Ontology of Human and Mouse Craniofacial Musculoskeletal System

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Abstract

We developed and enhanced the craniofacial component of the human musculoskeletal system in the Foundational Model of Anatomy Ontology (FMA)[1], and imported this component as an updated version of the Craniofacial Human Ontology (CHO), which is a sub-ontology of our evolving Ontology of Craniofacial Development and Malformation (OCDM)[2]. The CHO was then used as the ontology template for the Craniofacial Mouse Ontology (CMO) sub-ontology of the OCDM, the content of which was updated to include only structures that are found in mice, but which can be correlated to those in humans. Both sub-ontologies provide the structural foundation for a unifying framework that organizes and integrates craniofacial data ranging from genes to clinical phenotypes. In this study, we focus on the spatio-structural representation of the anatomical entities related to cranial dysmorphologies, such as craniosynostosis and midface hypoplasia in both species.

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 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 both the human and the mouse species and the associated processes involved in their development and malformation. We will later extend the ontology to include the zebrafish.

We initially augmented the craniofacial content of the FMA with extensive spatio-structural representations of anatomical entities and relations pertinent to the scope of FaceBase 2. 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) and EMAP. The underlying ontological framework is therefore designed to facilitate integration and interoperability of craniofacial data from multiple sources and multiple levels of granularity.

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