SOPHiA GENETICS™

SOPHiA DDM™ for Twist Human Comprehensive Exome

SOPHiA DDM™ for Twist Human Comprehensive Exome on Illumina NovaSeq™ 6000 is a fully integrated bioinformatic workflow (FASTQ to Report) that utilizes the analytical capabilities and advanced features of the SOPHiA DDM™ Platform to streamline the detection, analysis, and interpretation of genomic variants.

Main Features

Gene Panel

- 21,093 genes
- Entire mitochondrial genome

Variants Called

- SNVs
- Indels
- CNVs (96.3% of target regions)
- mtDNA variants

Full access to analytical output

- At-a-glance quality display
- Access to source files
- Fully optimized for NovaSeq data

Analytical Performance[1]

The SOPHiA DDM™ Platform analyzes complex NGS data by detecting, annotating and pre-classifying genomic variants.

Nuclear DNA	
Sensitivity for SNVs/Indels	>99%
Precision for SNVs/Indels	>99%
Average % coverage of target region with depth >50x	>99%
Sensitivity for CNVs 2-4 exons (1-2 exons)	>99% (98%)
Mitochondrial DNA	
Sensitivity for SNVs/Indels	>99%
Precision for SNVs/Indels	>99%
Average % coverage of target region with depth >100x	>99%

One Simple Intuitive Platform: Beyond Analytics

Accelerated assessment and reporting of genomic variants

Dedicated features in SOPHiA DDM™ reduce the complexity associated with determining the significance of genomic variants and facilitate the interpretation process for streamlined turnaround times:

- **GRCh38/hg38 based analytics -** Annotate variants accurately
- **Dual Variant Pre-classification -** Improve assessment of variant pathogenicity with both ACMG scores and SOPHiA GENETICS machine learning-based predictions
- **Virtual Panels -** Restrict interpretation to genes of interest using the HPO or OMIM® browser
- **Cascading Filters -** Apply custom filtering options to pinpoint relevant variants and save strategies for future analyses
- **Familial Variant Analysis (trio-analysis) -** Identify disease-associated variants considering different modes of inheritance, through a family-based approach.

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 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.

Product code: DL4013ILLRGL

CNV, copy number variation; mtDNA, mitochondrial DNA; NGS, next-generation sequencing; SNV, single nucleotide variant.

1. Analytical performance metrics were calculated from at least 3 blood samples run on an Illumina NovaSeq™ 6000 instrument with 80M reads per sample, using a dilution of mtDNA:nDNA of 1:100. For mtDNA, numbers were calculated for all samples pooled together due to the smaller number of variants (188 SNVs, 3 Indels).

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