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

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,

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 2309 mentions in open access literature.

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

Bagger FO, et al. (2024) Whole genome sequencing in clinical practice. BMC medical genomics, 17(1), 39.

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.

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.

Xue H, et al. (2024) Genetic testing for fetal loss of heterozygosity using single nucleotide polymorphism array and whole-exome sequencing. Scientific reports, 14(1), 2190.

Deng X, et al. (2024) Novel heterozygous mutations of TNFRSF13B in EBV-associated T/NK lymphoproliferative diseases (EBV-T/NK-LPDs). Blood science (Baltimore, Md.), 6(1), e00180.

Köro?lu M, et al. (2024) Cerebrotendinous Xanthomatosis patients with late diagnosed in single orthopedic clinic: two novel variants in the CYP27A1 gene. Orphanet journal of rare diseases, 19(1), 53.

Mao YX, et al. (2024) Transport mechanism of human bilirubin transporter ABCC2 tuned by the inter-module regulatory domain. Nature communications, 15(1), 1061.

Eriksson A, et al. (2023) Somatic Exonic Deletions in RUNX1 Constitutes a Novel Recurrent Genomic Abnormality in Acute Myeloid Leukemia. Clinical cancer research : an official journal of the American Association for Cancer Research, 29(15), 2826.

Diaz-Lombana N, et al. (2023) Case report: Novel frameshift mutation in LAMA2 gene causing congenital muscular dystrophy type 1A. Frontiers in genetics, 14, 1158350.

Bandres-Ciga S, et al. (2023) NeuroBooster Array: A Genome-Wide Genotyping Platform to Study Neurological Disorders Across Diverse Populations. medRxiv : the preprint server for health sciences.

Hou JW, et al. (2023) Loss-of-function CFTR p.G970D missense mutation might cause congenital bilateral absence of the vas deferens and be associated with impaired spermatogenesis. Asian journal of andrology, 25(1), 58.

Zhou X, et al. (2023) Novel biallelic mutations in TMEM126B cause splicing defects and lead to Leigh-like syndrome with severe complex I deficiency. Journal of human genetics, 68(4), 239.

Xu X, et al. (2023) The Pathology of Primary Familial Brain Calcification: Implications for Treatment. Neuroscience bulletin, 39(4), 659.

Li S, et al. (2023) Compound heterozygous loss-of-function variants in BRAT1 cause lethal neonatal rigidity and multifocal seizure syndrome. Molecular genetics & genomic medicine, 11(1), e2092.

Yen CC, et al. (2023) Clinical Characteristics and Responses to Immune Checkpoint Inhibitors in RET-Aberrant Digestive Tract Tumours. Targeted oncology, 18(4), 611.

Qiao F, et al. (2023) A female of progressive familial intrahepatic cholestasis type 3 caused by heterozygous mutations of ABCB4 gene and her cirrhosis improved after treatment of ursodeoxycholic acid: a case report. BMC medical genomics, 16(1), 171.

Wang XC, et al. (2023) A novel missense mutation in SPAST causes hereditary spastic paraplegia in male members of a family: A case report. Molecular medicine reports, 27(4).

Chen YC, et al. (2023) TOMM40 Genetic Variants Cause Neuroinflammation in Alzheimer's Disease. International journal of molecular sciences, 24(4).

Ivanoshchuk D, et al. (2023) The Mutation Spectrum of Rare Variants in the Gene of Adenosine Triphosphate (ATP)-Binding Cassette Subfamily C Member 8 in Patients with a MODY Phenotype in Western Siberia. Journal of personalized medicine, 13(2).

Deng L, et al. (2023) Shared molecular signatures between coronavirus infection and neurodegenerative diseases provide targets for broad-spectrum drug development. Scientific reports, 13(1), 5457.