Tutorial
The Human Disease Ontology (DO)

http://www.disease-ontology.org/
What is an **ONTOLGY**?

- 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 DO 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