



Testing for Hereditary Breast and Gynecologic Cancer



Hereditary Cancer Risk Assessment and/or Testing Are Recommended By:

American College of Obstetrics & Gynecology (ACOG)¹

US Preventive Services Task Force (USPSTF)²

American College of Medical Genetics and Genomics (ACMG) & National Society of Genetic Counselors (NSGC)³

NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®)⁴⁻⁵

GUIDELINES RECOMMEND GENETIC TESTING IF YOUR PATIENTS HAVE PERSONAL OR FAMILY HISTORIES OF ANY OF THE FOLLOWING SIGNS* OF HEREDITARY CANCER:



MULTIPLE CANCERS

- > 2 or more primary cancers in the same person
- > 3 or more cancers on the same side of the family



RARE CANCERS

- > Triple negative breast cancer
- > Male breast
- > Ovarian
- > Pancreatic
- > Metastatic prostate



EARLY-ONSET CANCERS

- > Breast cancer diagnosed before 50
- > Uterine cancer diagnosed before 50



ANCESTRY

- > Ashkenazi Jewish with breast cancer

*Adapted from national published guidelines

HEREDITARY CANCER TESTING OPTIONS*

14-21
DAYS

39
GENES

CancerNext®

39-gene test addressing the most common hereditary cancers; National consensus management guidelines provide recommendations regarding risk management for **all genes** in the panel⁴⁻⁵

14-21
DAYS

13
GENES

BRCAPlus®

13-gene STAT test addressing hereditary breast cancer risk; National consensus management guidelines provide recommendations regarding risk management for **all genes**⁴

14-21
DAYS

19-26
GENES

BRCANext®

19-gene test addressing hereditary breast and gynecologic cancers; National consensus management guidelines provide recommendations regarding risk management for **all genes** on the panel⁴

Option to add 7 additional limited evidence genes**

*Additional testing options available

**Genes are considered "limited evidence" if association with cancer has been suggested but not confirmed due to limited evidence. Management guidelines are not available regarding risk management.

Add +RNAinsight® for More Accurate Results⁶⁻¹⁰

- Identifies more positive results
- Reduces variant of uncertain significance (VUS) rate
- Helps address evidence gaps in non-White populations

+RNAinsight Product Overview

- Has no impact on turnaround time or out-of-pocket cost to patient
- Is available for all tests except for BRCAPlus®
- Kit includes 1 EDTA tube (DNA) and 1 PAXgene® tube (RNA)

5%

Patients Impacted⁹

RNA evidence has been useful for 5% of hereditary cancer testing patients at Ambry.

~1/25

Positive Patients Would Be Missed Without +RNAinsight¹⁰

Results without +RNAinsight would be missed or inconclusive

References

1. ACOG COMMITTEE OPINION, Number 793. Obstet Gynecol. 2019
2. US Preventive Services Task Force. (2019). BRCA-related cancer: risk assessment, genetic counseling and genetic testing
3. Hampel H, ACMG, NSGC, et al., Genetics in Medicine. 2014
4. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V1.2025. ©National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed September 13, 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.
5. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. V2.2024 ©National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed October 19, 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.
6. Landrith T et al. npj Precision Oncology. 2020.
7. Karam R et al. JAMA Network Open. 2019.
8. Horton C et al. NPJ genomic medicine. 2022.
9. Horton C et al. JAMA Oncology. 2023
10. Horton C et al. Expanding the reach of paired DNA and RNA sequencing: Results from 450,000 consecutive individuals from a hereditary cancer cohort; (Oral Presentation Session 84 - Strategies to Interpret Germline Variants in Cancer Predisposition Genes). Presented at the Annual Meeting of The American Society of Human Genetics, November 68, 2024, in Denver, CO.