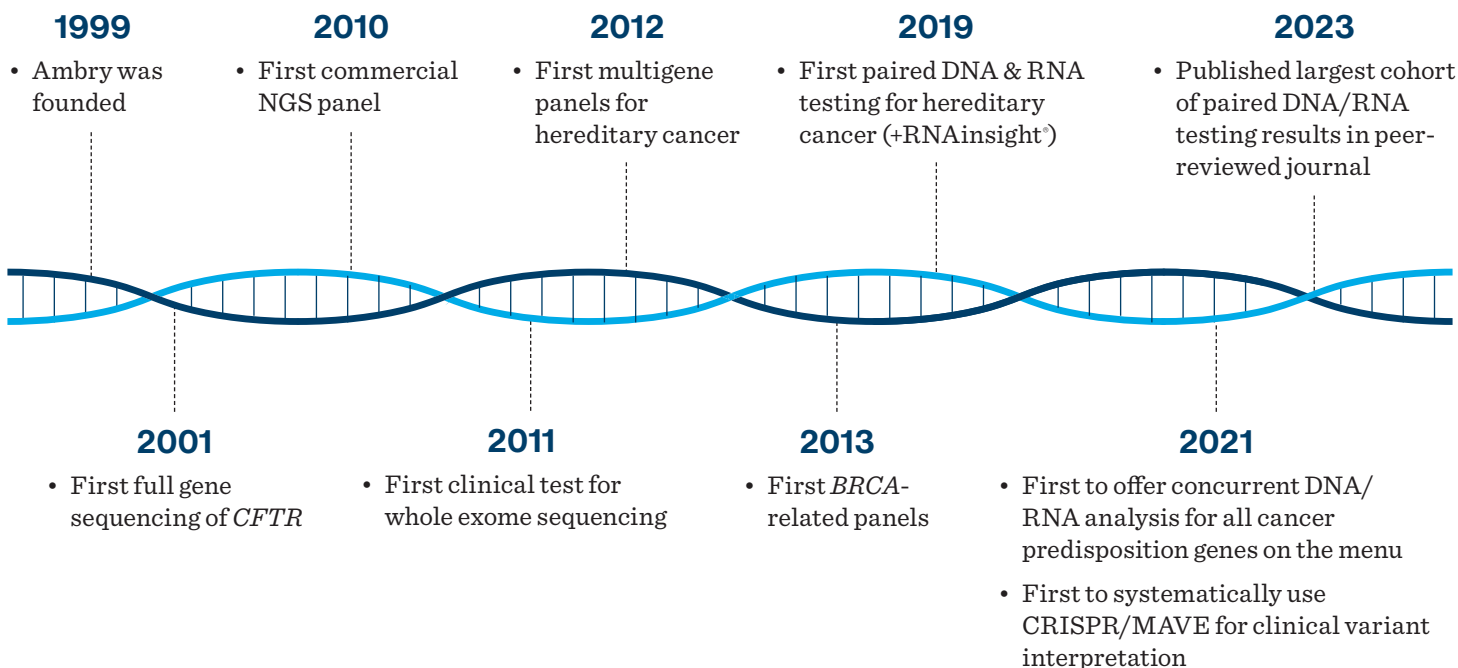


## Hereditary Cancer Portfolio Overview

### Setting the Standard in Genetic Testing

25 YEARS OF INNOVATION AND DISCOVERIES



### +RNAinsight®

Paired DNA/RNA genetic testing with +RNAinsight analyzes functional RNA data to help classify DNA variants. It also identifies deep-intronic mutations that may go undetected with a DNA-only or reflexive RNA testing approach. As a result, diagnostic yield is higher and variant of uncertain significance rate is lower, providing clarity for patients and healthcare providers.<sup>1</sup> This novel functional evidence is especially important in non-White populations that have been underrepresented in research and clinical testing.

