

SOPHiA DDM™ for KAPA HyperExome

Enabling molecular laboratories to maximize the utility of KAPA HyperExome

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

Analytical performance*

Detection of:

- CNVs in 18,578 genes¹
- SNVs & Indels in all 24,377 genes of the panel²

Full access to analytical output:

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

	Performance metrics ³	Observed	Lower CI (95%)
Genomic variations			
SNV & Indels ^{3,4}	Sensitivity	99.2%	99.2%
	Precision	99.2%	99.2%
CNVs ⁵	Sensitivity	89.3%	-

The pipeline is compatible with Illumina NextSeq® 550 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:

- **GRCh38/hg38 based analytics** - Annotate variants accurately
- **Dual-Variant Preclassification** - Improve assessment of variants pathogenicity based on both ACMG scores and SOPHiA GENETICS' 5 algorithm-supported predictions
- **Virtual Panel** - 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
- **Familial Variant Analysis (trio-analysis)** - Identify disease causing variants considering different modes of inheritance, through a family-based approach

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 available globally. Our Set Up Program provides assistance with assay set up for fast and worry-free transition to routine testing. All along the workflow, our robust support, including re-analysis and dedicated bioinformaticians, help save time and resources to ensure fast resolution of workflow disruptions.

Secure and unlimited data storage

SOPHiA DDM™ platform provides unlimited and unrestricted storage, while keeping data safe by applying the highest industrial standards of encryption in compliance with local data security policies.

Access to SOPHiA community

Through the SOPHiA DDM™ platform genomics experts from >1000 healthcare institutions interpret their findings and flag the pathogenicity level of variants. This highly valuable information enriches the variant knowledge base and is safely shared among the members of the community, supporting their decision-making process for research purposes.

1. CNV detection with resolution of 2-5 exons, depending on the applied sequencing depth per sample.
 2. SNV and indel detection depends on applied sequencing depth per sample.
 3. SNV and indel performance metrics are based on 68906 variants in the high confidence regions of three reference samples. For each sample, 80 M 101 bp reads (40 M read pairs) were used. Sequencing was performed using an Illumina NextSeq instrument.
 4. Analytical performance for SNV and indel detection was verified using three difference reference samples in an independent sequencing run. Sensitivity: 99.8% (CI 99.8%); precision: 99.6% (CI 99.5%).
 5. CNV detection with resolution of 2 exons in 80 M 150 bp reads.

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