Resource Summary Report

Generated by RRID on Aug 5, 2026

Human Gene Mutation Database

RRID:SCR_001621
Type: Tool

Proper Citation

Human Gene Mutation Database (RRID:SCR_001621)

Resource Information

URL: https://www.hgmd.cf.ac.uk/ac/introduction.php?lang=english

Proper Citation: Human Gene Mutation Database (RRID:SCR_001621)

Description: Curated database of known (published) gene lesions responsible for human inherited disease.

Abbreviations: HGMD

Synonyms: The Human Gene Mutation Database, The Human Gene Mutation Database at the Institute of Medical Genetics in Cardiff

Resource Type: database, data or information resource

Defining Citation: PMID:22948725, PMID:20368137, PMID:20038494, PMID:19348700, PMID:18428754, PMID:18245393, PMID:12754702, PMID:10612821, PMID:9399854, PMID:9066272, PMID:8882888

Keywords: gene, disease, gene lesion, mutation, deletion, insertion, duplication, rearrangement, nuclear gene, functional polymorphism, bio.tools

Related Condition: Inherited disease

Availability: Free, Freely available

Resource Name: Human Gene Mutation Database

Resource ID: SCR_001621

Alternate IDs: nlx_153887, SCR_001888, biotools:hgmd, nif-0000-10459, OMICS_00281

Alternate URLs: http://www.hgmd.cf.ac.uk/ac/index.php, https://bio.tools/hgmd,

Record Creation Time: 20220129T080208+0000

Record Last Update: 20260805T044025+0000

Ratings and Alerts

No rating or validation information has been found for Human Gene Mutation Database.

No alerts have been found for Human Gene Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 2462 mentions in open access literature.

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

Qu X, et al. (2026) Investigation of epilepsy-related genes in a Drosophila model. Neural regeneration research, 21(1), 195.

Di Pierro E, et al. (2026) New cases of ?-aminolevulinic acid dehydratase deficiency: Functional insights into gene variants using an innovative mouse liver model. Journal of internal medicine, 299(1), 126.

Treger TD, et al. (2025) Predisposition Footprints in the Somatic Genome of Wilms Tumors. Cancer discovery, 15(2), 286.

Karali M, et al. (2025) Variants in the AGBL5 gene are responsible for autosomal recessive Retinitis pigmentosa with hearing loss. European journal of human genetics : EJHG, 33(6), 727.

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.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Kim JA, et al. (2025) Systematic genetic assessment of hearing loss using whole-genome sequencing identifies pathogenic variants. Experimental & molecular medicine, 57(4), 775.

Srivastava R, et al. (2025) Advancing precision oncology with AI-powered genomic analysis. Frontiers in pharmacology, 16, 1591696.

Mighell TL, et al. (2025) A small molecule stabilizer rescues the surface expression of nearly all missense variants in a GPCR. Nature structural & molecular biology, 32(12), 2429.

Ullah N, et al. (2025) Identification of missense DMC1 variants in males with non-obstructive azoospermia. Journal of assisted reproduction and genetics, 42(11), 3723.

Li J, et al. (2025) Osimertinib plus chemotherapy versus osimertinib for patients with advanced NSCLC with concomitant EGFR and TP53 mutations: a prospective cohort study. Scientific reports, 15(1), 20952.

Fang W, et al. (2025) Genetic analysis of a pedigree with hereditary coagulation factor XII deficiency. Annals of hematology, 104(2), 1281.

Buianova AA, et al. (2025) Clinical and Allelic Heterogeneity in a Small Cohort of Patients with Inherited Epidermolysis Bullosa. International journal of molecular sciences, 26(12).

Assing K, et al. (2025) Functional Characterization of an IL2RG Variant, a Case Report of X-Linked T- B + NK + SCID. Immunity, inflammation and disease, 13(12), e70307.

Shan S, et al. (2025) Animal models of methylmalonic acidemia: insights and challenges. Orphanet journal of rare diseases, 20(1), 583.

Tachida Y, et al. (2025) Systematic empirical evaluation of individual base editing targets: Validating therapeutic targets in USH2A and comparison of methods. Molecular therapy : the journal of the American Society of Gene Therapy, 33(4), 1466.

Wang Y, et al. (2025) A Novel Variant in Dentin Sialophosphoprotein (DSPP) Gene Causes Dentinogenesis Imperfecta Type III: Case Report. Molecular genetics & genomic medicine, 13(3), e70087.

Kouri C, et al. (2025) Oligogenic analysis across broad phenotypes of 46,XY differences in sex development associated with NR5A1/SF-1 variants: findings from the international SF1next study. EBioMedicine, 113, 105624.

Nadeali Z, et al. (2025) Investigating TSHR gene variants in consanguineous families: novel insights into variable expression in familial congenital hypothyroidism. Frontiers in endocrinology, 16, 1559281.

Lee SJ, et al. (2025) Gene signatures and genotype-phenotype correlations of sensorineural hearing loss in Noonan syndrome and related RASopathies. Scientific reports, 15(1), 12102.