CELEMICS

PRODUCTS & SERVICES 2022

CELEMICS

Innovative NGS-Based Products with Novel Sequencing Technology

Celemics Takes Customization to the Next Level

NGS industries are rapidly evolving and our customers are in greater need of technical support in various research fields and markets.
Just like a chameleon utilizing its colored pigments and crystals to adapt to its surroundings, we proactively pioneer and adapt to the everchanging NGS markets with our cutting-edge technology to support our customer's needs.
We are keenly focused on bringing the right solution to customers by elaborately customizing to individual customer's needs and goals.

CELEMICS

Celemics confidently offers customization services
and preset panels of the utmost quality

CONTENTS

CELEMICS' PRODUCTS & SERVICES

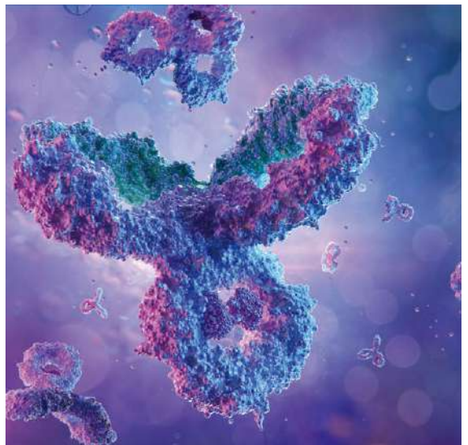
CELEMICS PRODUCTS & SERVICES OVERVIEW

CANCER RESEARCH

- Customized Targeted Sequencing
- BRCA 1/2 (Breast Cancer)
- OncoRisk
- CancerScreen / CancerMaster
- G-Mendeliome Clinical Exome Sequencing
- G-Mendeliome Disease Specific
- Customized RNA Sequencing
- Customized Methylation Sequencing
- Circulating Tumor DNA - Colorectal, Lung, Breast

MOLECULAR BREEDING (ANIMAL / PLANT RESEARCH)

- Customized High-Throughput Genotyping
- Africa Swine Fever Virus



ANTIBODY DISCOVERY

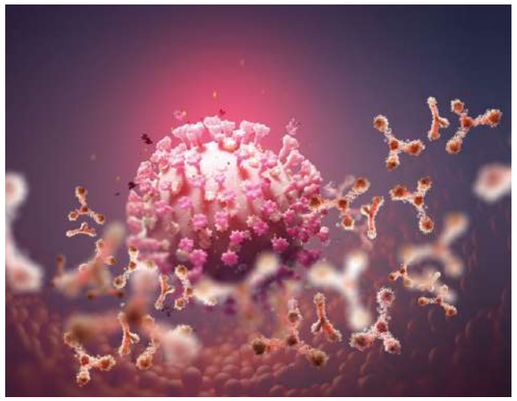
- TrueRepertoire™
- Immune Repertoire Profiling

DIAGNOSTICS & INHERITED DISEASES

- Customized Targeted Sequencing
- G-Mendeliome Clinical Exome Sequencing
- G-Mendeliome Disease Specific
- Cancer Panels (Somatic, Germline, ctDNA)
- Customized RNA Sequencing
- Customized Methylation Sequencing
- PharmacoScreen Panel
  : Standard / Epilepsy / Anti-tuberculosis

MICROBIOLOGY & VIRUS RESEARCHES

- Customized Targeted Sequencing
- Comprehensive Respiratory Virus
- Aftican Swine Fever Virus
- Customized 16S V4 NGS
- BTSeq™ – Standard
- BTSeq™ – Viral Analysis



SYNTHETIC BIOLOGY

- BTSeq™ Full Plasmid Sequencing
- BTSeq™ – Standard

DISTRIBUTORS & COLLABORATIONS

North America

[Collaborations]

South America

LK Global S.A.

Europe

INTRON · ISOGEN · LabLife · Refer

Asia Pacific

TreeCODE · funakoshi

South Korea(H.Q)

- Animal and Plant Quarantine Agency
- Korea Disease Control and Prevention Agency
- 국방과학연구소 Agency for Defense Development
- GC Genome
- Gangnam Severance Hospital
- Korea University Medical Center · KAIST

TARGETED SEQUENCING SOLUTION

CELEMICS PRODUCTS & SERVICES 2022

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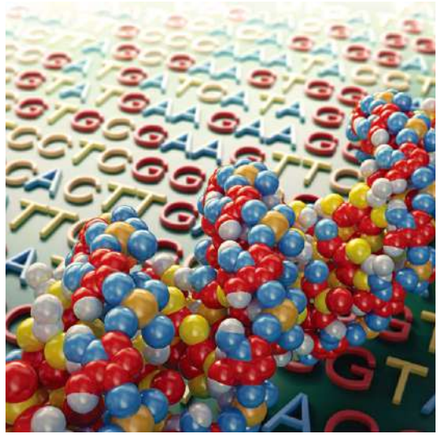
Targeted Sequencing Overview

Celemics has developed and delivered over 1,000 different customized panels. Our target enrichment method is capable of specifically isolating your genomic loci of interest out of the whole genome and increasing the sensitivity of detecting genetic mutations by producing higher coverage & in-depth sequencing data.

END-TO-END CUSTOMIZATION



PANEL DESIGN

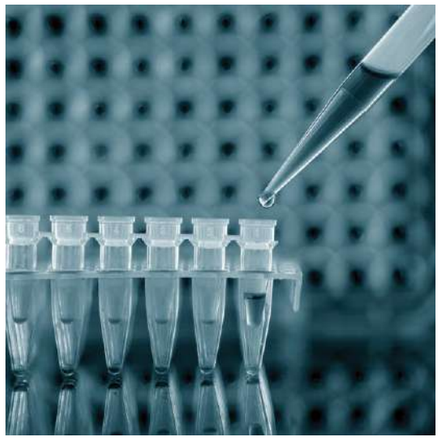
- Elaborately designed NGS panels comprised of your genes of interest
- Interactive discussion with customer prior to designing the panel (e.g., GC-rich, Homologous regions)
- Supported by advanced technology for probe design and reagent optimization
- Panel expansion possible through simple gene addition
- Alternative protocols in case required instruments are not available

SUPERIOR PERFORMANCE

- Market leading target enrichment kits
- Maximized cost-effectiveness
- Pre-capture pooling and high panel performance enables additional cost and labor savings

IN-HOUSE TEST & REBALANCING

- Adjustments to performance and functionality through thorough in-house validation test for every designed panel
- Detailed QC results encompassing wet-lab experiments, NGS run, and bioinformatics analysis provided to customer
- Rebalancing service possible through request
- Able to increase depth and coverage of a specific area if requested
- Finalize your order after reviewing QC results



DATA ANALYSIS

- Technical support available for customers new to NGS analysis
- Provides bioinformatics analysis services and tools from FASTQ to clinical report by request

OUTSTANDING PERFORMANCE OF TARGETED SEQUENCING

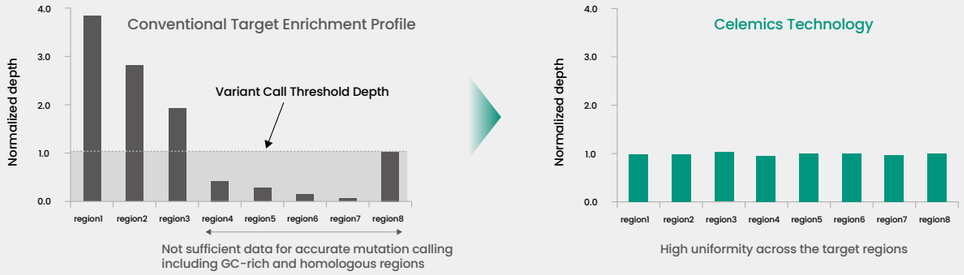
At Celemics, we support our customers through target hybridization-based NGS services and products individually designed and manufactured by experienced researchers and technicians. We have established a robust system for customized design panels and developed a variety of kits according to our customer's needs. All Ready-to-Use kits are completely validated and provide the best performance in the market. Our research team has designed and manufactured over a thousand customized panels and promises to offer the best quality product and service to our customers.

Key Features

1. Exceptional panel performance achieved by hybridization-based target capture method	Overcome limitations of amplicon-based NGS analysis with thoroughly validated hybridization-based target capture method High uniformity and coverage achieved by Celemics proprietary probe design technology
2. Assess all types of mutations with high sensitivity and specificity	Superior analytical performance compared to competitor products in detecting SNV, InDel, CNV, and rearrangement in a single NGS run with maximized sensitivity and specificity and minimized NGS noise enabled by Celemics unique molecular barcode assay and robust bioinformatics pipeline
3. Robust performance of assessing DNA and RNA across various specimen quality	Compatible with poor-quality and low-amount specimens such as FFPE, solid tumor, liquid biopsy, etc.
4. Efficient capture of 'Hard-to-Capture' regions	Analyze the clinically significant mutations embedded in GC rich or homologous regions, which are frequently masked by competitors
5. Wide compatibility with NGS instruments and automation platforms	Compatible with all NGS Instruments from Illumina, Thermo Fisher Scientific, Pacific Bioscience, MGI, and Oxford Nanopore Provides enzymes for DNA fragmentation as a substitute for sonicators
6. Flexible panel content: number of reactions of your choice and Gene Add-on Service	Save costs by ordering the number of reactions required for your experiment Expand your panel with minimum cost, time, and effort by simply adding or combining panels and genes of your interest

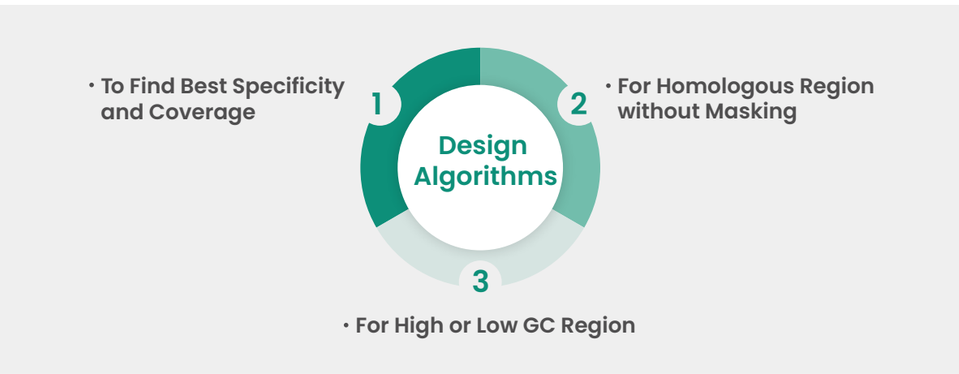
PROBE DESIGN TECHNOLOGY

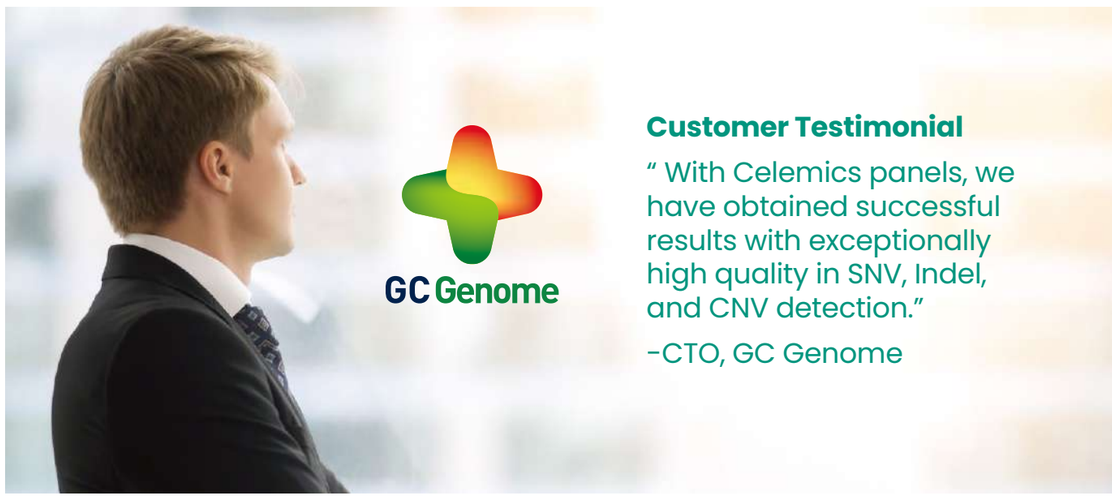
Market Problem and Celemics' Answer



Proprietary Probe Design Algorithm

Based on extensive wet-lab target capture experimentation for every customized panel



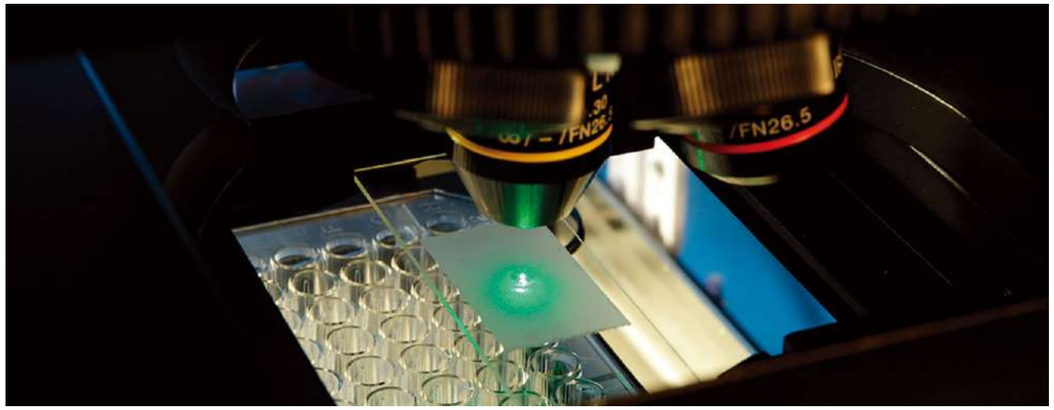


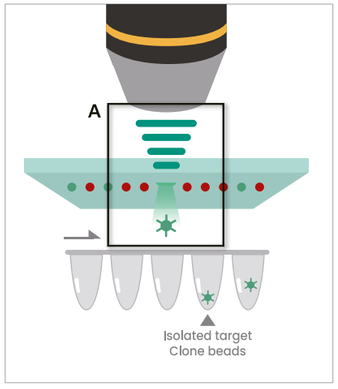
PROBE DESIGN TECHNOLOGY

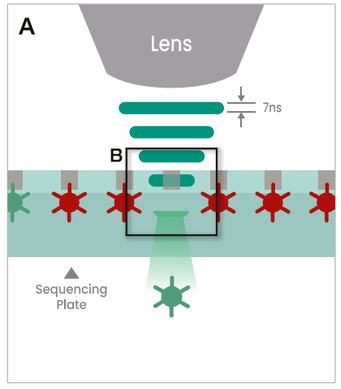
Proprietary Probe Manufacturing Technology

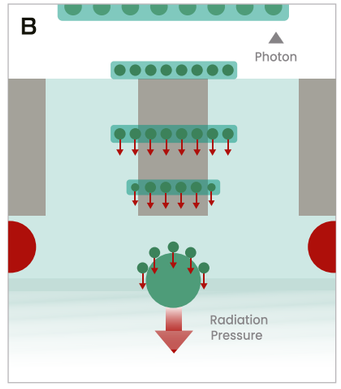
- Reduces complexity in handling complex oligo pools
- Enables extremely low-biased probe pool with handling individual probe sets
- Allows for cost-effectiveness and high-performance: advantage from pool-based probes and individually synthesized probes
- Achieves superior lot-to-lot uniformity for repeated orders due to proprietary 2-step probe synthesis technology

MSSIC Technology: Massively Separated and Sequence Identified Cloning

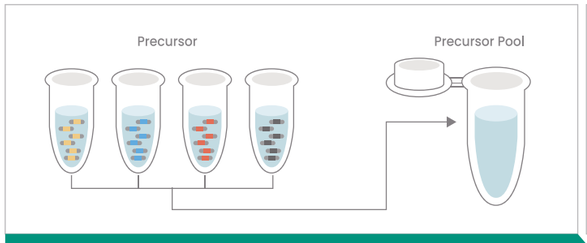




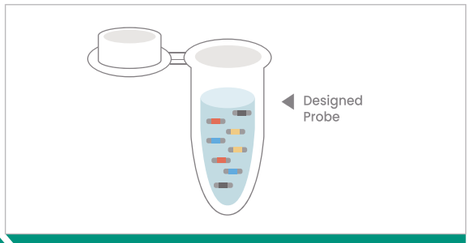




Two step probe manufacturing



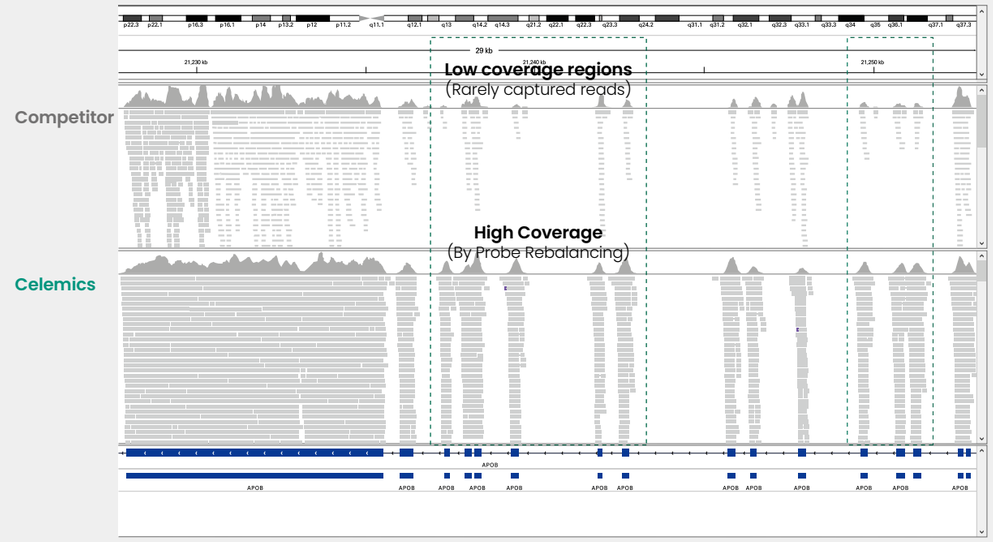
STEP 1. Manufacturing and Pooling of Precursor



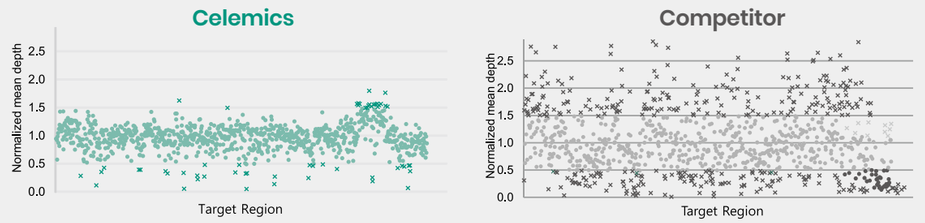
STEP 2. Optimized NGS Panel

TARGETED SEQUENCING PANEL PERFORMANCE

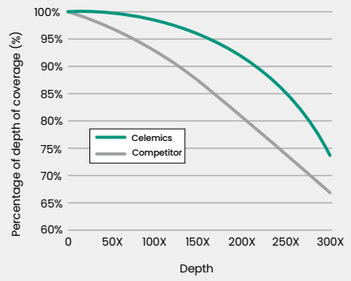
1. High Coverage Panel Compared to Competitor Products



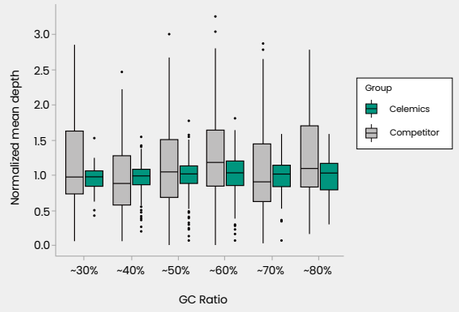
2. Higher Uniformity Across Target Regions



3. Superior Coverage Depth Over Target Regions



4. Superior Capture Performance Across GC Percentage

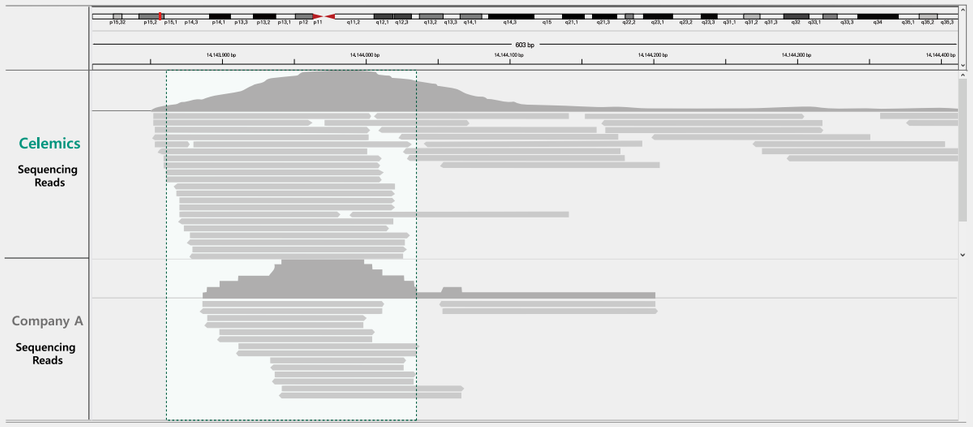


** Target region of both panels (BED file) are identical.
** Number of reads are the same for the results from both panels.

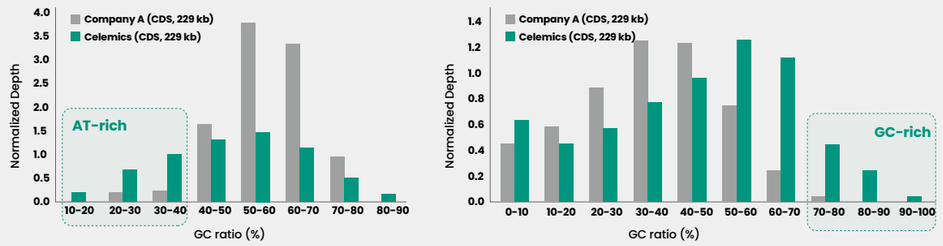
Targeted sequencing allows for sequencing with higher accuracy by specifically targeting the genomic regions of interest. The optimization process of the probes and reagents is essential for each of the different NGS platform types. Celemics has established the design technologies for the probes and reagents for various applications and achieved superior uniformity and depth of coverage compared to competitor products.

SEQUENCING PERFORMANCE OF CELEMICS PANEL FOR HARD-TO-CAPTURE REGIONS

1. Higher Depth compared to Company A Targeting Against the Same Target Area



2. Better Uniformity across AT- and GC-rich Regions

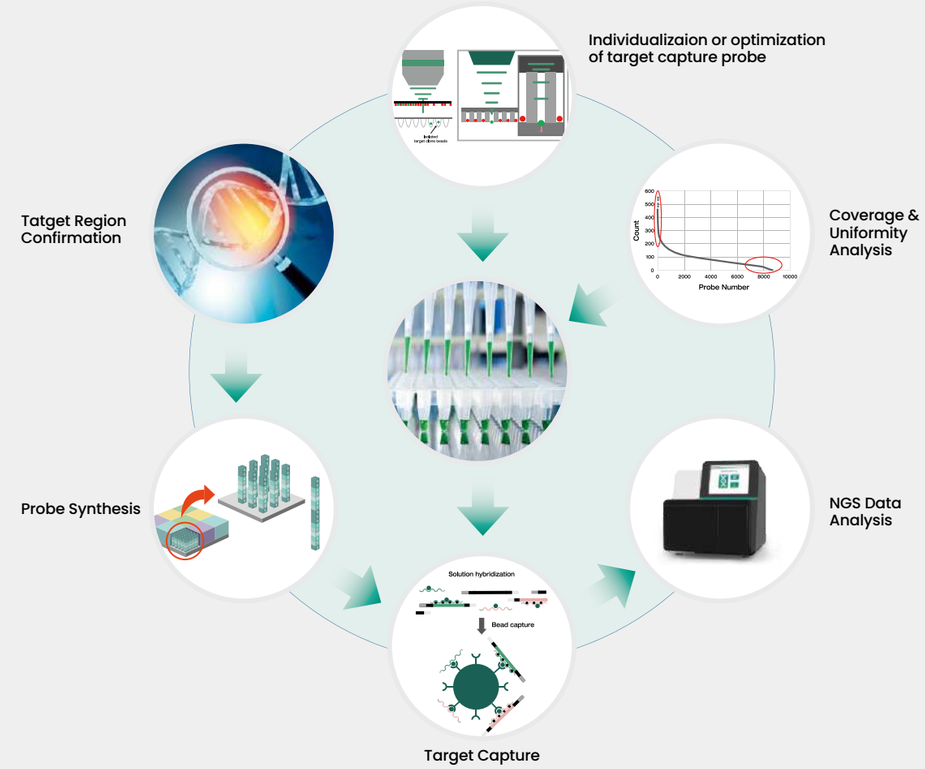


Even the most advanced NGS techniques have been challenged by GC-rich and homologous regions that are often masked or omitted by competitor services. Such a challenge is overcome by Celemics proprietary probe design technology which enables successful sequencing of GC-rich, AT-rich or homologous regions upon request. We also provide Homolog Report when the requested region includes homologous regions. Customers can then decide whether to include the regions in the order.

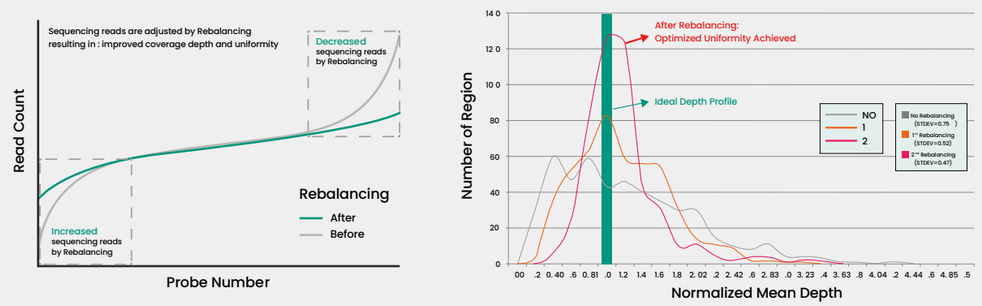
Our customers performed validation tests comparing Celemics' customized panels with our competitors'. For the competitor product, they performed validation tests based on competitor's recommended protocols for the same target regions. They also used the same sequencing amount for the fair experiment. As a result, customers selected our customized panels due to the high capture efficiency even with a lower amount of sequence data.

PILOT TEST & REBALANCING

Overview of Celemics Rebalancing Technology



Capture Uniformity Analysis



For customized targeted sequencing panels, we conduct in-house performance tests of requested panels and deliver the test results to customers. We also provide rebalancing services in case the customer requests for a specific area or overall performance improvement. The service includes redesigning probes against the requested regions and optimizing reagents to best meet our customers' needs.

EXAMPLE OF ctDNA ANALYSIS USING PROPRIETARY MOLECULAR BARCODES

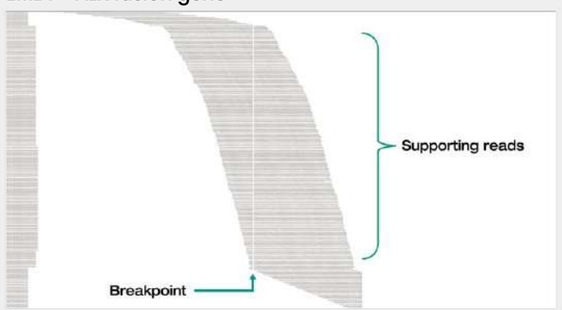
Performance Verification using Reference Material: 100% Sensitivity and Specificity

				0.5% VAF			1% VAF			WT		
	Gene	DNA change	AA change	VAF	VAF	VAF	VAF	VAF	VAF	VAF	VAF	VAF
Seracare	NRAS	c.182A>G	p.Q61R	0.96%	0.55%	0.78%	1.09%	0.98%	1.44%	0.06%	0.00%	0.00%
	PIK3CA	c.1633G>A	p.E545K	0.57%	0.69%	0.24%	1.18%	1.13%	0.38%	0.00%	0.00%	0.00%
	PIK3CA	c.3140A>G	p.H1047R	0.42%	0.33%	0.45%	0.81%	0.93%	0.94%	0.00%	0.00%	0.00%
	PIK3CA	c.3204_3205insA	p.N1068fs*4	0.51%	0.45%	0.51%	0.86%	0.95%	0.87%	0.00%	0.00%	0.00%
	EGFR	c.2310_2311insGGT	p.D770_N771insG	0.38%	0.36%	0.42%	0.48%	0.86%	0.78%	0.00%	0.00%	0.00%
	EGFR	c.2369C>T	p.T790M	0.44%	0.48%	0.48%	0.77%	1.23%	1.05%	0.00%	0.00%	0.00%
	EGFR	c.2573T>G	p.L858R	0.56%	0.51%	0.74%	1.58%	1.39%	0.85%	0.00%	0.00%	0.00%
	BRAF	c.1799T>A	p.V600E	0.51%	0.52%	0.47%	0.78%	0.70%	0.45%	0.00%	0.00%	0.00%
	PTEN	c.741_742insA	p.P248fs*5	0.31%	0.55%	0.51%	1.16%	1.30%	1.52%	0.00%	0.00%	0.00%
	KRAS	c.35G>A	p.G12D	0.43%	0.34%	0.62%	1.16%	0.89%	0.91%	0.00%	0.00%	0.00%
	ATK1	c.49G>A	p.E17K	0.69%	0.37%	0.35%	0.65%	0.66%	1.01%	0.00%	0.00%	0.00%
	TP53	c.818G>A	p.R273H	0.40%	0.47%	0.41%	1.84%	1.14%	0.86%	0.03%	0.05%	0.00%
	TP53	c.743G>A	p.R248Q	0.47%	0.44%	0.50%	0.90%	0.88%	0.85%	0.02%	0.07%	0.00%
	TP53	c.723delC	p.C242fs*5	0.43%	0.40%	0.41%	0.87%	0.85%	0.72%	0.00%	0.00%	0.00%
	TP53	c.524G>A	p.R175H	0.71%	0.66%	0.71%	1.19%	1.13%	1.02%	0.06%	0.05%	0.03%
	TP53	c.263delC	p.S90fs*33	0.50%	0.81%	0.53%	1.31%	1.55%	1.37%	0.09%	0.01%	0.06%
			Avg. (%)	0.52%	0.50%	0.51%	1.04%	1.04%	0.94%	0.02%	0.01%	0.01%

Accurate CNV and Gene Rearrangement Analysis with FFPE Samples Due to High Coverage Uniformity

	Panel A (400 kb)		Panel B (2.2 Mb)	
CNV	100 ng	50 ng	100 ng	50 ng
MET	Detected	Detected	Detected	Detected
MYCN	Not in 400 kb panel target region		Detected	Detected

EML4 - ALK fusion gene



We have conducted complete validation test for each Ready-to-Use panel and proved its superior performance compared to competitor products. The products are highly optimized for accurate and efficient assays even with poor quality and low-amount samples such as FFPE, ctDNA, etc. As shown in the table above, we have successfully performed CNV and rearrangement analysis from 50 ng of FFPE samples.

COST-EFFECTIVE SEQUENCING

Significantly reduced cost in Sample Prep, Target Enrichment Kit, and Sequencing

1. Sample Prep consumables developed and provided by Celemics for the highest optimization include CeleMag™ Clean-up Bead, CeleMag™ Streptavidin Bead, CLM Polymerase, and EP-kit (one-step workflow from Fragmentation to End-repair and A-tailing).
2. Pre-capture pooling reduces costs per sample.
3. Celemics has secured technology for proprietary probe design and manufacturing, significantly reducing costs of our Target Enrichment Kit.
4. Celemics panels have shown superior performance compared to competitor product in terms of uniformity and on-target ratio, enabling high-quality, cost-effective sequencing.

