Translational Medicine Ontology: A Patient-Centric Ontology for Drug Development and Clinical Practice

Patricia L. Whetzel (B.Sc., Ph.D.)1, Colin Batchelor (M.Chem., D.Phil.)2, Christine Denney (B.Sc.)3, Elgar Pichler (M.Sc., Ph.D.)4, Bosse Andersson (B.Sc., M.Sc.)5, Olivier Bodenreider (B.Sc., Ph.D.)6, Tim Clark (B.Sc., Ph.D.)7, Christopher Domarew (MBChB, PharmD, RPh)8, Michel Dumontier (B.Sc., Ph.D.)9, Anja Jentzsch (B.Sc.)10, Vipul Kashyap (B.Sc., Ph.D.)11, Julia Kozlovsky (B.Sc., M.Sc.)12, Joanne Luciano (B.Sc, M.Sc., Ph.D.)12, Chimezie Oguji (B.Sc.)13, Matthias Samwald (B.Sc., Ph.D.)14, Jun Zhao (B.Sc., Ph.D.)15, Susie Stephens (B.Sc., Ph.D.)16

1Stanford University, Stanford, CA, USA; 2Royal Society of Chemistry, Cambridge, UK; 3Eli Lilly, Indianapolis, IN, USA; 4AstraZeneca, Waltham, MA, USA; 5AstraZeneca, Lund, Sweden; 6National Library of Medicine, Bethesda, MD, USA; 7Harvard Medical School, Cambridge, MA, USA; 8Warrington Hospital, Warrington, UK; 9Carleton University, Ottawa, ON, Canada; 10Free University Berlin, Berlin, Germany; 11Cigna, Hartford, CT, USA; 12Predictive Medicine, Inc., Belmont, MA, USA; 13Cleveland Clinic, Cleveland, OH, USA; 14DERI Galway, Galway, Ireland & KLI Austria; 15University of Oxford, Oxford, UK; 16Johnson & Johnson, Radnor, PA, USA

Abstract
The aim of personalized medicine is for patients to receive the right drug at the right time and at the right dose. To achieve this aim, traditionally separate data sets, in drug discovery, hypothesis management, formulation, clinical trials and clinical research need to be integrated, queried and analyzed collectively to identify biomarkers necessary to tailor pharmaceuticals. Ontologies, a formal representation of domain knowledge, can drive data integration; however, at present few ontologies exist that bridge genomics, chemistry and medicine. We present a high-level, patient-centric ontology to facilitate integration of data relating to translational medicine and describe prototype applications within both pharmaceutical research and clinical practice. The ontology has been developed by participants in the World Wide Web Consortium’s Semantic Web for Health Care and Life Sciences Interest Group and members of the National Center for Biomedical Ontology.

Introduction
The Translational Medicine Ontology (TMO) is an application ontology that draws on existing open domain ontologies and provides a framework to describe patient-centric information.

Methods
We identified roles played by people across the fields of healthcare and life sciences, collected questions that people in these roles typically ask, and identified relevant domain ontologies1 that can be used to formulate these questions. This guided our initial selection of classes within the TMO. We developed drug discovery and clinical practice use cases to help refine the ontology2. The TMO has been aligned with the Basic Formal Ontology3, an upper-level ontology, to facilitate interoperability with other ontologies.

Results
The TMO is available under the Creative Commons 3.0 BY license for download.

Discussion
We have developed an initial version of a patient-centric application ontology for translational medicine, in a collaborative effort between industry and academia. It will serve as a framework for enabling scientists to more easily answer interesting but difficult questions, especially those that span currently unconnected data ‘silos’. The presentation will highlight our methodology, work to date, and future steps.

References
1. http://esw.w3.org/topic/HCLSIG/PharmaOntology/Roles
2. http://esw.w3.org/topic/HCLSIG/PharmaOntology/UseCases