## 1-90 Genes

### For Maximum Flexibility

Order +RNAinsight with analysis of any genes on the hereditary cancer menu\*

**5%**

#### Hereditary Cancer Cases Leverage RNA Data

RNA data can benefit all Ambry patients, even those with DNA-only testing<sup>1</sup>

**1/25**

#### Positive Patients Would Be Missed Without RNA

Results without +RNAinsight would be negative or inconclusive<sup>2</sup>

**~6%**

#### Reduction in VUS Rate

+RNAinsight reduces ambiguity for providers and patients\*\*

**12,000**

#### Non-White Patients Have Benefited

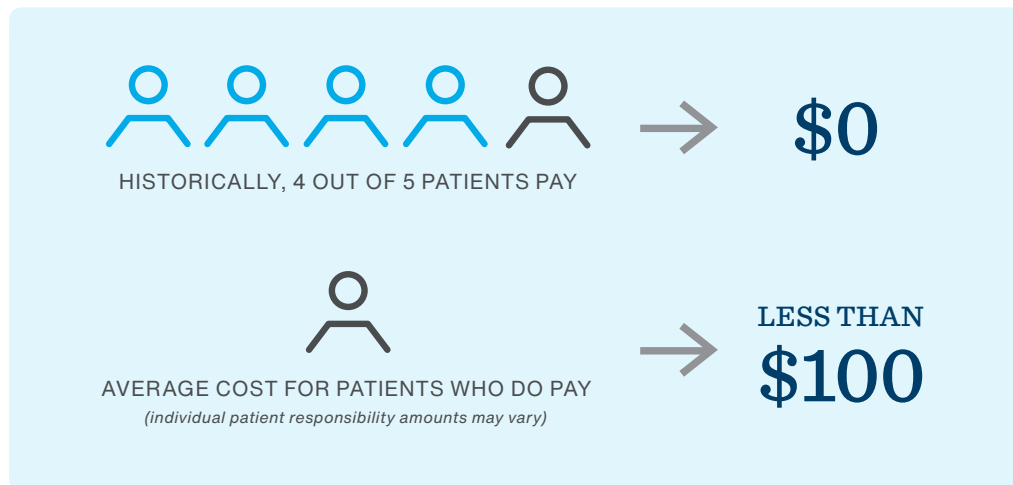
+RNAinsight provided functional data to address known data gaps\*\*

**Technical Details** +RNAinsight analyzes transcripts for up to 90 genes depending on which Ambry Genetics DNA-based Hereditary Cancer Panel it is paired with, and depending on the absence or presence of RNA transcripts expressed in the blood. The results from +RNAinsight are used to provide functional RNA information to further support classification of DNA variants. It is not intended to be used as a stand-alone diagnostic test.

