

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: 20260801T190927+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 [ASWG](#) .

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.

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.

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

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

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

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*.

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.

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.

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.

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

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.

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.

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.

Curtis D, et al. (2025) Assessment of ability of a DNA language model to predict pathogenicity of rare coding variants. *Journal of human genetics*, 70(11), 603.

Merepa SS, et al. (2025) GJA8-associated developmental eye disorders: a new multicentre study highlights mutational hotspots and genotype-phenotype correlations. *European journal of human genetics : EJHG*, 33(7), 860.