

The genomic application that integrates a capture-based target enrichment kit with the advanced analytics of the SOPHiA DDM™ Platform. The SOPHiA DDM™ Enhanced Whole Exome Solution builds on a comprehensive Whole Exome backbone, adding targeted probe enhancements for hereditary cancer, carrier screening, and pharmacogenomics indications to boost coverage in critical regions, alongside robust analytical modules designed to reliably detect difficult-to-call variants.

Main Features

The SOPHiA DDM™ Enhanced Whole Exome Solution fully covers the coding regions (±5bp of intronic regions) of 20,072 genes, the entire mitochondrial genome, and non-coding variants known to be associated with rare and inherited disorders. The hereditary cancer, carrier screening, and pharmacogenomics enhancements boost coverage of 94, 155, and 74 genes, respectively.

Gene Panel	Variants Called	Recommendations	Wet Lab
<ul style="list-style-type: none"> 20,072 genes Entire mitochondrial genome > 1750 non-coding variants with known pathogenicity in deep introns/enhancer/promoter genes 	<p>General variant types: SNVs, Indels, CNVs, mitochondrial variants, Alu insertions</p> <p>Specific variants: Large deletions in <i>HBA1</i> and <i>HBA2</i>, Gene/pseudogene analysis for <i>PMS2</i>, <i>SMN1</i>, <i>CYP21A2</i>, and <i>TNXB</i>, <i>MSH2</i> Boland inversions, <i>CFTR</i> polyTGT tract</p>	<p>Starting material 50 ng</p> <p>Sample type Blood</p> <p>Samples per run for > 50x coverage depth¹ 24 and 36 for NextSeq® 1000/2000 P3 and P4 XLEAP reagents 48 for NovaSeq™ S4 flow cell (1 lane) 32 and 192 for NovaSeq™ X 1.5B and 10B flow cells 120 for MGI DBNSEQ-T7 (1 lane) 16 for Element AVITI, High Output</p>	<p>Day 1: Library Preparation</p> <p>Day 2: Capture and Sequencing</p> <p>Hands-on library preparation time: 2.5 hours</p>

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

Dedicated features in SOPHiA DDM™ reduce the complexity of determining the significance of genomic variants and facilitate the interpretation process, thus reducing turnaround time:

- GRCh38/hg38 based analytics** – Select your transcript of choice and leverage insights from frequently updated variant annotation catalogs and zygosity annotation
- Variant pathogenicity prediction** – Prioritize variants for further analysis with point-based ACMG classification and SOPHiA DDM™ machine learning-based ABCD prediction
- Virtual Panels** – Restrict interpretation to sub-panels of genes of interest using the HPO or OMIM®
- Cascading Filters** – Apply custom filtering options and save strategies for future analyses
- Familial Variant Analysis (trio-analysis)** – Identify pathogenic variants considering different modes of inheritance, through a family-based approach
- Alamut™ Visual Plus** – Streamline your interpretation with enhanced genomic visualization and comprehensive variant annotation
- Reporting** – Generate customized CAP- and CLIA-compliant reports

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

Secure and unlimited data storage

Access to the SOPHiA DDM™ Platform is restricted to registered users only. The Platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with your local data security policies.

Analytical Performance

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic variants across all genes in the panel.

Analysis time from FASTQ: 11 hours²

	Observed
Sensitivity for SNVs/Indels ³	99.4%
Precision for SNVs/Indels ³	98.5%
Average on target region >25x (>50x)	99.7% (98.9%)
Spike-in / exome coverage ratio	3.5-4

- For best performance of the gene conversion modules for *SMN1*, *CYP2D6* and *CYP21A2*, a minimum capture size of 12 samples is recommended.
- For 24 samples of ~150M reads. Analysis time may vary depending on server load and the number of multiplexed samples.
- SNV and Indel performance metrics are based on six well-characterised reference samples with more than 26,700 confirmed variants each. Four samples were sequenced with 150M and two with 115M reads.

The SOPHiA GENETICS Community

Join one of the largest global networks of healthcare institutions to securely and anonymously share knowledge with your peers and confidently assess even challenging variants.

Product code: BS0141ILLRGLY10

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