

Get the most out
of your exome

SOPHiA Platform streamlines your workflow for accurate analysis of exome data to detect multiple variants, including CNVs.



Universal Platform

Custom-tailored for each and every exome technology



Set Up Program

Support to go-live in record time



SOPHiA Community

Faster, more informed interpretation

*“My experience with SOPHiA has demonstrated that the SOPHiA Platform accurately detects single nucleotide variants, indels, and **copy number variants** in gene panels and exome-sized applications.”*

– **Daniel Bellissimo**, PhD Director University Pittsburgh Medical Center Clinical Genomics Laboratory

High performance variant detection

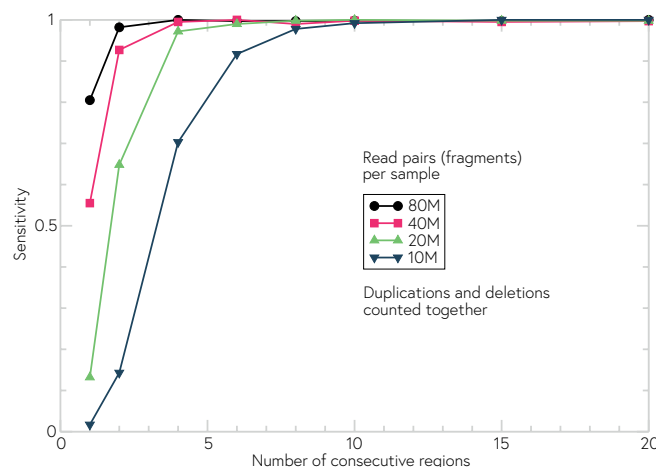
Tailored specifically to each exome application, SOPHiA's AI-powered technology ensures high quality data and improved accuracy for secondary analysis, and offers extremely uniform coverage even in complex regions, including GC-rich areas.

SOPHiA Platform provides accurate CNV detection down to the **exonic** resolution in a single workflow, including SNVs and Indels.

- ✓ SNVs
- ✓ Indels
- ✓ CNVs

> 90% sensitivity in CNV detection

including Twist™ Human Core Exome powered by SOPHiA*



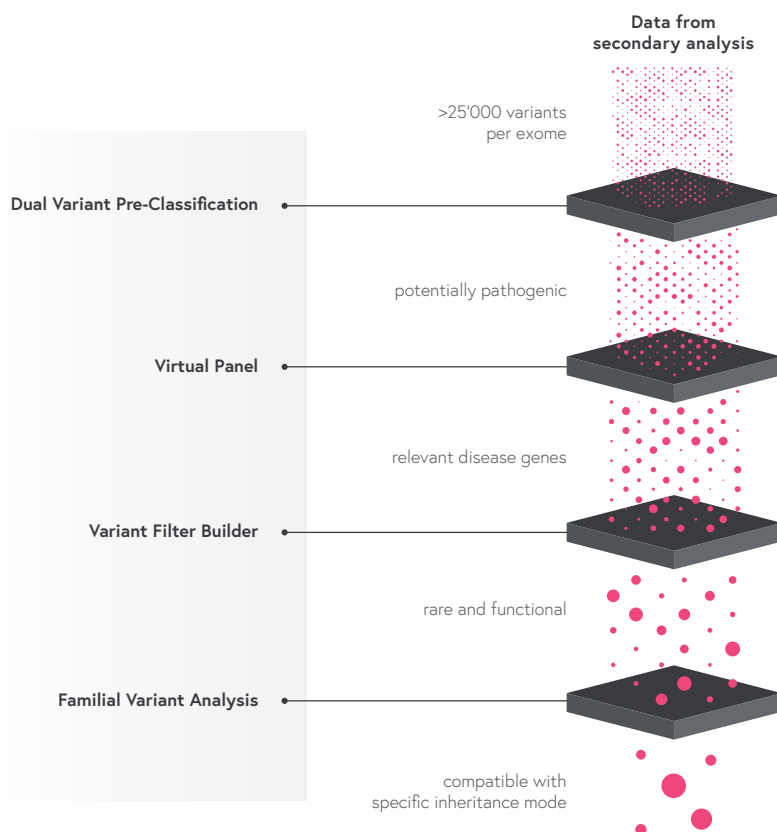
* > 90% Sensitivity of CNV detection in two consecutive regions (exons) with 40 million fragments (80 million reads) based on internal data.

Efficient variant prioritization to streamline your interpretation

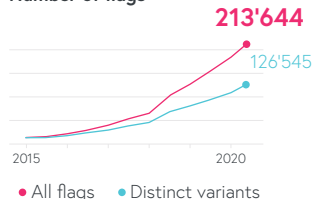
SOPHiA Platform features advanced filtering options to manage large sets of data and narrow the search down to relevant variants, starting with SOPHiA and ACMG pre-classification, Virtual Panel to limit the interpretation to a subset of genes, Variant Filter Builder for custom filtering strategies, and Familial Variant Analysis to select different inheritance modes.

SOPHiA Community for secure knowledge sharing

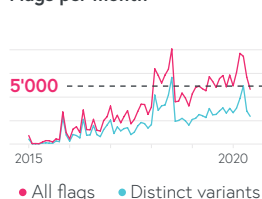
In SOPHiA Platform, experts in institutions from around the world can flag the pathogenicity level of variants in accordance to their knowledge. This highly valuable information feeds the variant knowledge base and is anonymously and safely shared among the members of the community. Institutions retain complete control of the data and all the information are processed ensuring security and privacy.



Number of flags



Flags per month



19.7% of the flagged variants in SOPHiA Platform are not found in public databases

Want to know more? info@sophiagenetics.com

SOPHiA GENETICS products are for Research Use and not for use in diagnosis.

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