CELEMICS FEATURES & BENEFITS

1. Hybridization-based capture	2. Maximized Efficiency allows Market Leading Capture Performance	3. Hybridization Enhancer Technology and Enzymatic Library Preparation
4. User-friendly Bioinformatics Software	5. Reduced NGS costs by Pre-capture pooling with no compromise on quality	6. Molecular barcode and bioinformatics for ultra-low VAF mutations
7. CAS for bioinformatics analysis	8. Flexible panel content with Gene Add-on Service	9. Default wet-lab QC for every customized panel
10. Minimal lot variation due to proprietary 2-step probe manufacturing technology	11. Compatible with all NGS instruments and automation platforms	12. Capture the 'Hard-to-Capture' regions
13. Optimization of species-specific blockers for maximum performance for agriculture and animal research	14. Improved Probe Design by Rebalancing Service only available in Celemics	15. Robust, Rapid, Reliable Customization

READY-TO-USE PANELS FOR ONCOLOGY

CELEMICS PRODUCTS & SERVICES 2022

BRCA 1/2 Panel
OncoRisk Panel
CancerScreen Panel - Core / 50 / 100 / 400
CancerMaster Panel

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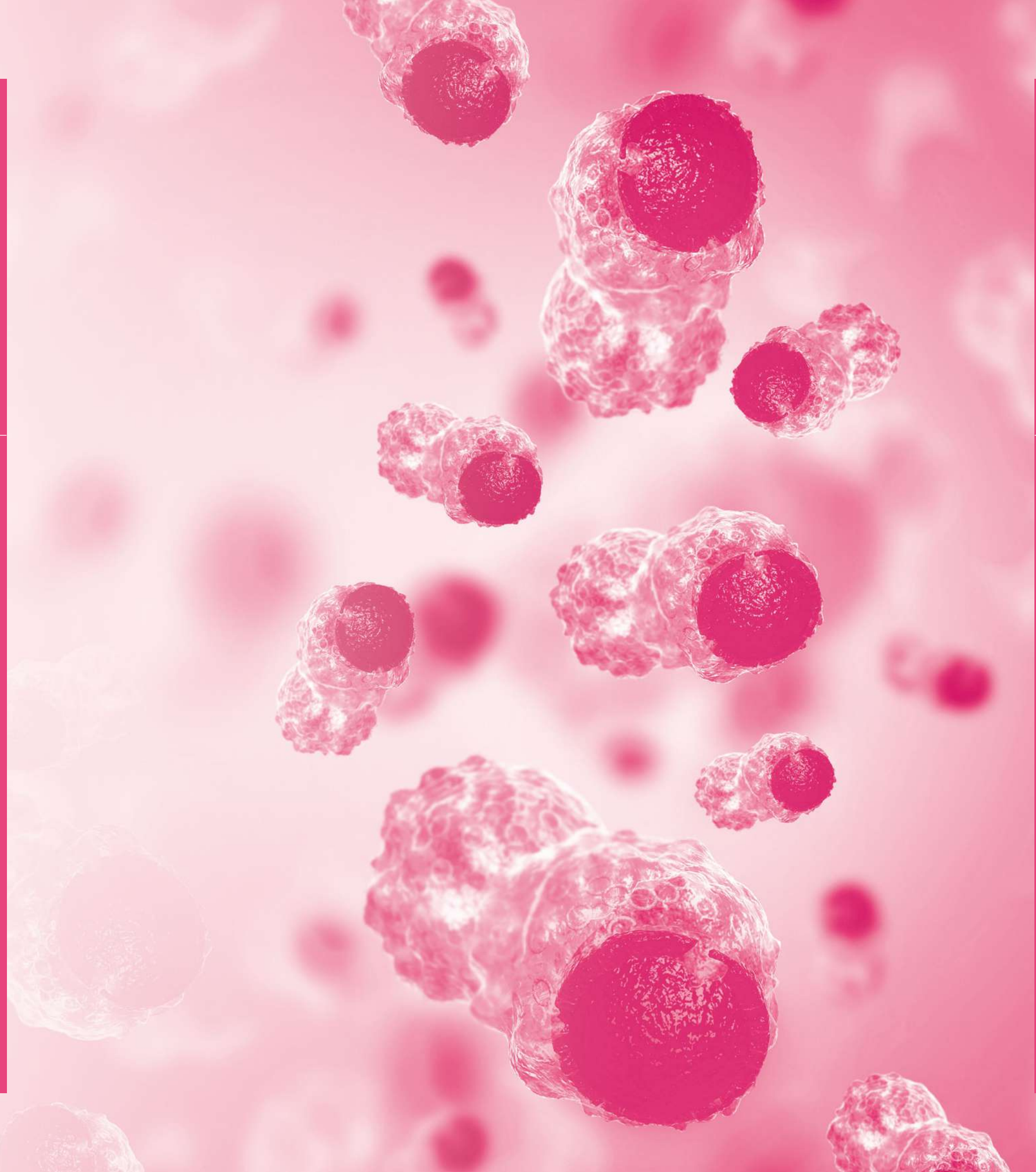


BRCA 1/2 Panel

Germline and Somatic Cancer

KEY FEATURES

1. Targets the whole CDS (+/- 40) and promoter regions of BRCA 1/2 with high specificity	Target regions not only covering the CDS regions but expanded to +40 and -40 of CDS to detect splicing site variants 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

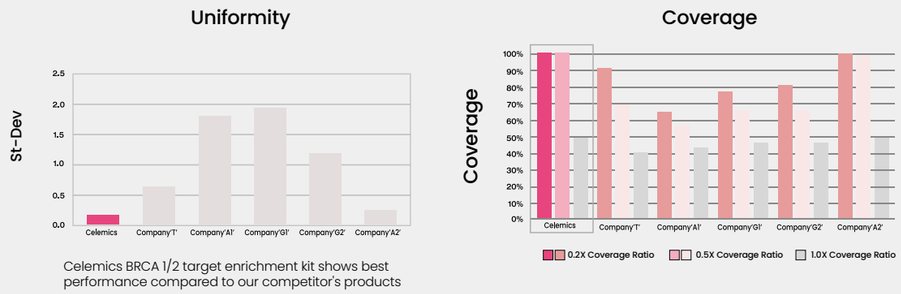
SPECIFICATION

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 by customer's request

PANEL PERFORMANCE

1. Superior Panel Performance Compared to Competitor Product

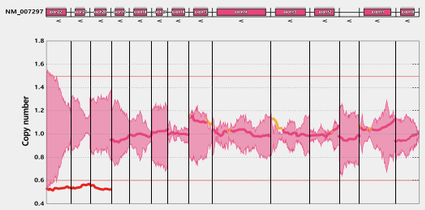


Celemics BRCA 1/2 target enrichment kit shows best performance compared to our competitor's products

2. SNV, CNV Analysis

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

Gene	Mutation Type	Amino Acid Change	Sequencing			Variant Allele Frequency
			Total	Ref.	Alt.	
BRCA1	Non-SYN	p.S1556G	634	315	301	48.71%
BRCA1	SYN	p.S1389S	876	501	370	42.48%
BRCA2	Non-SYN	p.N372H	396	213	181	45.94%
BRCA2	SYN	p.L1521L	289	0	281	99.29%



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

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 by customer's request

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								

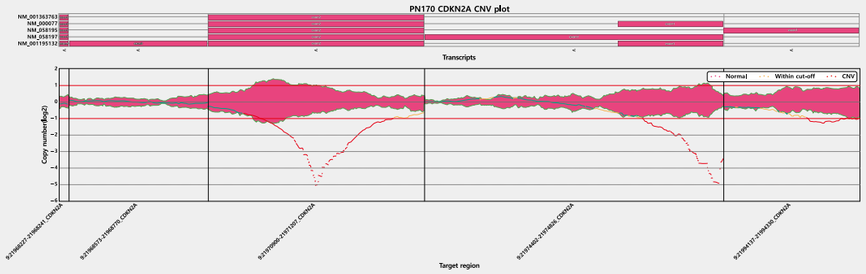
PANEL PERFORMANCE

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

CancerScreen Panel

Core/50/100/400

Somatic Cancer

KEY FEATURES

1. Optimized panel for solid cancer	Assess DNA, RNA, and the whole CDS regions (RefSeq) of up to 407 genes and rearrangement regions associated with solid cancer
2. High sensitivity and specificity	Detect low-frequency and rare variants with high sequencing depths Capture the GC rich and homologous regions with Celemics proprietary design technology
3. Cost-effective sequencing	Lower sequencing costs for 3 Gb sequencing amount compared to competitor product
4. Assess all variant types	Detect all mutation types including SNV, Indel, Large Indel, CNV, Rearrangement, MSI, and TMB in a single assay

SPECIFICATION

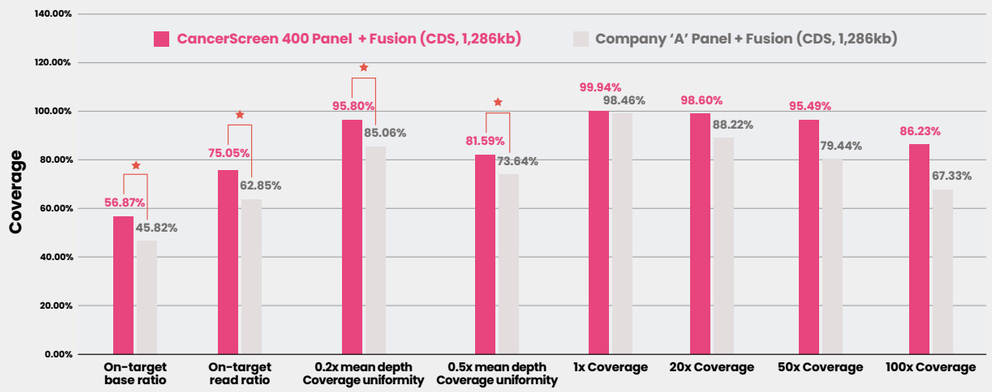
Gene count*	13 / 54 / 99 / 407 genes
Target size	61 / 197 / 299 / 1,123 kb + Rearrangement
Mutation type	SNV, Indel, CNV, Rearrangement, MSI, TMB
Sample type	FFPE, frozen tissue, cfDNA, RNA
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

* Gene Add-On Service: Genes can be added by customer's request

PANEL PERFORMANCE

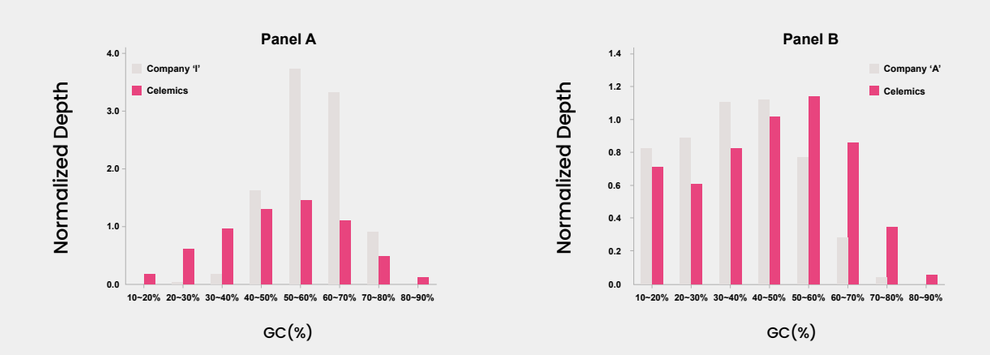
Performance Comparison with Competitor Product

Higher on-target ratio, uniformity, and coverage at 100X compared to competitor product over the target regions including exons and introns (Compared with the same sequencing depth)



Performance Comparison over GC-rich Regions

Higher uniform read depths over GC-rich regions compared to competitor product (Compared with the same sequencing depth)



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

CancerScreen Panel
Core

DESCRIPTION

The CancerScreen Core Panel is an NGS assay designed to detect all types of variants in 13 genes associated with somatic cancer. Targeting the selected genes with high sensitivity and specificity enables saving cost and effort. The report consists of the primary, secondary, and tertiary results for the In-depth understanding and interpretation of sequencing data.

GENE LIST

CancerScreen Core	**ALK**	APC	BRAF	EGFR	ERBB2	KRAS	MET	NRAS	PIK3CA	**RET**	**ROS1**	SMAD4	TP53

* Genes in bold indicate fusion analysis

CancerScreen Panel
50

DESCRIPTION

The CancerScreen 50 Panel is an expanded NGS assay designed to detect all types of variants in over 50 genes associated with somatic cancer. Targeting the selected genes with high sensitivity and specificity enables saving cost and effort. The report consists of the primary, secondary, and tertiary results for the In-depth understanding and interpretation of sequencing data.

GENE LIST

CancerScreen 50	ABL1	AKT1	**ALK**	APC	ATM	BRAF	BRCA1	BRCA2	CDH1	CDK4	CDK6	CDKN2A	CSF1R
	CTNNB1	DDR2	EGFR	ERBB2	ERBB4	ESR1	FGFR1	FGFR2	FGFR3	GNA11	GNAQ	GNAS	HRAS
	IDH1	IDH2	JAK2	KDR	KIT	KRAS	MAP2K1	MET	MLH1	MTOR	MYC	MYCN	NOTCH1
	NRAS	**NTRK1**	PDGFRA	PIK3CA	PTCH1	PTEN	PTPN11	RB1	**RET**	**ROS1**	SMAD4	SMO	SRC
	STK11	TP53											

* Genes in bold indicate fusion analysis

CancerScreen Panel
100

DESCRIPTION

The CancerScreen 100 Panel is an NGS assay for the comprehensive analysis of around 100 genes associated with somatic cancer. All types of variants are detected with high sensitivity and specificity. The report consists of the primary, secondary, and tertiary results for the In-depth understanding and interpretation of sequencing data.

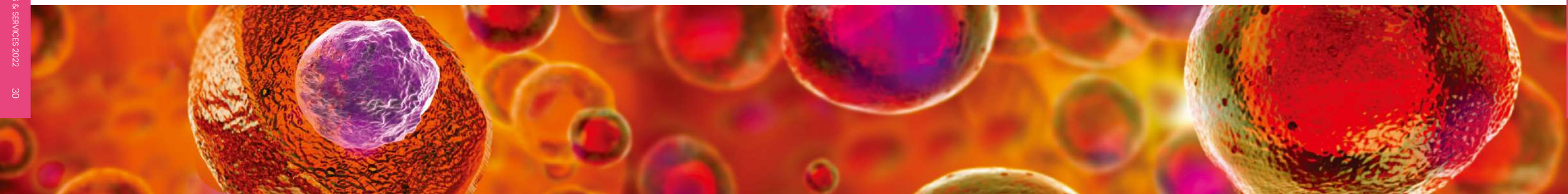
GENE LIST

CancerScreen 100	ABL1	AKT1	AKT2	AKT3	ALK	APC	ARID1A	ARID1B	ARID2	ATM	ATRX	AURKA	AURKB
	BARD1	BCL2	BLM	BMPR1A	BRAF	BRCA1	BRCA2	BRIP1	CDH1	CDK4	CDK6	CDKN2A	CHEK2
	CSF1R	CTNNB1	DDR2	EGFR	EPCAM	EPHB4	ERBB2	ERBB3	ERBB4	EZH2	FBXW7	FGFR1	FGFR2
	FGFR3	FLT3	GNA11	GNAQ	GNAS	HNF1A	HRAS	IDH1	IDH2	IGF1R	ITK	JAK1	JAK2
	JAK3	KDR	KIT	KRAS	MDM2	MET	MLH1	MPL	MRE11	MSH2	MSH6	MTOR	MUTYH
	NBN	NF1	NOTCH1	NPM1	NRAS	NTRK1	PALB2	PDGFRA	PDGFRB	PIK3CA	PIK3R1	PMS2	PRSS1
	PTCH1	PTCH2	PTEN	PTPN11	RAD50	RAD51C	RAD51D	RB1	RET	ROS1	SLX4	SMAD4	SMARCB1
	SMO	SRC	STK11	SYK	TERT	TOP1	TP53	VHL					

CancerScreen Panel
400

DESCRIPTION

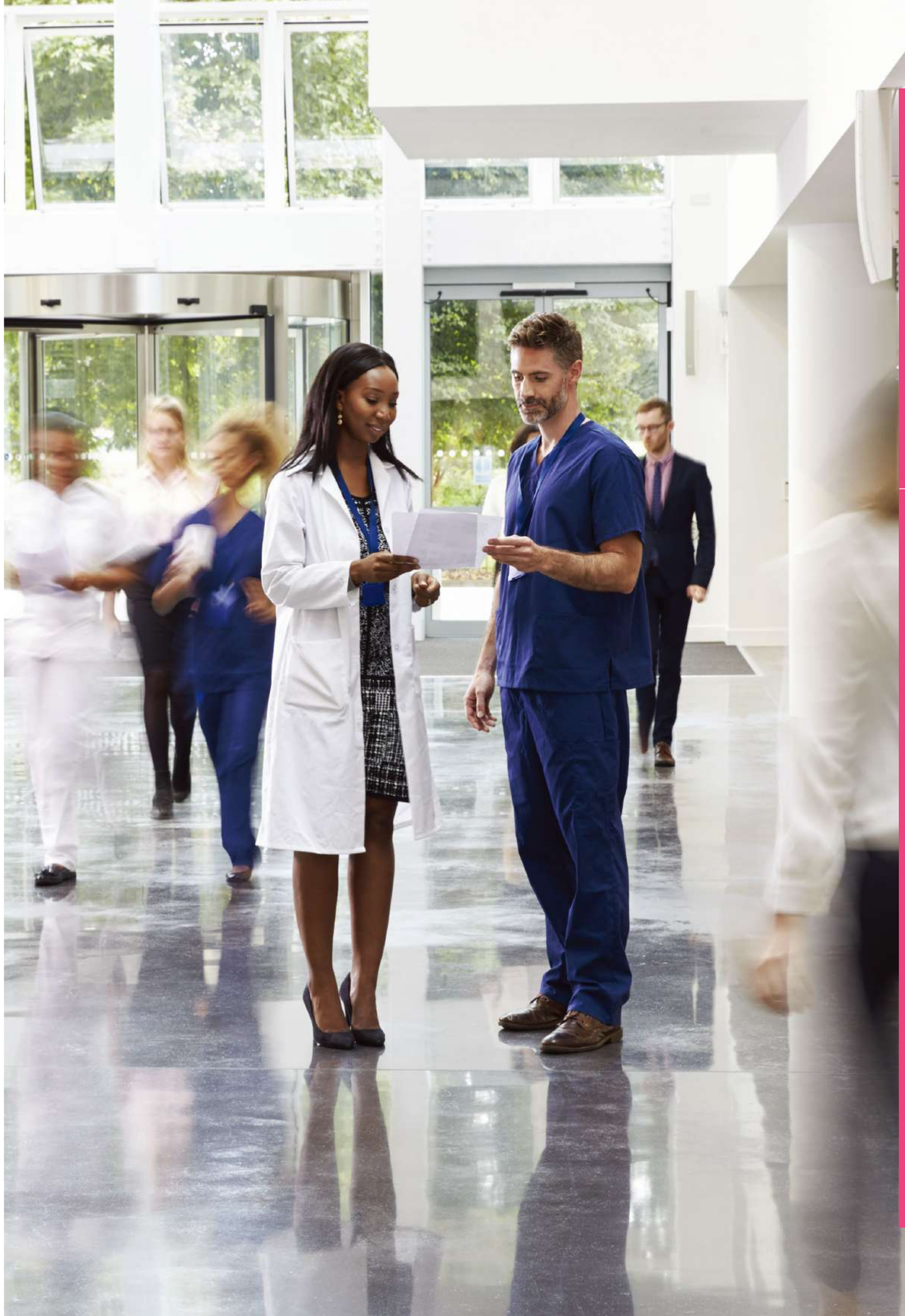
The CancerScreen 400 Panel is an NGS assay designed to detect all types of variants in over 400 genes associated with somatic cancer. Targeting the selected genes with high sensitivity and specificity enables saving cost and effort. The report consists of the primary, secondary, and tertiary results for the In-depth understanding and interpretation of sequencing data.



CancerScreen Panel 400

GENE LIST

CancerScreen 400	ABL1	ABL2	ADGRA2	AKT1	AKT2	AKT3	ALK	AMER1	APC	APCDD1	APEX1	APOB	APOBEC1
	AR	ARAF	ARFRP1	ARID1A	ARID1B	ARID2	ASXL1	ATM	ATP11B	ATR	ATRX	AURKA	AURKB
	AXIN1	AXL	B2M	B3GAT1	BACH1	BAP1	BARD1	BCL2	BCL6	BCL9	BCOR	BCR	BIRC2
	BIRC3	BLM	BRAF	BRCA1	BRCA2	BRD2	BRD3	BRD4	BRIP1	BTG1	BTK	BTLA	CARD11
	CASP5	CASP8	CBFB	CBL	CDK12	CDK4	CDK6	CDK8	CDKN1A	CDKN1B	CDKN2A	CDKN2B	CDKN2C
	CDX2	CEBPA	CHD1	CHD2	CHD4	CHEK1	CHEK2	CHUK	CIC	CRBN	CREBBP	CRKL	CRLF2
	CSF1R	CSF2	CSF2RA	CSF2RB	CSNK2A1	CTCF	CTLA4	CTNNA1	CTNNB1	CUL3	CUL4A	CUL4B	CXCL10
	CXCL11	CXCL9	CXCR3	CYLD	CYP17A1	DAXX	DCUN1D1	DDR2	DICER1	DIS3	DNMT1	DNMT3A	DOCK2
	DOT1L	EGFR	ELMO1	EML4	EMSY	EP300	EPHA3	EPHA5	EPHA6	EPHA7	EPHB1	EPHB4	EPHB6
	ERBB2	ERBB3	ERBB4	ERCC1	ERCC2	ERG	ERRFI1	ESR1	ETV1	ETV4	ETV5	ETV6	EWSR1
	EYA2	EZH2	FANCA	FANCC	FANCD2	FANCE	FANCF	FANCG	FANCI	FANCL	FANCM	FAS	FAT1
	FAT3	FBXW7	FGF1	FGF10	FGF12	FGF14	FGF19	FGF2	FGF23	FGF3	FGF4	FGF6	FGF7
	FGFR1	FGFR2	FGFR3	FGFR4	FH	FLCN	FLT1	FLT3	FLT4	FOXA1	FOXL2	FOXO3	FOXP3
	FRS2	FUBP1	GABRA6	GAS6	GATA1	GATA2	GATA3	GATA4	GATA6	GID4	GLI1	GNA11	GNA13
	GNAQ	GNAS	GRIN2A	GRM3	GSK3B	GUCY1A2	GZMA	GZMB	GZMH	H3F3A	HGF	HIST1H3B	HNF1A
	HOXA3	HRAS	HSD3B1	HSP90AA1	IDH1	IDH2	IDO1	IDO2	IFITM1	IFITM3	IFNA1	IFNB1	IFNG
	IGF1	IGF1R	IGF2	IGF2R	IKBKE	IKZF1	IL12A	IL12B	IL2	IL23A	IL6	IL7R	INHBA
	INPP4B	INSR	IRF2	IRF4	IRS2	ITGAE	ITK	JAK1	JAK2	JAK3	JUN	KAT6A	KDM5A
	KDM5C	KDM6A	KDR	KEAP1	KEL	KIT	KLF4	KLHL6	KMT2A	KMT2B	KMT2C	KNSTRN	KRAS
	LAG3	LMO1	LRP1B	LRP6	LTK	LYN	LZTR1	MAGI2	MAGOH	MAML1	MAP2K1	MAP2K2	MAP2K4
	MAP3K1	MAP3K13	MAPK1	MAX	MCL1	MDM2	MDM4	MED12	MEF2B	MEN1	MET	MITF	MLH1
	MPL	MRE11	MSH2	MSH6	MTOR	MUTYH	MYB	MYC	MYCL	MYCN	MYD88	MYO18A	NCOA3
	NCOR1	NF1	NF2	NFE2L2	NFKBIA	NOTCH1	NOTCH2	NOTCH3	NOTCH4	NPM1	NRAS	NSD1	NSD3
	NTRK1	NTRK2	NTRK3	NUP93	NUTM1	PAK3	PAK5	PALB2	PARP1	PARP2	PARP3	PARP4	PAX5
	PBRM1	PDCD1	PDCD1LG2	PDGFRA	PDGFRB	PDK1	PGR	PHF6	PHLPP2	PIK3C2B	PIK3C3	PIK3CA	PIK3CB
	PIK3CG	PIK3R2	PKHD1	PLCG1	PLCG2	PMS2	PNP	PNRC1	POLD1	POLE	PPARG	PPP2R1A	PRDM1
	PREX2	PRF1	PRKAR1A	PRKC1	PRKDC	PRPF40B	PRSS8	PTCH1	PTCH2	PTEN	PTK2	PTPN11	PTPRC
	PTPRD	QKI	RAB35	RAC1	RAC2	RAD17	RAD50	RAD51	RAD52	RAD54L	RAF1	RANBP2	RARA
	RB1	RBM10	REL	RET	RHEB	RHOA	RHOB	RICTOR	ROBO1	ROBO2	ROS1	RPA1	RPS6KB1
	RPTOR	RUNX1	RUNX1T1	RUNX3	SDHA	SDHB	SDHC	SDHD	SEMA3A	SEMA3E	SET	SETBP1	SETD2
	SF3A1	SF3B1	SH2B3	SKP2	SLIT2	SMAD2	SMAD3	SMAD4	SRSF2	SRSF7	STAG2	STAT3	STAT4
	TERT	TET2	CD274	TP53									



CancerMaster Panel

Somatic Cancer

DESCRIPTION

The CancerMaster Panel is designed to detect all variant types and immuno-oncology markers (MSI and TMB), which are crucial biomarkers for cancer immunotherapy. For CNV analysis, different cut-offs are applied according to the ratio of cancer cells. The panel is also designed to detect Epstein-Barr virus (EBV) and Human Papillomaviruses (HPV), allowing for the comprehensive analysis of cancer-associated genes.

KEY FEATURES

1. Comprehensive analysis of cancer-associated genes	A broad range of targeting elements including somatic variants, IO-signatures (TMB, MSI), EBV and HPV, for clinical diagnoses of different cancer types and applications to precision medicine
2. Extensive validation studies	Robust panel performance supported by extensive validation tests with reference and clinical specimens

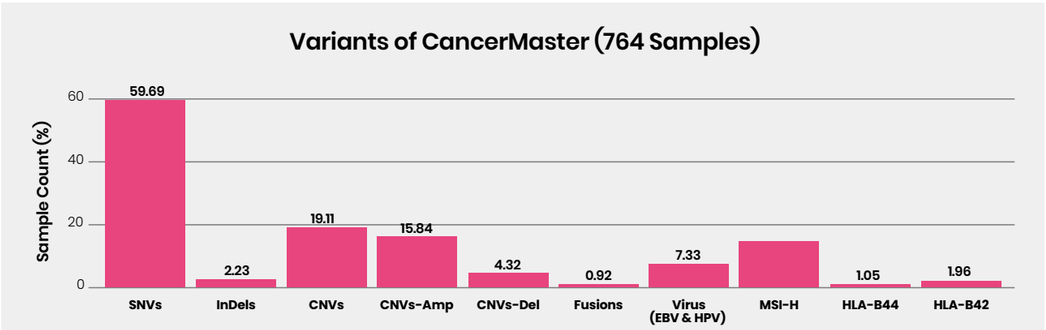
SPECIFICATION

Gene count*	524 genes
Covered region	Whole CDS, custom regions of oncogenes, immune response genes, and EBV & HPV viruses
Target size	2.5 Mb
Mutation type	SNV, Indel, CNV, Rearrangment, TMB, MSI, EBV, HPV
Sample type	FFPE, Fresh frozen tissue (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)
Publication	Molecular Characterization of Biliary Tract Cancer Predicts Chemotherapy and PD-1/PD-L1 Blockade Responses, Hepatology, 2021

* Gene Add-On Service: Genes can be added by customer's request

PANEL PERFORMANCE

The probes are designed to include the intron regions as well as clinically significant biomarkers. By conducting extensive validation studies with clinical samples, the panel was examined to show its performance with high sensitivity and specificity in detecting the variants in cancer-associated genes.

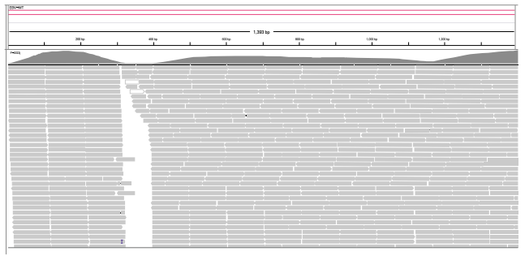


ANALYSIS OF EBV & HPV

EBV (Epstein-Barr Virus)

- Related disease – Lymphoma
- Genes – EBV type 1 (EBNA-2)

Validation for detection of EBV type 1 (EBNA-2) in control specimens



HPV (Human Papillomavirus)

- Related disease – Cervical cancer
- Genes – HPV L1 gene
  (Analysis of a total of 24 types is possible)

Analysis of the following 11 types of HPV types was completed using clinical specimens

Human infection HPV list
Human papillomavirus type 178
Human papillomavirus type 136
Human papillomavirus type 140
Human papillomavirus type 154
Human papillomavirus type 156
Human papillomavirus type 179
Human papillomavirus type 201
Human papillomavirus type 49
Human papillomavirus type 9
Human papillomavirus type 92
Human papillomavirus type 96

PACKAGE COMPOSITION

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

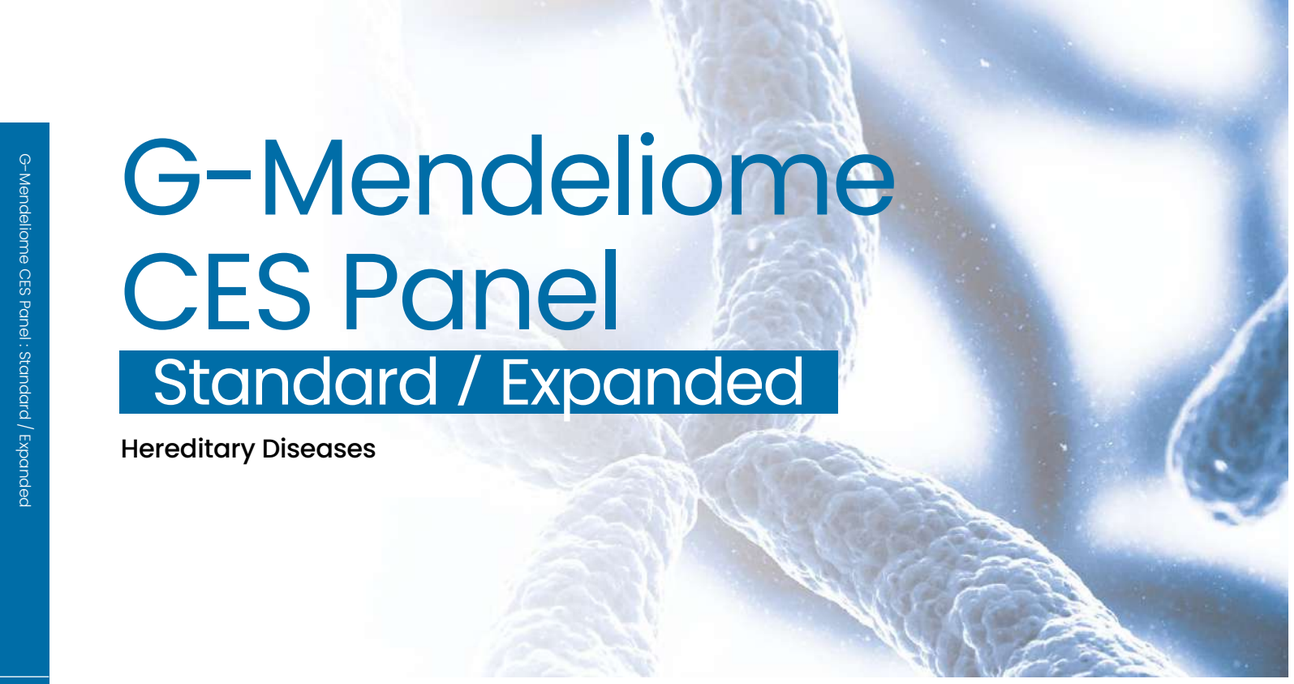
Package option	Options	
Pooling method	Single Reaction	Pre-capture Pooling
Library Preparation kits	Standard Kit	EP-kit
Hybridization Enhancer	Included	Not included

READY-TO-USE PANELS FOR INHERITED DISEASE

CELEMICS PRODUCTS & SERVICES 2022

G-Mendeliome CES Panel
: Standard / Expanded
G-Mendeliome Disease-Specific Panel

CELEMICS



G-Mendeliome CES Panel

Standard / Expanded

Hereditary Diseases

DESCRIPTION

The G-Mendeliome CES Panel has overcome the limitations of analyzing clinical diseases with whole exome sequencing. By selectively targeting the clinically significant genes, the panel enables comprehensive analysis with the most effective sequencing throughput.

