Resource Summary Report

Generated by NIF on Apr 18, 2025

OMIM

RRID:SCR_006437
Type: Tool

Proper Citation

OMIM (RRID:SCR_006437)

Resource Information

URL: http://omim.org

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Description: Online catalog of human genes and genetic disorders, for clinical features, phenotypes and genes. Collection of human genes and genetic phenotypes, focusing on relationship between phenotype and genotype. Referenced overviews in OMIM contain information on all known mendelian disorders and variety of related genes. It is updated daily, and entries contain copious links to other genetics resources.

Abbreviations: OMIM, MIM

Synonyms: Online Mendelian Inheritance in Man, OMIM - Online Mendelian Inheritance in Man, MIM, The Online Mendelian Inheritance in Man Morbid Map

Resource Type: database, catalog, data or information resource

Defining Citation: PMID:22477700, PMID:22470145, PMID:21472891, PMID:19728286, PMID:18842627, PMID:18428346, PMID:17642958, PMID:17357067, PMID:15608251, PMID:15360913, PMID:11752252, PMID:10845565, PMID:10612823, PMID:9805561, PMID:7937048, PMID:1867277

Keywords: gene, genetics, phenotype, genotype, genetic loci, mutation, clinical, trait, disorder, umls, ontology, gold standard, FASEB list

Related Condition: Genetic disorder, Mendelian disorder, Developmental disorder

Funding:

Availability: Restricted

Resource Name: OMIM

Resource ID: SCR_006437

Alternate IDs: nif-0000-03216, OMICS_00278

Alternate URLs: http://www.ncbi.nlm.nih.gov/sites/entrez?db=omim, http://www.ncbi.nlm.nih.gov/Omim/, http://purl.bioontology.org/ontology/OMIM

License URLs: http://omim.org/help/agreement http://omim.org/help/copyright

Record Creation Time: 20220129T080236+0000

Record Last Update: 20250418T055132+0000

Ratings and Alerts

No rating or validation information has been found for OMIM.

No alerts have been found for OMIM.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 4819 mentions in open access literature.

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

Mutai H, et al. (2025) Genetic landscape in undiagnosed patients with syndromic hearing loss revealed by whole exome sequencing and phenotype similarity search. Human genetics, 144(1), 93.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Yan K, et al. (2025) Using network pharmacology and molecular docking technology, proteomics and experiments were used to verify the effect of Yigu decoction (YGD) on the expression of key genes in osteoporotic mice. Annals of medicine, 57(1), 2449225.

MacLaren RE, et al. (2025) XOLARIS: A 24-Month, Prospective, Natural History Study of

201 Participants with Retinitis Pigmentosa GTPase Regulator-Associated X-Linked Retinitis Pigmentosa. Ophthalmology science, 5(1), 100595.

Zhang L, et al. (2025) The Therapeutic Mechanisms of Huayu Quban Capsule in Treating Acne Vulgaris Are Uncovered Through Network Pharmacology and Molecular Docking. Journal of cosmetic dermatology, 24(1), e16632.

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

Smith JR, et al. (2025) Standardized pipelines support and facilitate integration of diverse datasets at the Rat Genome Database. Database : the journal of biological databases and curation, 2025.

Scherer N, et al. (2025) Coupling metabolomics and exome sequencing reveals graded effects of rare damaging heterozygous variants on gene function and human traits. Nature genetics, 57(1), 193.

Spedicati B, et al. (2025) Scent of COVID-19: Whole-Genome Sequencing Analysis Reveals the Role of ACE2, IFI44, and NDUFAF4 in Long-Lasting Olfactory Dysfunction. Life (Basel, Switzerland), 15(1).

Zhang J, et al. (2025) Fracture-healing effects of Rhizoma Musae ethanolic extract: An integrated study using UHPLC-Q-Exactive-MS/MS, network pharmacology, and molecular docking. PloS one, 20(1), e0313743.

Hu L, et al. (2025) Network pharmacology combined with experimental verification for exploring the potential mechanism of phellodendrine against depression. Scientific reports, 15(1), 1958.

Li Z, et al. (2025) Effects of the Salvia miltiorrhiza, Ligustrum lucidum, and Taraxacum mongolicum ultra-fine powder formula on meat quality of aged layers by multi-omics. Poultry science, 104(2), 104783.

Benslama O, et al. (2025) Silymarin as a Therapeutic Agent for Hepatocellular Carcinoma: A Multi-Approach Computational Study. Metabolites, 15(1).

Zhang Y, et al. (2025) Exploring the Underlying Mechanism of Weiling Decoction Alleviates Cold-Dampness Diarrhea Based on Network Pharmacology, Transcriptomics, Molecular Docking and Experimental Validation. Pharmaceuticals (Basel, Switzerland), 18(1).

Halligan NLN, et al. (2025) Variants in the ?-globin locus are associated with pneumonia in African American children. HGG advances, 6(1), 100374.

McGivern B, et al. (2025) MGA-related syndrome: A proposed novel disorder. HGG advances, 6(1), 100387.

Zeng Y, et al. (2025) Prenatal genetic detection in foetus with gallbladder size anomalies: cohort study and systematic review of the literature. Annals of medicine, 57(1), 2440638.

Hu Y, et al. (2025) DisGeNet: a disease-centric interaction database among diseases and various associated genes. Database : the journal of biological databases and curation, 2025.

Wang D, et al. (2025) Ginkgo biloba extract mediates HT22 cell proliferation and migration after oxygen-glucose deprivation/reoxygenation via regulating RhoA-ROCK2 signalling pathway. Metabolic brain disease, 40(1), 91.

Bouzid A, et al. (2025) Whole exome sequencing identifies ABHD14A and MRNIP as novel candidate genes for developmental language disorder. Scientific reports, 15(1), 367.