BRCAPlus® 13 genes	BRCANext® 19-26 genes^	ColoNext® 21-26 genes^	CancerNext® 40 genes	CancerNext-Expanded® 77-90 genes^	CustomNext-Cancer® Choose from 90 genes^^
	BRIP1		BRIP1	BRIP1	BRIP1
ATM	ATM		ATM	ATM	ATM
BARD1	BARD1		BARD1	BARD1	BARD1
BRCA1	BRCA1		BRCA1	BRCA1	BRCA1
BRCA2	BRCA2		BRCA2	BRCA2	BRCA2
CHEK2	CHEK2		CHEK2	CHEK2	CHEK2
NF1	NF1		NF1	NF1	NF1
PALB2	PALB2		PALB2	PALB2	PALB2
RAD51C	RAD51C		RAD51C	RAD51C	RAD51C
RAD51D	RAD51D		RAD51D	RAD51D	RAD51D
CDH1	CDH1	CDH1	CDH1	CDH1	CDH1
PTEN	PTEN	PTEN	PTEN	PTEN	PTEN
STK11	STK11	STK11	STK11	STK11	STK11
TP53	TP53	TP53	TP53	TP53	TP53
	MLH1	MLH1	MLH1	MLH1	MLH1
	MSH2	MSH2	MSH2	MSH2	MSH2
	MSH6	MSH6	MSH6	MSH6	MSH6
	PMS2	PMS2	PMS2	PMS2	PMS2
	EPCAM	EPCAM	EPCAM	EPCAM	EPCAM
		APC	APC	APC	APC
		AXIN2	AXIN2	AXIN2	AXIN2
		BMPR1A	BMPR1A	BMPR1A	BMPR1A
		SMAD4	SMAD4	SMAD4	SMAD4
		GREM1	GREM1	GREM1	GREM1
		MBD4	MBD4	MBD4	MBD4
		MSH3	MSH3	MSH3	MSH3
		MUTYH	MUTYH	MUTYH	MUTYH
		NTHL1	NTHL1	NTHL1	NTHL1
		POLD1	POLD1	POLD1	POLD1
		POLE	POLE	POLE	POLE
		RPS20	RPS20	RPS20	RPS20
			BAP1	BAP1	BAP1
			CDKN2A	CDKN2A	CDKN2A
			FH	FH	FH
			FLCN	FLCN	FLCN
			HOXB13	HOXB13	HOXB13
			MET	MET	MET
			TSC1	TSC1	TSC1
			TSC2	TSC2	TSC2
			VHL	VHL	VHL
				AIP	AIP
				ALK	ALK
				CDC73	CDC73
				CDK4	CDK4
				CDKN1B	CDKN1B
				CEPBA	CEPBA
				CTNNA1	CTNNA1
				DDX41	DDX41
				DICER1	DICER1
				EGFR	EGFR
				ETV6	ETV6
				GATA2	GATA2
				KIT	KIT
				LZTR1	LZTR1
				MAX	MAX
				MEN1	MEN1
				MITF	MITF
				NF2	NF2
				PDGFRA	PDGFRA
				PHOX2B	PHOX2B
				POT1	POT1
				PRKAR1A	PRKAR1A
				PTCH1	PTCH1
				RB1	RB1
				RET	RET
				RUNX1	RUNX1
				SDHA	SDHA
				SDHAF2	SDHAF2
				SDHB	SDHB
				SDHC	SDHC
				SDHD	SDHD
				SMARCA4	SMARCA4
				SMARCB1	SMARCB1
				SMARCE1	SMARCE1
				SUFU	SUFU
				TMEM127	TMEM127
				WT1	WT1

## Excellent Coverage, Personalized Support

At Ambry, we understand that there is no “one-size-fits-all” in healthcare. We will work with your patients’ insurance coverage and provide options for accessing our high-quality genetic testing based on your patients’ individualized needs.



## Patient Assistance Program



If your patient needs help with the cost of testing, they can request support through our Patient Assistance Program (PAP). The PAP considers their individual financial situation and provides personalized payment options based on a personalized assessment of their financial need.



Have your patients call or email our Billing Support team at +1 949-900-5795 or [billing@ambrygen.com](mailto:billing@ambrygen.com) with any questions.

## Let us be your trusted partner

All hereditary cancer tests utilize Ambry’s Classifi® program, a proprietary, knowledge-driven engine for gene classification, variant analysis & interpretation, and reporting. The Classifi program delivers the highest quality test results and ensures we leave no stone unturned in getting answers for you and your patients.

Ambry  
**Classifi®**

### References

1. Horton, C., Hoang, L., Zimmermann, H., Young, C., Grzybowski, J., Durda, K., Vuong, H., Burks, D., Cass, A., LaDuca, H., Richardson, M. E., Harrison, S., Chao, E. C., & Karam, R. (2023) Diagnostic outcomes of concurrent DNA and RNA sequencing in individuals undergoing hereditary cancer testing. JAMA Oncology. <https://doi.org/10.1001/jamaoncol.2023.5586>
2. 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 8, 2024, in Denver, CO.

\* Not available for STAT Testing or BRCAplus.

\*\* Based on internal data