

BARD1 Mutations

Cancer Risks and General Management Recommendations

There are currently no national consensus guidelines outlining specific clinical management recommendations for individuals who carry a *BARD1* gene mutation. Additionally, exact lifetime cancer risks associated with *BARD1* mutations are unknown at this time. Current literature may be specific to one particular *BARD1* mutation and/or ethnic population.

Cancer Type	<i>BARD1</i> Mutation Carrier Cancer Risks	General Population Lifetime Cancer Risks	Surveillance/Management Recommendations
Female Breast Cancer	Potential Increase (including triple negative breast cancer)	12.4%	<ul style="list-style-type: none"> • Insufficient evidence to support intervention based on <i>BARD1</i> mutation status alone.¹ • Discuss family history and personal risk factors with a physician to determine appropriate surveillance and management options. • Limited data has been published regarding an increased risk of breast cancer associated with <i>BARD1</i> gene mutations²⁻⁵; two large case-control studies reported approximately two fold risk.^{6,7}
Ovarian Cancer	Unknown	1.3%	<ul style="list-style-type: none"> • Insufficient evidence to offer ovarian cancer screening or risk reducing surgery based on <i>BARD1</i> mutation status alone.¹ • Discuss family history and personal risk factors with a physician in ovarian cancer risk management decision-making. • Studies have reported <i>BARD1</i> mutations in patients with ovarian cancer but the data is limited at this time and may be impacted by type of gene mutation.⁸⁻¹¹

Other Cancer Risks: It is currently unknown if *BARD1* mutations cause a predisposition to other cancers. At this time, there are no known cancer risks for men with a *BARD1* mutation. Patients are encouraged to contact our office every 1-2 years to determine if there is any new information related to the associated risks and clinical management of individuals with *BARD1* mutations.

Implications for Family Members/Reproductive Considerations

- First-degree relatives (i.e., parents, siblings, and children) have a 50% chance to have the familial *BARD1* mutation. Second-degree relatives (i.e., nieces/nephews, aunts/uncles, and grandparents) have a 25% chance to have the familial mutation.
- For carriers of a known mutation, assisted reproduction (with or without egg or sperm donation), pre-implantation genetic testing, and prenatal diagnosis options exist.
- All family members are encouraged to pursue genetic counseling to clarify their risks. Family members can visit www.FindAGeneticCounselor.com to find genetic services near them.

References

*please note that some references may be specific to one particular mutation and/or ethnic population

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