BRIP1

The *BRIP1* 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 *BRIP1* is the unwinding of damaged DNA so that it can be repaired.

Like most genes, each person has two copies of the *BRIP1* gene: one inherited from each parent. A mutation in a single *BRIP1* gene inherited from either parent is known to increase risk of breast and ovarian cancer over a lifetime.

In very rare cases, a person can inherit two *BRIP1* mutations, one from each parent. This causes a blood condition called Fanconi anemia, which is associated with bone marrow failure, physical disabilities, and childhood cancers.

How common are mutations in the *BRIP1* gene?

Mutations in the *BRIP1* gene are rare—the exact frequency is not yet known. Studies to establish the frequency of *BRIP1* mutations are ongoing.

How mutations in this gene impact risk

Women

If a woman has a mutation in the *BRIP1* gene, her chances of developing breast and ovarian cancer are 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[1]	With *BRIP1* mutation
Breast	10%	Elevated (12-32%)[2,3]
Ovarian	1%	Elevated (5-14%)[4,5]

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

[1] 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.
[2] Seal S, Thompson D, Renwick A, et al. Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. *Nat Genet.* 2006 Nov;38(11):1239-41.
[3] Easton DF, Pharoah PD, Antoniou AC, et al. Gene-panel sequencing and the prediction of breast-cancer risk. *N Engl J Med.* 2015;372(23):2243-57.
[4] 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.
[5] Rafnar T, Gudbjartsson DF, Sulem P, et al. Mutations in BRIP1 confer high risk of ovarian cancer. *Nat Genet.* 2011 Oct 2;43(11):1104-7. doi: 10.1038/ng.955.

Men

If a man has a mutation in the *BRIP1* gene, his chance of developing cancer is not known to be increased.

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.[6] They are for women who have a mutation in the *BRIP1* gene, except as noted. If you have a mutation in this gene, your healthcare provider may use these NCCN Guidelines® to help create a customized screening plan for you.

Women

Breast cancer[7]

- There are currently no breast cancer screening guidelines specific to women with *BRIP1* mutations from the NCCN. Therefore, these NCCN Guidelines® are for women who have the same breast cancer risk as the average US woman. However, your healthcare provider may recommend additional breast cancer screening and risk reduction options, such as earlier and more frequent screening, screening using breast MRI, and medications to reduce the risk of breast cancer.
- **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.

Ovarian cancer[8]

- **Starting at age 45-50, or earlier based on family history of ovarian cancer:** Your healthcare provider may discuss a risk-reducing salpingo-oophorectomy (the surgical removal of the ovaries and fallopian tubes) with you to lower the risk of developing ovarian cancer.

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[6] Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast and Ovarian V.1.2017 and 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.

[7] National Comprehensive Cancer Network. Breast Cancer Screening and Diagnosis. *NCCN Guidelines Version 1.2016.* Available at www.nccn.org. Published July 2016.

[8] National Comprehensive Cancer Network. Genetic/Familial High-Risk Assessment: Breast and Ovarian. *NCCN Guidelines Version 1.2017*. Available at www.nccn.org. Published Setpember 2016.

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

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

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.
www.komen.org/

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