

Mouse Anatomy Ontologies and GXD

Terry F. Hayamizu, Martin Ringwald

Mouse Genome Informatics, The Jackson Laboratory, Bar Harbor, ME, USA

Extended Abstract

Anatomy ontologies are essential for the standardized description and integration of many types of biological data pertinent to anatomy, including gene expression, biological process, phenotype and pathology data. From its inception as a community resource for gene expression information for the laboratory mouse, the Gene Expression Database (GXD) has recognized this critical need, actively promoting and contributing to the development of detailed and comprehensive anatomy ontologies for both the developing and postnatal mouse.

The ontology for mouse embryo anatomy was developed by the Edinburgh Mouse Atlas Project (EMAP), with input and feedback from GXD. The EMAP ontology standardizes nomenclature for anatomical structures in the embryonic mouse and provides stage-specific partonomic hierarchies for relationships between developing structures. EMAP ontology development continues as a collaborative effort between EMAP and GXD. The Adult Mouse Anatomy (MA) ontology, developed by GXD, provides standardized anatomy nomenclature for the postnatal mouse, and is organized from both spatial and anatomical system perspectives. The plan is to eventually combine and integrate the ontologies to generate an anatomy ontology covering the entire lifespan of the laboratory mouse.

Both developmental and adult mouse anatomy ontologies have been incorporated into GXD and are currently being used to describe and integrate an extensive range of mouse gene expression data. Currently, GXD includes data for over 50,000 experimental

assays, with greater than 1 million expression results annotated to specific mouse anatomy terms, providing expression information for approximately 12,000 mouse genes. The mouse anatomy ontologies continue to be expanded and refined according to the requirements of accurate and precise data curation by GXD and other resources, as well as in response to input from others in the scientific community.

GXD is an integral part of Mouse Genome Informatics (MGI), which provides integrated access to a wide range of information pertinent to the laboratory mouse. Since many types of data have anatomic attributes, anatomy ontologies are also being used to integrate different kinds of data available for the mouse. For example, Mammalian Phenotype Ontology (MP) development is guided by both MA and EMAP ontologies, by incorporating anatomy term names, confirming term placement within the ontology, and using anatomy terms in logical definitions of MP terms. Furthermore, Gene Ontology (GO) curators use anatomy ontologies as a guide for adding biological process terms that refer to anatomical structures, as well as to indicate specific mouse structures within contextual annotations. Thus, the mouse anatomy ontologies are being used to annotate and integrate gene expression and phenotype data, contributing to an integrated description of biological phenomena in the mouse. Furthermore, we are working on establishing cross-references with anatomy ontologies for other species to support cross-species queries and comparative analysis.

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