

Resource Summary Report

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Genetics Home Reference

RRID:SCR_007681

Type: Tool

Proper Citation

Genetics Home Reference (RRID:SCR_007681)

Resource Information

URL: <http://ghr.nlm.nih.gov/>

Proper Citation: Genetics Home Reference (RRID:SCR_007681)

Description: Genetics Home Reference provides consumer-friendly information about the effects of genetic variations on human health. Genetics Home Reference contains condition summaries (describing major features of genetic conditions), gene summaries (describing normal function, chromosomal location, etc), and gene family summaries.

Synonyms: Genetics Home Reference

Resource Type: data or information resource, database

Keywords: gene summary, genetic condition, health, human genetics, FASEB list

Resource Name: Genetics Home Reference

Resource ID: SCR_007681

Alternate IDs: nif-0000-02889

Record Creation Time: 20220129T080243+0000

Record Last Update: 20260811T043314+0000

Ratings and Alerts

No rating or validation information has been found for Genetics Home Reference.

No alerts have been found for Genetics Home Reference.

Data and Source Information

Usage and Citation Metrics

We found 63 mentions in open access literature.

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

Hanami RN, et al. (2025) Semantic classification of Indonesian consumer health questions. *Journal of biomedical semantics*, 16(1), 13.

Zhao B, et al. (2025) Paternal mosaicism in ASXL3-related bainbridge-roopers syndrome: implications for genetic counseling and prenatal diagnosis. *Frontiers in pediatrics*, 13, 1655021.

Liu H, et al. (2025) A Comprehensive Analysis of Sex-Biased Gene Expression in the Aging Human Retina Through a Combination of Single-Cell and Bulk RNA Sequencing. *Investigative ophthalmology & visual science*, 66(1), 28.

Lian R, et al. (2024) Clinical cases series and pathogenesis of Lamb-Shaffer syndrome in China. *Orphanet journal of rare diseases*, 19(1), 281.

Halim-Fikri H, et al. (2024) Global Globin Network and adopting genomic variant database requirements for thalassemia. *Database : the journal of biological databases and curation*, 2024.

Zygmunt-Górska A, et al. (2024) Comparison of clinical characteristics of a pediatric cohort with combined pituitary hormone deficiency caused by mutation of the PROP1 gene or of other origins. *Hormones (Athens, Greece)*, 23(1), 69.

Swanson GM, et al. (2023) Phthalates impact on the epigenetic factors contributed specifically by the father at fertilization. *Epigenetics & chromatin*, 16(1), 3.

Lin ZB, et al. (2023) Expanded phenotypic spectrum of FOXL2 Variant c.672_701dup revealed by whole-exome sequencing in a rare blepharophimosis, ptosis, and epicanthus inversus syndrome family. *BMC ophthalmology*, 23(1), 446.

Mikec Š, et al. (2022) Syndromic male subfertility: A network view of genome-phenome associations. *Andrology*, 10(4), 720.

Tejada Moreno JA, et al. (2022) Mutations in SORL1 and MTHFDL1 possibly contribute to the development of Alzheimer's disease in a multigenerational Colombian Family. *PloS one*, 17(7), e0269955.

Owen N, et al. (2022) Identification of 4 novel human ocular coloboma genes ANK3, BMPR1B, PDGFRA, and CDH4 through evolutionary conserved vertebrate gene analysis. *Genetics in medicine : official journal of the American College of Medical Genetics*, 24(5), 1073.

Li Z, et al. (2021) Germline and somatic mutation profile in Cancer patients revealed by a medium-sized pan-Cancer panel. *Genomics*, 113(4), 1930.

Olga T, et al. (2021) Uterine smooth muscle tumour of uncertain malignant potential and in vitro fertilization treatment in an infertile patient. *SAGE open medical case reports*, 9, 2050313X211012516.

Wang J, et al. (2021) Disease Spectrum of Breast Cancer Susceptibility Genes. *Frontiers in oncology*, 11, 663419.

Dong Y, et al. (2021) First Chinese patient with mental retardation-40 due to a de novo CHAMP1 frameshift mutation: Case report and literature review. *Experimental and therapeutic medicine*, 22(2), 907.

Das AB, et al. (2021) Lung disease network reveals impact of comorbidity on SARS-CoV-2 infection and opportunities of drug repurposing. *BMC medical genomics*, 14(1), 226.

Wishart DS, et al. (2021) MarkerDB: an online database of molecular biomarkers. *Nucleic acids research*, 49(D1), D1259.

Sanyaolu A, et al. (2020) Current modalities of sickle cell disease management. *Blood science (Baltimore, Md.)*, 2(4), 109.

Acharya KS, et al. (2020) Epigenetic alterations in cytochrome P450 oxidoreductase (Por) in sperm of rats exposed to tetrahydrocannabinol (THC). *Scientific reports*, 10(1), 12251.

Shen N, et al. (2020) Importance of early detection of juvenile polyposis syndrome: A case report and literature review. *Medicine*, 99(50), e23494.