SOPHiA GENETICS™

SOPHiA DDM™ Whole Exome Solution v2

The genomic application that combines a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform. SOPHiA DDM™ Whole Exome Solution v2 offers enhanced probe design and increased detection capabilities for a deeper investigation of Mendelian disorders.

Main Features

SOPHiA DDM™ Whole Exome Solution v2 covers the coding regions of 19,425 genes and the entire mitochondrial genome. Probe design is highly optimized to guarantee a high on-target reads percentage and coverage uniformity even in GC-rich regions, including the first exon.

Gene Panel	Variants Called	Recommendations	Wet Lab
• 19,425 genes • Entire mitochondrial genome	SNVs Indels CNVs (97% genes)	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run for > 50x coverage depth** 12 for Illumina NextSeq® 500/550 High Output v2 (2x150bp) 12 (per lane) for Illumina NovaSeq® 6000 (SP) 24 (per lane) for Illumina NovaSeq® 6000 (S1) 64 (per lane) for Illumina NovaSeq® 6000 (S2) 8[1] for MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x200)	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Hands-on library preparation time: 2.5 hours**

Analytical Performance

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

Analysis time[2] from FASTQ: Overnight

	Observed
Sensitivity for SNVs/Indels[3]	>99%
Precision for SNVs/Indels[3]	99%
Sensitivity for CNVs 2-4 exons[4]	>99%
Sensitivity for mitochondrial SNVs/ Indels (limit of detection 5% heteroplasmy)[5]	>99%
Precision for mitochondrial SNVs/Indels (limit of detection 5% heteroplasmy)[5]	100%
Coverage uniformity	>99%
Average % of target region with depth >20x	>99%

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 -** Annotate variants accurately
- **Dual Variant Pre-classification -** Improve assessment of variant pathogenicity with both ACMG scores and SOPHiA GENETICS machine learning-based predictions
- **Virtual Panels -** Restrict the interpretation to sub-panels of genes of interest using the HPO or OMIM® browser
- **Cascading Filters -** Apply custom filtering options for quicker screening of relevant variants and save strategies for future analyses
- **Familial Variant Analysis (trio-analysis) -** Identify pathogenic variants considering different modes of inheritance, through a family-based approach

After interpretation, you can generate a customizable variant report that includes valuable information to support decision making.

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 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.

Access to the SOPHiA GENETICS community

In the SOPHiA DDM™ Platform, experts from hundreds of healthcare institutions interpret their results and flag the pathogenicity level of variants according to their knowledge and experience. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.

Product code: BS0122ILLRGLY10

1. Theoretical estimated maximum number of samples to be multiplexed, assuming 900 million reads per lane, and considering available kit size.
2. Analysis time may vary depending on the number of samples multiplexed and server load.
3. Performance metrics are based on high confidence regions in a reference sample, with 80M reads per sample. Sequencing was performed using an Illumina NextSeq® 550 instrument.
4. Analytical performance for CNVs was calculated with 80M reads per sample. Sequencing was performed using an Illumina NovaSeq® instrument.
5. Analytical performance for mitochondrial SNVs/Indels was calculated on 96 variants (93 SNPs and 3 Indels), with 80M reads per sample. Sequencing was performed using an Illumina NextSeq® 550 instrument.

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