Tutorial-style Workshop: Ontological Realism for Biomedical Ontologies and Electronic Health Records

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Abstract. Three internationally recognized experts in biomedical ontology will offer a tutorial style workshop to demystify the principles of Ontological Realism as they are applied in the Open Biomedical Ontology Foundry. The goal is to raise awareness about the value of these principles on top of formal languages and to improve skills in building, evaluating and using biomedical ontologies.

Keywords. Ontologies, Ontological Realism, Electronic Health Records

Background

Ontological Realism is a methodology for ontology development which combines elements from (1) scientific realism, i.e. the doctrine according to which scientific theories are (broadly) true accounts of reality, and (2) metaphysical realism which states that observer-independent types of entities (also called universals) exist [1]. The methodology invites ontologists to conceive the world as including entities of two sorts, respectively called 'particulars' and 'universals'. Particulars are concrete individual entities (entities that exist in space and time and that exist only once); universals are to be understood as repeatable types, which means that for each universal we can discover many particulars, which are its instances.

The distinction between universals and particulars accounts for the division of labor between - respectively - realism-based biomedical ontologies (e.g. from the OBO foundry) [2] on the one hand, and clinically relevant particular entities referred to by the EHR with emphasis on the Referent Tracking methodology [3] on the other hand. The OBO Foundry seeks to be a collection of high quality ontologies that follow the principles of Ontological Realism and describe domains relevant for biomedicine and the life sciences thereby resorting to the Basic Formal Ontology (BFO) [4] and the Relation Ontology [5]. Currently 75 projects are using BFO for this goal (www.ifomis.org/bfo/users), examples being the Gene Ontology and the Ontology of General Medical Science. Referent Tracking provides a framework for the unambiguous representation of what is observed on the side of the patient and of the state-

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ments that are made about such findings, for example in the context of adverse events [6] or substance intolerances [7].

Goal of the workshop

Despite the success of the methodology, a number of critiques have recently been published [8-10]. A common element in these critiques, as discussed in [1] and several forthcoming commentaries on [10], is (1) the *perceived* complexity of the methodology for researchers not familiar with basic notions of ontology from a philosophical perspective, and (2) the overestimation of the power of description logics when the semantics of languages such as OWL is not well understood. The former leads to inconsistent ontological commitment [11] while the latter to ontologies whose axioms constitute a huge source for unintended models, rendering logic-based reasoning unreliable [12].
The learning objective of this tutorial style workshop is to demystify the methodology and provide practical examples of how it should be applied in building or evaluating ontologies or reporting adverse events from EHR data.

Format and speakers

The three speakers have extensive experience in realism-based ontology design, evaluation and application and have given several lectures and tutorials on the topic.

Tutorial components:

- ***Ontology and ontologies*** (5 min. - Werner Ceusters): clarifying the distinction between Ontology as scientific discipline and ontologies as representational artifacts [1];
- ***Ontological Realism & ontologies*** (25 min. - Mathias Brochhausen): Ontological Realism [1] as basis for upper ontologies, focusing on Basic Formal Ontology [4]. Entities and relations in biomedical ontologies as recognized by Ontological Realism (e.g. ACGT [13]), and discussion of the quality criteria for OBO Foundry Ontologies [2, 14];
- ***Ontology authoring and evaluation*** (30 min. - Stefan Schulz): application of ontological realism in detecting and avoiding common mistakes in formal representations, taking examples from SNOMED CT [11], the NCI Thesaurus [12], and OBO;
- ***Ontological Realism and EHRs*** (20 min. - Werner Ceusters): how to improve EHR data by means of ontologies [3], using problem lists, diagnoses [15] and adverse event reporting as examples [6];
- ***Roundup and future collaborations*** (10 min. - all): final question answering and plans for future collaboration with audience.

The papers referenced in 'Tutorial components' will serve as background reading and are available on http://www.referent-tracking.com/RTU/?page=classes.phtml. Attendees are invited to send questions to the tutorial organizers up to one month prior to the event. These questions will be explicitly addressed in the tutorial.

Expected achievements and outcomes

After the tutorial, the audience will be able to (1) understand better the added value of the Ontological Realism principles over mere computational and logical frameworks, (2) apply the principles to build or evaluate ontologies, (3) assess how to optimally use such ontologies in EHR systems, and (4) make recommendations to clinicians and biomedical informaticians to improve the systems they are working with.

Statement of participation

All speakers are committed to register as conference participants.

References

[1] Smith B, Ceusters W. Ontological Realism as a Methodology for Coordinated Evolution of Scientific Ontologies. Applied Ontology. 2010;5(3-4):139-88.

[2] Smith B, Ashburner M, Ceusters W, Goldberg L, Mungall C, Shah N, et al. The OBO Foundry: coordinated evolution of ontologies to support biomedical data integration. Nature Biotechnology. 2007;25:1251-5.

[3] Ceusters W, Smith B. Strategies for Referent Tracking in Electronic Health Records. Journal of Biomedical Informatics. 2006 June;39(3):362-78.

[4] Bittner T, Smith, B. . Normalizing Medical Ontologies Using Basic Formal Ontology. Tagungsband der 49 Jahrestagung der Deutschen Gesellschaft für Medizinische Informatik, Biometrie und Epidemiologie. 2004.

[5] Smith B, Ceusters W, Klagges B, Köhler J, Kumar A, Lomax J, et al. Relations in biomedical ontologies. Genome Biology. 2005;6(5):R46.

[6] Ceusters W, Capolupo M, Moor GD, Devlies J, Smith B. An Evolutionary Approach to Realism-Based Adverse Event Representations. Methods of Information in Medicine. 2011;50(1):62-73.

[7] Hogan W. Towards an Ontological Theory of Substance Intolerance and Hypersensitivity. Journal of Biomedical Informatics. 2011;44(1):26-34.

[8] Lord P, Stevens R. Adding a Little Reality to Building Ontologies for Biology. Plos ONE. 2010;5(9):e12258.

[9] Dumontier M, Hoehndorf R. Realism for scientific ontologies. Proceedings of the 6th International Conference on Formal Ontology in Information Systems (FOIS). 2010:387-99.

[10] Maojo V, Crespo J, Garcia-Remesal M, Iglesia Ddl, Pérez-Rey D, Kulikowski C. Biomedical Ontologies: Toward Scientific Debate. Methods of Information in Medicine. 2011;50(3):in print.

[11] Schulz S, Cornet R. SNOMED CT's Ontological Commitment. In: Smith B, editor. ICBO: International Conference on Biomedical Ontology. Buffalo NY: National Center for Ontological Research; 2009. p. 55-8.

[12] Schulz S, Schober S, Tudose I, Stenzhorn H. The Pitfalls of Thesaurus Ontologization - the Case of the NCI Thesaurus. AMIA Annual Symposium Proceedings. Washington D.C.: AMIA; 2010. p. 727-31.

[13] Brochhausen M, Spear A, Cocos C, Weiler G, Martín L, Anguita A, et al. The ACGT Master Ontology and its applications - Towards an ontology-driven cancer research and management system. Journal Biomedical Informatics. 2011;44(1):8-25.

[14] Smith B, Brochhausen M. Putting biomedical ontologies to work. Methods of Information in Medicine. 2010;49(2):135-40.

[15] Scheuermann RH, Ceusters W, Smith B. Toward an Ontological Treatment of Disease and Diagnosis. Proceedings of the 2009 AMIA Summit on Translational Bioinformatics, San Francisco, California, March 15-17, 2009: American Medical Informatics Association; 2009. p. 116-20.