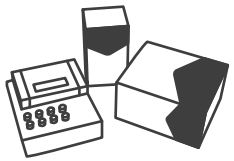
CLINICAL EXOME SOLUTION™ BY SOPHiA GENETICS

The Clinical Exome Solution (CES) by SOPHiA GENETICS is a molecular diagnostic application that bundles the analytical power of SOPHiA™ AI with a capture-based target enrichment kit and full access to SOPHiA DDM® platform.



Knowledge-Driven Kit Design

SOPHiA™

Collective AI for Data-Driven Medicine

SaaS Analytical Platform

The CES panel covers the coding regions (± 5 bp of intronic regions) of more than 4,900 genes with known inherited disease-causing mutations and spans 11 Mb of target region. It guarantees superior coverage uniformity, high on-target reads percentage and exceptional coverage in GC-rich regions, even in the first exon.

Gene panel

4,900 genes

Recommendations

Starting material: 200 ng

Sample source: Blood

Samples per run: Depending on sequencing platform[1]

Sequencer	Flow Cell/ Sequencing Kit	Recommended reads per sample	Samples per run
Illumina MiSeq	Kit v3 (2x300bp)	12M	4
Illumina NextSeq 500/550	Mid Output Kit v2 (2x150bp)	16M	16
	High Output Kit v2 (2x150bp)	16M	48
Illumina HiSeq 2500	High Output (2x125bp)	19M	24 (per lane)
	Rapid Run Mode (2x150bp)	19M	16 (per lane)

Wet lab

Day 1: Library Preparation

Day 2: Capture and Sequencing

Total hands-on time: 8 hours

SOPHiA analyses complex genomic NGS data by detecting, annotating and pre-classifying genomic variants such as SNVs and Indels to help clinicians better diagnose their patients.

SOPHiA leads to excellent clinical grade analytical performances:

	Observed
Sensitivity	> 99%[2]
Precision	> 99%[2]
Repeatability	> 99%
Average on-target rate	> 90%
Coverage uniformity	> 98%
Average percentage of target region > 50x	> 96%

Analysis time from FASTQ files: Overnight[3]

(1) Illumina®, MiSeq®, NextSeq® 500/550 and HiSeq® 2500 are registered trademarks of Illumina, Inc., which are not affiliated with SOPHiA GENETICS®

(2) Performance metrics are based on high confidence regions in a reference sample. Values have been calculated on a reference sample and 10 M fragments per sample (300bp read length)

(3) Analysis time may vary depending on the number of samples multiplexed and server load

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.

- **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
- **Interpretation Projects:** create interpretation projects on datasets by restricting the analysis to a specific set of genes, associated to a defined disease or reflecting patient's consent

Access to the World's Largest Clinical Genomics Community

Through SOPHiA DDM, experts from hundreds of healthcare institutions can easily interpret the variants and flag them with the appropriate level of pathogenicity. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.


SOPHiA™
The AI Democratizing Data-Driven Medicine
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