SOPHiA GENETICS™

SOPHiA DDM Dx Hereditary Cancer Solution

CE IVD

The molecular diagnostic application that streamlines the analysis of the complex mutational landscape associated with major hereditary cancer disorders by combining a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

The CE-IVD marked SOPHiA DDM Dx Hereditary Cancer Solution covers the coding regions and splicing junctions of 26 genes (target region of 105 kb) associated with Hereditary Breast and Ovarian Cancer (HBOC), Lynch syndrome and various intestinal polyposis syndromes. Probe design is optimized to guarantee high on-target rate and coverage uniformity, even in GC-rich regions, with proven clinical-grade performance.

Gene Panel	Variants Called	Recommendations	Wet Lab
ABRAXAS1, APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PIK3CA, PMS2, PMS2CL[1]*, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2*	SNVs Indels	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run for >250x coverage depth / Sequencer (Flow Cell)**[2] 48 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 and Indels. The SOPHiA DDM Platform™ desktop access mode 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[3]

	Observed (%)	Lower 95% CI
Sensitivity	100	99.20
Specificity	99.99	99.99
Accuracy	99.99	99.99
Precision	99.86	96.42
Repeatability	99.98	99.98
Reproducibility	99.93	99.92
Average on-target rate	79.39	
Coverage uniformity	99.72	

*CDS results are not part of the CE-IVD claim.
**Performance values have been calculated based on SNVs and Indels in 159 samples processed on Illumina MiSeq™.

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:
BS0102ILLCGLL01-016; BS0102ILLCGLL01-032
BS0102ILLCGLL01-048; BS0102ILLCGLL01-096

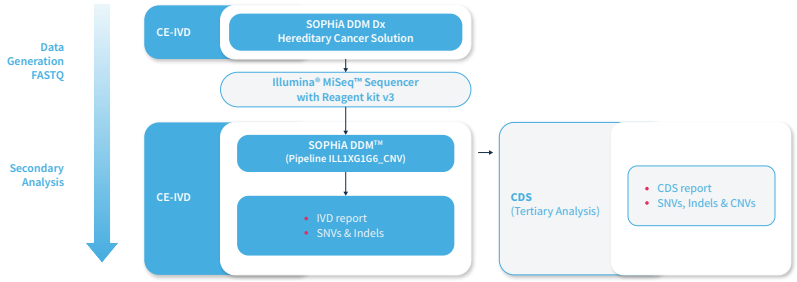

Figure 1. IVD workflow.CDS results are not part of the CE-IVD claim.

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; SNV, single nucleotide variation.
1. The pseudogene *PMS2CL* is part of the analysis but not a gene responsible for disease.
2. Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform.
3. Analysis time may vary depending on the number of samples multiplexed and server load.