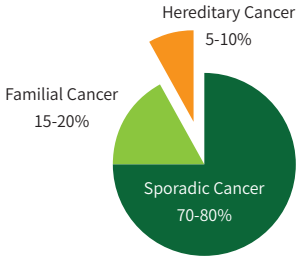
AmoyDx® HANDLE Hereditary Cancer NGS Panel

Why Hereditary Cancer Testing Matters?



Approximately 5–10% of all cancers are caused by inherited mutations in cancer predisposition genes. [1] Multigene panel testing detects more hereditary cases than syndrome-specific approaches. [2]

Clinical guidelines [3,4] recommend germline testing for individuals with personal or family histories of breast, ovarian, colorectal, endometrial, or related cancers.

Comprehensive Hereditary Cancer Testing in One Integrated Workflow

Comprehensive Gene Coverage

- 150 cancer predisposition genes analyzed
- Detection of SNVs/InDels across all genes + CNVs in 8 key genes

Broad Scope of Utility

- Identifies all major hereditary cancer syndromes, including: Hereditary Breast and Ovarian Cancer, Lynch Syndrome, Li-Fraumeni Syndrome, PTEN Hamartoma Tumor, Syndrome (PHTS), and other high-risk genetic disorders
- One test evaluates hereditary risks for pan-cancer types

Automated Reporting with Actionable Insights

Automated Reports delivering actionable insights for tailored surveillance and prevention

- Hereditary cancer syndrome linked to each result
- Cancer types with elevated risk
- Curated interpretation of key variants

Fast and Streamlined Workflow

- Proprietary HANDLE technology with one-tube workflow
- <1 hour hands-on time, <5 hours total library preparation

Addressing Key Gaps in Hereditary Cancer Research

The AmoyDx® HANDLE Hereditary Cancer Panel overcomes the limits of broad or narrow testing approaches:

AmoyDx Panel Vs. Whole Exome Sequencing

- Focuses only on hereditary cancer genes, avoiding irrelevant data
- More affordable and efficient for cancer research
- Easier to interpret, accelerating variant analysis

AmoyDx Panel Vs. Small Gene Panels

- Includes all guideline-recommended genes for solid tumors
- Increases chances of finding actionable variants
- Minimizes need for repeat or expanded testing

Effortless Reporting and Insights with ARAS

ARAS: AmoyDx Report Automation System

>2.1 Result Summary

Genetic Test Results			
Gene	Variant	Zygosity	Interpretation
BRCA1	c.3718C>T:p.(Q1240*) NM_007294.4	Heterozygous	Pathogenic
Clinical Overview			
The Genetic Test Result is Associated with the Following Hereditary Cancer Syndrome (Hereditary Pattern):		The Genetic Test Result is Associated with Elevated Risks for the Following Cancers (Hereditary Pattern):	
Hereditary Breast and Ovarian Cancer Syndrome (AD)		Breast Cancer (AD) Ovarian Cancer (AD) Pancreatic Cancer (AR)	

