OncoRisk Panel

Hereditary Cancer
(Germline cancer risk)

KEY FEATURES

1. Comprehensive analysis of oncogenes	Analyze 31 oncogenes associated with inherited cancer and precisely selected from contract research organizations and numerous research studies
2. Robust bioinformatics system for large deletion analysis	Receive bioinformatics results for large deletion analysis provided by Celemics proprietary bioinformatics analysis system
3. Used for Homologous Recombination Deficiency (HRD) testing	Provides information for HDR grade computation to aid precision medicine for tumor treatment

SPECIFICATION

Gene count*	31 genes
Covered region	Whole CDS
Target size	96 kb
Mutation type	SNV, Indel, CNV, Rearrangment
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	99.90% (SNV), 99.50% (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

GENE LIST

OncoRisk Panel	APC	ATM	BARD1	BLM	BMPR1A	BRCA1	BRCA2	BRIP1	CDH1	CDK4	CDKN2A	CHEK2	EPCAM
	MLH1	MRE11A	MSH2	MSH6	MUTYH	NBN	PALB2	PMS2	PRSS1	PTEN	RAD50	RAD51C	RAD51D
	SLX4	SMAD4	STK11	TP53	VHL								

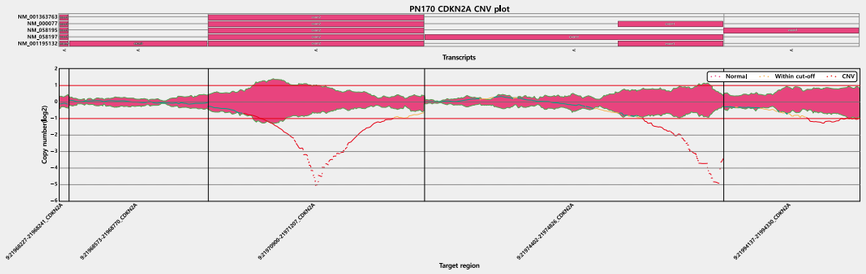
PANEL PERFORMANCE

1. SNV Analysis Example

Gene	Mutation Type	Amino Acid Change	Total Depth	REF Depth	ALT Depth	Variant Allele Frequency
APC	SYN	p.S1738S	1008	590	415	41.17%
ATM	Non-SYN	p.D1853N	417	200	217	52.04%
BARD1	Non-SYN	p.R658C	829	435	394	47.53%
BMPR1A	Non-SYN	p.P2T	621	309	311	50.08%
BRCA1	SYN	p.S1389S	802	460	342	42.64%
BRCA2	SYN	p.V2171V	1026	0	1026	100%
BRIP1	SYN	p.Y1137Y	844	3	840	99.53%
PMS2	Non-SYN	K541E	686	0	646	100%
PRSS1	SYN	p.N246	921	0	921	100%
RAD51D	Non-SYN	p.L152IL	971	0	971	100%

2. CNV Analysis Example

Higher sequencing depths in the target regions, enabling accurate CNV analysis



PACKAGE COMPOSITION

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

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

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