SOPHiA GENETICS™

SOPHiA GENETICS™ Pharmacogenomics Community Panel

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

Main Features

The SOPHiA GENETICS™ Pharmacogenomics Community Panel covers coding and non-coding regions of 41 genes associated with pharmacogenomics. Probe design is highly optimized to guarantee a high on-target reads percentage and coverage uniformity even in GC-rich regions, including the first exon. A specialized analytical module covers CNVs, star alleles, and promoters/UTRs in *CYP2D6*, and the pharmacogenomics-specific output files report the genotype status of all genes in the panel.

Gene Panel	Variants Called	Recommendations	Wet Lab
ABCB1, ABCC2, ABCC4, ABCG2, ACE, ADRB1, ADRB2, BCHE, CDA, COMT, CYP1A2, CYP2B6, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, DRD2, G6PD, HTR2A, ITPA, MTHFR, NAT2, NR1I2, NR1I3, NUDT15, OPRM1, P2RY12, POR, RYR1, SLC22A1, SLCO1B1, TPMT, TYMS, UGT1A1, UGT1A4, UGT2B7, VKORC1	SNVs Indels CNVs Star alleles (*CYP2D6* only)	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run for > 50x coverage depth** 48 for Illumina MiSeq® V3 32 for Illumina MiSeq® V2 48 for Illumina MiniSeq™ High Output Kit 16 for Illumina MiniSeq™ Mid Output Kit	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Hands-on library preparation time: 8 hours**

Analytical Performance[1]

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

Analysis time from FASTQ: 2 hours

	Observed	Lower 95% CI
Sensitivity	100%	96.34%
Accuracy	100%	99.99%
Precision	100%	96.34%
Specificity	100%	99.99%
Repeatability	99.98%	99.97%
Reproducibility	99.98%	99.97%
Average on-target rate	67%	
Coverage uniformity	99.78%	
Average % of target region with depth >200x	99.83%	

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 Cascading Filters for speedy screening of relevant variants
- **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. In addition, our Set Up Program provides assistance with assay set up for a fast and worry-free transition to routine testing.

Secure and unlimited data storage

Access to the SOPHiA DDM™ Platform is restricted to registered users only. The Platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with your 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. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.

Product code: CS2532ILLRGLY10

1. Excluding *CYP2D6* analytical module. Analytical performance metrics were calculated from 2 runs with 12 samples on an Illumina MiSeq® instrument, and 3 runs with 12 samples on an Illumina MiniSeq™ instrument.

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