SOPHiA GENETICS™

From data to insights to confident care

SOPHiA DDM™ PLATFORM FOR HEREDITARY DISORDERS

The SOPHiA DDM™ platform offers streamlined workflows (from FASTQ file to variant report) to accurately assess genomic variants associated with hereditary disorders. Designed to be secure, the platform leverages machine learning-patented algorithms and intuitive features to confidently detect, annotate and pre-classify multiple types of variants in a single workflow. As a result, clinical researchers save time and effort in spotting the variants of interest from complex and noisy genomic data sets.

HIGHLIGHTS

Achieve complete genomic analysis

SOPHiA DDM™ offers optimized read alignment to confidently call multiple types of variants in one unique experiment. The platform utilizes complete GRCh38/hg38-based analytics for variant annotation and performs comprehensive transcript annotation with MANE*.

SNVs/Indels	✓	*Alu* insertions*	✓
CNVs	✓	Boland inversion*	✓
Mitochondrial variants*	✓	Pseudogene variants*	✓

*Depending on the gene panel

Ensure data security

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 platform, experts from hundreds of healthcare institutions interpret the 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.

Complete variant annotation and exploration on a full-genome level

The SOPHiA DDM™ platform pulls information from a broad range of databases, allowing comprehensive annotation and visualization of genomic variants. In addition, the platform offers dedicated features for efficient variant prioritization, facilitating the interpretation process. For advanced tertiary analyses, users can also access Alamut Visual Plus™, a full-genome browser that integrates numerous curated genomic and literature databases, guidelines, and missense and splicing predictors. The software offers advanced visualization, customizable reporting, and local data management, providing a comprehensive solution for accelerating complex variant assessment.

Scalable

From targeted to comprehensive exome applications

Efficient

Multiple types of variants detected in a single experiment

User friendly

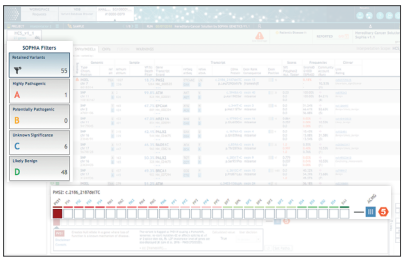
No bioinformatics expertise needed

SOPHiA DDM™ for Hereditary Disorders

Fast, easy, and intuitive workflow

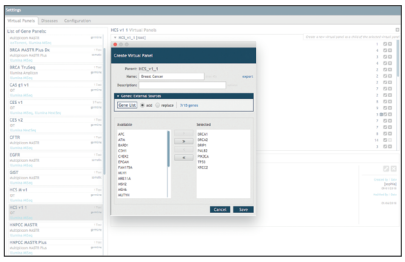
SOPHiA DDM™ offers comprehensive workflows with dedicated functionalities, enabling experts to manage the large amount of data coming from NGS-based applications (from targeted panels to whole exomes) and easily identify, visualize, and report any variants of interest, thus reducing turnaround time.

Integrated features for efficient variant prioritization and interpretation



Dual Variant Pre-Classification

Improve assessment of variants' pathogenicity based on both ACMG scores and our machine learning-based predictions.

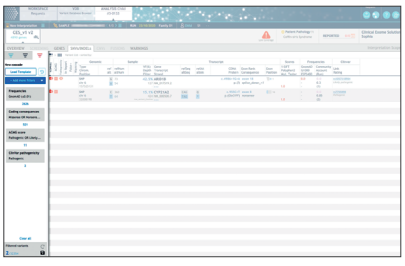


Virtual Panel

Restrict interpretation to sub-panels of genes using the HPO or OMIM® browser.

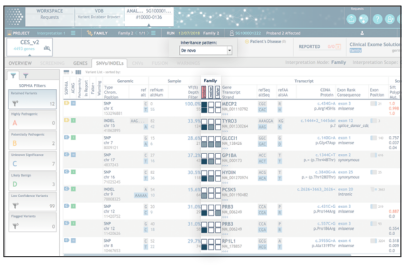
KEY ADVANTAGES

- Focus only on relevant variants
- Streamline variant analysis
- Shorten turnaround time



Cascading Filters

Apply custom filtering options for quicker screening of relevant variants and save strategies for future analyses.



Familial Variant Analysis (trio-analysis)

Quickly identify causative variants by selecting different inheritance modes with a single mouse click and shortening the candidate variant list accordingly.

Filtering utility