



The Human Disease Ontology project's 1<sup>st</sup> Quarterly newsletter, sharing highlights with our user community.

Buzz Newsletter October 2018

## Upcoming Conferences and Workshops

**(October 15-16, 2018)**

**ASHG conference (BioHackathon)**

**2018 CIViC Hackathon and Curation Jamboree (pre-ASHG), Scripps, La Jolla, CA**

The focus of this meeting is to continue to develop community consensus on data standards for interpretation of variants in cancer, engage software developers for improved variant interpretation software, engage clinical experts for curation of content and enhance integration and interoperability of resources.

**(October 16-20, 2018)**

**American Society of Human Genetics (ASHG), San Diego, CA**

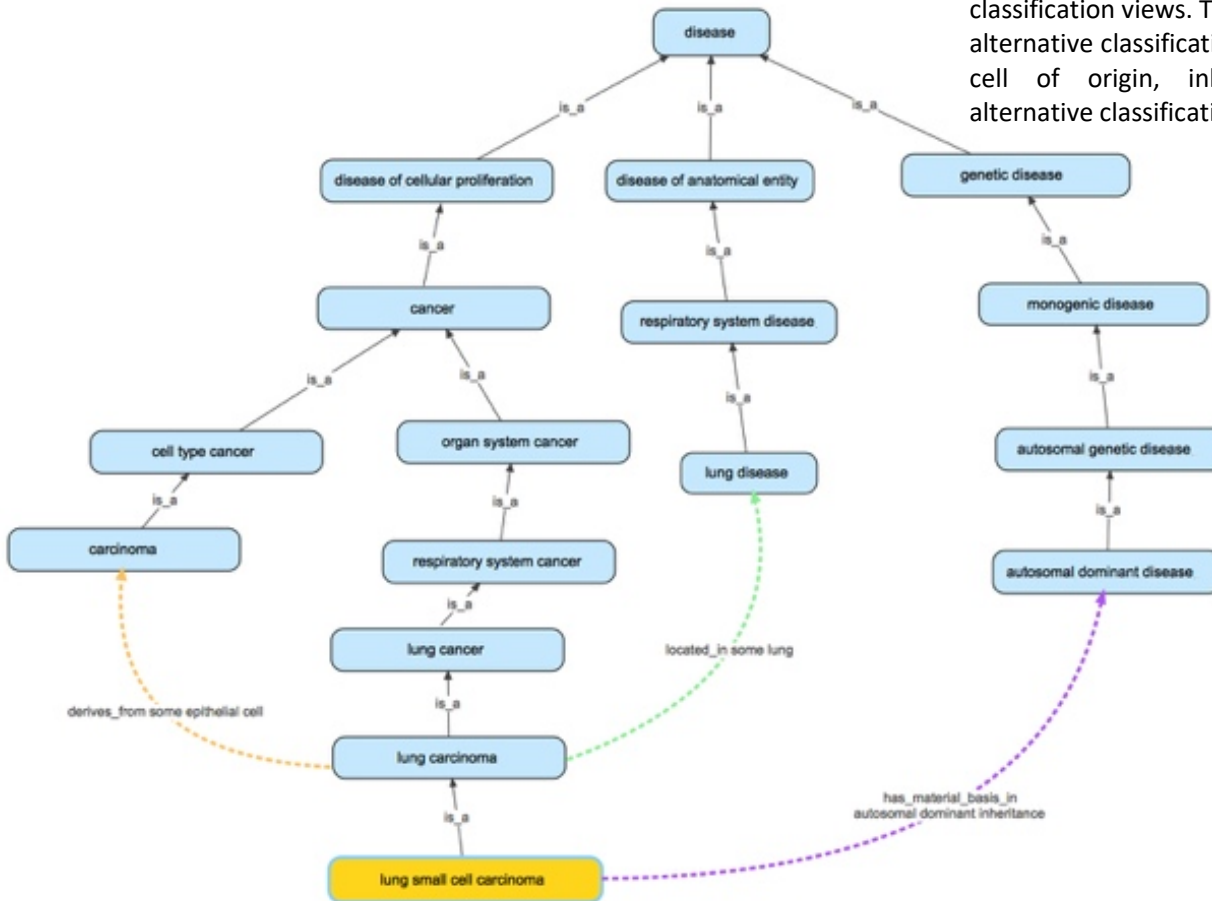
**Poster Sessions: 2:00 pm, October 17, 2018**

