SOPHiA GENETICS™

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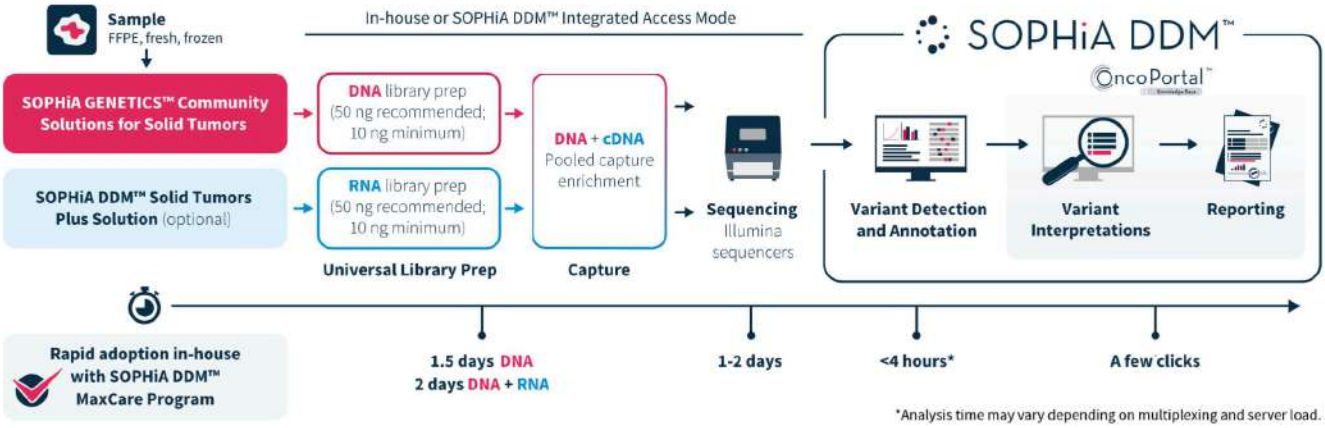
Genique Lifesciences

# SOPHiA GENETICS™ Community Solutions for Solid Tumors

Accelerate discovery with expert-designed applications

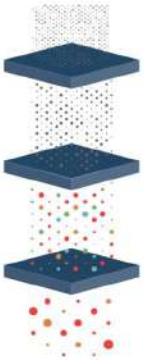
SOPHiA GENETICS™ Community Solutions for Solid Tumors are **expert-designed, capture-based** applications, minimizing setup time and accelerating research. The solutions, combined with the analytical power of the SOPHiA DDM™ Platform, deliver fast, accurate, and cost-effective insights from targeted regions of interest.

## STREAMLINED SAMPLE-TO-REPORT WORKFLOW




*Analysis time may vary depending on multiplexing and server load.

## RELIABLE VARIANT DETECTION WITH OPTIONAL FUSION CALLING



**Variant calling and annotation**

Detect **SNVs**, **Indels**, **known fusions**, **CNVs**[1] and **MSI** in a single workflow, with comprehensive and reliable **annotation.**

**Prioritization**

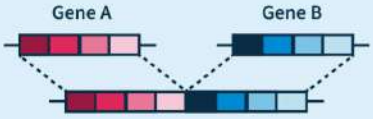
**Pre-classify** variant pathogenicity based on recent guidelines and machine learning.

**Filtering**

Easily apply multiple filtering functions to focus on **disease-specific variants** and **create custom filtering** strategies.

**ADD-ON**

### SOPHiA DDM™ Solid Tumor Plus Solution




Detect **143** fusion pairs from **RNA** samples

Our fusion detection technology is designed to **minimize false positives** and allow the **accurate detection of fusions** even in degraded samples.

## BENEFITS OF SOPHiA GENETICS™ COMMUNITY SOLUTIONS FOR SOLID TUMORS

- **Uniform coverage of target regions**
- **High-quality probe design to optimize on-target rate**
- **Gene content curated by experts in solid tumor analysis**
- **Simple and reliable data analysis and interpretation**

1. All solutions detect gene amplifications. Whole gene deletion available for specific solutions (CSTS_55 and CSTS_57).

CNV, copy number variant; Indel, insertions and deletions; MSI, microsatellite instability, SNV, single nucleotide variant.

