Development of a BFO-Based Informed Consent Ontology (ICO)

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***Abstract* —** An Informed Consent Ontology (ICO) was developed to support informed consent data integration and reasoning. ICO is aligned with the Basic Formal Ontology (BFO), and logically represent the terms and their relations related to informed consent processes and contents. ICO contains 471 terms including 137 ICO-specific terms and the other terms imported from existing reliable ontologies. The ontology is available at http://ico-ontology.googlecode.com/.

Keywords— Informed consent; ontology; ICO; OBO Foundry, Basic Formal Ontology (BFO), OBI ontology

I. INTRODUCTION

The informed consent process is one of the fundamental pillars of human research [1]. A human subject research study must undergo informed consent process, which cannot be conducted unless approved by a regulatory body. The entire informed consent process involves giving a subject adequate information of the study, providing adequate opportunity for the subject to consider all options, ensuring that the subject has comprehended this information, obtaining the subject's voluntary agreement to participate and, continuing to provide information as the subject or situation requires. The signed documents will then be archived for possible future usage. Adoption of electronic consent documents has been appealing to the clinical research community [1]. Yet, there is no coherence of representing informed consent in various electronic systems, which impede productive data integration and sharing when interoperability among those systems become mandatory. To tackle this challenge, we initiated a community-driven effort to develop an Informed Consent Ontology (ICO), to enable Semantic Web technology that allows integration, sharing and meaningful information extraction while keeping related consent information distributed, dynamic and diverse in different systems.

ICO was developed using a combination of top-down and bottom-up approaches. The 'backbone' of ICO is formed by adopting the Basic Formal Ontology (BFO) 2 as the upper level ontology [2]. Using the tools Ontodog [3] or OntoFox [4], related ontology terms were imported from reliable ontologies including the Ontology for Biomedical Investigations (OBI) [5] and Information Artifact Ontology (IAO).

To further extend ICO, we started with manually identifying and extracting a list of candidate terms by concept extraction from the informed consent templates used at the University of Michigan, which covers clinical research study, behavioral research study and biobank areas. The candidate terms were then mapped to several pre-identified resources, especially the National Cancer Institute Thesaurus (NCIt) [6]. This manual mapping process allowed us to reuse terms and definitions vetted by others and judged to be consistent with the use of the term in consent documents. The candidate terms were then categorized and organized based on BFO2's structure. Authors held face to face discussions to review those terms and their definitions. If appropriate, logical axioms were defined to provide restrictions to these terms.

ICO was generated using the Web Ontology Language (OWL2). Protégé-OWL 4.2 was used for the ontology authoring and editing. New terms were generated using new ICO IDs with the prefix of "ICO_" followed by seven auto-incremental digital numbers.

II. RESULT

A. *ICO availability and statistical summary*

ICO is released under creative commons by 3.0 License. It has been deposited into the Ontobee program [7]: http://www.ontobee.org/browser/index.php?o=ICO. Ontobee is the default program for dereferencing ICO ontology terms. ICO has also been deposited into NCBO BioPortal: http://bioportal.bioontology.org/ontologies/ICO.

As of Aug. 14, 2014, ICO contains 471 terms (137 ICO-specific), including 385 classes (131 ICO-specific), 55 object properties (3 ICO-specific), 30 annotation properties (3 ICO-specific), and one datatype property.

B. *BFO-aligned ICO hierarchy*

As shown in Fig. 1, the ICO informed consent related terms are ultimately placed under '*continuant*' and '*occurent*' branches of BFO2. The top level ICO terms are placed under either OBI terms (e.g. informed consent process related terms) or IAO terms (e.g. informed consent document related terms).

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Various types of informed consent forms and their elements are represented in ICO.

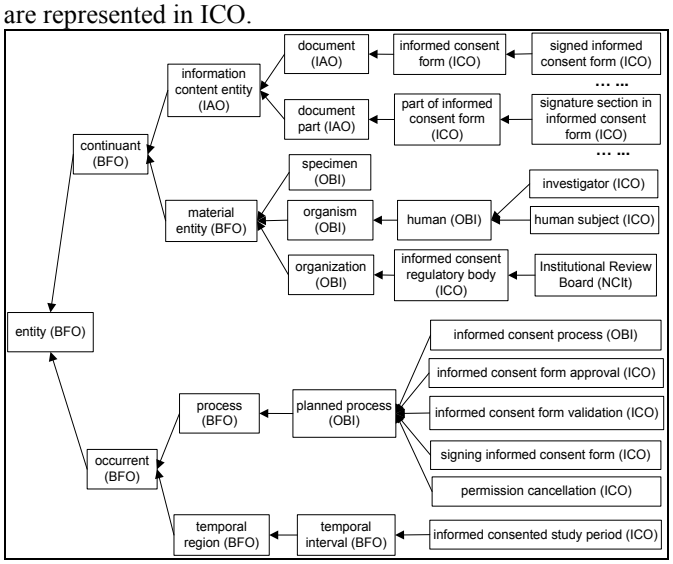

Fig. 1. The top level hierarchical structure and key terms of ICO.

