SOPHiA GENETICS™

SOPHiA Solid Tumor Solution

The genomic application that characterizes the complex mutational landscape of the major solid tumors by combining a capture-based target enrichment kit with the analytical performance and advanced features of the SOPHiA DDM™ platform.

Main Features

SOPHiA Solid Tumor Solution covers **42 genes** associated with solid tumors, including lung, colorectal, skin, and brain cancers. It also covers 6 unique loci to detect MSI[1] status associated with colorectal cancer. Probe design is optimized to guarantee high on-target rate and coverage uniformity throughout the entire target regions.

Gene Panel	Variants Called	Recommendations	Wet Lab
AKT1 (3), ALK (21-25), BRAF (11,15), CDK4 (2), CDKN2A (1,2,3), CTNNB1 (3), DDR2 (17), DICER1 (24,25), EGFR (18-21), ERBB2 (8,17,20), ERBB4 (10,12), FBXW7 (7-11), FGFR1 (12,14), FGFR2 (7,12,14), FGFR3 (7,9,14,16), FOXL2 (1*), GNA11 (4,5), GNAQ (4,5), GNAS (8), H3F3A(2*), H3F3B (2*), HIST1H3B (1), HRAS (2-4), IDH1 (4), IDH2 (4), KIT (8-11,13,17,18), KRAS (2-4), MAP2K1 (2,3), MET (2,14-20), MYOD1 (1), NRAS (2-4), PDGFRA (12,14,18), PIK3CA (2*,3,6*,8,10,21), PTPN11 (3), RAC1 (3), RAF1 (7,10,12,13*,14*,15*), RET (11,13,15,16), ROS1 (38*,41*), SF3B1 (15-17), SMAD4 (8-12), TERT (promoter*,1*,8*,9*,13*), TP53 (2-11)* **Hotspots only*	• SNVs and Indels in all genes of the panel • *MET* large deletions • *TERT* promoter mutations C228T and C250T • MSI[1] status in 6 unique loci associated with colorectal cancer: BAT-25, BAT-26, CAT-25, NR-21, NR-22 and NR-27 • Gene amplification events in 24 genes: *ALK, BRAF, CDK4, CDKN2A, EGFR, ERBB2, FBXW7, FGFR1, FGFR2, FGFR3, HRAS, KIT, KRAS, MET, MYOD1, NRAS, PDGFRA, PIK3CA, RAF1, ROS1, RET, SF3B1, TERT and TP53*	**Starting material** ≥10 ng (50 ng recommended) **Sample type** FFPE, fresh-frozen tissue **Samples per run for 1000x coverage depth / Sequencer (Flow Cell/Ion Chip Kit)[2]** 8 / Illumina MiniSeq™ Mid Output Kit (2x150) 24 / Illumina MiSeq® v3 (2x300bp)* 12 / Ion Torrent™ Ion S5 System (Ion 530™) *2x150-cycle sequencing run (paired-end) is recommended	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time:** 1.5 days

Analytical Performance

The SOPHiA DDM™ platform analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic alterations in one unique experiment.

Analysis time from FASTQ: from ≥ 4 hours[3]

	Observed (%)	Lower 95% CI
Sensitivity	98.77	93.31
Specificity	100	99.92
Accuracy	99.97	99.85
Precision	100	96.25
Repeatability	96.45	96.41
Reproducibility	89.13	89.05
Coverage uniformity	98.70	92.50*

Performance values have been calculated based on SNVs and Indels only in 150 samples processed on Illumina MiSeq® using KAPA™ HyperPlus KK8512 library preparation reagents.
*5% quantile

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

The platform helps users to immediately focus on relevant genomic alterations. Several features facilitate their interpretation process:

- Hotspot screening
- Algorithm-supported variant pre-classification
- Fully customizable filters
- Comprehensive report

Confident decision-making

The SOPHiA DDM™ platform integrates the OncoPortal™.
This feature provides the latest scientific evidence on the significance of each genomic alteration to support informed decision-making for research purposes.

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 fast and worry-free transition to routine testing.

Secure and unlimited data storage

The SOPHiA DDM™ platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with local data security policies.

Access to the SOPHiA GENETICS community

Through the SOPHiA DDM™ platform genomics experts from hundreds of healthcare institutions interpret their findings and flag the pathogenicity level of variants. This highly valuable information enriches the variant knowledge base and is safely shared among the members of the community, supporting their decision-making process for research purposes.

1. MSI: Microsatellite Instability
2. Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform.
3. Analysis time may vary depending on the number of samples multiplexed and server load.

For Research Use Only. Not for Use in Diagnostic Procedures.