varsome

VARIANT INTERPRETATION

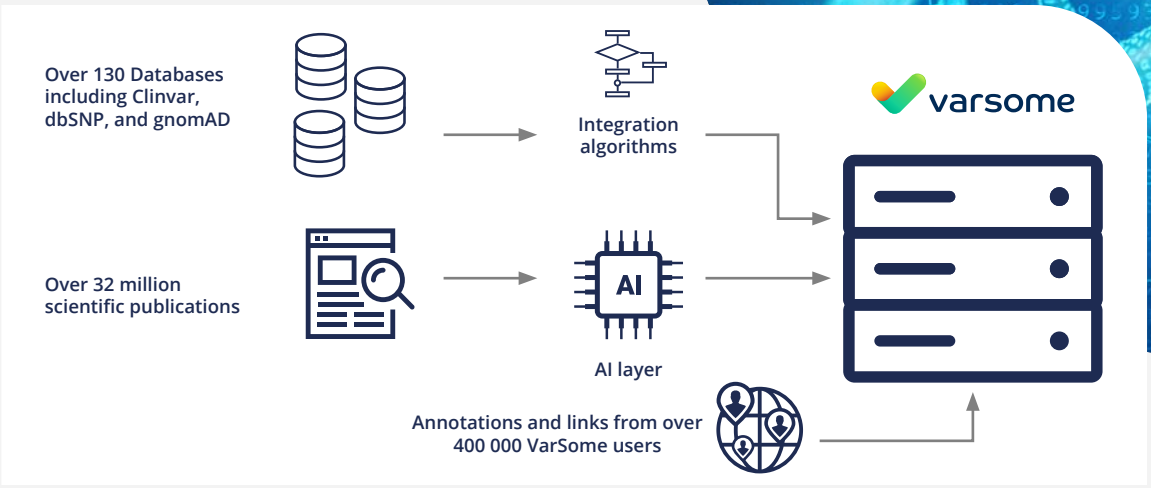
FIND ANSWERS FASTER

VarSome

The Global Genomics Community

VarSome compliance and certifications
CE-IVD, HIPAA & GDPR Compliance, Hébergeur de Données de Santé Compliance, ISO 13485 (Quality Management Systems for Medical Devices), ISO 27001 (Information Security Management), IEC 62304 (Software Life Cycle Processes for Medical Devices).

What is VarSome



Your Variant Interpretation Engine

VarSome maintains, harmonizes, and integrates over 130 genomic databases and over 32 million publications to give you the most comprehensive variant interpretation engine there is. The complete list of data sources can be viewed at https://varsome.com/datasources.

VarSome's Network Effect
Over 400 000 users around the world add variant classifications, link publications, and provide additional annotation.

Industry-Leading Variant Classification

Cited in over 1 700 peer-reviewed publications, VarSome's proprietary ACMG*, AMP**, and CNV classifiers apply industry standard variant interpretation guidelines to help you understand your variants. Unlike other static platforms, our classifiers are dynamic and work in real-time making it possible to classify even novel variants. To ensure high levels of confidence, VarSome's classifiers provide transparency on evidence used and rules triggered as well as giving users the ability to adjust rule weighting.

Why VarSome VarSome fits around you

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Search
For labs that only need to look up known variants of interest.

varsomeclinical

Analyze
For labs generating NGS data needing fast, scalable, and powerful variant interpretation.

varsomeapi

Code
For Bioinformatics groups needing to integrate the VarSome database and classifiers into existing in-house pipelines.

*https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4544753/
**https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5707196/

Search

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Who is it for: Individuals in need of individual gene or variant lookups.

VarSome.com is a free-to-use search engine for querying the VarSome database. Users can search by HGVS nomenclature, rsID, gene name, transcript symbol or genome location. However, the VarSome database contains much more than just variant frequencies. Our complete search functionality supports word searches allowing you to find publications, diseases, phenotypes, genes, ClinVar & UniProt variants, and user comments.

Classifiers

The VarSome database powers the classification algorithms that apply ACMG* and AMP** interpretation guidelines to germline, somatic, and copy number variants. Every result provides clear natural language explanations as to why specific rules were triggered and the accompanying evidence. Similarly, a full explanation is provided when threshold criteria have not been met. As the rules were originally designed to be applied by clinicians with the benefit of case knowledge, VarSome allows users to adjust the weighting of rules to better fit their case.

VarSome and ClinVar
ClinVar links directly to VarSome result pages. Similarly, you can make ClinVar Submissions directly from VarSome.

Community

Over 400 000 users have visited VarSome, adding comments, linking publications, and providing advice. As many variants are rarely seen by any one individual, the VarSome Community ensures that knowledge and experience is shared. Users around the world have connected via VarSome to share expertise.

