SOPHiA GENETICS™

SOPHiA DDM™ Extended Myeloid Solution

The genomic application that streamlines the interpretation of complex genomic variants by combining a capture-based target enrichment kit with the analytical performance and advanced features of the SOPHiA DDM™ Platform.

Main Features

SOPHiA DDM™ Extended Myeloid Solution (ExtMYS) covers the complete coding sequences of **98 relevant genes** associated with leukemia, myelodysplastic syndromes, and myeloproliferative neoplasms. Probe design is optimized to guarantee high on-target rate and coverage uniformity even in GC-rich regions. The technical limitations related to the analyses of key biomarkers such as *CEBPA*, *ASXL1*, *CALR*, *FLT3* and *KMT2D* (including detection of tandem duplications) are addressed.

Gene Panel	Variants Called	Recommendations	Wet Lab
ABL1, ANKRD26, ***ASXL1****, ASXL2, ATM, ATRX, BCOR, BCORL1, BRAF, BRCC3,* ***CALR****, CBL, CBLB, CBLC, CCND2, CDKN2A,* ***CEBPA****, CHEK2, CREBBP, CSF3R, CSMD1, CSNK1A1, CTCF, CUX1, DDX41, DHX15, DNMT3A, ELANE, ETNK1, ETV6, EZH2, FANCA, FANCL,* ***FLT3****, GATA1, GATA2, GNAS, GNB1, HNRNPK, HRAS, IDH1, IDH2, IKZF1, JAK1, JAK2, JAK3, KDM6A, KIT, KMT2A, KMT2D, KRAS, LUC7L2, MECOM, MET, MPL, MYC, NF1, NOTCH1, NOTCH2, NPM1, NRAS, PAX5, PDGFRA, PHF6, PIGA, PML, PPM1D, PTPN11, RAD21, RAF1, RB1, RBBP6, RPS19, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SETBP1, SF3B1, SH2B3, SMC1A, SMC3, SOS1, SRP72, SRSF2, STAG1, STAG2, STAT3, STAT5B, TERC, TET, TET2, TP53, U2AF1, WT1, ZBTB7A, ZRSR2*	SNVs Indels CNVs *FLT3* ITDs *KMT2D* PTDs	**Starting material** 50 ng DNA **Sample type** Blood and bone marrow **Samples per run / Sequencer**[1] 16 on Illumina NextSeq® 500/550 Mid Output v2 (2x150bp) 36 on Illumina NextSeq® 500/550 High Output v2 (2x150bp)	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time:** 2 days

Analytical Performance

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

Analysis time from FASTQ: from 6 hours[2]

	Observed (%)	Lower 95% CI
Sensitivity	100	98.13
Specificity	99.99	99.99
Accuracy	99.99	99.99
Precision	97.91	91.6
Repeatability	99.95	99.92
Reproducibility	99.96	99.92

The SOPHiA DDM™ Platform reaches advanced analytical performance. The values have been calculated based on 143 samples processed on Illumina NextSeq® 500/550.

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:
BS2201ILLRSMY10
BS0118ILLRSMY10[3] (with RNA sequencing)

Confident decision-making

The OncoPortal™ Plus add-on module for SOPHiA DDM™ Platform matches tumor molecular profiles with clinical associations andavailable 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 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; CNVs, copy number variations; Indels, insertions/deletions; ITDs, internal tandem duplications; PTDs, partial 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 BS2201ILLRSML01 and BS0118ILLRSML01.

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.