

# Resource Summary Report

Generated by SPARC SAWG on Sep 17, 2026

## 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:** data or information resource, database

**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:** 20260912T200217+0000

### Ratings and Alerts

No rating or validation information has been found for PhenoDB.

No alerts have been found for PhenoDB.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 5 mentions in open access literature.

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

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.