C. *Modeling informed consent workflows*

An example of a workflow starting form an '*informed consent form design*' to the archiving of a '*signed informed consent form*' is modeled using ICO (Fig. 2).

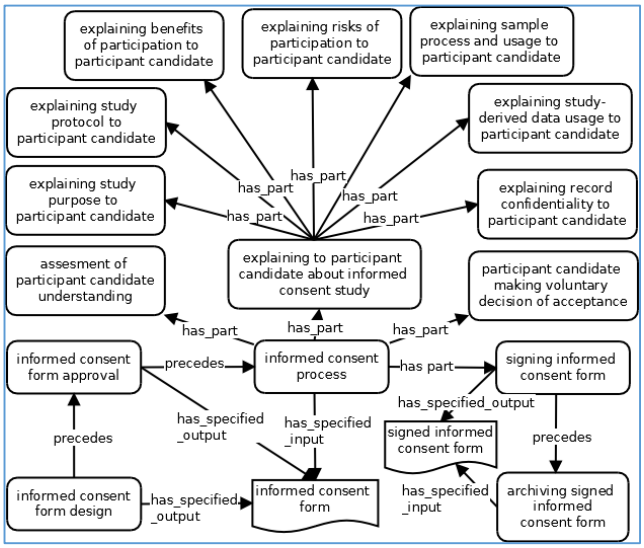

Fig. 2. ICO modeling of an informed consent process.

After an informed consent form is drafted, it must be approved by the '*informed consent regulatory body*'. An approved informed consent form will then be subjected to the '*informed consent form process*', which includes: 1) '*explaining to participant candidate about informed consent study*' indicated in the informed consent form; 2) '*assessment of participant candidate's understanding*' of the explanation; 3) '*participant candidate making voluntary decision of acceptance*'; and 4) '*signing the informed consent form*'. To ensure the participant make voluntary decision, it is necessary to explain to him/her adequate information. Those were captured in ICO as parts of '*explaining to participant candidate about informed consent study*', including: study purpose, study protocol, benefits and risks of participating, sampling procedure, sample usage, data derived from the study, data usage, and record confidentiality. Finally, the '*signed informed consent form*' will be archived ('*archiving signed informed consent form*') for future use (Fig. 2).

III. Discussion

We presented a BFO-based informed consent ontology. Importing BFO and selected OBI and IAO terms provided a basic syntactic and semantic framework for further ICO development. ICO intends to capture both the depth and breadth of informed consent in clinical study research domain, and to be used in the following example applications: 1) Automatic generation of electronic informed consent forms; 2) Informed consent validation; 3) Biobank biospecimen storage, processing, and data release. The development of ICO is in its early stage. We look forward to future collaborations with other related ontologies and research efforts [8, 9].

Acknowledgment

We thank Dr. Nicholas H. Steneck and Blake J. Roessler for their valuable discussions and feedback. This research was supported by a University of Michigan interdisciplinary research award (MCubed) and by the National Center for Advancing Translational Sciences of the National Institutes of Health (NIH) under Award Number 2UL1TR000433-06. The content is solely the responsibility of the authors and does not necessarily represent the official views of the funding sources.

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Abstract

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Introduction

The informed consent process is one of the fundamental pillars of human research [1]. The entire informed consent process involves giving a subject adequate information of the study, providing adequate opportunity for the subject to consider all options, ensuring that the subject has comprehended this information, obtaining the subject's voluntary agreement to participate and, continuing to provide information as the subject or situation requires. The signed documents will then be archived for possible future usage. Adoption of electronic consent documents has been appealing to the clinical research community [1]. Yet, there is no coherence of representing informed consent in various electronic systems, which impedes productive data integration and sharing when interoperability among those systems becomes mandatory. To tackle this challenge, we initiated a community-driven effort to develop an Informed Consent Ontology (ICO), to enable Semantic Web technology that allows integration, sharing and meaningful information extraction while keeping related consent information distributed, dynamic and diverse in different systems.

