Resource Summary Report

Generated by ASWG on Aug 6, 2026

Nomenclature for the description of sequence variants

RRID:SCR_010261
Type: Tool

Proper Citation

Nomenclature for the description of sequence variants (RRID:SCR_010261)

Resource Information

URL: http://www.hgvs.org/mutnomen/

Proper Citation: Nomenclature for the description of sequence variants (RRID:SCR_010261)

Description: Database of gene mutation nomenclature.

Abbreviations: MUTNOMEN

Resource Type: data or information resource, database

Defining Citation: PMID:10612815

Resource Name: Nomenclature for the description of sequence variants

Resource ID: SCR_010261

Alternate IDs: nlx_156915

Record Creation Time: 20220129T080257+0000

Record Last Update: 20260806T054520+0000

Ratings and Alerts

No rating or validation information has been found for Nomenclature for the description of sequence variants.

No alerts have been found for Nomenclature for the description of sequence variants.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 430 mentions in open access literature.

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

Despas L, et al. (2025) Development of a functional assay for the characterisation of SMAD4 variants from the French haemorrhagic hereditary telangiectasia cohort. Journal of medical genetics, 62(12), 741.

Hartung TI, et al. (2025) Two Different NF1 Pathogenic Variants in a Family With Neurofibromatosis Type 1. Cancer genomics & proteomics, 22(1), 41.

Bayanova M, et al. (2025) Whole-Exome Sequencing Followed by dPCR-Based Personalized Genetic Approach in Solid Organ Transplantation: A Study Protocol and Preliminary Results. Methods and protocols, 8(2).

Yao SQ, et al. (2025) Contrasting pathophysiological mechanisms of OPA1 mutations in autosomal dominant optic atrophy. Cell death discovery, 11(1), 259.

Bayanova M, et al. (2025) Genetic landscape and phenotypic spectrum of osteogenesis imperfecta in the Kazakhstani pediatric population. Scientific reports, 15(1), 11223.

Yang J, et al. (2025) A homozygous PRDX3 pathogenic variant in a paediatric case of spinocerebellar ataxia type 32. Neurogenetics, 26(1), 86.

Mao Y, et al. (2025) Exploring the clinical implications of novel SRD5A2 variants in 46,XY disorders of sex development. Asian journal of andrology, 27(2), 211.

Mazouji O, et al. (2025) Mutational profiling using liquid biopsy to guide targeted therapy in patients with metastatic cancer. Scientific reports, 15(1), 11135.

Fan X, et al. (2024) Possible germline mosaicism in a pedigree with Treacher Collins syndrome: A case report and brief review. Science progress, 107(2), 368504241242278.

Andjelkovic M, et al. (2024) Characterization of 13 Novel Genetic Variants in Genes Associated with Epilepsy: Implications for Targeted Therapeutic Strategies. Molecular diagnosis & therapy, 28(5), 645.

Hu D, et al. (2024) Clinical and genetic characteristics of 100 consecutive patients with Birt-Hogg-Dubé syndrome in Eastern Chinese region. Orphanet journal of rare diseases, 19(1), 348.

Caillot C, et al. (2024) Phenotypic characterisation of SMAD4 variant carriers. Journal of medical genetics, 61(8), 734.

Chen P, et al. (2024) Exploring the impact of a KCNH2 missense variant on Long QT syndrome: insights into a novel gender-selective, incomplete penetrance inheritance mode. Frontiers in genetics, 15, 1409459.

Wang Y, et al. (2023) Exons 1-3 deletion in FLCN is associated with increased risk of pneumothorax in Chinese patients with Birt-Hogg-Dubé syndrome. Orphanet journal of rare diseases, 18(1), 115.

Bayanova M, et al. (2023) Whole-Genome Sequencing Among Kazakhstani Children with Early-Onset Epilepsy Revealed New Gene Variants and Phenotypic Variability. Molecular neurobiology, 60(8), 4324.

Jinda W, et al. (2023) Identification of Genomic Alterations in Thai Patients With Colorectal Cancer Using Next-Generation Sequencing-Based Multigene Cancer Panel. Cureus, 15(5), e39067.

Chong JX, et al. (2023) Variants in ACTC1 underlie distal arthrogryposis accompanied by congenital heart defects. medRxiv : the preprint server for health sciences.

Geberhiwot T, et al. (2023) Consensus clinical management guidelines for acid sphingomyelinase deficiency (Niemann-Pick disease types A, B and A/B). Orphanet journal of rare diseases, 18(1), 85.

Thanprasertsuk S, et al. (2023) Levodopa-induced dyskinesia in early-onset Parkinson's disease (EOPD) associates with glucocerebrosidase mutation: A next-generation sequencing study in EOPD patients in Thailand. PloS one, 18(10), e0293516.

Tian W, et al. (2023) A novel missense variant in OTUD5 causes X-linked multiple congenital anomalies-neurodevelopmental syndrome. Molecular genetics & genomic medicine, 12(1), e2325.