SOPHiA GENETICS™

SOPHiA DDM™ for Whole Genome Sequencing

Unearth genetic revelations with WGS

A unified workflow from data to discovery

Find answers from the depths of your data with our fast and accurate application that enables you to analyze, visualize, and explore your genome results in a single, streamlined workflow.

Powered by:

NEW GENERATION SOPHiA DDM™ PLATFORM

Rapid Upload

(CLI or background uploader)

FASTQ → QC → Align

Accurate, AI-powered Variant Calling



Coding & non-coding variants, gDNA and mtDNA

DEEPPER™

SNV/Indels

MUSKAT™

CNVs | RoH | Structural variants

Comprehensive Tertiary Analysis



Add on alamut™ VISUAL PLUS

MOKA™

Annotation → Filtration → Interpretation

Intuitive Visualization and Reporting



Genome browser

Customizable Reports

30x and low-pass WGS applications will be available.

NVIDIA

Powerful **NVIDIA GPUs** process computationally heavy workloads to **provide a scalable and efficient solution**

Microsoft Azure

Samples are run in parallel on their own Microsoft Azure nodes for **high scalability**

Advanced analytical performance

SNV/Indels: >99% sensitivity

RoH: 99% sensitivity (for events ≥5Mb)

Constitutional CNVs: >99% sensitivity (for events ≥10kb)

Mosaic CNVs: 95% sensitivity (for events ≥5Mbp and mosaic fraction as low as 30%)

Structural variants: >75% F-score

HPO and AI-driven variant prioritization

Including ACMG score

Community-shared pathogenicity flagging

With the highest standards of data privacy and security

WANT TO KNOW MORE

Contact us at → info@sophiagenetics.com

CLI, command line interface; CNV, copy number variation; gDNA, genomic DNA; GPU, graphics processing unit; mtDNA, mitochondrial DNA; QC, quality control; RoH, runs of homozygosity; SNV, single nucleotide variant.

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