## COMMUNITY SOLUTIONS FOR SOLID TUMORS

| Solution name<br>Reference | Genes covered and associated diseases[1] | Panel size<br>Reads/sample<br>Samples per run/Sequencer[2] | Product codes |
|---|---|---|---|
| **SOPHiA DDM™ Solid Tumor Solution (STS)**<br>Backbone for community solutions | **42 genes:**<br>*AKT1, ALK, BRAF, CDK4, CDKN2A, CTNNB1, DDR2, DICER1, EGFR, ERBB2, ERBB4, FBXW7, FGFR1, FGFR2, FGFR3, FOXL2, GNA11, GNAQ, GNAS, H3F3A, H3F3B, HIST1H3B, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MET, MYOD1, NRAS, PDGFRA, PIK3CA, PTPN11, RAC1, RAF1, RET, ROS1, SF3B1, SMAD4, TERT, TP53*<br><br>**Covered diseases:**<br>Lung, melanoma, colorectal, thyroid, GIST, and glioma | **Panel size:** 21.6Kb<br><br>**Reads/sample:** 2.1 M<br><br>**Samples per run/Sequencer:**<br>8 on Illumina MiniSeq™ Mid Output (2x150)<br>24 on Illumina MiSeq ® v3 (2x300bp)<br>12 on Ion Torrent™ Ion S5 System (Ion 530™)<br>96 on MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x150bp) | BS0105ILLRSM<br><br>**STS+ (DNA&RNA):**<br>BS0115ILLRSM |
| **SOPHiA DDM™ Community Solid Tumor Solution 49 genes (CSTS_49)**<br>CSTS_L_v1 | **49 genes:**<br>*AKT1, ALK, BRAF, CDK4, CDKN2A, CTNNB1, DDR2, DICER1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FOXL2, GNA11, GNAQ, GNAS, H3F3A, H3F3B, HIST1H3B, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MED12, MET, MYOD1, NRAS, PDGFRA, PIK3CA, POLE, PTEN, PTPN11, RAC1, RAF1, RET, ROS1, SF3B1, SMAD4, STK11, TERT, TP53*<br><br>**Covered diseases in addition to those covered by STS:**<br>Endometrial cancer | **Panel size:** 29.5Kb<br><br>**Reads/sample:** 3 M<br><br>**Samples per run/Sequencer:**<br>16 on Illumina MiSeq® v3 (2x150bp)<br>Also compatible with Illumina NextSeq® 550 | CS2479ILLRSM<br><br>**CSTS_49+ (DNA&RNA):**<br>CS2490ILLRSM |
| **SOPHiA DDM™ Community Solid Tumor Solution 51 genes (CSTS_51)**<br>CSTS_A_V1 | **51 genes:**<br>*AKT1, ALK, ARID1A, BRAF, BRCA1, BRCA2, CDK4, CDKN2A,CTNNB1, DDR2, DICER1, EGFR, ERBB2, ERBB4, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FOXL2, GNA11, GNAQ, GNAS, H3F3A, H3F3B, HIST1H3B, HRAS, IDH1, IDH2, KIT, KMT2A, KMT2D, KRAS, MAP2K1, MAP2K2, MET, MTOR, MYOD1, NRAS, PDGFRA, PIK3CA, PTPN11, RAC1, RAF1, RET, ROS1, SF3B1, SMAD4, TERT, TGFBR2, TP53*<br><br>**Covered diseases in addition to those covered by STS:**<br>Endometrial cancer | **Panel size:** 44Kb<br><br>**Reads/sample:** 3.7 M<br><br>**Samples per run/Sequencer:**<br>12 on Illumina MiSeq® v3 (2x150bp) | CS2155ILLRSM<br><br>**CSTS_51+ (DNA&RNA):**<br>CS2443ILLRSM |
