MYELOID TUMOR SOLUTION™ BY SOPHiA GENETICS

The Myeloid Solution (MYS) by SOPHiA GENETICS is a molecular diagnostic application that bundles the analytical power of SOPHiA™ AI with a capture-based target enrichment kit and full access to SOPHiA DDM™ platform.

SOPHiA GENETICS

Knowledge-Driven Kit Design

SOPHiA™

Collective AI for Data-Driven Medicine

SaaS Analytical Platform

The MYS panel covers the coding regions and splicing junctions (± 25 bp) of 30 genes associated to Myelodysplasic Syndroms (MDS), Myeloproliferative Neoplasms (MPN) and Leukemia including Internal Tandem Duplications (ITD). It guarantees superior coverage uniformity, high on-target reads percentage and exceptional coverage in GC-rich regions, even in the first exon. Technical limitations related to the analysis of key biomarkers such as *CEBPA*, *FLT3* and *CALR* are overcome with our solution.

Gene panel

ABL1 (4-9), *ASXL1* (9,11,12), *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)

Recommendations

Starting material: 200 ng

Sample source: Blood and bone marrow

Samples per run: 24(1)

Wet lab

Day 1: Library Preparation

Day 2: Capture and Sequencing

Total hands-on time: 8 hours

CDS excluding UTRs

Additional gene content and pack sizes are under development

(1) Sequenced on Illumina MiSeq® v3 (600 cycles). 12 samples may be sequenced using v2 (500 cycles) chemistry. However, the usage of v3 is highly recommended

Compatible with all Illumina Instruments: MiniSeq™, MiSeq®, NextSeq® 500/550 and HiSeq® 2500

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(2) Analysis time may vary depending on the number of genes, samples multiplexed and server load

SOPHiA analyses complex genomic NGS data by detecting, annotating and pre-classifying genomic variants such as SNVs, Indels and CNVs in 30 genes associated to haematological disorders to help clinicians better diagnose their patients.

SOPHiA leads to excellent clinical grade analytical performances:

	Observed
Sensitivity	100%
Reproducibility	100%
Average on-target rate	86,64%
Coverage uniformity	99,31%
Average percentage of target region > 1000x	98,87%
Reported variant	1%

Analysis time from FASTQ files: 4 hours(2)

All results are presented in SOPHiA DDM, the platform of choice for clinicians performing routine diagnostic testing. Thanks to its intuitive user interface and integrated features, variants visualization and interpretation are facilitated, while assuring protection of clinical genomic data.

Dedicated features

Dedicated features in SOPHiA DDM reduce the complexity of determining the clinical significance of genomic variants. Its pre-classified variants makes it easy and quick to interpret the data.

OncoPortal

SOPHiA DDM integrates the OncoPortal, providing actionable information on Haematological malignancies. Leveraging on precision medicine intelligence and combining curated content from professional association guidelines and publicly available databases, it contains relevant somatic classifications, based on variant-drug-disease association, best available treatments and relevant ongoing clinical trials.

Access to the World's Largest Clinical Genomics Community

Through SOPHiA DDM, experts from hundreds of healthcare institutions can easily interpret the variants and flag them with the appropriate level of pathogenicity. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.

SOPHiA™

The AI Democratizing Data-Driven Medicine

sophiagenetics.com | info@sophiagenetics.com | @SOPHiAGENETICS