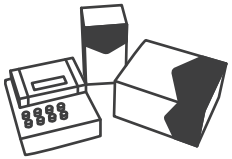
CLINICAL EXOME SOLUTION™ BY SOPHiA GENETICS

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™

The AI Democratizing Data-Driven Medicine

SOPHiA DDM®

SaaS Analytical Platform

The CES panel covers the coding regions (± 5bp of intronic regions) of more than 4,900 genes with known inherited disease-causing mutations and spans 11 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
4,900 genes

Recommendations
Starting material: 200 ng
Sample source: Blood
Samples per run: Depending on sequencing platform(1)

Sequencer	Flow Cell/ Sequencing Kit	Recommended samples per run (for 150x median coverage)
Illumina MiSeq®	v3 (2x300bp)	4
Illumina NextSeq® 500/550	Mid Output Kit v2 (2x150bp)	16
	High Output Kit v2 (2x150bp)	48
Illumina HiSeq® 2500	High Output (2x125bp)	24 (per lane)
	Rapid Run Mode (2x150bp)	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, Indels and CNVs(2) to help clinicians better diagnose their patients.

SOPHiA leads to excellent clinical grade analytical performance:

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

Analysis time from FASTQ files: Overnight(4)

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. The CES is also compatible with Thermo Fisher Scientific platforms
(2) The resolution of CNV detection, ranging from 2-5 exons, depends on the applied sequencing depth per sample
(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

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 with 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.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