Resource Summary Report

Generated by RRID on Aug 15, 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, software resource, data or information resource, web service

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: 20260815T042923+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 335 mentions in open access literature.

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

Alimoradi E, et al. (2026) Identification of a Novel Missense Homozygous Variant in LINS1 in Two Distinct Iranian Families With Consanguineous Marriage. Molecular genetics & genomic medicine, 14(2), e70184.

Vettiatil D, et al. (2026) Ultrarare Variants in DNA Damage Repair and Mitochondrial Genes in Pediatric Acute-Onset Neuropsychiatric Syndrome and Acute Behavioral Regression in Neurodevelopmental Disorders. Developmental neuroscience, 1.

Wang Y, et al. (2026) Clinical and genetic basis of congenital gonadotropin deficiency. Human reproduction open, 2026(2), hoag017.

Jiang F, et al. (2026) Functional Screening of NDUFAF6 Variants in Knockout Cells and Complementary Computational Analysis. Journal of clinical laboratory analysis, 40(1), e70147.

Amaral RAS, et al. (2026) Expanding the clinical and genetic spectrum of RHO-associated retinitis pigmentosa. Experimental biology and medicine (Maywood, N.J.), 251, 10893.

Koparir A, et al. (2026) Clinical and genetic heterogeneity of syndromic hearing loss and its non-syndromic hearing loss mimics. Molecular medicine (Cambridge, Mass.), 32(1).

Zhang PQ, et al. (2026) Biological Context-Informed and Population-Stratified Strategies Improve Genetic Diagnosis of CCDC22-Related Disorder. Genetics research, 2026(1), e8137770.

Mancuso G, et al. (2026) Whole-Exome Sequencing Identifies Novel Genetic Variants Associated with Unexplained Neurodevelopmental Disorders in Children. International journal of molecular sciences, 27(2).

Smaili F, et al. (2026) DYSF gene variant spectrum in Arab populations across eight countries: A systematic review. Biomolecules & biomedicine, 26(9), 1511.

Ferrer M, et al. (2026) Clinical and molecular characterization of a novel pathogenic AIFM1 E336K mutation connecting mitochondrial dysfunction and neurodegeneration. Cell communication and signaling : CCS, 24(1).

Semenova V, et al. (2026) Genetic and Epigenetic Drivers of Wilms Tumor Predisposition in Russian Pediatric Patients: A Multicenter Study. International journal of molecular sciences, 27(9).

Lai M, et al. (2026) A novel ANK1 gene mutation associated with hereditary spherocytosis: a case report. Frontiers in pediatrics, 14, 1760131.

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

Balestrini S, et al. (2026) Clinical and genetic landscape of epilepsies with absence seizures and single-gene etiology. Epilepsia, 67(1), 272.

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.

Musante L, et al. (2026) A novel spliceosomopathy caused by de novo SF3B3 variants. Genome medicine, 18(1).

Moushib LI, et al. (2026) Exploring the strengths and limitations of AI-driven variant prioritization versus manual curation in inborn errors of immunity. Frontiers in genetics, 17, 1713299.

Khurana S, et al. (2026) Clinical and genetic determinants of survival in amyotrophic lateral sclerosis patients from North India. Brain communications, 8(1), fcag003.

Janková N, et al. (2026) Identification and functional assessment of a KCNH2 compound heterozygosity in a patient with presumed idiopathic ventricular fibrillation ascertains the diagnosis of long QT syndrome type 2. Europace : European pacing, arrhythmias, and cardiac electrophysiology : journal of the working groups on cardiac pacing, arrhythmias, and cardiac cellular electrophysiology of the European Society of Cardiology, 28(2).

Marsza?ek-Kruk BA, et al. (2026) Congenital Temporomandibular Joint Ankylosis: Investigating Potential Genetic Etiologies with Whole Exome Sequencing. Journal of clinical medicine, 15(4).