Resource Summary Report

Generated by ASWG on Aug 3, 2026

Human Genome Variation Society: Databases and Other Tools

RRID:SCR_006876
Type: Tool

Proper Citation

Human Genome Variation Society: Databases and Other Tools (RRID:SCR_006876)

Resource Information

URL: http://www.hgvs.org/dblist/dblist.html

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Description: A list of various databases freely available to the public, including several mutation and variation resources, such as education resources for teachers students provided by the Human Genome Variation Society. Databases listed include: * Locus Specific Mutation Databases * Disease Centered Central Mutation Databases * Central Mutation and SNP Databases * National and Ethnic Mutation Databases * Mitochondrial Mutation Databases * Chromosomal Variation Databases * Other Mutation Databases (i.e. your round holes don""t fit our square pegs) * Clinical and Patient Aspects Databases * Non Human Mutation Databases * Artificial Mutations Only * Other Related Databases * Education Resources for Teachers and Students

Abbreviations: HGVS Databases & Other Tools

Synonyms: HGVS: Databases and Other Tools

Resource Type: data set, data or information resource

Keywords: genome, artificial, chromosome, clinical, disease, human, mitochondrial, non human, snp, mutation, genetic variation, education, ethnic

Availability: Public

Resource Name: Human Genome Variation Society: Databases and Other Tools

Resource ID: SCR_006876

Alternate IDs: nif-0000-02959

Record Creation Time: 20220129T080238+0000

Record Last Update: 20260801T191132+0000

Ratings and Alerts

No rating or validation information has been found for Human Genome Variation Society: Databases and Other Tools.

No alerts have been found for Human Genome Variation Society: Databases and Other Tools.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 13 mentions in open access literature.

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

Wang P, et al. (2025) Functional characterization of SOX5 variant causing Lamb-Shaffer syndrome and literature review of variants in the SOX5 gene. Orphanet journal of rare diseases, 20(1), 300.

Zhao Q, et al. (2023) Compound heterozygous splicing variants in KIAA0586 cause fetal short-rib thoracic dysplasia and cerebellar malformation: Use of exome sequencing in prenatal diagnosis. Molecular genetics & genomic medicine, 11(3), e2124.

He T, et al. (2023) A novel splicing variation in L1CAM is responsible for recurrent fetal hydrocephalus. Molecular genetics & genomic medicine, 11(11), e2253.

Wang P, et al. (2022) L1 Syndrome Prenatal Diagnosis Supplemented by Functional Analysis of One L1CAM Gene Missense Variant. Reproductive sciences (Thousand Oaks, Calif.), 29(3), 768.

Erdo?an M, et al. (2021) The Genetic Analysis of Cystic Fibrosis Patients With Seven Novel Mutations in the CFTR Gene in the Central Anatolian Region of Turkey. Balkan medical journal, 38(6), 357.

Yang M, et al. (2021) Genetic diagnoses in pediatric patients with epilepsy and comorbid intellectual disability. Epilepsy research, 170, 106552.

Yang M, et al. (2021) Identification of a novel EXT2 frameshift mutation in a family with hereditary multiple exostoses by whole-exome sequencing. Journal of clinical laboratory analysis, 35(9), e23968.

Kondratyeva E, et al. (2020) Clinical and genetic characterization of patients with cystic fibrosis and functional assessment of the chloride channel with the pathogenic variant

c.831G>A (p.Trp277*), described for the first time. Gene, 761, 145023.

Sánchez-Chaparro MM, et al. (2020) Genetic Variants in the 3'UTR of BRCA1 and BRCA2 Genes and their Putative Effects on the microRNA Mechanism in Hereditary Breast and Ovarian Cancer. Diagnostics (Basel, Switzerland), 10(5).

Astuti D, et al. (2017) Monogenic diabetes syndromes: Locus-specific databases for Alström, Wolfram, and Thiamine-responsive megaloblastic anemia. Human mutation, 38(7), 764.

Richards S, et al. (2015) Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in medicine : official journal of the American College of Medical Genetics, 17(5), 405.

Gong S, et al. (2014) NECTAR: a database of codon-centric missense variant annotations. Nucleic acids research, 42(Database issue), D1013.

Sheykholeslami K, et al. (2013) A new mutation of the Atoh1 gene in mice with normal life span allows analysis of inner ear and cerebellar phenotype in aging. PloS one, 8(11), e79791.