# OncoRisk Panel

A target enrichment panel designed to analyze 31 genes that are associated with Breast, Ovarian, Colorectal, Endometrial, Melanoma, Pancreatic, Gastric, Prostate and Lung cancers. Utilizing Next Generation Sequencing, our target enrichment method allows scientists to specifically isolate whole CDS region of oncogenes and thereby increases the sensitivity of detecting genetic mutations

Performance

- **Clinical Grade of Target Enrichment**
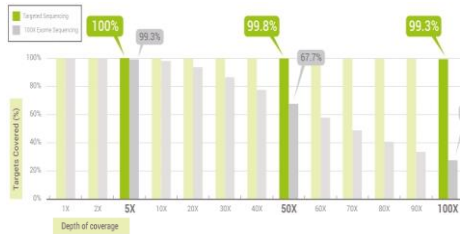
100.0% of coverage with proven test data. (seq. amount: 200Mb)
Possible number of sample in a single Miseq run : approx. 32ea

Source : Celemics, Performance of OncoRisk Kit. Jan.2015

- **Analysis of Single Nucleotide Variation (SNV) and Copy Number Variation (CNV)**

Gene	Mutation Type	Amino Acid Change	Total Depth	REF Depth	ALT Depth	Variant Allele Frequency
APC	SYN	p.S 1738 S	1008	590	415	41.17%
ATM	Non-SYN	p.D 1853 N	417	200	217	52.04%
BARD1	Non-SYN	p.R 658 C	829	435	394	47.53%
BMPR1A	Non-SYN	p.P 2 T	621	309	311	50.08%
BRCA1	SYN	p.S 1389 S	802	460	342	42.64%
BRCA2	SYN	p.V 2171 V	1026	0	1026	100%
BRIP1	SYN	p.Y 1137 Y	844	3	840	99.53%
CDH1	SYN	p.A 692 A	732	398	334	45.63%
EPCAM	Non-SYN	p.M 115 T	889	441	448	50.39%
MSH6	SYN	p.T 1102 T	292	143	149	51.03%
MUTYH	Non-SYN	p.Q 324 H	331	167	164	49.55%
NBN	SYN	p.P 672 P	604	299	305	50.50%
PMS2	SYN	p.K 541 E	646	0	646	100%
PRSS1	SYN	p.N 246 N	921	0	921	100%
RAD51D	SYN	p.R 53 Q	971	0	971	100%
SLX4	SYN	p.N 1500 N	993	493	500	50.35%
TP53	Non-SYN	p.P 33 R	850	438	410	48.24%
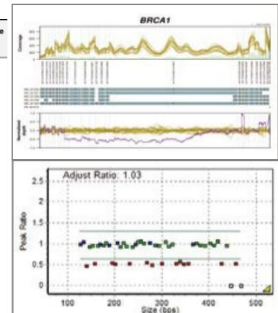
Studies have shown the specific genes carrying mutation have potential risk to develop certain types of cancers.

CDHI / Hereditary diffuse GASTRIC CANCER

Lifetime risk of developing cancer BY AGE 80

MEN 68%

WOMEN 87%
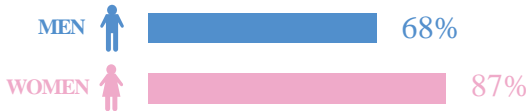
TP53 / Li-Fraumeni syndrome

lifetime risk of developing cancer BY AGE 30 21~49%

LIFETIME RISK lifetime risk of developing cancer 68~93%

1. Pharoah, P.D., et al., Incidence of gastric cancer and breast cancer in CDH1 (E-cadherin) mutation carriers from hereditary diffuse gastric cancer families. Gastroenterology, 2001. 121(6): p. 1348-53.
2. Hwang, S.J., et al., Germline p53 mutations in a cohort with childhood sarcoma: sex differences in cancer risk. Am J Hum Genet, 2003. 72 (4): p. 975-83.

Reference

Specifications

Targets	
Covered Region	CDS + Intron(Gene fusions)
Target size	97 kb
Target Enrichment	In-solution Hybridization
Minimum input DNA	> 50ng
Multiplexing	Up to 384 indexing available
Compatible platforms	All Illumina, Ion Torrent, MGIseq
Assay Genes	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