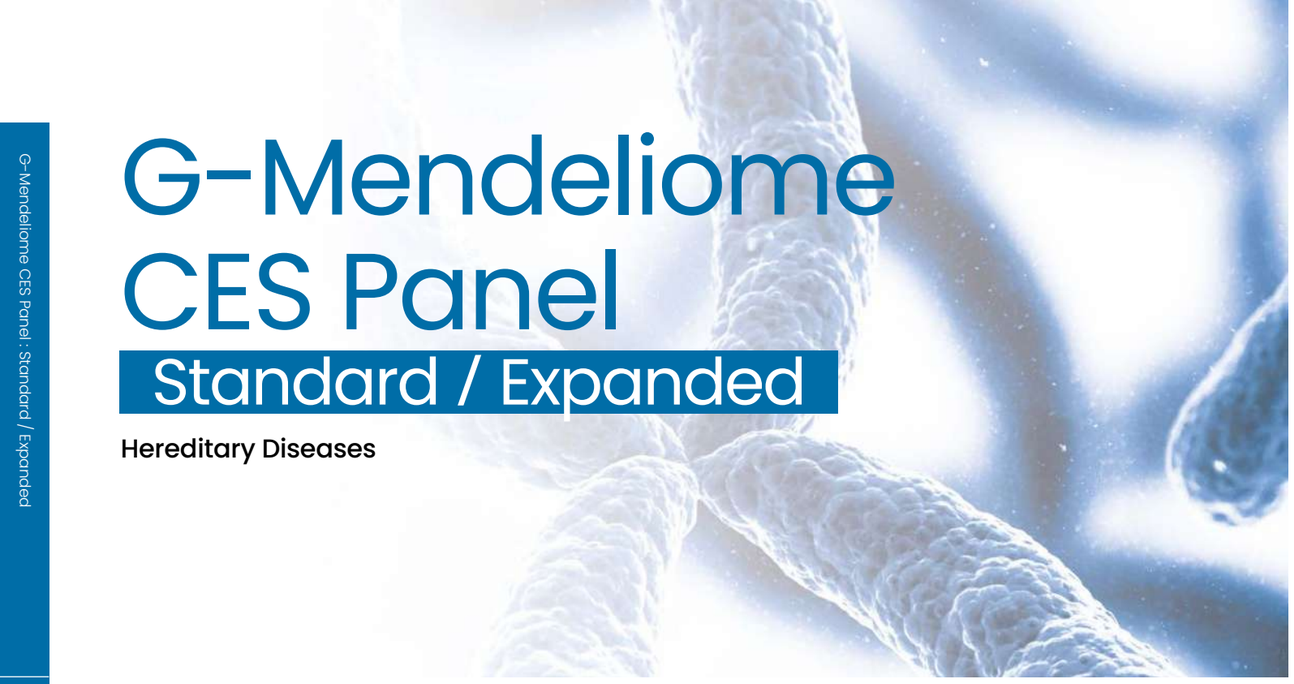


G-Mendeliome CES Panel

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

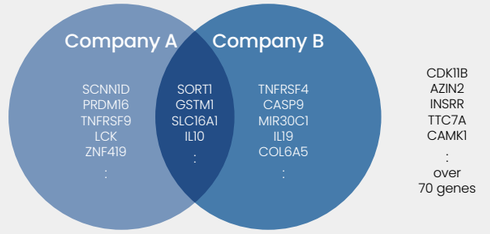
SPECIFICATION

Gene count*	5,508 / 7,513 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

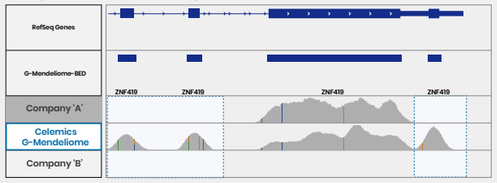
	Celemics	Company A	Company B
On-Target Read Ratio	82.8%	65.9%	80.8%

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

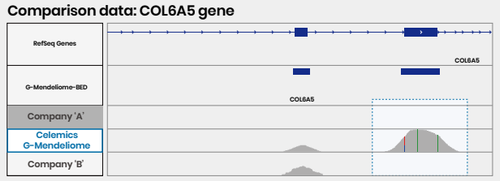


The CES panel was developed from the needs of GC Genome, the largest clinical NGS service provider in Korea, to solve the problem of poor diagnosis and high costs

