

Cross-species Mapping of Human and Mouse Craniofacial Structures

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1. Introduction

The Ontology of Craniofacial Development and Malformation (OCDM) serves as a unifying framework for integrating multiple forms of data generated by craniofacial researchers in FaceBase. It provides terminology standards and ontological semantics, to associate and correlate disparate FaceBase data by formally representing multispecies canonical craniofacial anatomy, embryological development, malformation, and cross-species anatomy homology mappings. Biomedical research involving development and malformation requires knowledge derived from model organisms, such as mice and zebrafish, which serve as invaluable models for acquiring information that cannot directly come from human subjects. The wealth and availability of information from model organisms can be leveraged and utilized to advance research at the human level. In the OCDM we therefore provided a comprehensive representation of model organism anatomy, such as that of the mouse (Craniofacial Mouse Ontology [CMO]) that can be correlated and mapped to human anatomy (Craniofacial Human Ontology [CHO]). We then created the Craniofacial Human Mouse Mapping Ontology (CHMMO), a subontology of the OCDM to record the homology mappings of craniofacial structures between humans and mice and later on, between humans and zebrafish.

2. Objective

Ontologically record homology mappings between:

human and mouse (CHMMO)

3. Methodology

A. Data sources

- Craniofacial Human Ontology (CHO)
- Craniofacial Mouse Ontology (CMO)
- Literature/publications
- Ontologies (Uberon, MA, EMAP)

B. Mapping approach

- provisional mappings based on similarities in existing nomenclature
- mapping at gross anatomical level

(except early developmental structures)

