

BARD1 gene

Associated Syndrome Name: **BARD1-associated cancer risk (Women only)**

BARD1 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Breast	High Risk

BARD1 gene Overview

BARD1-associated cancer risk (Women only)^{1, 2, 3, 4, 5}

- Women with *BARD1* mutations have a risk for breast cancer that is significantly increased over the 12.5% risk for women in the general population of the United States. Most studies have found that the risk is approximately doubled, with some studies suggesting that the risk could be much higher in certain populations or in women with a family history of breast cancer.
- Studies have also shown that breast cancers in women with *BARD1* mutations are more likely to be Triple Negative Breast Cancer (TNBC). This type of breast cancer lacks estrogen and progesterone receptors, and does not express Her2. It can be more aggressive than other types of breast cancer.
- At this time, there are no known cancer risks for men due to mutations in *BARD1*.
- Although there is an increased risk for cancer in women with mutations in *BARD1*, there are interventions that may reduce these risks. Guidelines from the National Comprehensive Cancer Network (NCCN) that may apply are listed below. Since information about the cancer risks associated with *BARD1* mutations is relatively new, and there is still some uncertainty about the best ways to reduce these risks, it may be appropriate to interpret these results in consultation with cancer genetics experts in this emerging area of knowledge.

BARD1 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Female Breast	To age 80 ^{2, 3, 6, 7, 8}	22%, with a particularly increased risk for triple negative breast cancer (TNBC)	10.7%

BARD1 Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Female Breast	Breast awareness - Women should be familiar with their breasts and promptly report changes to their healthcare provider. Periodic, consistent breast self-examination (BSE) may facilitate breast awareness. ⁹	Individualized	NA
	Clinical encounter, including clinical breast exam, ongoing risk assessment and risk-reduction counseling ⁹	When genetic risk is identified, but not before age 21	Every 6 to 12 months

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
	Mammography and consideration of breast MRI with contrast ¹⁰	Age 40, or modified to a younger age based on the family history of breast cancer	Annually
	Consider additional risk-reduction strategies. ^{9, 10}	Individualized	NA
For Patients With A Cancer Diagnosis	For patients with a gene mutation and a diagnosis of cancer, targeted therapies may be available as a treatment option for certain tumor types (e.g., PARP-inhibitors). ¹¹	NA	NA

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the *BARD1* gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

At this time, there are no known cancer risks for men due to mutations in *BARD1*.

References

- Hall, M et al. 2020 Triple-negative breast cancer risk with pathogenic variants in hereditary cancer predisposition genes. Presented at San Antonio Breast Cancer Symposium 2020.
- Suszynska M, et al. *BARD1* is A Low/Moderate Breast Cancer Risk Gene: Evidence Based on An Association Study of the Central European p.Q564X Recurrent Mutation. *Cancers (Basel)*. 2019 11:740. PMID: 31142030.
- Weber-Lassalle N, et al. Germline loss-of-function variants in the *BARD1* gene are associated with early-onset familial breast cancer but not ovarian cancer. *Breast Cancer Res*. 2019 21:55. PMID: 31036035.
- Shimelis H, et al. Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing. *J Natl Cancer Inst*. 2018 110:855-862. PMID: 30099541.
- Kurian AW, et al. Breast and Ovarian Cancer Penetrance Estimates Derived From Germline Multiple-Gene Sequencing Results in Women. *JCO Precis Oncol*. 2017 Nov;1:1-12. PMID: 35172496.
- Breast Cancer Association Consortium, et al. Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes. *JAMA Oncol*. 2022 8(3):e216744. PMID: 35084436.
- Breast Cancer Association Consortium, et al. Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *N Engl J Med*. 2021 384:428-439. PMID: 33471991.
- SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2023 Mar 24]. Available from <https://seer.cancer.gov/explorer/>.
- Bever TB, et al. NCCN Clinical Practice Guidelines in Oncology®: Breast Cancer Screening and Diagnosis. V 1.2022. Jun 2. Available at <https://www.nccn.org>.
- Daly M et al. NCCN Clinical Practice Guidelines in Oncology®: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. V 3.2023. Feb 13. Available at <https://www.nccn.org>.
- Schaeffer E, et al. NCCN Clinical Practice Guidelines in Oncology®: Prostate Cancer. V 1.2023. Sep 16. Available at <https://www.nccn.org>.

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