Resource Summary Report

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HGMD

RRID:SCR_001888
Type: Tool

Proper Citation

HGMD (RRID:SCR_001888)

Resource Information

URL: http://www.hgmd.cf.ac.uk/ac/index.php

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Description: It represents an attempt to collate known (published) gene lesions responsible for human inherited disease. More specifically, this database is a comprehensive core collection of germline mutations in nuclear genes that underlie or are associated with human inherited disease. This database, whilst originally established for the study of mutational mechanisms in human genes (Cooper and Krawczak 1993), has now acquired a much broader utility in that it embodies an up-to-date and comprehensive reference source to the spectrum of inherited human gene lesions. Thus, HGMD provides information of practical diagnostic importance to (i) researchers and diagnosticians in human molecular genetics, (ii) physicians interested in a particular inherited condition in a given patient or family, and (iii) genetic counselors. The Human Gene Mutation Database comprises various types of mutation within the coding regions, splicing and regulatory regions of human nuclear genes causing inherited disease. Somatic mutations and mutations in the mitochondrial genome are thus not included, although in the latter case, links to Mitomap are now provided. Each mutation is entered only once in order to avoid confusion between recurrent and identical-by-descent lesions. Mutations inferred from amino acid sequencing have been excluded since, in the absence of direct DNA analysis, some ambiguity may exist as to the DNA sequence changes involved. Silent mutations within the coding region which do not alter the encoded amino acid are also not recorded. If such mutations are known to adversely affect mRNA splicing or gene expression, or have been reported in significant association with disease, they may be included. Sponsors: This Database is supported by Celera, Macmillan, The Genome Database, DFG, BIOS Scientific Publishers, Research Genetics, ScotLab bioscience, MB Biochemicals, Phzer, Sun Life, GFH, Springer, SmithKline Beecham, Hybaid.

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:9399854

Keywords: encode, expression, gene, genetic, amino acid, coding region, disease, dna sequence, human, inherited disease, mitochondrial genome, molecular, mrna, mutation, nuclear gene, regulatory region, splicing

Resource Name: HGMD

Resource ID: SCR_001888

Record Creation Time: 20220129T080210+0000

Record Last Update: 20260903T234507+0000

Ratings and Alerts

No rating or validation information has been found for HGMD.

No alerts have been found for HGMD.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 845 mentions in open access literature.

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

Yoo S, et al. (2023) The first case of novel variants of the FSHR mutation causing primary amenorrhea in 2 siblings in Korea. Annals of pediatric endocrinology & metabolism, 28(1), 54.

Lenarduzzi S, et al. (2023) Whole-exome sequencing: Clinical characterization of pediatric and adult Italian patients affected by different forms of hereditary cardiovascular diseases. Molecular genetics & genomic medicine, 11(5), e2143.

Lu L, et al. (2023) Molecular pathogenesis of a novel Met394Thr variant causing hemophilia B. Molecular genetics & genomic medicine, 11(5), e2147.

Walker LC, et al. (2023) APPLICATION OF THE ACMG/AMP FRAMEWORK TO CAPTURE EVIDENCE RELEVANT TO PREDICTED AND OBSERVED IMPACT ON SPLICING: RECOMMENDATIONS FROM THE CLINGEN SVI SPLICING SUBGROUP. medRxiv : the preprint server for health sciences.

Yang K, et al. (2023) Assessment of a novel variation in DHODH gene causing Miller syndrome: The first report in Chinese population. Molecular genetics & genomic medicine,

11(7), e2186.

Br??nger T, et al. (2023) Conserved patterns across ion channels correlate with variant pathogenicity and clinical phenotypes. Brain : a journal of neurology, 146(3), 923.

Alizadehmohajer N, et al. (2023) Using In Silico Bioinformatics Algorithms for the Accurate Prediction of the Impact of Spike Protein Mutations on the Pathogenicity, Stability, and Functionality of the SARS-CoV-2 Virus and Analysis of Potential Therapeutic Targets. Biochemical genetics, 61(2), 778.

Ataei Z, et al. (2023) Novel in-frame duplication variant characterization in late infantile metachromatic leukodystrophy using whole-exome sequencing and molecular dynamics simulation. PloS one, 18(2), e0282304.

Roht L, et al. (2023) AXIN2-related oligodontia-colorectal cancer syndrome with cleft palate as a possible new feature. Molecular genetics & genomic medicine, 11(6), e2157.

Zanoni P, et al. (2023) The genetic landscape and clinical implication of pediatric Moyamoya angiopathy in an international cohort. European journal of human genetics : EJHG, 31(7), 784.

Udupa P, et al. (2023) Genome sequencing identifies a large non-coding region deletion of SNX10 causing autosomal recessive osteopetrosis. Journal of human genetics, 68(4), 287.

Fazelifar AF, et al. (2023) Identification of a novel pathogenic variant in KCNH2 in an Iranian family with long QT syndrome 2 by whole-exome sequencing. Journal of arrhythmia, 39(3), 430.

Sharo AG, et al. (2023) ClinVar and HGMD genomic variant classification accuracy has improved over time, as measured by implied disease burden. Genome medicine, 15(1), 51.

Wang S, et al. (2023) Clinical and genetic analysis of a case of late onset carbamoyl phosphate synthase I deficiency caused by CPS1 mutation and literature review. BMC medical genomics, 16(1), 145.

Ni M, et al. (2023) What is the appropriate genetic testing criteria for breast cancer in the Chinese population?-Analysis of genetic and clinical features from a single cancer center database. Cancer medicine, 12(12), 13019.

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.

Li W, et al. (2023) De Novo Mutations Contributes Approximately 7% of Pathogenicity in Inherited Eye Diseases. Investigative ophthalmology & visual science, 64(2), 5.

Sun KY, et al. (2023) A deep catalog of protein-coding variation in 985,830 individuals. bioRxiv : the preprint server for biology.