varsome

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VARSOME IS FREE!

The Human Genomics Community

***VarSome* is a community-driven project aiming at sharing global expertise on human variants. It is FREE and features variant search engine and aggregated knowledge base consisting of more than 30 cross-referenced public data resources and contributions from its community of more than 100’000 users worldwide.**

100'000-strong Community

VarSome is a community-driven project aiming at sharing global expertise on human genomic variants. It features a variant search engine and aggregated knowledge base consisting of more than 30 cross-referenced public data resources, as well as real-time functional annotation of any variant.

Its world-wide community of more than 100'000 healthcare and life sciences professionals, who classify, link research findings and publications, and share evidence on variants, creates a unique resource in the genomics landscape. *VarSome* also facilitates introductions between users who wish to exchange notes on search results, without compromising its strict privacy policy. of both parties.

30+ Genomic Databases

VarSome offers a massive cross-referenced knowledge base consisting of 30+ public genomic databases, representing over 33 billion data points. But there is more to it: whenever a public database is updated, *VarSome* processes it and makes it available to the whole genomics community for annotation and classification!

Integrated resources:
ClinVar, dbSNP, gnomAD, HPO, MONDO, Ensembl, RefSeq, GWAS, CGD, HGNC, UniGene, Orphanet, CIViC genes, GERP, dbNSFP, COSMIC, IARC TP53, ICGC, Kaviar, DANN scores, CIViC mutations, UniProt variants, UniProt domains, GHR, CPIC, DGV, DECIPHER, ExAC CNVs, ExAC genes, PanelApp, Mondo, PMKB.

ACMG Classification

VarSome displays automated variant classification according to the guidelines of the American College of Medical Genetics and Genomics (Richards et al. 2015). Each ACMG rule is explained, along with why it has been triggered, or why not. If you have additional evidence, you can manually turn on other ACMG rules and easily reach the final verdict for your variant.

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Versatile Variant Search

You can search *VarSome* by HGVS nomenclature, rsID, gene name, transcript symbol or genomic location. *VarSome* can also parse single lines from VCF files to look up the variant they describe. The results are not limited to known variants, you can query any possible variant.

Full-text Search

VarSome full-text search functions like other search engines with one important difference: the search query returns only entries from the *VarSome* aggregated knowledge base, thus showing you only the results your are looking for. It enables you to perform targeted searches not just for variants, but over the entire contents of *VarSome*, such as articles, diseases, phenotypes, genes, etc. Importantly, this includes content provided by the entire *VarSome* user global community.

Application Programming Interface (API)

VarSome provides a powerful API which allows you to integrate its knowledge base in your own software at a fraction of the cost that would take to integrate data from such a multitude of sources and perform real-time functional annotation.

Cite VarSome!

If you use VarSome for your work please cite it in your articles and all other communications.

VarSome: The Human Genomic Variant Search Engine. Christos Kopanos, Vasilis Tsiolkas, Alexandros Kouris, Charles E Chapple, Monica Albarca Aguilera, Richard Meyer, and Andreas Massouras. Oxford Bioinformatics, bty897, 30 October 2018.

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