



## DO Buzz Newsletter September 2022

### Upcoming Meetings

#### [15th Annual Biocuration Conference](#)

Careers in Biocuration Workshop:

Sep 21

Session 3: Oct 4

Virtual

#### [ICBO 2022](#)

Sep 25-28

Virtual / In-person Hybrid

Ann Arbor, MI, USA

#### [US2TS: US Semantic Tech Symp](#)

Sep 29-Oct 1

Ann Arbor, MI, USA

### Latest News

#### ◇ *The Disease Ontology Project has received a new funding award!!!*

The Human Disease Ontology (DO) Knowledgebase (DO-KB) has been funded under a new NHGRI U24 5-year award of \$3,726,982 to provide a comprehensive disease feature similarity network for clinical differential diagnosis and biomedical data exploration.

The DO-KB diseaseome will: (1) aggregate disease information across genomic resources, model complex diseases and define the disease-environmental exposome; (2) establish federated disease/genomic/proteomic resource querying; (3) provide multi-lingual DO disease knowledge resources; (4) establish ML/AI ready disease mechanistic datasets; and (5) establish a clinical training nosology program for UMSOM medical, genetic counseling, dental, nursing and pharmacy trainees and residents.

#### ◇ Human Disease Ontology at a glance:

<b>Disease Classes</b>	<b>11,137</b>
Definitions	8,706 (78%)
Logical Axioms	7,934 subclassOf / 722 equivalentClass
Imports	15 sources / 4,467 classes
Cross-references	13 sources / 36,626 xrefs

Review the “Latest Release Notes” (p. 2) for a description of changes.

#### ◇ Web resource updates:

- A new [FAQ](#) has been added to describe the Disease Ontology’s relation to BFO & OGMS.
- The [Curation](#) page has been updated with a list of medical vocabulary browsers and a link to a list of [common logical axiom patterns](#) in the ontology.

### Citations & Use of the Disease Ontology

Scholarly works that cite or use the Disease Ontology are assessed quarterly by resource.

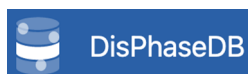
Source	Viewable list	Scholarly Works
Pubmed Cited By		834
PubMed (manually curated)	<a href="#">MyNCBI collection</a>	876
Scopus	<a href="#">Cited by list</a> (subscription required)	1,351
lens.org	<a href="#">Works Citing the Disease Ontology</a>	1,708

### New Community Resources

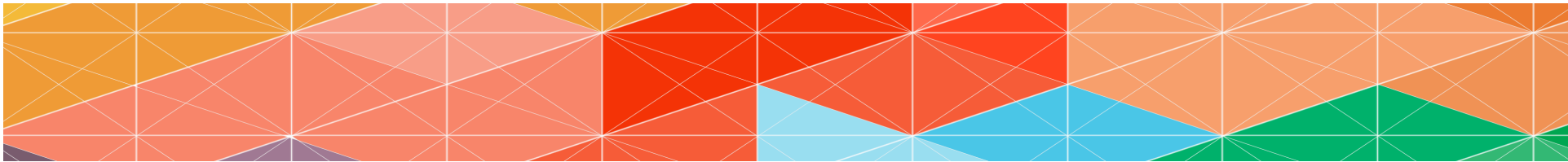
In this quarter (Mar-May), scholarly works described **11** new resources and methodologies designed for use by the biomedical community that used the Disease Ontology. A few are described below:



**iLncDA-LTR:** A Learning to Rank (LTR)-based method to predict diseases related with newly-found lncRNAs; doi: [10.1016/j.combiomed.2022.105605](https://doi.org/10.1016/j.combiomed.2022.105605), PMID: [35594681](https://pubmed.ncbi.nlm.nih.gov/35594681/).



