SOPHiA GENETICS™

SOPHiA DDM™ Hereditary Cancer Solution (HCS) v2.0

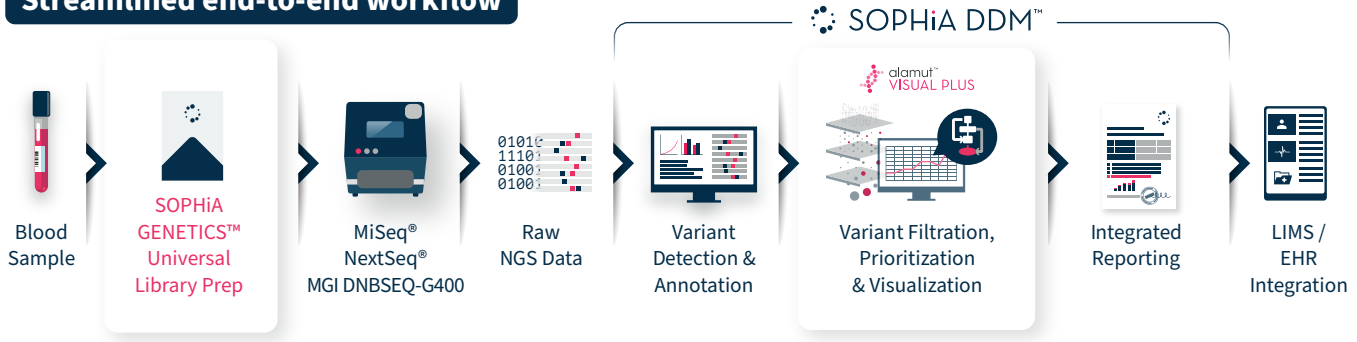
Confidently assess genetic variants predisposing to cancer

The **SOPHiA DDM™ HCS v2.0** is a capture-based NGS application that characterizes challenging variants associated with hereditary cancers. It enables researchers to accelerate their research in-house with high-quality content and the advanced analytical features of the SOPHiA DDM™ Platform.

Highlights

- Includes **guideline-driven content** targeting coding and non-coding regions in **83 biologically actionable genes.**
- Contains **hg38**-based analytics, **built-in sample check ID**, and selected **UTRs** and **promoter regions.**
- Detects **SNVs, Indels** and **CNVs**, and more complex variants such as ***Alu* insertions, Boland Inversion** and ***PMS2/PMS2CL*** gene conversion.
- Improves **laboratory logistics** and **resource management** with **SOPHiA GENETICS™ Universal Library Prep[a]**.

Streamlined end-to-end workflow



Guideline-driven content

covers 83 biologically actionable genes — △ *Full gene* □ *Gene hotspots* ○ *CNVs* ☆ *Promoters/UTRs*

AIP △○	*CDKN2A* △□○	*MDH2* △○	*PALB2* △○	*RB1* △□○	*SPINK1* △○☆
APC △□○☆	*CHEK2* △□○	*MEN1* △□○	*PDGFRA* △○	*RET* △○	*STK11* △○
ATM △□○	*CTNNA1* △○	*MET* △○	*PIK3CA* △○	*RNF43* △○	*SUFU* △○
AXIN2 △○	*DICER1* △□○	*MITF* △○	*PMS2* △□○	*SDHA* △○	*TERC* △□○
BAP1 △○	*DLST* △○	*MLH1* △□○☆	*PMS2CL** ○	*SDHAF2* △○	*TERT* △□○☆
BARD1 △○	*EPCAM* △○	*MLH3* △○	*POLD1* △○	*SDHB* △○	*TGFBR2* △○
BMPR1A △□○	*FAM175A* △○☆	*MSH2* △□○	*POLE* △○	*SDHC* △○	*TMEM127* △○
BRCA1 △□○☆	*FH* △○	*MSH3* △○	*POT1* △○	*SDHD* △○	*TP53* △□○
BRCA2 △□○☆	*FLCN* △□○	*MSH6* △○	*PRKAR1A* △□○	*SEC23B* △○	*TSC1* △□○
BRIP1 △□○	*GREM1* △○☆	*MUTYH* △○	*PRSS1* △○	*SLC25A11* △○	*TSC2* △□○
CDC73 △○	*HOXB13* △○	*NBN* △○	*PTCH1* △○	*SMAD4* △○	*VHL* △□○
CDH1 △○	*KIT* △○	*NF1* △□○	*PTEN* △○☆	*SMARCA4* △○	*WRN* △○
CDK4 △○	*LZTR1* △□○	*NF2* △○	*RAD51C* △○	*SMARCB1* △□○	*WT1* △○
CDKN1B △□○	*MAX* △○	*NTHL1* △□○☆	*RAD51D* △○	*SMARCE1* △○	

*pseudogene.

