

An Ontology for Human Craniofacial Development

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Abstract

In this study we develop and extend the human development content in the Foundational Model of Anatomy Ontology (FMA)¹, with emphasis on the craniofacial region. This content is a requirement to support the Ontology of Craniofacial Development and Malformation (OCDM). The OCDM aims to serve as a unifying framework for organizing and integrating disparate craniofacial data generated by the different members of the FaceBase Consortium. Here we elaborate on the embryological development of anatomical structures that are related to our use case for this project, the cleft lip and/or palate (CL/P).

Craniofacial development consists of complex embryological events that are influenced and controlled by both genetic and environmental factors. Any disturbance at any time in the development can result in structural malformations, such as facial clefts. In this study we selected cleft lip and/or cleft palate (CL/P), the most common type of facial cleft, as our primary use case. We augmented the craniofacial content of the FMA with extensive spatio-structural and temporal representations of anatomical entities and relations relating to structures involved in CL/P (e.g. lips, alveolar ridge, incisor teeth and palate). We extended the description of entities and relations to granularity levels that would allow us to account for where and when malformation can occur during normal development. Where applicable, we leveraged existing ontologies, such as HPO and PATO, to enhance the content development.

The main objective of this project is to provide support for the FaceBase Consortium (<https://www.facebase.org/>), which primarily deals with craniofacial abnormalities. To date no existing ontology is sufficient enough to comprehensively represent its domain, which includes both normal and abnormal anatomy and processes and their associated embryological development. The OCDM was built for this purpose, with one of its main components, craniofacial development, extracted from the FMA to address both the normal and abnormal development of anatomical structures.

The approach we describe here serves three practical purposes: 1) to use a reference ontology such as the FMA to provide semantic standards for FaceBase communication and annotations of development-related data, 2) to provide a developmental ontology template for model organisms, such as the mouse, which can be used to cross correlate with the human version, and 3) to leverage this underlying ontological framework to facilitate and promote integration and interoperability, reuse of knowledge and data discovery among applications that use different schemes.

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References

1. Rosse C, Mejino JLV. 2007. The Foundational Model of Anatomy Ontology, In: Burger A, Davidson D, Baldock R (eds). Anatomy Ontologies for Bioinformatics: Principles and Practice, London: Springer, pp59-117.

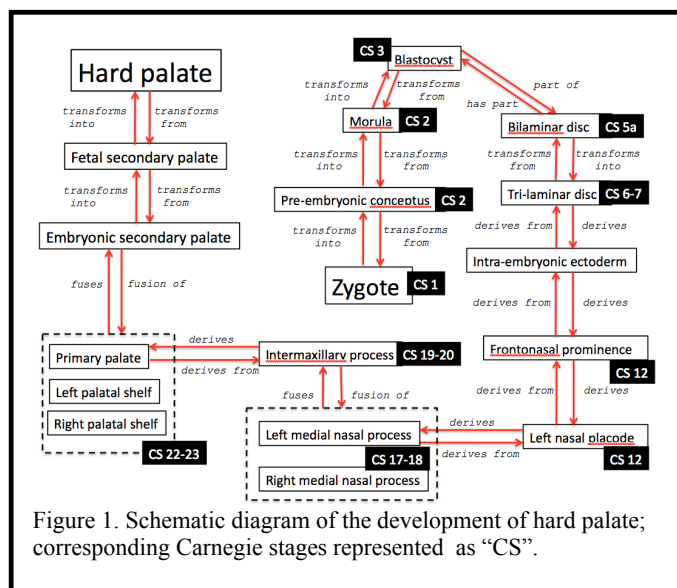


Figure 1. Schematic diagram of the development of hard palate; corresponding Carnegie stages represented as “CS”.