EXTENDED CARDIO SOLUTION™ BY SOPHiA GENETICS

The Extended Cardio Solution (ExtCAS) by SOPHiA GENETICS is a molecular diagnostic 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™

The AI Democratizing Data-Driven Medicine

SOPHiA DDM®

SaaS Analytical Platform

The ExtCAS panel covers the coding regions and splicing junctions (± 5bp) of 128 most clinically relevant genes (target region of 470 kb), associated with arrhythmias (e.g. Long/Short QT syndrome or Brugada syndrome) and cardiomyopathies. It guarantees high on-target rate and coverage uniformity even in GC-rich regions, including the first exon.

Gene panel

128 genes

For more information on the covered genes, visit: sophiagenetics.com/cardiosolutions

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)	12
Illumina NextSeq® 500/550	Mid Output Kit (2x150bp)	48
Ion S5™	Ion 540	16

Wet lab

Day 1: Library Preparation

Day 2: Capture and Sequencing

Total hands-on time: 8 hours

SOPHiA analyzes complex genomic NGS data by detecting, annotating and pre-classifying genomic variants to help clinicians better diagnose their patients. It enables accurate and comprehensive detection of SNVs, Indels and CNVs(2) in all genes of the panel.

SOPHiA reaches excellent clinical-grade analytical performance:

	Observed
Sensitivity	98.60%
Specificity	99.99%
Accuracy	99.96%
Precision	99.06%
Repeatability	99.96%
Average on-target rate	94.10%
Coverage uniformity	99.82%
Average percentage of target region with depth > 200x	100%

Analysis time from FASTQ files: 4 hours(3)

The results are presented in SOPHiA DDM, the platform of choice for clinicians performing routine diagnostic testing. Its intuitive user interface and advanced features facilitate the visualization and interpretation of genomic variants. Patient's 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 clinical 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, search for phenotype-related genes (HPO browser integrated)

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) CNV detection not available for exon 47 of *FLNC* and exons 172-197 of *TTN*

(3) Analysis time may vary depending on the number of samples multiplexed and server load