SOPHiA GENETICS™

SOPHiA DDM™ Solid Tumor Plus Solution

The genomic application that characterizes the complex mutational landscape of the major solid tumors by combining DNA target capture and RNA target amplicon with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

SOPHiA DDM™ Solid Tumor Plus Solution (STS+) targets DNA variants and RNA transcripts of fusion genes associated with solid tumors, including lung, colorectal, skin, and brain cancers. The DNA panel covers **42 genes** including 6 unique loci to detect MSI status associated with colorectal cancer, and the RNA panel targets **136 clinically relevant gene fusions**. 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*

RNA Gene Fusion Panel

136 clinically relevant RNA gene fusions, involving genes such as ALK, BRAF, EGFR, FGFR1, FGFR2, FGFR3, NTRK1, NTKR3, PPARG, RET and ROS1

View RNA gene fusion panel list

Variants Called

- SNVs and Indels
- *TERT* promoter mutations C228T and C250T
- MSI status in BAT-25, BAT-26, CAT-25, NR-21, NR-22 and NR-27
- 136 gene fusions in addition to *MET* exon 14 skipping and *EGFR* variant III
- Gene amplification in 24 genes including *EGFR*, *ERBB2*, *FGFR2* and *MET*

Recommendations

Starting material
10-50 ng DNA
100-200 ng RNA

Sample type
FFPE, fresh-frozen tissue

Samples per run / Sequencer[1]
24 DNA + 24 RNA on Illumina MiSeq® v3 (2x300bp)
48 DNA + 48 RNA on Illumina NextSeq® 500/550 Mid-Output (2x150bp)

Wet Lab

Day 1:
DNA and RNA Library Preparation

Day 2:
Capture and Sequencing of DNA and RNA libraries in 1 run

Total library preparation time:
1.5 days for DNA,
6 hours for RNA

Analytical Performance*

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting, annotating and pre-classifying genomic alterations and RNA fusions in the genes of this panel.

Analysis time from FASTQ: from 4 hours[2]

	DNA		RNA
	Observed (%)	Lower 95% CI	Observed (%)
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	98.70	92.50*	

Performance values have been calculated based on SNVs and Indels only in 150 FFPE samples processed on Illumina MiSeq® for DNA and reference samples for RNA.
*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

Product code:
BS0115ILLRSMY10[3]

Confident decision-making

The OncoPortal™ Plus add-on module for SOPHiA DDM™ Platform matches tumor molecular profiles with clinical associations and available clinical trials, leveraging expertly curated evidence powered by JAX-CKB. After interpretation, the flexible reporting tools enable users to prepare push-button, comprehensive reports that are customizable to their needs.

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 MaxCare Program provides assistance with assay set up for fast and worry-free transition to routine testing.

Access to the SOPHiA GENETICS Community

Through the SOPHiA DDM™ Platform genomics experts from from hundred 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.

CI, confidence interval; FFPE, formalin-fixed paraffin-embedded; Indels, insertions/deletions; MSI, microsatellite instability; NGS, next-generation sequencing; SNVs, single nucleotide variants.
1. Number of samples per run is recommended for 1000x coverage depth. Sequencing recommendations and pipelines for other sequencing kits and instruments are available upon request. Delivery time may vary according to the selected sequencing platform.
2. Analysis time may vary depending on the number of genes, samples multiplexed, and server load.
3. Product code for SOPHiA GENETICS™ Universal Library Prep (ULP), replacing BS0115ILLRSMY05 and BS0115ILLRSML03.
Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB).

