# Towards the development of a biomechanical ontology to support the initiatives of the Parametric Human Project


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## Abstract

The creation of a conceptual schema that symbolically represents the collective knowledge of a particular domain is a daunting task, particularly to capture and unify the breadth and complexity of human form and function. Much like other modelling endeavours, a number of fundamental challenges exist when developing a formal knowledge base. The primary goal here is to identify the key elements that must be considered during ontology creation and evolution, so that we can facilitate the desired compilation and sharing of multi-modal, multi-dimensional anatomical data. What is required to accommodate a growing, iteratively-authored, and searchable data-driven knowledge base? How do we annotate and compile multiple instances of anatomical structures into a probabilistic framework that accounts for inter-individual variability, including the potential existence of geometrical asymmetries, anomalies, abnormalities and pathologies? A lack of common standards can result in massive information loss, both in terms of what is (or can be) analytically gleaned from the data, as well as the contextual information that guides observations and analyses. Thus, we strive to develop a flexible ontology that incorporates sufficient granularity to support the assembly of a "complete" human model that enables multi-purpose, multi-scale modelling and simulation.



**Keywords**: biomechanical ontology, anatomical variation, semantics, knowledge representation, knowledge sharing.


## 1. Introduction

Perusal of the literature indicates how pervasive the use of biomechanical models is becoming nowadays. Modelling and simulation now seem to play an increasingly central role in decision-making processes, be it to inform clinical- or research-based choices in health-related fields (e.g., orthopaedics, surgery, rehabilitation), or safety and design in fields related to engineering. In many cases, a scalable model of a representative human is used, or morphed to represent a specific clinical scenario, to garner information that can be generalized across the population of interest. Just how reasonable such assumptions are remain unclear.

Most existing human models rely on the reconstructed anatomy of a single individual (e.g. Visible Human), or are comprised of data gathered and compiled from multiple sources. The latter effectively results in some sort of Frankenstein-like human – not what most would consider normal, or representative. Thus, most specimens represent a convenient rather than random sample of the population, and might or might not be prototypical. That said, the level and modes of human anatomical variation within the population are not generally known. Moreover, sparse datasets are often supplemented with data garnered from animal models. These facts are somewhat troubling, particularly given the use of models in planning and decision-making in clinical and industrial settings. Understanding human anatomical variation, and how it impacts behaviour, is paramount to the fidelity of results. Just how variable the anatomy is across individuals, and how this might impact the results generated using models remains uncertain. The Parametric Human Project (PHP) was established to work towards addressing these concerns, through the aggregation and construction of a probabilistic digital atlas of the human musculoskeletal anatomy. Conceptually, such an atlas will catalogue data and models that can be used to define and construct any individual that resides within the comprised *virtual population*.

The vision of creating a unified, formal knowledge base requires not only the collection and compilation of data, but also the framework to support accessibility and usability (i.e., an ontology). The nature of collaboration necessitates that we, as a group, agree upon a common language base so that data, information and knowledge can be shared and transferred, while remaining semantically rich. This will help to eliminate ambiguity in scientific discourse, enable knowledge sharing, and support external reasoning. Given the scope of the PHP, we envision



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developing an ontological framework that will support knowledge regarding human anatomy and biomechanics, and the relationship between form and function.

As such, the purpose of the current paper is threefold: 1) to provide an overview of the envisioned importance of developing a new ontological framework to support the PHP consortium; 2) to identify and highlight the salient features that we must incorporate into the proposed ontology; and 3) to identify pertinent resources that will help to guide the development of the proposed ontology.

## 2. The importance of an ontology

Historically, the term *ontology* refers to the philosophical study of the nature of being, or reality. As such, any systematic account of existence aims to identify all entities that do (or can be said to) exist, as well as how those entities can be grouped or categorized, and how they relate to one another within a hierarchy. Consequently, such an existential framework should enable further subdivision of entities according to their similarities and differences.

