

**EDITORIAL:****DISEASE ONTOLOGY**SC Mohapatra<sup>1</sup> and Meghkanta Mohapatra<sup>2</sup>

The term *ontology* is originally used in philosophy and has been applied in many different subjects thereafter. It is branch of metaphysics that studies the nature of existence. The word element *onto-* comes from the Greek "being". Thus Ontology means "science of being". From philosophy ontology is an explicit formal specification of how to represent the objects, concepts and other entities that are assumed to exist and the relationships they have with each other. When the knowledge about a domain is represented in a declarative language, the set of objects are called the universe of discourse. We can describe the ontology of a program by defining a set of representational terms. Definitions associate the names of entities in the population of discourses (e.g. classes, relations, functions or other objects).

Formally, an ontology is the statement of a logical theory. We say that an agent commits to an ontology if its observable actions are consistent with the definitions. The idea of ontological commitment is based on the "Knowledge-Level perspective". In case of disease it makes sense that the 'Science of being a disease'. The core meaning within computer science is a model for describing the world that consists of a set of types, properties, and relationship. There is also generally an expectation that the features of the model in an ontology should closely resemble the real world (related to the object). What may ontologies have in common in both computer science and in philosophy is the representation of entities, ideas, and events, along with their properties and relations, according to a system of categories. In both fields, there is considerable work on problems of ontological relativity and concerning debates. Other fields' make assumptions are sometimes explicitly not biologically plausible e.g. statistical models. Thus elaborated and explored for ontology of disease need to be established more strongly. Such concerns intersect with those of information science when a simulation or model is intended to enable decisions in epidemiology to determine risk management. Some claim all social sciences have explicit ontology issues also.

The traditional goal of ontological inquiry in particular is to divide the world "at its joints" to discover those fundamental categories or kinds into which the world's objects naturally fall. During the second half of the 20th century, philosophers extensively debated the possible methods or approaches to building ontologies. By contrast, computer scientists were building some large and robust ontologies such as WordNet and Cyc, with comparatively. Since the mid-1970s, researchers in the field of artificial intelligence in the early 1990s, the widely cited Web page and paper "Toward Principles for the Design of Ontologies Used for Knowledge Sharing" by Tom Gruber is credited with a deliberate definition of *ontology* as a technical term in computer science. Gruber introduced the term to mean a specification of a conceptualization: An ontology is a description of the concepts and relationships that can formally exist for an agent or a community of agents.

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According to Gruber (1993): Ontologies are often equated with taxonomic hierarchies of classes, class definitions, and the subsumption relation, but ontologies need not be limited to these forms. Ontologies are also not limited to conservative definitions- that is, definitions in the traditional logic sense that only introduce terminology and do not add any knowledge about the world. To specify a conceptualization, one needs to state axioms that do constrain the possible interpretations for the defined terms.

### What is an Ontology?

This definition was originally proposed in 1992 "Ontology by definition is a branch of metaphysics concerned with the nature and relations of being a particular theory about the nature of being or the kinds of things that have existence an ontology is a specification of a conceptualization. The word "ontology" seems to generate a lot of controversy in discussions. It is also often confused with 'epistemology', which is about knowledge and knowing. In the context of knowledge sharing, means a *specification of a conceptualization*. What is important is how an ontology is designed. It is for the purpose of enabling knowledge sharing and reuse. For pragmatic reasons, we choose to write an ontology *as a set of definitions of formal vocabulary*. It has some nice properties for knowledge sharing among software (e.g., semantics independence).

### Ontologies as a specification mechanism:

A subject of formally represented knowledge is based on a conceptualization. A conceptualization is an abstract, simplified view of the world that we wish to represent for some purpose. An ontology is an explicit specification of a conceptualization and a systematic account of existence. When the knowledge of a domain is represented in a declarative formalism, the set of objects that can be represented is called the universe of discourse. This set of objects, and the describable relationships among them, are reflected in the representational vocabulary with which a knowledge-based program represents knowledge. We can describe the ontology of a program by defining a set of representational terms.

We use common ontologies to describe ontological commitments for a set of agents so that they can communicate about a domain of discourse without necessarily operating on a globally shared theory. We say that an agent **commits** to an ontology if its observable actions are consistent with the definitions in the ontology. The idea of ontological commitments is based on the Knowledge-Level perspective as started Newell (1982). Pragmatically, a common ontology defines the vocabulary with which queries and assertions are exchanged among agents. Ontological commitments are agreements to use the shared statements in a coherent and consistent manner. The agents sharing a description need not render a knowledge base, and an agent that commits to an ontology is not required to answer all queries. In short, a commitment to a common ontology is a guarantee of consistency, but not completeness, with respect to queries and assertions using the vocabulary defined in the ontology. Most ontologies describe individuals (instances), classes (concepts), attributes.

