

HyperPETE panel

Panel Name	Panel Size (kb)	General Application Space	Details
Common Hereditary Oncology Panel	144	Hereditary Genetics & Oncology	Hereditary Breast and Ovarian Cancers, BRCA1 & BRCA2, Hereditary Colorectal Cancer
Newborn Screening Panel	215	Hereditary Genetics & Oncology	Fits well with carrier screening & NIPT offered by NIPT IBT
Lung Cancer Fusion Panel	20 (~150 fusions)	Somatic Oncology	RNA Fusions
Oncology Hot Spot Panel	25	Somatic Oncology	Actionable targets (gene/mutations)
Pan Cancer Panel	200	Somatic Oncology	5441 target regions and 80 cancer related genes

Common Hereditary Oncology Panel gene list

Gene List				
APC	CDK4	MEN1	PALB2	SDHA
ATM	CDKN2A	MLH1	PDGFRA	SDHB
AXIN2	CHEK2	MSH2	PMS2	SDHC
BARD1	CTNNA1	MSH3	POLD1	SDHD
BMPR1A	DICER1	MSH6	POLE	SMAD4
BRCA1	EPCAM	MUTYH	PTEN	SMARCA4
BRCA2	GREM1	NBN	RAD50	STK11
BRIP1	HOXB13	NF1	RAD51C	TP53
CDH1	KIT	NTHL1	RAD51D	TSC1
TSC2	VHL			

Primary bed file - ~144 Kb
Capture bed file - ~200 Kb

Newborn Screening Panel gene list

Gene List					
ABCD1	CD3D	FAH	IL2RG	NBN	SLC46A1
ACADM	CD3E	FOXN1	IL7R	NHEJ1	SMN1
ACADVL	CD3G	GAA	IVD	ORAI1	STAR
ACAT1	CD40LG	GALE	JAK3	PAH	STAT5B
ADA	CFTR	GALK1	LIG4	PAX8	STIM1
AK2	CHD7	GALT	MCCC1	PCCA	TBX1
ASL	CORO1A	GATA2	MCCC2	PCCB	TSHR
ASS1	CYP11A1	GCDH	MCEE	PNP	WAS
ATM	CYP11B1	HADHA	MMAA	POR	ZAP70
BCKDHA	CYP17A1	HADHB	MMAB	PRKDC	
BCKDHB	CYP21A2	HBB	MMADHC	PTPRC	
BLNK	DBT	HLC5	MMUT	RAC2	
BTD	DCLRE1C	HMGCL	MTHFD1	RAG1	
BTK	DKC1	HSD3B2	MTHFR	RAG2	
CBS	DOCK2	IDUA	MTR	RMRP	
CD247	DOCK8	IGHM	MTRR	SLC22A5	

Primary bed file - ~215 Kb
Capture bed file - ~300 Kb

Oncology HotSpot Panel gene list

Gene List				
AKT1	EGFR	GNAQ	NF1	RET
ALK	ERBB2	GNAS	NRAS	STK11
APC	ESR1	HRAS	NTRK3	TP53
ATM	FBXW7	IDH1	PDGFRA	TSC1
BRAF	FGFR1	IDH2	PIK3CA	TSC2
BRCA1	FGFR2	JAK2	POLE	
CDKN2A	FGFR3	KIT	PTCH1	
CTNNB1	GNA11	KRAS	PTEN	

Primary bed file - ~25 Kb
Capture bed file - ~36 Kb

Pan Cancer Panel v1

Gene List					
ABL1	CTNNB1	FGFR3	MAP2K2	NTRK3	SETD2
AKT1	DDR2	FLT3	MET	PDGFRA	SMAD4
ALK	DNMT3A	GNA11	MLH1	PIK3CA	SMARCB1
APC	EGFR	GNAQ	MPL	PMS2	SMO
ARID1A	ERBB2	GNAS	MSH2	POLE	SRC
ATM	ERBB3	HNF1A	MSH6	PTCH1	STK11
ATR	ERCC2	HRAS	MTOR	PTEN	TERT
BRAF	ERCC3	IDH1	NF1	PTPN11	TP53
BRCA1	ERCC4	IDH2	NF2	RAD50	TSC1
BRCA2	ESR1	JAK1	NFE2L2	RAD51C	TSC2
BRIP1	EZH2	JAK2	NOTCH1	RAD51D	VHL
CDH1	FANCA	JAK3	NPM1	RAF1	
CDK4	FBXW7	KIT	NRAS	RB1	
CDKN2A	FGFR1	KRAS	NTRK1	RET	
CSF1R	FGFR2	MAP2K1	NTRK2	ROS1	

Primary bed file - ~200 Kb
Capture bed file - ~300 Kb
Includes 190 MSI Loci

Lung Cancer Fusion Panel

Target	Gene	Target Exons																											
5'	ALK	2	4	6	10	16	17	18	19	20	21	22	23	26															
5'	PPARG	1	2	3																									
5'	BRAF	2	7	8	9	10	11	12	15	16																			
3'	BRAF	1	3	7	8	10	13																						
5'	EGFR	7	8	9	16	19	20																						
3'	EGFR	1	24	25																									
5'	FGFR1	2	3	4	5	6	7	8	9	10	11	17																	
3'	FGFR1	12	17																										
5'	FGFR2	2	5	7	8	9	10																						
3'	FGFR2	16	17																										
5'	FGFR3	3	5	8	9	10																							
3'	FGFR3	16	17	18																									
5'	MET	2	4	5	6	13	14	15	16	17	21																		
3'	MET	2	13																										
5'	NRG1	1	2	3	4	6	8																						
3'	NRG1	1																											
5'	NTRK1	1	2	4	6	8	10	11	12	13	14																		
5'	NTRK2	5	7	9	11	12	13	14	15	16	17																		
5'	NTRK3	4	7	10	12	13	14	15	16																				
3'	NTRK3	13	14	15																									
5'	RET	2	4	6	8	9	10	11	12	13	14																		
5'	ROS1	2	4	7	31	32	33	34	35	36	37																		
3'	AXL	19	20																										
5'	PDGFRA	10	11	12	13	14																							
3'	PDGFRA	7																											
5'	PDGFRB	8	9	10	11	12	13	14																					

Housekeeping Genes

■ VCP
 ■ RAB7A
 ■ CHMP2A
 ■ HPRT1

Used as positive controls

- Captured library generation
- Captured library metrics