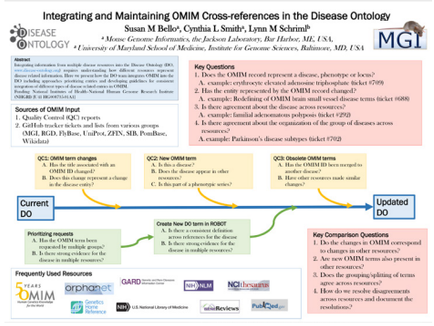
DISEASE ONTOLOGY

DO Buzz Newsletter August 2019

Upcoming Conferences

(July 9 - August 2, 2019) Buffalo, New York
International Conference on Biomedical Ontologies
Poster Presentation by Susan M Bello:
"Integrating and Maintaining OMIM Cross-references in the Disease Ontology"



Latest News:

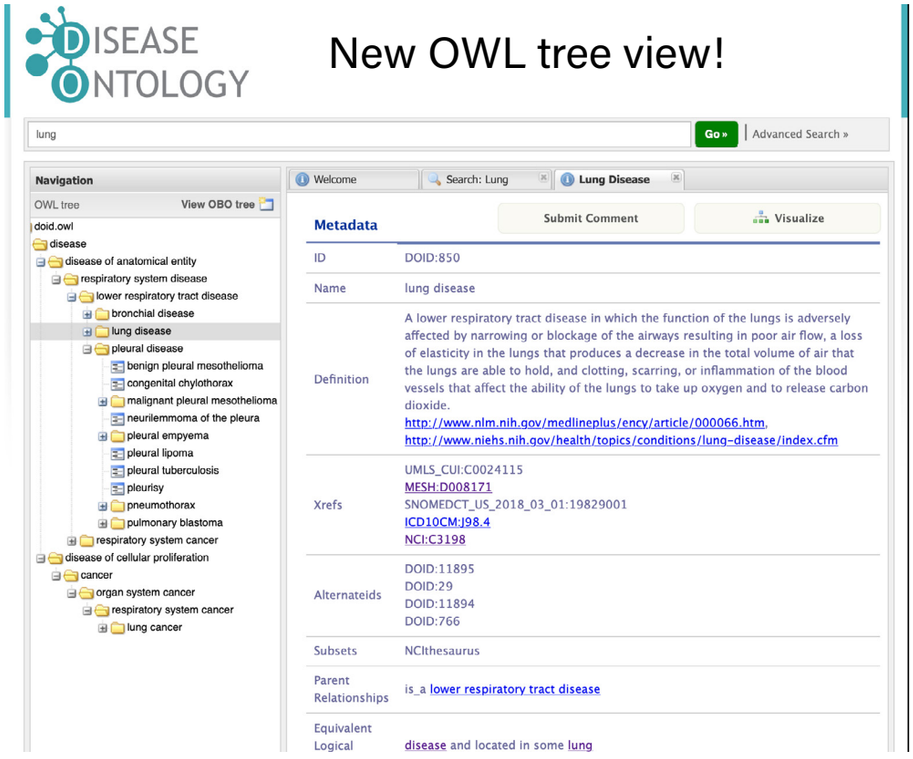
DO has completed the sarcoma branch review and revision, guided by Dr. Radoslav Davidovic at the Center for Multidisciplinary Research, Institute of Nuclear Sciences VINA, Belgrade, Serbia.

Melody Swen joins the Human Disease Ontology Team.
Melody joined the DO team in June, 2019 as an Ontology Analyst. She comes to us from the Wendy Klag Center for Autism and Developmental Disabilities in the Department of Mental Health, Johns Hopkins Bloomberg School of Public Health.

DO Spotlight of the Quarter: DO Website Relaunch with Added Accessibilities and Tree
The DO website has been updated with new features, including new Community, Curation, Documentation, and Outreach pages. Site usage statistics will be posted to the DO website along with quarterly release notes.

A new tree view has been added to the DO website. The alternative tree view, based on the doid.owl file, will enable searches that include any of the DO imported files (e.g. cell types, anatomy, inheritance). For example, to retrieve diseases that are located in the same body site (anatomical location), or to retrieve all diseases that share a common mode of inheritance (e.g. autosomal recessive inheritance).

Users can now submit searches against either the OBO tree (current public view) or the OWL tree (by clicking the toggle button (top of tree box)).



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Latest Release Notes:

DO Data Release: Available in DO's GitHub repository: (previous release notes)
Release # 61:
July 15, 2019 Release Notes:
The DO's July 2019 release includes a revised classification for early-onset & late onset Parkinson's disease (coordinated with MGI and OMIM), the addition of new OMIM subtypes and term definitions. E.g. Angelman syndrome, leukocyte adhesion deficiency diseases, Boucher-Neuhauser syndrome, geroderma osteodysplasticum, hyaline body myopathy, autosomal recessive hyaline body myopathy, autosomal dominant hyaline body myopathy, isolated sulfite oxidase deficiency and Olive-McFarlane syndrome. This release includes 9,384 DO classes (67% defined: 6372/9384).

Recent Conference Presentation

(April 2-6, 2019) Seattle, Washington
Annual Clinical Genetics Meeting (ACMG)
Poster presentation by Katharine Bisordi, MS, MGC, DO Clinician Team, UMSOM Genetic Counselor: "What the Disease Ontology (DO) Can DO to Improve Communication across Health-Related Datasets"
(April 7-10, 2019) Cambridge, UK
12th International Biocuration Conference
▪ April 10th talk: "Expanding the MIxS Genomic Minimal Information Standards" (Lynn Schriml) ▪ Two Posters:
▪ "DO: The FAIR human disease ontology domain standard" (Lynn Schriml)
▪ "Structured Design Patterns in the Human Disease Ontology for Enhanced Genetic Disease Classification" (Michael Sinclair)

Disease Ontology Citations:

The DO team has identified a body of 351 DO project citations (as of July 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, inclusion 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