KEY FEATURES

1. Comprehensive genomic profiling of a variety of genetic diseases	Includes 7,000 genes associated with clinically significant genetic diseases
2. A wide range of target regions	Includes all clinically significant regions that are not covered from competitor panels
3. Cost-effective analysis	Able to provide accurate analysis with reduced sequencing costs compared to WES

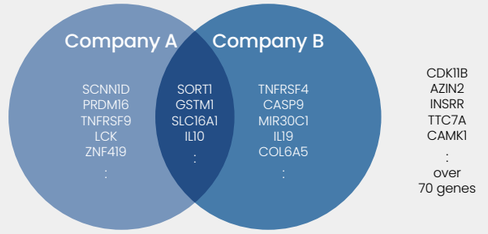
SPECIFICATION

Gene count*	5,516 / 7,563 genes
Covered region	CDS, hotspots, Mitochondrial genome
Target size	13.8 / 19.6 Mb
Mutation type	SNV, Indel, CNV
Sample type	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

PANEL PERFORMANCE

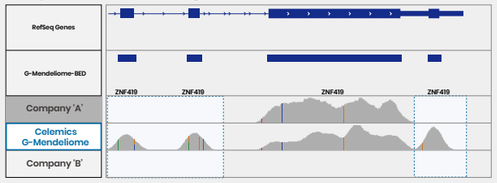
	Celemics	Company A	Company B
On-Target Read Ratio	82.8%	65.9%	80.8%

G-Mendeliome CES: Company A + Company B + over 70 genes

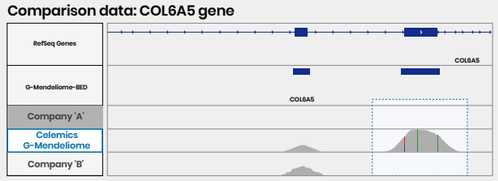


The CES panel was developed from the needs of GC Genome, the largest clinical NGS service provider in Korea, to solve the problem of poor diagnosis and high costs

Comparison data : ZNF419 gene



Comparison data: COL6A5 gene



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

LIST OF DISEASES ASSESSED BY G-MENDELIOME CES PANEL

Category	Related Diseases
Cardiology	Aortopathy and connective tissue disorders
	Arrhythmia
	Cardiomyopathy
	Congenital heart defect
	Dyslipidemia
	Other cardiovascular diseases
	Pulmonary hypertension
Dermatology	Adams-Oliver syndrome
	Albinism
	Cardiofaciocutaneous syndrome
	Cutis laxa
	Dyskeratosis congenita
	Ectodermal dysplasia
	Ehlers-Danlos syndrome
	Epidermolysis bullosa
	Hereditary acrodermatitis enteropathica
	Hermansky-Pudlak syndrome
	Hypotrichosis
	Ichthyosis
	Neurofibromatosis
	Pachyonychia congenita
	Palmoplantar keratoderma
	Progeria and Progeroid Syndromes
	Skin cancer
	Tuberous sclerosis
	Waardenburg syndrome
	Xeroderma pigmentosum
Endocrinology	Adrenal hyperplasia
	Diabetes
	Hyperinsulinism
	Hyperparathyroidism
	Hypothyroidism
	Kallmann syndrome
	Multiple endocrine neoplasia
	Obesity
	Pancreatitis
	Premature ovarian failure
ENT	Hearing loss
GI/Hepatology	Cholestasis
	Congenital diarrhea
	Congenital hepatic fibrosis
	Gastrointestinal atresia
	Hirschsprung disease
	Polycystic liver disease
Hematology	Anemia
	Bleeding&Thrombotic disorder
	Bone marrow failure
	Congenital neutropenia
	Hemochromatosis
	RBC membrane disorder
Immunology	Antibody deficiencies
	Autoinflammatory disorders
	Combined T/B cell deficiencies
	Complement deficiencies
	Defects in intrinsic and innate immunity
	Immune dysregulation
	Phagocytic defects

Category	Related Diseases
Metabolism	Aminoacidopathies
	Carbohydrate disorders
	Congenital disorders of glycosylation
	Creatine biosynthesis disorders
	Fatty acid oxidation defects
	Lipodystrophy
	Lysosomal storage disorders
	Organic acidemias
	Peroxisomal disorders
	Porphyria
	Purine/Pyrimidine metabolism disorders
	Pyruvate metabolism and tricarboxylic acid cycle defects
	Urea cycle disorders
Nephrology	Bartter syndrome
	Ciliopathies
	Diabetes insipidus
	Hemolytic uremic syndrome
	Hypokalemia
	Hypomagnesemia
	Hypophosphatemic rickets
	Nephrolithiasis
	Nephrotic syndrome/Focal glomerulonephrosis
	Pseudohypoaldosteronism
	Renal malformation
	Renal tubular acidosis
Neurology	Autism
	Movement disorders
	Neurodegenerative disorders
	Neuromuscular disorders
	Neuropathies and related disorders
	Seizures and Brain abnormalities
Oncology	Breast and gynecological cancer
	Colorectal cancer
	Endocrine cancer
	Gastrointestinal cancer
	Hematologic malignancy
	Lung cancer
	Nervous system/brain cancer
	Pancreatic cancer
	Prostate cancer
	Renal cancer
	Sarcoma
	Skin cancer
Ophthalmology	Albinism
	Cataract/Ectopia lentis
	Corneal dystrophy
	Glaucoma
	Microphthalmia/Anophthalmia
	Nystagmus
	Ophthalmoplegia/Oculomotor apraxia
	Optic atrophy
	Retinal dystrophy
	Retinoblastoma
Pulmonology	Bronchiectasis
	Central hypoventilation/Apnea
	Cystic fibrosis
	Cystic lung disease
	Hermansky-Pudlak syndrome
	Interstitial lung disease
	Primary ciliary dyskinesia
	Surfactant dysfunction
Skeletal disorders	Amelogenesis imperfecta
	Arthrogryposes
	Cleft lip palate
	Craniosynostosis
	Exostosis
	Facial dysostosis
	Macrocephaly/Overgrowth syndrome
	Osteopetrosis
	Short stature syndrome
	Skeletal dysplasia

G-Mendeliome Disease-Specific Panel

KEY FEATURES

1. Comprehensive analysis of a broad range of diseases	Identifying diseases associated with: Acute lymphatic leukemia, Acute Myeloid Leukemia, Cardiac disease, Coagulation, Epilepsy, Hearing loss, Inborn errors of metabolism, Lymphoma, Lysosomal storage disease, Common hereditary cancer for a medical checkup, Neuromuscular disease, Parkinson's disease, Alzheimer's disease, Dementia, Dystonia, RASopathies, Retinitis pigmentosa, Short stature, Skin disease, and Somatic cancer
2. Collaboration with the leading CRO in the country	Developed 17 different panels for assessing genes of related diseases

SPECIFICATION

Gene count*	Ranges from 61 to 321 genes
Covered regions	Whole CDS, hotspots
Target size	109-1,173 kb
Mutation type	SNV, Indel, CNV
Sample type	Differs by somatic or germline panel
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

* Gene Add-On Service: Genes can be added by customer's request

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

LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Gene List											
Alzheimer-Parkinson-Dementia Panel (*101 genes, 244.8 Kb*) **Related Diseases:** Alzheimer's disease, Parkinson's disease, Dystonia	AARS	ABCA13	ABCA7	ABCB11	ADCY5	ALS2	ANG	ANO3	APP	ATP13A2	ATP1A3	ATP7B
	C19orf12	CACNA1B	CHCHD10	CHMP2B	CHRNA4	CIZ1	COG1	COL4A4	COL6A3	DAO	DCTN1	DNMT1
	EVC	FERMT1	FIG4	FREM2	FUS	GBA	GCH1	GNAL	GNAO1	GRM1	GRN	HNRNPA1
	HNRNPA2B1	HPCA	HPSE2	IL12RB2	KCTD17	KMT2B	L2HGDH	LAMA3	LRRK2	MAPT	MATR3	MECR
	NDUFV3	NEK1	NPHS2	OPTN	PANK2	PARK7	PDP1	PINK1	PLA2G6	PNKD	PRKN	PRKRA
	PRNP	PRRT2	PSEN1	PSEN2	RELN	SERPIND1	SETX	SGCE	SIGMAR1	SLC12A6	SLC19A3	SLC2A1
	SLC30A10	SLC6A3	SNCA	SOD1	SORL1	SOX6	SPG11	SQSTM1	SRY	SUMF1	TAF1	TAF15
	TARDBP	TBK1	TDRD7	TH	THAP1	TIMM8A	TOR1A	TREM2	TUBA4A	TUBB4A	UBQLN2	VAC14
	VAPB	VCAN	VCP	VPS13A	WNK1							
Bleeding Disorder-Coagulopathy Panel (*147 genes, 339.2 Kb*) **Related Diseases:** Bleeding Disorder, Coagulation	AARS	ABCA1	ABCA13	ABCB11	ACTN1	ANKRD26	ANO6	AP3B1	ARHGAP35	BLOC1S3	BLOC1S6	BRCA1
	BRCA2	BRIP1	CD36	CDAN1	COG1	COL4A4	CPNE1	CYCS	DDX41	DKC1	DNMT1	DTNBP1
	ELANE	ERCC4	ETV6	EVC	F10	F11	F13A1	F13B	F2	F5	F7	F8
	F9	FANCA	FANCB	FANCC	FANCD2	FANCE	FANCF	FANCG	FANCI	FANCL	FANCM	FERMT1
	FERMT2	FGA	FGB	FGG	FLI1	FREM2	FYB1	GATA1	GATA2	GFI1	GFI1B	GP1BA
	GP1BB	GP6	GP9	GRM1	HAX1	HOXA11	HPS1	HPS3	HPS4	HPS5	HPS6	HPSE2
	IFNG	IL12RB2	ITGA2B	ITGB3	KIAA1244	L2HGDH	LAMA3	LMAN1	LYST	MASTL	MCFD2	MLPH
	MPL	MYH9	MYO5A	NBEAL2	NBN	NDUFV3	NHP2	NOP10	NPHS2	P2RY12	PALB2	PCNXL2
	PDP1	PLA2G4A	PLAU	PRF1	PRKACG	PRUNE2	RAB27A	RAD51C	RASGRP2	RBM8A	RIPK3	RPL11
	RPL35A	RPL5	RPS10	RPS19	RPS24	RPS26	RPS7	RUNX1	SBDS	SEC23B	SERPIND1	SERPINE1
	SERPINF2	SLC12A6	SLFN14	SLX4	SOX6	SP4	SRC	SRP72	SRY	STIM1	SUMF1	TBXA2R
	TBXAS1	TDRD7	TERC	TERT	TINF2	UBE2T	USP6NL	VCAN	VIPAS39	VPS33B	VWF	WAS
	WIPF1	WNK1	XRCC2									
Cardiovascular Panel (*207 genes, 694.8 Kb*) **Related Diseases:** Cardiac diseases	AARS	ABCA13	ABCB11	ABCC9	ABCG5	ABCG8	ACTA1	ACTA2	ACTC1	ACTN2	AKAP9	ALMS1
	ANK2	ANKRD1	APOA4	APOA5	APOB	APOC2	APOE	ARHGAP35	BAG3	BRAF	CACNA1C	CACNA2D1
	CACNB2	CALM1	CALR3	CASQ2	CAV3	CBL	CBS	CETP	COG1	COL3A1	COL4A4	COL5A1
	COL5A2	COX15	CPNE1	CREB3L3	CRELD1	CRYAB	CSRP3	CTF1	DES	DMD	DNAJC19	DNMT1
	DOLK	DPP6	DSC2	DSG2	DSP	DTNA	EFEMP2	ELN	EMD	EVC	EYA4	FBN1
	FBN2	FERMT1	FHL1	FHL2	FKRP	FKTN	FREM2	FXN	GAA	GATAD1	GCKR	GJA5
	GLA	GPD1L	GPIHBP1	GRM1	HADHA	HCN4	HFE	HPSE2	HRAS	HSPB8	IL12RB2	ILK
	JAG1	JPH2	JUP	KCNA5	KCND3	KCNE1	KCNE2	KCNE3	KCNH2	KCNJ2	KCNJ5	KCNJ8
	KCNQ1	KIAA1244	KLF10	KRAS	L2HGDH	LAMA2	LAMA3	LAMA4	LAMP2	LDB3	LDLR	LDLRAP1
	LMF1	LMNA	LPL	LTBP2	MAP2K1	MAP2K2	MIB1	MURC	MYBPC3	MYH11	MYH6	MYH7
	MYL2	MYL3	MYLK	MYLK2	MYO6	MYOZ2	MYPN	NDUFV3	NEXN	NKX2-5	NODAL	NOTCH1
	NPHS2	NPPA	NRAS	PCNXL2	PCSK9	PDLIM3	PDP1	PKP2	PLN	PRDM16	PRKAG2	PRKAR1A
	PRUNE2	PTPN11	RAF1	RANGRF	RBM20	RIPK3	RYR1	RYR2	SALL4	SCN1B	SCN2B	SCN3B
	SCN4B	SCN5A	SCO2	SDHA	SEPN1	SERPIND1	SGCB	SGCD	SGCG	SHOC2	SLC12A6	SLC25A4
	SLC2A10	SMAD3	SMAD4	SNTA1	SOS1	SOX6	SP4	SREBF2	SRY	SUMF1	TAZ	TBX20
	TBX3	TBX5	TCAP	TDRD7	TGFB2	TGFB3	TGFBR1	TGFBR2	TMEM43	TMPO	TNNC1	TNNI3
	TNNT2	TPM1	TRDN	TRIM63	TRPM4	TTN	TTR	TXNRD2	USP6NL	VCAN	VCL	WNK1
	ZBTB17	ZHX3	ZIC3									
Common Hereditary Cancer Panel (*94 genes, 231.5 Kb*) **Related Diseases:** Medical checkup	AARS	ABCA13	ABCB11	APC	ARHGAP35	ATM	ATRX	BARD1	BMPR1A	BRAF	BRCA1	BRCA2
	BRIP1	CDH1	CDKN2A	CHEK2	COG1	COL4A4	CPNE1	DNMT1	EGLN1	EGLN2	EPAS1	EPCAM
	EVC	FERMT1	FGFR1	FH	FREM2	GRM1	H3F3A	HPSE2	HRAS	IDH2	IL12RB2	KIAA1244
	KIF1B	KMT2D	L2HGDH	LAMA3	MAX	MDH2	MEN1	MERTK	MET	MLH1	MRE11	MSH2
	MSH6	MUTYH	NBN	NDUFV3	NF1	NF2	NPHS2	PALB2	PCNXL2	PDP1	PMS2	POLD1
	POLE	PRSS1	PRUNE2	PTEN	RAD50	RAD51C	RAD51D	RB1	RET	RIPK3	SDHA	SDHAF2
	SDHB	SDHC	SDHD	SERPIND1	SLC12A6	SMAD4	SOX6	SP4	SPINK1	SRY	STK11	SUMF1
	TDRD7	TMEM127	TP53	TSC1	TSC2	USP6NL	VCAN	VHL	WNK1	WT1		
Epilepsy Panel (*150 genes, 414.5 Kb*) **Related Diseases:** Epilepsy	AARS	ABCA13	ABCB11	ADGRV1	ADSL	ALDH7A1	ALG13	ARHGAP35	ARHGEF15	ARHGEF9	ARX	ASAH1
	ATP1A2	ATP6AP2	CACNA1A	CASK	CDKL5	CHD2	CHRNA2	CHRNA4	CHRNA7	CHRNB2	CLCN4	CLN3
	CLN5	CLN6	CLN8	CNTNAP2	COG1	COL4A4	CPNE1	CSTB	CTSD	DCX	DEPDC5	DLG3
	DNAJC5	DNM1	DNMT1	DOCK7	DYRK1A	EEF1A2	EPM2A	EVC	FERMT1	FOLR1	FOXG1	FREM2
	GABRA1	GABRA2	GABRB3	GABRG2	GAMT	GATM	GNAO1	GOSR2	GRIN1	GRIN2A	GRIN2B	GRM1
	HCN1	HDAC4	HNRNPU	HPSE2	IL12RB2	IQSEC2	KANSL1	KCNA2	KCNB1	KCNH5	KCNJ10	KCNMA1
	KCNQ2	KCNQ3	KCNT1	KCTD7	KIAA1244	L2HGDH	LAMA3	LGI1	MAGI2	MBD5	MECP2	MEF2C
	MFSD8	NDUFV3	NECAP1	NHLRC1	NPHS2	NR2F1	NRXN1	PCDH19	PCNXL2	PDP1	PIGA	PIGO
	PIGQ	PIGV	PLCB1	PNKP	PNPO	POLG	PPT1	PRICKLE1	PRICKLE2	PRRT2	PRUNE2	QARS
	RELN	RIPK3	SCARB2	SCN1A	SCN1B	SCN2A	SCN8A	SCN9A	SERPIND1	SLC12A6	SLC13A5	SLC25A22
	SLC2A1	SLC35A2	SLC6A8	SLC9A6	SMS	SOX6	SP4	SPTAN1	SRPX2	SRY	ST3GAL3	STXBP1
	SUMF1	SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TDRD7	TPP1	TSC1	TSC2	UBE3A
	USP6NL	VCAN	WDR45	WNK1	WWOX	ZEB2						

LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Gene List											
Hearing Loss-Deafness Panel (*63 genes, 143.9 Kb*) **Related Diseases:** Hearing loss, Deafness	AARS	ABCA13	ABCB11	ARHGAP35	CDH23	CLRN1	COCH	COG1	COL11A1	COL2A1	COL4A4	CPNE1
	DIAPH1	DNMT1	EDNRB	EVC	EYA1	FERMT1	FREM2	GJB2	GJB6	GRM1	HPSE2	IL12RB2
	KCNE1	KCNQ1	KCNQ4	KIAA1244	L2HGDH	LAMA3	MITF	MYO15A	MYO7A	NDUFV3	NPHS2	OTOF
	PAX3	PCNXL2	PDP1	POU3F4	PRUNE2	RIPK3	SERPIND1	SIX5	SLC12A6	SLC26A4	SNAI2	SOX10
	SOX6	SP4	SRY	SUMF1	TDRD7	TECTA	TMC1	TMIE	TMPRSS3	USH1C	USH2A	USP6NL
	VCAN	WFS1	WNK1									
Lymphoid Leukemia Panel (*85 genes, 139.9 Kb*) **Related Diseases:** Acute lymphatic leukemia	AARS	ABCA13	ABCB11	ABL1	AMELX	AMELY	ARHGAP35	BRAF	BTG1	CDKN2A	COG1	COL4A4
	CPNE1	CREBBP	CRLF2	DNM2	DNMT1	DNMT3A	EP300	ETV6	EVC	EZH2	FBXW7	FERMT1
	FLT3	FREM2	GATA3	GRM1	HPSE2	IDH1	IDH2	IKZF1	IL12RB2	IL7R	JAK1	JAK2
	JAK3	KDM6A	KIAA1244	KMT2A	KMT2D	KRAS	L2HGDH	LAMA3	LEF1	LMO1	MAPK1	NDUFV3
	NF1	NOTCH1	NPHS2	NRAS	NSD2	NT5C2	NUDT15	PAX5	PCNXL2	PDP1	PHF6	PRUNE2
	PTEN	PTPN11	RB1	RIPK3	RUNX1	SERPIND1	SETD2	SH2B3	SLC12A6	SOX6	SP4	SRY
	STAG2	STAT3	STAT5B	SUMF1	TBL1XR1	TCF3	TDRD7	TP53	TPMT	USP6NL	VCAN	WNK1
	WT1											
Lymphoma Panel (*83 genes, 131.2 Kb*) **Related Diseases:** Lymphoma	AARS	ABCA13	ABCB11	ALK	ARHGAP35	ATM	B2M	BCL6	BIRC3	BRAF	BTK	CARD11
	CD79A	CD79B	COG1	COL4A4	CPNE1	CREBBP	CXCR4	DNMT1	EGR2	EP300	EVC	EZH2
	FAS	FAT4	FBXO11	FERMT1	FREM2	GRM1	HPSE2	ID3	IDH2	IKBKB	IKZF1	IL12RB2
	JAK3	KIAA1244	KLF2	L2HGDH	LAMA3	MYC	MYD88	NDUFV3	NFKBIE	NOTCH1	NOTCH2	NPHS2
	PCNXL2	PDP1	PLCG1	PLCG2	POT1	PRDM1	PRUNE2	RHOA	RIPK3	RPS15	RRAGC	SERPIND1
	SF3B1	SLC12A6	SOCS1	SOX6	SP4	SRY	STAT3	STAT5B	SUMF1	TBL1XR1	TCF3	TDRD7
	TET2	TNFAIP3	TNFRSF14	TP53	TP63	TRAF3	UBR5	USP6NL	VCAN	WNK1	XPO1	
Lysosomal Storage Diseases Panel (*118 genes, 209.4 Kb*) **Related Diseases:** Lysosomal storage disease	AARS	ABCA13	ABCB11	ABCD1	ACOX1	AGA	AGL	ALDOA	ALDOB	ARHGAP35	ARSA	ARSB
	ATP13A2	ATP7A	ATP7B	CLN3	CLN5	CLN6	CLN8	COG1	COL4A4	CPNE1	CTNS	CTSA
	CTSD	CTSF	DNAJC5	DNMT1	EVC	FERMT1	FREM2	FUCA1	G6PC	GAA	GALC	GALE
	GALK1	GALK2	GALNS	GALT	GBA	GBE1	GJB2	GLA	GLB1	GNPTAB	GNPTG	GNS
	GRM1	GRN	GUSB	GYS1	GYS2	HEXA	HEXB	HGSNAT	HPRT1	HPSE2	HYAL1	IDS
	IDUA	IL12RB2	KCTD7	KIAA1244	L2HGDH	LAMA3	LDHA	LIPA	MAN2B1	MANBA	MCOLN1	MFSD8
	NAGA	NAGLU	NDUFV3	NEU1	NPC1	NPC2	NPHS2	PCNXL2	PDP1	PEX1	PEX10	PEX12
	PEX13	PEX14	PEX16	PEX19	PEX2	PEX26	PEX3	PEX5	PEX6	PFKM	PHKA2	PHKB
	PHKG2	PPT1	PRUNE2	PYGL	PYGM	RIPK3	SERPIND1	SGSH	SLC12A6	SLC17A5	SLC2A2	SLC37A4
	SMPD1	SOX6	SP4	SRY	SUMF1	TDRD7	TPP1	USP6NL	VCAN	WNK1		
Metabolic Disorders Panel (*104 genes, 151.8 Kb*) **Related Diseases:** Inborn errors of metabolism	AARS	ABCA13	ABCB11	ABCD1	ACAD8	ACADM	ACADS	ACADSB	ACADVL	ACAT1	AHCY	ARG1
	ARHGAP35	ASL	ASS1	AUH	BCKDHA	BCKDHB	BTD	CBS	COG1	COL4A4	CPNE1	CPS1
	CPT1A	CPT2	DBT	DECR1	DHCR7	DLD	DNMT1	ETFA	ETFB	ETFDH	EVC	FAH
	FERMT1	FREM2	GALE	GALK1	GALT	GAMT	GATM	GCDH	GCH1	GNMT	GRM1	HADH
	HADHA	HADHB	HLCS	HMGCL	HPD	HPSE2	HSD17B10	IL12RB2	IVD	KIAA1244	L2HGDH	LAMA3
	LMBRD1	MAT1A	MCCC1	MCCC2	MLYCD	MMAA	MMAB	MMACHC	MMADHC	MMUT	MTHFR	MTR
	MTRR	NDUFV3	NPHS2	OPA3	OTC	PAH	PCBD1	PCCA	PCCB	PCNXL2	PDP1	PRUNE2
	PTS	QDPR	RIPK3	SERPIND1	SLC12A6	SLC22A5	SLC25A13	SLC25A20	SLC6A8	SOX6	SP4	SRY
	SUMF1	TAT	TAZ	TCN2	TDRD7	USP6NL	VCAN	WNK1				
Myeloid Leukemia Panel (*84 genes, 117 Kb*) **Related Diseases:** Acute myeloid leukemia	AARS	ABCA13	ABCB11	AMELX	AMELY	ANKRD26	ARHGAP35	ASXL1	ATRX	BCOR	BCORL1	BRAF
	CALR	CBL	CBLB	CEBPA	COG1	COL4A4	CPNE1	CSF3R	DDX41	DNMT1	DNMT3A	ETV6
	EVC	EZH2	FERMT1	FLT3	FREM2	GATA1	GATA2	GRM1	HPSE2	HRAS	IDH1	IDH2
	IL12RB2	JAK2	JAK3	KDM6A	KIAA1244	KIT	KRAS	L2HGDH	LAMA3	MPL	NDUFV3	NOTCH1
	NPHS2	NPM1	NRAS	PCNXL2	PDGFRA	PDP1	PHF6	PPM1D	PRUNE2	PTPN11	RAD21	RIPK3
	RUNX1	SERPIND1	SETBP1	SF3B1	SLC12A6	SMC1A	SMC3	SOX6	SP4	SRSF2	SRY	STAG1
	STAG2	STAT3	SUMF1	TDRD7	TET2	TP53	U2AF1	USP6NL	VCAN	WNK1	WT1	ZRSR2
Neuromuscular Panel (*321 genes, 1.2 Mb*) **Related Diseases:** Neuromuscular disease	AARS	ABCA13	ABCB11	ABCB7	ABCD1	ABHD12	ACAD9	ACADL	ACADM	ACO2	ACTA1	ADCK3
	AFG3L2	AGL	AIFM1	ALDH3A2	AMPD1	ANO10	ANO5	AP4B1	AP4E1	AP4M1	AP4S1	AP5Z1
	APTX	ARHGAP35	ARSA	ATCAY	ATL1	ATM	ATP2A1	ATP7A	ATP7B	ATP8A2	BAG3	BEAN1
	BIN1	BSCL2	C10orf2	C12orf65	C19orf12	CACNA1A	CACNA1S	CACNB4	CAPN3	CASK	CAV3	CCDC78
	CCDC88C	CFL2	CHAT	CHRNA1	CHRNB1	CHRND	CHRNE	CHRNG	CLCN1	CLCN2	CLN5	CNTN1
	COG1	COL4A4	COL6A1	COL6A2	COL6A3	COLQ	CPNE1	CPT1B	CPT2	CRYAB	CTDP1	CWF19L1
	CYP27A1	CYP2U1	CYP7B1	DAG1	DCTN1	DDHD1	DDHD2	DES	DMD	DNAJB2	DNAJB6	DNM2
	DNMT1	DOK7	DYNC1H1	DYSF	EEF2	EGR2	ELOVL4	ELOVL5	EMD	ERLIN2	ETFA	ETFB
	EVC	FA2H	FAM134B	FERMT1	FGD4	FGF14	FHL1	FIG4	FKRP	FKTN	FLNC	FLVCR1
	FREM2	FRMD7	FUS	FXN	GAA	GAD1	GALC	GAN	GARS	GBA2	GDAP1	GJB1
	GJC2	GLA	GLE1	GNB4	GNE	GOSR2	GPR143	GRID2	GRM1	GYS1	HADHA	HADHB

Panel Name	Gene List											
Neuromuscular Panel (*321 genes, 1.2 Mb*) **Related Diseases:** Neuromuscular disease	HINT1	HOXD10	HPSE2	HSPB1	HSPB8	HSPD1	HSPG2	IGHMBP2	IKBKAP	IL12RB2	ISPD	ITGA7
	ITPR1	JPH3	KBTBD13	KCNA1	KCNC3	KCND3	KCNE3	KCNJ10	KCNJ18	KIAA0196	KIAA1244	KIF1A
	KIF1B	KIF1C	KIF5A	KLHL40	KLHL41	L1CAM	L2HGDH	LAMA1	LAMA2	LAMA3	LARGE	LDB3
	LITAF	LMNA	LPIN1	LRSAM1	MARS	MARS2	MATR3	MED25	MFN2	MPZ	MRE11A	MTM1
	MTMR14	MTMR2	MTPAP	MTTP	MUSK	MYF6	MYH2	MYH7	MYOT	NDRG1	NDUFV3	NEB
	NEFL	NGF	NIPA1	NOP56	NPHS2	NTRK1	OPA1	OPA3	OPHN1	PABPN1	PANK2	PCNXL2
	PDK3	PDP1	PDYN	PEX7	PFKM	PGAM2	PHKA1	PHYH	PLEC	PLEKHG5	PLP1	PMM2
	PMP22	PNKP	PNPLA6	POLG	POLG2	POMGNT1	POMT1	POMT2	PRKCG	PRPS1	PRUNE2	PRX
	PTF1A	PTRF	PYGM	RAB7A	RAPSN	REEP1	RIPK3	RNF216	RRM2B	RTN2	RUBCN	RYR1
	RYR2	SACS	SBF2	SCN4A	SCN9A	SEPN1	SERPIND1	SETX	SGCA	SGCB	SGCD	SGCE
	SGCG	SH3TC2	SIL1	SLC12A6	SLC16A2	SLC1A3	SLC33A1	SLC39A4	SLC52A2	SLC9A1	SLC9A6	SMN1
	SNX14	SOD1	SOX6	SP4	SPAST	SPG11	SPG20	SPG21	SPG7	SPTBN2	SPTLC1	SPTLC2
	SRY	STAC3	STUB1	SUCLA2	SUMF1	SYNE1	SYNE2	SYT14	TBP	TCAP	TDP1	TDRD7
	TECPR2	TGM6	TK2	TMEM240	TNNI2	TNNT1	TPM2	TPM3	TPP1	TRIM32	TRPV4	TTBK2
	TTN	TTPA	TTR	TUBB4A	TYMP	USP6NL	VAMP1	VCAN	VCP	VLDLR	VPS13A	VPS37A
	VRK1	WFS1	WNK1	WWOX	XK	YARS	ZFYVE26	ZFYVE27	ZNF592			
Retinitis Pigmentosa Panel (*111 genes, 325.3 Kb*) **Related Diseases:** Retinitis pigmentosa	ABCA4	ABHD12	ADAM9	ADGRA3	AGBL5	AIPL1	ARHGEF18	ARL2BP	ARL3	ARL6	BBS1	BBS2
	BEST1	C2orf71	C8orf37	CA4	CABP4	CACNA1F	CACNA2D4	CDHR1	CERKL	CLRN1	CNGA1	CNGB1
	CNGB3	CNNM4	CRB1	CRX	CWC27	CYP4V2	DHDDS	DHX38	ELOVL4	EMC1	EYS	FAM161A
	FLVCR1	FSCN2	GNAT2	GUCA1A	GUCA1B	GUCY2D	HGSNAT	HK1	IDH3B	IFT140	IFT172	IMPDH1
	IMPG2	KCNV2	KIAA1549	KIZ	KLHL7	LRAT	MAK	MERTK	MVK	NEK2	NEUROD1	NR2E3
	NRL	OFD1	PDE6A	PDE6B	PDE6C	PDE6G	PDE6H	PITPNM3	POMGNT1	PRCD	PRKCG	PROM1
	PRPF3	PRPF31	PRPF4	PRPF6	PRPF8	PRPH2	RAB28	RAX2	RBP3	RDH12	RDH5	REEP6
	RGR	RGS9	RGS9BP	RHO	RIMS1	RLBP1	ROM1	RP1	RP2	RP9	RPE65	RPGR
	RPGRIP1	SAG	SEMA4A	SLC7A14	SNRNP200	SPATA7	SPP2	TOPORS	TRNT1	TTC8	TULP1	UNC119
	USH2A	ZNF408	ZNF513									
Short Stature Panel (*193 genes, 629 Kb*) **Related Diseases:** Short stature	AARS	ABCA13	ABCB11	ACTA2	ADAMTS10	ADAMTS2	ADAMTSL4	AGPS	ALPL	ARHGAP35	ARSE	ATP6V0A2
	ATP7A	ATRX	B3GALT6	B4GALT7	BGN	BLM	BRAF	CBL	CBS	CDC6	CDT1	CHST14
	COG1	COL10A1	COL11A1	COL1A1	COL1A2	COL2A1	COL3A1	COL4A4	COL5A1	COL5A2	COL9A1	COL9A2
	COL9A3	COMP	CPNE1	CREBBP	CRTAP	CTSK	CUL7	DHCR7	DLL3	DNMT1	DYNC2H1	DYRK1A
	EBP	EFEMP2	ELN	EP300	ERCC6	ERCC8	EVC	EVC2	EXT1	EXT2	FBLN5	FBN1
	FBN2	FERMT1	FGD1	FGF23	FGFR1	FGFR2	FGFR3	FKBP10	FLNA	FLNB	FOXE3	FREM2
	GH1	GHR	GHRHR	GLI2	GLI3	GNAS	GNPAT	GRM1	HESX1	HPSE2	HRAS	HSPG2
	IFITM5	IFT80	IGF1	IGF1R	IL12RB2	INPPL1	KCNJ2	KCNJ8	KDM6A	KIAA1244	KMT2D	KRAS
	L2HGDH	LAMA3	LBR	LHX3	LIFR	LOX	LTBP2	LZTR1	MAP2K1	MAP2K2	MAT2A	MATN3
	MED12	MFAP5	MYH11	MYLK	NBAS	NBN	NDUFV3	NEK1	NF1	NIPBL	NPHS2	NRAS
	NSDHL	OBSL1	ORC1	ORC4	ORC6	P3H1	PCNT	PCNXL2	PDP1	PEX7	PHEX	PLOD1
	POR	POU1F1	PPIB	PPP1CB	PRKG1	PROP1	PRUNE2	PTPN11	PYCR1	RAF1	RIN2	RIPK3
	RIT1	RMRP	ROR2	RPS6KA3	RUNX2	SBDS	SERPIND1	SERPINH1	SHOC2	SKI	SLC12A6	SLC26A2
	SLC2A10	SLC34A3	SLC35D1	SLC39A13	SMAD3	SMARCAL1	SMC1A	SMC3	SMS	SOS1	SOS2	SOX3
	SOX6	SOX9	SP4	SPRED1	SRCAP	SRY	SUMF1	TDRD7	TGFB1	TGFB2	TGFB3	TGFBR1
	TGFBR2	THRB	TRIM37	TRIP11	TRPS1	TRPV4	TTC21B	USP6NL	VCAN	WDR19	WDR35	WNK1
	WRN											
Skin Disorder Panel (*152 genes, 545.7 Kb*) **Related Diseases:** Skin diseases	ABCA12	ABCB6	ABCC6	ABHD5	ADAMTS2	ADAR	ALAD	ALAS2	ALDH3A2	ALOX12B	ALOXE3	APIS1
	ATM	ATP2A2	ATP2C1	ATP6V0A2	BLM	CARD14	CDH3	CDSN	CLDN1	COL17A1	COL1A1	COL1A2
	COL3A1	COL5A1	COL5A2	COL7A1	CPOX	CTC1	CTSC	CYP4F22	DDB2	DKC1	DOCK8	DSG1
	DSG4	DSP	DST	EBP	ECM1	EDA	EDAR	EDARADD	EFEMP2	ELN	ERCC2	ERCC3
	ERCC4	ERCC5	EXPH5	FANCA	FANCC	FANCG	FECH	FERMT1	FLCN	FLG	GJB2	GJB3
	GJB4	GJB6	GNAS	GORAB	GPR143	GSN	GTF2H5	HFE	HMBS	HR	IL36RN	ITGA3
	ITGA6	ITGB4	JUP	KIT	KRT1	KRT10	KRT14	KRT16	KRT17	KRT2	KRT5	KRT6A
	KRT6B	KRT6C	KRT81	KRT83	KRT86	KRT9	LAMA3	LAMB3	LAMC2	LIPH	LIPN	LOR
	LPAR6	LYST	MBTPS2	NF1	NF2	NHP2	NIPAL4	NOP10	NSDHL	OCA2	PKP1	PLEC
	PLOD1	PNPLA1	POFUT1	POGLUT1	POLH	POMP	PPOX	PRKAR1A	PTCH1	PTCH2	PYCR1	RECQL4
	RTEL1	SLC27A4	SLC39A4	SLC45A2	SLURP1	SNAP29	SPINK5	SPRED1	ST14	STAT3	STS	SUFU
	TERC	TERT	TGM1	TGM5	TINF2	TNXB	TRPV3	TSC1	TSC2	TTR	TYK2	TYR
	TYRP1	UROD	UROS	WAS	WRAP53	XPA	XPC	ZMPSTE24				
Solid Tumor Panel (*61 genes, 109.4 Kb*) **Related Diseases:** Somatic cancer	ABL1	AKT1	ALK	APC	ATM	BRAF	BRCA1	BRCA2	CDH1	CDKN2A	CSF1R	CTNNB1
	DLC1	EGFR	ERBB	ERBB2	ERBB4	ESR1	FBXW7	FGFR1	FGFR2	FGFR3	FTSJ3	GNA11
	GNAQ	GNAS	HNF1A	HRAS	IDH1	IDH2	JAK2	JAK3	KCNB2	KDR	KIT	KRAS
	MET	MLH1	MYC	MYCN	NOTCH1	NRAS	NRXN1	PDGFRA	PIK3CA	PTEN	PTPN11	RB1
	RBAK	RET	ROS1	SMAD4	SMARCB1	SMO	SMURF1	SRC	SSFA2	STK11	TP53	VHL
	ZNF594											

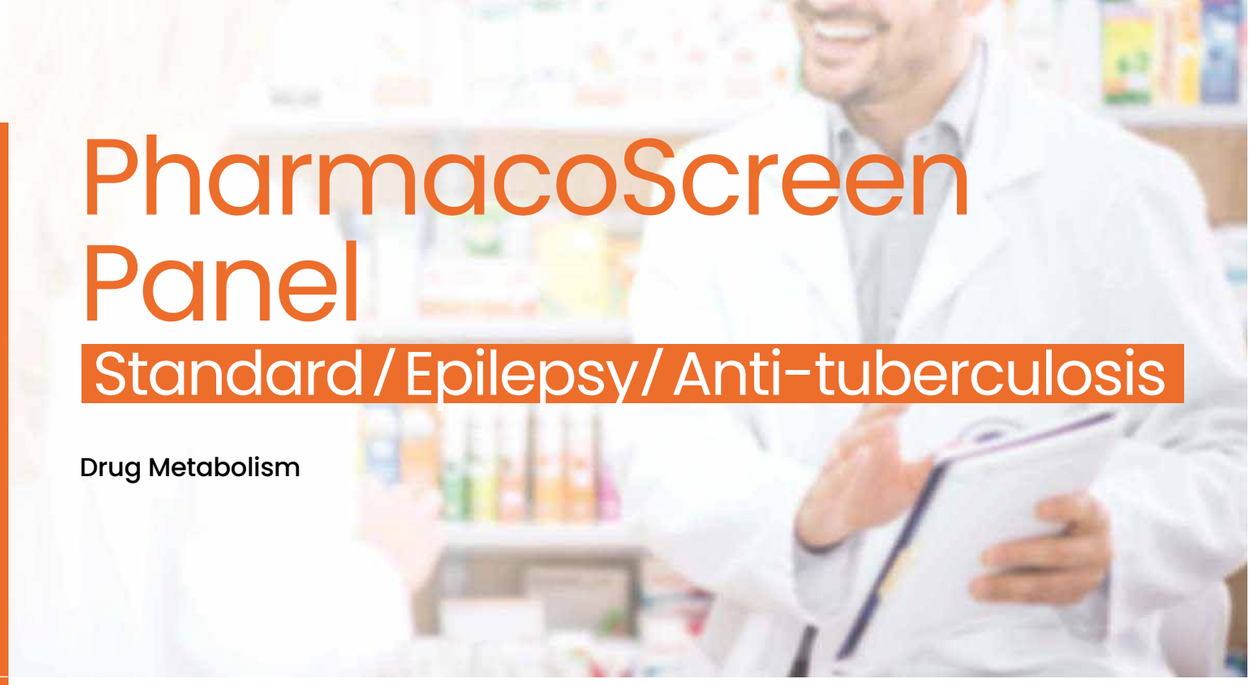
READY-TO-USE PANELS FOR PHARMACO-GENOMICS

CELEMICS PRODUCTS & SERVICES 2022

PharmacoScreen Panel

· Standard / Epilepsy / Anti-tuberculosis

CELEMICS



PharmacoScreen Panel

Standard / Epilepsy/ Anti-tuberculosis

Drug Metabolism

DESCRIPTION

The main target of PharmacoScreen Panel is the genes associated with prescribed drugs of the corresponding diseases. The assay allows for precise selection and dosage of prescribed drugs, and detection of genetic variants associated with drug metabolism, epilepsy and anti-tuberculosis.

