

New Era of Developing and Using Ontologies for Microbiology Research and Diagnosis

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Originated from philosophy, the term ontology is used in bioinformatics field to refer to a representative vocabulary that describes the knowledge of some domain, typically a commonsense knowledge domain [1]. The term *formal ontology* used in biomedicine today typically means the definition of sets of terms and relations including logical assumptions and inference machinery [2]. Biomedical (formal) ontologies have been used intensively. For example, the Gene Ontology (GO) as a tool for unification of biology was first published in 2000 [3]. As of May 16, 2012, a search of “Gene Ontology” in PubMed retrieved more than 4000 papers. GO plays an important role in interpreting biological data to rational biological meanings. For instance, the GO annotations have served as a backbone for current OMICS data analysis. Currently over 300 biomedical ontologies have been stored in the Biportal ontology repository of the National Center for Biological Ontology (NCBO): <http://biportal.bioontology.org/ontologies>.

To facilitate integrative research in infectious diseases, a community-based Infectious Disease Ontology (IDO) has been developed [4]. IDO is aimed to include a suite of interoperable ontologies that jointly cover the entire infectious disease domain, spanning infectious disease specialties and the clinical care, public health, and biomedical research. Under the framework of IDO, we have recently developed Brucellosis Ontology (IDOBURU), the first reported bacterial infectious disease ontology [5]. Brucellosis is a zoonotic infectious disease caused by intracellular facultative Gram-negative bacteria *Brucella* spp. As a brucellosis knowledgebase, IDOBURU represents different aspects of brucellosis, including host infection, zoonotic disease transmission, symptoms, virulence factors and pathogenesis, diagnosis, intentional release, vaccine prevention, and treatment [5]. As a formal ontology developed using the Web Ontology Language (OWL) [6], IDOBURU provides both machine-readable and human-readable vocabulary and supports computer-assisted automated reasoning. For example, based on biological knowledge captured within the ontology, simple scripts can be developed to query IDOBURU and identify 229 *Brucella* virulence factors, 29 protective antigens, and one protein that is both virulence factor and protective antigen [4]. IDOBURU also includes different *Brucella* diagnostic methods such as specimen culturing, immunoassays, and Polymerase Chain Reaction (PCR) assays. These methods are organized in logical and specifically defined hierarchies. As an example, a specific PCR assay in IDOBURU includes the assayed *Brucella* species, gene name and information for obtaining gene sequence, PCR primers and product details, the specimen used for diagnosis, and the assertion to make a diagnosis based on a PCR result. These details are represented logically and interlinked with computer-understandable relations. Such a meta data design can be used to represent various PCR assays. The IDOBURU knowledge base is also integrated with other biomedical ontologies such as GO, IDO-core, and other specific IDO-core extensions. Such an integrative approach supports complex data exchange, data integration, and automated reasoning.

The new era of developing and using biomedical ontologies in microbiology research and diagnosis has just started. The preliminary results out of IDOBURU and other biomedical ontologies have demonstra-

ted their significant power and novelty of integrating data and creation of knowledge in the field of microbiology. More efforts are required for further development and applications of biomedical ontologies.

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