SOPHiA™

SOPHiAGENETICS.COM

SOPHiA SOLID TUMOR SOLUTION™ CE IVD

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

SMART KIT DESIGN

The STS application covers 42 clinically relevant genes associated with solid tumors such as lung, colorectal, skin and brain cancers. It also covers 6 unique loci to detect MSI status associated with colorectal cancer. Probe design is highly-optimized to provide exceptional coverage uniformity throughout the entire target regions, resulting in superior data quality.

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: 10ng min (50ng recommended)
Sample source: FFPE, fresh-frozen tissue
Samples per run: Depending on sequencing platform(1)

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

Wet lab

Day 1: Library Preparation
Day 2: Capture and Sequencing
Total library preparation time: 1.5 days

ANALYTICAL POWER

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying genomic alterations to help experts streamline data interpretation.

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, ROS1, RET, SF3B1, TERT* and *TP53*

SOPHiA reaches clinical-grade performance(2):

	Observed	Lower 95% CI
Sensitivity	100%	94.82%
Reproducibility	99.97%	99.92%
Repeatability	99.99%	99.95%
Accuracy	100%	95.77%
Precision	100%	93.18%
Coverage uniformity	98.6%	93.5%*

*5% quantile

Analysis time from FASTQ files: 4 hours(3)

UNIVERSAL PLATFORM

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

Main features

SOPHiA DDM offers several features that make variant analysis more efficient. 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 databases to determine the actionability and clinical significance of genomic alterations. Moreover, the OncoPortal uses inclusion and exclusion criteria to maximize clinical trial matching that may benefit the patient, both locally and at the global level.

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.

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform
(2) Performance values have been calculated based on SNVs and Indels in 394 samples processed on Illumina MiSeq®
(3) Analysis time may vary depending on samples multiplexed and server load