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 14 to 293 genes
Covered regions	Whole CDS, hotspots
Target size	37-1,159 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

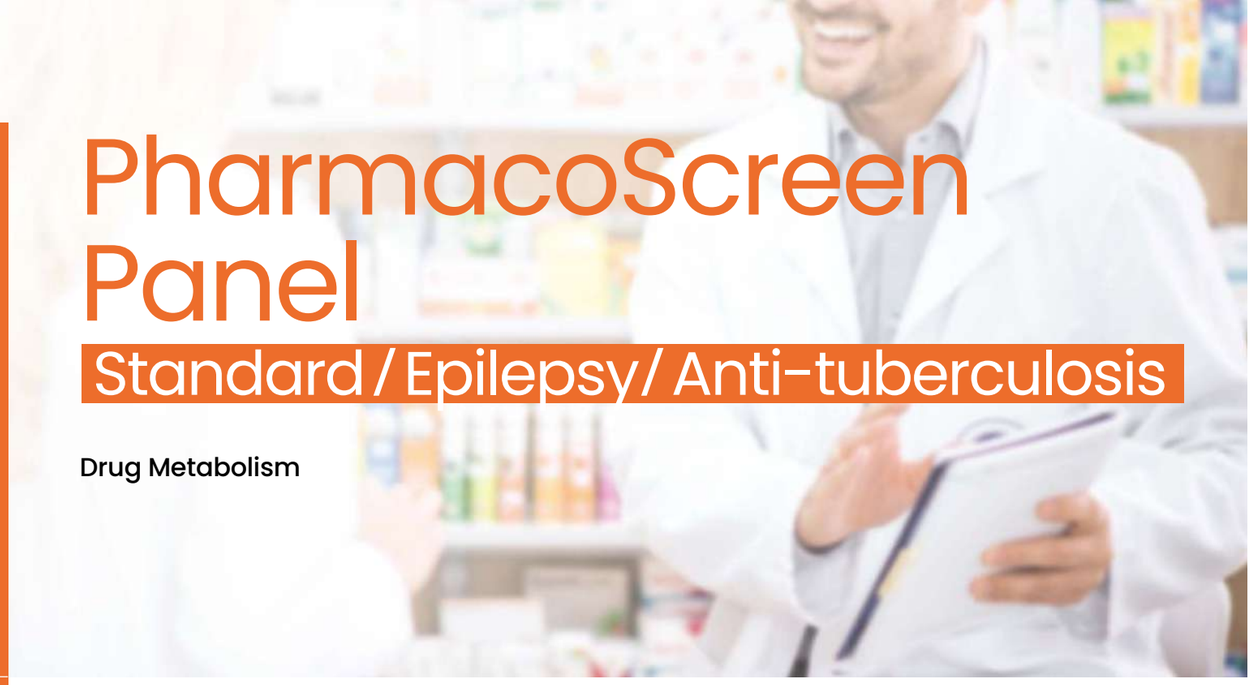
LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Related Diseases	Gene List								
Alzheimer-Parkinson-Dementia Panel	Alzheimer's disease, Parkinson's disease, Dementia, 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	Bleeding Disorder, Coagulation	AARS	ABCA1	ABCA13	ABCB11	ACTN1	ANKRD26	ANO6	AP3B1	BLOC1S3
		BLOC1S6	BRCA1	BRCA2	BRIP1	CD36	CDAN1	COG1	COL4A4	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	L2HGDH	LAMA3	LMAN1	LYST	MASTL	MCFD2	MLPH
		MPL	MYH9	MYO5A	NBEAL2	NBN	NDUFV3	NHP2	NOP10	NPHS2
		P2RY12	PALB2	PDP1	PLA2G4A	PLAU	PRF1	PRKACG	RAB27A	RAD51C
		RASGRP2	RBM8A	RPL11	RPL35A	RPL5	RPS10	RPSI9	RPS24	RPS26
		RPS7	RUNX1	SBDS	SEC23B	SERPIND1	SERPINE1	SERPINF2	SLC12A6	SLFN14
		SLX4	SOX6	SRC	SRP72	SRY	STIM1	SUMF1	TBXA2R	TBXAS1
		TDRD7	TERC	TERT	TINF2	UBE2T	VCAN	VIPAS39	VPS33B	VWF
		WAS	WIPF1	WNK1	XRCC2					
Cardiovascular Panel	Cardiac diseases	ABCC9	ABCG5	ABCG8	ACTA1	ACTA2	ACTC1	ACTN2	AKAP9	ALMS1
		ANK2	ANKRD1	APOA4	APOA5	APOB	APOC2	APOE	BAG3	BRAF
		CACNA1C	CACNA2D1	CACNB2	CALM1	CALR3	CASQ2	CAV3	CBL	CBS
		CETP	COL3A1	COL5A1	COL5A2	COX15	CREB3L3	CRELD1	CRYAB	CSRP3
		CTF1	DES	DMD	DNAJC19	DOLK	DPP6	DSC2	DSG2	DSP
		DTNA	EFEMP2	ELN	EMD	EYA4	FBN1	FBN2	FHL1	FHL2
		FKRP	FKTN	FXN	GAA	GATAD1	GCKR	GJA5	GLA	GPD1L
		GPIHBP1	HADHA	HCN4	HFE	HRAS	HSPB8	ILK	JAG1	JPH2
		JUP	KCNA5	KCND3	KCNE1	KCNE2	KCNE3	KCNH2	KCNJ2	KCNJ5
		KCNJ8	KCNQ1	KLF10	KRAS	LAMA2	LAMA4	LAMP2	LDB3	LDLR
