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.

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<th>Cancer Type</th>
<th>BARD1 Mutation Carrier Cancer Risks</th>
<th>General Population Lifetime Cancer Risks</th>
<th>Surveillance/Management Recommendations</th>
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</table>
| Female Breast Cancer | Potential Increase (including triple negative breast cancer) | 12.4% | • Insufficient evidence to support intervention based on BARD1 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 BARD1 gene mutations²-⁵; two large case-control studies reported approximately two fold risk.⁶,⁷ |
| Ovarian Cancer    | Unknown                             | 1.3% | • Insufficient evidence to offer ovarian cancer screening or risk reducing surgery based on BARD1 mutation status alone.¹  
- Discuss family history and personal risk factors with a physician in ovarian cancer risk management decision-making.  
- Studies have reported BARD1 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.
*please note that some references may be specific to one particular mutation and/or ethnic population