SOPHiA™

SOPHiAGENETICS.COM

SOPHiA HOMOLOGOUS RECOMBINATION SOLUTION™

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

SMART KIT DESIGN

SOPHiA Homologous Recombination Solution covers coding regions and splicing junctions of 15 genes involved in the homologous recombination pathway associated with a wide range of malignancies, such as breast, ovarian and prostate cancers. Probe design is optimized to guarantee high on-target rate and coverage uniformity throughout the entire target regions.

Gene panel

ATM, BARD1, BRCA1, BRCA2, BRIP1, CDK12, CHEK1, CHEK2, FANCL, PALB2, PPP2R2A, RAD51B, RAD51C, RAD51D, RAD54L, TP53

Recommendations

Starting material: 10 ng minimum (50 ng recommended)
Sample source: FFPE, fresh-frozen tissue
Samples per run: Depending on sequencing platform(1)

Sequencer	Flow Cell/ Ion Chip Kit	Recommended samples per run (for 1000x coverage depth)
Illumina MiSeq®	v3 (2x300bp)*	8
	v2 (2x150bp)	4
Ion Torrent™ Ion S5™ System	Ion 540™	16

*2x150-cycle sequencing run (paired-end) is recommended

Wet lab

Day 1: DNA Library Preparation
Day 2: Capture and Sequencing
Total library preparation time: 1.5 days

ANALYTICAL POWER

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying genomic alterations to support experts for data-informed decision making. The application enables accurate and comprehensive detection of SNVs, Indels as well as CNV screening in all genes of the panel.

SOPHiA reaches advanced analytical performance(2):

	Observed	Lower 95% CI
Sensitivity	100%	96.05%
Specificity	100%	99.99%
Accuracy	100%	99.99%
Precision	100%	96.05%
Repeatability	99.97%	99.91%
Reproducibility	99.98%	99.97%

Analysis time from FASTQ files: 4 hours(3)

(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) Performance values have been calculated based on SNVs and Indels in samples processed on Illumina MiSeq®.
(3) Analysis time may vary depending on the number of genes, samples multiplexed and server load.

UNIVERSAL PLATFORM

The results are presented in SOPHiA DDM®, the platform of choice for experts performing routine genomic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic alterations. Data is kept safe by applying the highest industrial standards of encryption.

Main features

SOPHiA Platform offers several features, such as variant pre-classification and customized filtering options, that facilitate the data interpretation process.

OncoPortal™

SOPHiA OncoPortal is the intuitive interface for researchers, enabling access to relevant medical knowledge databases and clinical trial registries to determine the actionability and significance of genomic alterations.

Access to SOPHiA's Community

In SOPHiA, experts from more than one thousand healthcare institutions interpret the results and flag the pathogenicity level of variants according to their knowledge and experience. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community.

For Research Use Only. Not for use in diagnostic procedures.
SOPHiA GENETICS products are for Research Use and not for use in diagnostic procedures.
Somatic gene variant annotations and related content have been powered by, without limitation. The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).