| **SOPHiA DDM™ Community Solid Tumor Solution 55 genes (CSTS_55)**<br>CSTS_N_v2 | **55 genes:**<br>*AKT1, ALK, ARID1A, ARID5B, BRAF, BRCA1, BRCA2, CDK4, CDKN2A, CTCF, CTNNB1, DDR2, DICER1, EGFR, ERBB2, ERBB4, FBXW7, FGFR1, FGFR2, FGFR3, FOXL2, GNA11, GNAQ, GNAS, H3F3A, H3F3B, HIST1H3B, HRAS, IDH1,IDH2, KEAP1, KIT, KRAS, MAP2K1, MET, MYOD1, NF1, NRAS, PDGFRA, PIK3CA, PIK3R1, POLE, PPP2R1A, PTEN, PTPN11, RAC1, RAF1, RET, ROS1, RPL22, SF3B1, SMAD4, STK11, TERT, TP53*<br><br>**Covered diseases in addition to those covered by STS:**<br>Breast and ovarian cancers | **Panel size:** 77Kb<br><br>**Reads/sample:** 6.6 M<br><br>**Samples per run/Sequencer:**<br>36 on Illumina NextSeq® 500/550 Mid-Output (2x150bp)<br>112 on Illumina NextSeq® 2000 P2 (2x150bp) | CS2505ILLRSM<br><br>**CSTS_55+ (DNA&RNA):**<br>CS2541ILLRSM |
| **SOPHiA DDM™ Community Solid Tumor Solution 57 genes (CSTS_57)**<br>CSTS_N_v3 | **57 genes:**<br>*AKT1, ALK, ARID1A, ARID5B, BRAF, BRCA1, BRCA2, CDK4, CDKN2A, CTCF, CTNNB1, DDR2, DICER1, EGFR, ERBB2, ERBB4, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FOXL2, GNA11, GNAQ, GNAS, H3F3A, H3F3B, HIST1H3B, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MET, MYOD1, NF1, NRAS, PDGFRA, PIK3CA, PIK3R1, POLD1, POLE, PPP2R1A, PTEN, PTPN11, RAC1, RAF1, RET, ROS1, RPL22, SF3B1, SMAD4, STK11, TERT, TP53*<br><br>**Covered diseases in addition to those covered by STS:**<br>Breast, ovarian, endometrial cancers | **Panel size:** 83.6Kb<br><br>**Reads/sample:** 8.3 M<br><br>**Samples per run/Sequencer:**<br>24-32 on Illumina NextSeq® 2000 P1 (2x150bp)<br>24-32 on Illumina NextSeq® 550 Mid-Output (2x150bp)<br>96 on Illumina NextSeq® 2000 P2 (2x150bp) | CS2552ILLRSM<br><br>**CSTS_57+ (DNA&RNA):**<br>CS2603ILLRSM |
| **SOPHiA DDM™ Community Solid Tumor Solution 106 genes (CSTS_106)**<br>CLSO_v5 | **106 genes:**<br>*AKT1, ALK, AR, ARAF, ARID1A, ARID2, ATM, ATRX, B2M, BAP1, BARD1, BRAF, BRCA1, BRCA2, BRIP1, CARD11, CCND1, CCNE1, CDK12, CDK4, CDK6, CDKN2A, CDKN2B, CHEK1, CHEK2, CTNNB1, DAXX, DDR2, EGFR, EIF1AX, ERBB2, ERBB3, ERBB4, ERCC2, ESR1, EZH2, FANCA, FANCL, FBXW7, FGFR1, FGFR2, FGFR3, GATA3, GNA11, GNAQ, GNAS, H3F3A, HIST1H3B, HRAS, IDH1, IDH2, JAK1, JAK2, KDM6A, KEAP1, KIT, KMT2C, KRAS, MAP2K1, MAP2K2, MAP3K1, MC1R, MDM2, MET, MITF, MTOR, MYC, MYCN, NBN, NCK1, NF1, NOTCH1, NOTCH2, NRAS, NTRK1, NTRK3, PALB2, PBRM1, PDGFRA, PIK3CA, PIK3R1, POLD1, POLE, PPP2R2A, PTCH1, PTEN, PTPN11, RAC1, RAD51B, RAD51C, RAD51D, RAD54L, RAF1, RB1, RET, RICTOR, ROS1, SF3B1, SMAD4, SMARCA4, STK11, TERT, TP53, TSC1, TSC2, VHL*<br><br>**Covered diseases in addition to those covered by STS:**<br>Breast, ovarian, endometrial cancers | **Panel size:** 324Kb<br><br>**Reads/sample:** 21 M<br><br>**Samples per run/Sequencer:**<br>12 on Illumina NextSeq® 550 Mid-Output (2x150bp)<br>36 on Illumina NextSeq® 550 High-Output (2x150bp) | CS2429ILLRSM<br><br>**CSTS_106+ (DNA&RNA):**<br>CS2515ILLRSM |

