

# Representation of parts within the Foundational Model of Anatomy ontology

A critique and proposal for enhanced structure and consistency of regional parts

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**Abstract**— As biomedical ontologies grow in size and complexity it is crucial to develop methods for detecting inconsistencies within ontologies. The Foundational Model of Anatomy (FMA) ontology represents knowledge of human anatomy, with structural organization provided by class and part relationships. Using a manual audit, I identify types of inconsistencies arising from class and regional part relationships for regions of the body and the parts of organs. Inconsistencies arise from both explicitly declared relationships and relationships that are implied by the lexical constructs of class names. The purpose of this work is to propose methods of structural organization and lexical consistency that will make the FMA more compatible with computational auditing and increase its usability.

**Keywords**—ontology; paratomy; anatomy

## I. INTRODUCTION

As biomedical ontologies grow in size, specificity and complexity, maintaining internal consistency of the representation becomes increasingly difficult. Because ontologies can contain tens of thousands of classes, computational methods for detecting inconsistencies are necessary for quality assurance efforts. One step toward designing computational auditing methods is to identify patterns within the content that are useful for identifying inconsistencies in modeling or possible errors.

This paper examines part and class relationships within the Foundational Model of Anatomy (FMA) ontology—both those explicitly declared and those implied by the lexical constructs of class names. The purpose of this work is to identify types of inconsistencies and to propose methods of structural organization and lexical consistency that will make the FMA more compatible with computational auditing and increase its usability.

## II. BACKGROUND

The FMA represents knowledge of human anatomy using a series of organizing units at different levels of granularity. These units include “Cardinal body part”, “Organ system”, and “Organ” (Fig. 1) [1]. The FMA is one of the largest biomedical ontologies, with over 104,000 classes and 140 types of relations. In addition to the *subClass\_of* relation defining the class hierarchy, the main organizational structure is provided by the *regional\_part* relation (for spatial divisions of the body) and the *constitutional\_part* relation (describing divisions for which the parts are simpler in composition than the whole) [2].

## III. METHODS

This work is based on version 4.3 of the FMA, released in February 2016. The OWL file was manually inspected in Protégé (version 4.3.0).

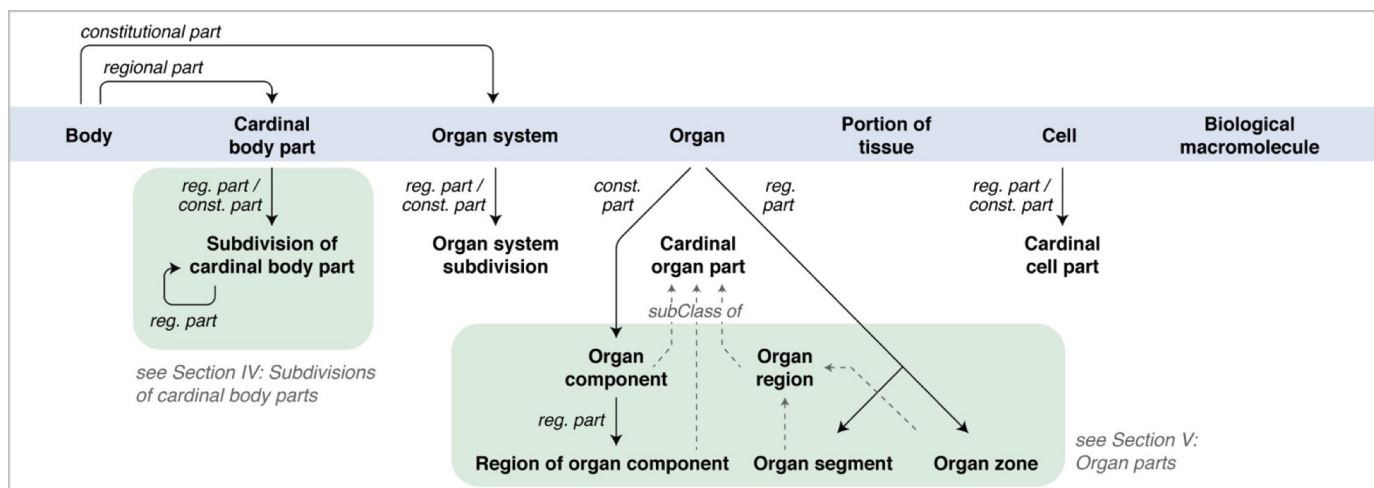


Fig. 1. Overview of part relationships using high-level classes within the FMA. Classes representing organizing units are in the blue bar. Typical *regional\_part* and *constitutional\_part* relationships among the subclasses are indicated with solid black arrows. *SubClass\_of* relationships are indicated with gray dashed arrows.