- Precision etiology for complex disease classification. (Lynn Schriml) Poster # 1413/W ([abstract](#))
- Wikidata for biomedical knowledge integration and curation. (Greg Stupp) Poster # 1599/W ([abstract](#))
- New features facilitating search and analysis of models of human disease. Mouse Genome Informatics and The Alliance of Genome Resources.(Cynthia Smith) Poster # 1595/F ([abstract](#))



## DO Spotlight of the Quarter:

**DO - OWL view** -- By defining inferred parent relationships, the DO redefines etiology complexity and enables alternative disease classification views. The DO has built an anatomy alternative classification and is currently building cell of origin, inheritance and syndrome alternative classifications.



## Latest Release Notes

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

Release Cycle: every two weeks

### October 5, 2018 Release Notes

DOID term count: 9,092 disease terms, 5682/9092 terms defined (62%).

In doid.owl file: 940 Equivalent To axioms and 4,071 SubClass Of statements.

Added new human disease terms from MGI: retinitis pigmentosa 77, CLOVES syndrome, early infantile epileptic encephalopathy 39. Defining additional cell of origin axioms for cell type cancer.

### September 2018 Release Notes

DOID term count : 9,079 disease terms, with 62% (5,655/9,079) of DO terms having textual definitions.

Additions: 833 textual definitions produced from the DO UMSOM medical students, 1491 GARD rare disease xrefs and 665 MeSH xrefs identified by Wikidata users and validated by the DO team. Equivalent To axioms populating inferred genetic disease, monogenic disease, autosomal dominant disease, cell type cancer parent to child relationships, with 730 Equivalent To axioms and 3612 SubClass Of statements. Creation of complex disease logical definitions, e.g. Prader-Willi syndrome.

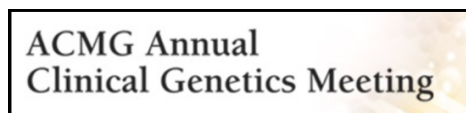
## Outreach Highlights



ICBO 2018 (August 2018) Oregon State University, Corvallis, Oregon  
The Disease Ontology team organized the Big-Data Disease Workshop: Biomedical Health Knowledge Integration and Dissemination: Human Disease Ontology (Lynn Schriml, Michelle Giglio, Becky Tauber). <http://icbo2018.cgrb.oregonstate.edu>



11<sup>th</sup> International Biocuration Conference (April 2018)  
Fudan University, Shanghai, China. The Wikidata - Human Disease Ontology team (Greg Stupp) presented: Wikidata Crowdsourcing Model for Enhancing DO Curation  
<http://biocuration2018.cn/biocuration2018/>



Annual Clinical Genetics Meeting (April 2018)  
Charlotte, NC. Poster: Disease Ontology: Improvements for Clinical Care and Research Applications (Linda Jeng, M.D., Ph.D.)  
<https://www.acmgmeeting.net/acmg2018/>



Cancer Biomarkers Data Commons Think-Tank meeting (February 2018)  
National Cancer Institute, a talk was presented on: Cancer, Ontologies, and Data Integration. (Lynn Schriml)  
<https://nciphub.org/projects/cbdcworkshop/info>

## [Disease Ontology Citations](#)

The DO team has identified a body of **336 DO project citations** (as of August 2018), an increase from ~50 citations in 2015. This set of citations has been compiled as a public PubMed **MyNCBI collection (DO\_citing\_papers)**:

<https://www.ncbi.nlm.nih.gov/sites/myncbi/lynn.schriml.1/collections/49204559/public/>.

This MyNCBI collection represents the growing number of instances of integration of DO in databases, research studies, bioinformatics tools. The DO Citations, identified through PubMed data mining (direct DO paper citations, inclusion of 'Disease Ontology', DO URL or DOID).

## [Publications](#)

### [Augmenting the disease ontology improves and unifies disease annotations across species](#)

Susan M. Bello, Mary Shimoyama, Elvira Mitraka, Stanley J. F. Laulederkind, Cynthia L. Smith, Janan T. Eppig, Lynn M. Schriml Disease Models & Mechanisms 2018:

dmm.032839 doi: 10.1242/dmm.032839 Published 27 February 2018.