Resource Summary Report

Generated by RRID on May 16, 2025

Disease Phenotype Ontology

RRID:SCR_008687
Type: Tool

Proper Citation

Disease Phenotype Ontology (RRID:SCR_008687)

Resource Information

URL: http://human-phenotype-ontology.github.io/

Proper Citation: Disease Phenotype Ontology (RRID:SCR_008687)

Description: The Disease Ontology group has developed a set of standard representations of phenotypes associated with diseases useful in bioinformatics applications. These are formalized into an ontological structure and are encoded in OWL. Neurodegenerative diseases have a wide and complex range of biological and clinical symptoms. While neurodegenerative diseases share many pathological features in common, they also contain unique signatures. Animal models of these disorders are key to translational research. However, animal models typically replicate only a subset of disease features or display features that are only indirectly related to a given disorder, whose relationship to the human condition may be across several diseases. Matching animal models to human diseases is therefore a significant informatics challenge. We have been working to develop ontologies that capture essential features of neurodegenerative diseases and associated animal models in a way that allows more flexible matching of animal models to human disorders and in a way that makes explicit commonalities and differences among animal models and human neurodegenerative disease. Creating ontologies for diseases and disorders is a very challenging task (Gupta et al., 2003) because of the complexity of the disorders and because of the limitations of current ontology formalisms. In order to simplify the approach and make it practical for use in information systems, we have focused on formal descriptions of phenotypes associated with diseases and animal models rather than on a formal model of the disease process itself. We employ the modular ontologies developed as part of the Neuroscience Information Framework (NIF: http://nif.nih.gov) and the Phenotype and Trait Ontology (PATO), an ontology of qualities associated with biological phenotypes, to create a flexible template for creating phenotypic statements at the class and instance levels. We show how these phenotypes can be used to look for commonalities across multiple neurodegenerative conditions and animal models.

Synonyms: DPO

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

Funding:

Resource Name: Disease Phenotype Ontology

Resource ID: SCR_008687

Alternate IDs: nif-0000-35922

Record Creation Time: 20220129T080248+0000

Record Last Update: 20250516T053919+0000

Ratings and Alerts

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

No alerts have been found for Disease Phenotype Ontology.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 40 mentions in open access literature.

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

Halim-Fikri H, et al. (2024) Global Globin Network and adopting genomic variant database requirements for thalassemia. Database : the journal of biological databases and curation, 2024.

Schlüter A, et al. (2023) ClinPrior: an algorithm for diagnosis and novel gene discovery by network-based prioritization. Genome medicine, 15(1), 68.

Opladen T, et al. (2021) U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet journal of rare diseases, 16(1), 95.

Uliana V, et al. (2021) Deciphering the pathogenesis of the COL4-related hematuric nephritis: A genotype/phenotype study. Molecular genetics & genomic medicine, 9(2), e1576.

Liu Y, et al. (2021) Whole-Exome Sequencing in a Cohort of High Myopia Patients in

Northwest China. Frontiers in cell and developmental biology, 9, 645501.

Rodríguez-López R, et al. (2021) Immune Deficiency in Jacobsen Syndrome: Molecular and Phenotypic Characterization. Genes, 12(8).

Xu J, et al. (2020) Network pharmacology to dissect the mechanisms of Yinlai Decoction for pneumonia. BMC complementary medicine and therapies, 20(1), 168.

Zhou R, et al. (2020) Discovery of Herbal Pairs Containing Gastrodia elata Based on Data Mining and the Delphi Expert Questionnaire and Their Potential Effects on Stroke through Network Pharmacology. Evidence-based complementary and alternative medicine : eCAM, 2020, 4263591.

Deaton AM, et al. (2019) Rationalizing Secondary Pharmacology Screening Using Human Genetic and Pharmacological Evidence. Toxicological sciences : an official journal of the Society of Toxicology, 167(2), 593.

Zou J, et al. (2019) Autoinflammatory characteristics and short-term effects of delivering high-dose steroids to the surface of the intact endolymphatic sac and incus in refractory Ménière's disease. Journal of otology, 14(2), 40.

Huang L, et al. (2019) Genetic factors define CPO and CLO subtypes of nonsyndromicorofacial cleft. PLoS genetics, 15(10), e1008357.

Nguyen PA, et al. (2019) Phenotypes associated with genes encoding drug targets are predictive of clinical trial side effects. Nature communications, 10(1), 1579.

Gao C, et al. (2019) Diagnostic Yields of Trio-WES Accompanied by CNVseq for Rare Neurodevelopmental Disorders. Frontiers in genetics, 10, 485.

Xu HY, et al. (2019) ETCM: an encyclopaedia of traditional Chinese medicine. Nucleic acids research, 47(D1), D976.

Pagnamenta AT, et al. (2019) Delineation of dominant and recessive forms of LZTR1-associated Noonan syndrome. Clinical genetics, 95(6), 693.

Dong Z, et al. (2019) Genome Sequencing Explores Complexity of Chromosomal Abnormalities in Recurrent Miscarriage. American journal of human genetics, 105(6), 1102.

Naumova OY, et al. (2018) DNA methylation alterations in the genome of a toddler with cri-du-chat syndrome. Clinical case reports, 6(1), 14.

Barbato E, et al. (2018) Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. Archives of oral biology, 91, 96.

Vitali F, et al. (2018) ONS: an ontology for a standardized description of interventions and observational studies in nutrition. Genes & nutrition, 13, 12.

van Setten J, et al. (2018) PR interval genome-wide association meta-analysis identifies 50 loci associated with atrial and atrioventricular electrical activity. Nature communications, 9(1),

2904.