SOPHiA GENETICS™

# 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 GENETICS™ Extended Myeloid Solution covers the complete coding sequences of **98 relevant genes** associated with myeloid neoplasms such as myelodysplastic syndromes (MDS), myeloproliferative neoplasms (MPN), overlap syndromes MDS/MPN and acute myeloid 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, 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<br>Indels<br>CNVs<br>FLT3 ITDs[1] | **Starting material**<br>200 ng DNA<br><br>**Sample type**<br>Blood and bone marrow<br><br>**Samples per run / Sequencer (Flow Cell / Ion Chip Kit)[3]**<br>Up to 16 for Illumina NextSeq® 500/550 Mid Output v2 (2x150bp)<br>36 for Illumina NextSeq® 500/550 High Output v2 (2x150bp) | **Day 1:**<br>Library Preparation<br><br>**Day 2:**<br>Capture and Sequencing<br><br>**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[2] from FASTQ: from 6 hours**

| | 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

### 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™).


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