Revisions to the Disease Ontology to Support the Alliance of Genome Resources

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Model organism databases are one of the cornerstones of biomedical research, serving thousands of users daily. Each database curates and integrates vast amounts of genetic, functional, evolutionary, molecular, physiological and other biological data, information, and knowledge from the scientific literature, individual researchers, and a variety of publicly available sources. Six model organism databases and the GO consortium have formed a partnership to build the Alliance of Genome Resources (AGR), an integrated interspecies genome resource in support of translational research. The AGR will offer a unified resource to interrogate model organism data, including information related to human diseases, in order to advance genome biology and genomic medicine. To accomplish this, unifying data standards have been adopted for cross-model organism database use, including the use of the Disease Ontology (DO) as the standard vocabulary for human disease. Mouse Genome Database (MGD, http://www.informatics.jax.org) and the Rat Genome Database (RGD, http://rgd.mcw.edu), founding members of the AGR, are collaborating with DO in order to, on one hand, align their data with the disease classification model used by the DO, and, on the other other hand, to expand and enrich DO to include all the terms and relationships to cover their needs. As a result of this collaboration, the DO is now generating disease terms for individual members of OMIM's phenotypic series, has expanded the number of diseases with inferred anatomy based relationships, and has implemented a new relation to include database entries that show susceptibility to a particular disease. This will deeply enrich DO, while at the same time provide MGD and RGD - and the other members of the AGR - with a robust resource that will foster interoperability and provide the human genetics/genomics community with a consistent way to query disease associations.