

SOPHiA SOLID TUMOR SOLUTION™

The genomic application that bundles a capture-based target enrichment kit with the analytical power of SOPHiA™ AI and full access to the SOPHiA DDM™ platform.



SOPHiA Solid Tumor Solution covers 42 relevant genes associated with solid tumors such as 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

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

Recommendations

Starting material: 10 ng min (50 ng recommended)

Sample source: FFPE, fresh-frozen tissue

Samples per run: Depending on sequencing platform⁽²⁾

Sequencer	Flow Cell/ Ion Chip Kit	Recommended samples per run (for 1000x coverage depth)
Illumina MiniSeq™	Mid Output Kit (2x150bp)	8
Illumina MiSeq®	v3 (2x300bp)*	24
Ion Torrent™ Ion S5™ System	Ion 530™ Chip	12

*2x150-cycle sequencing run (paired-end) is recommended

Wet lab

Day 1: Library Preparation

Day 2: Capture and Sequencing

Total library preparation time: 1.5 days

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying genomic alterations to support experts with their data-informed decision making.

SOPHiA accurately detects:

- SNVs and Indels in all the genes of the panel
- *MET* large deletions
- *TERT* promoter mutations C228T and C250T
- MSI 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*, *RET*, *ROS1*, *SF3B1*, *TERT* and *TP53*

SOPHiA reaches advanced analytical performance⁽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	98.7%	92.5%*

*5% quantile

Analysis time from FASTQ files: 4 hours⁽⁴⁾

- (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) Performance values have been calculated based on SNVs and Indels in samples processed on Illumina MiSeq[®] using KAPA[™] HyperPlus KK8512 library preparation reagents.
- (4) Analysis time may vary depending on samples multiplexed and server load.

The results are presented in SOPHiA DDM, the platform of choice for experts performing genomic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic alterations. Data are kept safe by applying the highest industrial standards of encryption.

Main features

SOPHiA DDM offers several features that make variant analysis more efficient, such as hotspot screening which streamlines the visualization of mutated and wild type hotspot positions. With variant pre-classification and customized filtering options, experts can easily accelerate the data interpretation process.

OncoPortal™

SOPHiA DDM integrates the OncoPortal, a decision support functionality based on precision medicine intelligence. It enables experts to access relevant therapeutic, prognostic and diagnostic information to help determine actionability and clinical significance of detected genomic alterations. Moreover, the OncoPortal uses genes and disease association to maximize clinical trial matching.

Access to SOPHiA's Community

In SOPHiA DDM, experts from hundreds of healthcare institutions interpret the 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.

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