ICO overview

Besides a full import of BFO 2 as our framework, we started from identifying reusable component from OBO foundry ontologies, especially OBI and IAO ontologies. Based on this top-down procedure, we further expanded the ontology by extracting related terms from three informed consent templates used in University of Michigan. The terminology expertise then mapped the extracted terms to other existing resources: UMLS®, NCIt, BRIDG, OCRe, CHV, UCSD permission ontology, NCBO Bioportal repository and Ontobee repository. The definitions and relations of ICO terms were finalized by manual review during ICO developers' meeting.

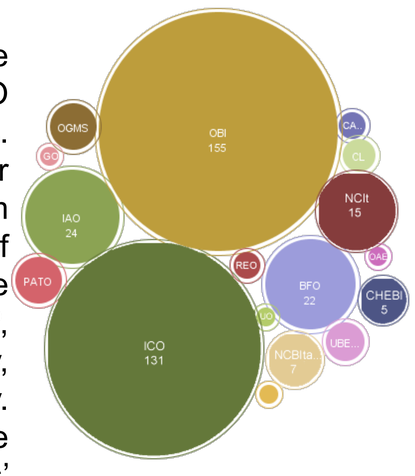

Fig.1 Portions of imported classes and ICO classes in current ICO.

The current ICO (v.53) is written in RDF/OWL syntax. It contains 471 terms (137 ICO-specific), including 385 classes (131 ICO-specific), 55 object properties (3 ICO-specific), 30 annotation properties (3 ICO-specific), and one datatype property.

BFO aligned ICO hierarchy

ICO terms are ultimately placed under '*continuant*' and '*occurent*' branches of BFO2. The top level ICO terms are placed under either OBI terms (*e.g.* informed consent process related terms) or IAO terms (*e.g.* informed consent document related terms) (Fig.2).

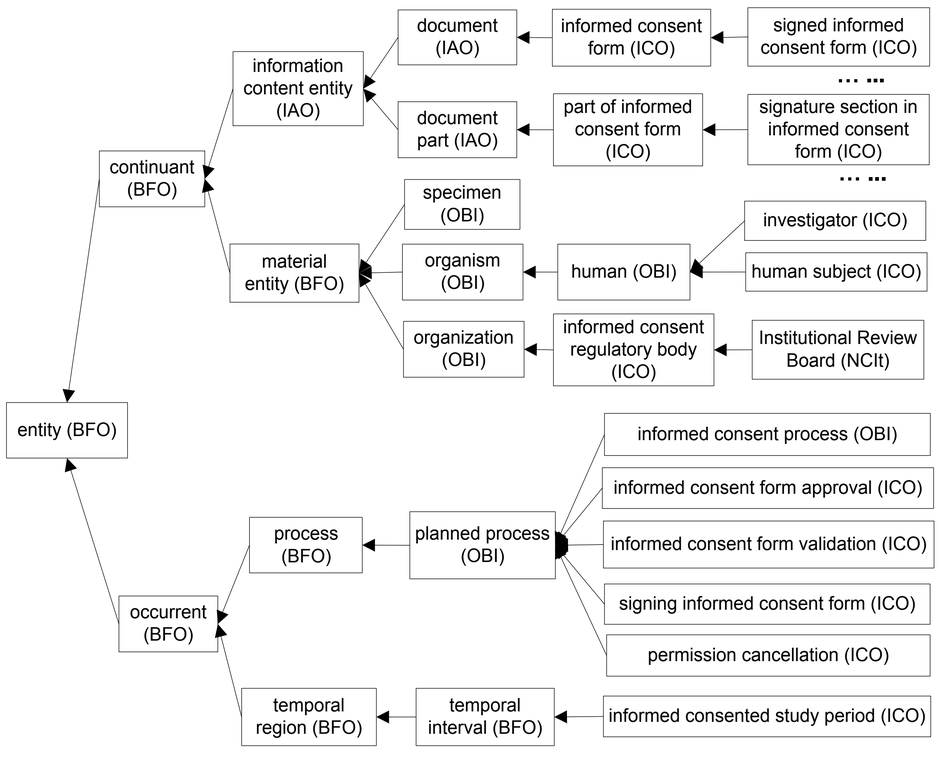

Fig.2 Hierarchy of ICO terms placed under BFO's framework.

