



Upcoming Conferences

May 17-20, 2020: Biocuration 2020 in Bar Harbor, Maine

September 16-19, 2020: ICBO 2020 in Bozen-Bolzano, Italy

Recent Conference Presentation

November, 2019: Open Science Symposium, Carnegie Mellon University, Pittsburgh, PA. Open Genomic Metadata Tools & Data

Latest Release Notes:

DO Data Release: Available in DO's [GitHub repository](#): ([previous release notes](#))

Release # 67: December 12, 2019 Release Notes

In the DO November 2019 release: Includes 9,690 DOIDs with 69% of DO terms defined. This release includes the addition over >200 more definition source types from Evidence and Conclusion Ontology, updates to the sources for definitions, and replacing lost `located_in` axioms for two branches. Early and late onset symptoms were also added to Alzheimer subtypes.

Latest News: Happy Holidays from the Disease Ontology Team! 🎄

Upcoming changes to the website's OWL tree coming January:, the OBO tree in the DO website will be generated from the `doid.owl` file and will thus include both asserted and inferred disease parents. We hope that this will achieve a more synonymous relationship between our OWL and OBO files.

DO Spotlight of the Quarter: Recent DO Use-Cases 2019

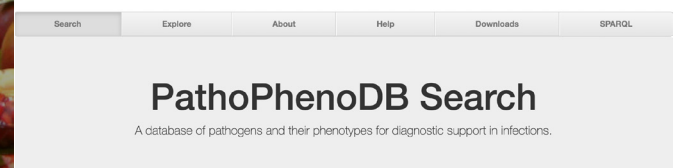
The Disease Ontology is being used in wider applications – this quarter, two new publications have been released detailing DO use cases. FoodOn is a harmonized food ontology aiming to increase global food traceability, quality control, and data integration. A consortium-driven project ontology about food that addresses food product terminology gaps, FoodOn focuses on human and domesticated animal food description. FoodOn has a “has food substance analog” relation that connects any two food source items or products. For food source terms related to allergic hypersensitivity diseases, FoodOn uses DO as a reference.

PathoPhenoDB is a database containing pathogen-to-phenotype associations and adheres to the FAIR data principles. PathoPhenoDB created their pathogen-to-phenotype association by finding at least one possible human pathogen for every infectious disease class in the DO. As DO expands with future releases, PathoPhenoDB intends to maintain coverage of new infectious disease classes. More diseases were manually gathered from CDC, Wikipedia, and literature in PubMed. Lexical matching was used to map the additionally gathered diseases to DO.

Links to mentioned publications:

FoodOn: <https://www.ncbi.nlm.nih.gov/pubmed/31304272>

PathoPhenoDB: <https://www.ncbi.nlm.nih.gov/pubmed/?term=pathophenodb>



Citations:

Dooley, D. M., Griffiths, E. J., Gosal, G. S., Buttigieg, P. L., Hoehndorf, R., Lange, M. C., ... & Hsiao, W. W. (2018). FoodOn: a harmonized food ontology to increase global food traceability, quality control and data integration. *npj Science of Food*, 2(1), 23.

Kafkas, Ş., Abdelhakim, M., Hashish, Y., Kulmanov, M., Abdellatif, M., Schofield, P. N., & Hoehndorf, R. (2019). PathoPhenoDB, linking human pathogens to their phenotypes in support of infectious disease research. *Scientific data*, 6(1), 79.



Disease Ontology's New [YouTube Video!](#)

In this video tutorial we look at the basic and advanced search functions that allow one to explore the Human Disease Ontology web interface. We discuss the metadata associated with ontology terms and compare and contrast the two options for graphically viewing the ontology terms (OWL & OBO).

Explore the Human Disease Ontology Project information & resources

The screenshot shows the Disease Ontology website with a teal header. The navigation menu includes: Home, About, Community, Tools & Resources, Outreach, Curation, Downloads, and social media icons for Facebook, Twitter, and LinkedIn. Below the header, there are four main dropdown menus:

- About:** The Disease Ontology Project, DO Team, Scientific Advisory Board, Release Notes on GitHub, How is DO FAIR, Statistics, DO's Logo, FAQ.
- Community:** Community calendar, Publications, Collaborators, Newsletter.
- Tools & Resources:** DO Resources, Related Resources, Citing DO.
- Outreach:** Contact Us, Tutorial, Slideshare.
- Curation:** Curation Tools and Documentation, DO SPARQL queries, ROBOT, DO Style Guide, Design Patterns.
- Downloads:** GitHub, OBO Foundry, DO Releases, Imports.

The main content area features the Disease Ontology logo and a paragraph of text: "Disease Ontology has been developed as a standardized ontology for human disease with the purpose of providing the biomedical community with consistent, reusable and sustainable descriptions of human disease terms, phenotype characteristics and related medical vocabulary disease concepts through collaborative efforts of researchers at Northwestern University, Center for Genetic Medicine and the University of Maryland School of Medicine, Institute for Genome Sciences. The Disease Ontology semantically integrates disease and medical vocabularies through extensive cross mapping of DO terms to MeSH, ICD, NCI's thesaurus, SNOMED and OMIM. To get started please visit the [tutorial page](#)."

Disease Ontology Citations:

The DO team has identified a body of 602 DO project citations (as of December 2019). This set of citations has been compiled as a public [PubMed MyNCBI](#) collection (DO citing papers). This MYNCBI collection represents the growing number of instances of integration of DO in databases, research studies, and bioinformatics tools. The DO Citations are identified through PubMed data mining (direct DO paper citations, inclusions of 'Disease Ontology', DO URL or DOID).

Publications:

[Human Disease Ontology 2018 update: classification, content and workflow expansion.](#)

Schriml LM, Mitraka E, Munro J, Tauber B, Schor M, Nickel L, Felix V, Jeng L, Bearer C, Lichenstein R, Bisordi K, Campion N, Hyman B, Kurland D, Oates CP, Kibbey S, Sreekumar P, Le C, Giglio M, Greene C. *Nucleic Acids Res.* 2019 Jan 8;47(D1):D955-D962. doi: 10.1093/nar/gky1032.

PDF: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6323977/pdf/gky1032.pdf>

[ECO, the Evidence & Conclusion Ontology: community standard for evidence information.](#)

Giglio M, Tauber R, Nadendla S, Munro J, Olley D, Ball S, Mitraka E, Schriml LM, Gaudet P, Hobbs ET, Erill I, Siegele DA, Hu JC, Mungall C, Chibucos MC.

Nucleic Acids Res. 2019 Jan 8;47(D1):D1186-D1194. doi: 10.1093/nar/gky1036.

PDF: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6323956/pdf/gky1036.pdf>