

Tutorial

The Human Disease Ontology (DO)



<http://www.disease-ontology.org/>

What is an ONTOLOGY ?

- Set of textual definitions and categories in a subject area or domain, including the properties and relations between the ontology terms.
- Organizes biomedical concepts to describe domains e.g. anatomy, environment, phenotype, taxonomy, protein, cell
- Explicitly defines concepts, relations, attributes and constraints.
- A knowledge network with nodes representing concepts and edges representing semantic relationships.
- Knowledge representation.

The project

- The DO was created to be a community resource in 2003 at Northwestern University. The DO team expanded in 2005-2008, with the addition of team members at the University of Maryland School of Medicine.
- DO's initial Wiki site: http://do-wiki.nubic.northwestern.edu/do-wiki/index.php/Main_Page
- The DO website was launched in 2008 with the website content updated periodically with DO's regular data releases.
- DO Releases are available in GitHub: <https://github.com/DiseaseOntology/HumanDiseaseOntology/releases>
- DOs standardized descriptions of human diseases facilitates the communication of health-related data across biomedical databases, bioinformatics tools, genomic and cancer resources.

Cross-references (Xrefs)

The Disease Ontology includes direct cross-references to other clinical vocabularies.

MESH - Medical Subject Headings



OMIM - Online Mendelian Inheritance in Man -



ICD9CM - International Classification of Diseases, 9th edition, Clinical Modification



ICD10CM - International Classification of Diseases, 10th edition, Clinical Modification

ICD-O - International Classification of Diseases for Oncology



NCI Thesaurus – NCI Reference Terminology

GARD - Genetic and Rare Diseases Information Center



Orphanet - Rare Diseases



SNOMED CT - Systematized Nomenclature of Medicine -- Clinical Terms