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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 genes [1]
- SNVs & Indels in all 24,377 genes of the panel [2]

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 variant pathogenicity with both ACMG scores and SOPHiA GENETICS™ machine learning-based 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

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 interpretation, you can generate a customizable variant report that includes 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 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 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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