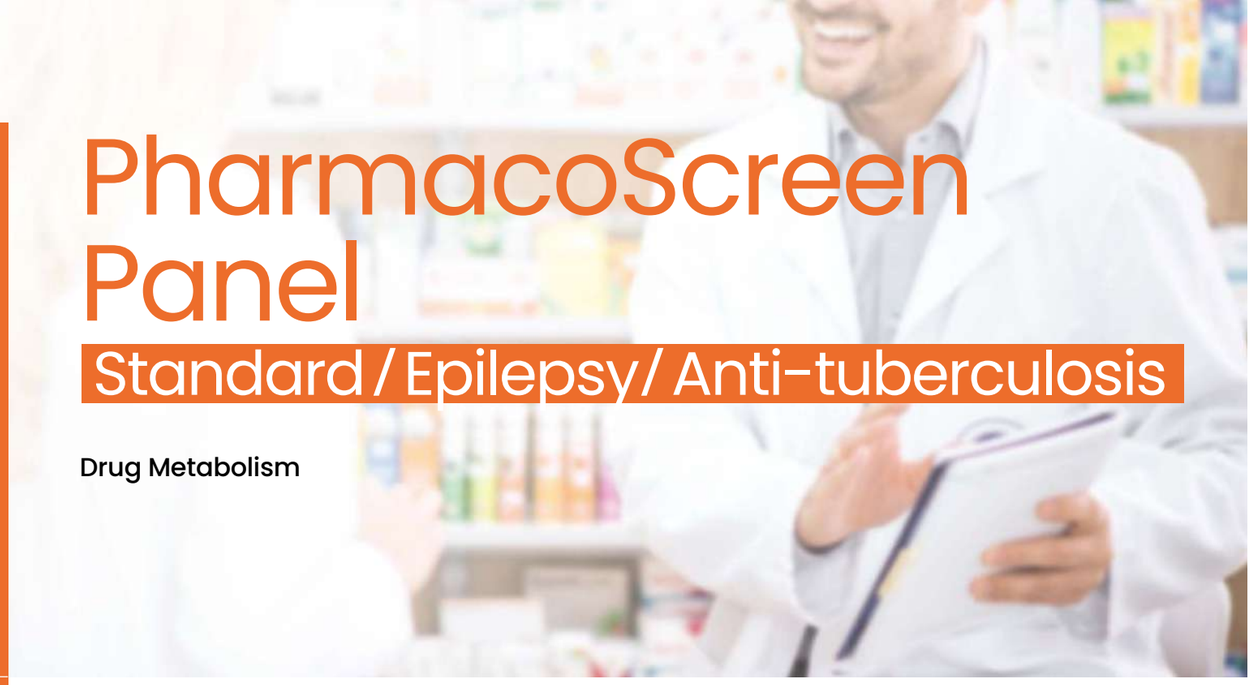


PharmacoScreen Panel

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

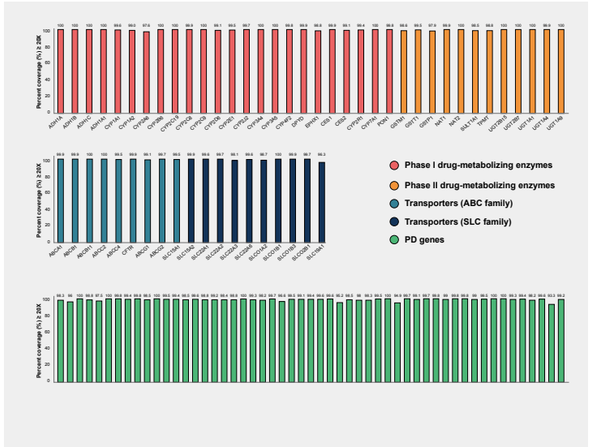
The panel performance test resulted in 99.9% specificity and 99.7% sensitivity.

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