**DisPhaseDB:** A database of mutations found in proteins involved in membrane-less organelles and liquid-liquid phase separation in human diseases; doi: [10.1016/j.csbj.2022.05.004](https://doi.org/10.1016/j.csbj.2022.05.004), PMID: [35685370](https://pubmed.ncbi.nlm.nih.gov/35685370/).

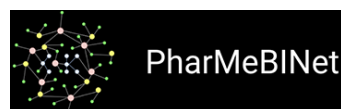


### New Community Resources (continued)



**DES System**

**DES-Amyloidoses:** A knowledgebase developed for exploring information related to human amyloid-related diseases; doi: [10.1371/journal.pone.0271737](https://doi.org/10.1371/journal.pone.0271737), PMID: [35877764](https://pubmed.ncbi.nlm.nih.gov/35877764/).



**PharMeBINet**

**PharMeBINet:** A pharmacological medical biochemical network with heterogeneous information on drugs, adverse drug reactions, genes, proteins, gene variants, and diseases as an open-source Neo4j database; doi: [10.1038/s41597-022-01510-3](https://doi.org/10.1038/s41597-022-01510-3), PMID: [35821017](https://pubmed.ncbi.nlm.nih.gov/35821017/).



**Heml 2.0**

**Heml 2.0:** An online service for heatmap illustration and functional enrichment analysis with diseases, genes and more; doi: [10.1093/nar/gkac480](https://doi.org/10.1093/nar/gkac480), PMID: [35670661](https://pubmed.ncbi.nlm.nih.gov/35670661/).

A thorough list of Ontologies, Resources and Methodologies that use the DO is available at <https://disease-ontology.org/community/use-cases>.

Disclaimer: Article spotlights and community resource lists highlight utilization of the Human Disease Ontology and are not an endorsement of any person(s), resource(s), method(s), or finding(s).

### Latest Release Notes

Data releases are available in DO's [GitHub repository](#) ([previous release notes](#))

#### Release #117: v2022-08-29

DO's August release includes revised and updated disease classifications, updated NCBITaxon logical definitions, and 11,137 disease terms with the addition of: injection anthrax, 'hypotonia, ataxia, and delayed development syndrome', 'short stature, hearing loss, retinitis pigmentosa, and distinctive facies', Leber congenital amaurosis 19, HMG-CoA synthase 2 deficiency and DeSanto-Shinawi syndrome. The revised classifications/added subtypes are for: autosomal recessive intellectual developmental disorder, acromesomelic dysplasia, dilated cardiomyopathy, common variable immunodeficiency, agammaglobulinemia, 3-methylglutaconic aciduria, tetrahydrobiopterin (BH4)-deficient hyperphenylalaninemia, Mandibuloacral dysplasia and arthrogyriposis.

#### Release #116: v2022-07-27

This release includes 11,033 DO disease terms, added textual definitions and over 200 new axioms. New disease terms and their subtypes include: hot water epilepsy, keratosis palmoplantaris striata, Baraitser-Winter syndrome, benign familial infantile seizures, Graves ophthalmopathy, 'inclusion body myopathy and brain white matter abnormalities', Catel Manzke syndrome, X-linked mental retardation Gustavson type, hyperthyroidism, nasal cavity benign neoplasm and spastic paraplegia with deafness. Definitions have been added for biliary tract benign neoplasms, biliary tract diseases and dental diseases. Imports have been revised and SYMP symptom ontology terms augmented. *This release also includes a new annotation property, 'term replaced by', which we will now add to obsolete terms to relate the obsoleted term to an active term that it was merged into.*

### DO on Social Media

- Check out the **Latest Video** on the Disease Ontology's [YouTube Channel](#):  
"Advanced searches of the DO website using relation axioms"
- *Are you aware of videos describing use of the DO?* Please let us [know!](#)  
Help augment the "[Applications using DO](#)" playlist.
- Videos are also available in "The Human Disease Ontology" playlists at:



[YouTube Channel](#)

Biomedical Ontology  
World [YouTube Channel](#)

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