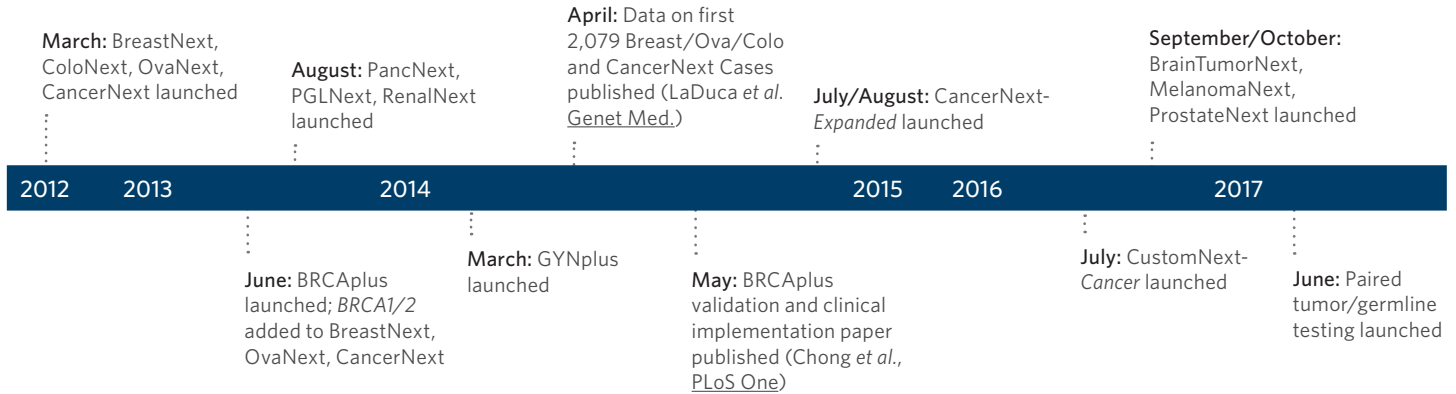


# Hereditary Cancer Panels

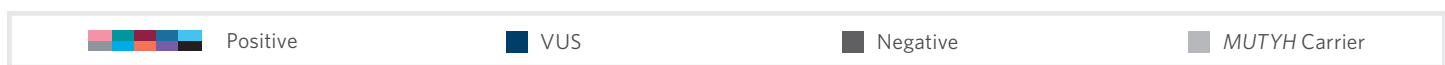
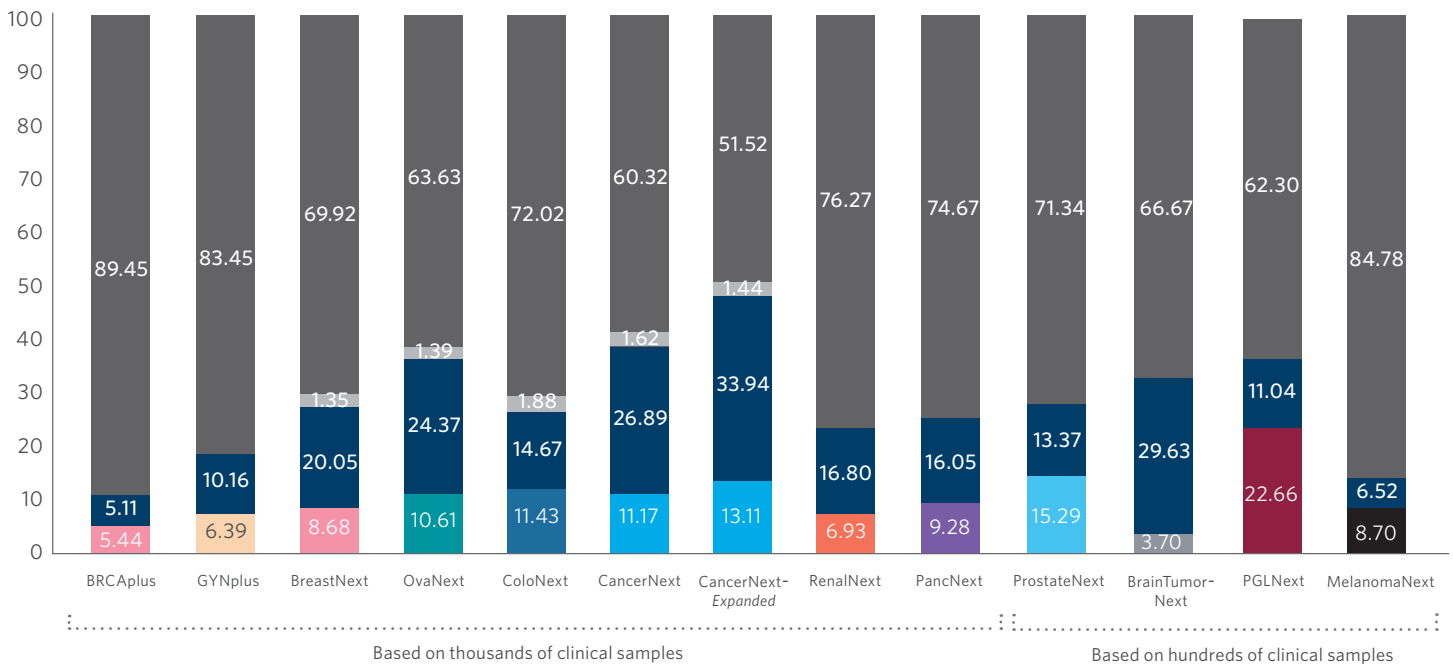
WHITE PAPER October 2017

