

## BARD1

The *BARD1* gene is a tumor suppressor gene. Tumor suppressor genes slow down cell division, repair DNA mistakes, or tell cells when to die. When they don't work properly, cells can grow out of control, which can lead to cancer. The primary role of *BARD1* is stabilizing and assisting the *BRCA1* gene in repairing damaged DNA before a cell divides to make copies of itself.

Like most genes, each person has two copies of the *BARD1* gene: one inherited from each parent. A mutation in a single *BARD1* gene inherited from either parent is known to increase risk of breast cancer over a lifetime.

### How common are mutations in the *BARD1* gene?

Mutations in the *BARD1* gene are rare—the exact frequency is not yet known. Studies to establish the frequency of *BARD1* mutations are ongoing.

### How mutations in this gene impact risk

#### Women

If a woman has a mutation in the *BARD1* gene, her chance of developing breast cancer is greater than that of the average US woman. This does not mean that she has a diagnosis of cancer or that she will definitely develop cancer in her lifetime

Cancer by age 80	Average US woman <sup>1</sup>	With <i>BARD1</i> mutation <sup>2,3,4</sup>
Breast	10%	Elevated

*Elevated: Risk is increased, but further research may clarify the exact risk figure.*

#### Men

If a man has a mutation in the *BARD1* gene, his chance of developing cancer is not known to be increased.

<sup>1</sup> Surveillance, Epidemiology, and End Results (SEER) Program, National Cancer Institute. 2010-2012. DevCan software (<http://surveillance.cancer.gov/devcan>) V 6.7.0, Accessed June 2015.

<sup>2</sup> De brakeleer S, De grève J, Loris R, et al. Cancer predisposing missense and protein truncating BARD1 mutations in non-BRCA1 or BRCA2 breast cancer families. *Hum Mutat.* 2010;31(3):E1175-85.

<sup>3</sup> Couch FJ, Hart SN, Sharma P, et al. Inherited mutations in 17 breast cancer susceptibility genes among a large triple-negative breast cancer cohort unselected for family history of breast cancer. *J Clin Oncol.* 2015;33(4):304-11.

<sup>4</sup> Walsh T, Casadei S, Lee MK, et al. Mutations in 12 genes for inherited ovarian, fallopian tube, and peritoneal carcinoma identified by massively parallel sequencing. *Proc Natl Acad Sci U S A.* 2011 Nov 1;108(44):18032-7.

## Additional information

Research on the *BARD1* gene is ongoing, especially research related to its impact on ovarian cancer risk.

## Screening guidelines

Below is a summary of screening guidelines from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) established by experts at the National Comprehensive Cancer Network (NCCN).<sup>5</sup> Because there are no published NCCN Guidelines® specific to women with *BARD1* mutations, these guidelines are for women who have the same breast cancer risk as the average US woman. However, if you have a mutation in this gene, your healthcare provider may recommend additional screening and risk reduction options, such as earlier and more frequent screening, screening with breast MRI, and medications to reduce the risk of breast cancer.

### Women

#### Breast cancer<sup>6</sup>

- Starting at age 25: Breast awareness - Women should be familiar with their breasts and promptly report changes to their healthcare provider.
- Between ages 25-39: Breast exam, risk assessment, and risk reduction counseling by your provider every 1-3 years.
- Starting at age 40: Breast exam, risk assessment, and risk reduction counseling by your provider and mammogram every year. Your provider may discuss screening with tomosynthesis.

## Useful resources

### FORCE

Providing support, education, research, and resources for survivors and people at increased risk of cancer due to an inherited mutation or family history of cancer.

[www.facingourrisk.org](http://www.facingourrisk.org)

### Bright Pink

Focused on the prevention and early detection of breast and ovarian cancer in young women, while providing support for high-risk individuals.

[www.brightpink.org](http://www.brightpink.org)

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<sup>5</sup> Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Breast Cancer Screening and Diagnosis V.1.2016. © National Comprehensive Cancer Network, Inc 2016. All rights reserved. Accessed November 1, 2016. To view the most recent and complete version of the guideline, go online to NCCN.org. NATIONAL COMPREHENSIVE CANCER NETWORK®, NCCN®, NCCN GUIDELINES®, and all other NCCN Content are trademarks owned by the National Comprehensive Cancer Network, Inc.

<sup>6</sup> National Comprehensive Cancer Network. Breast Cancer Screening and Diagnosis. *NCCN Guidelines Version 1.2016*. Available at [www.nccn.org](http://www.nccn.org). Published July 2016.

**Susan G. Komen**

Dedicated to reducing deaths from breast cancer by funding breast cancer research, ensuring access to care through community programs worldwide and supporting public health policies that help people facing breast cancer.

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