		LDLRAP1	LMF1	LMNA	LPL	LTBP2	MAP2K1	MAP2K2	MIB1	MURC
		MYBPC3	MYH11	MYH6	MYH7	MYL2	MYL3	MYLK	MYLK2	MYO6
		MYOZ2	MYPN	NEXN	NKX2-5	NODAL	NOTCH1	NPPA	NRAS	PCSK9
		PDLIM3	PKP2	PLN	PRDM16	PRKAG2	PRKAR1A	PTPN11	RAF1	RANGRF
		RBM20	RYR1	RYR2	SALL4	SCN1B	SCN2B	SCN3B	SCN4B	SCN5A
		SCO2	SDHA	SEPN1	SGCB	SGCD	SGCG	SHOC2	SLC25A4	SLC2A10
		SMAD3	SMAD4	SNTA1	SOS1	SREBF2	TAZ	TBX20	TBX3	TBX5
		TCAP	TGFB2	TGFB3	TGFBR1	TGFBR2	TMEM43	TMPO	TNNC1	TNNI3
		TNNT2	TPM1	TRDN	TRIM63	TRPM4	TTN	TTR	TXNRD2	VCL
		ZBTB17	ZHX3	ZIC3						
Common Hereditary Cancer Panel	Medical checkup	APC	ATM	ATRX	BARD1	BMPR1A	BRAF	BRCA1	BRCA2	BRIP1
		CDH1	CDKN2A	CHEK2	EGLN1	EGLN2	EPAS1	EPCAM	FGFR1	FH
		H3F3A	HRAS	IDH2	KIF1B	KMT2D	MAX	MDH2	MEN1	MERTK
		MET	MLH1	MRE11	MSH2	MSH6	MUTYH	NBN	NF1	NF2
		PALB2	PMS2	POLD1	POLE	PRSS1	PTEN	RAD50	RAD51C	RAD51D
		RB1	RET	SDHA	SDHAF2	SDHB	SDHC	SDHD	SMAD4	SPINK1
		STK11	TMEM127	TP53	TSC1	TSC2	VHL	WT1		
Epilepsy Panel	Epilepsy	AARS	ABCA13	ABCB11	ADGRV1	ADSL	ALDH7A1	ALG13	ARHGEF15	ARHGEF9
		ARX	ASAH1	ATP1A2	ATP6AP2	CACNA1A	CASK	CDKL5	CHD2	CHRNA2
		CHRNA4	CHRNA7	CHRNB2	CLCN4	CLN3	CLN5	CLN6	CLN8	CNTNAP2
		COG1	COL4A4	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	L2HGDH	LAMA3	LGI1	MAGI2	MBD5	MECP2	MEF2C
		MFSD8	NDUFV3	NECAP1	NHLRC1	NPHS2	NR2F1	NRXN1	PCDH19	PDP1
		PIGA	PIGO	PIGQ	PIGV	PLCB1	PNKP	PNPO	POLG	PPT1
		PRICKLE1	PRICKLE2	PRRT2	QARS	RELN	SCARB2	SCN1A	SCN1B	SCN2A
		SCN8A	SCN9A	SERPIND1	SLC12A6	SLC13A5	SLC25A22	SLC2A1	SLC35A2	SLC6A8
		SLC9A6	SMS	SOX6	SPTAN1	SRPX2	SRY	ST3GAL3	STXBP1	SUMF1
		SYN1	SYNGAP1	SYNJ1	SZT2	TBC1D24	TCF4	TDRD7	TPP1	TSC1
		TSC2	UBE3A	VCAN	WDR45	WNK1	WWOX	ZEB2		