- Gene available in SOPHiA DDM™ STS
- Additional gene vs SOPHiA DDM™ STS
- Additional gene vs SOPHiA DDM™ STS with high biological relevance

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1. The list of associated diseases are non-exhaustive; 2. Sequencing recommendations and pipelines for other sequencing kits and instruments are available upon request. The number of samples per run is recommended for 1000x coverage depth. Illumina NextSeq instruments require 2×150 bp sequencing on compatible flow cells.

## FUSION PAIRS INTERROGATED BY THE SOPHiA DDM™ SOLID TUMOR PLUS SOLUTIONS

SOPHiA DDM™ Solid Tumor Plus Solutions cover 143 relevant gene fusions in addition to MET exon 14 skipping and EGFR variant III.[1]

| Fusion partner 1 | Fusion partner 2 |
|---|---|
| ACPP(10) | FGFR1(4) |
| AGAP3(9) | BRAF(9) |
| AGAP3(10) | BRAF(9) |
| AGK(2) | BRAF(8) |
| AHCYL1(1) | FGFR2(18) |
| ATIC(7) | ALK(20) |
| C19orf12(2) | AKT2(9) |
| CCDC6(1) | BRAF(9) |
| CCDC6(1) | RET(10) |
| CCDC6(1) | RET(12) |
| CD74(6) | ROS1(33) |
| CD74(6) | ROS1(34) |
| CD74(7) | ROS1(33) |
| CD74(7) | ROS1(34) |
| CDK5RAP2(13) | BRAF(9) |
| CEP112(16) | FGFR2(17) |
| CEP112(16) | FGFR2(18) |
| CEP85L(2) | ROS1(18) |
| CLTC(31) | ALK(20) |
| CTNNA3(9) | FGFR2(17) |
| CUL1(7) | BRAF(9) |
| DCTN1(26) | ALK(20) |
| EGFR(1) | EGFR(8) |
| EGFR(24) | RAD51(4) |
| EML4(2) | ALK(20) |
| EML4(6) | ALK(19) |
| EML4(6) | ALK(20) |
| EML4(13) | ALK(20) |
| EML4(14) | ALK(20) |
| EML4(18) | ALK(20) |
| EML4(19) | ALK(20) |
| EML4(20) | ALK(20) |
| EML4(2) | NTRK3(14) |
| ERBB2(26) | C1orf87(10) |
| ERBB2(25) | GRB7(10) |
| ERBB2(26) | GRB7(2) |
| ETV6(4) | NTRK3(14) |
| ETV6(5) | NTRK3(14) |
| ETV6(5) | NTRK3(15) |
| EZR(9) | ROS1(32) |
| EZR(9) | ROS1(33) |
| EZR(9) | ROS1(34) |
| FAM131B(2) | BRAF(9) |
| FAM131B(3) | BRAF(9) |
| FGFR2(17) | BICC1(3) |
| FGFR2(17) | BICC1(10) |
| FGFR2(17) | BICC1(16) |
| FGFR2(17) | CCSER1(4) |
| FGFR2(17) | KCNH7(10) |
| FGFR2(17) | KIAA1217(3) |
| FGFR2(17) | KIF14(19) |
| FGFR2(17) | LAMC1(27) |
| FGFR2(17) | NOL4(7) |
| FGFR2(17) | NRAP(23) |
| FGFR2(17) | NRBF2(4) |
| FGFR2(17) | PPP1R21(16) |
| FGFR2(17) | RABGAP1L(20) |
| FGFR2(17) | RASAL2(3) |
| FGFR2(17) | ROCK1(2) |
| FGFR2(17) | SHC2(2) |