KEY FEATURES

1. Assess extensive target regions associated with pharmacogenomics	Target over 120 genes associated with pharmacokinetics and pharmacodynamics
2. Validated panel performance	Collaborated with 4 major university hospitals on a government project Complete validation for clinical application
3. Flexible panel contents	PharmacoScreen Panels for drug metabolism, epilepsy, and anti-tuberculosis.

PANEL PERFORMANCE

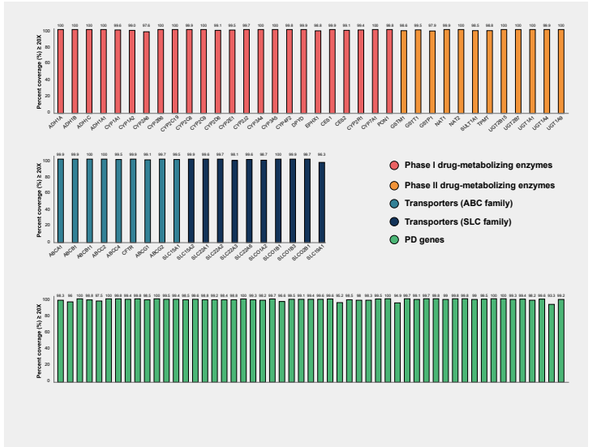
The panel performance test resulted in 99.9% specificity and 99.7% sensitivity.

1.1 Phase I/II drug-metabolizing enzyme (Drug metabolism)

1.2 ABC & SLC family transporter genes (Drug effect)

1.3 Pharmacodynamics genes (Drug biochemical and physiological)

1.4 Modifier genes (Drug ADME enhancement)



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

PharmacoScreen

Standard

DESCRIPTION

One of the major problems of organ transplantation is tissue damage by rejection and relapse of the disease after transplantation. Although applying immunosuppressive drugs can prevent rejection, determining the proper dosage of immunosuppressive drugs for an individual patient is challenging. The PharmacoScreen Standard Panel is an NGS assay, designed to assess 122 genes associated with pharmacogenomics ,including drug metabolism (Phase I, II), Transporters (ABC and SLC families), and Parkinson's disease-related genes (PD genes). The panel is not limited to 122 genes, and more genes of interest can be added through our Gene Add-on service.

SPECIFICATION

Gene count*	283 genes
Covered region	Whole CDS, UTR (-50 bp, +10 bp)
Target Size	634 kb
Mutation Type	SNV, Indel, CNV
Sample type (amount)	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Sensitivity / Specificity	100% / 94.5%
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)
Publications	Targeted Next-Generation Sequencing for Comprehensive Genetic Profiling of Pharmacogenes, Clinical Pharmacology & Therapeutics, 2016

* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

- ● Phase I drug-metabolizing enzymes
- ● Phase II drug-metabolizing enzymes
- ▲ Transporters (ABC family)
- ▲ Transporters (SLC family)
- ■ PD genes
- ◆ Modifier genes

● ADH1A	● GSTM1	▲ ABCA1	■ ACE	■ ADRB1	■ KCNH2	◆ AHR
● ADH1B	● GSTP1	▲ ABCB1	■ ADRB2	■ ALOX5	■ LDLR	◆ KCNJ11
● ADH1C	● GSTT1	▲ ABCB11	■ BRCA1	■ APOA1	■ MAOA	◆ NR1I3
● ALDH1A1	● NAT1	▲ ABCC2	■ COMT	■ ARID5B	■ NR3C2	◆ NR1I2
● CES1	● NAT2	▲ ABCC3	■ DRD2	■ BDNF	■ NTRK2	◆ POR
● CES2	● SULT1A1	▲ ABCC4	■ F5	■ CACNA1C	■ PEAR1	◆ SOD2
● CYP1A1	● TPMT	▲ ABCC7	■ HMGCR	■ CPS1	■ PTGS1	
● CYP1A2	● UGT1A1	▲ ABCG1	■ MTHFR	■ CRHR1	■ PTGS2	
● CYP2A6	● UGT1A4	▲ ABCG2	■ NQO1	■ DBH	■ RYR1	
● CYP2B6	● UGT1A9	▲ SLC10A1	■ P2RY1	■ DRD1	■ RYR2	
● CYP2C19	● UGT1A10	▲ SLC15A1	■ P2RY12	■ EGFR	■ SCN1A	
● CYP2C8	● UGT2B15	▲ SLC15A2	■ PTGIS	■ ESR1	■ SCN2A	
● CYP2C9	● UGT2B7	▲ SLC19A1	■ SCN5A	■ FKBP5	■ SLC47A1	
● CYP2D6		▲ SLC22A1	■ TYMS	■ GLCCI1	■ SLC47A2	
● CYP2E1		▲ SLC22A2	■ VDR	■ GRK4	■ SLC6A3	
● CYP2J2		▲ SLC22A3	■ VKORC1	■ GRK5	■ SLC6A4	
● CYP2R1		▲ SLC22A4		■ G6PD	■ TBXAS1	
● CYP3A4		▲ SLC22A5		■ HTR1A	■ ZNF423	
● CYP3A5		▲ SLC22A6		■ HTR2A		
● CYP4F2		▲ SLC22A8				
● CYP7A1		▲ SLC22A11				
● DPYD		▲ SLC22A12				
● EPHX1		▲ SLCO1A2				
● PON1		▲ SLCO1B1				
		▲ SLCO1B3				
		▲ SLCO2B1				

PharmacoScreen Panel Standard	ABCA1	ABCB1	ABCB11	ABCB4	ABCB7	ABCC1	ABCC2	ABCC3	ABCC4	ABCC5	ABCC6	ABCC8	ABCC9	ABCG1	ABCG2
	ABP1	ACE	ADH1A	ADH1B	ADH1C	ADH4	ADH5	ADH6	ADH7	ADRB1	ADRB2	AHR	AKAP9	ALB	ALDH1A1
	ALDH2	ALDH3A1	ALDH3A2	ALOX5	AOX1	APOA1	APOA2	ARID5B	ARNT	ARSA	ATP7A	ATP7B	BDNF	BRCA1	CA5P
	CACNA1C	CBR1	CBR3	CDA	CES1	CES2	CFTR	CHST1	CHST10	CHST11	CHST13	CHST2	CHST3	CHST4	CHST5
	CHST6	CHST7	CHST8	CHST9	COMT	CPS1	CRHR1	CROT	CYP11A1	CYP11B1	CYP11B2	CYP17A1	CYP19A1	CYP1A1	CYP1A2
	CYP1B1	CYP20A1	CYP21A2	CYP24A1	CYP26A1	CYP26C1	CYP27A1	CYP27B1	CYP2A13	CYP2A6	CYP2A7	CYP2B6	CYP2B7P1	CYP2C18	CYP2C19
	CYP2C8	CYP2C9	CYP2D6	CYP2E1	CYP2F1	CYP2J2	CYP2R1	CYP2S1	CYP39A1	CYP3A4	CYP3A43	CYP3A5	CYP3A7	CYP46A1	CYP4A11
	CYP4B1	CYP4F11	CYP4F12	CYP4F2	CYP4F3	CYP4F8	CYP4Z1	CYP51A1	CYP7A1	CYP7B1	CYP8B1	DBH	DCK	DPYD	DRD1
	DRD2	EGFR	EPHX1	EPHX2	ESR1	F5	FAAH	FKBP5	FMO1	FMO2	FMO3	FMO4	FMO5	FMO6	G6PD
	GLCCI1	GRK4	GRK5	GSTA1	GSTA2	GSTA3	GSTA4	GSTA5	GSTM1	GSTM2	GSTM3	GSTM4	GSTM5	GSTO1	GSTP1
	GSTT1	GSTT2	GSTZ1	HMGCR	HNMT	HTR1A	HTR2A	KCNH2	KCNJ11	LDLR	MAOA	MAOB	MAT1A	METTL1	MTHFR
	NAT1	NAT2	NNMT	NQO1	NR1I2	NR1I3	NR3C1	NR3C2	NTRK2	ORM1	ORM2	P2RY1	P2RY12	PEAR1	PGAP3
	PNMT	PON1	PON2	PON3	POR	PPARD	PPARG	PPP1R9A	PRSS53	PTGIS	PTGS1	PTGS2	QPRT	RALBP1	RPL13
	RXRA	RYR1	RYR2	SCN1A	SCN2A	SCN5A	SERPINA7	SETD4	SLC10A1	SLC10A2	SLC13A1	SLC15A1	SLC15A2	SLC16A1	SLC19A1
	SLC22A1	SLC22A11	SLC22A12	SLC22A13	SLC22A14	SLC22A2	SLC22A3	SLC22A4	SLC22A5	SLC22A6	SLC22A7	SLC22A8	SLC25A27	SLC28A1	SLC28A2
	SLC28A3	SLC29A1	SLC29A2	SLC47A1	SLC47A2	SLC5A6	SLC6A3	SLC6A4	SLC6A6	SLC7A5	SLC7A7	SLC7A8	SLCO1A2	SLCO1B1	SLCO1B3
	SLCO2B1	SLCO3A1	SLCO4A1	SLCO5A1	SOD2	SPG7	SPN	SULT1A1	SULT1A2	SULT1A3	SULT1B1	SULT1C2	SULT1C4	SULT1E1	SULT2A1
	SULT2B1	SULT4A1	TBXAS1	TPMT	TPSG1	TYMS	UGT1A	UGT1A1	UGT1A10	UGT1A3	UGT1A4	UGT1A5	UGT1A6	UGT1A7	UGT1A8
	UGT1A9	UGT2A1	UGT2B11	UGT2B15	UGT2B17	UGT2B28	UGT2B4	UGT2B7	UGT8	VDR	VKORC1	XDH	ZNF423	ZNF423	

PharmacoScreen

Epilepsy

DESCRIPTION

The PharmacoScreen Epilepsy Panel, designed for research studies on epilepsy, consists of 91 genes associated with anti-epileptic drugs. Epilepsy is one of the most common neurological disorders, with its estimated prevalence is one out of 100 worldwide and constantly increasing. Epilepsy is usually treated by consistent application of anti-epileptic drugs. The aim of the treatment is to prevent seizures with no issues of side effects. Although over 20 different anti-epileptic drugs have been developed, most of the drugs failed to prevent seizures, or faced challenges of determining the proper dosage for an individual patient. The genetic factor is one of clinical factors to be considered.

SPECIFICATION

Gene count*	91 genes
Covered region	Whole CDS + UTR (-50 bp, +10 bp)
Target size	575 kb
Mutation type	SNV, Indel, CNV
Sample type (amount)	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

PharmacoScreen Panel Epilepsy	ANKK1	CACNA1A	CACNA1B	CACNA1D	CACNA1E	CACNA1F	CACNA1G	CACNA1H	CACNA1I	CACNA1S	CACNA2D1	CACNA2D2	CACNA2D3
	CACNA2D4	CACNB1	CACNB2	CACNB3	CACNB4	CACNG1	CACNG2	CACNG3	CACNG4	CACNG5	CACNG6	CACNG7	CACNG8
	CDH13	CLCN2	EFHC1	GABRA1	GABRA2	GABRA3	GABRA4	GABRA5	GABRA6	GABRB1	GABRB2	GABRB3	GABRD
	GABRE	GABRG1	GABRG2	GABRG3	GABRP	GABRQ	GABRR1	GABRR2	GABRR3	GRIA1	GRIA2	GRIA3	GRIA4
	GRIK1	GRIK2	GRIK3	GRIK4	GRIK5	GRIN1	GRIN2A	GRIN2B	GRIN2C	GRIN2D	GRIN3A	GRIN3B	HNF4A
	HTR1B	KCNA2	KCNB1	KCNC1	KCND3	KCNH1	KCNJ10	KCNQ2	KCNQ3	KCNT1	KCNTD7	LEPR	MAOA
	MAOB	RBFOX1	SCN1A	SCN2A	SCN3A	SCN8A	STS	TPH1	TPH2	UGT1A10	UGT1A6	UGT1A7	UGT1A9

PharmacoScreen

Anti-tuberculosis

DESCRIPTION

The PharmacoScreen Anti-tuberculosis Panel assesses genes associated with liver injury. Drug-induced liver injury (DILI), which is an important cause of acute liver failure, can be a threat to a patient and a common reason why some drug development projects are discontinued. According to a spontaneous reporting database from a research network of pharmacovigilance institutions in Korea, anti-tuberculosis drugs are reported to be the most common factor that leads to DILI demanding precise and personalized medicine.

SPECIFICATION

Gene count*	132 genes
Covered regions	Whole CDS + UTR (-50 bp, +10 bp)
Target size	186 kb
Mutation type	SNV, Indel, CNV
Sample type (amount)	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

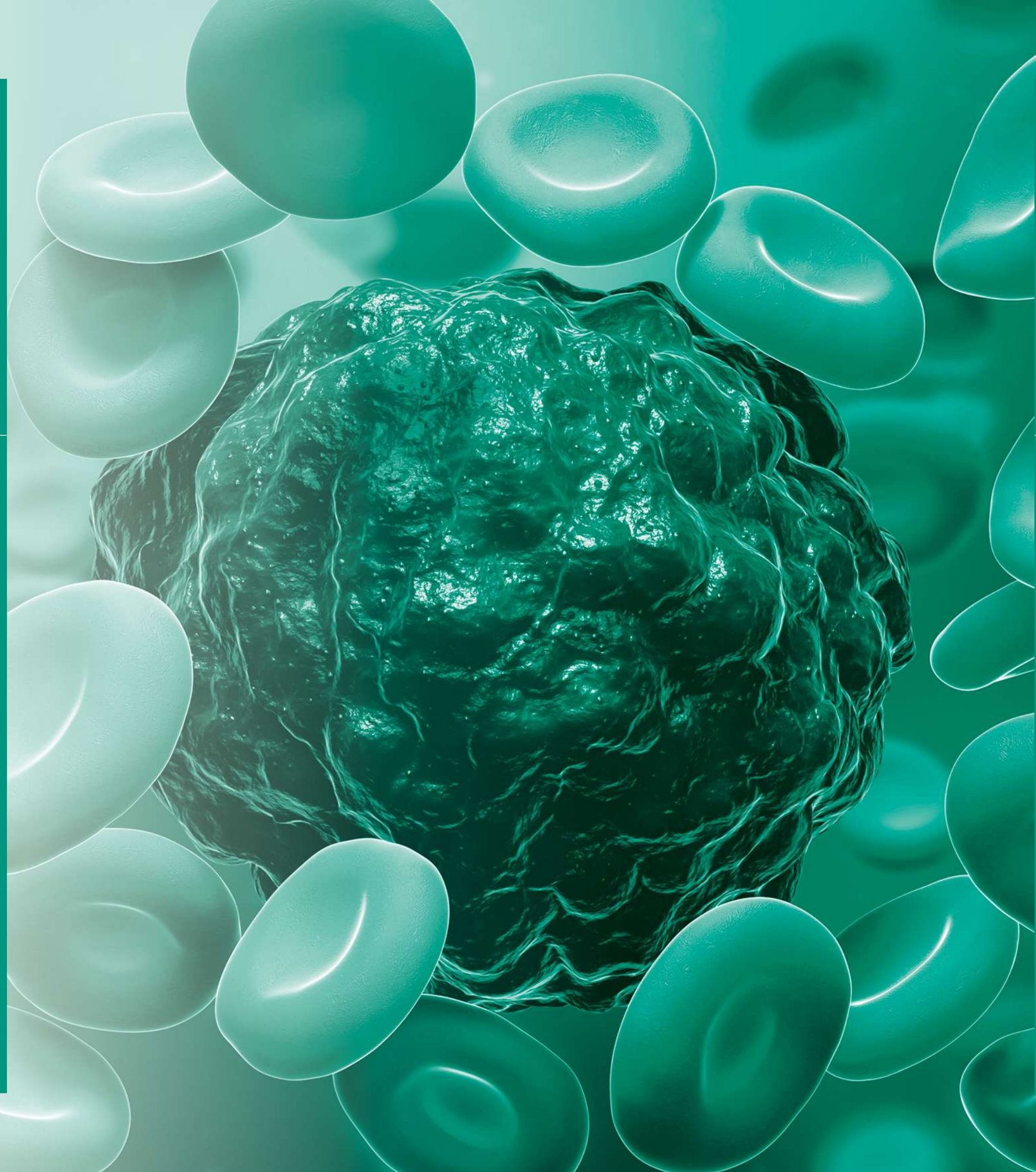
PharmacoScreen Panel Anti-tuberculosis	ABHD5	ADA	ADORA2A	ALAS1	ALPK2	ANO10	ASAH1	BACH1	BAX	BCL2	BTLA	CARD8	CASP1
	CASP3	CASP8	CASP9	CAT	CCL2	CD274	CD276	CD28	CD40	CD40LG	CD80	CD86	CPA6
	CTLA4	CYBA	DDX10	DPP4	ENTPD1	FAHD2A	FAS	FASLG	FBXW8	FOXP3	GCLC	GCLM	GGT1
	GPX1	GPX3	GPX4	GSR	GSS	GSTA1	GSTA2	GSTA3	GSTA4	GSTA5	GSTK1	GSTM2	GSTM3
	GSTM4	GSTM5	GSTO1	GSTO2	GSTT2	GSTZ1	HAVCR2	HIF1A	HMOX1	HMOX2	HSPA1L	ICOS	ICOSLG
	IDO1	IDO2	IFNG	IFNGR1	IFNGR2	IL10	IL10RA	IL12A	IL12B	IL12RB1	IL12RB2	IL17A	IL17RA
	IL18	IL18R1	IL18RAP	IL1A	IL1B	IL1R1	IL4	IL4R	IL6	IL6R	KCNE3	KCNIP3	KEAP1
	KSR2	LAG3	LGALS9	MAFK	MIR4272	MPO	NFE2L2	NLRP3	NOS1	NOS2	NOS3	NT5E	PDCD1
	PDCD1LG2	PLXNA4	POLD3	PROM2	PSD3	SOD1	SOD3	SRXN1	STAT3	TGFB1	TGFBR1	THSD7B	TNFRSF4
	TNF	TNFAIP3	TNFRSF14	TNFRSF1A	TNFRSF1B	TNFRSF9	TNFSF10	TNFSF14	TNFSF4	TNFSF9	TRIM43	TXNRD1	USP44
	VTCN1	ZNF804B											

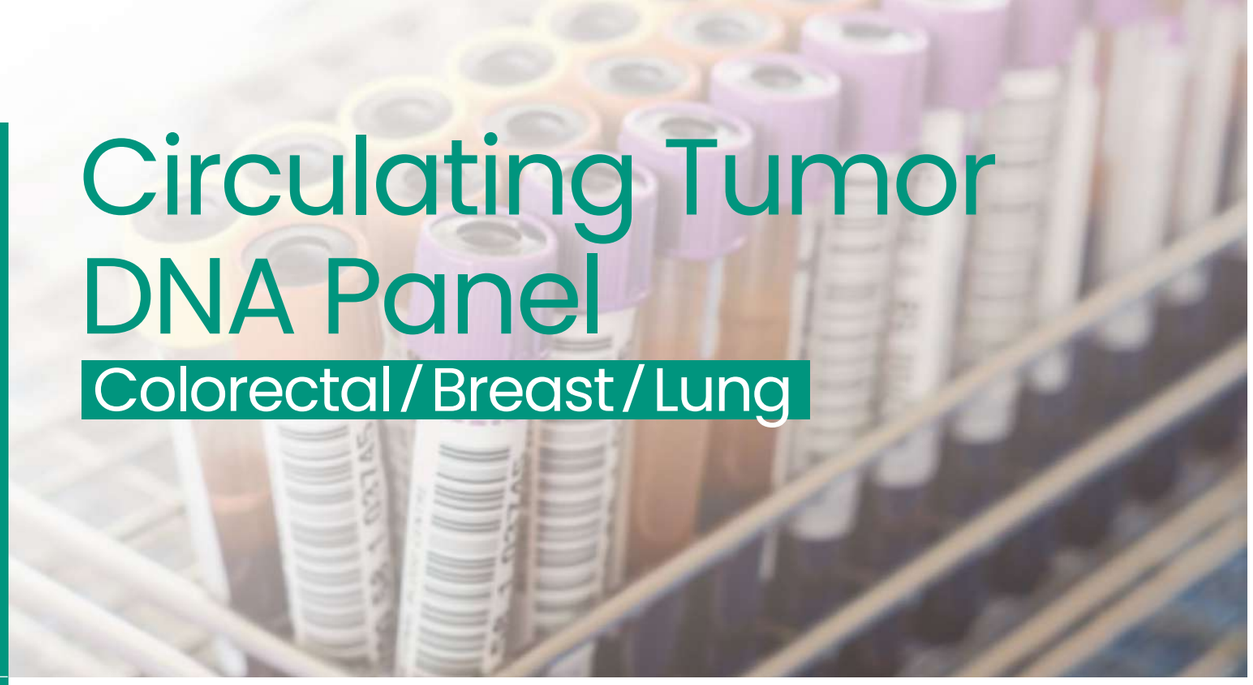
READY-TO-USE PANELS FOR LIQUID BIOPSY

CELEMICS PRODUCTS & SERVICES 2022

Circulating Tumor DNA Panel
: Colorectal / Breast / Lung

CELEMICS





Circulating Tumor DNA Panel

Colorectal / Breast / Lung

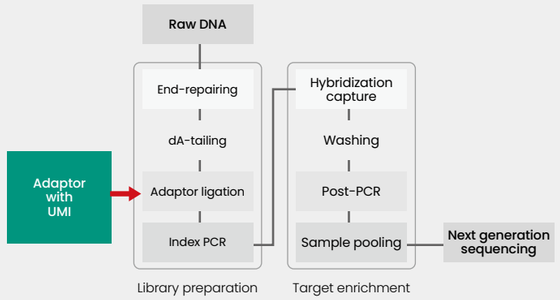
OVERVIEW

The detection sensitivity for low-frequency variants from a limited amount of sample is of great importance to ctDNA analysis kits. Celemics has developed ctDNA kits for colon, breast, and lung cancer assay through collaborative research with Seoul National University Hospital (SNUH) since 2017. We have integrated our market-leading proprietary technologies including probe design algorithms, noise removal techniques, and reagents optimization. The panels are thoroughly validated and ready to use for clinical diagnosis.

KEY FEATURES

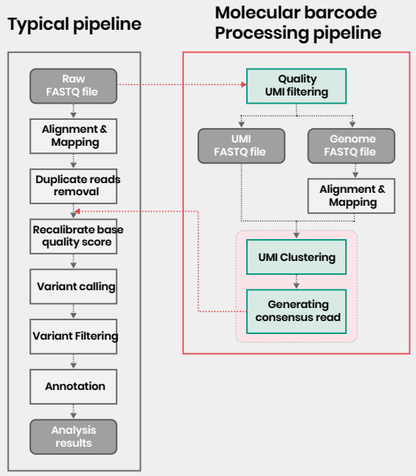
1. Detects ctDNA for colorectal cancer, breast cancer, and lung cancer	Assess 16 key genes for colorectal cancer, 27 for breast cancer, 28 for lung cancer
2. Highly optimized panel for clinical testing with exceptional accuracy	Complete validated panel performance conducted with patient samples through collaborative research with Seoul National University Hospital
3. Provides Unique Molecular Identifiers (UMI) and Bioinformatics Software	Receive high-quality data supported by Celemics proprietary UMI algorithms and analysis software, enabling efficient duplication removal and minimizing sequencing noise

MODULAR UNIQUE MOLECULAR IDENTIFIER



1. Able to assess ctDNA with ultra-low variant allele frequency (VAF)
2. Retrieves more unique reads than that from conventional duplication removal algorithm, reducing sequencing costs
3. Noise removal and accurate calls possible due to proprietary consensus sequence generation algorithm
4. Modular algorithm to be applied to the existing pipeline.

MODULAR BIOINFORMATICS PIPELINE



1. Molecular Barcode Analysis Program provided to the customers using Celemics ctDNA Panels. (Linux, CLI program)
2. Streamlined application of the Molecular Barcode Analysis Program to the standard pipeline.

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

Circulating-tumor DNA Colorectal Cancer Panel

SPECIFICATION

Gene count*	15 genes
Covered region	Whole CDS
Target size	49 kb
Mutation type	SNV, Indel
Sample type (amount)	Plasma (> 20 ng of cfDNA)
Platform	All sequencers from Illumina and MGI
Bioinformatics pipeline	1. Primary and Secondary analysis result (FASTQ to VCF) 2. Tertiary analysis result (VCF to Clinical report) 3. Linux-based consensus read generation software provided

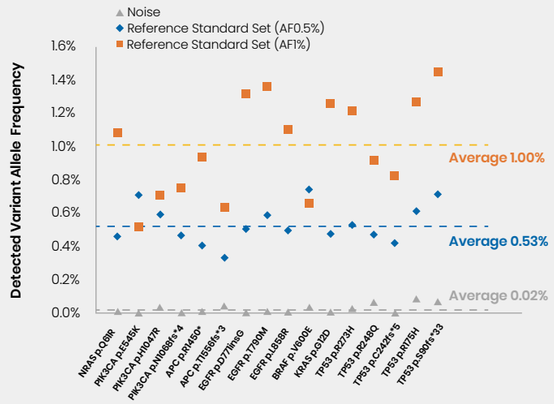
* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

ctDNA Panel Colorectal Cancer													
	APC	ATM	BRAF	EGFR	ERBB2	FBXW7	KRAS	MET	NIRAS	PDGFRA	PIK3CA	PTEN	SMAD4
	TCF7L2	TP53											

PANEL PERFORMANCE

Detection of 16 variants with 100% sensitivity and 97.9% specificity at 0.5% VAF and 1% VAF



Sensitivity, VAF 0.5%	100%
Sensitivity, VAF 1.0%	100%
Specificity	97.9%

Circulating-tumor DNA Breast Cancer Panel

SPECIFICATION

Gene count*	27 genes
Covered region	Whole CDS
Target size	99 kb
Mutation type	SNV, Indel
Sample type (amount)	Plasma (> 20 ng of cfDNA)
Platform	All sequencers from Illumina and MGI
Bioinformatics pipeline	1. Primary and Secondary analysis result (FASTQ to VCF) 2. Tertiary analysis result (VCF to Clinical report) 3. Linux-based consensus read generation software provided

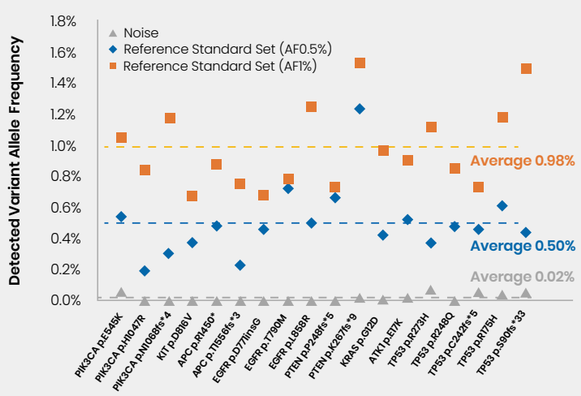
* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

ctDNA Panel Breast Cancer													
	AKT1	APC	AR	BRCA1	BRCA2	CCND1	CDH1	EGFR	ERBB2	ESR1	FGFR1	FGFR2	GATA3
	IGF1R	KIT	KRAS	MAP2K4	MAP3K1	MDM2	MYC	NF1	PIK3CA	PIK3R1	PTEN	RB1	TOP2A
	TP53												

PANEL PERFORMANCE

Detection of 27 variants with 96.3% specificity and 94.4% sensitivity at 0.5% VAF and 100% at 1% VAF



Sensitivity, VAF 0.5%	94.4%
Sensitivity, VAF 1.0%	100%
Specificity	96.3%

Circulating-tumor DNA Lung Cancer Panel

SPECIFICATION

Gene count*	28 genes
Covered region	Whole CDS for 8 genes and Hotspot exonic region for 20 genes
Target size	47 kb
Mutation type	SNV, Indel
Sample type (amount)	Plasma (> 20 ng of cfDNA)
Platform	All sequencers from Illumina and MGI
Bioinformatics pipeline	1. Primary and Secondary analysis result (FASTQ to VCF) 2. Tertiary analysis result (VCF to Clinical report) 3. Linux-based consensus read generation software provided

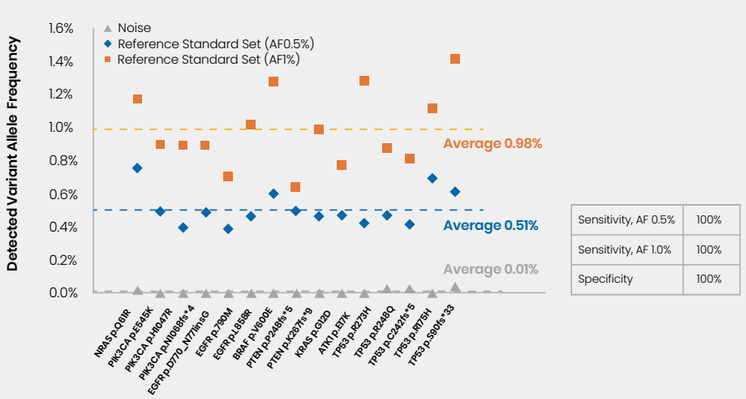
* Gene Add-On Service: Genes can be added by customer's request

GENE LIST

ctDNA Panel Lung Cancer	AKT1	ALK	ARAF	ARID1A	BRAF	CBL	CDKN2A	EGFR	ERBB2	HRAS	KEAP1	KRAS	MAP2K1
	MET	MTOR	NF1	NRAS	NTRK1	NTRK2	PIK3CA	PTEN	RB1	RIT1	ROS1	SETD2	STK11
	TP53	U2AF1											

PANEL PERFORMANCE

Detection of 28 variants with 100% sensitivity and 100% specificity at 0.5% VAF and 1% VAF detection



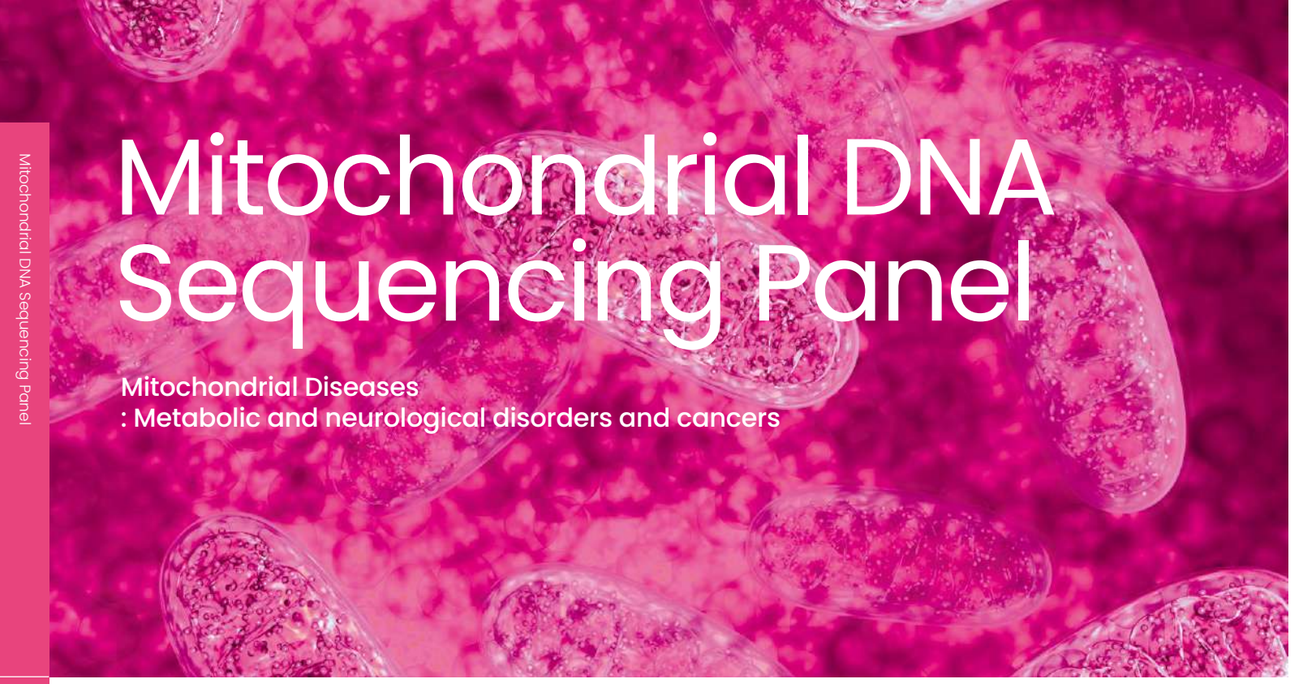
Sensitivity, AF 0.5%	100%
Sensitivity, AF 1.0%	100%
Specificity	100%

READY-TO-USE PANELS FOR MITOCHONDRIAL DNA

CELEMICS PRODUCTS & SERVICES 2022

Mitochondrial DNA Sequencing Panel

CELEMICS



Mitochondrial DNA Sequencing Panel

Mitochondrial Diseases
: Metabolic and neurological disorders and cancers

DESCRIPTION

Celemics has specifically designed capture probes and adjusted the concentration of the panel for each respective use with our own proprietary rebalancing technologies to provide complete, consistent coverage of the whole mitochondrial genome while taking into consideration small target regions. This enables the same high level of target capture efficiency regardless of small target sizes even with a stand-alone panel.