LIST OF PANELS FOR VARIOUS DISEASES

Panel Name	Related Diseases	Gene List								
Hearing Loss-Deafness Panel	Hearing loss, Deafness	CDH23	CLRN1	COCH	COL11A1	COL2A1	DIAPH1	EDNRB	EYA1	GJB2
		GJB6	KCNE1	KCNQ1	KCNQ4	MITF	MYO15A	MYO7A	OTOF	PAX3
		POU3F4	SIX5	SLC26A4	SNAI2	SOX10	TECTA	TMC1	TMIE	TMPRSS3
		USH1C	USH2A	WFS1						
Lymphoid Leukemia Panel	Acute lymphatic leukemia	AARS	ABCA13	ABCB11	ABL1	BRAF	BTG1	CDKN2A	COG1	COL4A4
		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	KMT2A	KMT2D
		KRAS	L2HGDH	LAMA3	LEF1	LMO1	MAPK1	NDUFV3	NF1	NOTCH1
		NPHS2	NRAS	NSD2	NT5C2	NUDT15	PAX5	PDP1	PHF6	PTEN
		PTPN11	RB1	RUNX1	SERPIND1	SETD2	SH2B3	SLC12A6	SOX6	SRY
		STAG2	STAT3	STAT5B	SUMF1	TBL1XR1	TCF3	TDRD7	TP53	TPMT
		VCAN	WNK1	WT1						
Lymphoma Panel	Lymphoma	AARS	ABCA13	ABCB11	ALK	ATM	B2M	BCL6	BIRC3	BRAF
		BTK	CARD11	CD79A	CD79B	COG1	COL4A4	CREBBP	CXCR4	DNMT1
		EGR2	EP300	EVC	EZH2	FAS	FAT4	FBXO11	FERMT1	FREM2
		GRM1	HPSE2	ID3	IDH2	IKBKB	IKZF1	IL12RB2	JAK3	KLF2
		L2HGDH	LAMA3	MYC	MYD88	NDUFV3	NFKBIE	NOTCH1	NOTCH2	NPHS2
		PDP1	PLCG1	PLCG2	POT1	PRDM1	RHOA	RPS15	RRAGC	SERPIND1
		SF3B1	SLC12A6	SOCS1	SOX6	SRY	STAT3	STAT5B	SUMF1	TBL1XR1
		TCF3	TDRD7	TET2	TNFAIP3	TNFRSF14	TP53	TP63	TRAF3	UBR5
		VCAN	WNK1	XPO1						
Lysosomal Storage Diseases Panel	Lysosomal storage disease	ABCD1	ACOX1	AGA	AGL	ALDOA	ALDOB	ARSA	ARSB	ATP13A2
		ATP7A	ATP7B	CLN3	CLN5	CLN6	CLN8	CTNS	CTSA	CTSD
		CTSF	DNAJC5	FUCA1	G6PC	GAA	GALC	GALE	GALK1	GALK2
		GALNS	GALT	GBA	GBE1	GJB2	GLA	GLB1	GNPTAB	GNPTG
		GNS	GRN	GUSB	GYS1	GYS2	HEXA	HEXB	HGSNAT	HPRT1
		HYAL1	IDS	IDUA	KCTD7	LDHA	LIPA	MAN2B1	MANBA	MCOLN1
		MFSD8	NAGA	NAGLU	NEU1	NPC1	NPC2	PEX1	PEX10	PEX12
		PEX13	PEX14	PEX16	PEX19	PEX2	PEX26	PEX3	PEX5	PEX6
		PFKM	PHKA2	PHKB	PHKG2	PPT1	PYGL	PYGM	SGSH	SLC17A5
		SLC2A2	SLC37A4	SMPD1	SUMF1	TPP1				
Metabolic Disorders Panel	Inborn errors of matabolism	ABCD1	ACAD8	ACADM	ACADS	ACADSB	ACADVL	ACAT1	AHCY	ARG1
