SOPHiA™

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MYELOID SOLUTION™ CE IVD
BY SOPHiA GENETICS

The molecular diagnostic application bundles a capture-based target enrichment kit with the analytical power of SOPHiA™ AI and full access to the SOPHiA DDM™ platform.

SMART KIT DESIGN

The MYS application covers complete coding sequence including ± 25 bp of exon-flanking regions of the 30 most clinically relevant genes associated with Myelodysplastic Syndromes (MDS), Myeloproliferative Neoplasms (MPN) and Leukemia. Probe design is highly optimized to provide exceptional coverage uniformity throughout the entire target regions, resulting in superior data quality. The technical limitations related to the analysis of key biomarkers such as *CEBPA*, *ASXL1*, *CALR* and *FLT3* (including detection of internal tandem duplications) are addressed with this CE-IVD marked solution.

Gene panel

ABL1 (4-9), ASXL1 (9,11,12,14), 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 (2-11), U2AF1 (2,6), WT1 (6-10), ZRSR2 (all)

Recommendations

Starting material: 200 ng
Sample source: Blood and bone marrow
Samples per run: Depending on sequencing platform[1]

Sequencer	Flow Cell Kit (Sequencing run)	Recommended samples per run (for 1000x coverage depth)
Illumina MiSeq®	v3 (2x300bp)	24
	v2 (2x250bp)	12

Wet lab

Day 1: Library Preparation
Day 2: Capture and Sequencing
Total library preparation time: 1.5 days

ANALYTICAL POWER

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying genomic variants such as SNVs, Indels and CNVs in all the genes of the panel, associated with hematological disorders to help experts better analyze and interpret genomic data.

SOPHiA reaches excellent clinical-grade performance[2]:

	Observed	Lower 95% CI
Sensitivity	99.85%	96.78%
Specifity	99.99%	99.98%
Accuracy	99.99%	99.98%
Precision	99.27%	96.78%
Repeatability	98.69%	
Reproducibility	99.30%	
Average on-target rate	87.41%	
Coverage uniformity	99.98%	
Average % of target region > 1000x	> 99%	
Limit of detection	2.5%	

Analysis time from FASTQ files: 4 hours[3]

A total of 419 clinical samples have been processed on a Illumina MiSeq® platform to calculate the above-mentioned metrics

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform
(2) Performance values have been calculated on SNVs and Indels only. The detection of CNVs is not part of the CE-IVD claim
(3) Analysis time may vary depending on the number of genes, samples multiplexed and server load

UNIVERSAL PLATFORM

The results are presented in SOPHiA DDM, the platform of choice for clinicians performing routine diagnostic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic alterations. Patient's data is kept safe by applying the highest industrial standards of encryption.

Main features

SOPHiA DDM offers several features that make variant analysis more efficient. With variant pre-classification and customized filtering options, experts can easily accelerate the data interpretation process.

OncoPortal™

SOPHiA DDM integrates the OncoPortal, a decision support functionality based on precision medicine intelligence. It enables experts to access relevant therapeutic, prognostic and diagnostic databases to determine the actionability and clinical significance of genomic alterations. Moreover, the OncoPortal uses inclusion and exclusion criteria to maximize clinical trial matching that may benefit the patient, both locally and at the global level.

Access to SOPHiA's Community

In SOPHiA DDM, experts from hundreds of healthcare institutions interpret the results and flag the pathogenicity level of variants according to their knowledge and experience. 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
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