SOPHiA GENETICS™

SOPHiA DDM™ Dx Solid Tumor Solution

CE IVD

The molecular diagnostic application that characterizes the complex mutational landscape of the major solid tumors by combining a capture-based target enrichment kit with the analytical performance and advanced features of the SOPHiA DDM™ Platform.

Main Features

The SOPHiA DDM™ Dx Solid Tumor Solution is intended to be used to identify variants occurring in **42 genes** involved in solid tumors by targeting specific mutation-prone positions within the genomic sequence. The function of the product is to serve as an aid to healthcare professionals to make a clinical decision related to solid tumors, 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
AKT1 (3), ALK (21-25), BRAF (11,15), CDK4 (2), CDKN2A (1,2,3), CTNNB1 (3), DDR2 (17), DICER1 (24,25), EGFR (18-21), ERBB2 (8,17,20), ERBB4 (10,12), FBXW7 (7-11), FGFR1 (12,14), FGFR2 (7,12,14), FGFR3 (7,9,14,16), FOXL2 (1*), GNA11 (4,5), GNAQ (4,5), GNAS (8), H3F3A(2*), H3F3B (2*), HIST1H3B (1), HRAS (2-4), IDH1 (4), IDH2 (4), KIT (8-11,13,17,18), KRAS (2-4), MAP2K1 (2,3), MET (2,14-20), MYOD1 (1), NRAS (2-4), PDGFRA (12,14,18), PIK3CA (2*,3,6*,8,10,21), PTPN11 (3), RAC1 (3), RAF1 (7,10*,12,13*,14*,15*), RET (11,13,15,16), ROS1 (38*,41*), SF3B1 (15-17), SMAD4 (8-12), TERT (promoter*,1*,8*,9*,13*), TP53 (2-11)*	SNVs and Indels in all genes of the panel	**Starting material** 50 ng **Sample type** FFPE **Samples per run for 1000x coverage depth / Sequencer (Flow Cell)[1]** 24 for Illumina® MiSeq™ v3 (2x300bp)*	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time:** 1.5 days

*Hotspots only

*2x150-cycle sequencing run (paired-end) is recommended

Analytical Performance

The SOPHiA DDM™ Platform analyzes complex NGS data with highly accurate detection of SNVs and Indels. SOPHiA DDM™ offers a Research Use Only (RUO) component that allows visualization and interpretation of variants in a single workflow.* The Platform reaches clinical-grade performance.**

Analysis time from FASTQ: < 4 hours[2]

	Observed (%)	Lower 95% CI
Sensitivity	98.77	93.31
Specificity	100	99.92
Accuracy	99.97	99.85
Precision	100	96.25
Repeatability	96.45	96.41
Reproducibility	89.13	89.05
Coverage uniformity	98.70	92.50***

*RUO results are not part of the CE-IVD claim.
**Performance values have been calculated based on SNVs and Indels only in 150 samples processed on Illumina® MiSeq® using KAPA™ HyperPlus KK8512 library preparation reagents.
***5% quantile

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

In Dx mode, 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 applications. 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 to support decision-making.

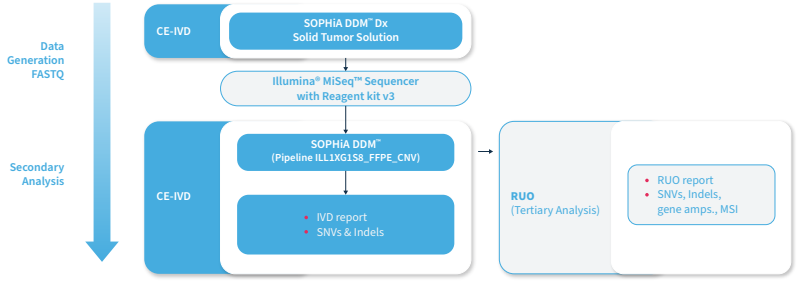

Figure 1. IVD workflow. RUO results are not part of the CE-IVD claim.

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 SOPHiA DDM™ MaxCare Program provides assistance with application set up for fast and worry-free transition to routine testing.

Secure data storage

The SOPHiA DDM™ Platform keeps data safe by applying the highest industrial standards of encryption in compliance with local data security policies.

Product codes:
BS0105ILLCSML03-016
BS0105ILLCSML03-032
BS0105ILLCSML03-048

amps., amplifications; FFPE, formalin-fixed, paraffin-embedded; IVD, in vitro diagnostic; MSI, microsatellite instability; SNV, single nucleotide variant.
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. Analysis time may vary depending on the number of samples multiplexed and server load.

Somatic gene and variant annotations and related content have been powered by Genomenon Cancer Knowledgebase (CKB).