Resource Summary Report

Generated by SPARC SAWG on Sep 13, 2026

Phenomizer

RRID:SCR_006157
Type: Tool

Proper Citation

Phenomizer (RRID:SCR_006157)

Resource Information

URL: http://compbio.charite.de/phenomizer/

Proper Citation: Phenomizer (RRID:SCR_006157)

Description: THIS RESOURCE IS NO LONGER IN SERVICE. Documented on March 31,2026. Phenomizer offers three different approaches to find the appropriate term for a phenotypic abnormality, indicated by the three tabs on the left hand side: Feature, Disease and Ontology. The Phenomizer is intended to be used by qualified and licensed physicians in order to provide assistance in reaching the correct diagnosis in patients with hereditary diseases and for use as a teaching aid. The Phenomizer does not make diagnoses. Rather, it produces a ranked list of possibilities that can be used by physicians as a part of the diagnostic workup. The Phenomizer does not contain information about all possible diagnoses or even all possible hereditary diseases. The Phenomizer should not be used to make medical decisions without the advice of a physician.

Synonyms: Phenomizer - Clinical Diagnostics with Similarity Searches in Ontologies

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

Defining Citation: PMID:19800049

Keywords: feature, disease, ontology, clinical, differential diagnoses

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: Phenomizer

Resource ID: SCR_006157

Alternate IDs: nlx_151657

Record Creation Time: 20220129T080234+0000

Record Last Update: 20260912T200141+0000

Ratings and Alerts

No rating or validation information has been found for Phenomizer.

No alerts have been found for Phenomizer.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 32 mentions in open access literature.

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

Harsono IW, et al. (2025) Clinical phenotype and trio whole exome sequencing data from a patient with glycogen storage disease IV in Indonesia. Data in brief, 58, 111231.

Abarca-Barriga HH, et al. (2025) Identification of intragenic variants in pediatric patients with intellectual disability in Peru. BMC medical genomics, 18(1), 76.

Martín López-Pardo B, et al. (2025) Transforming NICU care: rapid WES and transcriptomics-validation, social impact, and cost analysis. European journal of pediatrics, 184(7), 453.

Faviez C, et al. (2024) Objectivizing issues in the diagnosis of complex rare diseases: lessons learned from testing existing diagnosis support systems on ciliopathies. BMC medical informatics and decision making, 24(1), 134.

Reese JT, et al. (2023) Generalisable long COVID subtypes: findings from the NIH N3C and RECOVER programmes. EBioMedicine, 87, 104413.

Alsentzer E, et al. (2023) Simulation of undiagnosed patients with novel genetic conditions. Nature communications, 14(1), 6403.

El-Gazzar A, et al. (2023) Bi-allelic mutation in SEC16B alters collagen trafficking and increases ER stress. EMBO molecular medicine, 15(4), e16834.

Hsu RH, et al. (2023) Utility of whole-exome sequencing for patients with multiple congenital anomalies with or without intellectual disability/developmental delay in East Asia population. Molecular genetics & genomic medicine, 11(6), e2160.

Chen Z, et al. (2022) PhenoApt leverages clinical expertise to prioritize candidate genes via machine learning. American journal of human genetics, 109(2), 270.

Reese JT, et al. (2022) Generalizable Long COVID Subtypes: Findings from the NIH N3C and RECOVER Programs. medRxiv : the preprint server for health sciences.

Barbosa-Gouveia S, et al. (2021) Utility of Gene Panels for the Diagnosis of Inborn Errors of Metabolism in a Metabolic Reference Center. Genes, 12(8).

Pollini L, et al. (2020) KCND3-Related Neurological Disorders: From Old to Emerging Clinical Phenotypes. International journal of molecular sciences, 21(16).

Díaz-Santiago E, et al. (2020) Phenotype-genotype comorbidity analysis of patients with rare disorders provides insight into their pathological and molecular bases. PLoS genetics, 16(10), e1009054.

Thiffault I, et al. (2020) Pathogenic variants in KPTN gene identified by clinical whole-genome sequencing. Cold Spring Harbor molecular case studies, 6(3).

de Castro MJ, et al. (2020) Rapid Phenotype-Driven Gene Sequencing with the NeoSeq Panel: A Diagnostic Tool for Critically Ill Newborns with Suspected Genetic Disease. Journal of clinical medicine, 9(8).

Faviez C, et al. (2020) Diagnosis support systems for rare diseases: a scoping review. Orphanet journal of rare diseases, 15(1), 94.

Cheng L, et al. (2019) Computational Methods for Identifying Similar Diseases. Molecular therapy. Nucleic acids, 18, 590.

Zhang Y, et al. (2019) The Potential Mechanism of Bufadienolide-Like Chemicals on Breast Cancer via Bioinformatics Analysis. Cancers, 11(1).

Rao A, et al. (2018) Phenotype-driven gene prioritization for rare diseases using graph convolution on heterogeneous networks. BMC medical genomics, 11(1), 57.

Fujiwara T, et al. (2018) PubCaseFinder: A Case-Report-Based, Phenotype-Driven Differential-Diagnosis System for Rare Diseases. American journal of human genetics, 103(3), 389.