

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.



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 also 6 unique loci to detect MSI⁽¹⁾ status associated with colorectal cancer. The RNA fusion panel target 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*), GNAI1 (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 such as ALK, BRAF, EGFRvIII, FGFR1, FGFR2, FGFR3, NTRK1, NTRK3, PPARC, RET and ROS1

Recommendations

Starting material: 10 ng (50 ng recommended) DNA, 100-200ng RNA

Sample source: FFPE, fresh-frozen tissue

Samples per run: Depending on sequencing platform⁽²⁾

Sequencer	Flow Cell Kit (sequencing run)	Recommended samples per run (for 1000x coverage depth)
Illumina MiSeq®	v3 (2x150bp)	24 DNA + 24 RNA
Illumina NextSeq® 500/550	Mid Output Kit v2 (2x150bp)	48* DNA + 48* RNA

*Maximum number of indices available

Wet lab

Day 1: RNA and DNA 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

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

SOPHiA accurately detects:

- SNVs and Indels in all 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*
- 137 RNA fusions such as *MET* exon 14 skipping and *EGFR* variant III

SOPHiA reaches advanced analytical performance⁽³⁾:

	DNA		RNA**
	Observed	Lower 95% CI	
Sensitivity	100%	94.82%	100%
Reproducibility	99.97%	99.92%	
Repeatability	99.99%	99.95%	
Accuracy	100%	95.77%	
Precision	100%	93.18%	> 93%
Coverage uniformity	98.6%	93.5%*	

Analysis time from FASTQ files: 4 hours⁽⁴⁾

*5% quantile

** Calculated on reference samples

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 is 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 databases to determine the actionability and significance of genomic alterations. Moreover, the OncoPortal uses inclusion and exclusion criteria to maximize clinical trial matching, 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.

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(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) A total of 394 samples were processed on MiSeq® to obtain the above-mentioned metrics. Performance values have been calculated on SNVs and Indels only

(4) Analysis time may vary depending on the number of samples multiplexed and server load
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