DISEASE ONTOLOGY

DO Buzz Newsletter
February 2022

Upcoming Meetings

Rare Disease Day at NIH
Feb 28
Virtual

Genomics of Rare Disease
Mar 28–29
Virtual

NLM Curation at Scale Workshop
Mar 28-30
Virtual

15th Annual Biocuration Conference
Session 1: Apr 7
Session 2: Jun 2
Session 3: Oct (TBD)
Virtual

ISMB 2022 / BOSC 2022 /
Bio-ontologies COSI
Jul 10-14
Virtual / In-person Hybrid
Madison, Wisconsin, USA

CGC Annual Meeting 2022
Jul 31-Aug 3
Virtual / In-person Hybrid
St. Louis, Missouri, USA

Latest News: Check out the latest data updates!

◊ Disease Term updates (this quarter):

- Added more than 300 ICD-O (cancer, morphology only) cross-refs.
- Added more than 350 new has_symptom axioms.
- Added lymphatic system diseases.
- **Revised classifications** for cranial nerve neoplasms, diabetes, anemia, glutathione synthetase deficiency, sexual dysfunction and amblyopia terms.

Year in Review: 2021

The focus in 2021 was on expanding the DO's expressivity of the mechanisms of complex diseases, reviewing and augmenting complex disease classifications, enhancing DO's disease etiology representation through the expansion of symptom, phenotype, age of onset, and mode of transmission axioms, and automating workflow processes for identifying mappings to ICD-O cancers and new terms added to other clinical vocabularies.

Disease Ontology DATA

	Total	Net Increase	
Terms	10,924	253	(+2.4%)
Definitions	9,012	359	(+4.1%)
Imports	16,287	630	(+4.0%)
Class axioms	8,308	2,057	(+29.6%)
Cross-refs	36,105	158	(+0.4%)
TOTAL	**80,636**	**3,457**	

disease-ontology.org
Google Analytics Usage

Top 10 Countries (rank / change from 2020)		
🇺🇸 United States	1	
🇷🇺 Russia	2	**+1**
🇨🇳 China	3	**-1**
🇩🇪 Germany	4	**+1**
🇮🇳 India	5	**-1**
🇬🇧 United Kingdom	6	**+1**
🇺🇦 Ukraine	7	**+4**
🇭🇰 Hong Kong	8	**+13**
🇮🇶 Iraq	9	**-1**
🇪🇸 Spain	10	
Total Users	35,854 (+6.3%)	
New countries	161 (+3.9%)	

Human Disease Ontology Github Repository

		2021 Change	Total to Date
Forks		n.a.	96
Stars		n.a.	252
Commits		n.a.	2,577
Issues	Created	74	945
	Closed	91	919
	Open	n.a.	26

Latest Release Notes:

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

Release #108: v2022-02-21 Release Notes
This release includes 10,924 disease terms, an increase of 23 disease and 69 definitions; additional ICD-O xrefs for leukemias, revised classifications of cranial nerve neoplasms, diabetes, anemia and [glutathione synthetase deficiency; addition of > 350 symptom axioms.

Release #107: v2022-01-31 Release Notes
DO's January 2022 release includes 10,902 disease terms with 77% (8,419 defined). We have added over 300 ICDO xrefs, new has_symptom axioms, revised sexual dysfunction and amblyopia terms and added lymphatic system diseases.

Release #106: v2021-12-15 Release Notes
DO's December 2021 release includes the addition of has_symptom subClassOf axioms and term definitions.

DO Citations & Use

Publications Citing the DO

	Total to Date	Last year
Article	1,114	147
Conference Paper	212	21
Review	111	17
Book (Chapter)	42	1
Commentary*	17	2
Clinical Trial	1	n.a.
TOTAL	**1,497**	**188**

* includes letters, editorials, & historical articles.

For a list of articles see the DO's [PubMed MyNCBI collection](#) or [Scopus cited by list](#) (subscription required).

Quarterly Publication Spotlight

- Gillespie M, et al. **The reactome pathway knowledgebase 2022.** Nucleic Acids Res. 2022 Jan 7;50(D1):D687-D692. doi: [10.1093/nar/gkab1028](#). PMID: [34788843](#).

- Le DH. **A network-based method for predicting disease-associated enhancers.** PLoS One. 2021 Dec 8;16(12):e0260432. doi: [10.1371/journal.pone.0260432](#). PMID: [34879086](#).

- Wang CC, et al. **A machine learning-driven approach for prioritizing food contact chemicals of carcinogenic concern based on complementary in silico methods.** Food Chem Toxicol. 2022 Feb;160:112802. doi: [10.1016/j.fct.2021.112802](#). PMID: [34979167](#).

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

DO on Social Media

Disease Ontology Channel (2021)

Watch time	1,362 minutes
Total views	648
Most viewed video	"What is an ontology?"
Top countries for channel views	🇺🇸 United States
	🇮🇳 India
	🇨🇦 Canada

- 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" playlist at:

ISB International Society for Biocuration [YouTube Channel](#)

Biomedical Ontology World [YouTube Channel](#)

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