SOPHiA GENETICS™

SOPHiA DDM™ Myeloid Plus Solution

The genomic application that streamlines the interpretation of complex variants by combining the DNA target capture and RNA target enrichment kits with the analytical performance and advanced features of the SOPHiA DDM™ Platform.

Main Features

SOPHiA DDM™ Myeloid Plus Solution (MYS+) covers **30 relevant genes** (10 with complete coding sequences) associated with myelodysplastic syndromes, myeloproliferative neoplasms, and leukemia. Probe design is optimized to guarantee high on-target rate and coverage uniformity even in GC-rich regions. The RNA part covers **119 gene** fusions associated with leukemia. The technical limitations related to the analyses of translocations and key biomarkers such as *CEBPA*, *CALR* and *FLT3* (including detection of internal tandem duplications) are addressed.

DNA Gene Panel

ABL1 (4-9), ***ASXL1*** *(10,12,13), BRAF (15),* ***CALR*** *(9), CBL (8,9),* ***CEBPA*** *(all), CSF3R (all), DNMT3A (all), ETV6 (all), EZH2 (all),* ***FLT3*** *(13-15,20), HRAS (2,3), IDH1 (4), IDH2 (4), JAK2 (all), KIT (2,8-11,13,17,18), KRAS (2,3), MPL (10), NPM1 (10,11), NRAS (2,3), PTPN11 (3,7-13), RUNX1 (all), SETBP1 (4), SF3B1 (10- 16), SRSF2 (1), TET2 (all), TP53 (all), U2AF1 (2,6), WT1 (6-10), ZRSR2 (all)*

RNA Gene Fusion Panel

Variants Called

SNVs
Indels
CNVs
FLT3 ITDs
Gene fusion

Recommendations

Starting material
50 ng DNA, 500 ng RNA

Sample type
Blood and bone marrow

Samples per run / Sequencer[1]
24 DNA + 24 RNA on Illumina MiSeq® v3 (2x300bp)
12 DNA + 12 RNA on Illumina MiSeq® v2 (2x250bp)

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:
2 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]

	Observed (%)	Lower 95% CI
Sensitivity	99.92	97.49
Specificity	99.99	99.98
Accuracy	99.99	99.98
Precision	99.52	91.47
Repeatability	98.69	98.66
Reproducibility	99.30	99.27
Average on-target rate	87.41	
Coverage uniformity	99.98	
Mean % of target region > 1000x	> 99	
Limit of detection	2.5*	

The SOPHiA DDM™ Platform reaches advanced analytical performance. Performance values are based on SNVs and Indels in 237 DNA samples processed on Illumina MiSeq®.
*For SNVs and Indels; *FLT3*-ITD excepted.

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:
BS0112ILLRSMY10[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 Genomenon Clinical Knowledgebase (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 hundreds 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; CNVs, copy number variations; Indels, insertions/deletions; ITDs, internal tandem duplications; NGS, next-generation sequencing; SNVs, single nucleotide variants.

1. Number of samples per run is recommended for 1000x coverage depth. Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform.
2. Analysis time may vary depending on the number of samples multiplexed and server load.
3. Product codes for SOPHiA GENETICS™ Universal Library Prep (ULP), replacing BS0112ILLRSML01 and BS0112ILLRSMY01.

Somatic gene and variant annotations and related content have been powered by Genomenon Clinical Knowledgebase (CKB).

