SOPHiA GENETICS™

SOPHiA DDM™ 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™ Myeloid 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 technical limitations related to the analyses of key biomarkers such as *CEBPA*, *ASXL1*, *CALR* and *FLT3* (including detection of internal tandem duplications) are addressed.

Gene Panel	Variants Called	Recommendations	Wet Lab
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)*	SNVs Indels CNVs *FLT3* ITDs	**Starting material** 50 ng DNA **Sample type** Blood and bone marrow **Samples per run / Sequencer[1]** 24 on Illumina MiSeq® v3 (2x300bp) 96 on Illumina NextSeq® 500/550 Mid Output v2 (2x150bp) 4 on Illumina MiniSeq™ Mid Output (2x150bp) 16 on Illumina MiniSeq™ High Output (2x150bp) 32 on Ion S5™ using Ion 540™ chip 96† on MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x200)	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time:** 2 days

† theoretical estimated maximum number of samples to be multiplexed, assuming 900 million reads per lane, and considering available kit size.

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 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	99.88	
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 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:
BS0103ILLRSMY10
BS0103TFSRGLY10[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 BS0103ILLRSMY01, BS0103ILLRSML01, and BS0103TFSRGLL01.

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

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