SOPHiA GENETICS™

SOPHiA DDM™ Clinical Exome Solution v3

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™ Clinical Exome Solution v3 offers enhanced probe design and increased detection capabilities for a deeper investigation of Mendelian diseases.

Main Features

SOPHiA DDM™ Clinical Exome Solution v3 covers the coding regions (±5bp of intronic regions) of 5,500 genes, the entire mitochondrial genome and non-coding variants known to be associated with rare and inherited disorders (probe footprint of ~16 Mb). Probe design is highly optimized to guarantee high on-target reads percentage and coverage uniformity even in GC-rich regions, including the first exon.

Gene Panel

- 5,500 genes
- Entire mitochondrial genome
- ~ 200 non-coding variants with known pathogenicity in deep introns/enhancer/promoter genes

Variants Called

SNVs
Indels
CNVs

Recommendations

Starting material
200 ng DNA

Sample type
Blood

Samples per run for > 50x coverage depth
16 for Illumina NextSeq® 500/550 Mid Output v2 (2x150bp)
48 for Illumina NextSeq® 500/550 High Output v2 (2x150bp)
48 (per lane) for Illumina NovaSeq® 6000 (SP)
96 (per lane) for Illumina NovaSeq® 6000 (S1)
4 for Illumina MiSeq® v3 (300bp)
48[1] for MGI DNBSEQ-G400, FCL, 1 lane of 4 (2x200)

Wet Lab

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: 6 hours

	Observed
Sensitivity for SNVs/Indels[3]	99.4%
Precision for SNVs/Indels[3]	99.3%
Sensitivity for CNVs 2-4 exons (1-2 exons)[4]	98.2% (83.0%)
Sensitivity for mitochondrial SNVs/Indels detection (limit of detection 5% heteroplasmy)[5]	>99.9%
Coverage uniformity	99.4%
Average on target region >25x (>50x)	98.7% (88.7%)

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: BS0120ILLRGLY10

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. SNV and Indel performance metrics are based on more than 7,650 variants. For each sample, 16.25M reads per sample were used. Sequencing was performed using an Illumina NextSeq® instrument.
4. Analytical performance for CNVs has been calculated on 80 CNVs, sequenced on NextSeq® instrument.
5. Sensitivity for mitochondrial SNVs/Indels has been calculated on 96 variants (93 SNPs and 3 Indels), sequenced on NextSeq® instrument.

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