CONTACT US

SOPHiA DDM™ for Devyser CFTR

Enabling molecular laboratories to maximize the utility of Devyser CFTR

SOPHiA DDM™ for Devyser CFTR is a fully integrated bioinformatic workflow (FASTQ to Report) to streamline your variant interpretation and reporting.

Analytical performance

Detection of:

- SNVs
- Indels
- CNVs (23 CNVs, including direct detection of 8 common pathogenic CNVs, CFTRdele1, CFTRdele2, CFTRdele2ins182, CFTRdele2,3, CFTRdele14b–17b, CFTRdele17a–18, CFTRdele22,23, CFTRdele22–24)
- Polythymidine variants (5T/7T/9T) within intron 9 (IVS8) and TG-repeat number upstream of the poly-T region

Full access to analytical output:

- At-a-glance quality display
- Access to source files

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[1] from FASTQ: 4 hours

	Observed	Lower 95% CI
Sensitivity	100%	83.33%
Specificity	100%	99.96%
Accuracy	100%	99.96%
Precision	100%	83.33%
Average on-target reads	96%	
Coverage uniformity	99.67%	
Average % of target region with depth >200x	100%	

The pipeline is compatible with Illumina MiniSeq™ and MiSeq® v2 and v3 sequencers.

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
- **Cascading Filters** - Apply custom filtering options for quicker screening of relevant variants and save strategies for future analyses

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 a fast and worry-free transition to routine testing.

Secure and unlimited data storage

Access to the SOPHiA DDM™ platform is restricted to registered users only. The platform provides unlimited and unrestricted storage, while keeping 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. Analysis time may vary depending on the number of samples multiplexed and server load.

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