Resource Summary Report

Generated by ASWG on Aug 26, 2026

GeneReviews

RRID:SCR_006560
Type: Tool

Proper Citation

GeneReviews (RRID:SCR_006560)

Resource Information

URL: http://www.ncbi.nlm.nih.gov/books/NBK1116/

Proper Citation: GeneReviews (RRID:SCR_006560)

Description: Provides clinically relevant and medically actionable information for inherited conditions in standardized journal-style format, covering diagnosis, management, and genetic counseling for patients and their families. Searchable book of expert-authored, peer-reviewed disease descriptions presented in standardized format and focused on clinically relevant and medically actionable information on diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.

Abbreviations: GeneReviews

Resource Type: data or information resource, database

Defining Citation: PMID:20301295

Keywords: genetics, disease, clinical, diagnosis, management, genetic counseling, gene, chromosomal locus, phenotype, allele, locus, mutation

Related Condition: Inherited disease

Availability: Acknowledgement required, Protected by copyright

Resource Name: GeneReviews

Resource ID: SCR_006560

Alternate IDs: OMICS_00269

Record Creation Time: 20220129T080236+0000

Record Last Update: 20260821T193759+0000

Ratings and Alerts

No rating or validation information has been found for GeneReviews.

No alerts have been found for GeneReviews.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 127 mentions in open access literature.

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

Liu X, et al. (2026) Safetyome and specialized panels for over 3,000 phenotypes: a systematic and translational approach using human genetics and pharmacology. Toxicological sciences : an official journal of the Society of Toxicology, 209(3).

Ustach VD, et al. (2026) Multiscore, a gene ranker powered by artificial intelligence and real-world clinical data, shows high sensitivity for the molecular diagnosis of Mendelian disorders in nearly 10,000 exomes and genomes. Human genetics, 145(1), 22.

Ying D, et al. (2026) Population-scale genomic medicine with the Hong Kong Genome Project. Nature medicine, 32(6), 2277.

Allen JD, et al. (2026) Pharmacotherapy Risks in Rare Genetic Diseases: Cross-Referencing ACMG Secondary Findings v3.2 List With Clinical Databases. Clinical and translational science, 19(1), e70464.

Hossain WA, et al. (2026) Identification and molecular functional analysis of genes associated with addiction using integrated genetic web-based programs and databases. Frontiers in genetics, 17, 1763510.

J??ger K, et al. (2026) KLINSE: a comprehensive service model for rare disease information and care management support. Orphanet journal of rare diseases, 21(1).

Nkrumah E, et al. (2026) Burden of heterozygote carriers for autosomal recessive conditions in the Middle East: A study of 14,392 genomes. HGG advances, 7(3), 100625.

Azab B, et al. (2026) Clinical heterogeneity associated with Bardet-Biedl syndrome-related genes in presumed non-syndromic inherited retinal disease. Frontiers in cell and developmental biology, 14, 1802945.

Zhang Z, et al. (2026) Enhanced genetic diagnosis in early pregnancy loss: an integrated approach using CNV-Seq and STR genotyping. Reproductive biology and endocrinology : RB&E, 24(1).

Carstens N, et al. (2026) Implementation of an Inherited Diseases Gene Panel to Accelerate Precision Medicine in the South African Public Healthcare System. Molecular genetics & genomic medicine, 14(4), e70213.

Wanitsuwan W, et al. (2026) WGS identifies Lynch syndrome (LS) patients and uncovers a large family with MSH2-related LS in Southern Thailand. PloS one, 21(5), e0348867.

Kim YG, et al. (2025) Reassessment of variants of uncertain significance in tumor suppressor genes using new ClinGen PP1/PP4 criteria guidance. European journal of human genetics : EJHG, 33(10), 1368.

Bai J, et al. (2025) A novel intronic variant in the ASAH1 gene enhances aberrant splicing, causing spinal muscular atrophy with progressive myoclonic epilepsy. Italian journal of pediatrics, 51(1), 214.

Gold JI, et al. (2025) Exclusion-based exome sequencing in critically ill adults 18-40 years old has a 24% diagnostic rate and finds racial disparities in access to genetic testing. American journal of human genetics, 112(8), 1792.

Laurie S, et al. (2025) Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses. Nature medicine, 31(2), 478.

Kuhlen M, et al. (2025) Non-malignant features of cancer predisposition syndromes manifesting in childhood and adolescence: a guide for the general pediatrician. World journal of pediatrics : WJP, 21(2), 131.

Johnston JJ, et al. (2025) Specification of frequency criteria for secondary findings genes to improve variant classification concordance. bioRxiv : the preprint server for biology.

Matsukawa M, et al. (2025) Establishment of a comprehensive set of fact sheets for cancer predisposition genes for medical oncologists practicing cancer genome profiling. International journal of clinical oncology, 30(5), 827.

Othman AA, et al. (2025) Assessing Large Language Model Performance Related to Aging in Genetic Conditions. medRxiv : the preprint server for health sciences.

Othman AA, et al. (2025) Assessing large language model performance related to aging in genetic conditions. npj aging, 11(1), 33.