KEY FEATURES

1. High-fidelity sequencing	Guarantees maximum capture efficiency in custom panels without affecting target specificity
2. Highly uniform coverage and mean depth	High coverage and uniformity across the entire human mitochondrial genome
3. Flexible customization	Convenient addition to other Celemics target enrichment panels such as G-Mendeliome panels for further mtDNA-derived rare disease analysis

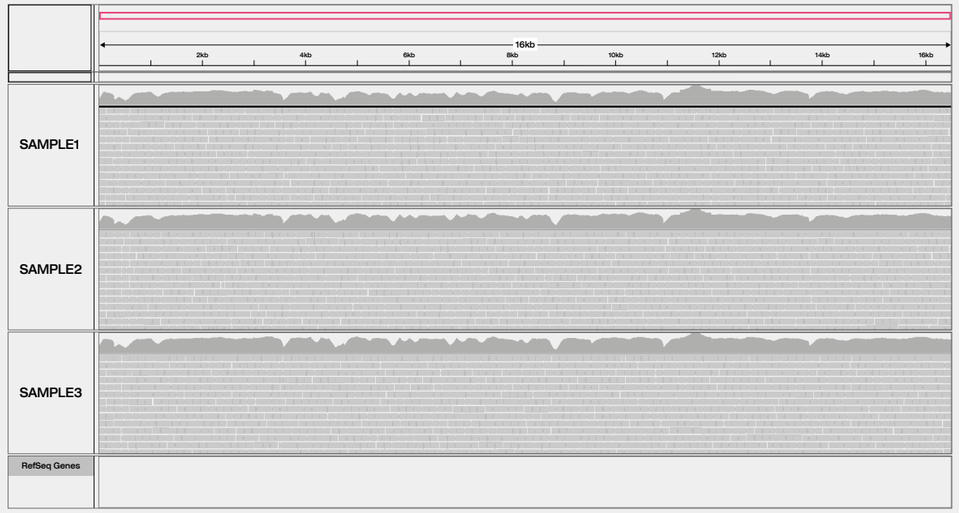
SPECIFICATION

Covered region*	Whole mitochondrial genome
Target size	16.6 kb
Mutation type	SNV, Indel
Sample type (amount)	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

PERFORMANCE

NGS Sequencing Amount	On-Target Base Ratio	Mean Depth	Coverage		
			10x	50x	100x
10Mb	97.93%	493x	99.98%	99.91%	99.87%

IGV EXAMPLE OF CELEMICS mtDNA SEQUENCING PANEL



Celemics mtDNA Sequencing Panel shows 99% coverage with uniformity

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

TARGET ENRICHMENT KITS FOR RNA SEQUENCING

CELEMICS PRODUCTS & SERVICES 2022

Targeted RNA Sequencing Panel

CELEMICS

Targeted RNA Sequencing Panel

Transcriptome Analysis

KEY FEATURES

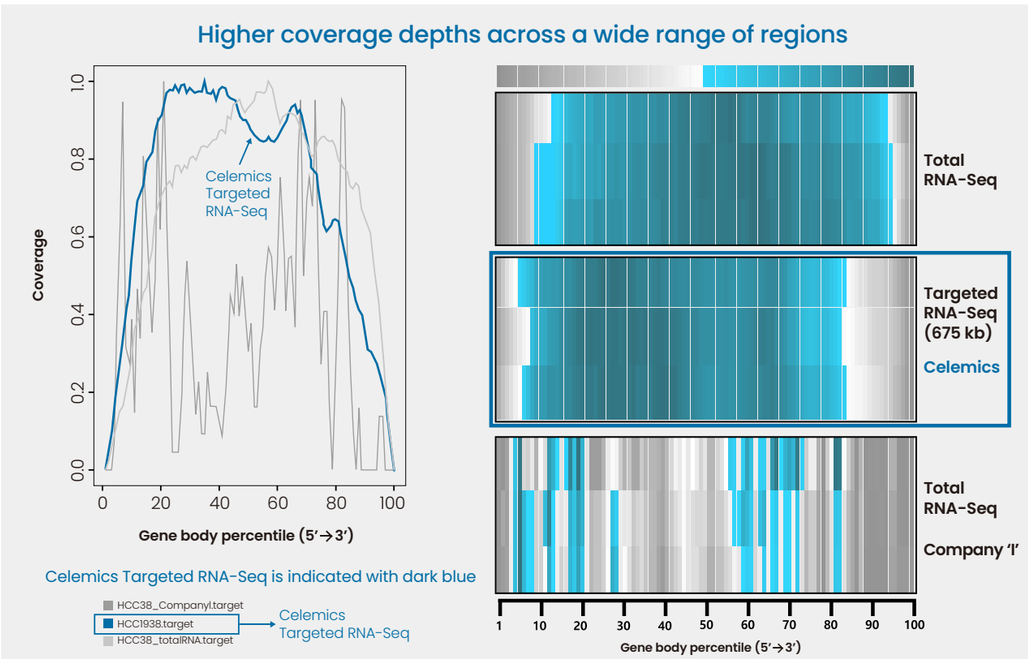
1. Cost-effective high-quality analysis	Accurate analysis of expression levels enabled by higher depth of coverage due to specific targeting of genes of interest, compared to total RNA sequencing
2. Compatible with a variety of sample types	Receive reliable results from poor-quality samples such as FFPE and low-amount samples such as cfRNA
3. Expression level in all regions of genes of interest	Covers all gene regions, allowing for the assessment of expression levels across all exons
4. Gene rearrangement analysis	Detects rearrangement and all other types of variants
5. Isoform analysis	Identify isoform expression levels by assessing the entire regions of targeted genes.

PACKAGE COMPOSITION

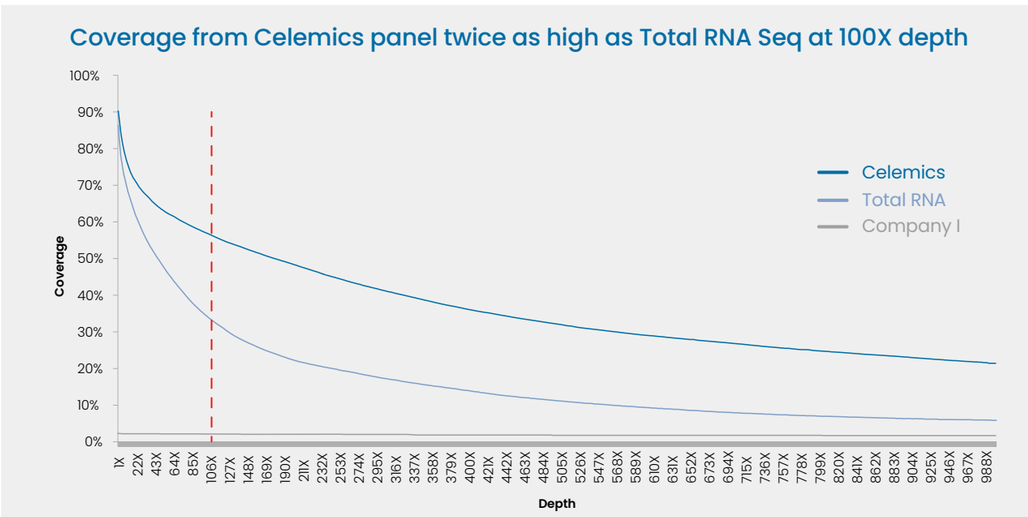
Package name	Compositions		
Target Enrichment	Target capture Probe	-	
Standard	Target Enrichment reagents	Library prep Kit	-
All-In-One			Beads / Polymerase

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

PANEL PERFORMANCE



Celemics Targeted RNA Sequencing assesses the expression level of selective genes with sufficient level of coverage depth that is higher than that of total mRNA sequencing. Compared to competitor products that targets only parts of an exon, the Targeted RNA Sequencing developed by Celemics showed relatively higher coverage across a wide range of regions.



The comparison test between Celemics Targeted RNA Sequencing and total RNA sequencing shows that the coverage from the Celemics product is 15% higher at 50X and twice as high at 100X.

TARGET ENRICHMENT KITS FOR EPIGENETICS

CELEMICS PRODUCTS & SERVICES 2022

Targeted Methylation Sequencing Panel

CELEMICS

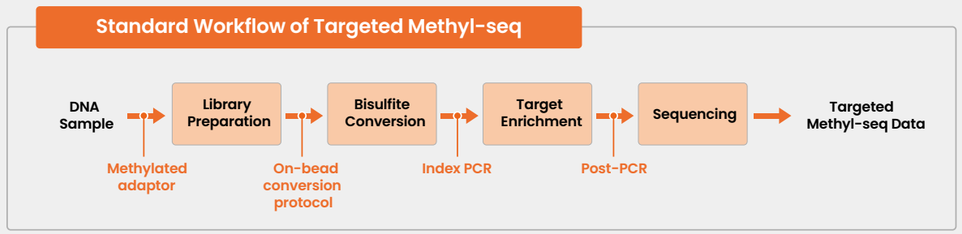
Targeted Methylation Sequencing Panel

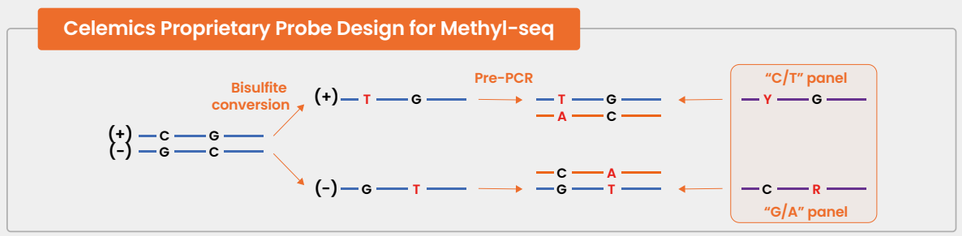
Epigenetics

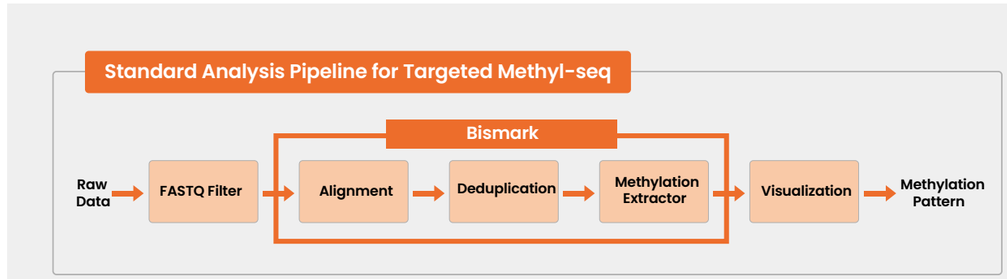
KEY FEATURES

1. Probe specifically designed for Methyl-seq	Elaborate design considering the sequence alteration by bisulfite conversion Thorough comparison analysis of the sequences before and after bisulfite conversion, enabling accurate detection of methylation sites
2. Compatible with all sample types	Perform methylation analysis with gDNA and cfDNA

PANEL PERFORMANCE

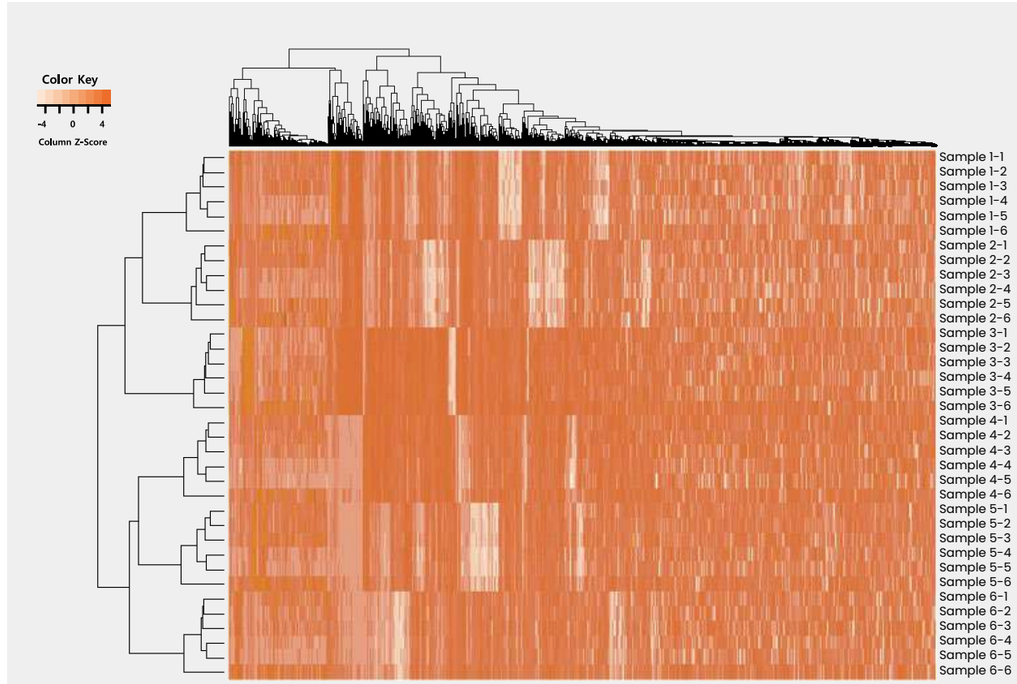






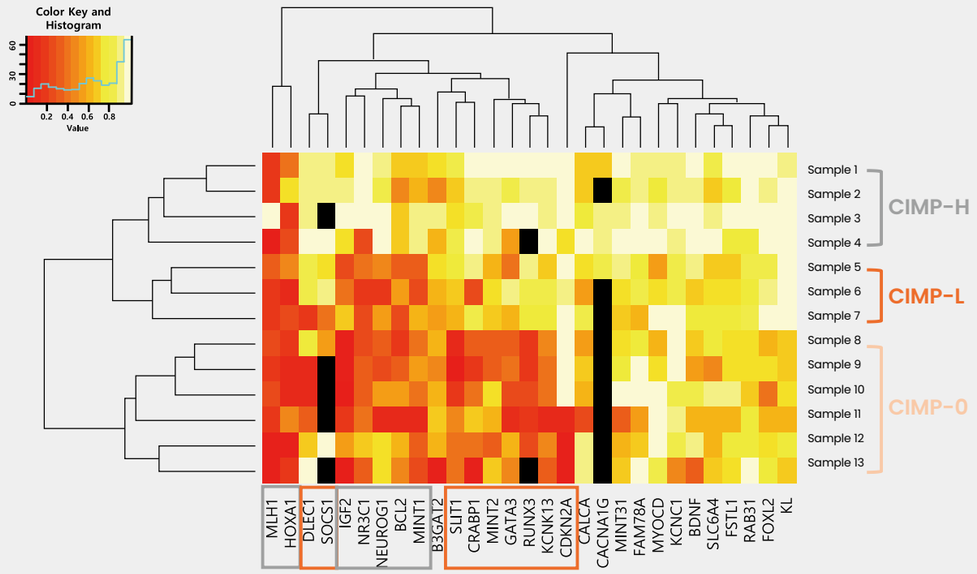
The Targeted Methylation Sequencing is proceeded with including a bisulfite conversion process in the NGS workflow.
The hybridization probe and methylated adapters are designed considering the sequence alteration by bisulfite conversion, enabling an accurate comparison analysis of the sequences before and after the conversion.
Selective genes are targeted for the analysis, allowing for cost-effective sequencing.

HIGH REPRODUCIBILITY OF METHYLATION PATTERN ANALYSIS



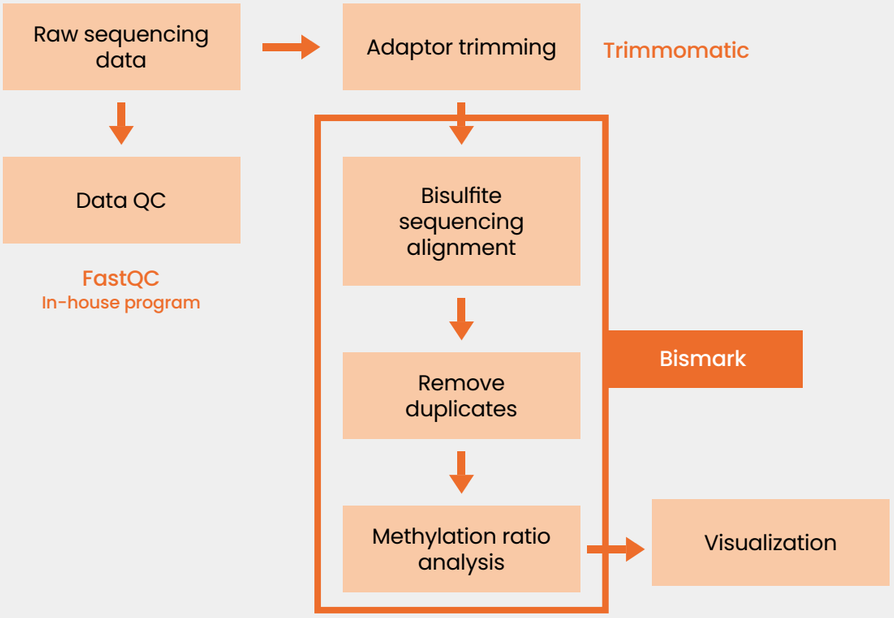
The results demonstrate high reproducibility of the analysis, yielding the same methylation patterns when repeatedly tested with the identical specimens.

HIGH CONCORDANCE OF METHYLATION PATTERN ANALYSIS WITH CLINICAL INFORMATION



The clustering result from pattern analysis showed high concordance with the clinical data information.

WORKFLOW OF TARGETED METHYLATION SEQUENCING ANALYSIS

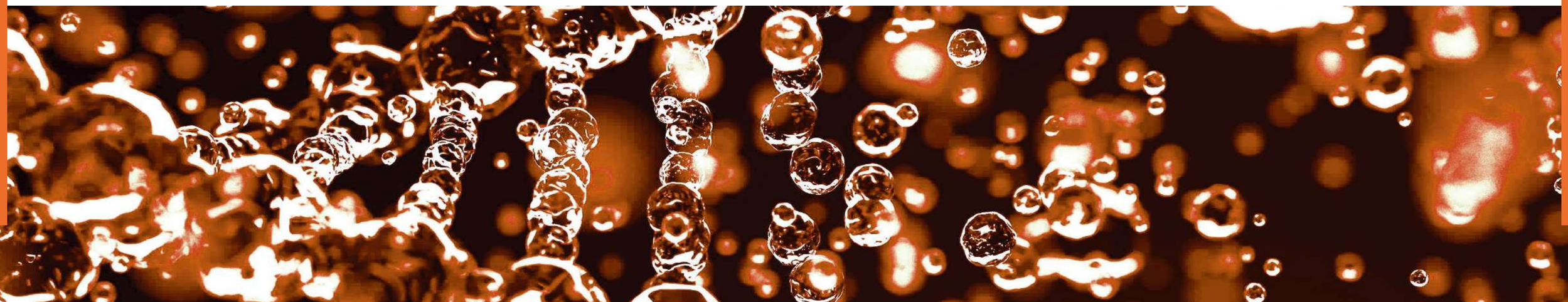


Customers who are new to methylation analysis are supported by Celemics bioinformatics software service for fast and accurate analysis.

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

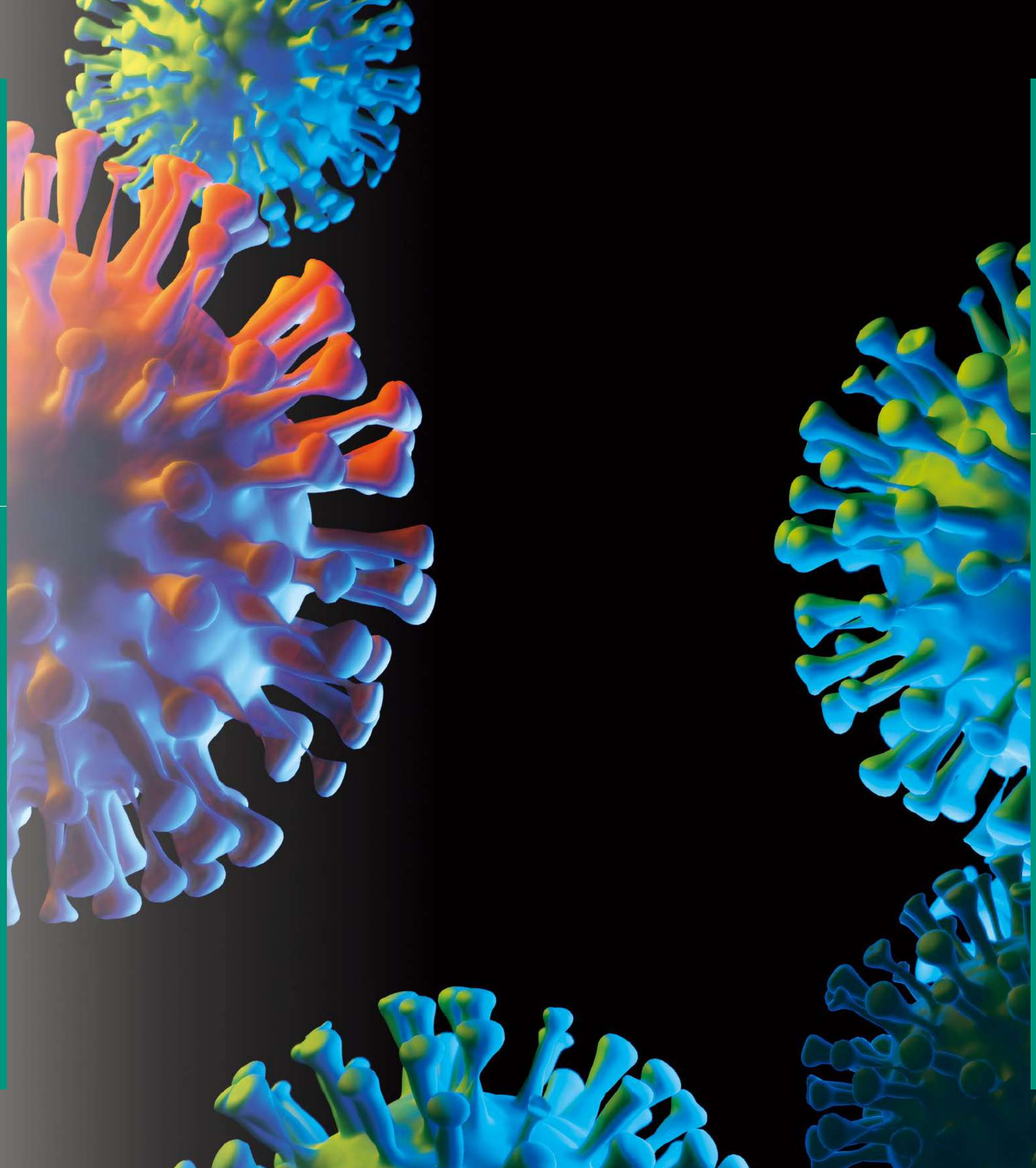


TARGET ENRICHMENT KITS FOR VIRUS RESEARCH

CELEMICS PRODUCTS & SERVICES 2022

Comprehensive Respiratory Virus Panel
African Swine Fever Virus Panel

CELEMICS



Comprehensive Respiratory Virus Panel

Research and Diagnosis of Respiratory viruses

DESCRIPTION

The CRV Panel is designed for the comprehensive analysis of clinically significant respiratory viruses that are widely assessed by medical institutions around the globe. The panel validation test with clinical samples showed superior whole genome sequencing (WGS) success rates compared to other competitor kits on the market. The panel tests for multiple infections by assessing all types of respiratory viruses including SARS-CoV-2. The panel includes all required kits including the RNA-to-cDNA Kit and cDNA-to-Captured Library Kit. The hybridization enhancer technology is used for rapid one-day workflow. Our customers can receive stand-alone bioinformatics software, 'Celemics Virus Verifier', which provides in-depth analysis information while ensuring the security of client sequence information.

KEY FEATURES

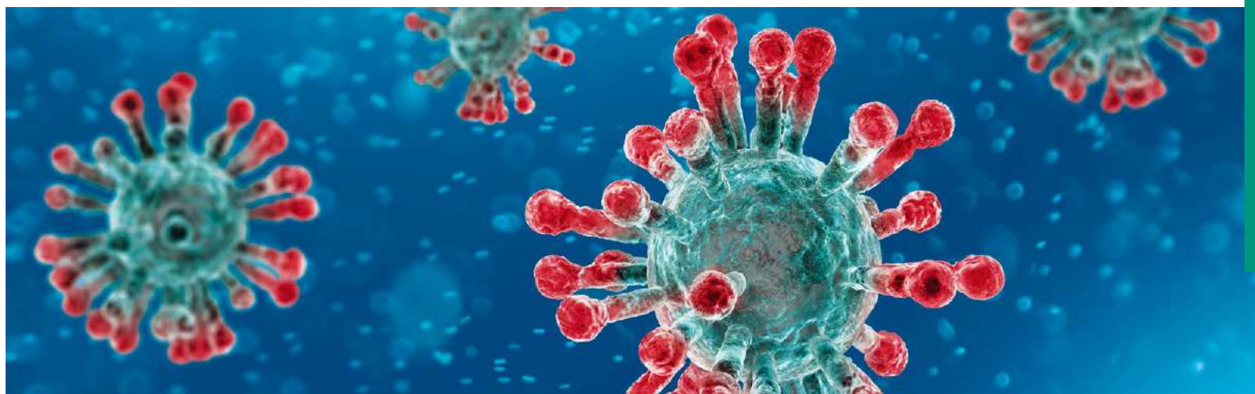
1. Coverage of wide range of respiratory pathogens	Assess WGS of 39 variants for 9 different virus types (SARS-CoV-2 solo analysis also available) Includes all types of respiratory viruses that are assessed by medical institutions around the globe
2. Superior WGS success rate even with poor quality specimen	Able to detect pathogens from patient specimens as well as poor quality environmental specimens Exceptional success rate of variant detection and WGS Significantly reduced gap formation
3. Double pandemic / coinfection detection	Detect all relevant viral strains in a single assay and test for multiple infections
4. Inclusion of Celemics Virus Verifier or bioinformatics analysis	Receive stand-alone bioinformatics SW Protect your easily-compromised data with our EU-GDPR compliant cloud system

SPECIFICATION

Target viruses*	9 types / 39 virus strains, including SARS-CoV-2
Target size	706 kb
Mutation type	Viral variants detection, Viral mutation (SNV, Indel) from generated Whole Genome Sequence
Sample type	Upper respiratory tract, Nasopharyngeal, Oropharyngeal specimens, and others
Platform	All sequencers from Illumina and Thermo Fisher
Kit composition	Provides all required reagents, including RNA to cDNA kit, cDNA to captured library kit, and bioinformatics software
Bioinformatics pipeline	Provides stand-alone bioinformatics software 'Celemics Virus Verifier' (FASTQ to Report)
Related publication	Evidence of long-distance droplet transmission of SARS-CoV-2 by direct air flow in a restaurant in Korea, J Korean Med Sci. (2020)

PATHOGEN LIST

Human Adenovirus	Coronavirus	Parainfluenza Virus	Respiratory Syncytial Virus
Human Adenovirus Type 1 (HAdV-C1)	Coronavirus HKU1	Parainfluenza 1 (PIV 1)	Respiratory Syncytial Virus A (RSV A)
Human Adenovirus Type 2 (HAdV-C2)	Coronavirus NL63	Parainfluenza 2 (PIV 2)	Respiratory Syncytial Virus B (RSV B)
Human Adenovirus Type 3 (HAdV-B3)	Coronavirus 229E	Parainfluenza 3 (PIV 3)	Human Metapneumovirus
Human Adenovirus Type 4 (HAdV-E4)	Coronavirus OC43	Parainfluenza 4 (PIV 4) A	
Human Adenovirus Type 5 (HAdV-C5)	SARS-CoV-2	Parainfluenza 4 (PIV 4) B	
Human Adenovirus 7 (HAdV-B7)			
Human Adenovirus 14 (HAdV-B14)		**Human Enterovirus**	**Human Rhinovirus (A/B/C)**
Human Adenovirus 21 (HAdV-B21)	**Influenza A**	EV-C104	Human Rhinovirus A
	Influenza A Virus (Flu A)	EV-C105	Human Rhinovirus B
Bocavirus 1/2/3/4 (HBoV)	Influenza A-H1 Virus (Flu A-H1)	EV-C109	Human Rhinovirus C
Human Bocavirus 1	Influenza A-H3 Virus (Flu A-H3)	EV-C117	
Human Bocavirus 2		EV-C118	
Human Bocavirus 3	**Influenza B**	CV-A21	
Human Bocavirus 4	Influenza B Virus (Flu B)	EV-D68	

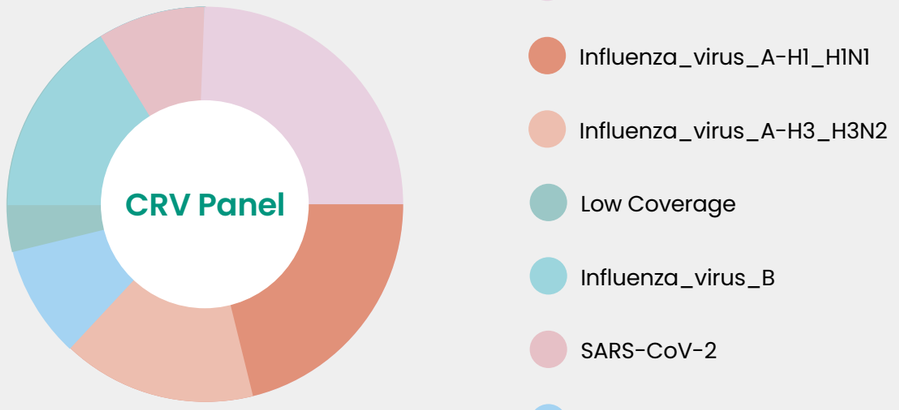


PERFORMANCE

High coverage of whole genome from reference samples using CRV Panel

Sample Type	Coverage [1X]	Coverage [10X]	Coverage [100X]
Reference sample (Illumina 2x75 bp)	99.95%	99.87%	98.95%

Example of viral strain identification in reference sample



CRV PANEL RESULTS GENERATED THROUGH CELEMICS VIRUS VERIFIER (STAND-ALONE SOFTWARE)

Celemics provides stand-alone software for bioinformatics analysis, allowing customers to access the detailed data analysis information and ensuring the security of client sequence information.

African Swine Fever Virus Panel

Virus Research, Virus WGS Analysis

DESCRIPTION

The high morbidity and mortality of African swine fever (ASF) has a severe impact on the global swine industry. However, currently there is no effective treatments or vaccines commercially available. The ASFV panel is designed to identify 26 strains of genotype II virus in a single NGS run. The panel can be utilized for identifying the cause and infection route.

