SOPHiA GENETICS™

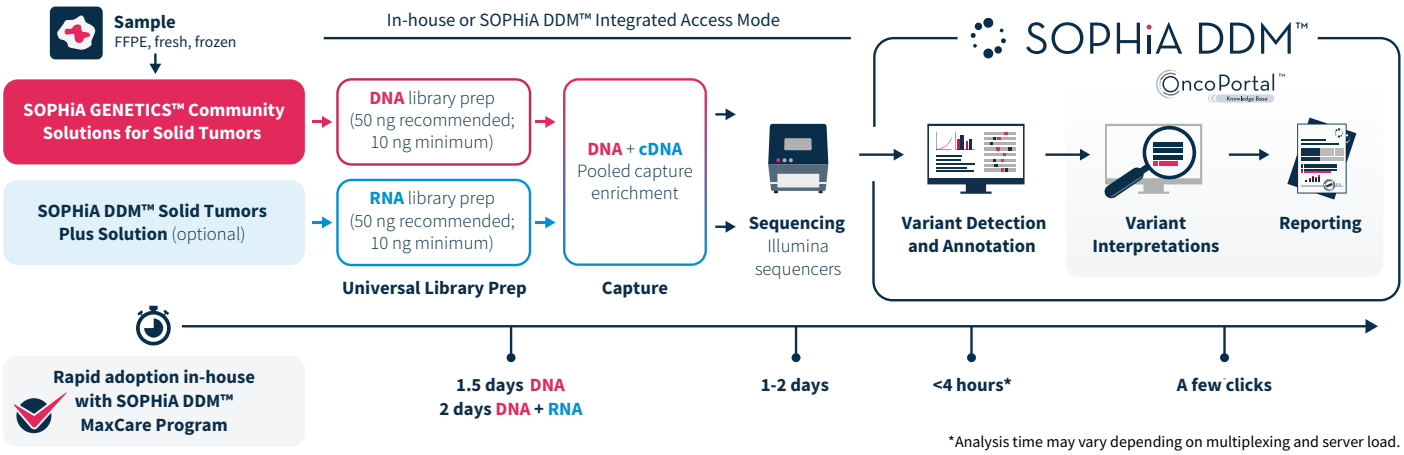
SOPHiA GENETICS™ Community Solutions for Solid Tumors

Accelerate discovery with expert-designed applications



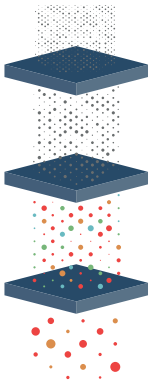
SOPHiA GENETICS™ Community Solutions for Solid Tumors are **expert-designed, capture-based** applications, minimizing setup time and accelerating research. The solutions, combined with the analytical power of the SOPHiA DDM™ Platform, deliver fast, accurate, and cost-effective insights from targeted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

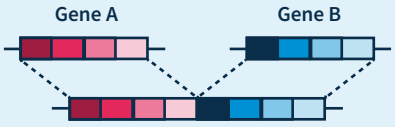
Pre-classify variant pathogenicity based on recent guidelines and machine learning.

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**

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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.

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COMMUNITY SOLUTIONS FOR SOLID TUMORS

Solution name Reference	**Genes covered and associated diseases[1]**	**Panel size** **Reads/sample** **Samples per run/Sequencer[2]**	**Product codes**
SOPHiA DDM™ Solid Tumor Solution (STS) Backbone for community solutions	**42 genes:** *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* **Covered diseases:** Lung, melanoma, colorectal, thyroid, GIST, and glioma	**Panel size:** 21.6Kb **Reads/sample:** 2.1 M **Samples per run/Sequencer:** 8 on Illumina MiniSeq™ Mid Output (2x150) 24 on Illumina MiSeq ® v3 (2x300bp) 12 on Ion Torrent™ Ion S5 System (Ion 530™) 96 on MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x150bp)	BS0105ILLRSM **STS+ (DNA&RNA):** BS0115ILLRSM
SOPHiA DDM™ Community Solid Tumor Solution 49 genes (CSTS_49) CSTS_L_v1	**49 genes:** *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* **Covered diseases in addition to those covered by STS:** Endometrial cancer	**Panel size:** 29.5Kb **Reads/sample:** 3 M **Samples per run/Sequencer:** 16 on Illumina MiSeq® v3 (2x150bp) Also compatible with Illumina NextSeq® 550	CS2479ILLRSM **CSTS_49+ (DNA&RNA):** CS2490ILLRSM
SOPHiA DDM™ Community Solid Tumor Solution 51 genes (CSTS_51) CSTS_A_V1	**51 genes:** *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* **Covered diseases in addition to those covered by STS:** Endometrial cancer	**Panel size:** 44Kb **Reads/sample:** 3.7 M **Samples per run/Sequencer:** 12 on Illumina MiSeq® v3 (2x150bp)	CS2155ILLRSM **CSTS_51+ (DNA&RNA):** CS2443ILLRSM
SOPHiA DDM™ Community Solid Tumor Solution 55 genes (CSTS_55) CSTS_N_v2	**55 genes:** *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* **Covered diseases in addition to those covered by STS:** Breast and ovarian cancers	**Panel size:** 77Kb **Reads/sample:** 6.6 M **Samples per run/Sequencer:** 36 on Illumina NextSeq® 500/550 Mid-Output (2x150bp) 112 on Illumina NextSeq® 2000 P2 (2x150bp)	CS2505ILLRSM **CSTS_55+ (DNA&RNA):** CS2541ILLRSM
SOPHiA DDM™ Community Solid Tumor Solution 57 genes (CSTS_57) CSTS_N_v3	**57 genes:** *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* **Covered diseases in addition to those covered by STS:** Breast, ovarian, endometrial cancers	**Panel size:** 83.6Kb **Reads/sample:** 8.3 M **Samples per run/Sequencer:** 24-32 on Illumina NextSeq® 2000 P1 (2x150bp) 24-32 on Illumina NextSeq® 550 Mid-Output (2x150bp) 96 on Illumina NextSeq® 2000 P2 (2x150bp)	CS2552ILLRSM **CSTS_57+ (DNA&RNA):** CS2603ILLRSM
SOPHiA DDM™ Community Solid Tumor Solution 106 genes (CSTS_106) CLSO_v5	**106 genes:** *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* **Covered diseases in addition to those covered by STS:** Breast, ovarian, endometrial cancers	**Panel size:** 324Kb **Reads/sample:** 21 M **Samples per run/Sequencer:** 12 on Illumina NextSeq® 550 Mid-Output (2x150bp) 36 on Illumina NextSeq® 550 High-Output (2x150bp)	CS2429ILLRSM **CSTS_106+ (DNA&RNA):** 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.

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