



DO Buzz Newsletter - Nov. 2021

Upcoming Meetings

<u>Biomedical Informatics</u> <u>Applications in Rare Diseases</u> (<u>BIA-in-RD</u>) <u>Workshop</u> Dec 9-12, 2021 Virtual

...with more to come in 2022.

Latest News: Check out the latest data and website updates!

- ♦ The Human Disease Ontology now includes 10,901 disease terms (77% with text definitions), plus 710 equivalent and 7,321 subClassOf disease axioms.
- ♦ Term updates:
 - Integrated UMLS bi-annual update: new MESH, NCI, SNOMEDCT xrefs.
 - Added over 2,600 has_symptom axioms.
 - Added new disease terms for OMIM pontocerebellar hypoplasia phenotypic series, WHO's critical, severe and non-severe COVID-19 terms, and parsley allergy, dill allergy, and RNASET2-deficient cystic leukoencephalopathy.
 - Updated biotin deficiency, dextro-looped transposition of the great arteries, glycogen storage diseases, and classification for sexual health disorders and fibromyalgia.
 - Added HPO phenotype axioms.
- New slim!! The DO_CFDE_slim (44 terms) was created for the <u>NIH Common Fund Data</u> <u>Ecosystem (CFDE)</u> -- a digital cloud environment integrating data from projects funded by the NIH Common Fund.
 - To explore CFDE datasets by disease enter the Data Portal from the <u>CFDE home page</u> and select Data Browser > Vocabularies > Disease from the drop down menu.
 - Example projects include:



NEW PUBLICATION!!

"The Human Disease Ontology 2022 update", published in the Database Issue of Nucleic Acids Research (DOI: <u>10.1093/nar/gkab1063</u>, PubMed ID: <u>34755882</u>), describes updates to the DO and disease-ontology.org since 2018.

The article's graphical abstract (right) highlights:

- 1. The increasing breadth of features and etiological factors used to describe disease complexity via axioms.
- 2. The actual disease terms and relationhips added since 2018 (red in network graph).
- 3. The increase in DO's use, as measured by direct citation or indirect citation of databases that incorporate the DO.







Latest Release Notes:

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

Release #105: v2021-11-17 Release Notes

This release addresses two issues, an IAO ID substitution for the rdf:label, and the revision of has_symptom property.

Release #104: v2021-11-16 Release Notes

The DO's November 2021 release includes 10,901 disease terms, the addition of: 32 disease terms, a new slim file: DO_CFDE_slim with 44 DO terms, for the Common Fund Data Ecosystem project, the addition of: over 2,600 has_symptom axioms, DO terms for the OMIM pontocerebellar hypoplasia phenotypic series, and additional definitions and xrefs.

Release #103: v2021-10-11 Release Notes

An extra release in early October to update imports and push out 11 new disease terms added by MGI.

Spotlight: Recent Publications

- Gogate N, Lyman D, Bell A, et al. COVID-19 biomarkers and their overlap with comorbidities in a disease biomarker data model. Brief Bioinform. 2021 Nov 5;22(6):bbab191. doi: 10.1093/bib/bbab191. PMID: 34015823.
- Baltoumas FA, Zafeiropoulou S, Karatzas E, et al. OnTheFly2.0: a text-mining web application for automated biomedical entity recognition, document annotation, network and functional enrichment analysis. NAR Genom Bioinform. 2021 Oct 6;3(4):lqab090. doi: 10.1093/nargab/lgab090. PMID: 34632381.
- Ringwald M, Richardson JE, Baldarelli RM, et al. Mouse Genome Informatics (MGI): latest news from MGD and GXD. Mamm Genome. 2021 Oct 26. doi: 10.1007/s00335-021-09921-0. Epub ahead of print. PMID: 34698891.

Disease Ontology Citations:

The DO team has identified > 1400 DO project citations. Articles citing or using the DO are now identified from PubMed, by collecting 'cited in' lists and reviewing automated search results, and Scopus, by collecting 'cited by' lists. This collection represents the growing integration of DO into databases, research studies, and bioinformatics tools.

Individual articles can be found in the DO's <u>PubMed MyNCBI collection</u> or <u>Scopus cited by list</u> (subscription required).

Disease Ontology on Social Media

Checkout videos about the Disease Ontology on its YouTube Channel, now with a new playlist!!!



The Applications using DO playlist has been seeded with videos highlighting the Rat Genome Database's (RGD) **YouTube** Multi-Ontology Enrichment Tool (MOET) and an example infectious disease analysis.

Are you aware of other videos describing use of the DO? The DO team would love to add them. Please let us know.

Videos made by the DO team are also available in "The Human Disease Ontology" playlist on:



Follow the Disease Ontology on Twitter for use cases, disease-related information, and the latest updates. 💆 @diseaseontology