

# Cross-species Mapping of Human and Mouse Craniofacial Structures

Jose LV Mejino Jr. MD<sup>1</sup>, Landon T Detwiler, MS<sup>1,2</sup>, Timothy C Cox, PhD<sup>3,4</sup>, and James F Brinkley, MD, PhD<sup>1,2</sup>

<sup>1</sup> Department of Biological Structure, <sup>2</sup> Department of Biomedical Informatics and Medical Education, University of Washington, <sup>3</sup> Division of Craniofacial Medicine, Department of Pediatrics, University of Washington, <sup>4</sup> Center for Developmental Biology and Regenerative Medicine, Seattle Children's Research Institute, Seattle, WA, USA

The Ontology of Craniofacial Development and Malformation (OCDM)<sup>1</sup> is an OWL application ontology for use by the NIH-sponsored FaceBase consortium, as well as by the larger craniofacial research community. The purpose of FaceBase is to collect and integrate data ranging from genomic to clinical in order to understand the causes and possible treatments of craniofacial birth defects such as cleft lip and cleft palate. The purpose of the OCDM is to provide the terminology standards, as well as the relationships (e.g. parts, developmental lineage, associated malformations), to associate and correlate disparate FaceBase data pertaining to the craniofacial complex by formally representing multi-species canonical anatomy, embryological development, malformation, and cross-species anatomy homology mappings.

Understanding development and malformation requires knowledge derived from research on model organisms, such as mice and zebrafish, that cannot directly come from human subjects. The wealth and availability of information from these organisms can be leveraged and utilized to advance research at the human level. In previous work on the OCDM we have provided comprehensive representations, not only of human, but also of mouse and zebrafish craniofacial anatomy. In the work reported here we investigated homology correspondences between human and mouse craniofacial structures, and represented them in the Craniofacial Human Mouse Mapping Ontology (CHMMO), one of several sub-ontologies of the OCDM.

Mappings include developmental entities from the zygote to perinatal structures at progressive stages of development, with emphasis on embryonic precursors of components of the musculoskeletal system of the head (muscles, bones, cartilages, skeletal ligaments and joints). Despite overtly apparent differences between mice and humans, these organisms are closely related mammals and share very similar biology. The structural similarity between the two organisms is the basis of much translational research. We initially carried out provisional mappings based on existing nomenclature, incorporating similar names, and then validated these, through consultation with domain experts and the literature, for homology evidence, either based on direct molecular studies or inferred vertebrate Bauplan. Bi-directional one-to-one and uni-directional one-to-null mappings were recorded using a Protégé mapping plugin we developed (Figure 1). Except for the very early stages of development (zygote to germ layers), mappings were largely implemented at the gross anatomical level.

Figure 1 Mapping editor Protégé plugin

These mappings (and mappings in progress relating human to zebrafish) will facilitate ontology-based searches of FaceBase model organism data, such as gene expression studies, that can give insight into the causes of clinical conditions such as cleft lip or palate. Funding: NIH DE24417.

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## References

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