SOPHiA™

SOPHiAGENETICS.COM

SOPHiA CARDIO 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

The CAS application covers the coding regions and splicing junctions (± 25bp) of 31 most relevant genes (target region of 131 kb), associated with arrhythmias (e.g. Long/Short QT syndrome or Brugada syndrome) and cardiomyopathies. Probe design is optimized to guarantee high on-target rate and coverage uniformity even in GC-rich regions, including the first exon.

Gene panel

CACNA1C, CACNA2D1, CACNB2, CASQ2, CTNNA3, DSC2, DSG2, DSP, HCN4, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LMNA, MYBPC3, MYH6, MYH7, MYL2, NKX2.5, PKP2, PLN, PRKAG2, RYR2, SCN10A, SCN5A, TMEM43, TNNI3, TNNT2, TRDN, TTR

Recommendations

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

Sequencer	Flow Cell / Ion Chip Kit	Recommended samples per run (for 250x median coverage depth)
Illumina MiSeq®	v3 (2x300bp)	32
Ion S5™	Ion 540	48

Wet lab

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

For Research Use Only. Not for use in diagnostic procedures

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, Indels and CNVs in all genes of the panel.

SOPHiA reaches advanced analytical performance:

	Observed	Lower 95% CI
Sensitivity	100%	97.36%
Specifity	100%	99.99%
Accuracy	100%	99.99%
Precision	100%	96.94%
Repeatability	100%	99.98%
Reproducibility	100%	99.97%
Average on-target rate	100%	
Coverage uniformity	99.51%	
Average % of target region with depth > 200x	99.78%	

Analysis time from FASTQ files: 4 hours(2)

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 variants. Data is kept safe by applying the highest industrial standards of encryption.

Main features

Dedicated features in SOPHiA DDM reduce the complexity of determining the significance of genomic variants.

- **Dual variant pre-classification:** Improve assessment of variants pathogenicity with the pre-classification of both ACMG guidelines and SOPHiA's prediction
- **Virtual Panels:** Restrict the interpretation to sub-panels of genes (e.g. focus on arrhythmias or cardiomyopathies)
- **Variant Filter Builder:** Define and edit custom filters for efficient analysis

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.

(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 samples multiplexed and server load