



DO Buzz Newsletter June 2022

Upcoming Meetings

[15th Annual Biocuration Conference](#)

Careers in Biocuration Workshop:

Sep 21

Session 3: Oct (TBD)

Virtual

[ISMB 2022](#) / [CAMDA](#) / [BOSC](#) / [Bio-ontologies](#)

Jul 10-14

Virtual / In-person Hybrid
Madison, Wisconsin, USA

- *Dr. Schriml will be a keynote speaker at CAMDA*

[CGC Annual Meeting 2022](#)

Jul 31-Aug 3

Virtual / In-person Hybrid
St. Louis, Missouri, USA

Latest News: Check out the latest data and web resource updates!

◇ Human Disease Ontology at a glance:

Disease Classes	11,003
Definitions	8,547 (78%)
Logical Axioms	7,755 subClassOf / 722 equivalentClass
Imports	15 sources / 4,230 classes
Cross-references	13 sources / 36,519 xrefs

◇ Additional data updates:

- *Spring cleaning has begun!* Broken definition source URLs are now being replaced (~ 200 so far).

◇ Web resource updates:

- DO's persistent URLs ([PURLs](#)) for *individual DO terms* now redirect to [disease-ontology.org](#).
 - This will enrich the experience for resources that incorporate DO and their end-users by providing additional, easier to read information about diseases.
 - *Try it!* DICER1 syndrome = http://purl.obolibrary.org/obo/DOID_0081063
- [disease-ontology.org](#)'s "Ontologies and Resources Using DO" have been moved from the Collaborators page to the new [Use Cases](#) page, where they've been grouped by type to facilitate review.
 - All of these resources were built with Disease Ontology data and designed for reuse by the scientific community. *Check out the page for more details!*
- [disease-ontology.org](#)'s advanced search field "DOID" has been renamed to "ID" to reflect its search of the entire DO ID-space including imports. *Refer to the [tweetorial](#) for details.*

◇ Revised classifications

- spermatogenic failure
 - hereditary spastic paraplegia
 - Opitz GBB/Teebi Hypertelorism
 - diabetes DM1
 - intellectual disability
- ◇ Review the "Latest Release Notes" (p. 2) for other disease additions.

Citations & Use of the Disease Ontology

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

- ◇ **1,551** scholarly works total (**76** published in 2022) have cited or used the Disease Ontology, according to PubMed (view DO's [MyNCBI collection](#)) and Scopus (view DO's [cited by list](#); subscription required).
- ◇ The DO team continues to review other sources and has now created a new, continuously updated collection at [lens.org](#) of "[Works Citing the Disease Ontology](#)" which includes 1,657 scholarly works, the largest number identified by a single resource thus far and more than PubMed and Scopus combined!



New Community Resources

Last quarter (Dec 2021 - Feb 2022), scholarly works described **17** new resources and methodologies designed for use by the biomedical community that used the Disease Ontology. In this quarter (Mar-May), **3** more were described:

- **Darling:** A web application for detecting disease-related biomedical entity associations with literature mining; doi: [10.3390/biom12040520](https://doi.org/10.3390/biom12040520), PMID: [35454109](https://pubmed.ncbi.nlm.nih.gov/35454109/).
- **DLMPM:** A disease and literature driven metabolism prediction model to identify the potential associations between metabolites and diseases; doi: [10.1186/s12864-022-08504-w](https://doi.org/10.1186/s12864-022-08504-w), PMID: [35387615](https://pubmed.ncbi.nlm.nih.gov/35387615/).
- **RGCNCDA:** A computational method using relational graph convolutional networks (R-GCNs) to predict potential circRNA-disease associations; doi: [10.1016/j.combiomed.2022.105322](https://doi.org/10.1016/j.combiomed.2022.105322).



Quarterly Primary Research Spotlight

- Chen Q, Chou WC, Lin Z. **Integration of Toxicogenomics and Physiologically Based Pharmacokinetic Modeling in Human Health Risk Assessment of Perfluorooctane Sulfonate.** Environ Sci Technol. 2022 Mar 15;56(6):3623-3633. doi: [10.1021/acs.est.1c06479](https://doi.org/10.1021/acs.est.1c06479). PMID: [35194992](https://pubmed.ncbi.nlm.nih.gov/35194992/).
- Martín Pérez-Pérez, et al. **A deep learning relation extraction approach to support a biomedical semi-automatic curation task: The case of the gluten bibliome.** Expert Systems with Applications, 2022;195:116616, doi: [10.1016/j.eswa.2022.116616](https://doi.org/10.1016/j.eswa.2022.116616).
- Stefanou IK, et al. **miRNAs expression pattern and machine learning models elucidate risk for gastric GIST.** Cancer Biomark. 2022;33(2):237-247. doi: [10.3233/CBM-210173](https://doi.org/10.3233/CBM-210173). PMID: [35213356](https://pubmed.ncbi.nlm.nih.gov/35213356/).
- Zhang F, et al. **Screening of Genes Related to Breast Cancer Prognosis Based on the DO-UniBIC Method.** Am J Med Sci. 2022 Apr 23:S0002-9629(22)00187-2. doi: [10.1016/j.amjms.2022.04.022](https://doi.org/10.1016/j.amjms.2022.04.022). PMID: [35472336](https://pubmed.ncbi.nlm.nih.gov/35472336/). Epub ahead of print.

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 #114: v2022-06-06

This release includes a revised diabetes DM1 classification and revision of synonyms for two DO terms.

Release #113: v2022-06-02

This release contains 11,003 disease terms and new definitions, xrefs, uberon anatomy terms, and has symptom axioms. Among the disease terms added were: leukemia molecular subtypes & their ICD-O codes; ectodermal dysplasia, immune deficiency, blastic plasmacytoid dendritic cell neoplasm, Marsili syndrome, and Teebi hypertelorism syndrome. The Opitz GBBB syndrome and intellectual disability classifications were revised.

Release #112: v2022-04-28

This release includes 10,960 human diseases with the addition of diffuse large B-cell lymphoma molecular and spondylocostal dysostosis subtypes, syndrome definitions, updated definition source URLs and new symptom SubClassOf axioms.

Release #111: v2022-04-01

This release includes 10,948 DO disease terms, 77% with textual definitions; new cancer syndrome synonyms, ICDO xrefs, DLBCL disease subtypes, DICER1 syndrome and symptom SubClassOf statements have been added.

Release #110: v2022-03-02

An extra release to support MGI, including new DO terms Tatton-Brown-Rahman syndrome, craniotubular dysplasia Ikegawa type, updated series for spermatogenic failure and hereditary spastic paraplegia.

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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