SOPHiA GENETICS™

Extended Cardio Solution

The genomic application that combines a capture-based target enrichment kit with the analytical capabilities and advanced features of the SOPHiA DDM™ Platform.

Main Features

SOPHiA DDM™ Extended Cardio Solution covers the complete coding sequence (± 5bp of exon-flanking regions) of 128 genes (target region of 470 kb) associated with arrhythmias (e.g. long/short QT syndrome or Brugada syndrome) and/or 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	Variants Called	Recommendations	Wet Lab
128 genes	SNVs Indels CNVs[1]	**Starting material** 200 ng DNA **Sample type** Blood **Samples per run for > 250x coverage depth / Sequencer (Flow Cell / Ion Chip Kit)** 8 for Illumina MiniSeq™ High Output Kit (2x150bp) 12 for Illumina MiSeq® v3 (2x300bp) 48 for Illumina NextSeq® 500/550 Mid Output Kit v2 (2x125bp) 96[2] for Illumina NextSeq® 500/550 High Output Kit v3 (2x150bp) 4 for Thermo Fisher Scientific Ion S5™ (Ion 530) 16 for Thermo Fisher Scientific Ion S5™ (Ion 540)	**Day 1:** Library Preparation **Day 2:** Capture and Sequencing **Total library preparation time: 1.5 days**

Analytical Performance[3]

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting, annotating and pre-classifying multiple types of genomic variants in all the genes of the panel.

Analysis time[4] from FASTQ: 4 hours

	Observed
Sensitivity	100%
Specificity	99.99%
Accuracy	99.99%
Precision	98.68%
Repeatability	99.99%
Reproducibility	99.98%
Average on-target rate	89.60%
Coverage uniformity	99.84%
Average % of target region with depth >200x	99.96%

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

Dedicated features in SOPHiA DDM™ reduce the complexity of determining the significance of genomic variants and facilitate the interpretation process, thus reducing turnaround time:

- **Dual Variant Pre-classification** - Improve assessment of variant pathogenicity with both ACMG scores and SOPHiA GENETICS™ machine learning-based predictions
- **Virtual Panels** - Restrict the interpretation to sub-panels of genes of interest using the HPO or OMIM® browser
- **Cascading Filters** - Apply custom filtering options for quicker screening of relevant variants and save strategies for future analyses

The platform can also provide access to Alamut™ Visual Plus, a full-genome browser that integrates numerous curated genomic and literature databases, guidelines, missense and slicing predictors, thus enabling a deeper variant exploration. After the interpretation, you can generate a customizable variant report, including valuable information to support decision making.

Global support at every step

We offer local support anywhere in the world. Our dedicated bioinformaticians help save time and resources, ensuring fast resolution of workflow disruptions. In addition, our Set Up Program provides assistance with assay set up for fast and worry-free transition to routine testing.

Secure data storage

Access to the SOPHiA DDM™ Platform is restricted to registered users only. The Platform keeps data safe by applying the highest industrial standards of encryption in compliance with your local data security policies.

Access to the SOPHiA GENETICS™ community

In the SOPHiA DDM™ Platform, experts from hundreds of healthcare institutions interpret their 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. CNV detection not available for exon 47 of *FLNC* and exons 172-197 of *TTN* due to the presence of homologous regions.
2. Maximum number of indices available.
3. Performance metrics were calculated on 177 distinct confirmed variants in 9 distinct samples. Sequencing was performed using an Illumina MiSeq® instrument.
4. Analysis time may vary depending on the number of samples multiplexed and server load.

For Research Use Only. Not for Use in Diagnostic Procedures.