased risk of cancer be managed?

- To **find breast cancer early**, women with a faulty CHEK2 gene should have breast cancer screening every year between age 40 to age 50 years. This involves having a mammogram, plus a breast check by a doctor. Some women with a faulty CHEK2 gene will need to begin breast cancer screening before age 40 years. After age 50 years, most women with a faulty CHEK2 gene should have breast cancer screening every 2 years. However, some women with a faulty CHEK2 gene will still need to continue breast cancer screening every year after age 50 years.
- To **reduce the chance of getting breast cancer**, women with a faulty CHEK2 gene may take medications such as tamoxifen or raloxifene.

What does this mean for family members?

Adult family members of someone with a faulty CHEK2 gene can have genetic testing to check who has the faulty gene and who does not. Their doctor can refer them to a genetic service to find out more about their chance of developing cancer and what genetic testing involves.

If a person **does have** the faulty CHEK2 gene:

- they can pass it on to their children
- each child has a 50% (1 in 2) chance of being born with it. Pregnancy planning options are available to people who want to prevent the faulty gene from being passed on.

If a person **does not have** the faulty CHEK2 gene:

- they have the same chance of developing cancer as the general population (unless there are other factors that increase this risk)
- they cannot pass it on to their children.

People who decide not to have genetic testing should still get advice about managing their chance of developing cancer.

More information and support

- Centre for Genetics Education NSW Health: Contact details for local genetics services –genetics.edu.au/SitePages/Genetic-Services.aspx
- Genetic Alliance Australia – geneticalliance.org.au
- Cancer Australia – canceraustralia.gov.au
- Breast Cancer Network Australia (BCNA) – bcna.org.au
- Facing Our Risk of Cancer Empowered (FORCE) – facingourrisk.org
- Pink Hope – pinkhope.org.au
- COSA - Medications to lower the chance of breast cancer: information for women – cosa.org.au/groups/cancer-genetics/resources/

History

Version 1

Date	Summary of changes
16/03/2022	New consumer information sheet developed following discussion of ID 3701 CHEK2 – risk management document at May 2021 cancer genetics reference committee meeting. Approved for publication as V.1. Review alongside ID 3701 at next scheduled review.

First approved: 16 March 2022
Review due: 7 March 2024

The currency of this information is guaranteed only up until the date of printing, for any updates please check:
https://www.eviq.org.au/p/3966
10 Jun 2022