CancerScreen Comprehensive Panel

CELEMICS

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Identifying and characterizing the molecular fingerprint for tumor cell is important as it can implicate with respect to diagnosis, prognosis, or treatment plans for each individuals. Celemics' CancerScreen Comprehensive Panel is designed to detect all types of variants, including SNV, Indel, CNV, rearrangements, and other immuno-oncology markers. The panel consists total of 765 genes enabling highly accurate and comprehensive analysis for identifying significant mutations and variants for solid tumors.

Key Features

Comprehensive analysis of most common genes related to solid tumors in a single NGS run	Sensitive detection with only 10 ng of gDNA	Market-leading on-target ratio and uniformity	Simple and time-saving workflow
Cost-effective methodology leading to excellent result quality	Complete Bioinformatics solution available	NGS panel applicable for companion diagnostics and drug therapy	Easy customization such as gene add-on services
Ability to detect rearrangement in NTRK, ALK, RET, and ROS1 genes	Superior capture performance in hard-to-capture regions, such as GC/AT-rich and homologous regions	Flexible integration on various sequencers, including Illumina, Thermo Fisher, and MGI	One-stop solution available; from sample prep to BI

Product Specifications

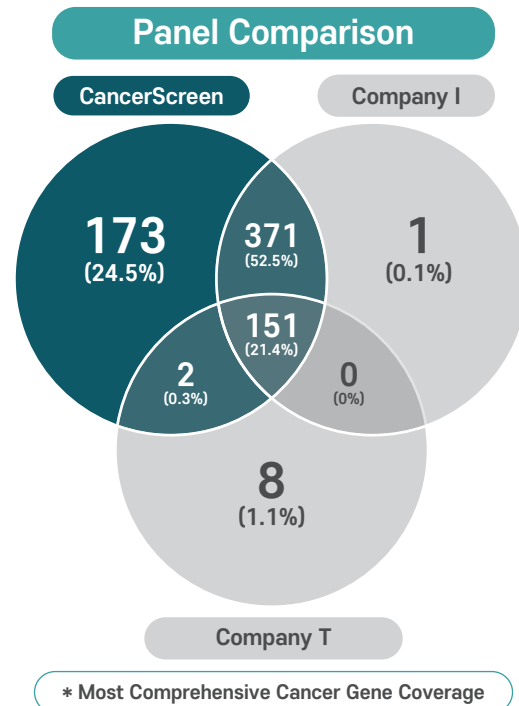| CancerScreen Comprehensive Panel	
Gene Numbers	765 genes (697 genes for DNA, 68 genes for RNA)
Target Regions	CDS, Hotspot variant, UTR, Intronic, Intergenic regions, and MSI markers
Variant Types	SNV, InDel, CNV, Fusion, MSI, TMB
Target Size	2.32 Mb (DNA) & 201 Kb (RNA)
Sample Types	Blood and Tissue (FFPE, Frozen Tissue)
Min. Input Amount of DNA	> 10 ng
NGS Platforms	All Illumina, Thermo Fisher, MGI Platforms

Expected throughput: ∗ NextSeq 550: ~14 samples / flowcell ∗ Ion 550 chip: ~3-4 samples / chip

⊙ Product Description

The CancerScreen Comprehensive panel is designed to detect all variant types and the immuno-oncology markers, microsatellite instability (MSI), and other possible relevant variants, which are crucial biomarkers for cancer immunotherapy. The panel consists of 697 genes for DNA and 68 genes for RNA, and is also designed to detect Epstein-Barr virus (EBV) and Human Papillomaviruses (HPV) allowing for in-depth analysis of cancer associated genes. For CNV analysis, different cut-off is applied according to the ratio of cancer cells.

