

Resource Summary Report

Generated by [NIF](#) on May 21, 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: database, data or information resource

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: 20250519T204807+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 [NIF](#).

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.