# BRCA 1/2 Panel

Germline and Somatic Cancer

### **KEY FEATURES**

1. Targets the whole CDS (+/- 40)	Target regions not only covering the CDS regions but expanded to +40 and -40 of CDS to detect splicing site variants		
and promoter regions of BRCA 1/2 with high specificity	Probes specifically designed for detecting deletion, duplication, and large rearrangement		
2. Compatible with a variety of sample types	No compromise on panel performance even with of using DNA from challenging specimen types such as blood and FFPE		
3. Market-leading panel performance in uniformity and coverage	Designed to target whole exon regions of BRCA 1, 2 gene with 100% coverage (RefSeq) and validated to yield 100% coverage		

### **SPECIFICATION**

Gene count*	BRCA 1/2 genes	
Covered region	Whole CDS (+/- 40bp), UTR, Promoter	
Target size	23 kb	
Mutation type	SNV, Indel, CNV	
Sample type(amount)	Blood (> 50 ng of fragmented DNA), FFPE	
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore	
Sensitivity	> 95% for all variant types at 5% VAF	
Specificity	99.9% (SNV), 99.5% (Indel)	
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)	

\* Gene Add-on Service: Genes can be added or removed by customer demand

### PANEL PERFORMANCE

## 1. Superior Panel Performance Compared to Competitor Product



#### 2. SNV, CNV Analysis

BRCA1, S1556G & S1389S / BRCA2, N372H & L1521L / BRCA1 CNV plot

Gene	Mutation	Amino AcidC	Sequencing			Vari - Alle
Gene	Туре	hange	Total	Ref.	Alt.	Frequ
BRCAI	Non-SYN	p.S1556G	634	315	301	48.7
BRCAI	SYN	p.\$1389\$	876	501	370	42.4
BRCA2	Non-SYN	p.N372H	396	213	181	45.9
BRCA2	SYN	p.L1521L	289	0	281	99.2

### **PACKAGE COMPOSITION**

Package name	Compositions		IS
Target Enrichment	Target capture Probe		-
Standard	Target Enrichment	Library prep Kit	-
All-In-One	reagents		Beads / Polymerase









Package option	Options		
Pooling method	Single Reaction	Pre-capture Pooling	
Library Preparation kits	Standard Kit	EP-kit	
Hybridization Enhancer	Included	Not included	