#### IV. SUBDIVISIONS OF CARDINAL BODY PARTS

Each of the cardinal body parts (“Head”, “Body proper”, left and right “Upper limb”, and left and right “Lower limb”) are divided into a hierarchy of smaller regions using the *regional part* relation. These divisions of the body are crucial in helping users understand and navigate the FMA and to provide classes that can be lexically modified to create other classes.

##### A. Alignment to the class hierarchy

As demonstrated in Fig. 2, there is an opportunity to tightly align the regional part hierarchy for cardinal body parts to the class hierarchy. Much of the structure for coordinating the two hierarchies is already in place, but some regions of the body are classified as types of anatomical clusters instead of subdivisions of the body. This obscures the relationship of those classes to other regions of the body.

There are two benefits to formalizing this organizational structure. First, it provides a place to classify all divisions of a particular region—even if a class represents an alternative partition which is not reflected in the *regional part* hierarchy. This is important for classes such as “Trunk”, which have more than one partition scheme. Second, users could more easily answer the question of what partitions are provided by the FMA for a particular region of the body by navigating the class hierarchy beneath “Subdivision of cardinal body part”.

For example, classes such as “Right side of head” and “Left side of head” do not currently have any relationships to other classes. But because they are subclasses of “Subdivision of head”, users can locate these terms to use in annotation.

##### B. Lexical consistency within class names

As shown in Fig. 3, some class names consist of a modifier (such as “Surface of...”) and a body region or high-level class for types of body regions. Consistency in referring to body regions would aid auditing and ontology users. In cases where another anatomical term is traditionally used, the term reflecting a consistent construct could be added as a synonym.

##### C. Consistency within the class hierarchy

Another source of inconsistency arises from the lexical implications of class names and their positions within the class hierarchy. As shown in Fig. 4, “Integument of chin” is a subclass of “Integument of subdivision of mouth.” From this a user would infer that “Chin” is a “Subdivision of mouth”, which is confirmed by the subclass relationship for “Chin” found in the FMA. However, as detailed in Fig. 4, contradictions can easily arise. For example, “Set of jaws” is a subclass of “Set of subdivisions of head”, but “Jaw” is a subclass of “Organ cluster.” Correcting these inconsistencies depends upon first establishing a comprehensive *regional part* hierarchy for subdivisions of cardinal body parts.

