

Resource Summary Report

Generated by [RRID](#) on Apr 8, 2025

PhenCode

RRID:SCR_010799

Type: Tool

Proper Citation

PhenCode (RRID:SCR_010799)

Resource Information

URL: <http://phencode.bx.psu.edu/>

Proper Citation: PhenCode (RRID:SCR_010799)

Description: A collaborative project to better understand the relationship between genotype and phenotype in humans that connects human phenotype and clinical data in various locus-specific mutation databases (LSDBs) with data on genome sequences, evolutionary history, and function in the UCSC Genome Browser. PhenCode is a collaboration among researchers at Penn State, UC Santa Cruz, and locus experts at other institutions.

Abbreviations: PhenCode

Synonyms: PhenCode: Paving the Path between Phenotype and Genome, Phenotypes for ENCODE

Resource Type: data or information resource, database

Defining Citation: [PMID:17326095](#)

Keywords: genotype, phenotype, mutation

Funding:

Availability: Acknowledgement requested, Free

Resource Name: PhenCode

Resource ID: SCR_010799

Alternate IDs: OMICS_00279

Record Creation Time: 20220129T080300+0000

Record Last Update: 20250404T060854+0000

Ratings and Alerts

No rating or validation information has been found for PhenCode.

No alerts have been found for PhenCode.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 7 mentions in open access literature.

Listed below are recent publications. The full list is available at [RRID](#).

Livesey BJ, et al. (2022) Interpreting protein variant effects with computational predictors and deep mutational scanning. *Disease models & mechanisms*, 15(6).

Zaucha J, et al. (2020) Family-specific analysis of variant pathogenicity prediction tools. *NAR genomics and bioinformatics*, 2(2), lqaa014.

Schaafsma GCP, et al. (2018) Representativeness of variation benchmark datasets. *BMC bioinformatics*, 19(1), 461.

van der Velde KJ, et al. (2017) GAVIN: Gene-Aware Variant INterpretation for medical sequencing. *Genome biology*, 18(1), 6.

De Baets G, et al. (2015) Increased Aggregation Is More Frequently Associated to Human Disease-Associated Mutations Than to Neutral Polymorphisms. *PLoS computational biology*, 11(9), e1004374.

Giollo M, et al. (2015) BOOGIE: Predicting Blood Groups from High Throughput Sequencing Data. *PloS one*, 10(4), e0124579.

Frousios K, et al. (2013) Predicting the functional consequences of non-synonymous DNA sequence variants--evaluation of bioinformatics tools and development of a consensus strategy. *Genomics*, 102(4), 223.