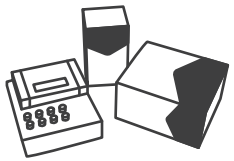
WHOLE EXOME SOLUTION™ BY SOPHiA GENETICS

The Whole Exome Solution (WES) 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 WES panel covers the coding regions (± 5 bp of intronic regions) of more than 19,000 RefSeq Genes and spans 39 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
>19,000 genes

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

Sequencer	Flow Cell/Sequencing Kit	Samples per run
Illumina NextSeq 500/550	Mid Output Kit v2 (2x150bp)	3
	High Output Kit v2 (2x150bp)	9
Illumina HiSeq 2500	High Output (2x125bp)	6 (per lane)
	Rapid Run Mode (2x150bp)	3 (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 to help clinicians better diagnose their patients. It enables accurate and comprehensive detection of SNVs and Indels for well-characterized regions of the panel.

SOPHiA leads to excellent clinical grade analytical performances:

	Observed
Sensitivity	> 99%(2)
Precision	> 99%(2)
Reproducibility	> 99%
Average on-target rate	> 90%
Coverage uniformity	> 98%
Average percentage of target region > 50x	> 96%

Analysis time from FASTQ files: Overnight(3)

All 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. intellectual disability or nephropathy) 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 variants and flag them with the appropriate level of pathogenicity. This highly valuable knowledge feeds the variant knowledge base and is anonymously and safely shared among all the members of the community.

(1) We recommend to have around 40M fragments per sample.
Illumina®, 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 40M fragments per sample (150bp read length)
(3) Analysis time may vary depending on the number of samples multiplexed and server load

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SOPHiA™
The AI Democratizing Data-Driven Medicine
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