Title: Working in Real Time: Building Ontologies While Annotating the Mouse from Genotype to Phenotype.

Authors: Judith A. Blake
Address: Mouse Genome Informatics
The Jackson Laboratory
600 Main Street
Bar Harbor, ME 04609
USA

Abstract: The Mouse Genome Informatics (MGI) system provides a comprehensive public resource about the laboratory mouse from sequence (genotype) to phenotype. A premier animal model for investigation of human biology and disease, the mouse is extensively manipulated genetically to study specific gene products and their contribution to particular phenotypes. Annotation of such information combines molecular details as well as information about the normal and dysfunctional phenotypes and the relationship of these phenotypes to those in humans. New data structures in MGI support a general DAG model for all ontologies that facilitates edits, views, and queries of these vocabularies.

MGI participates in the Gene Ontology (GO) consortium which provides ontologies for three molecular aspects of organisms. MGI curators contribute to the development of the GO and use the GO to annotate mouse data. We are also developing and using an ontology of phenotypes. Mouse phenotypes are associated via carefully defined relationships with human phenotypes, particularly diseases.

The challenge for ontology developers is to construct ontologies that are fully documented and that can be updated, expanded and maintained in forms accessible to the larger scientific community. I will discuss the MGI experience in ontology development which is occurring in parallel with on-going annotation of genotype and phenotype data for the laboratory mouse.