With Celemics' exclusive probe design and assay optimization technologies, the panel ensures superior performance in terms of detection sensitivity and specificity, enabling time-saving and cost-effective NGS in various oncology-related applications.
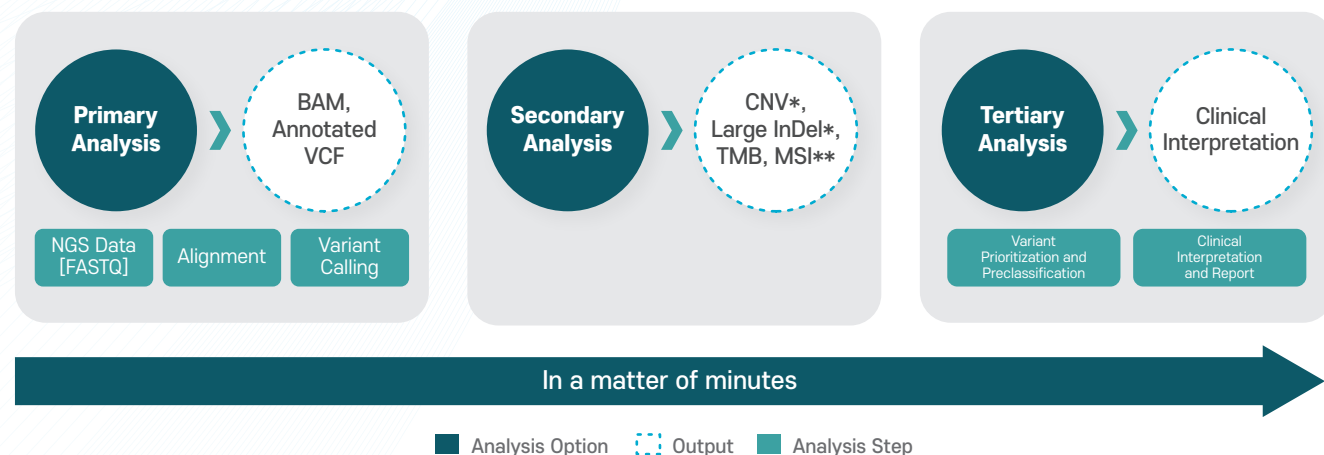
Along with the panels and reagents, Celemics also offers client-specific full bioinformatics support, with the report consisting of primary, secondary, and tertiary analyses for in-depth understanding and interpretation of sequencing data. Also, Celemics offers flexible customization of the ready-to-use panels; if the gene of interest does not exist in the panel or if you wish to expand the target region of interest, it can be added separately through our gene add-on service and can be integrated on to various sequencing platforms, such as Illumina, Ion Torrent, and MGI.

Panel Comparison

CancerScreen | Company I

173 (24.5%)
371 (52.5%)
1 (0.1%)
151 (21.4%)
2 (0.3%)
0 (0%)
8 (1.1%)

Company T

∗ Most Comprehensive Cancer Gene Coverage

● Workflow

Library Preparation → **Target Enrichment** → **Sequencing Run** → **Bioinformatics Analysis**

● CAS (Celemics Analysis Service) Workflow

Primary Analysis > BAM, Annotated VCF

NGS Data [FASTQ] | Alignment | Variant Calling

Secondary Analysis > CNV∗, Large InDel∗, TMB, MSI∗∗

Tertiary Analysis > Clinical Interpretation

Variant Prioritization and Preclassification | Clinical Interpretation and Report

In a matter of minutes

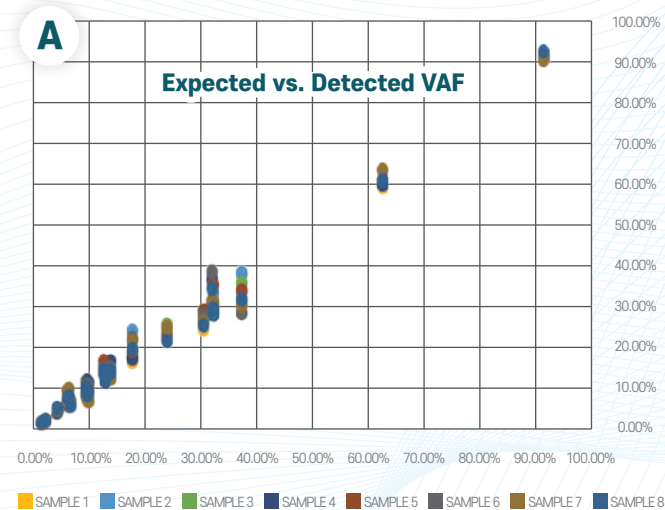■ Analysis Option ⌁ Output ■ Analysis Step

