Resource Summary Report

Generated by NIF on Apr 16, 2025

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

Funding:

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: 20250412T055105+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 101 mentions in open access literature.

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

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.

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.

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.

Radziwonik-Fraczyk W, et al. (2024) Next generation sequencing panel as an effective approach to genetic testing in patients with a highly variable phenotype of neuromuscular disorders. Neurogenetics, 25(3), 233.

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

Segura-Tudela A, et al. (2024) Enrichment of Immune Dysregulation Disorders in Adult Patients with Human Inborn Errors of Immunity. Journal of clinical immunology, 44(3), 61.

Wang Z, et al. (2024) VarCards2: an integrated genetic and clinical database for ACMG-AMP variant-interpretation guidelines in the human whole genome. Nucleic acids research, 52(D1), D1478.

Talebizadeh Z, et al. (2024) Landscape Analysis of Neurodevelopmental Comorbidities in Newborn Screening Conditions: Challenges and Opportunities. International journal of neonatal screening, 10(1).

Kerman BJ, et al. (2024) Processes and outcomes from a clinical genetics e-consultation service managed by a primary care physician champion. Genetics in medicine open, 2, 101831.

Gudmundsson S, et al. (2024) Exploring penetrance of clinically relevant variants in over 800,000 humans from the Genome Aggregation Database. bioRxiv : the preprint server for biology.

Gold J, et al. (2024) Universal Exome Sequencing in Critically Ill Adults: A Diagnostic Yield of 25% and Race-Based Disparities in Access to Genetic Testing. medRxiv : the preprint server for health sciences.

Wu R, et al. (2024) Phenotypic and genetic analysis of children with unexplained neurodevelopmental delay and neurodevelopmental comorbidities in a Chinese cohort using trio-based whole-exome sequencing. Orphanet journal of rare diseases, 19(1), 205.

Sawa YC, et al. (2024) Driver mutations associated with signatures of platinum sensitivity in germ cell tumors. NPJ precision oncology, 8(1), 249.

Groza T, et al. (2024) FastHPOCR: pragmatic, fast, and accurate concept recognition using the human phenotype ontology. Bioinformatics (Oxford, England), 40(7).

Baumgartner D, et al. (2024) Genetic Landscape of Amyotrophic Lateral Sclerosis in Czech Patients. Journal of neuromuscular diseases, 11(5), 1035.

Sultana T, et al. (2024) Computational exploration of SLC14A1 genetic variants through structure modeling, protein-ligand docking, and molecular dynamics simulation. Biochemistry and biophysics reports, 38, 101703.

Rao A, et al. (2024) Health-related quality of life in patients with diverse rare diseases: An online survey. Genetics in medicine open, 2, 101889.

Kim J, et al. (2023) Combining chromosomal microarray and clinical exome sequencing for genetic diagnosis of intellectual disability. Scientific reports, 13(1), 22807.