Multi-species Ontologies of the Craniofacial Musculoskeletal System

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***Abstract*— We created the Ontology of Craniofacial Development and Malformation (OCDM) [1] to provide a unifying framework for organizing and integrating craniofacial data ranging from genes to clinical phenotypes from multi-species. Within this framework we focused on spatio-structural representation of anatomical entities related to craniofacial development and malformation, such as craniosynostosis and midface hypoplasia. Animal models are used to support human studies and so we built multi-species ontologies that would allow for cross-species correlation of anatomical information. For this purpose we first developed and enhanced the craniofacial component of the human musculoskeletal system in the Foundational Model of Anatomy Ontology (FMA)[2], and then imported this component, which we call the Craniofacial Human Ontology (CHO), into the OCDM. The CHO was then used as a template to create the anatomy for the mouse, the Craniofacial Mouse Ontology (CMO) as well as for the zebrafish, the Craniofacial Zebrafish Ontology (CZO).**

Keywords—Foundational Model of Anatomy, Ontology of Craniofacial Development and Malformation, FaceBase Consortium, Craniofacial Congenital Abnormalities

I. Introduction

Craniofacial development is a complex process consisting of embryological events that are influenced and controlled by both genetic and epigenetic factors. Any disturbance during the course of development can result in structural malformations such as craniosynostosis (premature fusion of cranial sutures). The FaceBase Consortium [3, 4] was established to collect data ranging from genes to disease in order to understand the causes of these conditions. The purpose of the OCDM is to provide a semantic basis for integrating and understanding these diverse data, by providing a detailed description of structures resulting from normal and pathological developmental processes. One of the main components of the OCDM, canonical human craniofacial anatomy, is extracted from the FMA to provide the organizing framework for representing both normal and abnormal craniofacial structures in the human, the mouse and zebrafish species and the associated processes involved in their development and malformation. We restrict the description of anatomical entities to physical and spatio-structural properties only, and then use this description for identifying the structural correspondences between the different species.

II. Methods

We initially augmented the craniofacial content of the FMA with extensive spatio-structural representations of anatomical entities and relations, including development, pertinent to the scope of FaceBase. Hence, we extended the description to the components of the musculoskeletal system, which involves the muscles, bones, skeletal ligaments, cartilages and joints, to account for the possible sites of morphological changes observed in craniofacial malformations. The enhanced human craniofacial component of the FMA was then imported into the OCDM as the CHO, where it not only represents human craniofacial anatomy, but also provides the ontology template for various model organisms represented in the OCDM, such as the mouse and the zebrafish, which can be used to cross correlate with the human version. The mouse version, the CMO, was edited to assure that the content pertains only to mouse structures, and was mapped to other existing mouse ontologies such as the Mouse Adult Gross Anatomy Ontology (MA) [5] and EMAP [6]. The underlying ontological framework for both CHO and CMO, which consists of defined high level classes, was then used to build the CZO, with content derived from various sources, such as ZFA from ZFIN [7], authoritative publications, and domain experts from FaceBase. We used existing ontologies where applicable and created new content where needed. These ontologies were designed to facilitate integration and interoperability of craniofacial data from multiple sources, multiple levels of granularity and multiple species.

III. Results

Content development for all three species was carried out in the Protégé OWL version 4.3 authoring tool [8]. Figures 1-3 show Protégé screen captures of the class `Skull` in CHO, CMO and CZO. On the left panel is the

class hierarchy (taxonomy) and on the right, the different spatio-structural properties and annotations associated with the skull.

Fig 1. CHO ontology

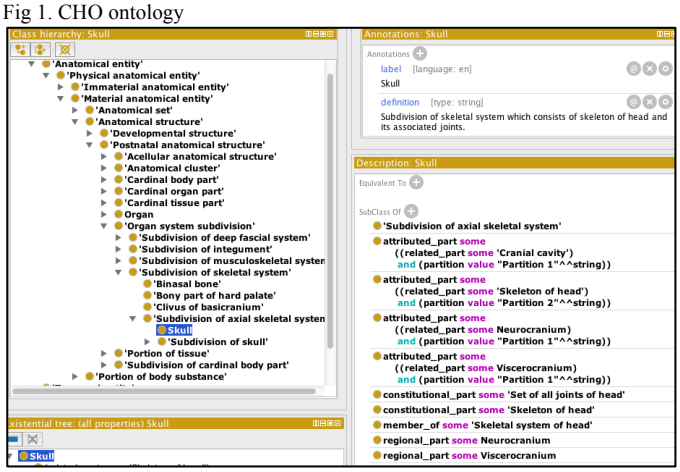


Fig. 2. CMO ontology

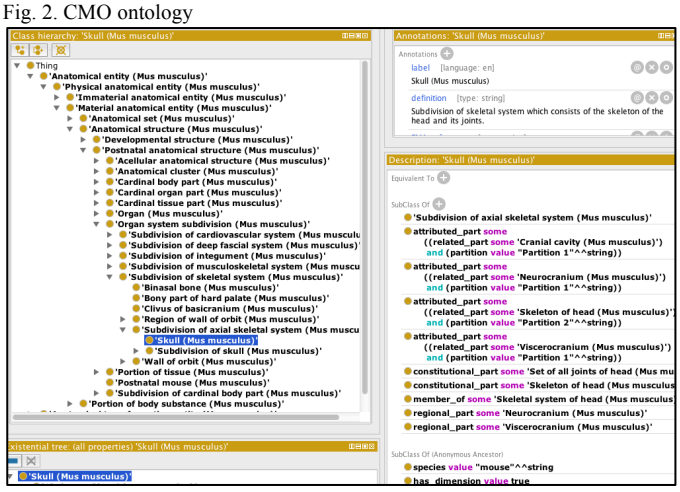
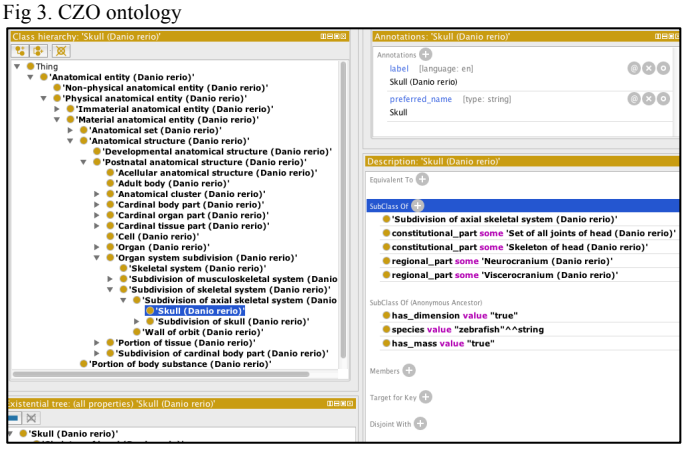


Fig 3. CZO ontology



IV. Conclusion

Studies in animal models provide data in support of human disease studies. The OCDM was created as a unifying system for integrating data from human and animal models, using anatomy as the organizing framework for representing craniofacial development and malformation. Because the same ontological principle was used to develop the human, mouse and zebrafish ontologies, more precise structural correlation between species is facilitated. The Protégé OWL version of the OCDM can be downloaded from the UWSIG site: http://www.si.washington.edu/projects/ocdm

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