		ASL	ASS1	AUH	BCKDHA	BCKDHB	BTD	CBS	CPS1	CPT1A
		CPT2	DBT	DECR1	DHCR7	DLD	ETFA	ETFB	ETFDH	FAH
		GALE	GALK1	GALT	GAMT	GATM	GCDH	GCH1	GNMT	HADH
		HADHA	HADHB	HLCS	**HMGCL**	**HPD**	**HSD17B10**	**IVD**	**LMBRD1**	**MAT1A**
		MCCC1	MCCC2	MLYCD	MMAA	MMAB	MMACHC	MMADHC	MMUT	MTHFR
		MTR	MTRR	OPA3	OTC	PAH	PCBD1	PCCA	PCCB	PTS
		QDPR	SLC22A5	SLC25A13	SLC25A20	SLC6A8	TAT	TAZ	TCN2	
Myeloid Leukemia Panel	Acute myeloid leukemia	ANKRD26	ASXL1	ATRX	BCOR	BCORL1	BRAF	CALR	CBL	CBLB
		CEBPA	CSF3R	DDX41	DNMT3A	ETV6	EZH2	**FLT3**	**GATA1**	**GATA2**
		HRAS	IDH1	IDH2	JAK2	JAK3	KDM6A	KIT	KRAS	MPL
		NOTCH1	NPM1	NRAS	PDGFRA	PHF6	PPM1D	PTPN11	RAD21	RUNX1
		SETBP1	SF3B1	SMC1A	SMC3	SRSF2	STAG1	STAG2	STAT3	TET2
		TP53	U2AF1	WT1	ZRSR2					
Neuromuscular Panel	Neuromuscular disease	AARS	ABCB7	ABCD1	ABHD12	ACAD9	ACADL	ACADM	ACO2	ACTA1
		ADCK3	AFG3L2	AGL	AIFM1	ALDH3A2	AMPD1	ANO10	ANO5	AP4B1
		AP4E1	AP4M1	AP4S1	AP5Z1	APTX	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	COL6A1	COL6A2	COL6A3	COLQ	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
		FA2H	FAM134B	FGD4	FGF14	FHL1	FIG4	FKRP	FKTN	FLNC
		FLVCR1	FRMD7	FUS	FXN	GAA	GAD1	GALC	**GAN**	**GARS**
		GBA2	GDAP1	GJB1	GJC2	GLA	GLE1	GNB4	GNE	GOSR2
		GPR143	GRID2	GRM1	GYS1	HADHA	HADHB	HINT1	HOXD10	HSPB1
		HSPB8	HSPD1	HSPG2	IGHMBP2	IKBKAP	ISPD	ITGA7	ITPR1	JPH3
		KBTBD13	KCNA1	KCNC3	KCND3	KCNE3	KCNJ10	KCNJ18	KIAA0196	KIF1A
		KIF1B	KIF1C	KIF5A	KLHL40	KLHL41	L1CAM	LAMA1	LAMA2	LARGE
		LDB3	LITAF	LMNA	LPIN1	LRSAM1	MARS	MARS2	MATR3	MED25
		MFN2	MPZ	MRE11A	MTM1	MTMR14	MTMR2	MTPAP	MTTP	MUSK
		MYF6	MYH2	MYH7	MYOT	NDRG1	NEB	NEFL	NGF	NIPA1
		NOP56	NTRK1	OPA1	OPA3	OPHN1	PABPN1	PANK2	PDK3	PDYN
		PEX7	PFKM	PGAM2	PHKA1	PHYH	PLEC	PLEKHG5	PLP1	PMM2
		PMP22	PNKP	PNPLA6	POLG	POLG2	POMGNT1	POMT1	POMT2	PRKCG
		PRPS1	PRX	PTF1A	PTRF	PYGM	RAB7A	RAPSN	REEP1	RNF216

