Resource Summary Report

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dbNSFP

RRID:SCR_005178
Type: Tool

Proper Citation

dbNSFP (RRID:SCR_005178)

Resource Information

URL: https://sites.google.com/site/jpopgen/dbNSFP

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Description: A database for functional prediction and annotation of all potential non-synonymous single-nucleotide variants (nsSNVs) in the human genome. Version 2.0 is based on the Gencode release 9 / Ensembl version 64 and includes a total of 87,347,043 nsSNVs and 2,270,742 essential splice site SNVs. It compiles prediction scores from six prediction algorithms (SIFT, Polyphen2, LRT, MutationTaster, MutationAssessor and FATHMM), three conservation scores (PhyloP, GERP++ and SiPhy) and other related information including allele frequencies observed in the 1000 Genomes Project phase 1 data and the NHLBI Exome Sequencing Project, various gene IDs from different databases, functional descriptions of genes, gene expression and gene interaction information, etc. Some dbNSFP contents (may not be up-to-date though) can also be accessed through variant tools, ANNOVAR, KGGSeq, UCSC Genome Browser''s Variant Annotation Integrator, Ensembl Variant Effect Predictor and HGMD.

Abbreviations: dbNSFP

Resource Type: data or information resource, database

Defining Citation: PMID:23843252, PMID:21520341

Keywords: non-synonymous single-nucleotide variant, function, annotation, functional prediction, non-synonymous mutation, splice site mutation, FASEB list

Availability: Acknowledgement requested, Free, Public

Resource Name: dbNSFP

Resource ID: SCR_005178

Alternate IDs: OMICS_00172

Record Creation Time: 20220129T080228+0000

Record Last Update: 20260905T133128+0000

Ratings and Alerts

No rating or validation information has been found for dbNSFP.

No alerts have been found for dbNSFP.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 708 mentions in open access literature.

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

Jang SK, et al. (2026) Leveraging protein language models to identify complex trait associations with previously inaccessible classes of functional rare variants. Cell genomics, 6(2), 101068.

Jeong R, et al. (2026) Functional analysis of NPR2 variants supports the therapeutic rationale for CNP in short stature. American journal of human genetics, 113(1), 117.

Rony R, et al. (2026) Putative breast cancer risk variants from populations of South Asian ancestry are under-represented in public variant classification databases. Breast cancer research : BCR, 28(1), 5.

Wang J, et al. (2026) Genetic diagnosis in fetal hydronephrosis: assessment using chromosomal microarray analysis and whole-genome sequencing. Human genomics, 20(1).

Shi T, et al. (2026) Functional reassessment of extended splice region variants in MYO7A with hearing loss and Usher syndrome. The Journal of pathology, 269(2), 222.

Carta MG, et al. (2026) Oncogenicity Variant Interpreter (OncoVI) Supports Harmonized Somatic Variant Interpretation in Precision Oncology. The Journal of molecular diagnostics : JMD, 28(6), 469.

Shea A, et al. (2026) SomaMutDB 2.0: A comprehensive database for exploring somatic mutations and their functional impact in normal human tissues. Nucleic acids research, 54(D1), D1291.

Ruan J, et al. (2026) MYOM1 deficiency leads to dilated cardiomyopathy by disrupting the homeostasis of sarcoplasmic reticulum. Redox biology, 91, 104072.

Lee DB, et al. (2026) Disease- and gene-specific deep learning for pathogenicity prediction of rare missense variants in cancer predisposition genes. BioData mining, 19(1).

Kim C, et al. (2026) Clinical and genetic features of idiopathic pulmonary fibrosis in a Korean cohort. Respiratory research, 27(1).

Wanitsuwan W, et al. (2026) WGS identifies Lynch syndrome (LS) patients and uncovers a large family with MSH2-related LS in Southern Thailand. PloS one, 21(5), e0348867.

Lee KL, et al. (2026) Non-BRCA germline pathogenic variants and response to neoadjuvant systemic therapy in triple-negative breast cancer patients enrolled in a prospective clinical trial. Breast cancer research and treatment, 217(2).

Islam N, et al. (2026) Single nucleotide polymorphisms affecting galantamine binding to acetylcholinesterase in Alzheimer's disease: a structural bioinformatics study. Journal of computer-aided molecular design, 40(1).

Reuter P, et al. (2026) Systematic functional evaluation of CNGA1 missense variants associated with retinitis pigmentosa. Molecular medicine (Cambridge, Mass.), 32(1).

Li K, et al. (2026) Mapping the Non-Canonical Splicing Variants: Decrypting the Hidden Genetic Architecture of Idiopathic Male Infertility. Advanced science (Weinheim, Baden-Wurttemberg, Germany), 13(9), e15512.

Zafar A, et al. (2026) Molecular dynamics simulations of intrinsically disordered protein regions enable biophysical interpretation of variant-effect predictors. HGG advances, 7(3), 100618.

Pan LY, et al. (2026) Genetic Variants and Clinical Characteristics of Young-Onset Parkinson's Disease in the Hakka Population of Western Fujian. Brain and behavior, 16(6), e71504.

Komrokji RS, et al. (2026) Impact of Mutational Landscape and Burden on RBC Transfusion Response in Patients With Lower-Risk Myelodysplastic Syndromes (LR-MDS) in the COMMANDS Study. American journal of hematology, 101(3), 427.

Buianova AA, et al. (2026) ACE-Dependent Alzheimer's Disease: Blood ACE Phenotyping of the Most Prevalent and Damaging ACE Missense Mutation-Y215C (rs3730025). Biomedicines, 14(2).

Hussain HMJ, et al. (2026) Bi-allelic variants in AP5Z1 and AP5B1 lead to retinal degeneration. HGG advances, 7(2), 100584.