

A deep learning-powered diagnostic application leveraging low-pass whole genome sequencing (WGS) to identify patterns of genomic instability in ovarian cancer samples. Supports accurate detection homologous recombination deficiency (HRD) status with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

The SOPHiA DDM™ Dx HRD Solution is an *in vitro* diagnostic medical device that can be used as an aid to determine the HRD status of tumors in patients with ovarian cancer by the semi-quantitative detection of the Genomic Instability biomarker. The device is intended to be used by healthcare professionals and is based on a next-generation sequencing (NGS) workflow taking genomic DNA extracted from FFPE treated tumor material as input.

Gene Panel ¹ (CDS only)	Variants Called ² (CDS only)	Recommendations	Wet Lab
AKT1*, ATM, BARD1, BRCA1 , BRCA2 , BRIP1, CCNE1, CDK12, CHEK1, CHEK2, ESR1*, FANCA, FANCD2, FANCL, FGFR1*, FGFR2*, FGFR3*, MRE11, NBN, PALB2, PIK3CA*, PPP2R2A, PTEN, RAD51B, RAD51C, RAD51D, RAD54L, TP53	SNPs Indels Gene amplifications	Starting material 50 ng DNA Sample type FFPE ovarian cancer tissue Samples per run / Sequencer 8 for Illumina® NextSeq® 500/550 Mid Output Kit 24 for Illumina® NextSeq® 500/550 High Output Kit	Day 1: Library Preparation Day 2: Capture and Sequencing Total library preparation time: 1.5 days

*Hotspot coverage only.

Analytical Performance

The web-based SOPHiA DDM™ Platform leverages low-pass WGS for the accurate analysis of genomic integrity. The Platform reaches clinical-grade performance.

Analysis time from FASTQ: < 8 hours²

Genomic Integrity Status	
Concordance with comparator NGS assay*	
Positive Percent Agreement	92.9%
Negative Percent Agreement	95.8%
Overall Percent Agreement	94.4%
Repeatability**	100%
Reproducibility***	100%
Limit of detection****	28.5% tumor content

*Based on 233 ovarian cancer FFPE samples sequenced on Illumina® NextSeq® 550, and for which a conclusive genomic instability result was obtained with the comparator NGS assay.
**Based on results obtained in independent experiments performed using multiple samples.
***Based on results obtained by seven independent laboratories provided with aliquots of the same reference samples.
****Established by measuring the lowest tumor content for which sensitivity is larger or equal to 95%.

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic integrity

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 portal to support decision-making.

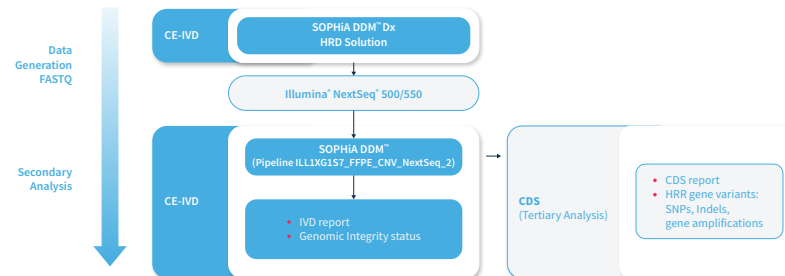


Figure 1. IVD workflow. CDS results are not part of the CE-IVD claim.

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.

Product codes:

BS01211LLCSMY08-32

CDS, clinical decision support; FFPE, formalin-fixed, paraffin-embedded; IVD, in vitro diagnostic; NGS, next-generation sequencing; SNP, single nucleotide polymorphism.

- Gene panel analysis is not covered by the CE-IVD claim.
- 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.