The disease ontology considers individuals, instances, objects (the basic or "ground level" objects). **Classes**: sets, collections, concepts, classes in programming, types of objects, or kinds of things. **Attributes**: aspects, properties, features, characteristics, or parameters that objects (and classes) etc those can have effect on

causation or not causation of disease. Thus it is a prerequisite of epidemiology of tomorrow. However, it has complex computer application for ontological modeling.

## TYPES

- (a) **Domain ontology:** A domain ontology (or domain-specific ontology) represents concepts which belong to part of the world. Particular meanings of terms applied to that domain are provided by domain ontology. For example the word *card* has many different meanings. An ontology about the domain of poker would model the "playing card" meaning of the word, while an ontology about the domain of computer hardware would model the "punched card" and "video card" meanings. Since domain ontologies represent concepts in very specific and often eclectic ways, they are often incompatible. Different ontologies in the same domain arise due to different languages. At present, merging ontologies that are not developed from common foundation ontology. Domain ontologies use the same foundation ontology to provide a set of basic elements with which specific meanings of the elements can be merged automatically. There are studies on generalized techniques for merging ontologies, but this area of research is still largely theoretical.
- (b) **Upper ontology:** An upper ontology (or foundation ontology) is a model of the common objects that are generally applicable across a wide range of domain ontologies. It usually employs a core glossary that contains the terms and associated object descriptions as they are used in various relevant domain sets. There are several standardized upper ontologies available for use, including BFO, Dublin Core, GFO, OpenCyc/ ResearchCyc, SUMO, etc. However, it has been employed as a linguistic tool for learning domain ontologies.
- (c) **Hybrid ontology:** The Gellish ontology is an example of a combination of an upper and a domain ontology.
- (d) **Visualization:** A survey of ontology visualization techniques is presented by Katifori et al. An evaluation of two most established ontology visualization techniques: indented tree and graph is discussed in.
- (e) **Ontology Engineering:** Ontology engineering (or ontology building) is a subfield of knowledge engineering. It studies the ontology development process, the ontology life cycle, the methods and methodologies for building ontologies, and the tool suites and languages that support them. Ontology engineering aims to make explicit software applications, and within enterprises and business procedures. It offers a direction towards solving the interoperability problems brought about by semantic obstacles, such as the obstacles related to the definitions of business terms and software classes. Ontology engineering is a set of tasks related to the development of ontologies for a particular domain.

- (f) **Ontology Learning:** Ontology learning is the automatic or semi-automatic creation including extracting a domain's terms from natural language text. As building ontologies manually is extremely labor-intensive and time consuming, there is great motivation to automate the process.
- (g) **Ontology Languages:** An ontology language is a formal language used to encode. There are a number of such languages for ontologies, both proprietary and standards-based. Common Algebraic Specification Language is a general logic-based specification language developed, and functions as a de facto standard in the area of software specifications. It is now being applied to ontology specifications in order to provide modularity and structuring mechanisms. Common logic is ISO standard 24707. Developing Ontology-Grounded Methods and Applications (DOGMA) adopts the fact-oriented modeling approach to provide a higher level of semantic stability.

The Gellish language includes rules for its own extension and thus integrates an ontology with an ontology language. IDEF5 is a software engineering method to develop and maintain usable, accurate, domain ontologies. KIF is a syntax for first-order logic that is based on S-expressions. MOF and UML are standards of the OMG. OBO, a language used for biological and biomedical ontologies. OntoUML is an ontologically well-founded profile of UML for conceptual modeling of domain ontologies. OWL is a language for making ontological statements, developed as a follow-on from RDF and RDFS, as well as earlier ontology language projects including OIL, DAML and DAML+OIL. OWL is intended to be used over the World Wide Web, and all its elements (classes, properties and individuals) are defined as RDF resources, and identified by URIs. Rule Interchange Format (RIF) and F-Logic combine ontologies and rules. Semantic Application Design Language (SADL) captures a subset of the expressiveness of OWL, using an English-like language entered via an Eclipse Plug-in. SBVR (Semantics of Business Vocabularies and Rules) is an OMG standard adopted in industry to build ontologies. TOVE, (Toronto Virtual Enterprise project).

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