Resource Summary Report

Generated by RRID on Aug 5, 2026

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: database, data or information resource

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: 20260805T044118+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 648 mentions in open access literature.

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

Peng T, et al. (2025) Individualized patient tumor organoids faithfully preserve human brain tumor ecosystems and predict patient response to therapy. Cell stem cell, 32(4), 652.

Fiorica PN, et al. (2025) Germline Pathogenic DROSHA Variants Are Linked to Pineoblastoma and Wilms Tumor Predisposition. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(8), 1491.

Yavas C, et al. (2025) Revealing Molecular Diagnosis With Whole Exome Sequencing in Patients With Inherited Retinal Disorders. Clinical genetics, 108(1), 14.

Rastogi R, et al. (2025) Critical assessment of missense variant effect predictors on disease-relevant variant data. Human genetics, 144(2-3), 281.

Vooren EV, et al. (2025) RPE65 variant p.(E519K) causes a novel dominant adult-onset maculopathy in 83 affected individuals. Research square.

Zafar A, et al. (2025) Molecular dynamics simulations of intrinsically disordered protein regions enable biophysical interpretation of variant effect predictors. bioRxiv : the preprint server for biology.

Mellone S, et al. (2025) Integrating genome and transcriptome analysis to decipher balanced structural variants in unsolved cases of neurodevelopmental disorders. Frontiers in genetics, 16, 1603513.

Hatzikotoulas K, et al. (2025) Translational genomics of osteoarthritis in 1,962,069 individuals. Nature, 641(8065), 1217.

Büyükgöl F, et al. (2025) Exome sequencing reveals low-frequency and rare variant contributions to multiple sclerosis susceptibility in Turkish families. Scientific reports, 15(1),

11682.

Scudeler MM, et al. (2025) ESR1 Variants and Subcontinental Genomic Ancestry: Insights from the 1000 Genomes Project and Native American Populations. Clinical pharmacology and therapeutics, 118(1), 232.

Tabata K, et al. (2025) Rare genetic variants involved in increased risk of paroxysmal atrial fibrillation in a Japanese population. Scientific reports, 15(1), 13216.

Veyhe SR, et al. (2025) A Case-Driven Multi-Omics Analysis for Longitudinal Ibrutinib Response Evaluation of Patients With Chronic Lymphocytic Leukemia. European journal of haematology, 114(6), 973.

Akimova D, et al. (2025) De Novo HNRNPU Pathogenic Variant Related to Developmental Epileptic Encephalopathy With Inherited KANSL1 Loss-of-Function Variant Resolved by RNA Analysis. Molecular genetics & genomic medicine, 13(9), e70127.

Orenbuch R, et al. (2025) Proteome-wide model for human disease genetics. Nature genetics, 57(12), 3165.

Koyama S, et al. (2025) Genetics and context for precision health in Greater Boston. Nature communications, 16(1), 11661.

Matsushita K, et al. (2025) Importance of EQA/PT for the detection of genetic variants in comprehensive cancer genome testing. Scientific reports, 15(1), 1036.

Toriello E, et al. (2025) Characterization of a WAS splice-site variant in a patient with Wiskott-Aldrich syndrome. Frontiers in immunology, 16, 1517347.

Nishioka M, et al. (2025) Mosaic variants of neurodevelopmental and mitochondrial genes in postmortem paraventricular thalamus in bipolar disorder detected by deep exome sequencing. Psychiatry and clinical neurosciences, 79(10), 653.

Zhong G, et al. (2025) PreMode predicts mode-of-action of missense variants by deep graph representation learning of protein sequence and structural context. Nature communications, 16(1), 7189.

Heo JY, et al. (2025) Assessing the performance of 28 pathogenicity prediction methods on rare single nucleotide variants in coding regions. BMC genomics, 26(1), 641.