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

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RNA Gene Fusion Panel

EML4(13)–ALK(20), EML4(20)–ALK(20), EML4(6)–ALK(20), TPM3(8)–ALK(20), RANBP2(18)–ALK(20), CLTC(31)-ALK(20), ATIC(7)-ALK(20), EML4(18)-ALK(20), EML4(14)-ALK(20), KIF5B(24)-ALK(20), VCL(16)-ALK(20), GCC2(19)-ALK(20), FN1(20)-ALK(19), EML4(2)-ALK(20), EML4(6)-ALK(19), EML4(19)-ALK(20), DCTN1(26)-ALK(20), KIAA1549(16)-BRAF(9), KIAA1549(15)-BRAF(9), KIAA1549(16)-BRAF(11), SND1(10)-BRAF(9), AGK(2)-BRAF(8), KIAA1549(15)-BRAF(11), FAM131B(3)-BRAF(9), CCDC6(1)-BRAF(9), OSBPL9(11)-BRAF(10), ZNF207(3)-BRAF(10), GIPC2(3)-BRAF(10), MYH14(19)-BRAF(10), PJA2(7)-BRAF(11), PRKAR2B(1)-BRAF(10), SND1(9)-BRAF(11), SND1(9)-BRAF(9), SND1(10)-BRAF(11), SND1(14)-BRAF(9), SND1(17)-BRAF(11), KIAA1549(19)-BRAF(9), KIAA1549(18)-BRAF(10), KIAA1549(16)-BRAF(10), KIAA1549(13)-BRAF(9), MKRN1(4)-BRAF(11), MKRN1(4)-BRAF(9), FAM131B(2)-BRAF(9), CUL1(7)-BRAF(9), AGAP3(9)-BRAF(9), AGAP3(10)-BRAF(9), RBM33(3)-BRAF(11), PRKAR1B(9)-BRAF(9), PHTF2(16)-BRAF(9), SCRIB(27)-BRAF(11), CDK5RAP2(13)-BRAF(9), CCDC6(1)-RET(12), NCOA4(7)-RET(12), KIF5B(15)-RET(12), PRKAR1A(7)-RET(12), KIF5B(16)-RET(12), KIF5B(18)-RET(12), RASSF4(3)-RET(12), NCOA4(8)-RET(12), NCOA4(11)-RET(12), CCDC6(1)-RET(10), TFG(4)-RET(11), PDCD10(4)-RET(11), SLC34A2(4)-ROS1(32), CD74(6)-ROS1(34), CD74(6)-ROS1(33), EZR(9)-ROS1(33), SDC4(2)-ROS1(32), CD74(7)-ROS1(34), GOPC(8)-ROS1(35), TPM3(9)-ROS1(35), ZCCHC8(2)-ROS1(36), GOLGB1(9)-ROS1(35), SLC4A4(21)-ROS1(34), CD74(7)-ROS1(33), SLC16A10(3)-ROS1(3), CEP85L(2)-ROS1(18), PLAGL1(6)-ROS1(32), EZR(9)-ROS1(34), EZR(9)-ROS1(32), FGFR2(17)-KIAA1217(3), FGFR2(17)-BICC1(10), FGFR2(17)-BICC1(16), FGFR2(17)-BICC1(3), FGFR2(17)-NRBF2(4), FGFR2(17)-RABGAP1L(20), FGFR2(17)-LAMC1(27), FGFR2(17)-KIF14(19), FGFR2(17)-ROCK1(2), FGFR2(17)-KCNH7(10), FGFR2(17)-PPP1R21(16), FGFR2(17)-NRAP(23), FGFR2(17)-VCL(10), FGFR2(17)-RASAL2(3), FGFR2(17)-NOL4(7), FGFR2(17)-SHC2(2), FGFR2(17)-CCSER1(4), FGFR2(17)-TFEC(2), FGFR3(17)-TACC3(11), FGFR3(17)-TACC3(10), FGFR3(17)-TACC3(6), FGFR3(17)-TACC3(14), FGFR3(17)-TNIP2(2), FGFR3(17)-TNIP2(1), FGFR3(17)-JAKMIP1(4), ACPP(10)-FGFR1(4), RAB11FIP1(3)-FGFR1(10), CTNNA3(9)-FGFR2(17), AHCYL1(1)-FGFR2(18), CEP112(16)-FGFR2(17), TPM3(8)-NTRK1(10), LMNA(2)-NTRK1(11), TPM3(8)-NTRK1(12), GON4L(20)-NTRK1(9), LMNA(2)-NTRK1(12), LMNA(4)-NTRK1(13), LMNA(8)-NTRK1(12), ETV6(5)-NTRK3(15), ETV6(4)-NTRK3(14), ETV6(5)-NTRK3(14), KHDRBS1(8)-NTRK3(12), MYO5A(23)-NTRK3(14), EML4(2)-NTRK3(14), ERBB2(25)-GRB7(10), ERBB2(26)-GRB7(2), ERBB2(26)-C1orf87(10), C19orf12(2)-AKT2(9), TSHZ3(1)-AKT2(7), EGFR(24)-RAD51(4), PAX8(10)-PPARG(2), PAX8(9)-PPARG(2), PAX8(8)-PPARG(2), C11orf95(3)-RELA(3), C11orf95(3)-RELA(2), C11orf95(2)-RELA(3), C11orf95(2)-RELA(2)