- validation by

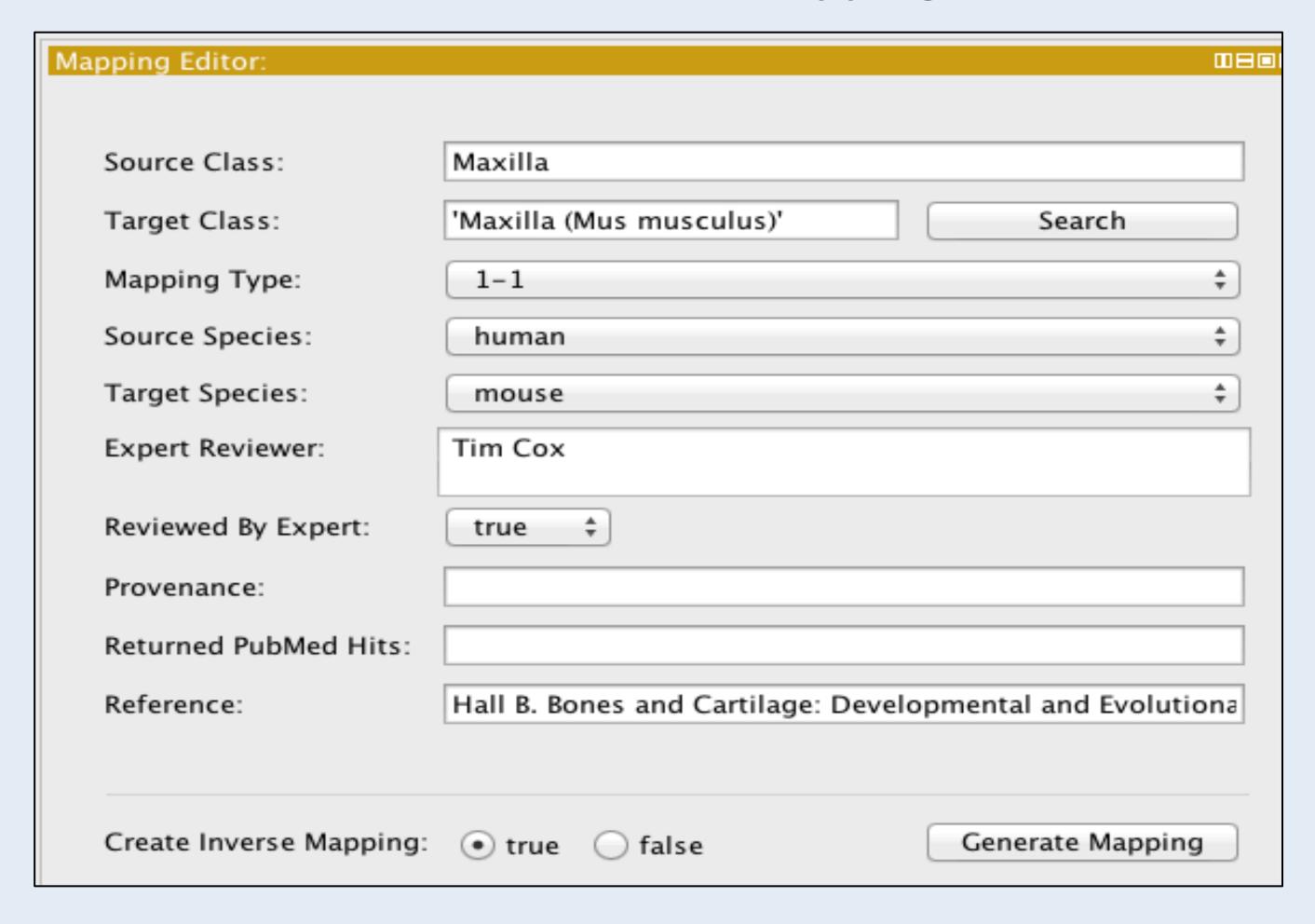
domain experts
literature/publication

C. Mapping types

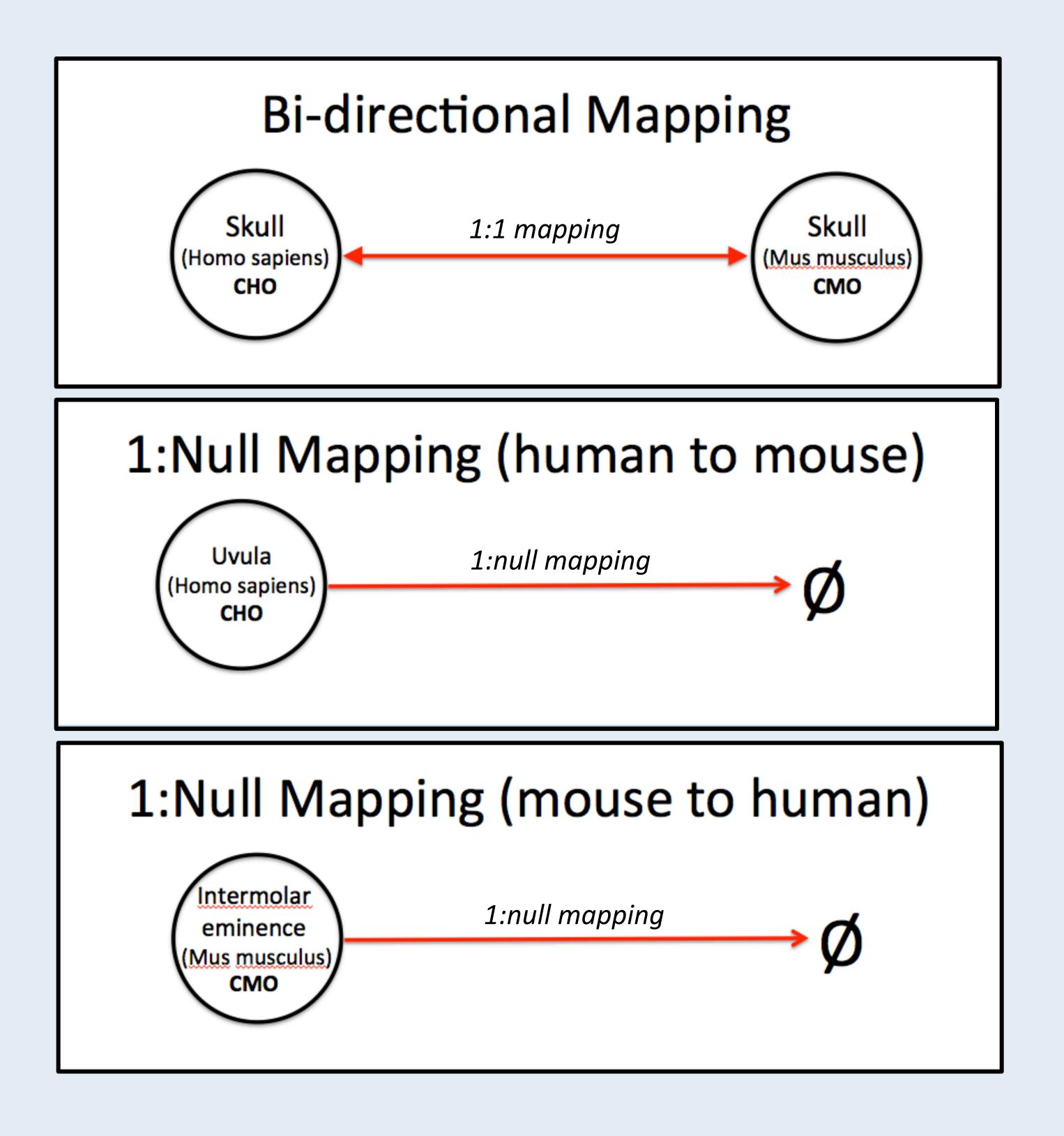
Bi-directional *one-to-one*Uni-directional *one-to-null* and *one-to-many*

D. Mapping tool (Protégé)

Human-mouse mapping



4. Results



Resolving one_to_many mapping

