SOPHiA™

SOPHiAGENETICS.COM

SOPHiA mini 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 mini Homologous Recombination Solution is a comprehensive application that covers the coding regions and splicing junctions (± 25bp min) of 4 genes involved in the homologous recombination pathway associated with but not limited to breast and ovarian cancers. Probe design is optimized to guarantee high on-target rate and coverage uniformity throughout the entire target regions.

Gene panel

BRCA1, BRCA2, RAD51C and 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)

Sequencer	Flow Cell Kit (sequencing run)	Recommended samples per run (for 1000x coverage depth)
Illumina MiniSeq™	High Output (2x150bp)	24
Illumina MiSeq®	v3 (2x150bp)	32
	v2 (2x150bp)	24

Wet lab

Day 1: DNA Library Preparation
Day 2: Capture and Sequencing
Total library preparation time: 8 hours

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

ANALYTICAL POWER

SOPHiA analyzes complex NGS data by detecting, annotating and pre-classifying genomic variants to support experts on data-informed decision making. It enables accurate and comprehensive detection of SNVs and Indels in all genes of the panel.

SOPHiA reaches advanced analytical performance:

	Observed
Sensitivity	99%
Reproducibility	100%
Average on-target rate	80%
Coverage uniformity	100%
Average % of target region with depth > 1000x	99%

Analysis time from FASTQ files: 4 hours(2)

(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) 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 is 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 databases to determine the actionability and significance of genomic alterations. Moreover, the OncoPortal uses inclusion and exclusion criteria to maximize clinical trial matching, both locally and at the global level.

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.

For Research Use Only. Not for use in diagnostic procedures.