KEY FEATURES

1. Swine-specific blocking reagent	Provides swine-specific blocking reagent that effectively blocks repetitive sequences and allows for selectively retrieving the ASFV sequence
2. Comprehensive analysis of ASFV subtypes	Detect genotype II virus subtypes with specifically designed probes
3. Convenient testing	Highly accurate results from blood samples, often considered more challenging due to the lower viral load compared to concentrated culture supernatant or spleen tissue sample

SPECIFICATION

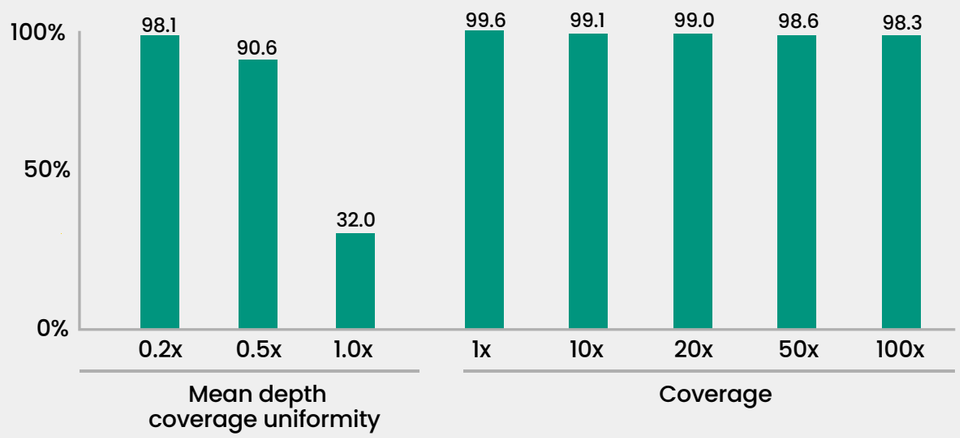
Target viruses*	ASFV 26 strains
Target size	192 kb
Mutation type	Virus detection, Virus genome assembly
Sample type (amount)	Swine blood (50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Celemics ASFV Pipeline (FASTQ to Result)

* Gene Add-On Service: Genes can be added by customer's request

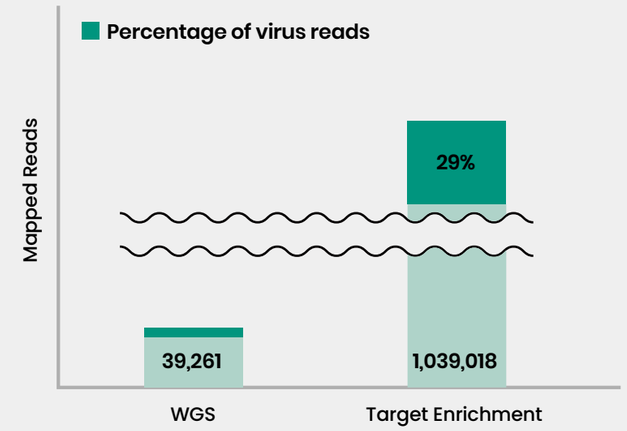
ASFV DETECTION WORKFLOW

PERFORMANCE

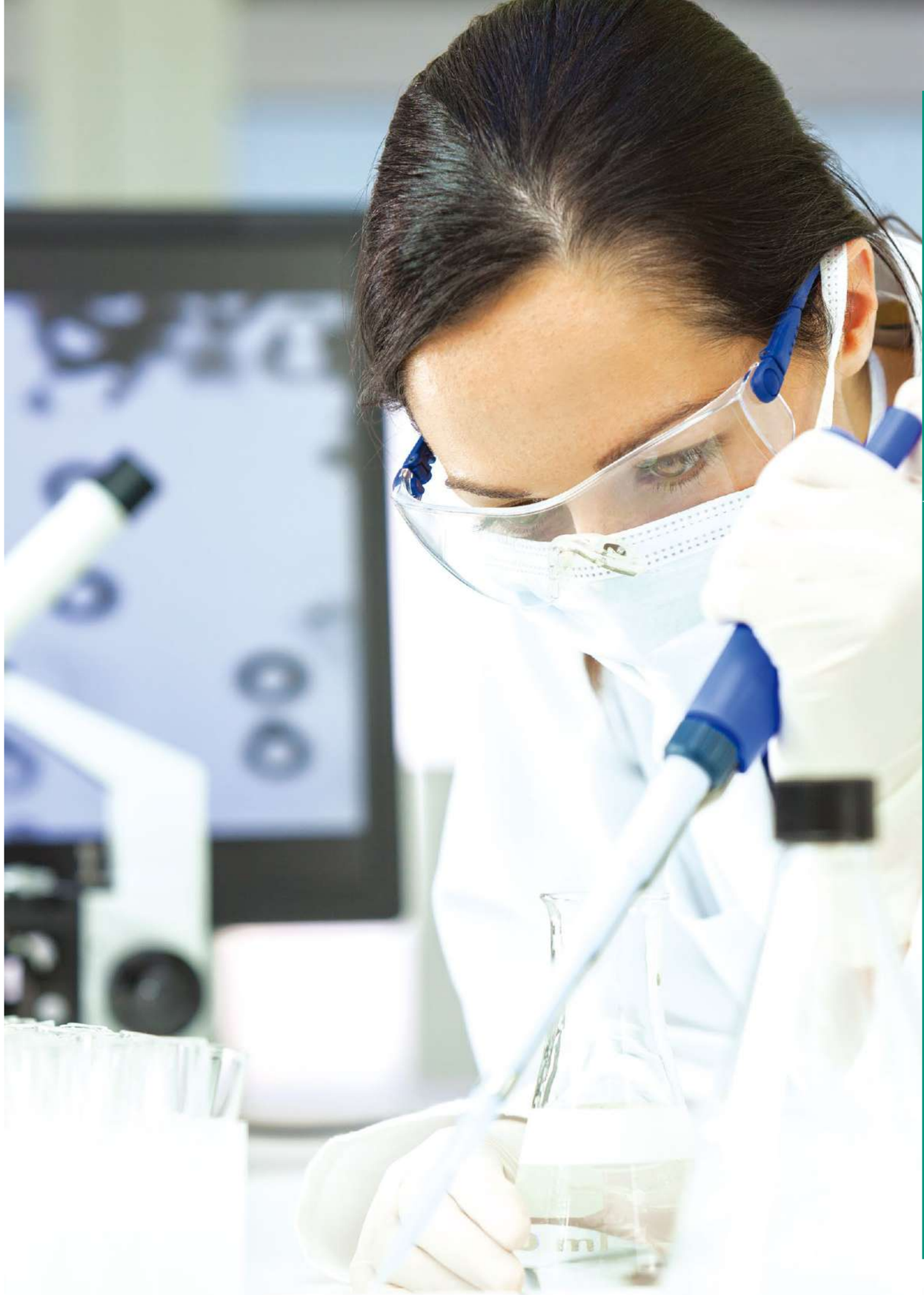
Advanced target enrichment technology enabling exceptional capture performance with high coverage and uniformity



The panel validation result shows high uniformity and high coverage at all levels.



With the same sequencing amount, target enrichment NGS yielded 29% virus reads out of a total of 1,039,018 reads, while whole genome sequencing (WGS) yielded 0.5% virus reads (green) out of a total of 39,261 reads.

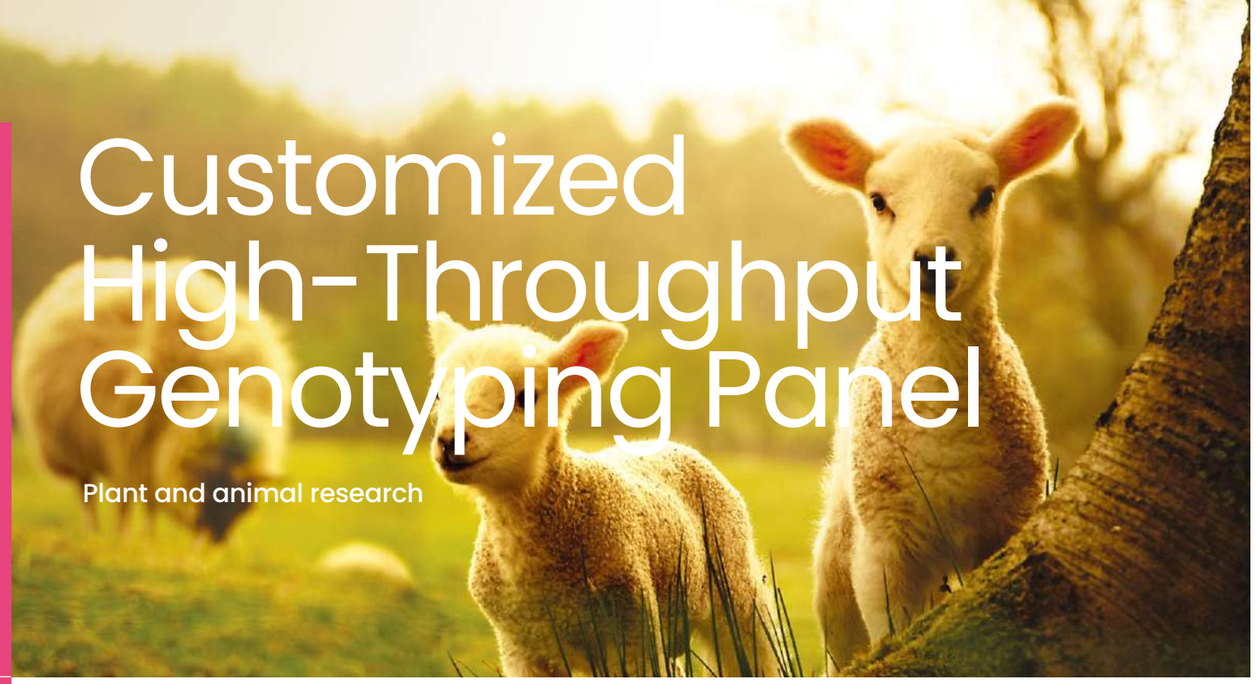


TARGET ENRICHMENT KITS FOR AGRICULTURE & ANIMAL RESEARCH

CELEMICS PRODUCTS & SERVICES 2022

Customized High-Throughput Genotyping Panel

CELEMICS



Customized High-Throughput Genotyping Panel

Plant and animal research

DESCRIPTION

For molecular breeding, the availability and easy accessibility of genomic resources is a prerequisite. Although technological advances have provided a range of resources like molecular markers, genetic linkage maps, whole genome sequences and transcriptomes, agricultural genomics has faced many challenges. Celemics provides a solution with the High-Throughput Genotyping Panel. We have utilized NGS methods, whereby a high number of regions of interest are simultaneously enriched using specifically designed probes to provide new insights into different agricultural genomics research.

KEY FEATURES

1. NGS-based target enrichment sequencing assay	Utilize NGS-based target enrichment methods for higher accuracy and cost-effectiveness compared to conventional methods such as conventional GBS, PCR, and microarray
2. Comprehensive analysis with high accuracy	Perform comprehensive assay of 100 to 10,000 markers with minimized false-negatives and false-positives Discover novel SNPs
3. Cost-effective analysis	Benefit from Celemics' library preparation kits, target capture technology, and multiplexing indices specifically designed for high-throughput genotyping
4. Outstanding performance regardless of various origins	Receive high-quality results enabled by species-specifically designed blocking oligos across all types of origins

PACKAGE COMPOSITION

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

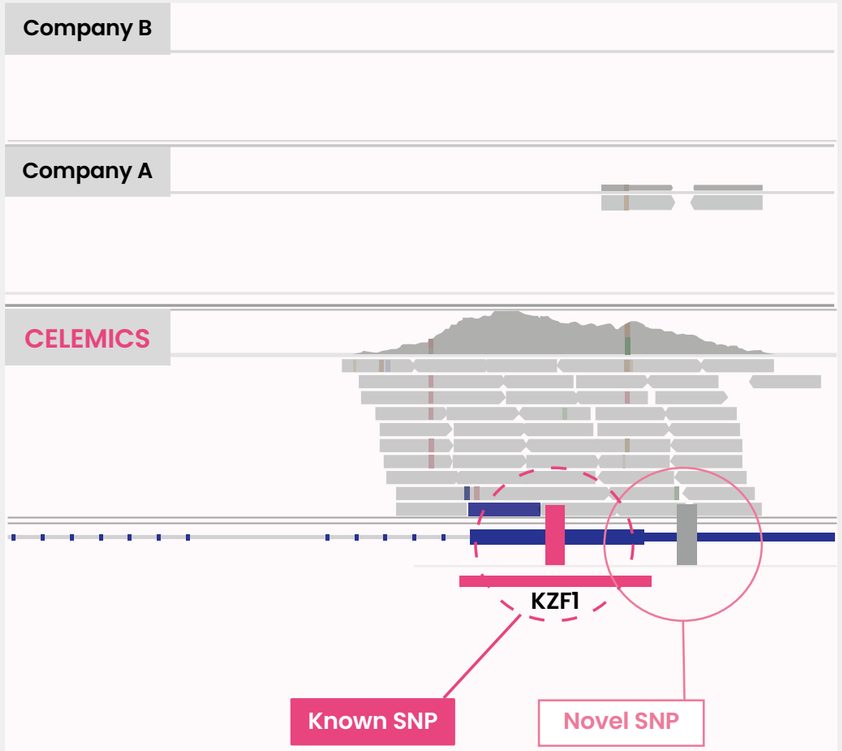
Package option	Options	
Pooling method	Single Reaction	Pre-capture Pooling
Library Preparation kits	Standard Kit	EP-kit
Hybridization Enhancer	Included	Not included

COMPARISON WITH CONVENTIONAL TECHNOLOGIES

	Advantages	Disadvantages
Conventional GBS	1. Sequencing of multiple samples due to lower amount of data required compared to WGS	1. Limited biomarkers available due to limited conserved regions, reducing overall resolution 2. Unable to detect SNPs in the restriction sites
Microarray	1. Higher reproducibility than conventional GBS	1. Hard to customize new targets (novel biomarkers) 2. Low flexibility to meet various kinds of genotyping
PCR	1. Cost-effective for low number of samples 2. Easy and fast analysis	1. Limited number of biomarkers to analyze at once 2. Inappropriate for mass-analysis of biomarkers
Celemics Target Enrichment	**1. Cost saving** : Highly cost-effective when assessing multiple samples **2. Flexible customization** : Novel biomarkers can be added or removed **3. Comprehensive analysis** : Including novel SNP discovery **4. Exceptional performance** : Celemics proprietary blocking oligo design technology **5. Wide compatibility** : Compatible with a wide range of sample types	

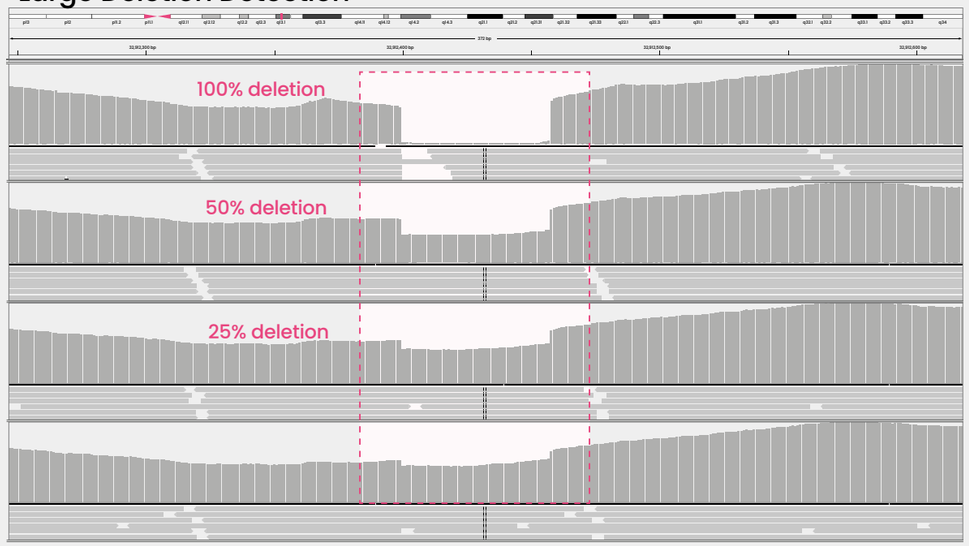
PERFORMANCE

Hybridization-based NGS target enrichment enables discovery of novel SNPs near target regions



PERFORMANCE

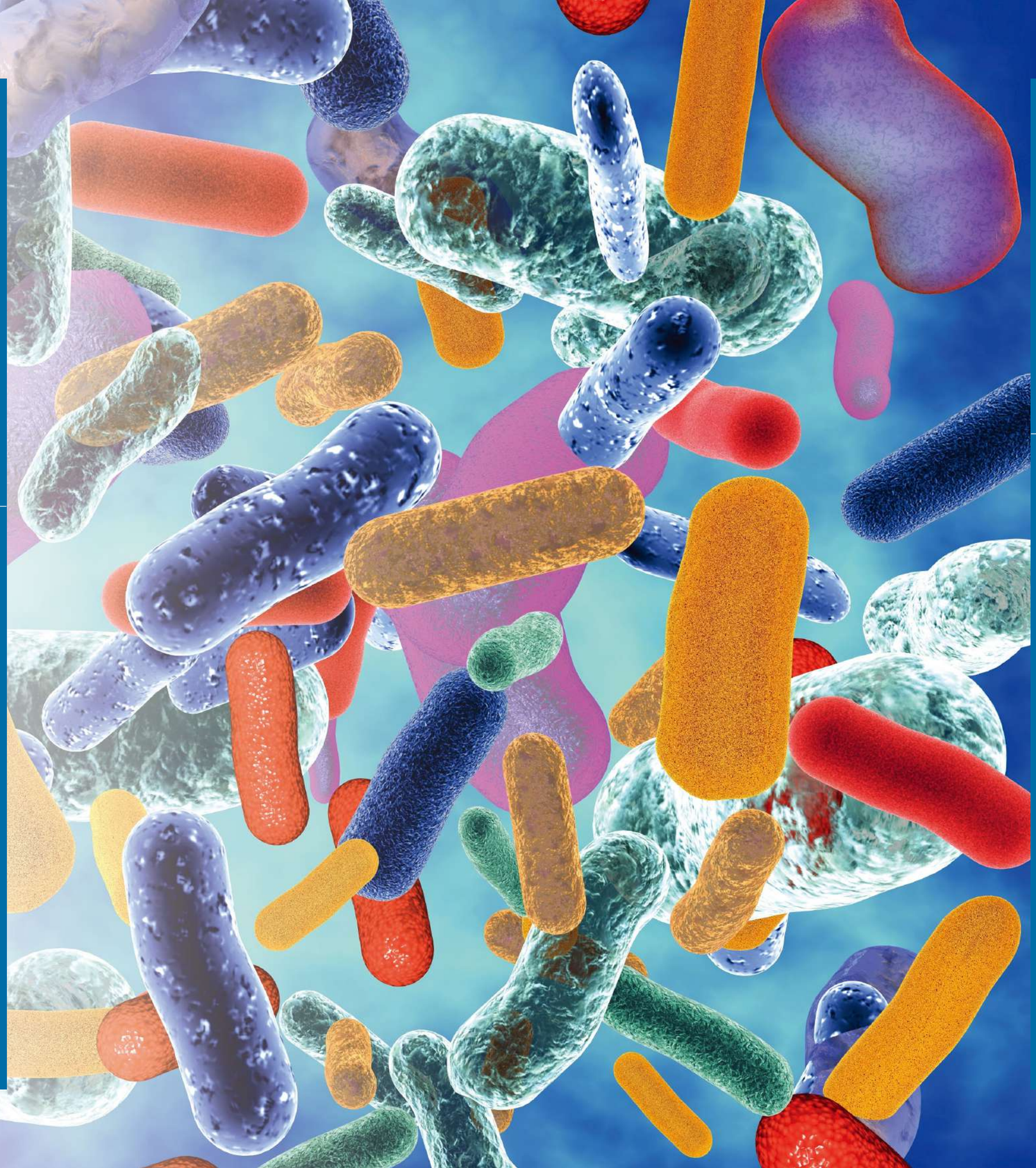
Hybridization-based NGS target enrichment enables accurate analysis of all mutation types including large deletion and rearrangement.



**Artificially generated data used*

CELEMICS SOLUTIONS FOR METAGENOMIC SEQUENCING

CELEMICS PRODUCTS & SERVICES 2022



Metagenomic Sequencing Service and Kit

CELEMICS

Metagenomic Sequencing Service and Kit

16S V4 / 16S V3-V4 / 18S ITS1 / 18S ITS1-ITS2

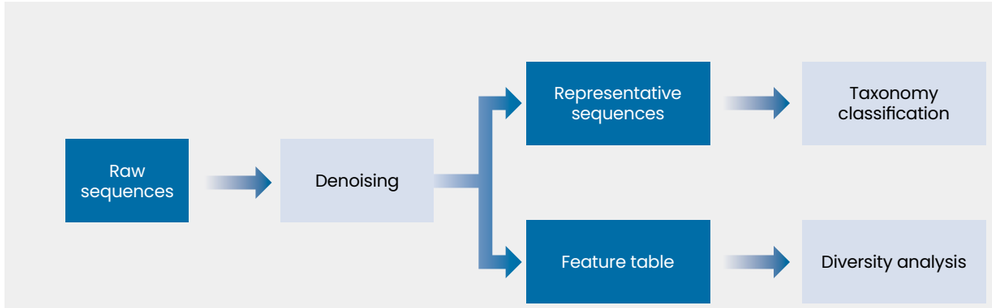
DESCRIPTION

Metagenomic Sequencing Service and Kit is used for microbiome and mycobiome studies. The service allows for characterizing and differentiating a myriad of microbial species. The 16S V4 (or V3-V4) region of bacteria and archaea and 18S ITS1 (or ITS1-ITS2) region of fungi is amplified by PCR. After cleaning up using CeleMag beads, the indices and adapters are attached for NGS and bioinformatics analysis. According to the purpose of customer's studies, various analysis reports are provided by the Celemics robust analysis pipeline. Please contact us for further information.

EXPERIMENT WORKFLOW

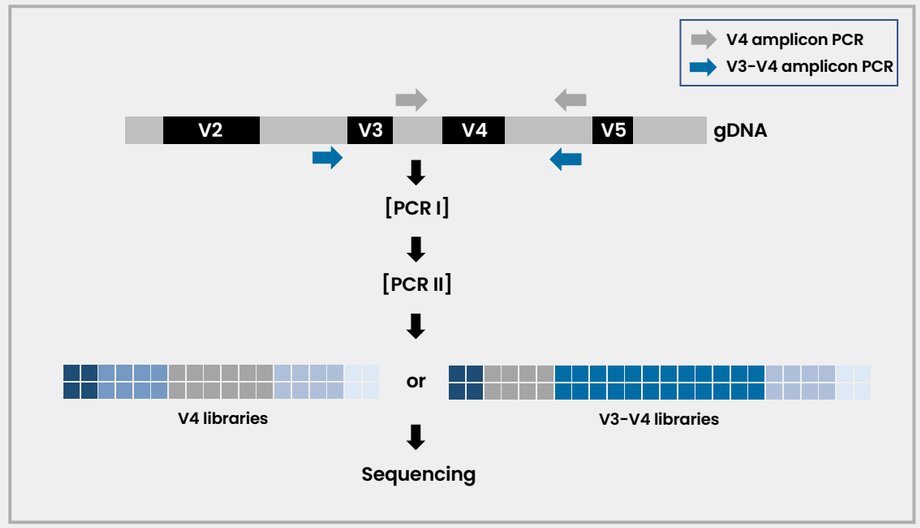
1. PCR amplification against gDNA using 16S region or ITS region specific primers
2. Bead cleanup
3. Index and adapter ligation with Nextera Index sets
4. Bead cleanup
5. Library pooling
6. NGS Sequencing

NGS-BASED METAGENOME ANALYSIS WORKFLOW

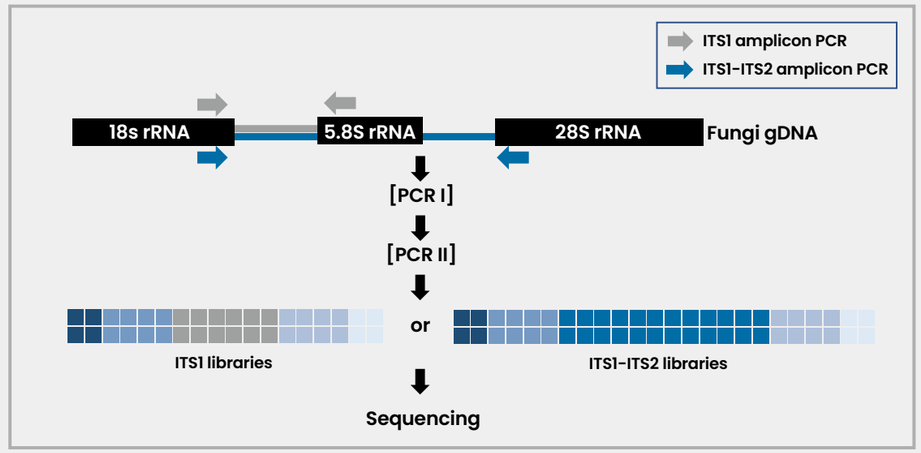


SEQUENCING WORKFLOW

16S rRNA V4 and V3-V4

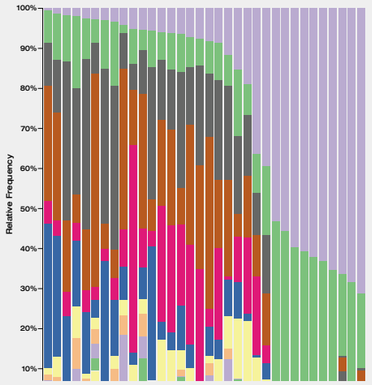


18S ITS1 and 18S ITS1-ITS2

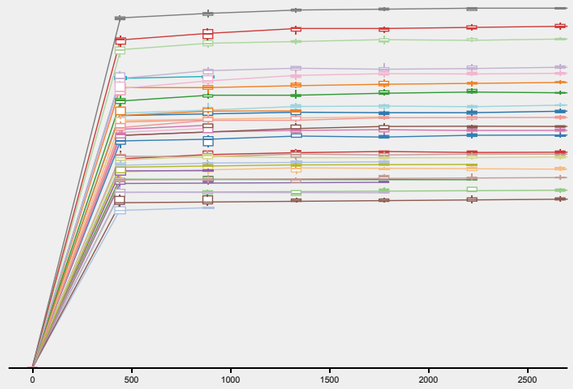


EXAMPLE OF METAGENOMIC SEQUENCING ANALYSIS REPORT

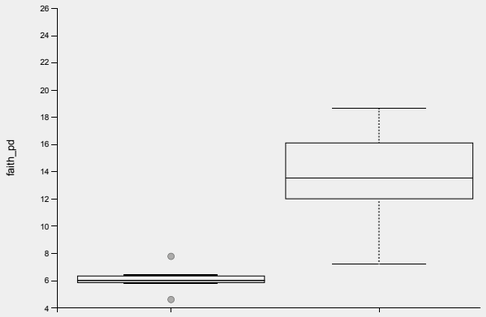
Taxonomy analysis



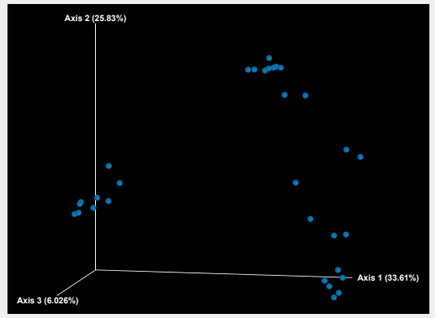
Alpha rarefaction



Alpha diversity



Beta diversity



Results presented above are a few selected examples of the metagenomics sequencing results that Celemics provides. Contact us for more information.

Barcode Tagged Sequencing™ (BTSeq™)

CELEMICS PRODUCTS & SERVICES 2022

BTSeq™ – Standard Service and Kit
BTSeq™ – Viral Analysis Service
BTSeq™ Mitochondrial DNA Sequencing Service
BTSeq™ Full Plasmid Sequencing Service

CELEMICS

Barcode-Tagged Sequencing™ (BTSeq™)

Wide Range of DNA Sizes

- No limitation of DNA size: 200 bp - 20 kb or longer
- Plasmid sequencing with large insert DNA

NGS-based, High Sequencing Accuracy

- NGS-based high sequencing quality
- Digitized sequencing results

Fast TAT, No Need for Primer Walking

- NGS-based result, within 24 hours after sample arrival
- No need for primer synthesis
- No need of repetitive Sanger sequencing cycles

Cost-effective

- Unparalleled cost-effectiveness compared to Sanger
- Only sequencing primer information required, eliminating the need for synthesizing the primers

No Limitation of Origin

- Sequencing samples of various species
- Virus, Bacteriophage, Mycobiome, etc.

No Need of High Concentration Sample

- Compatible with unpurified PCR products
- Low-amount sample requirements as little as 10 ng/μl

BTSeq™ SERVICE

- BTSeq™ – Viral Analysis Service
- Mitochondrial DNA Sequencing Service
- Full Plasmid Sequencing Service
- Microbial Identification Service

BTSeq™ – Standard Service and Kit

DESCRIPTION

For the last few decades, Sanger Sequencing has been the standard for analyzing DNA sequences. Due to its need for repetitive primer design, primer synthesis, and sequencing steps during Primer Walking when analyzing long sequences, however, it requires lengthy experimental time and large costs to perform. Additionally, issues such as high re-experimentation rates, intermittent errors, and a less than 1 kb read length limitation have made sequence analysis difficult for clients. To overcome these limitations, Celemics created an NGS-based molecular barcoding technology and NGS error elimination algorithm solution, allowing for the analysis of sequences with lengths greater than 1kb without the need of sequencing primers.

KEY FEATURES

Long DNA sequencing, No need of sequencing primer	Analyze from 200 bp to 20 kb and longer length in a single reaction No need of sequencing primer* No need of repetitive primer walking for long DNA de novo sequencing
Cost-effective, highly accurate, rapid turnaround time	Novel NGS-based proprietary enzyme and bioinformatics technology Cost-effective sequencing compared to Sanger sequencing Secure sequencing accuracy with NGS-based sequencing that yields more reliable results than Sanger sequencing Receive digitized results within 1-2 business days
Wide compatibility	Various applications with no limitation on DNA size or sample types across a broad range of origins Compatible with unpurified PCR products**

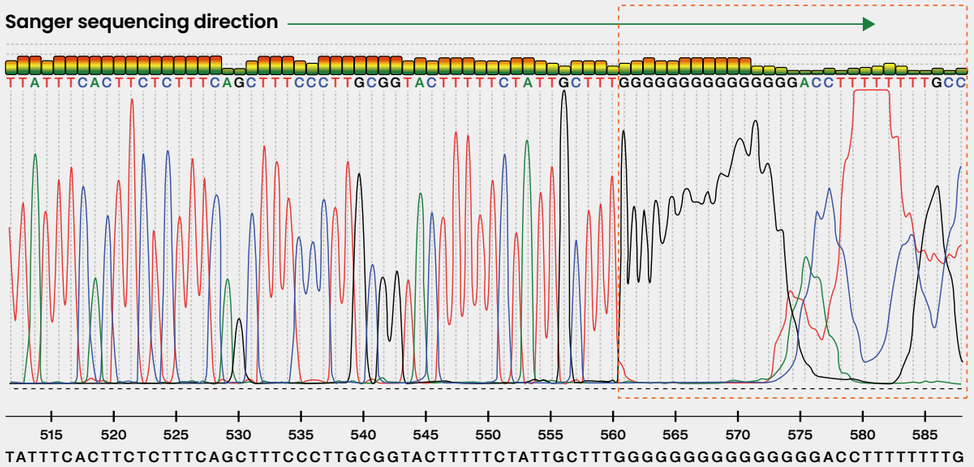
* Only primer sequence information is required
** Only samples that have single bands from gel electrophoresis are accepted

HIGH ACCURACY ACHIEVED BY NGS-BASED BTSeq™ SEQUENCING SERVICE

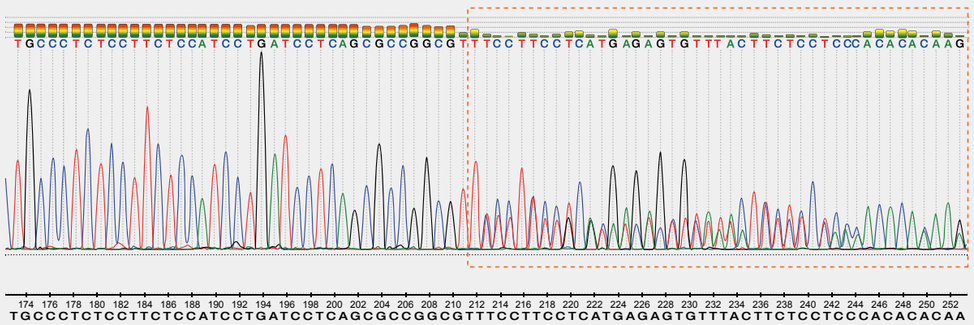
Sanger sequencing has been the gold standard sequencing method. Although Sanger sequencing service providers have supported researchers for several decades, the high competition among providers led to cost reduction in Sanger sequencing reagents. Most Sanger sequencing service providers started diluting the reagents and applying methods that are not recommended for the best quality result. This has resulted in inaccurate sequencing results and repetitive sequencing cycles.
While Sanger sequencing may have many limitations such as homopolymer sequencing, multi-peak problems, and detecting Indel or frameshift mutations, BTSeq™ overcomes such limitations and provides accurate sequencing data even from poor quality or low-amount samples.