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

RNA Gene Fusion Panel

ATF7IP(13)-JAK2(9,11,13,15,17,18,19), BCR(1,4,6,7,12,13,14,19)-ABL1(2,3,4), BCR(1,4,6,7,12,13,14,19)-FGFR1(11), BCR(1,4,6,7,12,13,14,19)-JAK2(9,11,13,15,17,18,19), BCR(1,4,6,7,12,13,14,19)-PDGFRA(12), BMP2K(14,15)-ZNF384(2,3,4,7), CBFA2T3(10,11)-GLIS2(4,5), CBFB(4,5)-MYH11(29,30,31,32,33,34,35), CCDC6(1,7)-PDGFRB(9,11), CHIC2(3)-ETV6(2,3), CNTRL(38)-FGFR1(11), CREBBP(4,5,6,7)-ZNF384(2,3,4,7), CUX1(11)-FGFR1(11), DEK(9)-NUP214(17,18), EBF1(10,13,14,15)-JAK2(9,11,13,15,17,18,19), EBF1(10,13,14,15)-PDGFRB(9,11), EML1(18)-ABL1(2,3,4), EP300(6)-ZNF384(2,3,4,7), ETV6(4,5,6,7)-ABL1(2,3,4), ETV6(4,5,6,7)-ARNT(3), ETV6(4,5,6,7)-JAK2(9,11,13,15,17,18,19), ETV6(4,5,6,7)-NTRK3(14), ETV6(4,5,6,7)-NTRK3(15), ETV6(4,5,6,7)-PDGFRB(9,11), ETV6(4,5,6,7)-RU NX1(1), ETV6(4,5,6,7)-RUNX1(3), FGFR1OP(5,6,7)-FGFR1(11), FIP1L1(12)-PDGFRA(12), FOXP1(19)-ABL1(2,3,4), INPP5D(8)-ABL1(2,3,4), KAT6A(16)-CREBBP(2,3), KMT2A(8,9,10,11)-AFDN(2), KMT2A(8,9,10,11)-AFF1(4,5,6,11), KMT2A(8,9,10,11)-AFF3(7,8,12), KMT2A(8,9,10,11)-AFF4(4,5,6), KMT2A(8,9,10,11)-ARHGAP26(19), KMT2A(8,9,10,11)-ARHGEF12(11,12,13), KMT2A(8,9,10,11)-ARHGEF17(2,3,4,5), KMT2A(8,9,10,11)-C2CD3(13,14,15,17), KMT2A(8,9,10,11)-CBL(10), KMT2A(8,9 ,10,11)-CIP2A(17), KMT2A(8,9,10,11)-CREBBP(2,3), KMT2A(8,9,10,11)-DCPS(2), KMT2A(8,9,10,11)-ELL(2,3,6), KMT2A(8,9,10,11)-EPS15(2,6), KMT2A(8,9,10,11)-FOXO3(2), KMT2A(8,9,10,11)-KMT2A(2), KMT2A(8,9,10,11)-KNL1(12), KMT2A(8,9,10,11)-MAML2(2,3), KMT2A(8,9,10,11)-MAPRE1(2,4,6), KMT2A(8,9,10,11)-MLLT1(2,4,5,6,7), KMT2A(8,9,10,11)-MLLT10(5,7,10,12,17), KMT2A(8,9,10,11)-MLLT11(2), KMT2A(8,9,10,11)-MLLT3(4,5,6,9,10), KMT2A(8,9,10,11)-MLLT6(8,9,12), KMT2A(8,9,10,11)-NRIP3(2), KMT2A(8,9,10,11)-RARA(2), KMT2A(8,9,10,11)-SEP5(2), KMT2A(8,9,10,11)-SEPT6(2), KMT2A(8,9,10,11)-SEPT9(2), KMT2A(8,9,10,11)-SEPT9(2), KMT2A(8,9,10,11)-TET1(9), MEF2D(7)-CSF1R(12), MN1(1)-ETV6(2,3), MNX1(1)-ETV6(2,3), MYB(8)-GATA1(5), NCOR1(35)-LYN(8), NDE1(6)-PDGFRB(9,11), NPM1(4,6)-MLF1(3), NPM1(4,6)-RARA(2), NUP214(23,26,28,29,30,31,32,34)-ABL1(2,3,4), NUP98(10,11,12,13,14)-DDX10(6,7), NUP98(10,11,12,13,14)-HOXA9(1,2), NUP98(10,11,12,13,14)-KDM5A(27), NUP98(10,11,12,13,14)-NSD1(6), NUP98(10,11,12,13,14)-RAP1GDS1(2,3), NUP98(10,11,12,13,14)-TOP1(8), OFD1(21)-JAK2(9,11,13,15,17,18,19), P2RY8(1)-CRLF2(1), PAG1(8)-ABL2(3,5), PAX5(4)-ETV6(2,3), PAX5(4)-JAK2(9,11,13,15,17,18,19), PAX5(5)-ETV6(2,3), PAX5(5)-JAK2(9,11,13,15,17,18,19), PCM1(26,36)-JAK2(9,11,13,15,17,18,19), PDE4DIP(16)-PDGFRB(9,11), PICALM(17,18,19)-MLLT10(5,7,10,12,17), PML(3,6)-RARA(2), RANBP2(18)-ABL1(2,3,4), RBM15(1)-MKL1(4,5), RCSD1(2)-ABL1(2,3,4), RCSD1(3)-ABL1(2,3,4), RUNX1(3)-RUNX1T1(6), SET(7)-NUP214(17,18), SFPQ(9)-ABL1(2,3,4), SNX2(3)-ABL1(2,3,4), SPAG9(26)-JAK2(9,11,13,15,17,18,19), SPTBN1(4)-FLT3(14), SPTBN1(4)-PDGFRB(9,11), SSBP2(5,6,8,10,16)-CSF1R(12), SSBP2(5,6,8,10,16)-JAK2(9,11,13,15,17,18,19), STAT5B(15,16)-RARA(2), STIL(1)-TAL1(3,4,6), STRN(6)-PDGFRA(12), STRN3(8,9)-JAK2(9,11,13,15,17,18,19), TAF15(6,9)-ZNF384(2,3,4,7), TCF3(11,13,15,16,17)-HLF(4), TCF3(11,13,15,16,17)-PBX1(3), TERF2(8)-JAK2(9,11,13,15,17,18,19), TPM3(8)-PDGFRB(9,11), TPR(22,39)-FGFR1(11), TRIM24(9,10,11)-FGFR1(11), ZBTB16(3,4)-ABL1(2,3,4), ZBTB16(3,4)-RARA(2), ZC3HAV1(12)-ABL2(3,5), ZEB2(9)-PDGFRB(9,11), ZMIZ1(18)-ABL1(2,3,4), ZMYM2(17)-FGFR1(11)