Within the realm of knowledge representation and artificial intelligence, an ontology is often described as an explicit *conceptualization* of a domain [1]. For anything that can be said to "exist" within a given domain, the nature of its existence can be represented and formalized within a knowledge base. To represent knowledge of a particular domain requires the construction of a controlled vocabulary (or terminology), as well as the definition of the relationships that exist between and amongst the defined terms (or *mereotopology*). It is these relational definitions that connect the various terms in a hierarchical fashion, and distinguish an ontology from a simple list of terms. Defined relations create granularity, and allow high-level information to be decomposed into lower-level bits (e.g., separate wholes into parts, parts of parts, and the boundaries between parts). Consideration of these defined relations is critical to constructing the desired semantic structure [2]. Consequently, any ontology constrains the meaning of a domain language, and creates a shared, formal semantics that describes how pieces of information interact with each other. Together, these characteristics help to structure logical inference and valid reasoning within a knowledge base.

The primary purpose of an ontology is standardisation, at both the syntactic and semantic levels (i.e., the terms, concepts, and relationships). Much like an underlying 3D coordinate system in imaging or modelling, an ontology creates a system of reference by which to unify its users. The lack of such a common frame of reference can result in dramatic information loss. For collaborative purposes, it is important that we agree upon, and properly implement a common language that incorporates mutually understood concepts and definitions. This will create *interoperability* across information systems, and allow us to effectively interact and communicate with each other, as well as with computational systems. By establishing a standard framework to document and archive metadata, we can more easily provide and propagate context when sharing data and/or processing and analysis tools. Beyond the vocabulary itself, ambiguity can only be avoided if we have a clear understanding of the relations that denote the specific connections between the various entities (e.g., *part_of*, *overlaps_with*). To achieve this requires that terms be embedded in a formal theory, so that we can analyse the connections between relations and their logical properties.

## 3. Considerations for the development of the ontology of human biomechanics

### 3.1 What is biomechanics?

Biomechanics is often described as existing where the study of biology intersects that of mechanics. Specifically, *biology* concerns the study of life and living organisms, including their structure, function and variation. To be explicit, living systems are biologically responsive to physical interactions, and have the capacity to sense the environment, respond and adapt over time. *Mechanics* involves the study of how physical bodies behave when forces or displacements are applied, and the subsequent effects of those bodies on their environment. As such, some consider biomechanics to involve the study of the mechanical aspects of biological systems. However, it has been argued that this is incorrect, "because biological systems do not have mechanical aspects. They only have *bio*mechanical aspects (otherwise mechanics, as it exists, would be sufficient to describe all phenomena which we now call biomechanical features of biological systems)." [3] Stated differently, mechanics is merely a set of concepts and approaches that can be used to study the structure and function of a given biological system. As such, we subscribe to the definition of *biomechanics* as "the study of the structure and function of biological systems by means of the methods of mechanics." [3]

The above definition captures the essence of our decision to use biomechanics as the domain for which to develop the current proposed ontology. Ultimately, it is the biology of the system (e.g., anatomy and physiology) that dictates its behaviour when forces (both internal and external) are applied. However, those same forces also cause adaptation, and lead to structural, and perhaps even functional, variability. The PHP supports the need to improve and formalize our understanding of such human anatomical and physiological variability. By creating a probabilistic atlas of the human anatomy, we can begin to quantitatively instantiate biological variability. The population of such an atlas might facilitate a stronger understanding of adaptation and the relationships between structure and function, since anatomical variability inherently informs us of the history of the forces that have been applied to the various tissues and structures.

Thus, we believe that the creation of such a framework will better support the use and description of mechanics to study structure and function of the human biological system. In essence, we want to add *metrology*, to allow us to mathematically define physical properties, so that we can classify and characterize measurable (and derivable) quantities, as they relate to the modelled anatomy. For example, we can mathematically describe how a muscle generates force, but also want to define its dependency on other biomechanical and physiological factors (e.g., tendon material properties, velocity of movement, fatigue). In this sense, the desire to integrate biomechanical properties is similar to the intent of the recently developed Ontology of Physics for Biology (OPB) [4]. Thus, a physics-based framework exists by which we can begin to conceptualize how to best incorporate biomechanical content into the proposed ontology. Importantly, the ontology of human anatomy will need to be extended to include lower-level details that will support and align with that of the new ontology of human biomechanics.

