

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 SOPHIA DDM™ Dx Hereditary Cancer Solution is an *in vitro* diagnostic test intended for the identification of germline variations occurring in **27 most common genes** involved in predispositions to breast and ovarian cancers as well as intestinal cancers in germline DNA samples isolated from peripheral blood. It is intended to be used as an aid to healthcare professionals and to provide molecular rationale to make a clinical decision related to germline mutations associated with breast and ovarian cancer, intestinal polyposis syndromes, and Lynch syndrome.

Gene Panel	Variants Called	Recommendations	Wet Lab
<i>ABRAXAS1, APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PIK3CA, PMS2, PMS2CL1, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2</i>	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. 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 hours³

	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

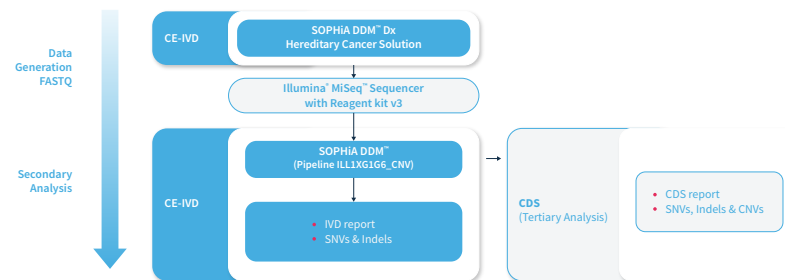


Figure 1. IVD workflow. CDS results are not part of the CE-IVD claim.

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 variant.

- The pseudogene *PMS2CL1* is part of the analysis but not a gene responsible for disease.
- Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform.
- Analysis time may vary depending on the number of samples multiplexed and server load.

This CE IVD-marked product is For In Vitro Diagnostic Use in Europe, Turkey and Israel markets. This product has not been cleared and approved by the U.S. FDA and may not be approved in some countries/regions. The CDS features are for Clinical Decision Support only and not for use as a primary diagnostic tool. Please contact SOPHIA GENETICS™ local Sales representatives to obtain the appropriate product information for your country of residence. All third party trademarks listed by SOPHIA GENETICS™ remain the property of their respective owners. Unless specifically identified as such, SOPHIA GENETICS™ use of third party trademarks does not indicate any relationship, sponsorship, or endorsement between SOPHIA GENETICS™ and the owners of these trademarks. Any references by SOPHIA GENETICS™ to third party trademarks is to identify the corresponding third party goods and/or services and shall be considered nominative fair use under the trademark law.