

Resource Summary Report

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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: data or information resource, catalog, database

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

Availability: Restricted

Resource Name: OMIM

Resource ID: SCR_006437

Alternate IDs: nif-0000-03216, r3d100010416, 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>,
<https://doi.org/10.17616/R3188W>

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

Record Creation Time: 20220129T080236+0000

Record Last Update: 20260729T044135+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 5456 mentions in open access literature.

Listed below are recent publications. The full list is available at [nidm-terms](#) .

Ghaemi B, et al. (2026) Precision radiolabeled B-cell maturation nanobody for targeted PET imaging and radioligand therapy of disseminated multiple myeloma. *Theranostics*, 16(6), 2748.

Vinogradov-Talyah E, et al. (2026) Human clearance systems have a layered architecture across tissues and cell types that supports varied proteome compositions. *Autophagy*, 22(1), 145.

Fischer MC, et al. (2026) Splicing and frameshift variants in QSER1 may be involved in developmental phenotypes. *HGG advances*, 7(1), 100539.

Yu Z, et al. (2026) Multi-trait genome-wide analysis identified risk loci and candidate drugs for heart failure. *HGG advances*, 7(1), 100540.

Huang L, et al. (2026) New insight into the epidemiological trends of respiratory syncytial virus infection and the underlying anti-respiratory syncytial virus mechanisms of andrographolide: integrating Global Burden of Disease database, network pharmacological analysis, and in vitro experiments. *Microbiology spectrum*, 14(1), e0234125.

Oh KK, et al. (2026) A roadmap to unveil the mechanism(s) of natural indole-derived molecules against NAFLD-derived HCC via systems pharmacology. *Translational oncology*, 63, 102618.

- Liu J, et al. (2026) Dietary supplementation with *Eucommia ulmoides* improves production performance, egg quality, meat characteristics and physiological status of aging laying hens. *Poultry science*, 105(1), 106156.
- Doss RM, et al. (2026) Exon-skipping due to bi-allelic splice site mutations in the neurodevelopmental disease gene *LNPK*. *HGG advances*, 7(1), 100543.
- Drost M, et al. (2026) Routine RNA-based analysis of potential splicing variants facilitates genomic diagnostics and reveals limitations of in silico prediction tools. *HGG advances*, 7(1), 100521.
- Namuli KL, et al. (2026) Unbiased human genomic characterization of polyglutamine disorder genes to guide biological understanding and therapeutic strategies. *HGG advances*, 7(1), 100547.
- Yin Q, et al. (2026) Genetic landscape of morphine response in BXD recombinant inbred mice. *HGG advances*, 7(1), 100535.
- Cheng M, et al. (2026) Targeting p38 MAPK signaling pathway:?? Quercetin as a novel therapy for TMJ synovitis. *International journal of molecular medicine*, 57(1).
- Leban T, et al. (2026) Multiomics Data Synthesis of FAM83H in Amelogenesis Imperfecta. *International dental journal*, 76(1), 109293.
- Esposito DV, et al. (2026) A non-coding ABO regulatory variant associated with VWF levels, thrombosis risk, and COVID-19 severity is topologically linked to ADAMTS13 in endothelial cells. *HGG advances*, 7(1), 100550.
- Epting D, et al. (2026) *PATJ* deficiency leads to cystic kidney disease and related ciliopathies. *HGG advances*, 7(1), 100514.
- Xiang F, et al. (2026) Integrative multiomics analysis reveals the ameliorative effects of Xiasangju on metabolic dysfunction-associated steatohepatitis. *Chinese medicine*, 21(1), 3.
- Li L, et al. (2026) Hyperoside prevents osteoporosis by activating the PI3K/AKT signaling pathway to inhibit oxidative stress and promote osteogenesis. *Molecular medicine reports*, 33(2).
- Zhao XC, et al. (2026) Unexpectedly high levels of normally spliced transcripts from the pathogenic *SLC10A7* alleles in a recessive form of skeletal dysplasia. *HGG advances*, 7(1), 100545.
- Liu W, et al. (2026) A national biobank framework for rare diseases: Standardized infrastructure and cross-institutional collaboration accelerating translational innovation in China. *HGG advances*, 7(1), 100544.
- Watanabe T, et al. (2026) Whole exome sequencing in Japanese spinocerebellar ataxia identifies novel variants. *Journal of human genetics*, 71(1), 35.