MYELOID PLUS SOLUTION BY SOPHiA GENETICS

The Myeloid Plus Solution (MYS+) by SOPHiA GENETICS is a molecular diagnostic application that bundles a smart DNA target capture and RNA target amplicon kits with the analytical power of SOPHiA™ AI and full access to SOPHiA DDM® platform.

SOPHiA GENETICS®

Smart Kit Design

SOPHiA™

The AI Democratizing Data-Driven Medicine

SOPHiA DDM®

SaaS Analytical Platform

The MYS+ targets DNA variants and RNA transcripts of fusion genes, associated with, but not limited to MDS(1), MPN(2) and Leukemia. The DNA panel covers the coding regions and splicing junctions of 30 clinically relevant genes associated with hematological malignancies. It guarantees high on-target reads percentage and coverage uniformity even in GC-rich regions. The RNA fusion panel covers over 119 gene fusions. Technical challenges related to the analysis of translocations and key biomarkers such as *CEBPA*, *FLT3* including internal tandem duplications and *CALR* are addressed with this solution.

DNA 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)

RNA fusion panel

For more information on fusion transcripts, visit:
https://www.sophiagenetics.com/hospitals/solutions/myeloid-plus-solution.html

Recommendations

Starting material: 200 ng DNA, 500 ng RNA
Sample source: Blood and bone marrow
Samples per run: Depending on sequencing platform(3)

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

Wet lab

Day 1: RNA and DNA Library Preparation
Day 2: Capture & Sequencing of DNA and RNA libraries in 1 run
Total library preparation time: 6 hours for RNA, 1.5 days for DNA

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

SOPHiA enables excellent clinical-grade analytical performance(4):

	DNA	RNA
Sensitivity	99.85%	100%
Specificity	99.99%	
Accuracy	99.99%	
Precision	99.27%	>95%
Repeatability	98.69%	
Reproducibility	99.30%	
Average on-target rate	87.41%	
Coverage uniformity	99.88%	
Average percentage of target region > 1000x	> 99%	
Limit of detection	2.5%	

Analysis time from FASTQ files: 4 hours(5)

(1) MDS: Myelodisplastic Syndroms
(2) MPN: Myeloproliferative Neoplasms
(3) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request
(4) A total of 419 clinical samples have been processed on a Illumina MiSeq® platform to calculate the above-mentioned metrics.Performance values have been calculated on SNVs and Indels only
(5) Analysis time may vary depending on the number of genes, samples multiplexed and server load

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

Dedicated features

SOPHiA DDM platform for Oncology enables experts to explore, interpret and report genomic alterations.
With pre-classified variants and customized variant filtering options, experts can easily accelerate the data interpretation process.

OncoPortal™

SOPHiA DDM platform for Oncology integrates the OncoPortal, a decision support functionality for experts who analyze cancer genomic profiles. Based on precision medicine intelligence, SOPHiA matches patients' genomic alterations with curated databases of evidence-based clinical associations. Such associations encompass a combination of genomic alterations, cancer types and therapies. This information highlights the clinical significance and actionability of the patient's tumor profile within their cancer and in other cancer types to increase potential treatment options. It also uses inclusion and exclusion criteria to identify clinical trials 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.


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