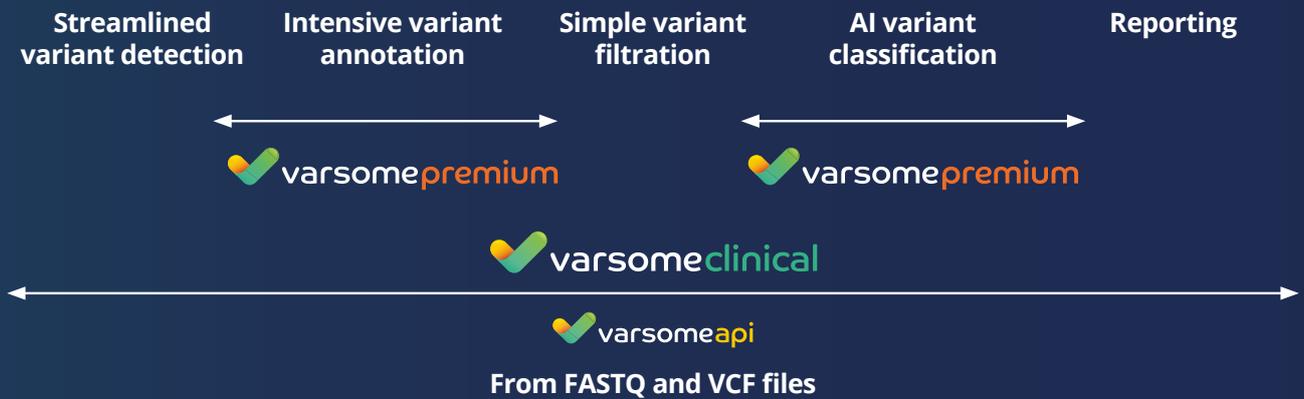




Your complete NGS data
analysis solution

Powering your daily analysis



VarSome Clinical is a variant annotation and interpretation platform for NGS data, for whole genomes, exomes, and gene panels. Explore its endless possibilities, which include:

Upload FASTQ or VCF

- Variant calling from FASTQ powered by Sentieon.
- Perform risk carrier screening for couples, analysis of tumor-normal samples, trio analysis, or small cohort analysis.

Dynamic filtering

- With our dynamic filters you have the ability to reduce your search space.
- Prioritize your causative variants with our fully customizable algorithmic filters.

Customizable reports

- Branding and layout of reports can be adjusted to suit your lab's branding policy.
- PharmCat integration offers pharmacogenomic reports.

Annotation and classification

- Over 140 databases including OMIM®, LOVD, ClinVar.
- Over 4 million linked publications.
- Curated publications.
- Automated real-time classification for germline*, somatic**, and CNV*** according to industry-standard guidelines.
- Splicing and missense predictions to decide whether some rules should be applied or to boost their strength.
- Tissue-specific variant expression data from the Genotype-Tissue Expression (GTEx) project.
- MANE, Ensembl, and RefSeq transcripts.
- Protein annotation.

VarSome Clinical is a medical device, from Annex III of IVDD 98/79/EEC.

VarSome Clinical aids in making an informed decision and diagnosis, and in the selection of appropriate treatments for cancer, Mendelian, and other genetic disorders by processing next generation DNA sequencing data.

Manufacturer: Saphetor SA

EC REP: Saphetor SA Greek Branch

Please read the user manual carefully.

Variant	Variant Type	Gene Symbol	ACMG Class	ACMG Rules	HGVS	HGVS Protein	HGVS Coding	Chromosome Position	Overlapping Scores	Interpretations	Severity
chr10:111111111	Deletion (del)	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111111del	chr10:111111111
chr10:111111112	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111112C>T	chr10:111111112
chr10:111111113	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111113G>A	chr10:111111113
chr10:111111114	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111114A>G	chr10:111111114
chr10:111111115	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111115T>C	chr10:111111115
chr10:111111116	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111116C>G	chr10:111111116
chr10:111111117	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111117G>T	chr10:111111117
chr10:111111118	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111118A>T	chr10:111111118
chr10:111111119	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111119T>G	chr10:111111119
chr10:111111120	SNP	BRCA1	Pathogenic (P)	...	NC_010907.4:g.111111120C>A	chr10:111111120

* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4544753/>

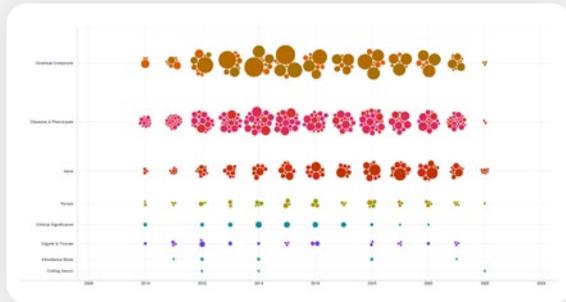
** <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5707196/>

*** <https://pubmed.ncbi.nlm.nih.gov/31690835/>

Visualize your results in a user-friendly interface

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.

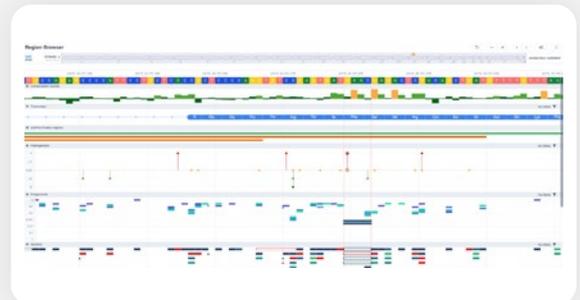


Publications Explorer

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

Region Browser

Our user-friendly browser brings together a wealth of omics data, including on-the-fly germline classification in a single view, so you can focus on what matters most: translating variant interpretation to clinical insight.



Supplement your VarSome



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



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



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.

Built with Swiss precision by

