Ambry Genetics®

Clinician Management Resource for *BRIP1*

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic variant in the *BRIP1* gene. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS[1]	AGE TO START	FREQUENCY
Ovarian Cancer		
Recommend risk-reducing salpingo-oophorectomy	45–50 years old (or earlier based on a specific family history of an earlier onset ovarian cancer)*	N/A
Female Breast Cancer		
Insufficient data for risk management; managed based on family history	N/A	Individualized
Other		
For patients of reproductive age, counsel about prenatal diagnosis and assisted reproduction, including pre-implantation genetic testing and donor gametes. Discussion should include known risks, limitations, and benefits of these technologies.	Individualized	N/A
Counsel for risk of autosomal recessive condition in offspring.	Individualized	N/A

* The current evidence is insufficient to make a firm recommendation as to the optimal age for this procedure. Based on the current, limited evidence base, a discussion about surgery should be held around age 45–50 years or earlier based on a specific family history of an earlier onset of ovarian cancer.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate. V2.2025. © National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed November 7,2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

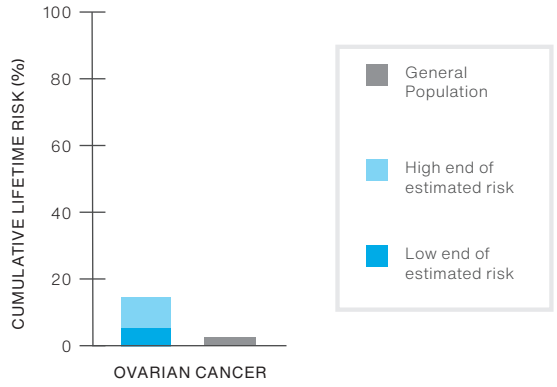
Understanding Your Positive *BRIP1* Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know

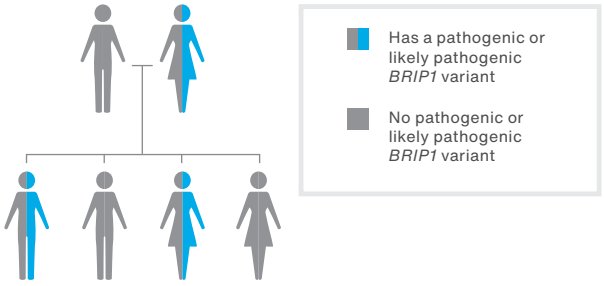
1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the *BRIP1* gene.
2	Cancer risks	You have an increased chance to develop ovarian cancer.
3	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
4	Other medical concerns	Individuals with a pathogenic or likely pathogenic *BRIP1* variant may have an increased risk to have a child with Fanconi anemia, but only if their partner also carries a pathogenic or likely pathogenic variant in the *BRIP1* gene. Fanconi anemia is a rare condition that can cause specific physical characteristics, bone marrow failure, and an increased risk of certain cancers.
5	Family	Family members may also be at risk – they can be tested for the pathogenic or likely pathogenic *BRIP1* variant that was identified in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

BRIP1 Lifetime Cancer Risks*



BRIP1 in the Family

There is a 50/50 random chance to pass on the pathogenic or likely pathogenic *BRIP1* variant to each of your children.



* Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history.

RESOURCES

- American Cancer Society cancer.org
- FORCE facingourrisk.org
- Imerman Angels imermanangels.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *BRIP1* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.

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