The Human Disease Ontology project's newsletter, sharing highlights with our user community.

DO Buzz Newsletter March 2019

Latest News

Michael Sinclair joins the Human Disease Ontology Team.
Michael joined the DO team in November, 2018. He comes to us from Karen Eilbeck's lab in the Department of Biomedical Informatics, University of Utah, where Michael earned a Masters in Biomedical Informatics and was a key developer on the Molecular Sequence Ontology as a National Library of Medicine postdoctoral fellow.

DO Spotlight of the Quarter:

DO Disease Pages in XenBase

Special issue of Frontiers in Physiology on *Xenopus* models of Organogenesis and Disease:
Nenni et al (2019) Xenbase: Facilitating the use of Xenopus to Model Human Disease.

At a fundamental level most genes, signaling pathways, biological functions and organ systems are highly conserved between man and all vertebrate species. Leveraging this conservation, researchers are increasingly using the experimental advantages of the amphibian *Xenopus* to model human disease. The online *Xenopus* resource, Xenbase, enables human disease modeling by curating the *Xenopus* literature published in PubMed and integrating these *Xenopus* data with orthologous human genes, anatomy, and more recently with links to the Online Mendelian Inheritance in Man resource (OMIM) and the Human Disease Ontology (DO). Here we review how Xenbase supports disease modeling and report on a meta-analysis of the published *Xenopus* research providing an overview of the different types of diseases being modeled in *Xenopus* and the variety of experimental approaches being used.

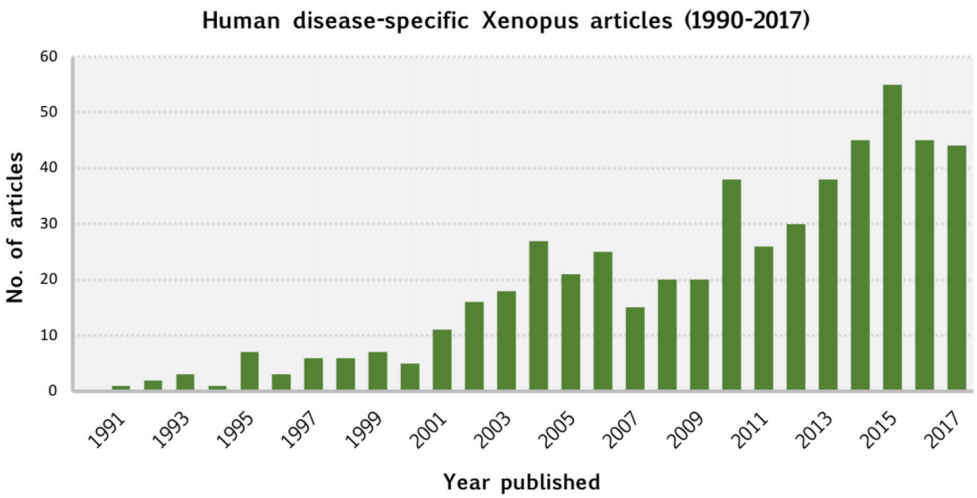

FIGURE 5 | Human disease-specific *Xenopus* articles (1990–2017). This chart shows the number of articles published, by year, between 1990 and 2017 that our curation identified as utilizing *Xenopus* as a model system for studying human disease. Publication dates were obtained from NCBI's PubMed database.

Nenni et al (2019) Xenbase: Facilitating the use of Xenopus to Model Human Disease.

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Recent Conference Presentation

(February 20-21, 2019) Orlando, Florida
Ontology for Precision Medicine: From Genomes to Public Health
Talk given by Michael Sinclair: "Cross Products for Clinical Information Retrieval from the Human Disease Ontology."

Upcoming Conferences

(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)

Latest Release Notes

DO Data Releases: Available in DO's GitHub repository: (previous release notes)
Release #56:
March 1, 2019 Release Notes:
DOID term count: 9,233 disease terms, 6,118/9233 terms defined (66%).
In doid.owl file: 678 Equivalent To axioms and 3,499 SubClass Of statements.
This release includes data and software updates, 41,172 vocabulary cross references (DO to OMIM, ICD9CM, ICD10CM, MeSH, NCI thesaurus, SNOMEDCT_US_2018, UMLS_CUI, Orphanet and NORD rare disease, the addition of 285 text definitions, revision of X-linked recessive disease logical definitions, and updates requested via GitHub tracker. Enhanced versions of the DO's Makefile and robot.jar file have been enhanced to improve our QC and release process.

Disease Ontology Citations

The DO team has identified a body of 347 DO project citations (as of March 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