Resource Summary Report

Generated by dkNET on May 19, 2025

PhenoDB

RRID:SCR_016551
Type: Tool

Proper Citation

PhenoDB (RRID:SCR_016551)

Resource Information

URL: https://phenodb.org/

Proper Citation: PhenoDB (RRID:SCR_016551)

Description: Database for phenotype genotype associations for humans. Used by clinical researchers to store standardized phenotypic information, diagnosis, and pedigree data and then run analyses on VCF files from individuals, families or cohorts with suspected Mendelian disease.

Resource Type: database, data or information resource

Defining Citation: PMID:25684268

Keywords: store, standardized, phenotype, genotype, Mendelian disease, mutation, next, generation, sequencing, data

Related Condition: Mendelian disease

Funding: NHGRI

Availability: Free, Registration required, Freely available for non commercial users

Resource Name: PhenoDB

Resource ID: SCR_016551

Record Creation Time: 20220129T080331+0000

Record Last Update: 20250519T204829+0000

Ratings and Alerts

No rating or validation information has been found for PhenoDB.

No alerts have been found for PhenoDB.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 5 mentions in open access literature.

Listed below are recent publications. The full list is available at dkNET.

von Hardenberg S, et al. (2024) Current genetic diagnostics in inborn errors of immunity. Frontiers in pediatrics, 12, 1279112.

Coban-Akdemir Z, et al. (2024) The impact of the Turkish population variome on the genomic architecture of rare disease traits. Genetics in medicine open, 2, 101830.

de Mello LEB, et al. (2022) Identification of NID1 as a novel candidate susceptibility gene for familial non-medullary thyroid carcinoma using whole-exome sequencing. Endocrine connections, 11(1).

Braunstein EM, et al. (2021) Germline ERBB2/HER2 Coding Variants Are Associated with Increased Risk of Myeloproliferative Neoplasms. Cancers, 13(13).

Bosio M, et al. (2019) eDiVA-Classification and prioritization of pathogenic variants for clinical diagnostics. Human mutation, 40(7), 865.