Resource Summary Report

Generated by <u>dkNET</u> on Apr 16, 2025

dbSNP

RRID:SCR_002338 Type: Tool

Proper Citation

dbSNP (RRID:SCR_002338)

Resource Information

URL: http://www.ncbi.nlm.nih.gov/SNP/

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Description: Database as central repository for both single base nucleotide substitutions and short deletion and insertion polymorphisms. Distinguishes report of how to assay SNP from use of that SNP with individuals and populations. This separation simplifies some issues of data representation. However, these initial reports describing how to assay SNP will often be accompanied by SNP experiments measuring allele occurrence in individuals and populations. Community can contribute to this resource.

Abbreviations: dbSNP

Synonyms: dbSNP: Database for Short Genetic Variations, Entrez SNP - Single Nucleotide Polymorphism, SNV Database, NCBI SNV Database, NCBI Short Genetic Variations Database, NCBI Short Genetic Variations, NCBI Single Nucleotide Polymorphism, Entrez SNP, dbSNP, NCBI Short Genetic Variations (SNV) database

Resource Type: database, data or information resource, service resource, data repository, storage service resource

Defining Citation: PMID:21154707

Keywords: insertion, polymorphism, short, deletion, single, nucleotide, genetic, variation, genomics, genotype, disease, allele, microsatellite, marker, multinucleotide, heterozygous, sequence, gold standard, bio.tools

Funding: NLM

Availability: Free, Freely available

Resource Name: dbSNP

Resource ID: SCR_002338

Alternate IDs: nif-0000-02734, biotools:dbsnp, OMICS_00264

Alternate URLs: http://www.ncbi.nlm.nih.gov/projects/SNP/, https://bio.tools/dbsnp

Record Creation Time: 20220129T080212+0000

Record Last Update: 20250416T063259+0000

Ratings and Alerts

No rating or validation information has been found for dbSNP.

No alerts have been found for dbSNP.

Data and Source Information

Source: <u>SciCrunch Registry</u>

Usage and Citation Metrics

We found 8217 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>dkNET</u>.

Tremmel R, et al. (2025) Translating pharmacogenomic sequencing data into drug response predictions-How to interpret variants of unknown significance. British journal of clinical pharmacology, 91(2), 252.

Wang J, et al. (2025) CAUSALdb2: an updated database for causal variants of complex traits. Nucleic acids research, 53(D1), D1295.

Tihagam RD, et al. (2025) The TRIM37 variant rs57141087 contributes to triple-negative breast cancer outcomes in Black women. EMBO reports, 26(1), 245.

Huang X, et al. (2025) Mutation spectra and genotype?phenotype analysis of congenital hypothyroidism in a neonatal population. Biomedical reports, 22(2), 30.

Verras GI, et al. (2025) Utility of Polygenic Risk Scores (PRSs) in Predicting Pancreatic Cancer: A Systematic Review and Meta-Analysis of Common-Variant and Mixed Scores with Insights into Rare Variant Analysis. Cancers, 17(2).

Alghamdi MA, et al. (2025) Genomic Insights into Blood Pressure Regulation: Exploring Ion Channel and Transporter Gene Variations in Jordanian Hypertensive Individuals. Medicina (Kaunas, Lithuania), 61(1).

Magistrati M, et al. (2025) De Novo DNM1L Pathogenic Variant Associated with Lethal Encephalocardiomyopathy-Case Report and Literature Review. International journal of molecular sciences, 26(2).

Cifaldi C, et al. (2025) Partial Loss of NEMO Function in a Female Carrier with No Incontinentia Pigmenti. Journal of clinical medicine, 14(2).

Yang F, et al. (2025) Acquired multiple EGFR mutations?mediated resistance to a third?generation tyrosine kinase inhibitor in a patient with lung adenocarcinoma who responded to afatinib: A case report and literature review. Oncology letters, 29(2), 81.

Kokuryo T, et al. (2025) Whole-genome Sequencing Analysis of Bile Tract Cancer Reveals Mutation Characteristics and Potential Biomarkers. Cancer genomics & proteomics, 22(1), 34.

Gao S, et al. (2025) Unraveling the genetic mysteries of spinal muscular atrophy in Chinese families. Orphanet journal of rare diseases, 20(1), 25.

Kzar WA, et al. (2025) Association of Polymorphism with Periodontitis and Salivary Levels of Hypoxia-Inducible Factor-1?. European journal of dentistry, 19(1), 133.

Mózner O, et al. (2025) Potential associations of selected polymorphic genetic variants with COVID-19 disease susceptibility and severity. PloS one, 20(1), e0316396.

Heimer G, et al. (2025) Biallelic PIGM Coding Variant Causes Intractable Epilepsy and Intellectual Disability Without Thrombotic Events. Clinical genetics, 107(2), 179.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Yang L, et al. (2025) A novel de novo GABRA2 gene missense variant causing developmental epileptic encephalopathy in a Chinese patient. Annals of clinical and translational neurology, 12(1), 137.

Mendeville MS, et al. (2025) Integrating genetic subtypes with PET scan monitoring to predict outcome in diffuse large B-cell lymphoma. Nature communications, 16(1), 109.

Koponen L, et al. (2025) A deep intronic PHEX variant associated with X-linked hypophosphatemia in a Finnish family. JBMR plus, 9(2), ziae169.

Wang Z, et al. (2025) Optimizing the NGS-based discrimination of multiple lung cancers from the perspective of evolution. NPJ precision oncology, 9(1), 14.

Ma SL, et al. (2025) Functional significance of some common oxytocin receptor SNPs involved in complex human traits. BMC molecular and cell biology, 26(1), 3.