Celemics Analysis Service (CAS) can provide client-specific BI solutions; ranging from simple annotated VCF even to complete clinical interpretations.

Performance Data - DNA

	On target read ratio	Uniformity (St. Dev)
Single sample	84.72%	0.56
8-pooling sample	84.53%	0.57

Table 1

CancerScreen Comprehensive DNA Panel performance statistics
DNA panel demonstrated on-target ratio of 84.72% and 84.53% for single and 8-pooling sample respectively, showing reproducible results regardless of single or pooled sample library with robust uniformity

A

Expected vs. Detected VAF

SAMPLE 1   SAMPLE 2   SAMPLE 3   SAMPLE 4   SAMPLE 5   SAMPLE 6   SAMPLE 7   SAMPLE 8

B

	Expected AF % (at COA)	Detected %
NRAS p.Q61K	12.11	14.39
ALK 3`UTR insertion	9.23	9.44
PIK3CA p.H1047R	17.36	19.63
EGFR p.E746_A750 (15bp del)	1.66	1.56
EGFR p.T790M	1.02	1.09
EGFR p.L858R	3.80	4.34
MET p.L238fs*25	5.82	7.03
BRAF p.V600E	12.45	12.38
KRAS p.G12D	5.73	7.09
TP53 p.P72R	90.92	91.41

Figure 1A&B **Expected vs. Detected VAF correlation and list of variants for AF comparison**

1A) The expected and actual VAF of variants. Each sample consists of 8-pooled sample libraries. The actual VAF showed relative concordance for the corresponding target genes, demonstrating the detection capability and reproducibility of the panel, regardless of the single or pooled sample libraries.

1B) Illustrate few of selected variants were assessed to have relatively similar allele frequencies.

Target genes: NRAS, ALK, CTNNB1, PIK3CA, KIT, FBXW7, APC, EGFR, MET, BRAF, NOTCH1, RET, KRAS, FLT3, BRCA2, TP53

Performance Data - RNA

LMNA   NTRK1

Gene 1	Gene 2	Gene 1	Gene 2
CCDC6	RET	LMNA	NTRK1
CD74	ROS1	NCOA4	RET
EGFR	SEPTIN14	PAX8	PPARG
EML4	ALK	SLC34A2	ROS1
ETV6	NTRK3	SLC45A3	BRAF
FGFR3	BAIAP2L1	TFG	NTRK1
FGFR3	TACC3	TMPRSS2	ERG
KIF5B	RET	TPM3	NTRK1
EGFR exon 2-7 skipping		MET exon 14 skipping	

Figure 2A **Fusion breakpoint IGV**

Generated with STAR-Fusion for renown rearrangement LMNA & NTRK1 pair. The IGV plot illustrates the successful capture and data generation in regions with gene rearrangements using CancerScreen Comprehensive Panel

Table 3 **List of fusion for RNA panel**

CancerScreen Comprehensive RNA panel can detect above listed fusion points for assessing RNA variants associated with somatic cancer

Order Information

Category	Sub-category	Sequencing Platform	Package option	Product unit
Ready-to-use	CancerScreen Comprehensive Panel	• Illumina • Thermo Fisher • MGI	1) All-in-one Package 2) Standard Package 3) Target Enrichment Package	• Reaction basis : 16, 48, 96 rxn • Sample basis : 16, 96, or more *Pre-capture pooling options: 4, 8, 12

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