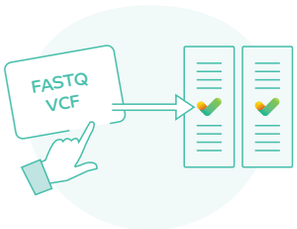
varsomeclinical

CLINICAL TOOL

End-to-End Solution for NGS

VarSome Clinical **is a clinically-certified platform allowing fast and accurate variant discovery, annotation, and interpretation of NGS data for whole genomes, exomes, and gene panels. *VarSome Clinical* helps molecular geneticists and clinicians reach faster and more accurate diagnoses and treatment decisions for genetic conditions.**



1 Upload FASTQ or VCF

VarSome Clinical accepts FASTQ and VCF files. You can upload the data easily and securely through its web interface or you can harness *VarSome*'s powerful API for an automated and secure data transfer. Once the data are uploaded, you can start the analysis!

JOINT CALLING | MULTI - SAMPLE ANALYSIS | QUALITY CONTROL REPORTS

2 Annotation & Classification

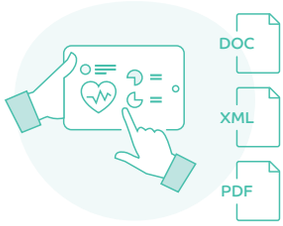
VarSome Clinical's robust pipeline is capable of analyzing whole genomes, exomes, and gene panels, for individual samples, trios, families, and cohorts in minutes. *VarSome Clinical* leverages the massive cross-referenced knowledge base of the free VarSome and offers also access to licensed databases.

OVER 30 DATA RESOURCES | ACMG CLASSIFICATION | PROPRIETARY DATA RESOURCES

3 Intuitive Web Portal

VarSome Clinical's feature-rich and intuitive web interface allows filtering variants according to pathogenicity, ACMG classification, allele frequency, gene list or phenotype, to name a few! Dynamic and algorithmic filters allow you to perform simple or advanced filtering, like segregation analysis, identification of de novo variants or variants in imprinted genes, and much more!

DYNAMIC FILTERS | ALGORITHMIC FILTERS | SAMPLE CROSS-REFERENCING



4 Clinical Report Generation

Once you have narrowed down the list of variants of interest, you can proceed with the generation of the clinical report, which includes all the details of your variants, including literature references and your custom comments. The layout of the report can be fully customized according to your unique branding policy.

CUSTOMIZABLE REPORTS | CE-IVD CERTIFICATION | AUDIT TRAIL

VarSome is brought to you by:
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Key Benefits

End-to-End Solution

VarSome Clinical provides validated and clinically certified genome-scale variant discovery, annotation and classification

Variant Knowledge Base

Cross-referenced data from 30+ genomic databases plus contributions from a 100'000-strong community.

Programming Interface

VarSome Clinical comes with a powerful API for automated data transfer in both directions: upload data and download annotated variants.

CE-IVD Mark

VarSome Clinical is certified as an In-vitro Diagnostic Medical Device according to the requirements of EC 93/42/EEC.

ISO 13485

We are certified as a company with ISO 13485 quality management system for medical devices.

Variant Sharing

Optionaly share selected information on variants with your partner institutions.

Did you know?

Precision Contests

VarSome Clinical's variant discovery rocks! It received top marks in the recent contest organized by the Food and Drug Administration, and it was steadily in the top ranks in all metrics, as in previous precision FDA contests. These results are a testament to the *VarSome* team's total commitment to excellence.

Variant Discovery

VarSome Clinical's variant discovery pipelines have been designed to achieve high quality standards, such as reproducibility, sensitivity and precision:

- Sensitivity: 99.8% for SNVs and 99.5% for indels
- Precision: 99.8% for SNVs and indels

Experts in Big Data

To date *VarSome* has aggregated and cross-referenced over 30 leading databases and other data resources, representing over 33 billion data points, and new ones are added constantly over time. But there is more to it: whenever a data resource is updated, *VarSome* quickly makes it available for annotation and classification of your variants!

Network of Geneticists

As *VarSome Clinical* has been deployed by dozens of institutional customers across many countries, you can benefit from *VarSome's* extensive network of molecular geneticists and health care professionals who contribute to the identification of likely causal variants, along with a summary of available therapeutic options.

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