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

Funding:

Availability: Acknowledgement requested, Free, Public

Resource Name: dbNSFP

Resource ID: SCR_005178

Alternate IDs: OMICS_00172

Record Creation Time: 20220129T080228+0000

Record Last Update: 20250422T055226+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 587 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

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.

Katsonis P, et al. (2025) Meta-EA: a gene-specific combination of available computational tools for predicting missense variant effects. Nature communications, 16(1), 159.

Scherer N, et al. (2025) Coupling metabolomics and exome sequencing reveals graded effects of rare damaging heterozygous variants on gene function and human traits. Nature genetics, 57(1), 193.

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

Huang C, et al. (2025) Comparative genetic analysis of blood and semen samples in sperm donors from Hunan, China. Annals of medicine, 57(1), 2447421.

Kock KH, et al. (2024) DNA binding analysis of rare variants in homeodomains reveals homeodomain specificity-determining residues. Nature communications, 15(1), 3110.

Tritto V, et al. (2024) Genetic/epigenetic effects in NF1 microdeletion syndrome: beyond the

haploinsufficiency, looking at the contribution of not deleted genes. Human genetics, 143(6), 775.

Guerrini-Rousseau L, et al. (2024) Medulloblastomas with ELP1 pathogenic variants: A weakly penetrant syndrome with a restricted spectrum in a limited age window. Neuro-oncology advances, 6(1), vdae075.

Zlotina A, et al. (2024) Characterization of pathogenic genetic variants in Russian patients with primary ciliary dyskinesia using gene panel sequencing and transcript analysis. Orphanet journal of rare diseases, 19(1), 310.

Carpentieri G, et al. (2024) Dominantly acting variants in ATP6V1C1 and ATP6V1B2 cause a multisystem phenotypic spectrum by altering lysosomal and/or autophagosome function. HGG advances, 5(4), 100349.

Schmidt A, et al. (2024) Systematic assessment of COVID-19 host genetics using whole genome sequencing data. PLoS pathogens, 20(12), e1012786.

Park Y, et al. (2024) Whole-Exome Sequencing Improves Understanding of Inherited Retinal Dystrophies in Korean Patients. Current issues in molecular biology, 46(10), 11021.

Danilov SM, et al. (2024) Carriers of Heterozygous Loss-of-Function ACE Mutations Are at Risk for Alzheimer's Disease. Biomedicines, 12(1).

Zhao H, et al. (2024) SIGMA leverages protein structural information to predict the pathogenicity of missense variants. Cell reports methods, 4(1), 100687.

Al-Mutairi DA, et al. (2024) Novel pathogenic variants of DNAH5 associated with clinical and genetic spectra of primary ciliary dyskinesia in an Arab population. Frontiers in genetics, 15, 1396797.

Idiiatullina E, et al. (2024) Heterozygous de novo dominant negative mutation of REXO2 results in interferonopathy. Nature communications, 15(1), 6685.

Grigore LG, et al. (2024) The Molecular Detection of Germline Mutations in the BRCA1 and BRCA2 Genes Associated with Breast and Ovarian Cancer in a Romanian Cohort of 616 Patients. Current issues in molecular biology, 46(5), 4630.

Wang Z, et al. (2024) VarCards2: an integrated genetic and clinical database for ACMG-AMP variant-interpretation guidelines in the human whole genome. Nucleic acids research, 52(D1), D1478.

Wang Y, et al. (2024) Whole-exome sequencing combined with postoperative data identify c.1614dup (CAMKK2) as a novel candidate monogenic obesity variant. Frontiers in endocrinology, 15, 1334342.

Kwak SH, et al. (2024) Genetic architecture and biology of youth-onset type 2 diabetes. Nature metabolism, 6(2), 226.