Resource Summary Report

Generated by <u>NIF</u> on May 15, 2025

Variant Effect Predictor

RRID:SCR_007931 Type: Tool

Proper Citation

Variant Effect Predictor (RRID:SCR_007931)

Resource Information

URL: http://www.ensembl.org/info/docs/tools/vep/index.html

Proper Citation: Variant Effect Predictor (RRID:SCR_007931)

Description: Data analysis service to predict the functional consequences of known and unknown variants.

Abbreviations: VEP

Synonyms: Ve!P

Resource Type: software resource, service resource, analysis service resource, production service resource, data analysis service

Keywords: perl, bio.tools

Funding:

Resource Name: Variant Effect Predictor

Resource ID: SCR_007931

Alternate IDs: biotools:ensembl_variant_effect_predictor

Alternate URLs: https://bio.tools/ensembl_variant_effect_predictor

Record Creation Time: 20220129T080244+0000

Record Last Update: 20250514T061435+0000

Ratings and Alerts

No rating or validation information has been found for Variant Effect Predictor.

No alerts have been found for Variant Effect Predictor.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 1629 mentions in open access literature.

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

Kentistou KA, et al. (2025) Rare variant associations with birth weight identify genes involved in adipose tissue regulation, placental function and insulin-like growth factor signalling. Nature communications, 16(1), 648.

Yao Y, et al. (2025) Whole-genome sequencing identifies novel loci for keratoconus and facilitates risk stratification in a Han Chinese population. Eye and vision (London, England), 12(1), 5.

Fan C, et al. (2025) Genome-wide association study of myofiber type composition traits in a yellow-feather broiler population. Poultry science, 104(1), 104634.

Bignotti E, et al. (2025) TP53 mutations and survival in ovarian carcinoma patients receiving first-line chemotherapy plus bevacizumab: Results of the MITO16A/MaNGO OV-2 study. International journal of cancer, 156(5), 1085.

Zhang Y, et al. (2025) A multiscale functional map of somatic mutations in cancer integrating protein structure and network topology. Nature communications, 16(1), 975.

Jung S, et al. (2025) Rare Variant Analyses in Ancestrally Diverse Cohorts Reveal Novel ADHD Risk Genes. medRxiv : the preprint server for health sciences.

Alfayyadh MM, et al. (2025) PathVar: A Customisable NGS Variant Calling Algorithm Implicates Novel Candidate Genes and Pathways in Hemiplegic Migraine. Clinical genetics, 107(2), 157.

Rostamzadeh Mahdabi E, et al. (2025) Comparative Analysis of Runs of Homozygosity Islands in Indigenous and Commercial Chickens Revealed Candidate Loci for Disease Resistance and Production Traits. Veterinary medicine and science, 11(1), e70074.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum

phenotype. American journal of human genetics, 112(1), 59.

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.

Gupta AA, et al. (2025) Durvalumab and tremelimumab in patients with advanced rare cancer: a multi-centre, non-blinded, open-label phase II basket trial. EClinicalMedicine, 79, 102991.

Shin HD, et al. (2025) Chromosome-level Genome Assembly of Korean Long-tailed Chicken and Pangenome of 40 Gallus gallus Assemblies. Scientific data, 12(1), 51.

Spedicati B, et al. (2025) Scent of COVID-19: Whole-Genome Sequencing Analysis Reveals the Role of ACE2, IFI44, and NDUFAF4 in Long-Lasting Olfactory Dysfunction. Life (Basel, Switzerland), 15(1).

Skystad Kvernebo M, et al. (2025) Genetic Variants in the SCN9A Gene are Detected in a Minority of Erythromelalgia Patients. Acta dermato-venereologica, 105, adv42022.

Turan B, et al. (2025) Investigating Sequence Variations in CNTNAP2 and SETBP1 Genes in Language Disorders. Clinical psychopharmacology and neuroscience : the official scientific journal of the Korean College of Neuropsychopharmacology, 23(1), 100.

Xu H, et al. (2025) Landscape of human protein-coding somatic mutations across tissues and individuals. bioRxiv : the preprint server for biology.

Avila MN, et al. (2025) Deleterious coding variation associated with autism is consistent across populations, as exemplified by admixed Latin American populations. medRxiv : the preprint server for health sciences.

Paris JR, et al. (2025) The Genomic Signature and Transcriptional Response of Metal Tolerance in Brown Trout Inhabiting Metal-Polluted Rivers. Molecular ecology, 34(1), e17591.

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

Kim JM, et al. (2025) Uncovering potential causal genes for undiagnosed congenital anomalies using an in-house pipeline for trio-based whole-genome sequencing. Human genomics, 19(1), 1.