SOPHiA™

SOPHiAGENETICS.COM

HEREDITARY DISORDER 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 HDS panel covers the coding regions (± 5bp of intronic regions) of 569 genes with known inherited disease-causing mutations and spans 2.5 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

569 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 >50x for >99.9% of target region)
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

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
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(3)

(1) Sequencing recommendations and specifications for other sequencing kits and instruments available upon request. The HDS 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) 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 family-based 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 large panels of genes

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.