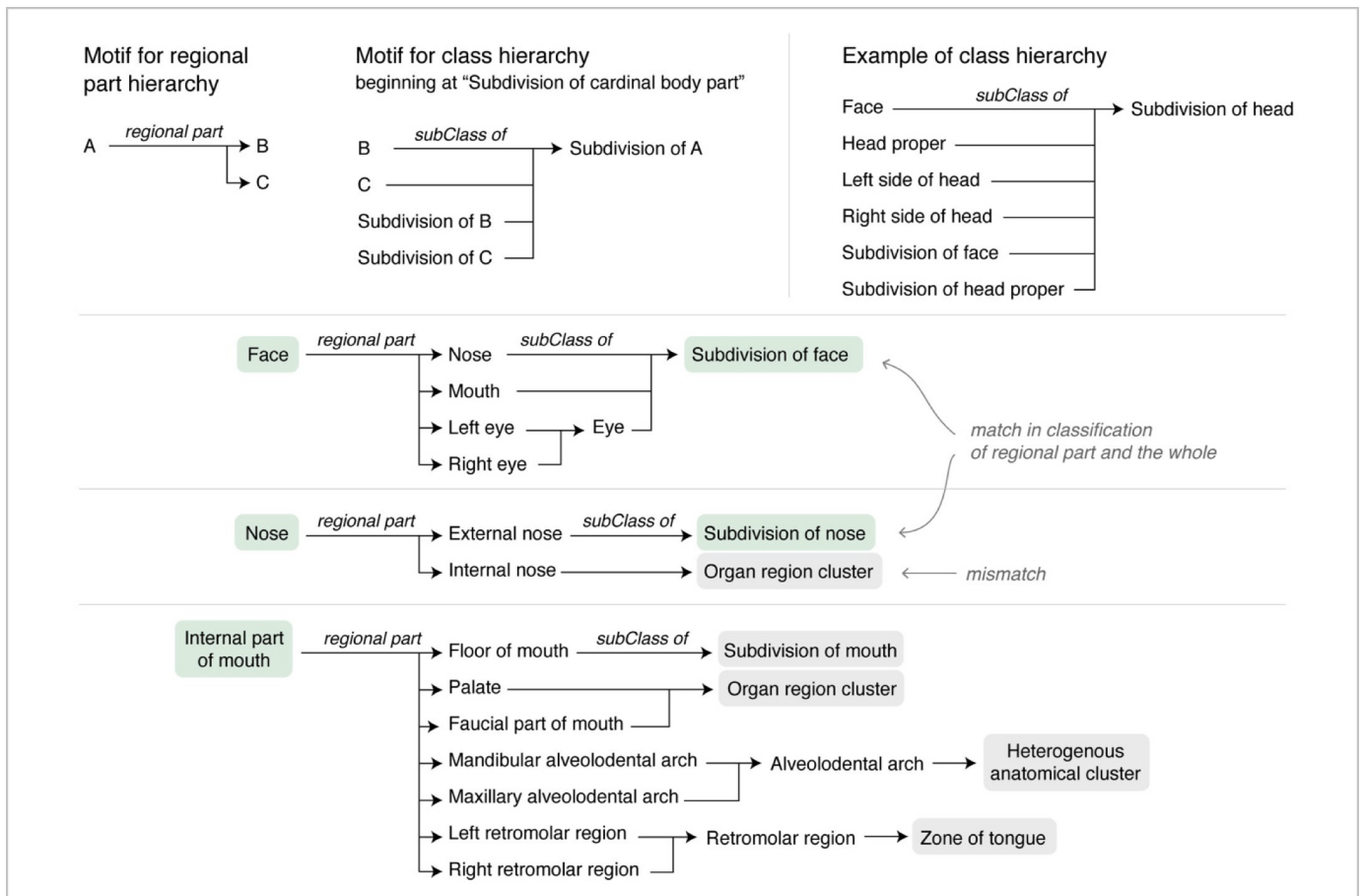


Fig. 2. Matches and mismatches in classification of parts in the regional part hierarchy and the class hierarchy. *Top left:* The motif for modeling regional parts is reflected in much of the class hierarchy beginning at “Subdivision of cardinal body part” (often using the phrase “Subdivision of...”). *Top right:* Example of class hierarchy, showing subclasses of “Subdivision of head”. Two of the classes, “Face” and “Head proper”, have corresponding classes beginning with the phrase “Subdivision of...”. *Bottom:* Examples of matches (green) and mismatches (gray) between classification in the regional part and class hierarchies.

| Examples of modifiers for body regions |  | Lexical variation in referring to “Cardinal body part” |  |
|--|--|--|--|
|  |  | phrase   | example of class                               |
| Cavity of...                           |  | ...of cardinal body part                               | “Vasculature of cardinal body part”            |
| Integument of...                       |  | ...of principal body part                              | “Set of capillary beds of principal body part” |
| Junction of...                         |  |  |  |
| Musculature of...                      |  |  |  |
| Neural network of...                   |  |  |  |
| Systemic capillary network of...       |  |  |  |
| Set of...                              |  |  |  |
| Set of capillary beds of...            |  |  |  |
| Surface of...                          |  |  |  |
| Vasculature of...                      |  |  |  |

| Lexical variation in referring to “Subdivision of cardinal body part” |  |
|---|--|
| phrase  | example of class   |
| ...of subdivision of cardinal body part                               | “Cavity of subdivision of cardinal body part”              |
| ...of cardinal body part subdivision                                  | “Neural network of cardinal body part subdivision”         |
| ...of body part subdivision   | “Vasculature of body part subdivision”                     |
| ...of subdivision of principal body part                              | “Surface of subdivision of principal body part”            |
| ...of principal body part subdivision                                 | “Set of capillary beds of principal body part subdivision” |

Fig. 3. Phrases in class names relevant to body regions. *Left*: Examples of modifiers paired with terms for regions of the body. *Right*: Variation in wording of terms which refer to high-level classes “Cardinal body part” and “Subdivision of cardinal body part”.



Fig. 4. Implications of class names referring to subdivisions of cardinal body parts. *Top*: Example of consistency in classification between “Integument of chin” and “Chin”, with “Chin” a “Subdivision of mouth” in both statements. *Bottom*: Examples of inconsistencies between the implied and actual placement of a class within the class hierarchy.