## 3.2 Specific characteristics of the ontology of human biomechanics

The development of any ontology is similar to that of a computer program. It is important to first identify the anticipated utility, and then keep the intended use(s) in mind prior to and during the development. Otherwise, no explicit framework exists to guide the inclusion, or exclusion, of the appropriate elements and structural relationships.

As with any modelling endeavour, the development of any ontology involves numerous build-test-revise cycles. The immediate importance of such an iterative process is to generate clarity and coherence, which is important to any ontology. Specifically, *clarity* is needed to effectively communicate the intended meaning of the included terms to all users, which is, in part, facilitated by making definitions as objective as possible. *Coherence* means that the ontology strives to generate inferences that are consistent with the definitions within the ontology. There are also some characteristics that we need to keep in mind throughout the development and testing of the current proposed ontology. These considerations, to be highlighted in this section, will ideally help to maintain the richness of the information used to generate the knowledge base, but also facilitate overall synergy of data within any aggregate model that is developed.

### *3.2.1 Maintenance of context and data provenance*

Importantly, there is a need to incorporate *metadata*, so that context can be documented and propagated throughout the lineage of the models. For example, there is a need to properly document and archive information regarding the experimental and/or imaging conditions under which the data were collected. This includes information about who collected the original data, when the data were collected, as well as the equipment that was used and the resolution of the data. As data are shared, it is important to identify whether, or to what extent, data were collected or derived. Archiving should include how the data were processed, the tools that were used to process and analyse the data, as well as how and which data were combined to form newly derived data and/or models. Such records should allow us to follow and understand the lineage of all of the data that comprise the knowledge base and/or the aggregate model, at any given time point. Otherwise, the risk of massive information loss arises, which can impact the fidelity of the knowledge base, as well as any activities related to modelling and simulation. The ability to track data and model *provenance* becomes particularly important when creating a knowledge base that compiles iteratively-authored datasets and models.

### *3.2.2 Extendibility, granularity, and the use of growing collections of statistical data*

The ontology should make as few claims as possible about the modelled human anatomy and biomechanics. For practical purposes, the ontology should support the *data-driven* nature of the PHP. It is unreasonable to expect that a rigid ontology can easily accommodate a growing, multi-scale knowledge base. As such, the ontology should be allowed to *evolve* as the supporting datasets grow, and knowledge and understanding progress. In other words, the ontology needs to be *extendible*, so that new conceptual entities, physical instances and parameterizations can be added, as necessary. Specifically, rather than define a rigid framework that only describes the prototypical, healthy human, we need to allow the freedom to specialize and instantiate the ontology, and atlas, as needed. This will allow us to continuously populate and update the atlas as new data are collected and aggregated. One challenge that might exist is the parallel evolution of the ontology and the atlas. Specifically, we want to populate statistics regarding the existence of specific objects and/or features (e.g., bony landmarks, tendon and ligament attachment sites); however, some of those same geometric and spatial features need to be defined, or represented, to determine existence under the framework of the ontology. This might be facilitated by incorporating spatial and geometric properties that do not rely on a pre-defined vocabulary. Overall, population of such an atlas (or atlases) will better quantify and distinguish anomalous, abnormal and/or pathological anatomy, to inform their impact using biomechanical principles.

Additionally, we need to be able to define new terms and relationships that might only exist for a few special cases, or instances, without requiring the revision of existing definitions. This becomes important for improving the overall *granularity* of the ontology. Specifically, as advances in biomedical imaging provide us with finer resolution images, we need to be able to handle and incorporate those lower-level anatomical details. This could present a challenge, as simple words are likely to be insufficient to capture the set of spatial and geometric properties and relationships that are desired in the proposed ontology. Consequently, it might be useful to consider how to best incorporate a common coordinate framework, so that the query language can support standard geometrical and topological concepts. Similarly, there is a desire to include specific tissue properties as they become available. Allowing extensible granularity in the proposed ontology is one feature that we expect will support the development of multi-purpose, multi-scale models. Ultimately, we need to consider how to extend the notion of the ontology to include explicit knowledge that describes a combination of anatomical structure and directions, as well as geometrical concepts.

