SOPHiA GENETICS™

SOPHiA DDM™ Dx Myeloid Solution

CE IVD

The molecular diagnostic application that streamlines the interpretation of complex genomic variants by combining a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

The SOPHiA DDM™ Dx Myeloid Solution is intended to be used to identify variants occurring in **30 genes** involved in myeloid neoplasms by targeting specific mutation-prone positions within the genomic sequence. The function of the product is to be an aid to healthcare professionals to make a clinical decision related to myeloid neoplasms, and to provide molecular rationale for appropriate therapy. The product is intended to be used for *in vitro* diagnostic and professional use only.

Gene Panel	Variants Called	Recommendations	Wet Lab
ABL1 (4-9), ***ASXL1*** *(10,12,13), 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)*	SNVs Indels *FLT3*-ITDs	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run / Sequencer (Flow Cell)[1]** 24 for Illumina® MiSeq™ v3 (2x300bp)	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time: 2 days**

Analytical Performance

The web-based SOPHiA DDM™ Platform analyzes complex NGS data with highly accurate detection of SNVs, Indels and *FLT3*-ITDs. SOPHiA DDM™ core offers a Clinical Decision Support (CDS) component that allows visualization and interpretation of variants in a single workflow.* The Platform reaches clinical-grade performance.**

Analysis time from FASTQ: < 6 hours[2]

	Observed (%)	Lower 95% CI
Sensitivity	99.92	97.49
Specificity	99.99	99.98
Accuracy	99.99	99.98
Precision	99.52	91.47
Repeatability	98.69	98.66
Reproducibility	99.30	99.27
Average on-target rate	87.41	
Coverage uniformity	99.88	
Mean % of target region > 1000x	> 99	
Limit of detection	2.5***	

*CDS results are not part of the CE-IVD claim.
**Performance values are based on SNVs and Indels in 237 samples processed on Illumina® MiSeq™.
***For SNVs and Indels; *FLT3*-ITD excepted.

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

The SOPHiA DDM™ Platform provides the user with a web-based portal and workspace to upload and analyze genomic sample data for our CE-IVD marked products. It enables a fully CE-IVD compliant workflow, from library preparation to variant identification (Figure 1). Once the samples are analyzed, IVD reports are created and can be downloaded from the web portal to support decision-making.

Product codes:

BS0103ILLCSML01-016
BS0103ILLCSML01-032
BS0103ILLCSML01-048
BS0103ILLCSML01-96

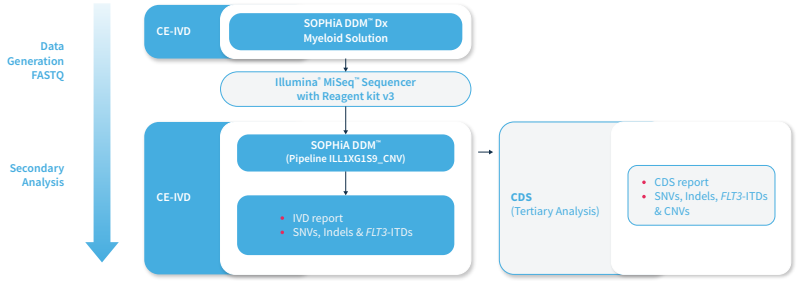

Figure 1. IVD workflow. CDS results are not part of the CE-IVD claim.

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 Set Up Program provides assistance with application set up for fast and worry-free transition to routine testing.

Secure and unlimited data storage

The SOPHiA DDM™ Platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with local data security policies.

CNV, copy number variation; IVD, in vitro diagnostic; ITD, internal tandem duplication; SNV, single nucleotide variant.
1. Sequencing recommendations and specifications for other sequencing kits and instruments are available upon request.
2. Varies depending on the number of genes, samples multiplexed and server load.

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