SOPHiA GENETICS™

SOPHiA DDM™ for Whole Genome Sequencing

SOPHiA DDM™ for Whole Genome Sequencing (WGS) is an all-in-one bioinformatic workflow (FASTQ → Report) for the analysis of variants associated with rare and inherited disorders.

Advanced Secondary Analysis

Compatible with

PCR-free WGS enrichment technologies

Illumina sequencers

Variant types

Detects coding & non-coding variants in genomic and mitochondrial DNA, covering constitutional & mosaic events including:

- SNVs/Indels
- CNVs
- RoH
- SVs

Analytical Performance

Performance metric	Observed
Sensitivity for SNVs/Indels	>99%
Sensitivity for RoH[1]	99%
Sensitivity for constitutional CNVs[2]	>99%
Sensitivity for mosaic CNVs[3]	95%
F-score for structural variants	>75%

1. For events ≥5Mb.
2. For events ≥10kb.
3. For events ≥5Mbp and mosaic fraction as low as 30%.

Accelerated Tertiary Analysis

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** – Select your transcript of choice and leverage insights from frequently updated variant annotation catalogs and zygosity annotation
- **Variant pathogenicity prediction** – Prioritize variants for further analysis with point-based ACMG classification, phenotypic classification, and SOPHiA DDM™ AI-driven ABCD prediction
- **Virtual Panels** – Restrict interpretation to sub-panels of genes of interest using the HPO or OMIM®
- **Cascading Filters** – Apply custom filtering options and save strategies for future analyses
- **IGV Explorer** – Intuitively visualize coverage and BAF plots for CNV and SV assessment
- **Alamut™ Visual Plus** – Streamline your interpretation with enhanced genomic visualization and comprehensive variant annotation
- **Reporting** – Generate customized CAP- and CLIA-compliant reports

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.

Secure data storage

The SOPHiA DDM™ Platform keeps data safe by applying the highest industrial standards while complying with 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. These expert perspectives can assist users in determining the true pathogenicity of their detected variants.

ACMG, American College of Medical Genetics and Genomics; CNV, copy number variation; HPO, Human Phenotype Ontology; OMIM, Online Mendelian Inheritance in Man; RoH, runs of homozygosity; SNV, single nucleotide variant; SV, structural variant; WGS, whole genome sequencing.

Product code:
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