Limitations of Sanger sequencing

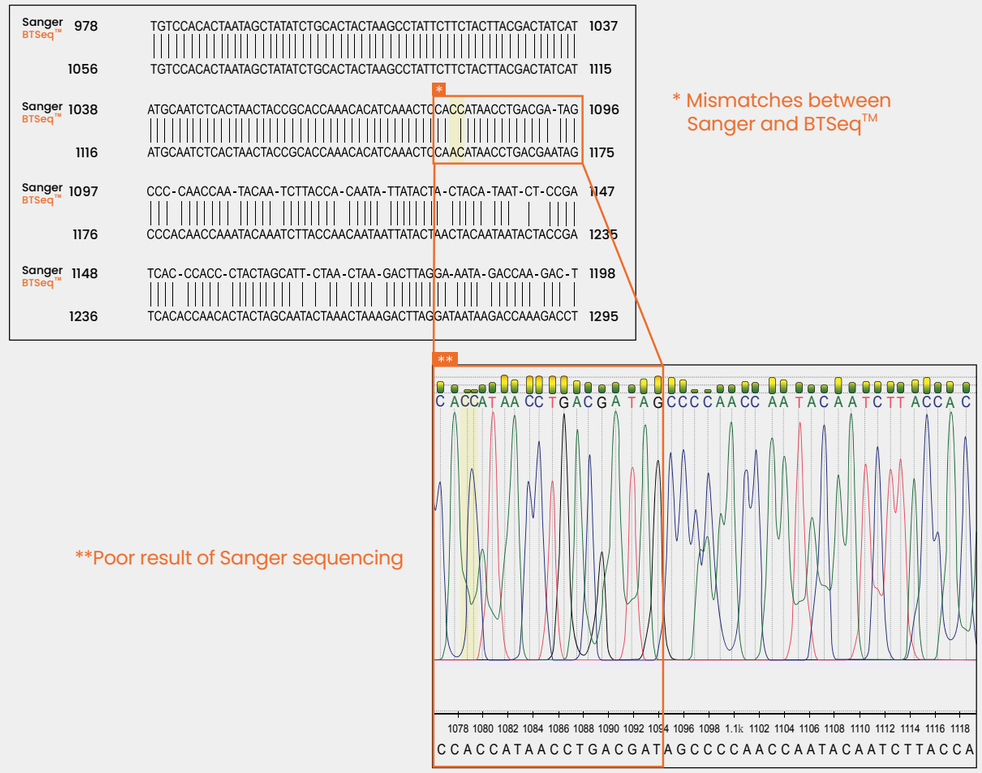
Example of homopolymer problem in Sanger sequencing



Example of multi-peak problem in Sanger sequencing



Accurate sequencing of BTSeq™



Most mismatches between BTSeq™ and Sanger sequencing results were due to the minor peaks or poor-quality results from Sanger sequencing.

Comparison between Sanger and BTSeq™

	Sanger	BTSeq™
Data type	Analog	Digital
Data quality	Ambiguous	Clear
Analysis size	Up to 1 kb	Up to 20 kb or longer
Sample concentration	> 100 ng/μl	> 10 ng/μl
Sample amount	> 20 μl	> 10 μl

COMPARISON TEST

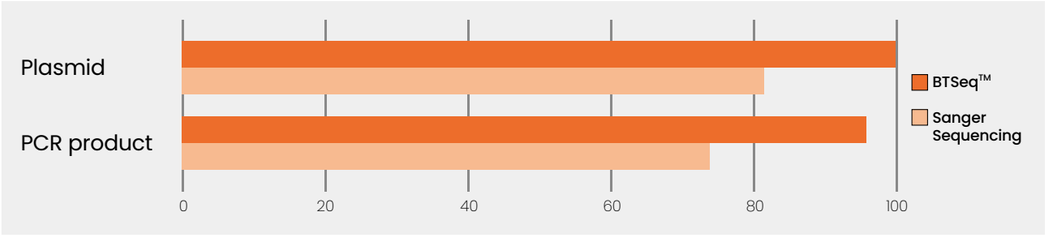
To assess the performance of BTSeq™, we have conducted multiple comparison tests with Sanger sequencing method. The samples that were sequenced by Sanger were provided by a Sanger Sequencing partner and randomly selected for BTSeq™ validation test. More than 80% of PCR product samples were not purified. The sample concentration ranged from 0.1 ng/μl to 200 ng/μl and 1 μl (0.1 ng – 200 ng) of each sample were used for BTSeq™. The results show high concordance of BTSeq™ with Sanger sequencing with even higher accuracy.

BTSeq™ shows errorless sequencing results

Number of Samples	Plasmid (n=454)		PCR product (n=801)	
Method	**BTSeq™**	**Sanger Sequencing**	**BTSeq™**	**Sanger Sequencing**
Unidentified	0	85**	36*	211**
Identified	454	369	765	590
Analysis success rate (%)	100.0%	81.3%	95.5%	73.7%

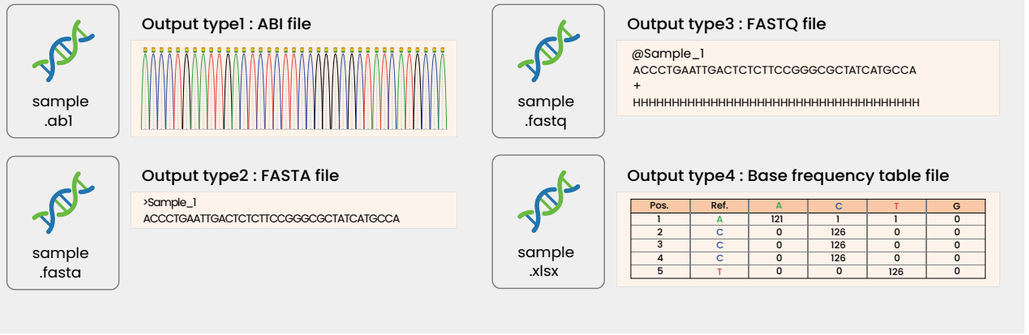
* Long repeated sequences
** Poor sequencing results

Analysis Success Rate (%)



DIGITIZED RESULTS*

The BTSeq™ service, an NGS-based sequencing service, provides digitized results by standalone bioinformatics analysis software enabling various options for result data



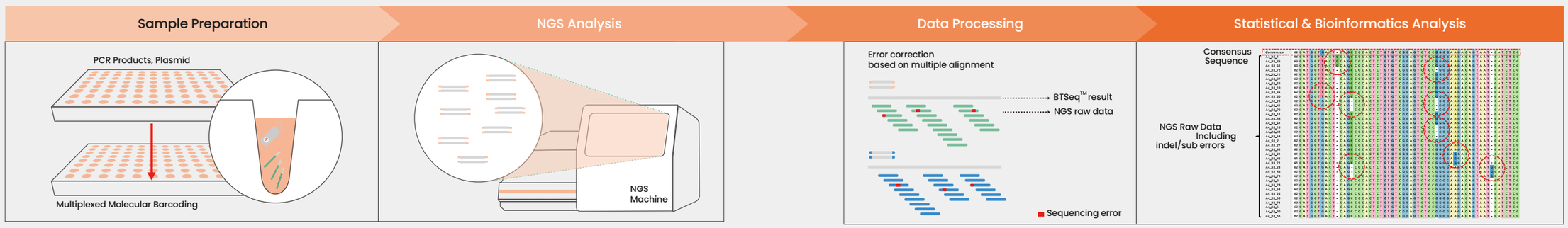
* Different options provided for different applications. Contact us for more information.

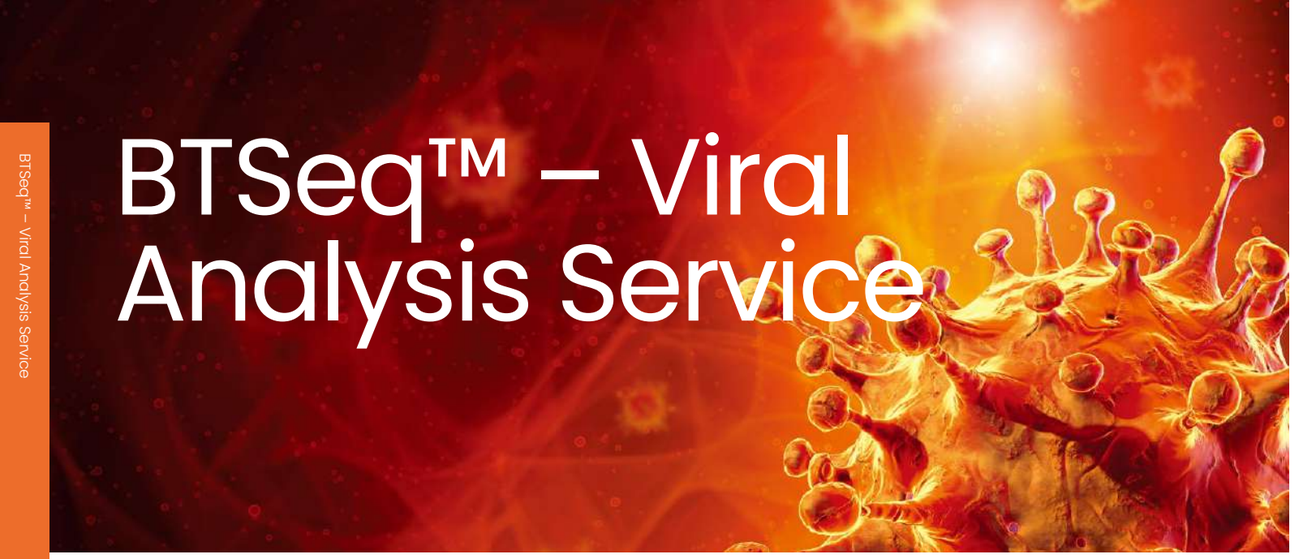
BTSeq™ SERVICE OPTIONS

Product Group	Service Option	Sample Type	Description
BTSeq™	BTSeq™ – Standard	PCR product / Plasmid	Primer sequence information is required*
	Plasmid Extraction	E. coli	-
	BTSeq™ – Raw Data	PCR product / Plasmid	Provides FASTQ file only

BTSeq™ SERVICE PROCESS

Celemics has developed sample preparation techniques and bioinformatics software enabling cost-effective workflow. The BTSeq™ sequencing provides highly accurate results with short turnaround time (TAT) by effectively correcting sequencing errors and generating consensus sequence with Celemics proprietary techniques.





BTSeq™ – Viral Analysis Service

DESCRIPTION

In most cases, RNA of the host cell is separated and purified along with viral RNA during extraction.
This leads to an excessive amount of data being required to perform typical Total RNA-seq compared to the entire viral genome, leading to low-quality data and high costs. Celemics solves this issue by developing extremely uniform amplification technology and bioinformatics software, which in turn provides quality data by efficiently eliminating any gaps generated from bias in the RT-PCR step.

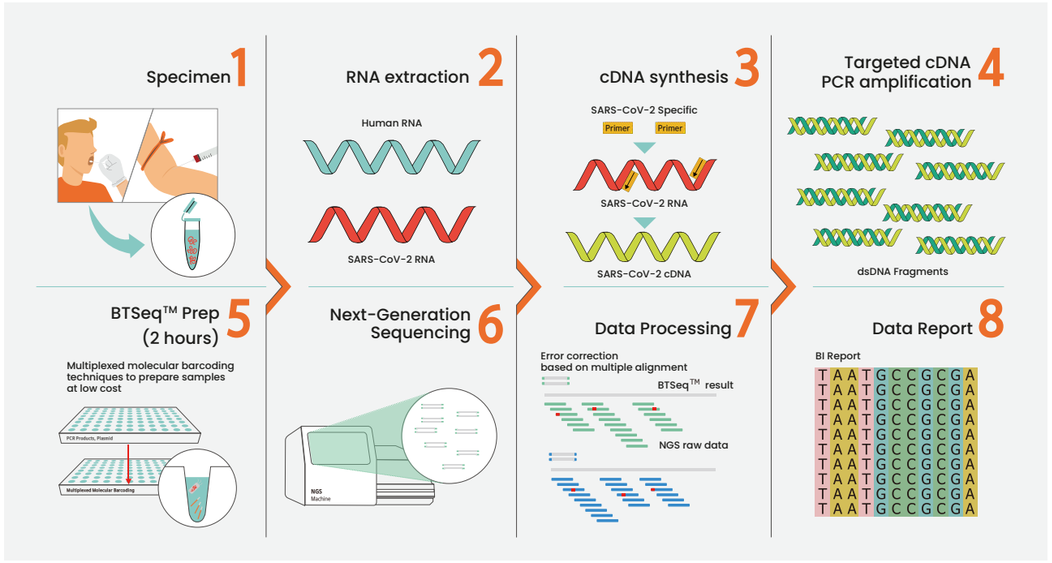
KEY FEATURES

1. High quality result generation from minuscule low-quality RNA samples	Enabling of high-quality whole genome analysis, even in minuscule low-quality RNA samples extracted from upper respiratory tract, nasopharyngeal, oropharyngeal swab clinical specimens
2. Results provided within 24 hours	Provision of whole novel coronavirus genome within 24 hours using Celemics' proprietary reagent and bioinformatics technology
3. High-quality data generation at cost-effective price	High-quality result generation, even from miniscule amounts of clinical samples

REQUIREMENTS

Sample type	RNA
Concentration	Ct value < 25
Volume	40 µl
Turnaround time	Within 3-5 business days from sample collection
Shipment	Shipping on dry ice (essential)

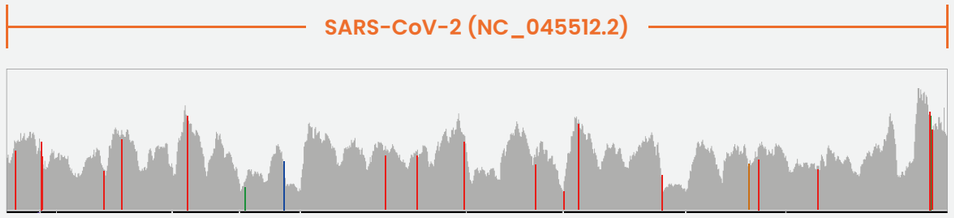
SERVICE PROCESS



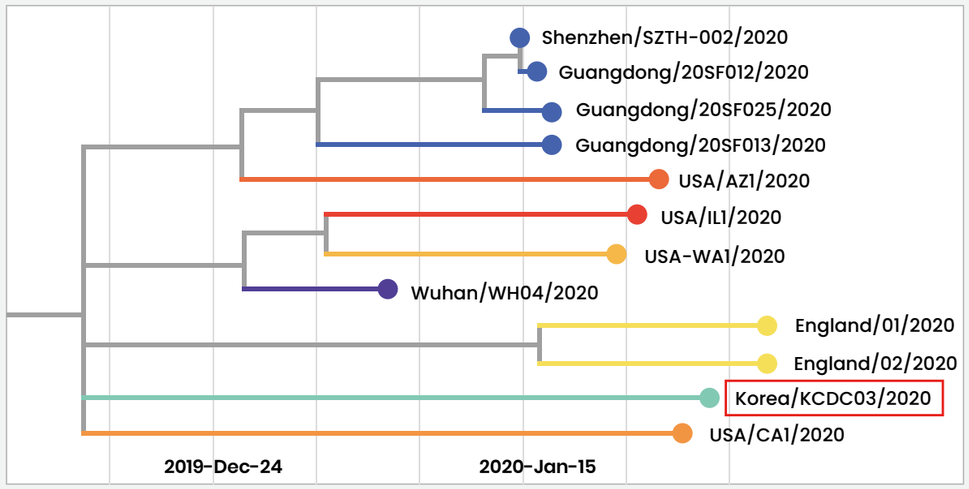
COMPARISON BETWEEN TOTAL RNA SEQUENCING AND BTSeq™

	Total RNA-Sequencing	BTSeq™
cDNA Synthesis	**Total RNA (host and viral) into cDNA** : Leads to unnecessary data and high sequencing cost	**Only viral RNA to cDNA** : Utilizes Virus specific multiplex primers to selectively amplify viral RNAs from total RNAs
Target Enrichment	**No target enrichment required** : Viral RNA coexists with host RNA when cDNA synthesis is performed	**Viral genome is specifically amplified** : Only a small amount of viral RNA is required
Library Preparation	RNA library prep using RNA library kit: **4 hours**	Simple library prep using BTSeq™ reagent: **2 hours**
Data Analysis	: Mapped to the viral genome : Read/assembly based classification	: Mapped to the viral genome : Read/assembly based classification
Turnaround Time	**2-3 weeks**	**1-2 days**

FULL COVERAGE OF SARS-COV-2 WGS ANALYZED BY BTSeq™ FROM PATIENT SPECIMENS

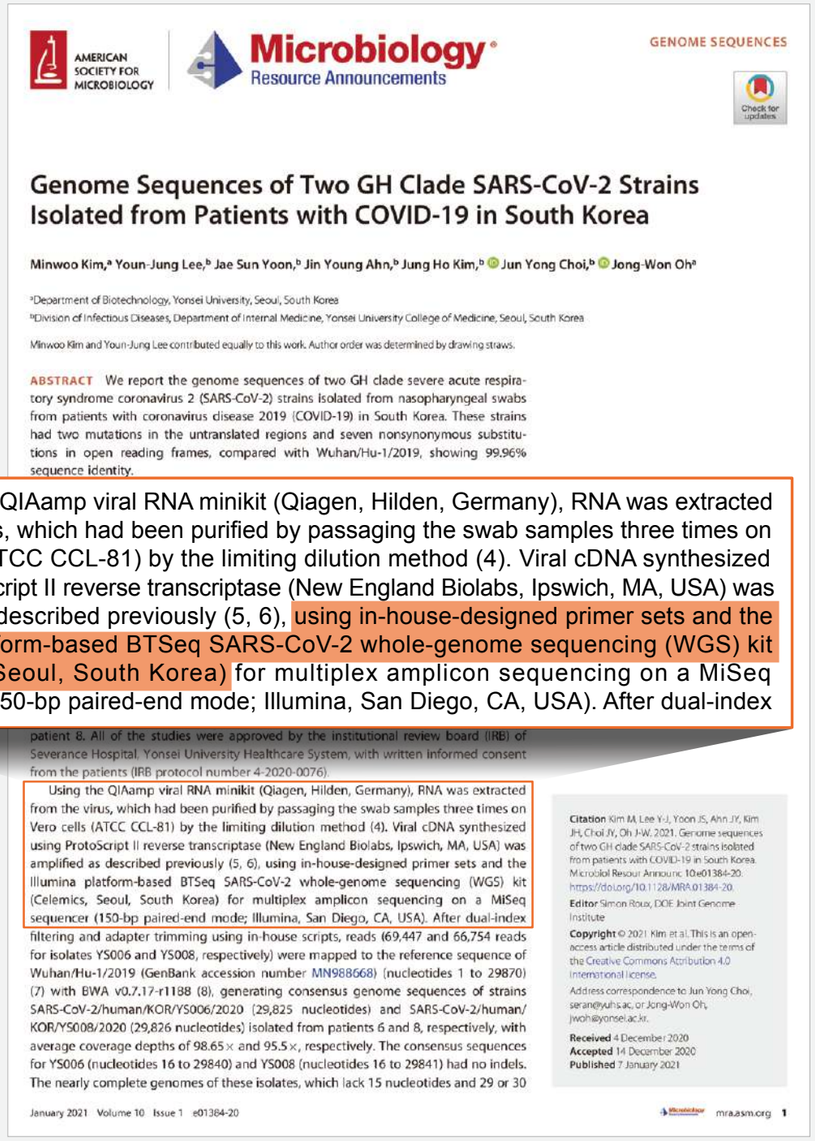


IDENTIFICATION OF KOREA/KCDC03/2020 USING BTSeq™ – VIRAL ANALYSIS SERVICE



Korea/KCDC03/2020	
Collection date	2020-01-26
Authors	Kim et al
Title	Newly discovered betacoronavirus, 2019-2020
Country	South Korea
Admin division	Gyeonggi
Host	Human
Location	Gyeonggi

REFERENCE OF BTSeq™ – VIRAL ANALYSIS SERVICE



AMERICAN SOCIETY FOR MICROBIOLOGY | Microbiology Resource Announcements

GENOME SEQUENCES

Genome Sequences of Two GH Clade SARS-CoV-2 Strains Isolated from Patients with COVID-19 in South Korea

Minwoo Kim,[a] Youn-Jung Lee,[b] Jae Sun Yoon,[b] Jin Young Ahn,[b] Jung Ho Kim,[b] Jun Yong Choi,[b] Jong-Won Oh[a]

[a]Department of Biotechnology, Yonsei University, Seoul, South Korea
[b]Division of Infectious Diseases, Department of Internal Medicine, Yonsei University College of Medicine, Seoul, South Korea

Minwoo Kim and Youn-Jung Lee contributed equally to this work. Author order was determined by drawing straws.

ABSTRACT We report the genome sequences of two GH clade severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) strains isolated from nasopharyngeal swabs from patients with coronavirus disease 2019 (COVID-19) in South Korea. These strains had two mutations in the untranslated regions and seven nonsynonymous substitutions in open reading frames, compared with Wuhan/Hu-1/2019, showing 99.96% sequence identity.

Using the QIAamp viral RNA minikit (Qiagen, Hilden, Germany), RNA was extracted from the virus, which had been purified by passaging the swab samples three times on Vero cells (ATCC CCL-81) by the limiting dilution method (4). Viral cDNA synthesized using ProtoScript II reverse transcriptase (New England Biolabs, Ipswich, MA, USA) was amplified as described previously (5, 6), using in-house-designed primer sets and the Illumina platform-based BTSeq SARS-CoV-2 whole-genome sequencing (WGS) kit (Celemics, Seoul, South Korea) for multiplex amplicon sequencing on a MiSeq sequencer (150-bp paired-end mode; Illumina, San Diego, CA, USA). After dual-index

patient 8. All of the studies were approved by the institutional review board (IRB) of Severance Hospital, Yonsei University Healthcare System, with written informed consent from the patients (IRB protocol number 4-2020-0076).

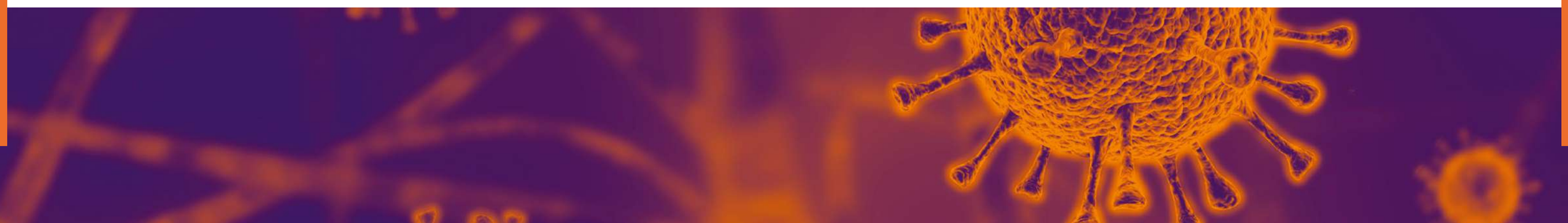
Using the QIAamp viral RNA minikit (Qiagen, Hilden, Germany), RNA was extracted from the virus, which had been purified by passaging the swab samples three times on Vero cells (ATCC CCL-81) by the limiting dilution method (4). Viral cDNA synthesized using ProtoScript II reverse transcriptase (New England Biolabs, Ipswich, MA, USA) was amplified as described previously (5, 6), using in-house-designed primer sets and the Illumina platform-based BTSeq SARS-CoV-2 whole-genome sequencing (WGS) kit (Celemics, Seoul, South Korea) for multiplex amplicon sequencing on a MiSeq sequencer (150-bp paired-end mode; Illumina, San Diego, CA, USA). After dual-index

filtering and adapter trimming using in-house scripts, reads (69,447 and 66,754 reads for isolates YS006 and YS008, respectively) were mapped to the reference sequence of Wuhan/Hu-1/2019 (GenBank accession number MN988668) (nucleotides 1 to 29870) (7) with BWA v0.7.17-r1188 (8), generating consensus genome sequences of strains SARS-CoV-2/human/KOR/YS006/2020 (29,825 nucleotides) and SARS-CoV-2/human/KOR/YS008/2020 (29,826 nucleotides) isolated from patients 6 and 8, respectively, with average coverage depths of 98.65× and 95.5×, respectively. The consensus sequences for YS006 (nucleotides 16 to 29840) and YS008 (nucleotides 16 to 29841) had no indels. The nearly complete genomes of these isolates, which lack 15 nucleotides and 29 or 30

Citation Kim M, Lee Y-J, Yoon JS, Ahn JY, Kim JH, Choi JY, Oh J-W. 2021. Genome sequences of two GH clade SARS-CoV-2 strains isolated from patients with COVID-19 in South Korea. Microbiol Resour Announc 10:e01384-20. https://doi.org/10.1128/MRA.01384-20.
Editor Simon Roux, DOE Joint Genome Institute

Address correspondence to Jun Yong Choi, seran@yuhs.ac, or Jong-Won Oh, jwoh@yonsei.ac.kr.
Received 4 December 2020
Accepted 14 December 2020
Published 7 January 2021

*Reference: Genome Sequences of Two GH Clade SARS-CoV-2 Strains Isolated from Patients with COVID-19 in South Korea, American Society Microbiology. (2020)



BTSeq™ Mitochondrial DNA Sequencing Service

DESCRIPTION

The BTSeq™ Mitochondrial DNA Sequencing enables accurate analysis of clinical variability and genetic heterogeneity. By sequencing 17 kb-long mtDNA with newly developed NGS-based technology, customers can decipher the instability and variations of mtDNA associated with many metabolic and neurologic disorders and cancers. The service provides highly accurate results with fast TAT and cost-effectiveness.

REQUIREMENTS

Sample Type	gDNA
Concentration	50 ng/μl
Volume	10 μl
Turnaround time	Within 4 business weeks from sample arrival
Shipment	Ship on ice

RESULT EXAMPLE OF BTSeq™ MITOCHONDRIAL DNA SEQUENCING

Example of mitochondrial variant analysis report

Sample	Gene	Amino Acid Change	Type	Allele		Sequencing Depth			VAF	Associated Disease
				Ref	Alt	Total	Ref	Alt		
Sample 1	ND2	p.Leu237Met	Missense	C	A	16370	0	16361	-	Blood iron metabolism
	ATP8	p.Leu17Phe	Missense	C	T	16155	8	16139	-	Longevity
	ND5	p.Asn30fs	Frameshift	-	A	17593	17197	197	1.12%	
Sample 2	ND2	p.Thr122Ala	Missense	A	G	16759	14	8005	-	AD, PD
	ATP6	p.Met58Thr	Missense	T	C	16909	15721	12	4.53%	-
	ND3	p.Thr114Ala	Missense	A	G	20141	8	219	1.24%	Breast cancer risk
Sample 3	ND2	p.Leu237Met	Missense	C	A	5100	0	5100	-	Blood iron metabolism
	ATP8	p.Leu17Phe	Missense	C	T	16353	6	16340	-	Longevity
	ND5	p.Asn30fs	Frameshift	-	A	16960	16625	193	1.14%	405

Example of summary report of NGS operation (target size: 16.6 kb)

Sample name	Raw read	Raw base	Total read	Filtered ratio	On target read ratio	On target base ratio	Uncovered	20x coverage	50x coverage	100x coverage
Sample 1	3,521,438	531,737,138	3,486,316	99.00%	90.78%	95.36%	0.00%	100.00%	100.00%	100.00%
Sample 2	3,514,296	530,658,696	3,479,540	99.01%	91.39%	95.82%	0.00%	100.00%	100.00%	99.99%
Sample 3	3,526,146	532,448,046	3,489,580	98.96%	90.12%	95.24%	0.00%	100.00%	100.00%	100.00%
Sample 4	3,500,420	528,563,420	3,463,806	98.95%	90.85%	95.67%	0.00%	100.00%	100.00%	100.00%

BTSeq™ Full Plasmid Sequencing Service

DESCRIPTION

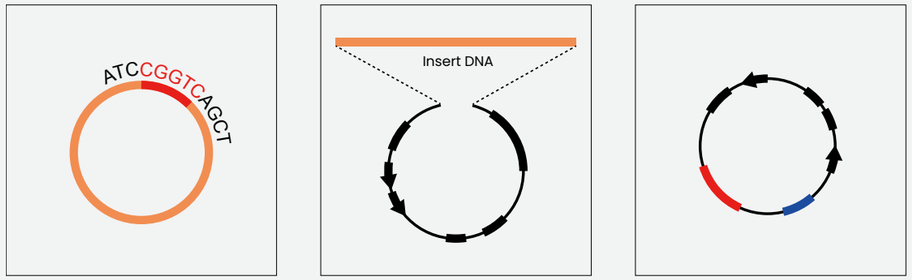
The BTSeq™ Full Plasmid Sequencing Service allows for the most effective analysis of the full-length sequencing of plasmids with shorter TAT and lower cost than Sanger sequencing. The service is ideal for protein engineering, vector engineering, antibody optimization, synthetic biology, and various other applications.

REQUIREMENTS

Sample type	PCR product, Plasmid*	
Concentration	10 ng/μl	
Volume	10 μl	
Turnaround time	Within 1 business day from sample arrival	
Packaging	1) RT 2) Ship on ice (Recommended)	

* Contact us for plasmids longer than 20kb

APPLICATIONS OF BTSeq™ FULL PLASMID SEQUENCING

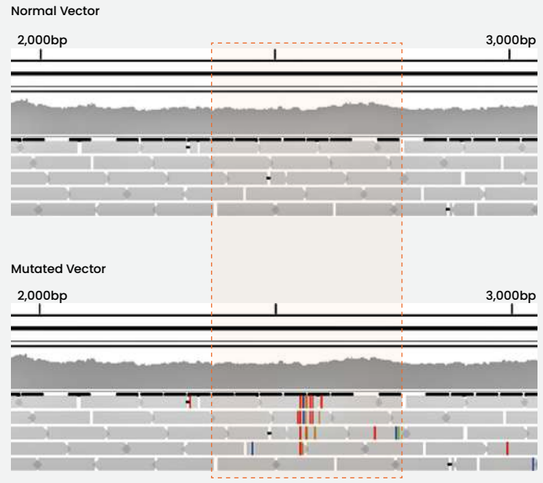


Confirmation of vector sequence

Sequencing large insert

Vector engineering

COMPARISON IGV DATA BETWEEN NORMAL AND MUTATED VECTORS



CELEMICS SOLUTIONS FOR IMMUNE REPERTOIRE SEQUENCING

CELEMICS PRODUCTS & SERVICES 2022

Immune Repertoire Profiling Service
TrueRepertoire™ Service

CELEMICS

Immune Repertoire Profiling Service

DESCRIPTION

Immune repertoire often represents an individual's current immunological status; whether the person is healthy, vaccinated, diseased, or infected. Only high-throughput NGS analysis can comprehensively profile an individual's immune repertoire. The Immune Repertoire Profiling Service provides effective data acquisition, integration, and interpretation for the customers.

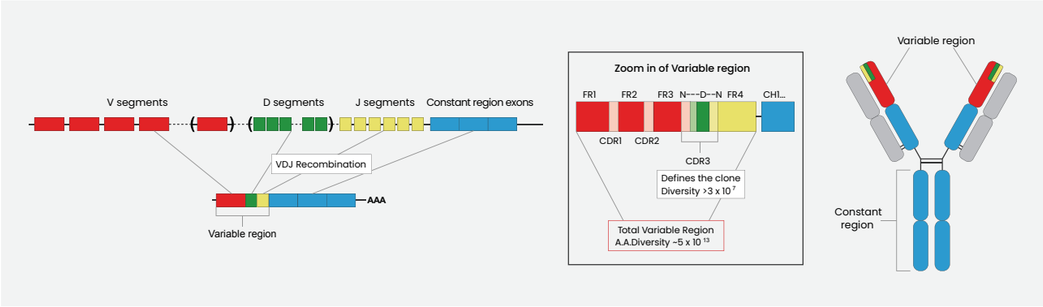
KEY FEATURES

1. Quantitative analysis of library diversity	- NGS-based analysis of complex antibody library consisting of millions (10^6~10^{12}) of sequences in a single experiment - Analysis of immunoglobulin and T-cell receptor repertoire; analysis of BCR/TCR for each clone - Frequency analysis of individual antibody clones within the library, identifying major and minor clones
2. Tracking of clonal frequencies for each sample	- For antibody discovery, analysis of library diversity according to its panning degree enabling monitoring changes in clonal frequency - Minimized omission of potentially significant antibody clones - Analysis of immune repertoire characteristics from blood sample and monitoring of each clone
3. Various analysis options for immune system studies	Perform the experiment with drastically reduced time and cost enabled by the advanced technology of MSSIC developed by Celemics

REQUIREMENTS

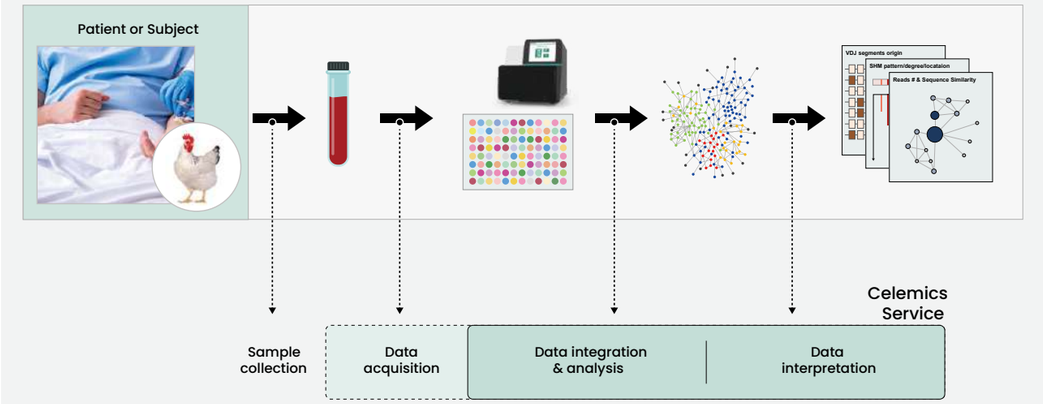
Sample type	Total RNA from B-Cell or/and T-Cell, DNA from B-Cell or/and T-Cell, DNA/RNA Amplicons
Concentration	100 ng/μl
Amount	1 μg
Turnaround time	Within 4-6 business weeks from sample collection
Temperature	RT for storage and shipment

DIVERSITY OF ANTIBODY



The antibody genes are composed of many different segments. The antibodies are presented in B cells with great diversity of 10^{13} repertoires.

