SOPHiA™

SOPHiAGENETICS.COM

SOPHiA CLINICAL EXOME SOLUTION™

The genomic application that bundles a capture-based target enrichment kit with the analytical power of SOPHiA™ AI and full access to the SOPHiA DDM™ platform.

SMART KIT DESIGN

SOPHiA Clinical Exome Solution covers the coding regions (±5bp of intronic regions) of 4,490 genes (target region of 12 Mb) related to rare inherited diseases. Probe design is optimized to guarantee high on-target reads percentage and coverage uniformity even in GC-rich regions, including the first exon.

Gene panel

4,490 genes

Recommendations

Starting material: 200 ng

Sample source: Blood

Samples per run: Depending on sequencing platform(1)

Sequencer	Flow Cell / Ion Chip Kit	Recommended samples per run (for >50x coverage depth)
Illumina NovaSeq® 6000	SP	48 (per lane)
	S1	96 (per lane)
Illumina MiSeq®	v3 (2x300bp)	4
Illumina HiSeq®2500	High Output (2x125bp)	24 (per lane)
	Rapid Run Mode (2x150bp)	16 (per lane)
Illumina HiSeq® 3000/4000	High Output (2x100bp)	24 (per lane)
	High Output (2x150bp)	32 (per lane)
Illumina NextSeq® 500/550	Mid Output Kit (2x150bp)	16
	High Output Kit (2x150bp)	48

Wet lab

Day 1: Library Preparation

Day 2: Capture and Sequencing

Total hands-on time: 8 hours

ANALYTICAL POWER

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic variants such as SNVs, Indels and CNVs(2) to support experts with their data-informed decision making.

SOPHiA reaches advanced analytical performance:

	Observed	Lower 95% CI
Sensitivity	99.45%	99.02%
Specifity	99.99%	99.99%
Accuracy	99.99%	99.99%
Precision	99.45%	99.02%
Reproducibility	99.99%	99.98%
Average on-target rate	> 90%	
Coverage uniformity	> 98%	
Average % of target region with depth > 50x	> 96%	

Analysis time from FASTQ files: Overnight(3)

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. Delivery time may vary according to the selected sequencing platform.
(2) CNV detection is available for 98.1% of genes with a resolution of 2-5 exons, depending on the applied sequencing depth per sample.
(3) Analysis time may vary depending on the number of samples multiplexed and server load.

UNIVERSAL PLATFORM

The results are presented in SOPHiA DDM, the platform of choice for experts performing genomic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic variants. Data is kept safe by applying the highest industrial standards of encryption.

Main features

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

- **Dual Variant Pre-Classification:** Improve assessment of variants pathogenicity with the pre-classification of both ACMG guidelines and SOPHiA's prediction
- **Familial Variant Analysis (trio-analysis):** Identify disease causing variants for different modes of inheritance, following a family-based approach
- **Virtual Panels:** Restrict the interpretation to sub-panels of genes of interest (e.g. eye disorders or hearing loss)
- **Variant Filter Builder:** Define and edit custom filters for efficient and dynamic analysis of exomes

Access to SOPHiA's Community

In SOPHiA DDM, experts from hundreds of healthcare institutions interpret the results and flag the pathogenicity level of variants in accordance 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.

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