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Creating a Translational Medicine Ontology

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Abstract

We, participants in the Translational Medicine Ontology activity of the World Wide Web Consortium's Health Care and Life Sciences Interest Group (http://esw.w3.org/topic/HCLSIG) and members of the National Center for Biomedical Ontology (http://bioontology.org/), are developing a high-level, patient-centric ontology for translational medicine which will draw on existing domain ontologies and allow the integration of data throughout the drug development process.

Introduction

The pharmaceutical industry has historically focused on the development of novel blockbuster drugs. There is now an increasing focus on personalized medicines, requiring the right patients to receive the right drug at the right dose. In order to develop a tailored drug, manufacturers need to identify biomarkers that will indicate how a given patient will respond to a particular treatment. Biomarkers can also be used to demonstrate the comparative effectiveness of drugs, which is increasingly required by payers. Such translational medicine strategies require that traditionally separate data sets from early drug discovery through to patients in the clinical setting be integrated, and presented, queried and analyzed collectively. Ontologies can be used to drive such data integration and analysis, however, at present few ontologies exist that bridge genomics, chemistry and the medical domain.

The Translational Medicine Ontology, an application ontology that bridges the diverse areas of translational medicine, draws on existing domain ontologies where appropriate and will provide a framework centered on less than 50 types of entities.

Goals

The Translational Medicine Ontology will facilitate data integration from diverse areas of translational medicine such as discovery research, hypothesis management, formulation, clinical trials, and clinical research. It will serve as a template for further ontology development, enabling scientists to answer interesting and currently difficult questions more easily, especially those about data that are typically hosted by different functional areas. The ontology will provide a framework for the modeling of patient-centric information, which is essential for tailoring drugs.

Methodology

We have identified a set of 17 roles played by people across health care and the life sciences and collected (1) relevant questions, (2) the entities that those questions involve, and (3) applicable extant domain ontologies.[1] Types of entities include: disease, drug, patient, target, gene, risk, pathway, population, compound, phenotype, and treatment.

Next steps will involve identifying use cases based on those questions, determining which entities to build into the ontology and aligning them with BFO,[2] an upper-level ontology, to aid interoperability between domain ontologies. We will use one use case to test the Translational Medicine Ontology by building a data integration application based on it.

Conclusion

This project seeks to develop a patient-centric application ontology for translational medicine, as a collaborative effort between groups in industry and academia. The presentation will highlight our methodology, work to date, and future steps.

1. http://esw.w3.org/topic/HCLSIG/PharmaOntology/Roles
2. http://www.ifomis.org/bfo

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