

Resource Summary Report

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Human Phenotype Ontology

RRID:SCR_006016

Type: Tool

Proper Citation

Human Phenotype Ontology (RRID:SCR_006016)

Resource Information

URL: <http://www.human-phenotype-ontology.org/>

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Description: Provides standardized vocabulary of phenotypic abnormalities encountered in human disease. Structured and controlled vocabulary for phenotypic features encountered in human hereditary and other disease. HPO is being developed in collaboration with members of OBO Foundry (Open Biological and Biomedical Ontologies), and logical definitions for HPO terms are being developed using PATO and a number of other ontologies including FMA, GO, ChEBI, and MPATH.

Abbreviations: HPO, HP

Synonyms: Human Phenotype Ontology (HPO), Human Phenotype Ontology

Resource Type: ontology, data or information resource, controlled vocabulary

Defining Citation: [PMID:20412080](#)

Keywords: phenotype, genetics, disease, phenomizer, obo, clinical, phenome, pathological, organismal, FASEB list

Related Condition: Monogenic disease, Hereditary disease

Availability: Free, Freely available

Resource Name: Human Phenotype Ontology

Resource ID: SCR_006016

Alternate IDs: SCR_006219, nlx_151406, nlx_151835

Alternate URLs: <http://purl.bioontology.org/ontology/HP>,
<http://compbio.charite.de/svn/hpo/trunk/src/ontology/human-phenotype-ontology.obo>

Record Creation Time: 20220129T080233+0000

Record Last Update: 20260805T044129+0000

Ratings and Alerts

No rating or validation information has been found for Human Phenotype Ontology.

No alerts have been found for Human Phenotype Ontology.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 73 mentions in open access literature.

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

Wang P, et al. (2025) The effect of ponacidin on CFA-induced chronic inflammatory pain and its mechanism based on network pharmacology and molecular docking. *Frontiers in medicine*, 12, 1510271.

Haffer N, et al. (2025) Genomics on FHIR - a feasibility study to support a National Strategy for Genomic Medicine. *NPJ genomic medicine*, 10(1), 57.

Godinez-Zamora GF, et al. (2025) Contribution of next generation sequencing to the diagnosis of inborn errors of immunity in a pediatric cohort. *Frontiers in immunology*, 16, 1638544.

Bhattacharya A, et al. (2025) Transcriptional pattern enriched for synaptic signaling is associated with shorter survival of patients with high-grade serous ovarian cancer. *eLife*, 13.

Coffey EL, et al. (2024) A Novel CARMIL2 Immunodeficiency Identified in a Subset of Cavalier King Charles Spaniels with Pneumocystis and Bordetella Pneumonia. *Journal of fungi (Basel, Switzerland)*, 10(3).

Groza T, et al. (2023) The International Mouse Phenotyping Consortium: comprehensive knockout phenotyping underpinning the study of human disease. *Nucleic acids research*, 51(D1), D1038.

Rosina E, et al. (2023) Comparison of first-tier whole-exome sequencing with a multi-step traditional approach for diagnosing paediatric outpatients: An Italian prospective study. *Molecular genetics & genomic medicine*, 12(1), e2316.

Sun Y, et al. (2023) MACdb: A curated knowledgebase for metabolic associations across human cancers. *Molecular cancer research : MCR*.

Nieminen M, et al. (2022) SODAR: managing multiomics study data and metadata. *GigaScience*, 12.

Crefcoeur LL, et al. (2022) Clinical characteristics of primary carnitine deficiency: A structured review using a case-by-case approach. *Journal of inherited metabolic disease*, 45(3), 386.

Fisher ME, et al. (2022) The *Xenopus* phenotype ontology: bridging model organism phenotype data to human health and development. *BMC bioinformatics*, 23(1), 99.

Schön U, et al. (2021) HPO-driven virtual gene panel: a new efficient approach in molecular autopsy of sudden unexplained death. *BMC medical genomics*, 14(1), 94.

Ren M, et al. (2020) Genotype-phenotype correlations of Berardinelli-Seip congenital lipodystrophy and novel candidate genes prediction. *Orphanet journal of rare diseases*, 15(1), 108.

Kerr M, et al. (2020) MITO-FIND: A study in 390 patients to determine a diagnostic strategy for mitochondrial disease. *Molecular genetics and metabolism*, 131(1-2), 66.

Yang L, et al. (2020) Novel and de novo point and large microdeletion mutation in PRRT2-related epilepsy. *Brain and behavior*, 10(5), e01597.

Pezzani L, et al. (2020) Double homozygosity in CEP57 and DYNC2H1 genes detected by WES: Composite or expanded phenotype? *Molecular genetics & genomic medicine*, 8(3), e1064.

Peterlin B, et al. (2020) Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study. *PloS one*, 15(9), e0239329.

Sun Q, et al. (2020) Whole-exome sequencing reveals two de novo variants in the RBM20 gene in two Chinese patients with left ventricular non-compaction cardiomyopathy. *Pediatric investigation*, 4(1), 11.

Liao Y, et al. (2019) WebGestalt 2019: gene set analysis toolkit with revamped UIs and APIs. *Nucleic acids research*, 47(W1), W199.

Köhler S, et al. (2019) Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic acids research*, 47(D1), D1018.