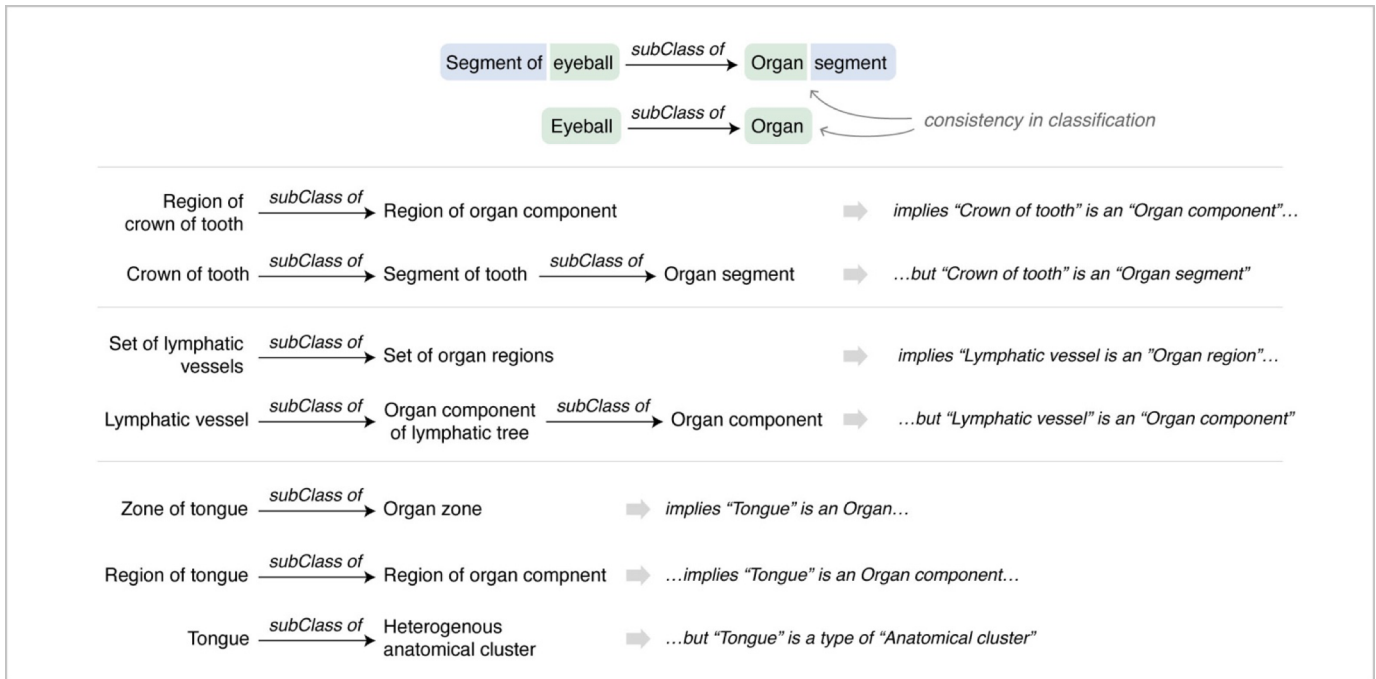


Fig. 5. Implications of class names referring to parts of organs. *Top*: Example of consistency in classification between “Segment of eyeball” and “Eyeball”, with “Eyeball” an “Organ” in both statements. *Bottom*: Examples of inconsistencies between the implied and actual placement of a class within the class hierarchy.

## V. ORGAN PARTS

The FMA has a complex representation of organ parts. Parts are classified as either an “Organ component” (bounded predominately by bona fide boundaries) or an “Organ region” (defined by fiat boundaries). Each organ region is further classified as an “Organ segment” (with anchored fiat boundaries) or an “Organ zone” (with floating fiat boundaries).

As shown in Fig. 5, inconsistencies arise when the lexical implications of class names do not match the class hierarchy. For example, “Region of crown of tooth” is a subclass of “Region of organ component”. However, “Crown of tooth” is classified as an “Organ segment”.

It is unclear if the complexity of representation for organ parts adds value to the FMA. But if retained, then auditing methods should be developed to ensure consistency.

## VI. BRANCHES AND TRIBUTARIES

Several types of organs have a tree structure, including arterial tree organs and venous tree organs. The regional parts of these trees can be described as a trunk plus either branches or tributaries. The FMA has two specific types of *regional\_part* relations (*branch* and *tributary*) to relate branch and tributary parts to the tree. However these relations are applied inconsistently—duplicating the *regional\_part* relationship, substituting for the general relationship, or not used at all. This inconsistency impacts efforts to access knowledge about tree organs and determine the completeness of representations.

## VII. KEYWORDS INDICATING REGIONAL PARTS

The terms “branch” and “tributary” have specific anatomical meanings in the FMA. But for other terms such as

“subdivision”, “region”, “segment”, and “subsegment” it is unclear whether these are synonyms or they carry specific meaning when used in class names.

The term “portion” should be reserved for subclasses of “Portion of tissue” and “Portion of body substance”, but has been applied to several regions of organs (such as “Intrapulmonary portion of pulmonary artery”).

## VIII. CONCLUSION

I have documented a variety of ways that inconsistencies in the representation of regional parts occur within the FMA. The first step to addressing these inconsistencies is to establish a robust regional part representation of the cardinal body parts.

Previous work to audit the FMA has used symmetric terms (for example, “left” and “right”, “superior” and “inferior”) [3]. This work identifies additional lexical modifiers that can be used to detect inconsistencies in the class hierarchy, regional part hierarchy, and class names.

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