REFERENCE GUIDE

Ambry Genetics®

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 DAY TAT

CancerNext®

Test addressing the most common hereditary cancers; National consensus management guidelines provide recommendations regarding risk management for **all genes** in the panel[4-5]

7-10 DAY TAT

BRCAplus®

STAT test addressing hereditary breast cancer risk; National consensus management guidelines provide recommendations regarding risk management for **all genes** in the panel[4]

14-21 DAY TAT

BRCANext®

Test addressing hereditary breast and gynecologic cancers; National consensus management guidelines provide recommendations regarding risk management for **all genes** on the base panel[4]

Option to add on select limited evidence genes**

Scan for more information on gene content.

*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 Results[6-10]

- **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 and little to no 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 Impacted[9]

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

~1/25

Positive Patients Would Be Missed Without +RNAinsight®[10]

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 Predispostion Genes). Presented at the Annual Meeting of The American Society of Human Genetics, November 8, 2024, in Denver, CO.

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MKT-ONCO-BRO-20048-EN v8 03.24.25