Panel Name	Related Diseases	Gene List								
Neuromuscular Panel	Neuromuscular disease	RRM2B	RTN2	RUBCN	RYR1	RYR2	SACS	SBF2	SCN4A	SCN9A
		SEPN1	SETX	SGCA	SGCB	SGCD	SGCE	SGCG	SH3TC2	SIL1
		SLC12A6	SLC16A2	SLC1A3	SLC33A1	SLC39A4	SLC52A2	SLC9A1	SLC9A6	SMN1
		SNX14	SOD1	SPAST	SPG11	SPG20	SPG21	SPG7	SPTBN2	SPTLC1
		SPTLC2	STAC3	STUB1	SUCLA2	SYNE1	SYNE2	SYT14	TBP	TCAP
		TDP1	TECPR2	TGM6	TK2	TMEM240	TNNI2	TNNT1	TPM2	TPM3
		TPP1	TRIM32	TRPV4	TTBK2	TTN	TTPA	TTR	TUBB4A	TYMP
		VAMP1	VCP	VLDLR	VPS13A	VPS37A	VRK1	WFS1	WNK1	WWOX
		XK	YARS	ZFYVE26	ZFYVE27	ZNF592				
RASopathy Panel	RASopathies	BRAF	CBL	HRAS	KRAS	MAP2K1	MAP2K2	NF1	NRAS	PTPN11
		RAF1	RIT1	SHOC2	SOS1	SPRED1				
Retinitis Pigmentosa Panel	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	Short stature	AARS	ABCA13	ABCB11	ACTA2	ADAMTS10	ADAMTS2	ADAMTSL4	AGPS	ALPL
		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	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	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	PDP1	PEX7
		PHEX	PLOD1	POR	POU1F1	PPIB	PPP1CB	PRKG1	PROP1	PTPN11
		PYCR1	RAF1	RIN2	RIT1	RMRP	ROR2	RPS6KA3	RUNX2	SBDS
		SERPIND1	SERPINH1	SHOC2	SKI	SLCI2A6	SLC26A2	SLC2A10	SLC34A3	SLC35D1
		SLC39A13	SMAD3	SMARCAL1	**SMC1A**	**SMC3**	**SMS**	**SOS1**	**SOS2**	**SOX3**
		SOX6	SOX9	SPRED1	SRCAP	SRY	SUMF1	TDRD7	TGFB1	TGFB2
		TGFB3	TGFBR1	TGFBR2	THRB	TRIM37	TRIP11	TRPS1	TRPV4	TTC21B
		VCAN	WDR19	WDR35	WNK1	WRN				
Skin Disorder Panel	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	Somatic cancer	ABL1	AKT1	ALK	APC	ATM	BRAF	BRCA1	**BRCA2**	**CDH1**
		CDKN2A	CSF1R	CTNNB1	DLC1	EGFR	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			



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

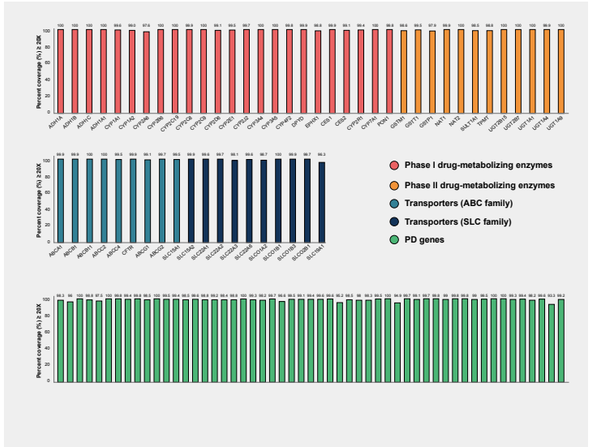
The panel performance test resulted in 99.9% specificity and 99.7% sensitivity.

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*	122 genes
Covered region	Whole CDS, UTR (-50 bp, +10 bp)
Target Size	534 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

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.