#### *3.2.3 Automated reasoning to annotate and audit data and models*

Reasoning requires that we access and use background (or pre-existing) knowledge to disambiguate information in appropriate ways. One goal of the proposed ontology is to create the sufficient knowledge and context to facilitate reasoning-based activities. The goal of reasoning will provide at least one way in which to audit the ontology during its development (e.g., are the defined relations sufficient to support automated reasoning?). A suitable structured knowledge base will be useful to annotate data and models using the common vocabulary and defined connectivity (or relations). This includes being able to index and interpret geometric and spatial data to confirm that imported data are indeed human. Similarly, this will serve to confirm that identified geometric and spatial anomalies or abnormalities do indeed exist in an individual model, and are not simply related to a processing and/or analysis error. Hence, automated reasoning can function not only as an internal check for the ontology itself, but also as a tool to prevent pollution of an aggregate model.

## 4. Borrowing from existing anatomical and biomedical frameworks

With the domain-of-interest and anticipated use of the proposed ontology identified, attention can now be focused on the useful ontology work that already exists, which can guide the endeavour. It is always possible that the work someone else has already done can inform the proposed work. In this section we highlight work that could create useful starting points, or at least resources, for the various elements and functionality that we wish to incorporate into the proposed biomechanical ontology.

### 4.1 The standardisation of anatomical terminology

As with many other domains, human anatomy consists of a rich and extensive set of terminology, with numerous terms existing for many of the anatomical entities and features. This can lead to great confusion, particularly when communication occurs between members of fields that use different terms to refer to the same entities. Ultimately, these challenges led to the development of an international standard of anatomical nomenclature, originally created as the *Nomina Anatomica* [5], before revision and expansion to become the *Terminologia Anatomica* (TA) [6]. In a general sense, the TA is a list of terms that designate the anatomical entities that comprise the human body. However, the TA is not arranged as an alphabetical list; rather, it is structured in a hierarchical-like list that helps to enhance the intrinsic meaning of each term, by implying a semantic relationship between terms. In other words, it is a structured vocabulary that is intended to communicate several layers of meaning [7]. In this sense, the structural arrangement of the TA can be likened to that of an ontological framework.

### 4.2 The Foundational Model of Anatomy (FMA)

Based on the terminology and structure of the TA, the FMA has become the seminal computable form of ontology of human anatomy. A special feature of the TA that facilitated its transition to the machine-readable form (the FMA) is the unique, numerical identifier associated with each of the defined anatomical entities. The FMA was designed to provide anatomical information needed by any health education-, research-, or medical practice-based user group. Intended to accommodate any viewpoint, the FMA is a high-level ontology that symbolically represents the ideal or prototypical anatomy to which each individual, and its constituent parts, should conform [8]. This means that it includes things like the (presumed) existence of an organ that might have been surgically removed. Consequently, certain instantiations fall outside of what would be deemed a quantifiable composition.

By making the concept of canonical anatomy explicit, the FMA inherently distinguishes between universal and instantiated anatomy, but avoided the need to explicitly define what is “normal.” Moreover, since the FMA is based on the canonical anatomy of the TA, the entities and relations defined therein are the result of anatomical generalizations that were qualitatively *inferred* from observable anatomical characteristics and features. The completeness and correctness of the TA have also been questioned; for example, noted as missing are a number of morphological features and an important articulation of the lower extremity [9]. Moreover, the spatial concepts that link the various entities are not the quantitative, point-based concepts of classical geometry. Instead they involve rather qualitative relations between the defined objects. Despite these limitations, the FMA does provide a general schema by which anatomical data can be stored in a computable form. Such a framework might allow rudimentary description and comparison of instantiated anatomy, but only at a relatively high-level. The existing upper ontology classes contained within the FMA need to be extended to allow inclusion of lower-level anatomical details, and to effectively create microstructure. This requires refinement of not only the geometrical description of landmark features, but also the inclusion of spatial relationships that quantify the relative locations of the various anatomical features that define structure.

