REFERENCE GUIDE

Ambry Genetics®
A TEMPUS COMPANY

Unlock the Full Genomic Picture to Personalize Cancer Care

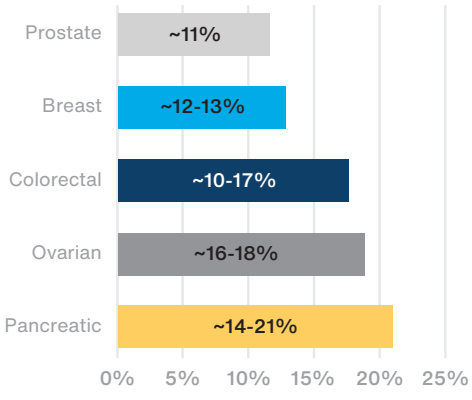
GERMLINE TESTING: AN ESSENTIAL TOOL IN PRECISION ONCOLOGY

Over the years, precision medicine has offered personalized care by leveraging a patient's unique genomic and molecular profile. Yet, germline testing—an essential tool in this approach—remains underutilized, especially in patients with cancer.[1]

Germline testing provides a more complete picture of a patient's genetic predispositions to cancer, which can help:

- Guide treatment decisions, such as:
  - Identifying patients eligible for targeted therapies (e.g., PARP inhibitors)
  - Deciding if risk-reducing surgical intervention is needed
  - Informing clinical trial eligibility for germline-focused therapies
- Inform survivorship care by:
  - Stratifying risk to anticipate future cancer occurrence
  - Guiding proactive screening and prevention measures
- Identify family members at increased risk for developing cancer

Percentage of patients with germline pathogenic variants in cancer predisposition genes[1,2]



By leveraging germline testing in your care strategies, you can enhance treatment personalization and improve patient management across every stage of the cancer care continuum.

Germline Testing For Patients With Cancer Is Recommended By:

- **American Society of Clinical Oncology (ASCO) Clinical Practice Guidelines**[3-6]
- **European Society for Medical Oncology (ESMO) Clinical Practice Guidelines**[7]
- **NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®)**[8-9]

GOING BEYOND TUMOR GENOMIC TESTING

- Tumor genomic testing may reveal findings suggestive of a germline variant. When this happens, guidelines recommend confirming the result through a CLIA-certified lab.
- Additionally, it is not a substitute for comprehensive germline testing. Guidelines recommend that patients meeting germline testing criteria have comprehensive testing performed in a CLIA-certified lab with established experience in hereditary cancer testing, as tumor genomic testing may miss clinically-significant germline variants.

AT MINIMUM, GUIDELINES RECOMMEND GERMLINE TESTING FOR PATIENTS WITH ANY OF THE FOLLOWING:*

EARLY-ONSET CANCERS

- Breast cancer diagnosed at or before age 50
- Colorectal cancer diagnosed before age 50
- Uterine cancer diagnosed before age 50

RARE CANCERS

- Ovarian
- Pancreatic
- Triple negative breast
- Male breast
- Metastatic and/or high-grade prostate

ANCESTRY/OTHER

- Ashkenazi Jewish ancestry
- Known gene mutation in family

SUSPICIOUS SOMATIC FINDINGS

- Suspected pathogenic/likely pathogenic variant in a cancer predisposition gene

ARE YOU TESTING *ALL* OF YOUR ELIGIBLE PATIENTS WHO MEET GUIDELINES?

**Adapted from national published guidelines. Not a comprehensive list. For example, it doesn't include family history-based indications for testing.*

HEREDITARY CANCER TESTING OPTIONS

14-21 DAY TAT

CancerNext®

Guideline-based[8-9], pan-cancer test for hereditary cancer predisposition, including genes associated with the most common hereditary cancers, including breast, ovarian, pancreatic, prostate, colorectal, endometrial, gastric, small bowel, urothelial, and renal.

14-21 DAY TAT

CancerNext-*Expanded*®

Comprehensive, pan-cancer test for hereditary cancer predisposition, including genes associated with a wide range of hereditary cancers, such as breast, ovarian, uterine, colorectal, gastric, pancreatic, prostate, melanoma, renal, central nervous system tumors, pheochromocytoma/paraganglioma, hematologic malignancy, and other rare cancer predisposition conditions.

Scan for more information on gene content.

We accept a variety of specimen types, including blood and saliva.

Add +RNAinsight® to Find More Answers

Testing with concurrent DNA/RNA analyzes functional RNA data to help detect and classify DNA variants, including deep intronic mutations that may be otherwise missed with a DNA-only or reflexive RNA testing approach.**

Identifies more positive patients who would otherwise be missed[10-11]

Reduces ambiguity by reducing the relative VUS rate[10,12]

Provides functional data to address known data gaps in non-white patients[10]

No impact on turnaround time[10]

**Not available for STAT testing.

Interested in Learning More?

Watch the “Guidance on Germline Testing Based on Somatic Test Results” webinar with Jessica Stoll, MS, CGC.

NEED GENETIC COUNSELING SUPPORT?

We recognize that not all organizations have access to a local genetics expert. Your Ambry representative can assist you with third-party pre- and post-test counseling resources, if needed.

READY TO ADD GERMLINE TO YOUR TESTING WORKFLOW?

Contact your Ambry Representative, or scan here and we’ll have your rep contact you.

References

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9. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. V1.2025 ©National Comprehensive Cancer Network, Inc. 2025. All rights reserved. Accessed October 29, 2025. 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.
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