Cloud deployments in

On-premises installation available upon request

VarSome Clinical is a Class C medical device per Rule 3 of Annex VIII of the Regulation (EU) 2017/746 on in vitro diagnostic Medical Devices (IVDR).

VarSome Clinical is an online software application that can process the results of Next-Generation Sequencing (NGS), generating genetic variation information based on standard guidelines and databases with clinical evidence.

Manufacturer: Saphetor SA | EC REP: Saphetor SA Greek Branch | Please read the user manual carefully.

VSC-BR-001-V01

SAPHETOR

We exist to enable anyone to find, share, and use the most comprehensive human genome data available.

We don't simply offer a Clinical Decision Support platform. We provide a home to a global community of connected healthcare professionals, enabling them to collaborate and improve lives around the world.

varsomeclinical

Your NGS data analysis solution

Built with Swiss precision by

SAPHETOR

IVDR CERTIFIED

varsomeclinical

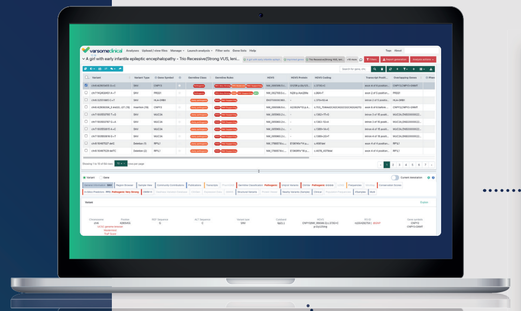
Powering your daily analysis

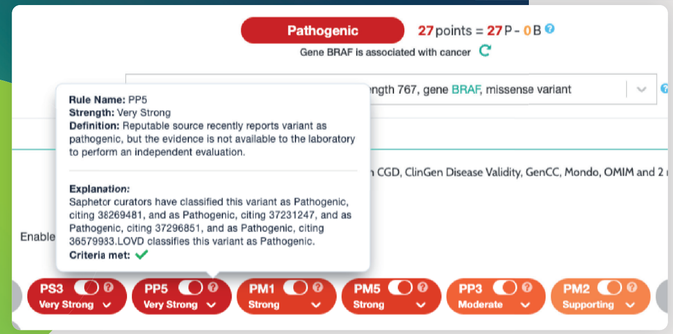
Annotation and classification

- Germline classifier based on the ACMG guidelines, providing pathogenicity predictions for all variants (including those that have not been previously described).
- Pathogenicity predictions with verdicts clearly explained.
- Supporting evidence of why classification rules have or have not been triggered.
- Integration of curated information from the VarSome curators available to enhance the classification of VUS variants.
- CNV classification based on the ACMG guidelines and somatic classification based on the AMP guidelines also available.
- Modify the variant classification manually and save it for future samples.
- Analysis results are frozen with annotation at time of processing. Live-annotation of variants is possible from within the analysis to see if new data has impacted predicted pathogenicity. Full-sample re-annotations are also possible.

Over 140 data sources

- LOVD
- ClinVar
- gnomAD
- OMIM®
- JAX CKB
- PharmGKB
- and more...





Upload FASTQ or VCF

- We support FASTQ files from Illumina and MGI sequencers.
- We support all VCF files (including tertiary analysis for long-read technologies).
- Analyze different types of files, from small panels to whole exome and genome sequences.
- Automate file uploads through our VarSome Clinical API or connect your Illumina BaseSpace account.

Workflows and filtering

- Create workflows for each analysis type and streamline your processes for you and your team.
- Adjust your team settings to give users permissions based on their expertise or role.
- Create and apply dynamic filters and gene lists to reduce your search space.
- Apply advanced filters to find *de novo* variants from a trio analysis, perform carrier screening for couples, and more.
- Prioritize variants that are more likely to be causative, with VarSome Picks; a phenotype-driven algorithmic filter.

Customizable reports

- Branding and layout of reports can be adjusted to suit your lab's branding policy.
- PharmCat integration offers pharmacogenomic reports.
- CIVIC and related Clinical Trials information, if available.

VarSome.com maintains, harmonizes, and integrates over 140 genomic databases, over 4 million publications, and robust classification tools to give you a comprehensive variant interpretation engine.

Visualize your sample

Sample View

The Sample View offers a comprehensive visualization of all the variants detected in the sample, integrating small variants and CNVs into a single display. Explore variant locations at the gene level using the MANE transcript, or include alternative transcripts in the view. Tailor your view by filtering variants according to their pathogenicity or coding impact.

CNV Browser

Discover challenging CNVs in your samples and visualize them with our new CNV Browser.

Intuitively designed to help you discern real CNVs from artefacts, our CNV browser offers a genome-wide view for all of your samples with one click.

Publications Explorer

Visualize publication trends with our publications timeline viewer, displaying data on PubMed articles relating to your gene, variant, or phenotype of interest.

Supplement your VarSome

varsomepremium

Understand candidate variants by using VarSome Premium. A powerful search tool to scour the full VarSome database to view comprehensive variant information and classifications.

varsomeapi

VarSome API is the perfect solution to integrate the VarSome database and classification tools using Python, R, Perl, Java, or any other programming language.