# Resource Summary Report

Generated by T1D on Sep 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:** data or information resource, database

**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:** 20260904T000042+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 2682 mentions in open access literature.

**Listed below are recent publications.** The full list is available at T1D .


Yadegari H, et al. (2026) Landscape and Spectrum of VWF Variants in Type 2 Von Willebrand Disease: Insights from a German Patient Cohort. Thrombosis and haemostasis, 126(2), 156.

Kihara M, et al. (2026) Medullary thyroid carcinoma exclusively associated with a homozygous RET V778I pathogenic variant: a case report with review of literature. Endocrine journal, 73(1), 109.

Deng X, et al. (2026) Genetic analysis of LRRK2 variants in Han Chinese patients with Parkinson's disease. PloS one, 21(1), e0340448.

Lyu P, et al. (2026) Database of CLCN5 Pathogenic Variants Causing Dent Disease. Kidney international reports, 11(5), 106475.

Geviti A, et al. (2026) Cumulative Incidence in Monogenic Alzheimer's Disease and Frontotemporal Dementia: Gene-Gene Interaction Effect. International journal of molecular sciences, 27(9).

Forte G, et al. (2026) Deciphering the Causative Role of a Novel APC Gene Variant in Attenuated Familial Adenomatous Polyposis Using Germline DNA-RNA Paired Testing. Biomedicines, 14(1).

Song S, et al. (2026) Disordered but Different: Functional and Evolutionary Divergence of Transcription Factor Intrinsically Disordered Regions. bioRxiv : the preprint server for biology.

Huang WH, et al. (2026) Identification of gene variants in 30 patients from southeastern China with severe hypospadias by whole-exome sequencing. Asian journal of andrology, 28(3), 276.

Varughese S, et al. (2026) Typical and atypical ADPKD: predicted pathogenic genetic variants and population frequencies. Nephrology, dialysis, transplantation : official

publication of the European Dialysis and Transplant Association - European Renal Association, 41(2), 286.

Dev P, et al. (2026) Short report: Targeted analysis of whole exome sequencing data in Indian cryptogenic stroke patients. PloS one, 21(2), e0326554.

Li Y, et al. (2026) Clinical Characteristics and Gene Mutations of Hereditary Spherocytosis in 59 Chinese Children. Molecular genetics & genomic medicine, 14(3), e70188.

Ye Y, et al. (2026) CNTNAP2: isoform- and context-specific functions in neurological disorders and cancer. Frontiers in cellular neuroscience, 20, 1803486.

Nelson N, et al. (2026) The Performance of In Silico Prediction Tools for Variant Curation in a Panel of Cancer Genes. Human mutation, 2026, 8817550.

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

Su J, et al. (2026) Identification of a new frameshift homozygous variant of PEX3 gene in a preterm infant with profound global developmental delay and bilateral ptosis: a case report and updated literature review. BMC pediatrics, 26(1), 81.

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.

Giannopoulou EZ, et al. (2026) The conundrum in diagnosing Maturity-Onset Diabetes of the Young (MODY) in a large German pedigree with early-onset diabetes and a novel HNF1A variant. Molecular and cellular pediatrics, 13(1).

Li YF, et al. (2026) Prenatal Diagnosis of Autosomal Recessive Primary Microcephaly Type 2 Caused by Compound Heterozygous WDR62 Variants in a Family With Two Recurrent Cases. Molecular genetics & genomic medicine, 14(4), e70203.

Pshenichnikova O, et al. (2026) Genetic and Clinical Characteristics of Russian Patients with Congenital Factor V Deficiency. International journal of molecular sciences, 27(8).

Shi T, et al. (2026) Functional reassessment of extended splice region variants in MYO7A with hearing loss and Usher syndrome. The Journal of pathology, 269(2), 222.