### 4.3 My Corporis Fabrica (MyCF): building upon the FMA

The desire to instantiate human anatomy using patient-specific data has resulted in efforts to incorporate greater granularity and more salient details of anatomical structure. Perhaps most notably, MyCF was developed to extend the structure of the FMA ontology, and to support the addition of topological, geometrical and functional aspects of individualized anatomy [10]. Rather than store a single geometric description for each anatomical entity, MyCF was designed to store multiple instances of the same anatomical entity. Thus, MyCF represents general variability in human anatomy based on the number of instances that exist in the database.

Just how granular MyCF enables the specific features to be geometrically described is somewhat unclear. However, based on its use to facilitate automatic image segmentation, it would appear to include relatively high-level descriptions (e.g., plane, sphere, cylinder, and cone) [11]. It should be possible to incorporate greater detail using geometrically invariant measures, as has been done to automate landmark identification on 3D bone models [12]. Specifically, measures of concavity and convexity (e.g., peak, ridge, pit, and ravine) were used to accomplish lower-level feature extraction, coupled with the use of a spatial adjacency matrix to define the relative landmarks with respect to each other. Nonetheless, methods exist to describe the more salient details of anatomical structure, and its variability, and could greatly improve the utility of existing ontologies of anatomy. These include a rich collection of morphometric tools and methods that are commonly used for biological shape analysis and feature extraction in the fields of physical anthropology and forensics [13]. Another critical addendum, and a goal of the proposed ontology, is to use such instantiated anatomy to inform and populate a probabilistic atlas. Ideally, this will allow us to better characterize spatial and geometric sources of variability, so that we can move towards quantifying what is "normal," prototypical, and/or representative human anatomy.

Importantly, we have begun to conceptualize how to extract and incorporate quantitative, lower-level spatial and geometric details that define anatomical structure (Figure 1). Current ontologies do not include such detailed descriptions of anatomical features. As such, existing ontologies of anatomy fail to capture the salient features that define the full structural richness and complexity of which the various entities are comprised. By including greater detail, we hope to move toward replicating the knowledge base that, for example, allows a physical anthropologist to distinguish the various bones, not only from one another, but between species or even gender.

