DO Buzz Newsletter

February 2023

Latest News

Our team grows, conferences and more!

- Claudia Sánchez-Beato Johnson joined the DO team in January, 2023. Claudia is from Madrid, Spain, bringing a unique international biomedical background and vision to the team. She obtained her Bachelor's degree in Kinesiology & Health Sciences at The College of William & Mary where she did Molecular & Cardiovascular Physiology research and later earned her Master's degree in Systems Medicine from Georgetown University.
- DO team members attended BIBM 2022 in Las Vegas (Dec 6-7, 2022), where at least 8 machine learning projects that directly use the Disease Ontology were described. Two additional projects of interest included the work of NCATS researchers to identify high reward areas for future rare disease research, building off of a 2020 publication that collected rare disease research and funding information, and the phenome-wide association study (PheWAS) work of keynote speaker Marylyn Ritchie.
- Are you attending ISB Biocuration?

  Meet the DO team at ISB Biocuration and learn about our work. Check out the DO talks and posters:
  - Assessing Resource Use: A Case Study with the Human Disease Ontology
  - A 20 year perspective on FAIR and TRUST-worthy Human Disease Knowledge Representation.
  - Join us at the DO co-hosted ISB Biocuration workshop, Monday, April 24th: Gaining perspective towards enhancing the intersection of biocuration and machine learning
- We are working on expanding our representation of polygenic and digenic diseases, keep an eye out for updates on our latest release!

Upcoming Meetings

The 2023 Ontology Summit
Every Wednesday
Jan 18, 2023 - May 3, 2023
Virtual

Rare Disease Day at NIH
Feb 28, 2023
Virtual / In-person Hybrid
NIH Main Campus (Natcher Conference Center)

16th Annual ISB Biocuration Conference
April 24-26, 2023
In-person
Padova, Italy

ISMB Bio-ontology and BOSC 2023
In-person
July 23-27, 2023

ICBO 2023
Aug 28-Sept 1st, 2023
In-person
Brasilia, DF Brazil

Human Disease Ontology at a glance:

Disease Classes	**11,207**
Definitions	8,820 (78.70 %)
Logical Axioms	8,076 subClassOf/ 725 equivalentClass
Imports	15 sources / 4,439 classes
Cross-references	14 sources / 37,269 xrefs

Web resource Update

◊ **OLIDA** is a curated database of oligogenic diseases and the variants in genes that have been published as causing these diseases. The combinations of variants that are contained in this database have been identified by researchers as being the cause of certain genetic diseases

- Nachtegael C. and Gravel B., Dillen A., Smits G., Nowé A., Papadimitriou S., Lenaerts T. Scaling up oligogenic diseases research with OLIDA: the Oligogenic Diseases Database. Database, April 2022. doi: 10.1093/database/baac023

Citations & New Community Resources Using the Disease Ontology

To date, 1,752 works citing the Disease Ontology are listed at lens.org. For example:

◊ Identification of the diagnostic genes and immune cell infiltration characteristics of gastric cancer using bioinformatics analysis and machine learning. doi: 10.3389/fgene.2022.1067524, PMID: 36685898.

◊ e-TSN: an interactive visual exploration platform for target-disease knowledge mapping from literature. doi: 10.1093/bib/bbac465, PMID: 36347537

◊ The Pharmacorank Search Tool for the Retrieval of Prioritized Protein Drug Targets and Drug Repositioning Candidates According to Selected Diseases. doi: 10.3390/biom12111559, PMID: 36358909

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 #123: v2023-01-30
This release includes 11,207 disease terms, with 8,820 (78.7%) textual definitions. 26 new disease terms, 76 new definitions. This release includes a new DO_infectious_disease_slim; updated xrefs from our bi-annual UMLS update, including all SNOMED_CT xrefs to the latest release; the annotation of trinucleotide repeat expansion disorders, digenic and polygenic disease annotations, a revised VEXAS syndrome, the addition of bronchopulmonary dysplasia and intellectual developmental disorder with ocular anomalies and distinctive facial features, numerous CNS neoplasm terms and their associated ICDO codes.

Release #122 v2022-12-15
This release includes 11,181 disease terms and the addition of ICDO codes for CNS cancers and breast cancers, 36 additional textual definitions and an updated disease name for monkeypox, now mpox.

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:

For the latest updates and other information, follow the Disease Ontology on **Twitter**!
@diseaseontology

ISB International Society for Biocuration YouTube Channel

Biomedical Ontology World YouTube Channel