GENERAL WORKFLOW



Celemics provides service for data acquisition, integration, and analysis, and interpretation.

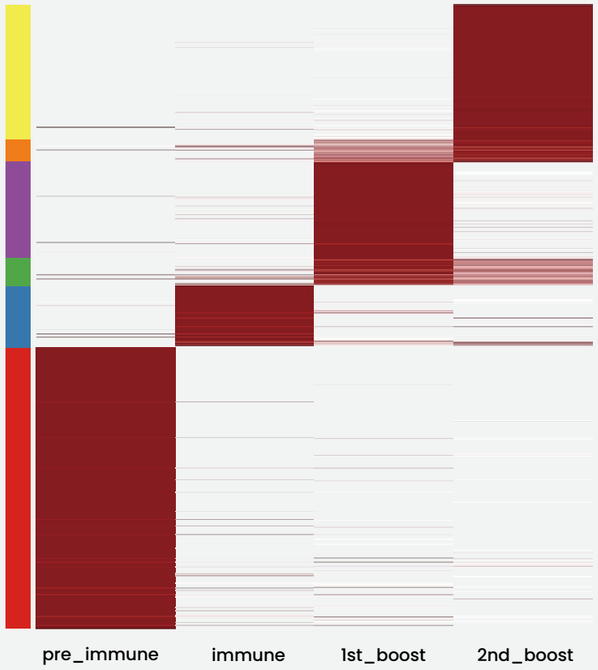
IMMUNE PROFILING EXAMPLES

Example 1 - Discovery of candidate antibodies from actively immunized chickens

CDR3 sorting results

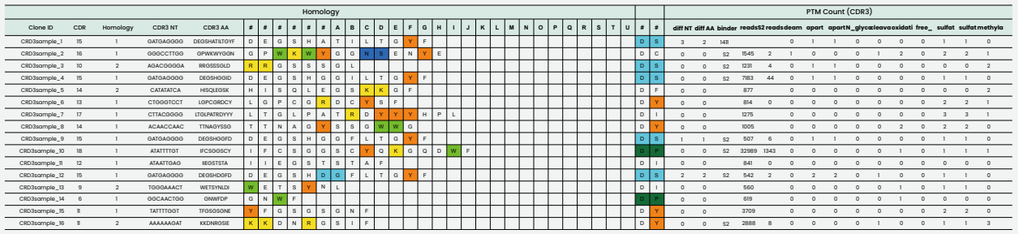
CDR3 ID	CDR3 AA	p_pre-immune	p_immune	p_1st_boost	p_2nd_boost	p_tissue	p_BM_2nd
CDR3sample_1	GSRDSSASTI	2433	773	31	0	2	461
CDR3sample_2	GSYDSSYVGI	1756	1444	2269	895	1058	789
CDR3sample_3	GSIDSSYVGI	1019	402	876	938	541	346
CDR3sample_4	ANFDSSSGAGI	46	25	338	483	1707	345
CDR3sample_5	GGYDSSAGI	231	268	934	207	966	7770
CDR3sample_6	GSFDSSTYAGI	3678	1034	425	290	547	431
CDR3sample_7	GSRDSSASTI	2433	773	31	0	2	461
CDR3sample_8	GSRDSSYVGI	6427	6370	10151	5756	10089	2680
CDR3sample_9	GGYDGSTYVGI	279	211	2047	178	271	88
CDR3sample_10	GSRDSNYVGI	407	567	974	749	868	224
CDR3sample_11	GSSSGTGI	1563	2580	899	1999	114	24702
CDR3sample_12	GSYDSSAGI	1195	875	1342	743	746	288
CDR3sample_13	GSRDSTYVGI	461	795	1355	998	983	136
CDR3sample_14	GGYDSSTDAGI	1167	1129	1353	1617	892	405

Sequence abundance clustering result



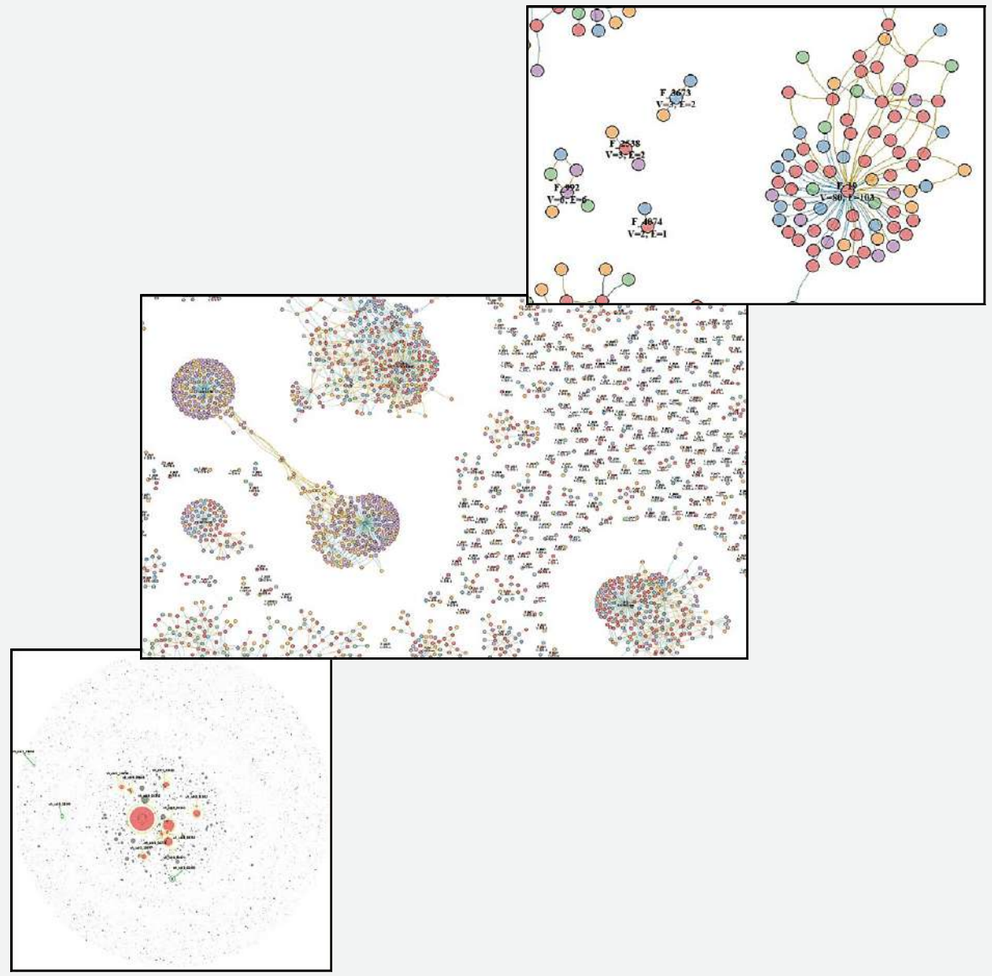
Example 2 - Discovery of novel drug candidates with antibody analysis from respiratory infection patient samples

CDR3 similarity analysis



Clone ID	CDR	Homology	CDR3 NT	CDR3 AA	PTM Count (CDR3)									
					diff NT	diff AA	binder	reads52	reads deam	apart	apart N_glycsleavoxidati	free_	sulfat	sulfat methyla
CRD3sample_1	15	1	GATGAGGGG	DEGSHATSTGYF	3	2	148		0	1	1	0	0	0	1	1	0
CRD3sample_2	16	1	GGGCCTTGG	GPWKWYGGN	0	0	52	1545	2	1	0	0	0	1	2	0	2	2	1
CRD3sample_3	10	2	AGACGGGGA	RRGSSSGLD	0	0	52	1231	4	0	1	1	0	0	0	0	0	0	2
CRD3sample_4	15	1	GATGAGGGG	DEGSHGGID	0	0	52	7183	44	0	1	1	0	0	0	0	1	1	0
CRD3sample_5	14	2	CATATATCA	HSQLEDSIK	0	0		877		0	0	0	0	0	0	0	0	0	2
CRD3sample_6	13	1	CTGGGTCCT	LGPCGRDCY	0	0		814	0	0	0	0	0	0	0	0	2	2	1
CRD3sample_7	17	1	CTTACGGGG	LTGLPATRDYYY	0	0		1275		0	0	0	0	0	0	0	3	3	1
CRD3sample_8	14	1	ACAACCAAC	TTNAGYSSG	0	0		1005		0	0	0	0	0	2	0	2	2	0
CRD3sample_9	15	1	GATGAGGGG	DEGSHGGFD	1	1	52	507	6	0	1	1	0	0	0	0	1	1	0
CRD3sample_10	18	1	ATATTTTGT	IFCSGDSCY	0	0	52	32989	1343	0	0	0	0	0	1	0	1	1	1
CRD3sample_11	12	1	ATAATTGAG	IEGSTSTA	0	0		841	0	0	0	0	0	0	0	0	0	0	0
CRD3sample_12	15	1	GATGAGGGG	DEGSHGDFD	2	2	52	542	2	0	2	2	0	1	0	0	1	1	0
CRD3sample_13	9	2	TGGGAAACT	WETEYNLDI	0	0		560		0	0	0	0	0	1	0	1	1	0
CRD3sample_14	6	1	GGCAACTGG	GNWFDF	0	0		619		0	0	0	0	0	1	0	0	0	0
CRD3sample_15	11	1	TATTTTGGT	TFGSGSGNE	0	0		3709		0	0	0	0	0	0	0	2	2	0
CRD3sample_16	11	2	AAAAAAGAT	KKDNRGSE	0	0	52	2888	8	0	0	0	0	1	0	0	1	1	3

Network Analysis Between Antibody Sequences



TrueRepertoire™ Service

DESCRIPTION

The TrueRepertoire™ is a NGS-based antibody library sequencing platform developed to overcome the key issues of existing methods such as sequencing error, short-read length, and high-cost gene synthesis for further characterization. Celemics has developed a cloning microchip, barcode assay technology, and laser-based non-contact clone retrieval system and integrated into the newly developed platform, TrueRepertoire™ assay. This service allows for full sequence analysis of over 10,000 clones in a single experiment and thereby discovering rare clones. The TrueRepertoire™ service contains the client's antibody clone of interest within the library itself, eliminating the need to perform new gene synthesis and significantly reducing time and cost.

KEY FEATURES

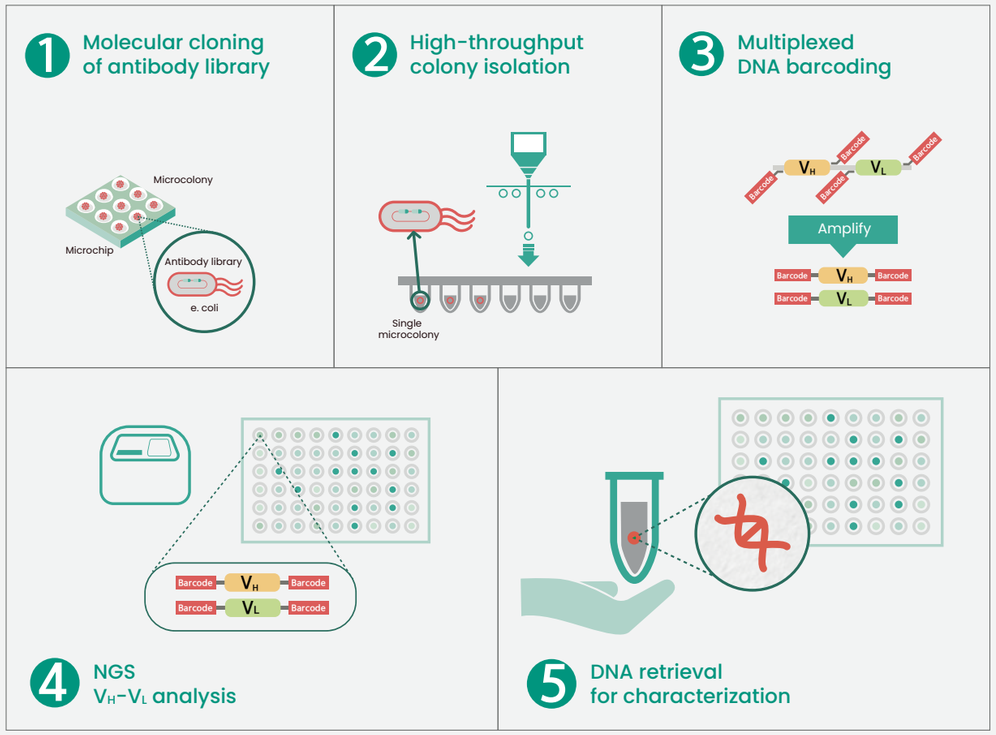
1. Provision of antibody DNA sequence library containing over 10,000 errorless strains	NGS-based sequence analysis and high-capacity clone separation and molecular barcode assays using Celemics proprietary MSSIC technology
2. V_H-V_L linkage analysis of each antibody	Receive V_H-V_L linkage information, an area difficult to analyze through NGS due to its short read length
3. Provision of physical property analysis of each antibody through bioinformatics analysis	Clone frequency distribution within the library V_H-V_L sequence length distribution, post-translation modification information, CDR and frame amino acid information, etc.
4. Retrieval of selected physical antibody allowing for convenient workflow	Eliminates the need to perform new gene synthesis and reduces time and cost due to the antibody clones within the library itself, enabling isolation of physical DNA for further characterization

REQUIREMENTS

Sample type*	Total RNA from B-Cell or/and T-Cell, DNA from B-Cell or/and T-Cell, DNA/RNA Amplicons
Concentration	100 ng/µl
Amount	1 µg
Turnaround time	Within 4-6 business weeks from sample collection**
Temperature	RT for storage and shipment

* ~30 bp of Consensus upstream & downstream sequence over V_H and V_L region required
** TAT depends on colony size

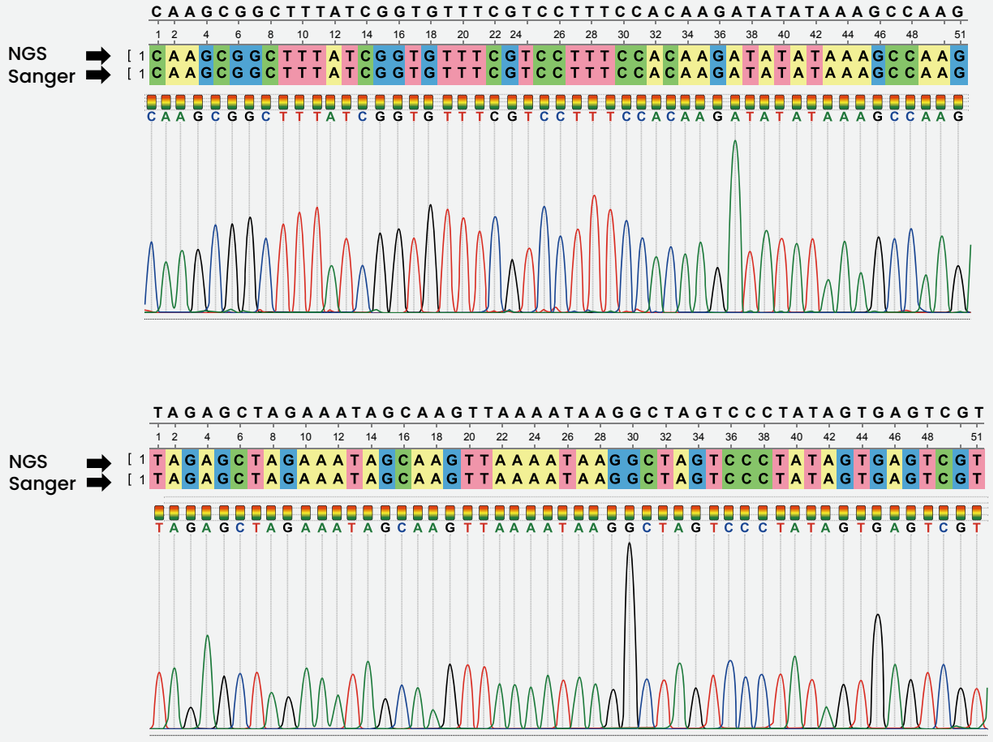
HOW TrueRepertoire™ WORKS



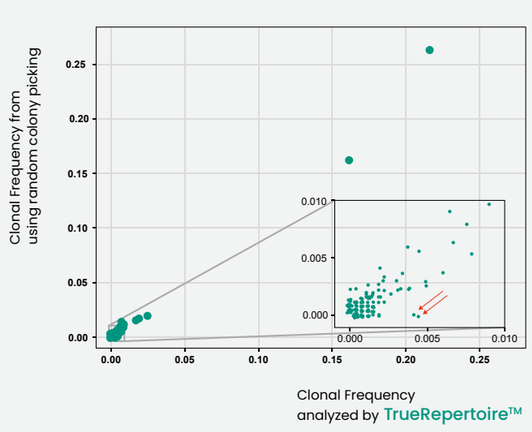
1. Celemics proprietary microcolony chip formation with high density, each colony starts from a single E. coli.
2. Extraction of the colonies from the microchip into microwell by Celemics' proprietary laser system
3. Multiplex PCR with barcoded primers from the isolated colonies
4. NGS and computation of the consensus sequences with cognate pairing of V_H and V_L
5. Clonal DNA retrieval based on the consensus sequence for further characterization

VALIDATION TESTS

Validation I. Result of 480 randomly selected antibody clones from TrueRepertoire™ perfectly matched (480/480) Sanger sequencing results of their physical DNA

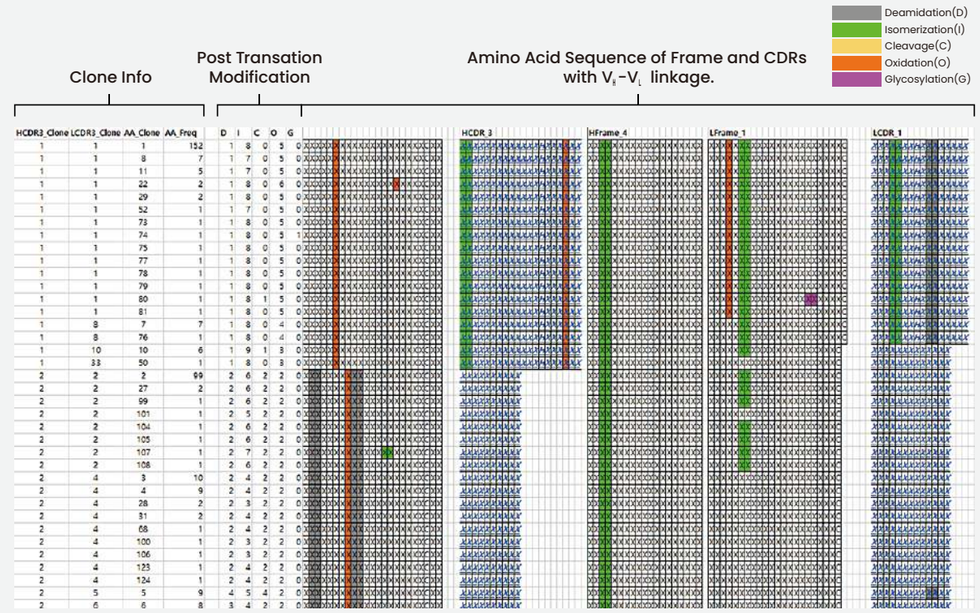


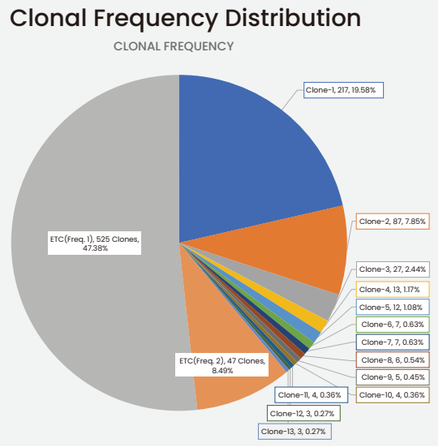
Validation II. Similar clonotype frequencies of major clones between TrueRepertoire™ and random colony picking

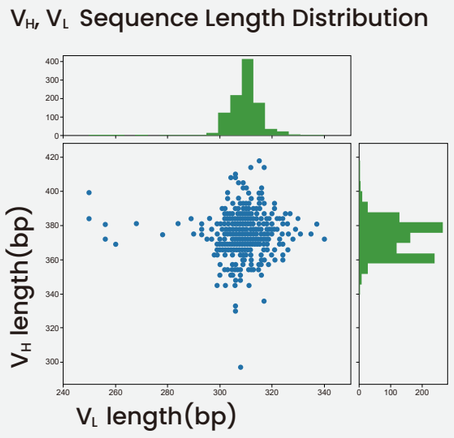


1. Major clones showed similar clonotype frequency in both platforms - random colony picking followed by Sanger sequencing and TrueRepertoire™
2. The result showed that there were newly identified clones found only in the TrueRepertoire™ results (red arrows)

USER FRIENDLY TrueRepertoire™ REPORT







MODULAR ACCESSORIES

CELEMICS PRODUCTS & SERVICES 2021

Library Preparation Kit - Standard / EP
Double-Stranded cDNA Synthesis Kit
Hybridization Enhancer
CeleMag™ Clean-up Bead
CeleMag™ Streptavidin Bead
CLM Polymerase
Bioinformatics Software

CELEMICS

Celemics Library Preparation Kit
Standard / EP

DESCRIPTION

Celemics Library Preparation Kit is optimized for high-efficiency Celemics panels. The Library Preparation Kits include End-repair, A-tailing enzyme mix, index primers (single or dual), adapters and buffers.

LIBRARY PREPARATION WORKFLOW FOR TARGET ENRICHMENT NGS

DNA Fragmentation		
Standard Fragmentation		EP Fragmentation
Option 1. Sonication	Option 2. Fragmentase	Fragmentase
Bead Purification & Quantification		
NGS Library Preparation		
ER/A	ER/A	ER/A
Adapter Ligation (Single/Dual Index)		
Bead Purification		
Index PCR		
Target Enrichment		

Celemics provides two methods for the library preparation step, Standard Library Preparation Kit and Enzymatic Preparation Kit (EP Kit). The Standard Library Preparation Kit includes all reagents for End repair (ER), A-tailing (A), and Adapter Ligation steps. For DNA fragmentation from Standard Library Preparation Kit, customers can use ultra-sonication devices or fragmentase. Fragmentase is provided by Celemics and included in the kit upon request. While the Standard Kit is composed of 4 different steps, the EP Kit includes all steps from enzymatic fragmentation to ER/A in a single reaction enabling convenient workflow. Since the purification step is not needed for EP Kit, the kit allows for minimal DNA loss which is a crucial factor for damaged DNA samples such as FFPE. EP Kit, provided by Celemics, includes all reagents required for library preparation.

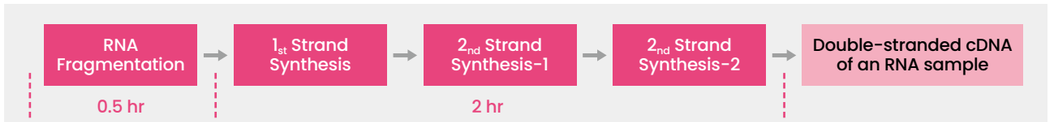
Note.
For Option 1, ultra sonicator is not provided with the kit.
For Option 2, the inclusion of the fragmentase in the kit is optional.

Celemics Double-Stranded cDNA Synthesis Kit

DESCRIPTION

Celemics Double-Stranded cDNA Synthesis Kit is optimized for NGS-based RNA sequencing. The kit includes all components from RNA fragmentation to double-stranded cDNA synthesis for NGS library preparation. The robust performance of the kit allows for the cDNA synthesis even from low amounts of RNA samples with high accuracy and reduced reaction time.

cDNA SYNTHESIS WORKFLOW



Sample amount : 10 ng to 1 µg *
Assay time : 30 minutes for RNA fragmentation and 2 hours for double-stranded cDNA synthesis
* Carrier RNA is required for sample amount < 25 ng

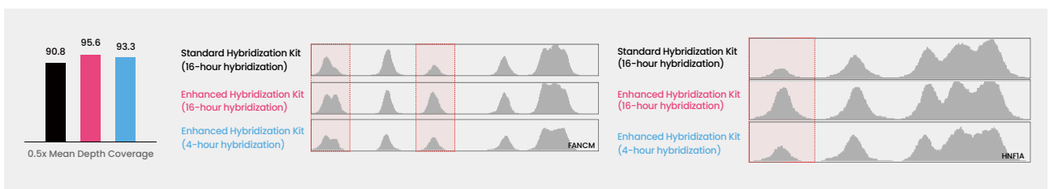
Celemics Hybridization Enhancer

DESCRIPTION

Celemics Hybridization Enhancer is developed for the hybridization step in the library preparation using Celemics Target Enrichment Kits (Enhanced Hybridization Kit). It enables 4 hours of hybridization with no compromise on the performance quality.

PERFORMANCE

Improved uniformity and coverage with Hybridization Enhancer



CeleMag™ Clean-up Bead

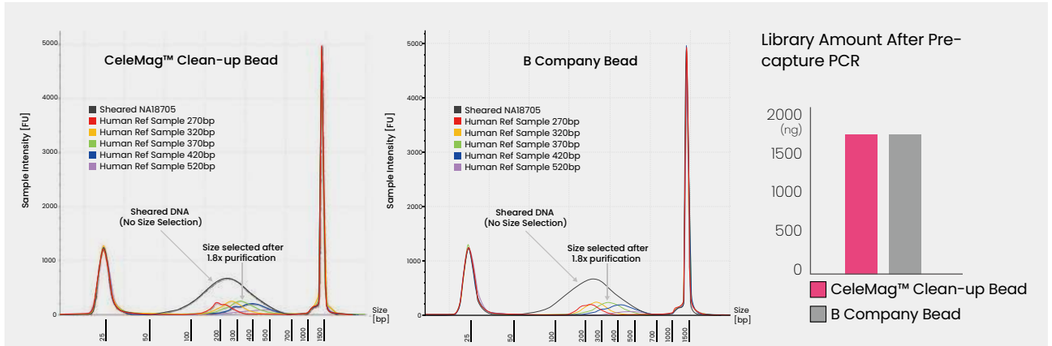
DESCRIPTION

The CeleMag™ Clean-up Bead utilizes unique magnetic bead-based chemistry enabling a simple, flexible and reproducible workflow for purification and size selection of nucleic acids.

KEY FEATURES

1. Market leading purification and size selection efficiency
2. Highly optimized with Celemics Target Enrichment Kits
3. Consistent size selection with flexibility

PERFORMANCE



CeleMag™ Clean-up Bead provides highly comparable performance to competitor product in size selection workflows, achieving consistent DNA size distributions and yielding desired library sizes.
CeleMag™ Clean-up Bead also provides equivalent NGS Library preparation recovery efficiency compared to competitor product.

CLM Polymerase

DESCRIPTION

The role of polymerase is critical in NGS process. Due to the complexity of the library, high performance polymerase is required for high uniformity and yield. As a service provider, Celemics has been providing CLM polymerase with market-leading performance, exhibiting high yield and accuracy with minimized PCR bias.
The product includes all reaction components for PCR. Contact us for more information.

CeleMag™ Streptavidin Bead

DESCRIPTION

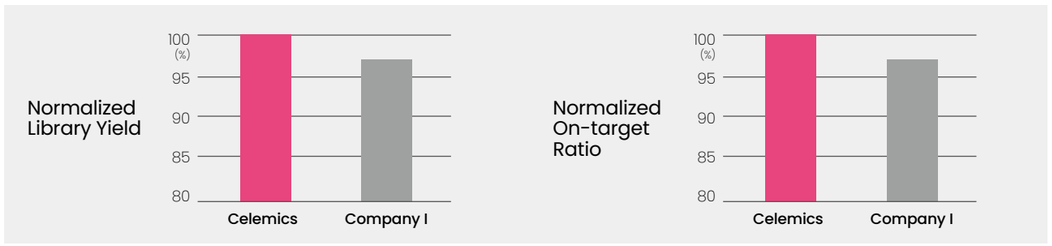
The CeleMag™ Streptavidin Bead selectively isolates biotinylated ligand, using binding properties of biotin. Its high performance enables isolating targeted genes that are bound to probes and minimizes DNA loss during the target enrichment process.

KEY FEATURES

1. High biotin-streptavidin binding capacity
2. Superior target enrichment efficiency

PERFORMANCE

Superior performance of CeleMag™ Streptavidin Bead compared to competitor product



Bioinformatics Software

DESCRIPTION

As a part of Celemics' intellectual property, a unique NGS bioinformatics pipeline is developed to process and analyze massive amounts of genomic data into a readable format with clinically significant biomarkers obtained through Next Generation Sequencing.

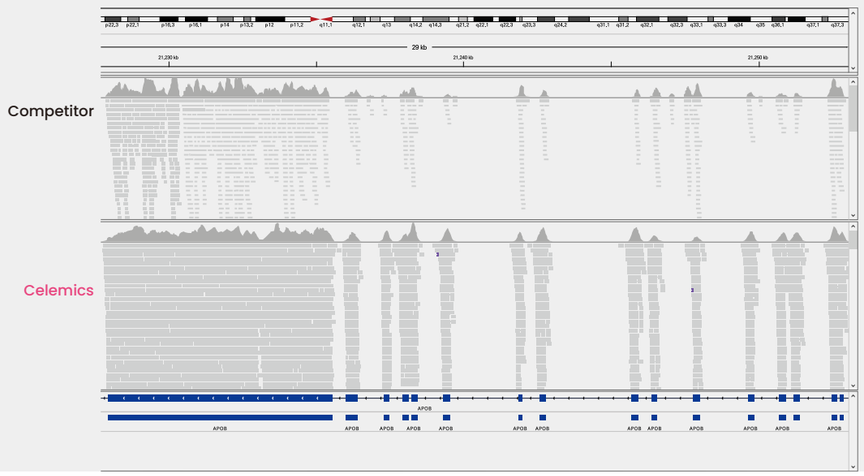
KEY FEATURES

1. Built-in service for all panel kits and services
2. Provides FASTQ to VCF and interpretation
3. Robust pipelines for detecting and analyzing all types of variants including SNV, Indel, CNV, Rearrangements, MSI, TMB, and ultra-low variants

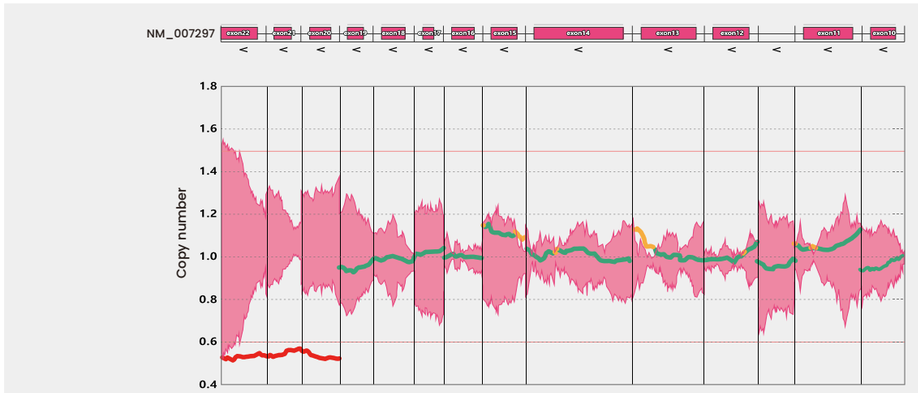
NGS DATA ANALYSIS PIPELINE

EXAMPLES OF BIOINFORMATICS ANALYSIS REPORT

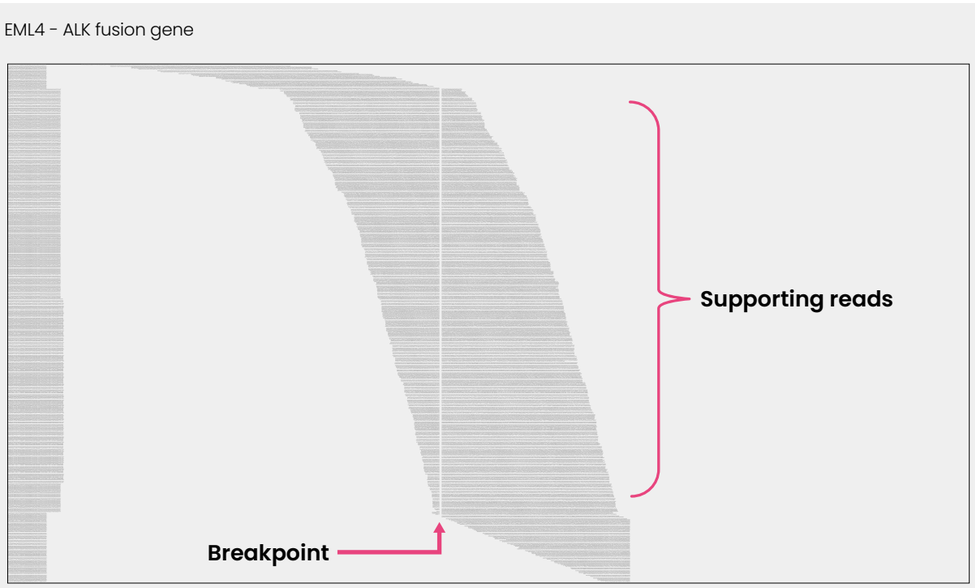
Comparison of IGV results from Celemics and competitor product



CNV Analysis Example – Deletion



Gene rearrangement analysis with FFPE samples



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