SOPHiA GENETICS™

SOPHiA DDM™ Lymphoma Solution

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

Main Features

SOPHiA DDM™ Lymphoma Solution covers **54 relevant genes** associated with many B- and T-Cell Lymphomas such as Diffuse Large B-Cell, Follicular, Mantle Cell and Burkitt Lymphomas. 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 *CREBBP*, *EP300* and *EZH2* are addressed. For specific needs, the gene content can be fully customized.

Gene Panel	Variants Called	Recommendations	Wet Lab
ARID1A, ATM (57-63), B2M, BCL2, BCL6 (8,9), BIRC3 (all,ex.2), BRAF (15), BTK (15), CARD11 (4-9), CCND1 (1), CCND3, CD58, CD79A (4,5), CD79B (5,6), CHD2, CDKN2A, CDKN2B, CIITA, CREBBP (27-30), CXCR4, EP300, EZH2 (16,18), FBXW7 (9,10), FOXO1, GNA13, ID3, IRF4, KMT2A, KMT2D, KRAS (2,3), MAL, MEF2B, MYC, MYD88, NFKBIE, NOTCH1 (34), NOTCH2 (26-28,34), NRAS (2,3), PAX5, PIM1, PLCG2 (17-23), POT1, PRDM1, PTEN (5), PTPN11, REL, SF3B1 (14,15), SOCS1, STAT6 (9-14), TCF3 (17-19), TNFAIP3, TNFRSF14, TP53, XPO1 (15-18)	SNVs Indels CNVs Gene amplifications	**Starting material** 50ng DNA **Sample type** FFPE, blood and bone marrow **Samples per run / Sequencer**[1] 8 on MiSeq® v3 (2x300bp), blood samples 4 on MiSeq® v2 (2x150bp), FFPE samples 36 on NextSeq® 500/550 Mid-Output (2x150bp), blood and FFPE samples	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time:** 1.5 days

Analytical Performance

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting, annotating and pre-classifying genomic alterations such as SNVs and Indels in 54 genes, and gene amplifications in 47 genes of the panel to support experts with their data-informed decision making.

Analysis time from FASTQ: from 4 hours[2]

	Observed (%)	Lower 95% CI
Sensitivity	100	92.5
Specificity	100	99.9
Accuracy	100	99.9
Precision	98	87.1
Average on-target rate	90.6	
Coverage uniformity	97.2	

The values have been calculated based on SNVs and Indels in 28 FFPE samples on 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 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 JAX-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.

Product code:
BS0116ILLRSMY10[3]

CNVs, copy number variants; FFPE, formalin-fixed paraffin-embedded; Indels, insertions/deletions; NGS, next-generation sequencing; SNVs, single nucleotide variants

1. Number of samples per run is recommended for 1000x coverage depth. Sequencing recommendations and pipelines for other sequencing kits and instruments are available upon request. Delivery time may vary according to the selected sequencing platform.
2. Analysis time may vary depending on the number of genes, samples multiplexed, and server load.
3. Product code for SOPHiA GENETICS™ Universal Library Prep (ULP), replacing BS0116ILLRSMY05.

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB).

For Research Use Only. Not for Use in Diagnostic Procedures.
OncoPortal™Plus is for Clinical Decision Support Use Only. Not intended as a primary diagnostic tool.