



DO Buzz Newsletter September 2020

## 2020 Virtual Conferences

- ◇ [ICBO - COB workshop](#)
- ◇ [ICBO-FoodOntologyWorkshop](#)
- ◇ [ISB Biocuration workshops](#)
- ◇ [Wikidata workshop, Oct. 29](#)

**Latest News:** Checkout the latest data and website updates !

- ◇ Expanded DO's 'coronavirus infectious disease' classification, addition of COVID-19 (DOID:0080600) and Middle East respiratory syndrome (DOID:0080642)
- ◇ View OBO tree - displaying doid.obo - enabling viewing of inferred, multi-parentage
- ◇ UMLS update: DO's bi-annual update of cross-references via the UMLS, integrates novel linkages to NCI thesaurus, MESH, SNOMEDCT US, ICD9CM and ICD10CM
- ◇ Human Disease Ontology Project integrated into the [UMB Data Catalog](#)
- ◇ DO's latest YouTube video: [Clinical Applications of the Human Disease Ontology](#)

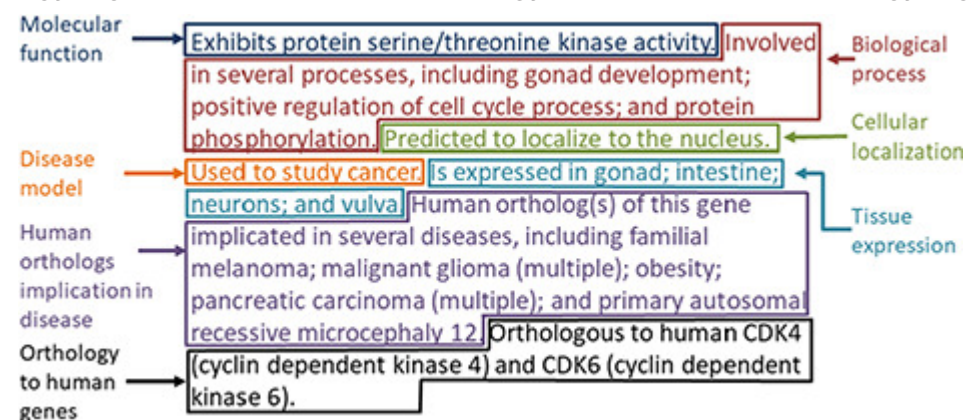
## Publication Highlight



### Automated generation of gene summaries at the Alliance of Genome Resources.

Kishore R, Arnaboldi V, Van Slyke CE, Chan J, Nash RS, Urbano JM, Dolan ME, Engel SR, Shimoyama M, Sternberg PW, Genome Resources TAO. Database (Oxford). 2020 Jan 1;2020:baaa037. doi: 10.1093/database/baaa037. PMID: 32559296; PMCID: PMC7304461.

Automated gene summaries are based on highly structured gene data such as annotations to ontology terms from several different ontologies, including the Gene Ontology (<http://geneontology.org/>) and the Disease Ontology (<http://disease-ontology.org/>).



- ◇ The Disease Ontology is utilized to generate the 'Disease Relevance,' 'Disease Biomarker' and 'Human ortholog implicated in disease' data categories used to generate the gene summaries.
- ◇ Over 121,000 gene summaries are available for the seven species (Alliance release 2.3) (<https://alliancegenome.org/downloads#gene-descriptions>)
- ◇ Total number of gene summaries for protein coding genes for the [see Table 4]:
  - Study of gene in human disease: 9,396**
  - Human ortholog implication in disease: 21,019**



### Latest Release Notes:

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

### Release # 85: September 15, 2020 Release Notes

This month's Disease Ontology release of 10,347 DO disease terms (73%, 7654 defined) includes our bi-annual UMLS update, which includes updating to the latest SNOMEDCT US release, and the addition of DO xrefs (ICD10CM, ICD9CM, MESH and NCI) and 191 OMIM IDs. This release includes new DO terms and term definitions, including lung sarcomatous carcinoma, autoimmune myocarditis and the addition of autoimmune, allergy disease logical definitions.

### Looking Ahead ...

#### September 2020 Website Updates:

New Web pages: DO Imports - view diversity of ontologies the DO uses to build logical axioms  
DO Subsets - view the 14 DO slim files ([in GitHub](#)) produced with each DO release

Revised Complex Disease Classifications: re-examining disease classification with the integration of triggers, environmental drivers and novel endotypes (mechanistic pathways)

#### Related Literature (Asthma):

Understanding Asthma Phenotypes, Endotypes, and Mechanisms of Disease  
Clin Rev Allergy Immunol. 2019 Apr;56(2):219-233. doi: 10.1007/s12016-018-8712-1. [PMID: 30206782](#)



### Disease Ontology Citations:

The DO team has identified 633 DO project citations (as of September 2020).

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>