ICO modeling of informed consent workflow

An example of an informed consent workflow starting form an '*informed consent form design*' to the archiving of a '*signed informed consent form*' is modeled using ICO (Fig.3). This model signifies the '*informed consent process*' by partitioning it into partial processes: '*explaining to participant candidate about informed consent study*','*assessment of participant candidate's understanding*' and '*participant candidate making voluntary decision of acceptance*'**.** To ensure the participant make voluntary decision, more detailed partial processes of explaining study protocol, study purpose, benefit and risks, sample collection and usage, data collection and usage, record confidentiality have been arranged as subclasses of '*planned process*'. It is also modeled that a '*informed consent process*' has specified input '*informed consent form*' and specified output '*signed informed consent form*' follows OBI's design pattern [2] (Fig.4).

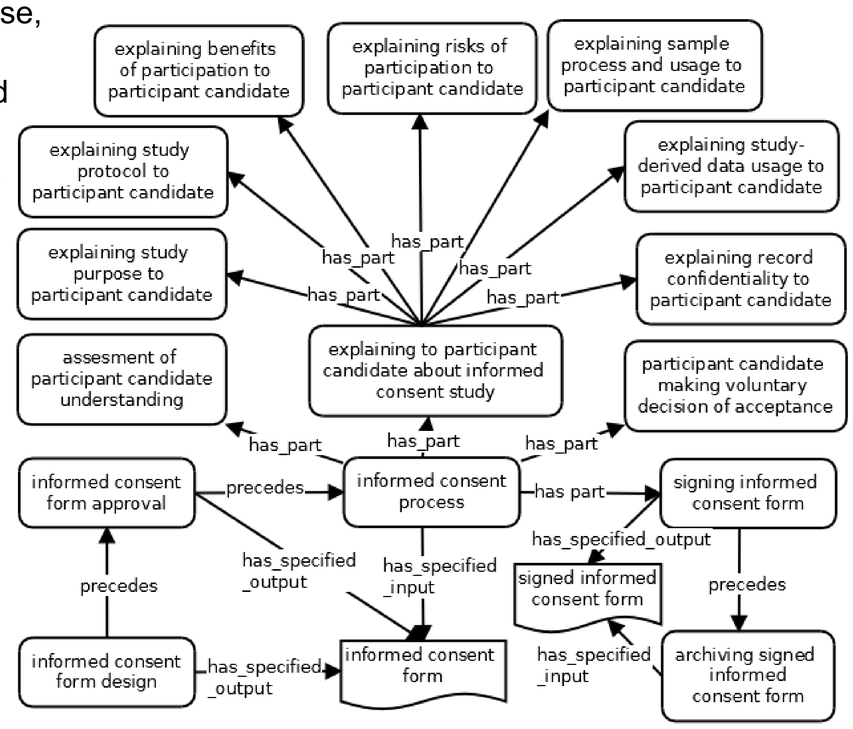

Fig.4 ICO modeling a typical informed consent in general.

Discussion

ICO intends to capture both the depth and breadth of informed consent in clinical study research domain, and to be used in the following example applications: 1) Automatic generation of electronic informed consent forms; 2) Informed consent validation; 3) Biobank biospecimen storage, processing, and data release.

ICO is in its early stage of development. we are aware of several other groups are working on informed consent related web application or *eConsent*, such as OMIABIS, d-acts ontology, OMRSE and some non-BFO aligned ontologies. As shown in Fig.5, the content of current ICO are rich in informed consent and informed consent form related terms. It requires substantial evaluation against workflows and decision making by multiple parties involved. We look forward to future collaborations with other related ontologies and research efforts to enrich and expand ICO.

Fig.5 Word cloud of ICO terms indicates that ICO is rich in informed consent and informed consent forms related terms.

Acknowledgements

We thank Dr. Nicholas H. Steneck and Blake J. Roessler for their valuable discussions and feedback. This research was supported by a University of Michigan interdisciplinary research award (MCubed) and by the National Center for Advancing Translational Sciences of the National Institutes of Health (NIH) under Award Number 2UL1TR000433-06. The content is solely the responsibility of the authors and does not necessarily represent the official views of the funding sources.

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List of Acronyms

OBO: Open Biological and Biomedical Ontologies, BFO: Basic Formal Ontology, OBI: Ontology for Biomedical Investigations; IAO: Information Artifact Ontology, UMLS®: Unified Medical Language System, NCIt: National Cancer Institute Thesaurus, NCBO: National Center for Biomedical Ontology, BRIDG: Biomedical Research Integrated Domain Group, OCRe: Ontology of Clinical Research , CHV: Consumer Health Vocabulary, UCSD:University of California San Diego, OMIABIS: Ontologized Minimum Information About BIobank data Sharing, OMRSE: Ontology of Medically Related Social Entities-