

Definitions in Biomedical Ontologies: The Method ONTODEF

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Submission: 📅 March 04, 2022

Published: 📅 April 14, 2022

Volume 3 - Issue 4

How to cite this article: Amanda Damasceno de Souza¹ and Mauricio Barcellos Almeida. A Systematic Overview of Bioelectronic Medicine and its Impact on Medical Technology. J Biotech Biores. 3(4). JBB. 000570. 2022.

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Abstract

Definitions are very important devices to build useful biomedical ontologies. The formulation of formal definitions should follow Aristotelian tennets. This paper aims to describe OntoDef, a method for the creation of suitable definitions for ontologies. The main contribution of the OntoDef method is to make explicit the identity of entities related to biomedical terms under definition, in establishing necessary and sufficient conditions.

Keywords: Definitions; Textual definitions; Biomedical ontology; Genus; Differentia; Ontodef method

Introduction

Textual definitions in ontologies are essential for the consistent use of specialized terms in interdisciplinary communities. Well-formulated definitions ensure consistency of human reasoning and support computational reasoning. These are requirements for an ontology to participate in biomedical repositories, such as the OBO Foundry [1-4].

In this article, the application of the OntoDef method for formulating textual definitions in biomedical ontologies is introduced and demonstrated. The method consists of systematizing principles to create definitions in natural language. It is developed within Information Science research and based on Aristotelian principles to define [5,6].

Principles to Elaborate Definitions

For Aristotle, each species of a “Genus” has a unique “Differentia” [7]. For example, the species “man” comprises the genus “animal” and a Differentia concerning other animals. Genus and Differentia define a term by providing the essence of an entity [8]. An Aristotelian definition is expressed in the form “ $\alpha = \text{def. } \beta$ ”, where α is the specie and β is the combination of the Genus + Differentia correspondent to α [7,9].

Principle of the unique inheritance

In ontologies, definitions are arranged hierarchically so that a term must not have two parents, that is, entities of a direct superior hierarchy. In having two parents we will incur multiple inheritance, which is not suitable for ontological definitions [5].

Principle of necessary and sufficient conditions

If a' is a necessary condition for an entity to be of type A, then every entity that is of type A will have the condition a' [8].

Principle of non-circularity

One should not define the term recursively; for example, laboratory examination is one performed in a clinical laboratory [10].

Principle of Intelligibility

The definition must use intelligible terms that are easy to understand so that the definition is not restricted to domain experts [5,10].

Applying Ontodef in Biomedical Ontologies

The OntoDef method is depicted in Figure 1. A full example of the OntoDef method to define a real term is presented in Table 1.

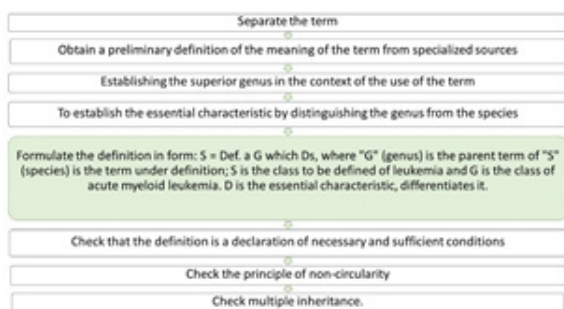


Figure 1: Algorithm for applying the OntoDef method. Source: Adapted from Souza AD [5] Souza AD et al. [6].

Table 1: Formulation of definition for the term “Acute erythroid leukemia” in Blood Ontology using OntoDef

OntoDef	Definition in Blood Ontology
1) Separate the term	Acute Erythroid Leukemia (AEL)
2) Obtain a preliminary definition	... is characterized by a immature erythroid population.
3) Establishing the superior genus	AEL <is-a> Acute Myeloid Leukemia (AML)
4) To establish the essential characteristics	Predominant immature erythroid population
5) Formulate the definition in form: S = Def. a G	AEL <is-a> AML with immature erythroid population.
6) Check necessary and sufficient conditions	Being an Acute erythroid leukemia is a necessary condition to be “characterized by the predominant presence of immature erythroid cells”. Being an erythroid leukemia s a sufficient condition to be “characterized by the predominant presence of immature erythroid cells”.
7) Check circularity	It is not circular
8) Check multiple inheritance	AEL <is-a> AML , both inheriting from myeloid lineage.
Textual definition to be formalized	An AEL is an acute myeloid leukemia characterized by a predominant immature erythroid population.

Final Remarks

The application of the OntoDef method brought a relevant contribution by addressing gaps in the way of creating definitions in ontologies. The method provides a view of the entity’s essence by establishing the necessary and sufficient conditions through methodological steps that condition the ontology creator to a reflection on the definition process.

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