



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.

Upcoming Conferences

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

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)).

Integrating and Maintaining OMIM Cross-references in the Disease Ontology
 Susan M Bello¹, Cynthia L Smith¹, Lynn M Schrimp²
¹ *Metastatic Genomic Informatics, The Jackson Laboratory, Bar Harbor, ME, USA*
² *University of Maryland School of Medicine, Institute for Genome Sciences and Policy, Baltimore, MD, USA*

Abstract
 Ontologies constructed from multiple disease resources into the Disease Ontology (DO) have been used to support clinical research, but OMIM cross-references (OMIM) have not been included. Here we present the DO team's approach to OMIM and the DO website updates to include OMIM cross-references and to support the integration of OMIM cross-references into the DO.

Key Questions

1. Does the OMIM record represent a disease, phenotype or locus?
2. Has the entity represented by the OMIM record changed?
3. Is there agreement about the disease access resources?
4. How do we resolve disagreements across resources and document the resolutions?

Sources of OMIM Input

1. Quality Control (QC) reports
2. Call-back notes and files from various groups (MGI, RGD, FlyBase, UniProt, ZFIN, SIB, Pombase, Wikidata)

QC: OMIM term changes

- A. Has the title associated with an OMIM ID changed?
- B. Does this change represent a change in the disease name?

QC: New OMIM term

- A. Is this disease?
- B. Does this disease appear in other resources?
- C. Is this part of a phenotype series?

QC: Disease OMIM term

- A. Has the OMIM ID been re-used in another disease?
- B. Has another resource made similar changes?

Key Comparison Questions

1. Do the changes to OMIM correspond to changes in other resources?
2. Are new OMIM terms also present in other resources?
3. Does the grouping/labeling of terms agree across resources?
4. How do we resolve disagreements across resources and document the resolutions?

Diagram: A flowchart showing the process from 'Current DO' to 'Updated DO'. It includes steps for 'Prioritize requests' (based on multiple groups, multiple groups, or disease in multiple resources), 'Create New DO term in RGD/OBO', and 'Key Comparison Questions'.

Frequently Used Resources: 5MIM, Ensembl, GARD, UniProt, NCI Thesaurus, PubMed, etc.



New OWL tree view!

lung [Advanced Search »](#)

Welcome Search: Lung **Lung Disease**

Navigation

OWL tree View OBO tree

doid.owl

- disease
 - disease of anatomical entity
 - respiratory system disease
 - lower respiratory tract disease
 - bronchial disease
 - lung disease
 - pleural disease
 - benign pleural mesothelioma
 - congenital chylothorax
 - malignant pleural mesothelioma
 - neurilemmoma of the pleura
 - pleural empyema
 - pleural lipoma
 - pleural tuberculosis
 - pleurisy
 - pneumothorax
 - pulmonary blastoma
 - respiratory system cancer
 - disease of cellular proliferation
 - cancer
 - organ system cancer
 - respiratory system cancer
 - lung cancer

Metadata

ID	DOID:850
Name	lung disease
Definition	A lower respiratory tract disease in which the function of the lungs is adversely affected by narrowing or blockage of the airways resulting in poor air flow, a loss of elasticity in the lungs that produces a decrease in the total volume of air that the lungs are able to hold, and clotting, scarring, or inflammation of the blood vessels that affect the ability of the lungs to take up oxygen and to release carbon dioxide. http://www.nlm.nih.gov/medlineplus/ency/article/000066.htm http://www.nlm.nih.gov/health/topics/conditions/lung-disease/index.cfm
Xrefs	UMLS_CUI:C0024115 MESH:D008171 SNOMEDCT_US_2018_03_01:19829001 ICD10CM:I98.4 NCI:C3198
Alternate IDs	DOID:11895 DOID:29 DOID:11894 DOID:766
Subsets	NCItHesaurus
Parent Relationships	is_a lower respiratory tract disease
Equivalent Logical	disease and located in some lung



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>