SOPHiA GENETICS™

SOPHiA DDM™ Community Extended Pharmacogenomics Solution

Developed and tested by genomic experts, the SOPHiA DDM™ Community Extended Pharmacogenomics Solution combines a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

The SOPHiA DDM™ Community Extended Pharmacogenomics Solution enables the analysis of 74 genes involved in drug metabolism, efficacy, and toxicity. It covers both coding and non-coding regions with optimized probe design for high on-target performance and uniform coverage, including GC-rich areas like first exons. The solution includes CNV and star allele calling, genotyping of key HLA alleles, and coverage of key mitochondrial DNA hotspots, promoters, and UTRs. Results are delivered in PGx-specific output files with clear genotype reporting.

Gene Panel

Core content
ABCB1, ABCC2, ABCC4, ABCG2, ACE, ADRB1, ADRB2, ALDH2, ALDH5A1, BCHE, CACNA1S, CDA, CEP72, CES1, CES2, COMT, CYP1A2, CYP2A6, CYP2A13, CYP2B6, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP2D7, CYP3A4, CYP3A5, CYP4F2, DPYD, DRD2, F2, F5, G6PD, GGCX, GRIK4, GSTP1, HCP5, HFE, HLA-A, HLA-B, HTR2A, HTR2C, IFNL3, IFNL4, ITPA, MT-RNR1, MTHFR, NAT2, NFIB, NR1I2, NR1I3, NUDT15, OPRM1, P2RY12, PEAR1, PON1, POR, PSORS1C1, RARG, RYR1, SLC22A1, SLC28A3, SLC6A4, SLCO1B1, SRY[1], TPMT, TYMS, UGT1A1, UGT1A4, UGT1A6, UGT2B15, UGT2B7, VKORC1, XRCC1

HLA genotyping[2]
HLA-A, HLA-B

Mitochondrial hotspots
MT-RNR1[3]

Star allele coverage (15 genes)[4]
CYP1A2, CYP2A6, CYP2A13, CYP2B6, CYP2C8, CYP2C9, CYP2C19, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, NAT2, NUDT15, SLCO1B1

Variants Called

SNVs
Indels
CNVs
Star alleles

Library Preparation

Starting material
50 ng DNA

Sample type
Blood

Library preparation time
Day 1:
Library Preparation
Day 2:
Capture and Sequencing

Total time: 2 days

Sequencer Compatibility

Reads per sample
4 million[5]

Samples per run[6]
12 for Illumina Miseq™ i100 25M flowcell
24 for Illumina Miseq™ i100 50M flowcell
48 for Illumina Miseq™ i100 100M flowcell
12 for Illumina MiSeq® V3
48 for Illumina NextSeq™ 500/550 Mid Output Kit
192 for Illumina NextSeq™ 500/550 High Output Kit
48 for Illumina NextSeq™ 1000/2000 P1
192 for Illumina NextSeq™ 1000/2000 P2
576 for Illumina NextSeq™ 1000/2000 P3
864 for Illumina NextSeq™ 1000/2000 P4

Analytical Performance[7]

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting and annotating multiple types of genomic variants in all genes of the panel.

Analysis time from FASTQ: 3 hours

	Observed	Lower 95% CI
Sensitivity	100%	99.36%
Accuracy	100%	99.99%
Precision	100%	96.36%
Specificity	100%	99.99%
Repeatability	99.80%	99.32%
Reproducibility	99.85%	99.66%
Coverage uniformity	99.37%	
Average % of target region with depth >200x	99.14%	
Consistency with ground-truth on *CYP2D6* star-allele calling[8]	100%	

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

Dedicated features in SOPHiA DDM™ reduce the complexity of determining the significance of genomic variants and facilitate the interpretation process, thus reducing turnaround time:

- **Variant annotation:** Using International guidelines such as ACMG and information from curated databases such as ClinVar
- **Variant filtering:** Virtual Panels and intuitive filters for speedy screening of relevant variants
- **Star allele calling:** An intuitive interface that presents wild-type and star alleles, displaying expandable details when selected.
- **Variant status report:** Pharmacogenomics-specific output files with genotype status

Global support at every step

We offer local support anywhere in the world. Our dedicated bioinformaticians help save time and resources, ensuring fast resolution of workflow disruptions.

Secure and unlimited data storage

The SOPHiA DDM™ Platform keeps data safe by applying the highest industrial standards while complying with local data security policies.

Access to the SOPHiA GENETICS Community

In the SOPHiA DDM™ Platform, experts from hundreds of healthcare institutions interpret their results and flag the pathogenicity level of variants according to their knowledge and experience. These expert perspectives can assist users in determining the true pathogenicity of their detected variants.

Product codes
CS2587ILLRGLY10-16; CS2587ILLRGLY10-32;
CS2587ILLRGLY10-48; CS2587ILLRGLY10-96

1. *SRY* is a control gene, not clinically relevant in the context of PGx.
2. High resolution *HLA* genotyping will become available later in the year.
3. CNV calling not available for mitochondrial hotspots.
4. Genes with star allele coding according to PharmVar nomenclature.
5. Recommendations based on the SOPHiA GENETICS Universal Library Prep protocol.
6. When working with more than 100 Gb or 96 samples, data upload to SOPHiA DDM™ is via the Command Line Interface (CLI) Tool.
7. Excluding the star allele calling and *CYP2D6* analytical modules. Analytical performance metrics were calculated from 2 runs with 35 samples on an Illumina MiSeq® instrument.
8. Data from 63 Coriell samples. The ground-truth is from the GeT-RM Program, U.S. CDC (www.cdc.gov/lab-quality/php/get-rm/) excluding reverse-hybrid.

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