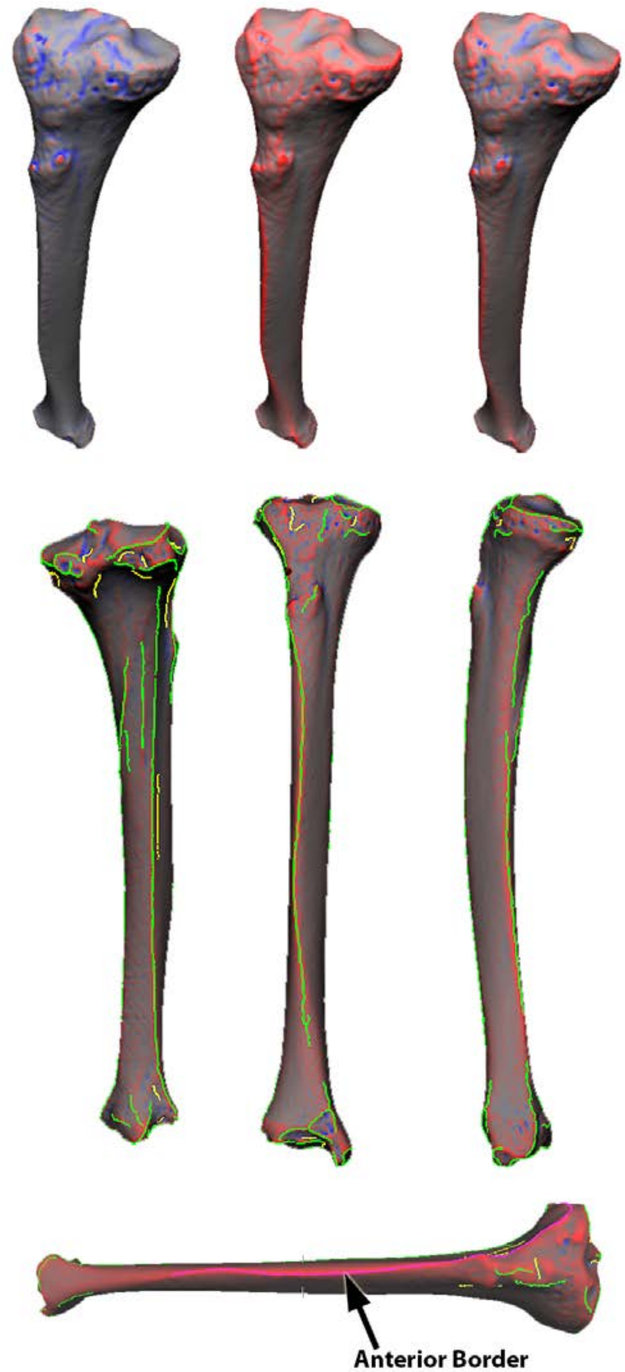


**Figure 1. The process of feature identification, extraction and annotation. (Top) Curvature analysis of the tibia to identify the locations of max, min, and mean curvature. (Middle) Feature extraction based on the curvature analysis. (Bottom) Manual or automated annotation of the anatomical features that define the structure of the tibia.**

Another aspect related to anatomical variability is that of function, particularly as it pertains to mechanical behaviours that become apparent during the performance of daily physical activities (e.g., walking). On this front, MyCF can produce not only a list of the anatomical entities required to create a patient-specific model, but also the mechanical parameters that are needed to perform biomechanical simulations. To facilitate this utility, the definition of certain entities (e.g., muscles) includes a specific classification based on its biomechanical action or function [14]. For example, the *rectus femoris*, a muscle that contributes to bend the knee, is given a new taxonomic relation that defines it to be *involved_in* knee flexion. Conceptually, such descriptions are of specific interest for the construction of the proposed ontology; however, their practical incorporation might not be so simple.

Certainly, there is some debate regarding how best to classify anatomical entities based on function, in a manner that enriches the organization of semantic content [15]. In part, many anatomical entities serve multiple functions, in multiple body systems; thus, function can vary according to the frame of reference. As it pertains to biomechanics, the function or line of action of a muscle can vary according to joint posture. For broad, fan-shaped muscles comprised of complex architectures, not all fascicles have the same line of action, and thus can vary in function even within a specific posture [16–18]. In these cases, detailed models of muscle structure must be combined with simulation to determine the individual, and potentially changing, contributions of the fascicles to specific activities or motions [19]. In other words, the granularity of the spatial and geometric characteristics is but one aspect that is likely to affect the interpretation of function. Additionally, based on biomechanical principles, force propagation between segments means that muscles can actually contribute to actions at joints that they do not even cross [20]. Thus, anatomical structure and location are not necessarily sufficient to correctly, or completely, infer overall biomechanical function. These types of challenges must be considered while establishing the framework of the proposed ontology.

### 4.4 Probabilistic atlases of anatomy

Some of the most extensive work to construct a multi-scale probabilistic atlas of human anatomy has focused specifically on neuroanatomy, and the desire to map the overall structure of the brain [21]. The ultimate vision the International Consortium for Brain Mapping (ICBM) is to create a representative atlas of the entire human species, while retaining information about the magnitude of biological variability in a quantitative manner. The statistical digital atlas supports visualization of the structure of the brain on multiple scales, but is also capable of generating statistical responses to user queries. Thus, it is possible to define and visualize specific subsets of the entire population. Great progress has been made on this front, resulting in an improved ability to automatically segment, register, and annotate 3D models. Additionally, a quantitative definition of a “normal, average brain” now exists, facilitating research of the structural variability between genders, as well as that which occurs with ageing, or in individuals with cognitive or psychological disorders.

