Resource Summary Report

Generated by NIF on Aug 3, 2026

VarSome

RRID:SCR_026165
Type: Tool

Proper Citation

VarSome (RRID:SCR_026165)

Resource Information

URL: https://varsome.com/

Proper Citation: VarSome (RRID:SCR_026165)

Description: Web service to search for variants, CNVs, genes, transcripts, publications, diseases. Annotation tool and search engine for human genomic variants, and platform enabling sharing of knowledge on specific variants. Enables users to look up variants in their genomic context, collects data from multiple databases in central location and most importantly, aims to enable community to freely and easily share knowledge on human variation.

Resource Type: data access protocol, data or information resource, web service, software resource

Defining Citation: PMID:30376034

Keywords: Human genomic variant search engine, genomic variant, search engine, search, variants, CNVs, genes, transcripts, publications, diseases

Availability: Free, Freely available

Resource Name: VarSome

Resource ID: SCR_026165

Record Creation Time: 20241211T053244+0000

Record Last Update: 20260803T040926+0000

Ratings and Alerts

No rating or validation information has been found for VarSome .

No alerts have been found for VarSome .

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 79 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF .

Marinakis NM, et al. (2026) Unraveling the Role of WDR91: Case Report of a Previously Unrecognized Clinical Entity. Clinical genetics, 109(1), 176.

Del Prete D, et al. (2026) Dual-Genetic Etiology in an Atypical Dent Disease Phenotype Which Combines Features of Focal Segmental Glomerulosclerosis and Ellis-Van Creveld-Like Syndrome: A Case Report. Case reports in nephrology and dialysis, 16(1), 1.

Simonati A, et al. (2026) Age at onset and gene variants predict lifespan and disease duration in childhood neuronal ceroid lipofuscinoses. Developmental medicine and child neurology, 68(2), 276.

Ncir WB, et al. (2026) First LDLRAP1 and Recurrent LDLR Mutations in Tunisian Families With Familial Hypercholesterolemia. Journal of cellular and molecular medicine, 30(1), e70997.

Sánchez-Fuentes A, et al. (2025) Diagnosis of Inherited Platelet Disorders: Clinical Evaluation and Functional and Molecular Assays. Biomolecules, 15(6).

Mikhaylenko DS, et al. (2025) Patients with Papillary Renal Cancer and Germline Duplication of MET Exons 5-21. Biomedicines, 13(6).

Ammar M, et al. (2025) Identification of a novel truncated pathogenic variant in PUS1 gene in two siblings of consanguineous Tunisian family: intrafamilial phenotypic variability related to mtDNA copy number. Annals of hematology, 104(2), 943.

Sudhindar PD, et al. (2025) Urinary renal epithelial cells can be used for NPHP1 phenotyping and a personalized therapeutic strategy. Journal of cell science, 138(20).

Shi X, et al. (2025) Utility of whole exome sequencing in the evaluation of isolated fetal growth restriction in normal chromosomal microarray analysis. Annals of medicine, 57(1), 2476038.

Li J, et al. (2025) The CFTR K464N variant in fetuses potential increases premature birth risk in Chinese families. Human genomics, 19(1), 25.

Lara Gutierrez A, et al. (2025) Robust Assessment of Homologous Recombination Deficiency Genomic Instability by OncoScan Microarrays. The Journal of molecular diagnostics : JMD, 27(6), 475.

Du W, et al. (2025) Case Report: Identification of a novel mutation, c.1067T > A, in the SERPING1 gene in a Chinese male with type 1 hereditary angioedema. Frontiers in

allergy, 6, 1554940.

Vasuri F, et al. (2025) Beyond histology: A tissue algorithm predictive of post-surgical recurrence in hepatocellular carcinomas, including TERT promoter mutation. Virchows Archiv : an international journal of pathology, 486(2), 365.

Zhou Q, et al. (2025) Molecular and Clinical Profiles of Pediatric Monogenic Diabetes Subtypes: Comprehensive Genetic Analysis of 138 Patients. The Journal of clinical endocrinology and metabolism, 110(8), 2314.

Vanoli A, et al. (2025) Adenomyoma/adenomyomatosis-associated mural intracholecystic neoplasms: analysis of clinico-pathologic, imaging, and molecular features of a consecutive case series. Virchows Archiv : an international journal of pathology, 487(1), 153.

Vasuri F, et al. (2025) Morpho-molecular approach (NGS plus digital PCR) in diagnosis of malignant biliary strictures. Pathologica, 117(1), 10.

Falsaperla R, et al. (2025) PPP5C pathogenic variant identified: a potential key to gaining insight into developmental and epileptic encephalopathy? Molecular and cellular pediatrics, 12(1), 3.

Redaelli V, et al. (2025) A novel SORL1 mutation in a pedigree affected by early-onset Alzheimer's disease. Journal of Alzheimer's disease reports, 9, 25424823241296017.

Martín-Bejarano P, et al. (2025) Functional characterization of BRCA1 variants of unknown significance using homologous recombination repair assays. Breast cancer research : BCR, 27(1), 174.

De Marchis L, et al. (2025) BRCA Screening and Identification of a Common Haplotype in the Jewish Community of Rome Reveal a Founder Effect for the c.7007G>C, p.(Arg2336Pro) BRCA2 Variant. Cancers, 17(12).