SOPHiA GENETICS™

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 GENETICS™ Myeloid Plus Solution 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 fusion part covers **119 key genes** 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 Fusion Panel

View RNA fusion panel list

Variants Called

SNVs
Indels
CNVs
FLT3 ITDs[1]
RNA fusions

Recommendations

Starting material
200 ng DNA, 500 ng RNA

Sample type
Blood and bone marrow

Samples per run / Sequencer (Flow Cell / Ion Chip Kit)[3]
24 DNA + 24 RNA Illumina MiSeq® v3 (2x300bp)
12 DNA + 12 RNA 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[2] from FASTQ: from 4 hours

	Observed (%)	Lower 95% CI
Sensitivity	99.85	96.78
Specificity	99.99	99.98
Accuracy	99.99	99.98
Precision	99.27	96.78
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. The values have been calculated based on SNVs and Indels in samples processed on Illumina MiSeq®.

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

Confident decision-making

The SOPHiA DDM™ platform integrates the OncoPortal™. This feature provides the latest scientific evidence on the actionability and significance of each genomic alteration to support informed decision-making for research purpose.

Access to the SOPHiA GENETICS community

Through the SOPHiA DDM™ platform genomics experts from >750 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.

1. Internal tandem duplications
2. Varies depending on the number of genes, samples multiplexed and server load.
3. Sequencing recommendations and specifications for other sequencing kits and instruments are available upon request. Number of samples per run is recommended for 1000x coverage depth.

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).
All product and company names are trademarks™ or registered® trademarks of their respective holders. Use of them does not imply any affiliation with or endorsement by them.

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

All third party trademarks listed by SOPHiA GENETICS remain the property of their respective owners. Unless specifically identified as such, SOPHiA GENETICS' use of third party trademarks does not indicate any relationship, sponsorship, or endorsement between SOPHiA GENETICS and the owners of these trademarks. Any references by SOPHiA GENETICS to third party trademarks is to identify the corresponding third party goods and/or services and shall be considered nominative fair use under the trademark law.

©SOPHiA GENETICS 2021 - PM_RUO_B.1.2.2.16_r7en

x

RNA 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)-RUNX1(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)