Endeavours such as that of the ICBM have generated ventures to compile statistical atlases of other specific human anatomy (e.g., heart, liver, bones). Of particular interest are projects that relate to the PHP’s growing catalogue of skeletal and muscle data. There are efforts to create and analyse statistical atlases of bony anatomy, based on large collections of computed tomography data [22–24]. Using registration techniques to establish point-based correspondences between subject-specific meshes, the anatomical modes of variation are determined, and the atlas constructed. Similar to the atlases being generated of the intricate fibre trajectories within the brain [25], there is also ongoing work to map the fibre orientation distribution of the myocardium [26]. Projects like these are of particular relevance to the PHP, given the growing catalogue of skeletal and muscle data now being generated within the consortium.

Importantly, we have only highlighted some specific anatomy-based applications for which statistical atlases have been built and used. However, given the purpose of the current paper, it is worthy to note that these atlases are typically comprised of three principal components. The first is a controlled vocabulary, or ontology, that is used to unify and provide a standard structure to the compiled data. The ontology, in turn, supports the remaining two components. Typically, there is a representative dataset (or individual) that is used to define the spatial extent and coordinates of the anatomy of interest. There is also a mapping between the representative and instantiated anatomy. Thus, while not always explicitly described in the literature, any probabilistic atlas requires a working ontology for users to realize the true utility of the atlas.

### 4.5 Ontology of Physics for Biology (OPB)

The OPB is being constructed to declaratively represent the formal structure of systems dynamics theory and thermodynamics, as they relate to biological processes [4]. As such, the OPB supports annotation of the biophysical content of biomedical datasets and analytical models, to facilitate knowledge aggregation and inform simulations. The OPB supplements existing ontologies for biological entities (e.g., molecules, cells, organs), and allows physical properties (e.g., energies, volumes, flow rates) to be assigned to those entities. It is also possible to establish dependency between those physical properties, to capture the spatial and temporal scales of specific biophysical processes. Overall, the intent of the OPB is not to represent physical “reality,” rather to represent the concepts and laws that provide the basis to *quantitatively explain* how the biological world works.

The specific aim of the OPB closely parallels that of the proposed biomechanical ontology, making it a valuable resource. Both the OPB and the proposed ontology share the concept of building a physics-based framework to support ontologies of biological content. Using a similar framework will allow us to define the physical properties associated with each of the anatomical entities included within the atlas (e.g., constitutive tissue properties, musculoskeletal mechanics, muscle contraction dynamics). In this way, we can semantically resolve and map the biomechanical content contained in the ontology of human anatomy. This will help to inform individual model development and simulations, as well as help to unify multi-scale components and characteristics into aggregate, whole-body human models.

## 5. Conclusion

Effective communication and knowledge sharing are central to collaborative research efforts, and can be facilitated by a common framework around which a formal working knowledge base can be structured. The current paper serves to highlight the utility of ontologies, and to propose a unifying foundational theory (i.e., biomechanics) on which to base a working ontology to serve the PHP group. Additionally, we identified the characteristics that the desired framework must possess, as well as numerous existing resources that provide useful starting points for the proposed ontology. The intent was to formalize and share these ideas and concepts, and inform the consortium of how we intend to approach this crucial component of developing a probabilistic atlas.

As a research consortium, it is important that we work together, not only to compile the common knowledge base, but also to inform how it should be structured, so that the needs of the community are met. On this front, the intent is to work directly with ontologists to develop a utilitarian ontology that will support individual- and consortium-level goals and initiatives. Just as we are not experts when it comes to ontology development, ontologists will not necessarily comprehend the specific applications that the proposed framework is meant to support. Hence, we promote exchange of ideas, not only within the context of data and knowledge sharing, but also related to the development of the infrastructure that will facilitate such sharing. As the ontology grows and evolves, consortium member-based testing will enable users to guide ontology revisions and additions. Ultimately, this will allow us to identify ways to improve how we store and represent data, information, and knowledge, while maintaining the richness of the syntactic and semantic content.

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