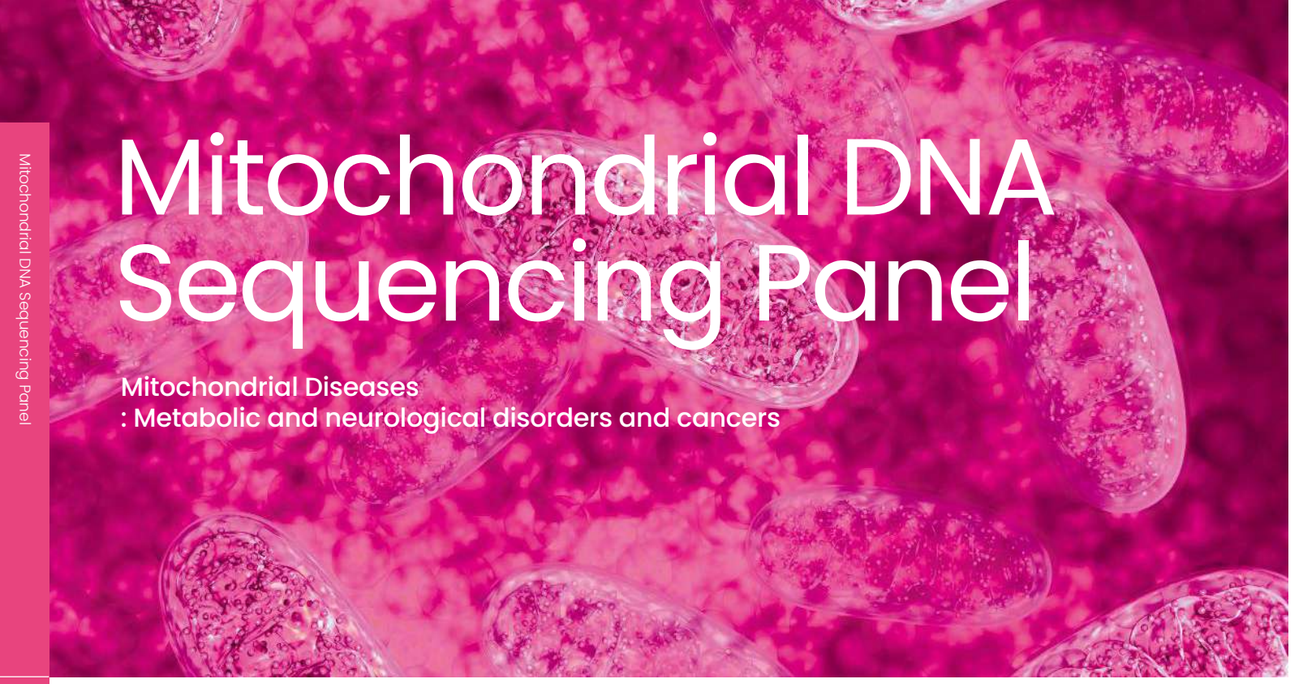
SPECIFICATION

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											



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

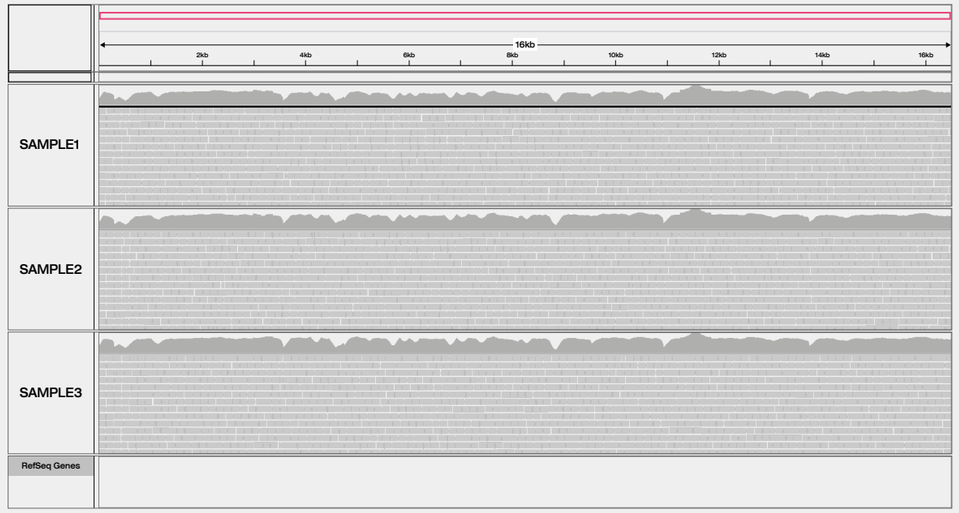
SPECIFICATION

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