

ONTOLOGIES LINK HUMAN DISEASES TO ANIMAL MODELS

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One of the major challenges facing biomedical researchers is to develop methods to link information obtained from studies of model organisms to the diagnosis and treatment of human diseases.

Model organisms provide a rich and deep understanding of gene functions particularly from studies of expression patterns and mutant phenotypes. Databases often store phenotypic information as text. Text alone, however, limits the ability to organize and search for specific information. It is much more preferable to use ontologies (structured vocabularies) because they define relationships between terms. Several existing ontologies provide terms and relationships useful for annotating phenotypes, including the Gene Ontology (GO) that classifies gene products, various model organism and human anatomical ontologies, and the Phenotype and Trait Ontology (PATO) that provides vocabulary and syntax for describing phenotypes.

We are using and further developing these resources and the syntax of PATO to design a set of orthogonal ontologies to describe mutant phenotypes and diseases. We are developing a curator interface and using it to annotate fruitfly and zebrafish mutant phenotypes and associated human diseases. Shared ontologies allow us to make links among different species.

We propose that this general strategy may be extended to annotate phenotypes in all species, thus providing a means to link human diseases to model organisms.

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