>2.2 Detailed Description of Genetic Predisposition Variants

Gene	Test Results			
BRCA1	Variant	c.3718C>T:p.(Q1240*) NM_007294.4	Zygosity Interpretation	Heterozygous Pathogenic
	Gene Description: BRCA1, breast cancer type 1 susceptibility protein, is a tumor suppressor (PMID: 30562755) involved in the DNA damage response and DNA repair (PMID: 21203981). BRCA1 germline mutations increase the risk of developing ovarian and/or breast cancer (PMID: 21285145) and somatic mutations are highest in NSCLC, pancreatic, and colon cancers (PMID: 27283171). **Variant Description and Evidence:** This variant is a nonsense mutation, which changes the 1240th amino acid from Glutamine to stop codon. This variant is expected to form functionally impaired or inactivated proteins. This variant is not recorded in the 1000G, ExAC, gnomAD or IJGVD databases; this variant is being reviewed by the BRCAExchange expert group as Pathogenic, the record of this variant in the ClinVar database (ID=54978) is Pathogenic in germline classification (review status: three stars). A multifactor likelihood analysis model based on clinical phenotypic data indicated the Likelihood ratio (LR) for this variant is 2.782 (Family-History LR:2.782) (PMID: 31853058). **Hereditary Cancer Risk:** Heterozygous loss-of-function pathogenic or likely pathogenic variants in BRCA1 are definitively associated with BRCA1-related hereditary cancer predisposition syndrome (autosomal dominant) (ClinGen). Breast cancer: 60%–72% absolute risk for primary breast cancer; 30%–40% 20-year cumulative risk for contralateral breast cancer; >20% 15-year cumulative risk in premenopausal women; 0.2%–1.2% risk of male breast cancer by age 70. Ovarian, pancreatic, and prostate cancers: 39%–58%, <5%, and 7%–26% absolute risks, respectively. Increased risks for cholangiocarcinoma and melanoma. (NCCN Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate V3.2025; Pancreatic Adenocarcinoma V2.2025; Biliary Tract Cancers V1.2025; Prostate Cancer V2.2025; Melanoma: Cutaneous V2.2025)			

>2.3 Variant(s) of Uncertain Significance

Gene	Variant	Zygosity	Interpretation
No variants of unknown clinical significance			

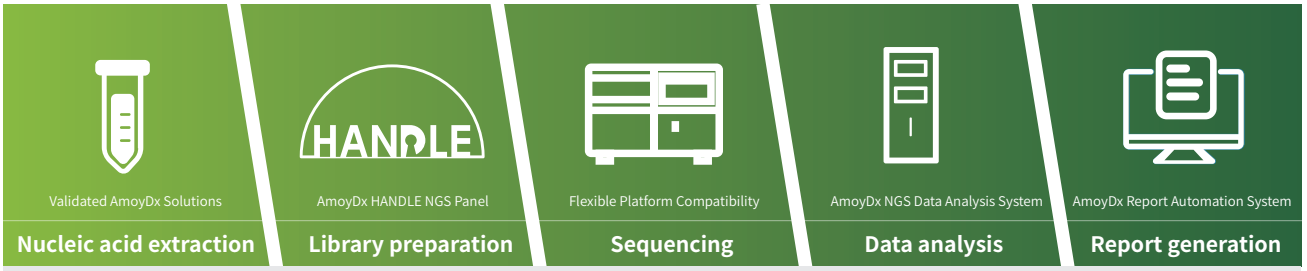
Note:
1.The test results derived from the detection range of the product, using the reference genome version hg19. Variants in this report are nomenclated according to the Human Genome Variation Society (HGVS) variant nomenclature guidelines (http://varnomen.hgvs.org).

>3 Quality Control

QC Parameter		Acceptance	QC Result
DNA	Total DNA Amount (ng)	⩾50	8280
	Total Library Amount (ng)	⩾150	300
Data	CleanQ30	⩾75%	90.56%
	Coverage(20x)	⩾99%	99.5%

Note:
1.Total DNA Amount：The total amount of DNA extracted from the submitted specimen.
2.Total Library Amount：The total amount of the constructed library.

Streamlined Workflow from Sample to Report in 3 days



1. Minimal hands-on time **2. Automated analysis** **3.Instant report generation**

Ordering Information

Catalog Number	8.06.0161
Kit Format	24 tests/kit
Instrument	NovaSeqv6000, NextSeq 500/550Dx, MiSeq / MiSeqDx
Storage & Shelf-life	-25°C to -15°C for 12 months

Citation:
1.Hum Mutat. 2020;41(8):e1-e6.
2.JAMA Oncol. 2021 Feb 1;7(2):312.
3.J Clin Oncol. 2024;42(21):2599-2615.
4.J Natl Compr Canc Netw. 2023;21(10):1000-1010.

For Research Use Only. Not for use in diagnostic procedures.

E-mail: info@amoydx.com

Address: 2 Science Park Drive, Ascent #01-08, Singapore 118222