Ambry is driven to give healthcare providers the most advanced genetic testing information available so they can determine the best treatment options for their patients. We are the first lab to take part in the largest study ever for hereditary cancer, to reduce the chance of either getting a false positive patient report or one that misses mutations. This is at the heart of why we do what we do: to deliver the most accurate information so all human disease can be understood.



## Hereditary Cancer Panel Experience

UNDERSTANDING DISEASE BETTER THROUGH DATA SHARING AND TRANSPARENCY



BRCA1 and BRCA2 VUS Rate: 3.09%

# Hereditary Cancer Multigene Panels

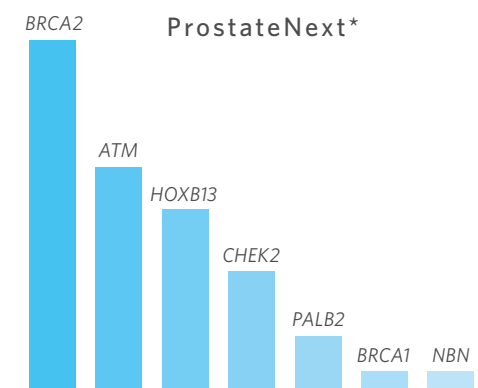
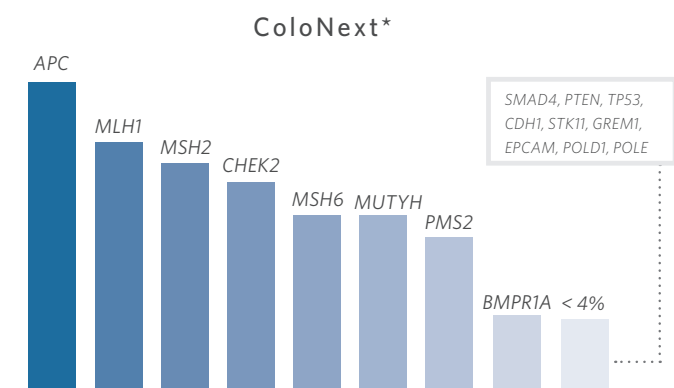
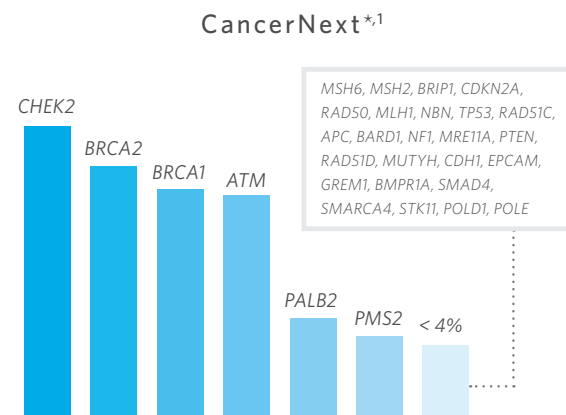
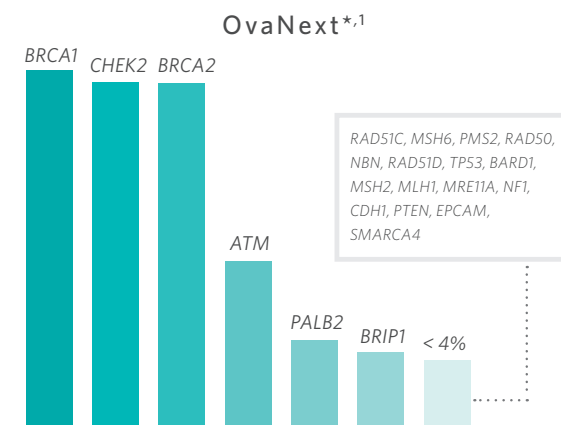
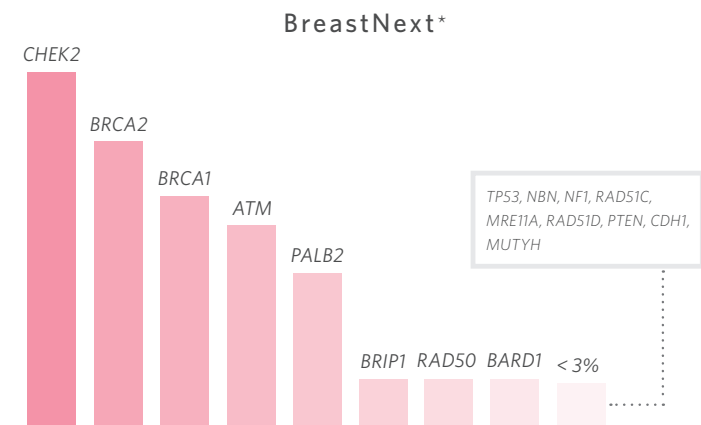
GENES	BRCAnext 8 genes	BreastNext 17 genes	GYNplus 13 genes	OvaNext 25 genes	ColoNext 17 genes	ProstateNext 14 genes	PancNext 13 genes	CancerNext 34 genes	BrainTumorNext 27 genes	MelanomaNext 8 genes	RenalNext 19 genes	PGLNext 12 genes	CancerNext-Exp 67 genes
BARD1		BARD1		BARD1				BARD1					BARD1
BRIP1		BRIP1	BRIP1	BRIP1				BRIP1					BRIP1
MRE11A		MRE11A		MRE11A				MRE11A					MRE11A
NBN		NBN		NBN		NBN		NBN	NBN				NBN
NF1		NF1		NF1				NF1	NF1			NF1	NF1
RAD50		RAD50		RAD50				RAD50					RAD50
RAD51C		RAD51C	RAD51C	RAD51C				RAD51C					RAD51C
RAD51D		RAD51D	RAD51D	RAD51D		RAD51D		RAD51D					RAD51D
ATM	ATM	ATM		ATM		ATM	ATM	ATM					ATM
PALB2	PALB2	PALB2	PALB2	PALB2		PALB2	PALB2	PALB2					PALB2
MUTYH		MUTYH		MUTYH	MUTYH			MUTYH					MUTYH
CHEK2	CHEK2	CHEK2		CHEK2	CHEK2	CHEK2		CHEK2					CHEK2
