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 16 genes involved in the homologous recombination pathway associated with a wide range of malignancies, such as breast and ovarian 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

Full coding regions for all genes

Recommendations

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

*The application can also be used with blood for germline analysis. For more information, please contact your local Sales and Business Development Manager

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 with their data-informed decision making. The application enables accurate and comprehensive detection of SNVs and Indels 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 genomic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic alterations. Data are kept safe by applying the highest industrial standards of encryption.

Main features

SOPHiA DDM offers several features that make variant analysis more efficient, such as hotspot screening which streamlines the visualization of mutated and wild type hotspot positions. With variant pre-classification and customized filtering options, experts can easily accelerate the data interpretation process.

OncoPortal™

SOPHiA DDM integrates the OncoPortal, a decision support functionality based on precision medicine intelligence. It enables experts to access relevant therapeutic, prognostic and diagnostic information to help determine actionability and clinical significance of detected genomic alterations. Moreover, the OncoPortal uses genes and disease association to maximize clinical trial matching.

Access to SOPHiA's Community

In SOPHiA DDM, experts from hundreds of 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.

Somatic gene variant annotations and related content have been powered by, without limitation, The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™).