Resource Summary Report

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Human Gene Mutation Database

RRID:SCR_001621
Type: Tool

Proper Citation

Human Gene Mutation Database (RRID:SCR_001621)

Resource Information

URL: http://www.hgmd.org

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

Funding:

Availability: Free for academic use, Free for non-profit institutional use, Account required, Commercial users required to purchase license

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: 20250426T055451+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 2388 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

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

Justice A, et al. (2025) Phenotypic findings associated with variation in elastin. HGG advances, 6(1), 100388.

Turvey AK, et al. (2025) Human disease-causing missense genetic variants are enriched in the evolutionarily ancient domains of the cytosolic aminoacyl-tRNA synthetase proteins. IUBMB life, 77(1), e2932.

Shu Y, et al. (2025) Dias-Logan syndrome with a de novo p.Leu360Profs*212 heterozygous pathogenic variant of BCL11A in a Chinese patient: A case report. SAGE open medical case reports, 13, 2050313X251314069.

Ishida Y, et al. (2025) A Novel Pathogenic CDC73 Gene Variant in Hyperparathyroidism-jaw Tumor Syndrome. JCEM case reports, 3(2), luaf016.

Minniakhmetov IR, et al. (2025) Genetic Structure of Hereditary Forms of Diabetes Mellitus in Russia. International journal of molecular sciences, 26(2).

Cui LM, et al. (2025) Analysis of a Series of 26 Cases With Prenatal Skeletal Dysplasia via Multiplatform Genetic Detection. Molecular genetics & genomic medicine, 13(1), e70062.

Bandres-Ciga S, et al. (2024) NeuroBooster Array: A Genome-Wide Genotyping Platform to Study Neurological Disorders Across Diverse Populations. Movement disorders : official journal of the Movement Disorder Society.

Nawaz H, et al. (2024) Brachyolmia, dental anomalies and short stature (DASS): Phenotype and genotype analyses of Egyptian and Pakistani patients. Heliyon, 10(1), e23688.

Kim SW, et al. (2024) Re-evaluation of a Fibrillin-1 Gene Variant of Uncertain Significance Using the ClinGen Guidelines. Annals of laboratory medicine, 44(3), 271.

Wei X, et al. (2024) Complex genotype-phenotype correlation of MYH11: new insights from monozygotic twins with highly variable expressivity and outcomes. BMC medical genomics, 17(1), 135.

Shang Q, et al. (2024) Clinical and genetic characteristics of Chinese patients diagnosed with chronic enteropathy associated with SLCO2A1 gene. Orphanet journal of rare diseases, 19(1), 201.

Johnson MB, et al. (2024) Human inherited PD-L1 deficiency is clinically and immunologically less severe than PD-1 deficiency. The Journal of experimental medicine, 221(6).

Fernández-Cancio M, et al. (2024) Clinical and molecular study of patients with thyroid dyshormogenesis and variants in the thyroglobulin gene. Frontiers in endocrinology, 15, 1367808.

Suzuki Y, et al. (2024) Familial and early recurrent pheochromocytoma in a child with a novel in-frame duplication variant of VHL. Clinical pediatric endocrinology : case reports and clinical investigations : official journal of the Japanese Society for Pediatric Endocrinology, 33(4), 229.

Khani M, et al. (2024) Biobank-scale characterization of Alzheimer's disease and related dementias identifies potential disease-causing variants, risk factors, and genetic modifiers across diverse ancestries. medRxiv : the preprint server for health sciences.

Sidoti A, et al. (2024) Exploring Trimethylaminuria: Genetics and Molecular Mechanisms, Epidemiology, and Emerging Therapeutic Strategies. Biology, 13(12).

Huang J, et al. (2024) Associations between genomic aberrations, increased nuchal translucency, and pregnancy outcomes: a comprehensive analysis of 2,272 singleton pregnancies in women under 35. Frontiers in medicine, 11, 1376319.

Chen WQ, et al. (2024) Identification of novel variations in three cases with rare inherited neuromuscular disorder. Experimental and therapeutic medicine, 27(6), 270.

Günbey C, et al. (2024) Horizontal gaze palsy with progressive scoliosis: Further expanding

the ROBO3 spectrum. Annals of clinical and translational neurology, 11(8), 2088.