SOPHiA™

SOPHiAGENETICS.COM

CLINICAL EXOME SOLUTION™ BY SOPHiA GENETICS

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

SMART KIT DESIGN

The CES panel covers the coding regions (± 5bp of intronic regions) of 4,490 genes and spans 12 Mb of target region. It guarantees 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 (sequencing run)	Recommended samples per run (for 250x coverage depth)
HiSeq® 3000/4000	High Output (2x100bp)	24 (per lane)
	High Output (2x150bp)	32 (per lane)
HiSeq®2500	High Output (2x125bp)	24 (per lane)
	Rapid run mode (2x150bp)	16 (per lane)
MiSeq®	v3 (2x300bp)	4
NextSeq® 500/550	Mid Output Kit (2x150bp)	16
	High Output Kit (2x150bp)	48
Ion Proton™	Ion P1 v3	4
Ion S5™	Ion 540	4

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 genomic variants such as SNVs, Indels and CNVs(2) to help clinicians better diagnose their patients.

SOPHiA reaches excellent clinical-grade performance:

	Observed
Sensitivity	> 99%(3)
Precision	> 99%(3)
Repeatability	> 99%
Reproducibility	> 99%
Average on-target rate	> 90%
Coverage uniformity	> 98%
Average % of target region with depth > 50x	> 96%

Analysis time from FASTQ files: Overnight(4)

(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) The resolution of CNV detection, ranging from 2-5 exons, depends on the applied sequencing depth per sample. CNV detection is available for 98.1% of the CES genes (4408 genes)
(3) Performance metrics are based on high confidence regions in a reference sample. Values have been calculated on a reference sample and 20 M reads per sample on a HiSeq® instrument (300bp read length)
(4) 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 clinicians performing routine diagnostic testing. Thanks to its intuitive user interface and integrated features, variants visualization and interpretation are facilitated, while assuring protection of clinical genomic data.

Main features

Dedicated features in SOPHiA DDM reduce the complexity of determining the clinical significance of genomic variants.

- **Dual variant pre-classification:** Pre-classify variants according to both ACMG guidelines and SOPHiA's prediction to offer a comprehensive set of information to clinicians for improved assessment of variants pathogenicity.
- **Familial Variant Analysis (trio analysis):** Identify disease causing variants for different modes of inheritance, following a familybased approach
- **Virtual Panels:** Restrict the interpretation to sub-panels of genes of interest (e.g. eye disorders or hearing loss) or according to patient's consent to prevent incidental findings
- **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. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.