STK11				STK11	STK11		STK11	STK11					STK11
CDH1	CDH1	CDH1		CDH1	CDH1			CDH1					CDH1
BRCA1	BRCA1	BRCA1	BRCA1	BRCA1		BRCA1	BRCA1	BRCA1					BRCA1
BRCA2	BRCA2	BRCA2	BRCA2	BRCA2		BRCA2	BRCA2	BRCA2		BRCA2			BRCA2
PTEN	PTEN	PTEN	PTEN	PTEN	PTEN			PTEN	PTEN	PTEN	PTEN		PTEN
TP53	TP53	TP53	TP53	TP53	TP53	TP53	TP53	TP53	TP53	TP53	TP53		TP53
MLH1			MLH1	MLH1	MLH1	MLH1	MLH1	MLH1	MLH1		MLH1		MLH1
MSH2			MSH2	MSH2	MSH2	MSH2	MSH2	MSH2	MSH2		MSH2		MSH2
MSH6			MSH6	MSH6	MSH6	MSH6	MSH6	MSH6	MSH6		MSH6		MSH6
PMS2			PMS2	PMS2	PMS2	PMS2	PMS2	PMS2	PMS2		PMS2		PMS2
EPCAM			EPCAM	EPCAM	EPCAM	EPCAM	EPCAM	EPCAM			EPCAM		EPCAM
SMARCA4				SMARCA4				SMARCA4	SMARCA4				SMARCA4
APC					APC		APC	APC	APC				APC
BMPR1A					BMPR1A			BMPR1A					BMPR1A
SMAD4					SMAD4			SMAD4					SMAD4
CDKN2A							CDKN2A	CDKN2A	CDKN2A	CDKN2A			CDKN2A
CDK4								CDK4		CDK4			CDK4
GREM1					GREM1			GREM1					GREM1
POLD1					POLD1			POLD1					POLD1
POLE					POLE			POLE					POLE
HOXB13						HOXB13		HOXB13					HOXB13
DICER1				DICER1				DICER1	DICER1				DICER1
BAP1										BAP1	BAP1		BAP1
FLCN											FLCN		FLCN
MET											MET		MET
MITF										MITF	MITF		MITF
TSC1									TSC1		TSC1		TSC1
TSC2									TSC2		TSC2		TSC2
VHL									VHL		VHL	VHL	VHL
FH											FH	FH	FH
SDHA											SDHA	SDHA	SDHA
SDHB											SDHB	SDHB	SDHB
SDHC											SDHC	SDHC	SDHC
SDHD											SDHD	SDHD	SDHD
SDHAF2												SDHAF2	SDHAF2
MAX												MAX	MAX
RET												RET	RET
TMEM127												TMEM127	TMEM127
MEN1									MEN1			MEN1	MEN1
AIP									AIP				AIP
ALK									ALK				ALK
CDKN1B									CDKN1B				CDKN1B
NF2									NF2				NF2
PHOX2B									PHOX2B				PHOX2B
