SOPHiA™

SOPHiAGENETICS.COM

SOPHiA SOLID TUMOR PLUS SOLUTION™

The genomic application that bundles DNA target capture and RNA target amplicon kits with the analytical power of SOPHiA™ AI and full access to the SOPHiA DDM™ platform.

SMART KIT DESIGN

SOPHiA Solid Tumor Plus Solution targets DNA variants and RNA transcripts of fusion genes associated with solid tumors such as lung, colorectal, skin, and brain cancers. The DNA panel covers 42 genes and the RNA panel targets 137 gene fusions. Probe design is optimized to guarantee high on-target rate and coverage uniformity throughout the entire target regions.

DNA 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*

RNA fusion panel

137 RNA fusions, involving genes such as *ALK, BRAF, EGFRvIII, FGFR1, FGFR2, FGFR3, NTRK1, NTKR3, PPARG, RET and ROS1*

Recommendations

Starting material: 10 ng (50 ng recommended) DNA, 100-200 ng RNA
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 MiSeq®	v3 (2x300bp)*	24 DNA + 24 RNA
Illumina NextSeq® 500/550	Mid Output Kit v2 (2x150bp)	48† DNA + 48† RNA

*2x150-cycle sequencing run (paired-end) is recommended
†Maximum number of indices available

Wet lab

Day 1: DNA and RNA Library Preparation
Day 2: Capture & Sequencing of DNA and RNA libraries in 1 run
Total library preparation time: 1.5 days for DNA, 6 hours for RNA

ANALYTICAL POWER

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 genes of the panel
- *TERT* promoter mutations C228T and C250T
- MSI(2) 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*
- 137 RNA fusions in addition to *MET* exon 14 skipping and *EGFR* variant III

SOPHiA reaches advanced analytical performance(3):

	DNA		
	Observed	Lower 95% CI	RNA†
Sensitivity	98.77%	93.31%	100%
Specificity	100%	99.92%	
Accuracy	99.97%	99.85%	
Precision	100%	96.25%	> 93%
Repeatability	96.45%	96.41%	
Reproducibility	89.13%	89.05%	
Coverage uniformity	98.7%	92.5%*	

Analysis time from FASTQ files: 4 hours(4)

*5% quantile
†Calculated on reference samples
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UNIVERSAL PLATFORM

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.

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).

(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) MSI: Microsatellite Instability
(3) Performance values have been calculated based on SNVs and Indels in samples processed on Illumina MiSeq®.
(4) Analysis time may vary depending on the number of samples multiplexed and server load.