Excellent analytical performance[1]

Median coverage uniformity[b]
99.99%

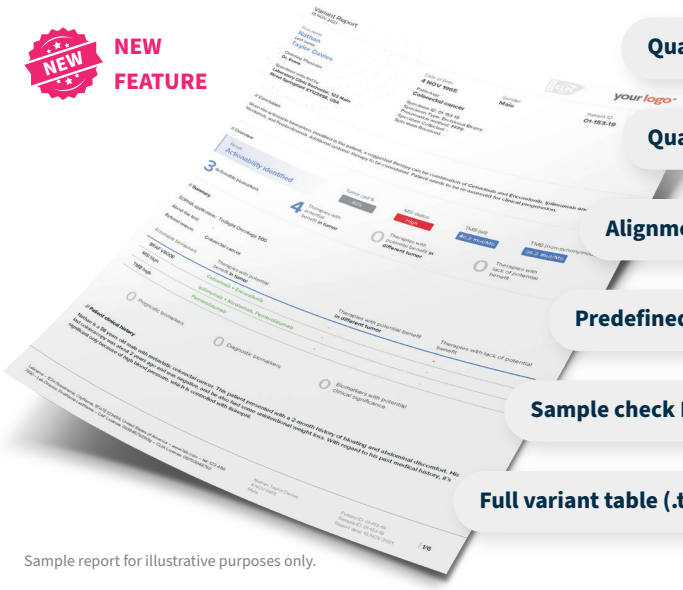
Median on-target rate[b]
(incl. flank target)
88.3%

Sensitivity[c]
100%
for SNV and Indel detection

Precision[c]
100% for SNV and **98.5%** for Indel detection

Comprehensive output files for analysis and reporting

NEW FEATURE



- Quality metrics (.pdf)
- New reporting tool (.pdf or .json) NEW
- Quality indicators NEW
- HGVS annotation NEW
- Indels status
- Alignment (.bam)
- Genotype (.vcf)
- CNVs status
- Predefined ID SNPs NEW
- Coverage depth
- Variant fraction
- Sample check ID NEW
- SNV status
- Alu status NEW
- Full variant table (.txt/.vcf)
- CNVs flagging & individual CNV reporting NEW

Sample report for illustrative purposes only.

"The SOPHiA DDM™ HCS v2.0 has comprehensive, guideline-driven content that ensures we're obtaining the most relevant insights from genomic data. The expanded capabilities available on the SOPHiA DDM™ Platform, including built-in sample check ID and HGVS annotation, makes it the optimal solution for timely, in-house results on complex mutational analysis.

Unimore | Modena, Italy
UNIMORE
UNIVERSITÀ DEGLI STUDI DI MODENA E REGGIO EMILIA

Specifications

Covered diseases	Breast, ovarian, endometrial, prostate, abdominal, endocrine and neuroendocrine, nervous, renal, and skin cancers
Input amount	50 ng DNA
Multiplexing for > 250x coverage depth	• 16 for Illumina MiSeq® v3 (2x200bp), 8-12 for v2 (2x150bp) • 48 for Illumina NextSeq® 2000 P1, 192 for P2 • 72 for Illumina NextSeq® 500/550 mid-output, 192 for high-output • 96* for MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x200) *theoretical estimated maximum number of samples to be multiplexed, assuming 900 million reads per lane, and considering available kit size.
Automation scripts	Available for Hamilton Clinical Starlet and Hamilton Star

Want to know more?

Contact us at: **info@sophiagenetics.com**

SOPHiA DDM™ Hereditary Cancer Solution (HCS) v2.0 is for research use only – not for use in diagnostic procedures.

[a]Available for Research Use Only SOPHiA DDM™ Oncology and Rare and Inherited Disease applications. [b]Based on analysis of 16 blood samples using Illumina NextSeq® 550 sequencer. [c]Based on analysis of 17 blood samples using Illumina NextSeq® 550 sequencer. [1]Data on File. CNV, copy number variation; HGVS, human genome variant society; NGS, next generation sequencing; SNV, single nucleotide variant.