| FGFR2(17) | TFEC(2) |
| FGFR2(17) | VCL(10) |
| FGFR3(17) | BAIAP2L1(2) |
| FGFR3(17) | JAKMIP1(4) |
| FGFR3(17) | TACC3(6) |
| FGFR3(17) | TACC3(10) |
| FGFR3(17) | TACC3(11) |
| FGFR3(17) | TACC3(14) |
| FGFR3(18) | TACC3(8) |
| FGFR3(18) | TACC3(9) |
| FGFR3(18) | TACC3(10) |
| FGFR3(18) | TACC3(11) |
| FGFR3(17) | TNIP2(1) |
| FGFR3(17) | TNIP2(2) |
| FN1(20) | ALK(19) |
| GCC2(19) | ALK(20) |
| GIPC2(3) | BRAF(10) |
| GOLGB1(9) | ROS1(35) |
| GON4L(20) | NTRK1(9) |
| GOPC(8) | ROS1(35) |
| KHDRBS1(8) | NTRK3(12) |
| KIAA1549(13) | BRAF(9) |
| KIAA1549(15) | BRAF(9) |
| KIAA1549(15) | BRAF(11) |
| KIAA1549(16) | BRAF(9) |
| KIAA1549(16) | BRAF(10) |
| KIAA1549(16) | BRAF(11) |
| KIAA1549(18) | BRAF(10) |
| KIAA1549(19) | BRAF(9) |
| KIF5B(24) | ALK(20) |
| KIF5B(15) | RET(12) |
| KIF5B(16) | RET(12) |
| KIF5B(18) | RET(12) |
| LMNA(2) | NTRK1(11) |
| LMNA(2) | NTRK1(12) |
| LMNA(4) | NTRK1(13) |
| LMNA(8) | NTRK1(12) |
| MET(13) | MET(15) |
| MKRN1(4) | BRAF(9) |
| MKRN1(4) | BRAF(11) |
| MYH14(19) | BRAF(10) |
| MYO5A(23) | NTRK3(14) |
| NCOA4(7) | RET(12) |
| NCOA4(8) | RET(12) |
| NCOA4(11) | RET(12) |
| OSBPL9(11) | BRAF(10) |
| PAX8(8) | PPARG(3) |
| PAX8(9) | PPARG(3) |
| PAX8(10) | PPARG(3) |
| PDCD10(4) | RET(11) |
| PHTF2(16) | BRAF(9) |
| PJA2(7) | BRAF(11) |
| PLAGL1(6) | ROS1(32) |
| PRKAR1A(7) | RET(12) |
| PRKAR1B(9) | BRAF(9) |
| PRKAR2B(1) | BRAF(10) |
| RAB11FIP1(3) | FGFR1(10) |
| RANBP2(18) | ALK(20) |
| RASSF4(3) | RET(12) |
| RBM33(3) | BRAF(11) |
| SCRIB(27) | BRAF(11) |
| SDC4(2) | ROS1(32) |
| SLC16A10(3) | ROS1(3) |
| SLC34A2(4) | ROS1(32) |
| SLC45A3(1) | BRAF(8) |
| SLC45A3(1) | BRAF(9) |
| SLC45A3(1) | BRAF(10) |
| SLC45A3(1) | BRAF(11) |
| SLC4A4(21) | ROS1(34) |
| SND1(9) | BRAF(9) |
| SND1(9) | BRAF(11) |
| SND1(10) | BRAF(9) |
| SND1(10) | BRAF(11) |
| SND1(14) | BRAF(9) |
| SND1(17) | BRAF(11) |
| TFG(4) | RET(11) |
| TMPRSS2(1) | ERG(2) |
| TPM3(8) | ALK(20) |
| TPM3(8) | NTRK1(10) |
| TPM3(8) | NTRK1(12) |
| TPM3(9) | ROS1(35) |
| TSHZ3(1) | AKT2(7) |
| VCL(16) | ALK(20) |
| ZCCHC8(2) | ROS1(36) |
| ZNF207(3) | BRAF(10) |

1. The list of gene fusions provided is non-exhaustive. The analytical pipeline is capable of detecting additional fusions.