POT1									POT1				POT1
PRKARIA									PRKARIA				PRKARIA
PTCH1									PTCH1				PTCH1
SMARCB1									SMARCB1				SMARCB1
SMARCE1									SMARCE1				SMARCE1
SUFU									SUFU				SUFU
RB1										RB1			RB1
XRCC2													XRCC2
BLM													BLM
FANCC													FANCC
GALNT12													GALNT12

AVERAGE TAT*	7-10	14.1	14.6	14	14.6	14.5	14	14.4	15.8	14.2	15	15.2	14.9
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\* Calendar days  
 CustomNext-Cancer allows you to choose your own combination of up to 68 genes. All genes on CancerNext-Expanded plus NTHL1 are available to order. TAT is 14-21 calendar days.  
 \* Amby's turnaround time represents the time that it takes for Amby to perform the requested testing. When all necessary clinical and family history information is provided with the sample, results are typically completed within 14 days. We will notify you in the unusual event that results will take longer than 21 days.

# Mutation Distributions

ORDERING THE RIGHT TEST CAN PROVIDE THE MOST ACCURATE AND COMPREHENSIVE ANSWERS



Our ongoing participation in numerous research studies and collaborations is so that we can better assess clinical validity of gene-disease relationships. For more information about our research visit: [ambrygen.com/research-and-collaborations](http://ambrygen.com/research-and-collaborations)

Mutation distribution data for our other hereditary cancer panels is available upon request.

\* Excludes MUTYH carriers, APC p.I1307K, and/or CHEK2 p.I157T depending on the genes included in the panel

<sup>1</sup> Data not yet available for DICER1 and/or HOXB13, added March 2017