Resource Summary Report

Generated by T1D on Aug 3, 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: database, data or information resource

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: 20260803T040441+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 115 mentions in open access literature.

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

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.

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.

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.

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.

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

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. npj aging, 11(1), 33.

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

K??nzel R, et al. (2025) Detecting monogenic obesity: a systematic exome-wide workup of over 500 individuals. International journal of obesity (2005), 49(7), 1400.

Carmona-Hidalgo B, et al. (2025) Efficacy of deep brain stimulation in treating monogenic dystonia symptoms: protocol for a systematic review. BMJ open, 15(4), e083127.

Alizadeh P, et al. (2025) Gene variant analysis in pediatrics with early-onset epilepsy: Identification of novel variants. Practical laboratory medicine, 45, e00462.

Singhal P, et al. (2025) Leveraging Open-Source Large Language Models to Identify Undiagnosed Patients with Rare Genetic Aortopathies. medRxiv : the preprint server for health sciences.

Gudmundsson S, et al. (2025) Exploring penetrance of clinically relevant variants in over 800,000 humans from the Genome Aggregation Database. Nature communications, 16(1), 9623.

Schwartz MLB, et al. (2024) Genetics Visit Uptake Among Individuals Receiving Clinically Actionable Genomic Screening Results. JAMA network open, 7(3), e242388.

Faria-Teixeira MC, et al. (2024) Craniofacial syndromes and class III phenotype: common genotype fingerprints? A scoping review and meta-analysis. Pediatric research, 95(6), 1455.

Li XY, et al. (2024) Genetic profiles of multiple system atrophy revealed by exome sequencing, long-read sequencing and spinocerebellar ataxia repeat expansion analysis. European journal of neurology, 31(12), e16441.

Liu JP, et al. (2024) Improving prenatal diagnosis with combined karyotyping, CNV-seq and QF-PCR: a comprehensive analysis of chromosomal abnormalities in high-risk pregnancies. Frontiers in genetics, 15, 1517270.

Gong Y, et al. (2024) Identification and functional characteristics of a novel splicing heterozygote variant of COL2A1 associated with Stickler syndrome type I. Frontiers in genetics, 15, 1308737.

Kim J, et al. (2024) AutoGVP: a dockerized workflow integrating ClinVar and InterVar germline sequence variant classification. Bioinformatics (Oxford, England), 40(3).