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VarSome Premium exists for professional users requiring a high volume of queries. Additionally, Premium provides access to additional databases and full FASTQ variant calling and VCF annotation.

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Analyze

varsomeclinical

Who is it for: Labs generating NGS data

VarSome Clinical is a CE IVD-certified and HIPAA-compliant platform that brings the power of VarSome to your Ilumina and BGI FASTQs and any VCFs.

As genomic approaches have become more widespread in research and in the clinic, there are several reasons why VarSome Clinical is a crucial addition to your analysis pipeline.

Illumina BaseSpace
You can integrate your Illumina BaseSpace account with VarSome Clinical for easy direct uploads of your FASTQs and VCFs.

Speed

VarSome Clinical employs dynamic and fully customizable algorithmic filters. This means that you can reduce your variant load dramatically to just your variants of interest – whether they be pathogenic variants, gene-list based, compound heterozygous, variants in imprinted genes or even *de novo* variants. VarSome Clinical's algorithmic filters put you and your workflow in control.

Interpret Novel Variants

VarSome applies ACMG* and AMP** interpretation guidelines to germline, somatic, and copy number variants. These are applied dynamically, rather than being looked up from a database as with other systems. This means that every variant will have the full weight of the VarSome database applied to it by the selected classifier algorithm to provide a suggested level of pathogenicity.

Family Analysis

VarSome Clinical's Trio analysis takes advantage of parental information to aid in diagnosing children with rare disease. Our Trio filters, combined with our variant classification filter, makes pediatric whole genomes and exomes much easier to interpret.

Customizable Reports

VarSome Clinical recognizes that every lab has its own way of doing things. For that reason, reports are fully customizable to include only the information you need and your own branding.

Your Data; Your Rules

Although we are HIPPA and GDPR compliant, CE-IVD and ISO 27001 and ISO 13485 certified, we recognize that every organization has different obligations when it comes to safeguarding data. If your country does not allow data to leave its borders, we offer an on-premises installation of VarSome Clinical to ensure data protection compliance.

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

Code

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Who is it for: Bioinformatics groups needing to integrate the VarSome database and classifiers into existing inhouse pipelines.

Free Your Team

VarSome was designed to support all types of users. As the field of genomics matures, we know that many groups have already built robust analysis pipelines. In these instances, maintaining databases can be a costly and time-consuming endeavour. VarSome API is the perfect solution for these groups. By connecting to our API, you can free your team to focus on analysis rather than database management.

Use The Best

The way we apply ACMG* and AMP** guidelines to classify pathogenicity of germline, somatic, and copy number variants makes it ideally suited to exomes and whole genomes. As a result, clinical labs, academic and industry research labs, and even other analysis platforms have accessed the VarSome API to integrate our industry-leading variant classifiers.

Easy To Use

The API has been designed to be as simple as possible. A REST API is provided and data requests are returned as JSON. This makes it ideal to integrate into Python, R, Perl, Java, or any other programming language.

Although we are HIPPA and GDPR compliant, CE-IVD and ISO 27001 and ISO 13485 certified, we recognize that every organization has different obligations when it comes to safeguarding data and metadata. For the highest levels of privacy, we offer the installation of the VarSome API on a dedicated server that we will update in line with our own API servers.

Find

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Who is it for: Clinical trial sponsors and managers

VarSome has over 400 000 users active in over 150 countries around the globe. Over 80% of users have opted-in to have their details shared with representatives of a clinical trial relevant to their VarSome searches.

Help ensure your patients have the opportunity to participate in the latest clinical trials

Supporting Clinical Trial Recruitment

Active Outreach
We can contact VarSome users, who have searched for specific genes or gene variants, to let them know that a clinical trial representative would like to speak to them to understand if they have a patient that may be eligible to enrol.

Digital Messaging
We can display prominent messaging on specific VarSome pages of interest, alerting viewers to an active trial or study. This messaging can be targeted to specific geographies.

In both instances we work with the trial sponsor to ensure all messaging and content is appropriate and complies with all laws and regulations in each country.

Global Distribution
We can analyze specific genes to produce a map of global distribution of relevant VarSome queries. This report is a powerful tool for identification of potential trial sites.

Where there is already an approved therapy available, VarSome Insights can also be used to support Medical Affairs and Commercial functions.

Getting Started with VarSome

Get in touch with our team at **sales@varsome.com** for a free consultation. Our team will work with you to define your variant interpretation needs and help you to test VarSome in your workflow.

Partnerships Opportunities

If you are a technology developer interested in partnering to further develop an opportunity with VarSome, or becoming a distribution partner, contact our Head of Strategic Alliances Tomáš Kučera at **tomas.kucera@varsome.com**

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SAPHETOR

VarSome Suite is brought to you by Saphetor SA.