Resource Summary Report

Generated by dkNET on Aug 7, 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: database, data or information resource

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: 20260807T042730+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 460 mentions in open access literature.

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

Han M, et al. (2026) Genetic spectrum and risk factor analysis for drug-resistant of early-onset epilepsy. Translational pediatrics, 15(3), 75.

Xu J, et al. (2026) Exploring the pathogenicity and genotype-phenotype correlation of 16p13.11 microduplication: a report of two cases. Translational pediatrics, 15(1), 23.

Chausova P, et al. (2026) Identification and Targeted Correction of a Pathogenic PMP22 Deep Intronic Variant. International journal of molecular sciences, 27(8).

Freyberg M, et al. (2026) Large variations in total and allele-specific transcript expression in a disease mutation-independent manner. Scientific reports, 16(1).

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

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.

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.

Imangaliyeva A, et al. (2025) Genetic Insights into Severe Obesity: A Case Study of MC4R Variant Identification and Clinical Implications. Genes, 16(5).

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

Bayanova M, et al. (2025) Whole-exome sequencing of kidney transplant recipients and donors: insights into end-stage renal disease and post-transplant genetic risk. BMC nephrology, 26(1), 617.

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.

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).

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

Nekrasov A, et al. (2025) Functional Characterization of HGD Gene Variants by Minigene Splicing Assay. International journal of molecular sciences, 26(21).

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

Ogorodova N, et al. (2024) A Comparative Evaluation of the Genetic Variant Spectrum in the USH2A Gene in Russian Patients with Isolated and Syndromic Forms of Retinitis Pigmentosa. International journal of molecular sciences, 25(22).

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.

Gisriel SD, et al. (2024) Optimization criteria for ordering myeloid neoplasm next-generation sequencing. EJHaem, 5(6), 1236.