Resource Summary Report

Generated by FDI Lab - SciCrunch.org on May 25, 2025

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

Funding:

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: 20250525T032341+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 422 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

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

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.

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.

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.

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.

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

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.

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.

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.

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.

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

Blue EE, et al. (2023) Rare variants in CAPN2 increase risk for isolated hypoplastic left heart syndrome. HGG advances, 4(4), 100232.

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.

Ralph D, et al. (2022) ENPP1 variants in patients with GACI and PXE expand the clinical and genetic heterogeneity of heritable disorders of ectopic calcification. PLoS genetics, 18(4), e1010192.

Fei H, et al. (2022) Exome sequencing and RNA analysis identify two novel CPLANE1 variants causing Joubert syndrome. Molecular genetics & genomic medicine, 10(3), e1877.

Almashagbah NA, et al. (2022) Pharmacogenetic Study of the Dihydropyridine Dehydrogenase Gene in Jordanian Patients with Colorectal Cancer. Asian Pacific journal of cancer prevention : APJCP, 23(9), 3061.

Spiegel R, et al. (2022) Molybdenum cofactor deficiency: A natural history. Journal of inherited metabolic disease, 45(3), 456.

Forny P, et al. (2022) Recovery of enzyme activity in biotinidase deficient individuals during early childhood. Journal of inherited metabolic disease, 45(3), 605.

Liccardo R, et al. (2022) Significance of rare variants in genes involved in the pathogenesis of Lynch syndrome. International journal of molecular medicine, 49(6).