Expanding the Representation of Permissions and Deontic Roles in the Informed Consent Ontology (ICO)

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Introduction

The Informed Consent Ontology (ICO), an OWL-based ontology, aims to represent the domain of informed consent, including the various processes surrounding consenting[1]. This includes, for example, informing participants of the key elements of a research study and what their participation will involve, and the voluntary nature of participation. We endeavor to represent the complex set of regulatory, legal, and policy processes and information flows involved in regulated research, as well as a representation of the permissions that accrue to a research team and various governing bodies (including funders, institutions, and collaborating groups) as a result of the consent process, and the realization of those permissions in permitted actions. The specific use case driving the expansion of ICO is the sharing of biospecimens and associated data. This requires a) an effective representation of permissions related to sharing biospecimens and associated data, and b) the definition of one or more deontic roles that inhere in an agent, such as a permission role that permits a research team or biobank to share specimens and data.

ICO relies on the upper level Basic Formal Ontology (BFO), and alignment with the Information Artifact Ontology (IAO), the Document Acts Ontology (D-Acts), the Ontology for Biobanking (OBIB), and the Data Use Ontology (DUO). BFO is a realism-based, top-level ontology that has been successfully employed in hundreds of projects, including those ontologies of the Open-Biomedical Ontology (OBO) Foundry. Since BFO is a true formal ontology containing only high-level terms like 'process', 'quality', and 'material entity', it is suitable for use across domains and facilitates semantic interoperability with other ontologies that import it[2]. IAO is a mid-level ontology that imports BFO and models information entities such as documents, data, and plan specifications[3]. D-Acts is a small mid-level ontology that imports the IAO and represents document acts (e.g. creating obligations through documents, such as a contract) and deontic roles – a role that inheres in an agent and which is externally grounded in normative expectations concerning how that agent should behave[4-6]. OBIB extends the Ontology for Biomedical Investigations (OBI)[7], and focuses on annotation terms relevant to biorepositories[8,9]. CRO represents the US Common Rule[10]. DUO uses consent codes to annotate data with usage permissions[11,12].

Methods

First, developers exported the label and definition of each term in the existing ICO ontology into a spreadsheet for group collaboration. Complex use cases, thirteen in total, were developed to test the boundaries of the ontology in order to revisit the necessity of terms asserted and whether to include more terms to improve expressivity. Literature on informed consent was consulted, including regulations and best practices for obtaining patient consent and consent form creation. Consent form templates with a diversity of presentation styles, participation descriptions, and procedure directives were annotated with ICO terms to show sufficient coverage, expressivity at different levels of granularity, and sensitivity to deontic logic (a reasoning paradigm for obligations and permissions)[13]. Terms were marked for revision when they represented entities that were necessary but could be more appropriately labeled or defined. Terms were deprecated if they fell outside of the domain of a reference ontology or did not adhere to BFO development principles (e.g., ill-formed genus or species terms, such as the process 'explaining to participant about research investigation'). We then tested the robustness of terms against the thirteen use cases using diagrams that expressed the complexity of the situation and representation available in the ontology. Collaborators from a variety of ontologies (D-ACTS, OBIB, DUO, etc.) provided feedback on the adequacy of the representation and necessary corrections for use in application ontologies. To test the adequacy of ICO's representation of 'document parts', we used NLP techniques to annotate a small set of consent forms representing different research designs and consent vs. assent. Sensitivity to the nuances of deontic logic was critical for the intended meaning of directives such as distinguishing either between permissible vs impermissible, between obligatory vs not-obligatory, etc.

Findings

We were able to expand the representation of the most general entities in tandem with the ontologies being imported (IAO, OBIB, D-Acts, etc.). We improved the representation of informed consent forms across phases of completion, as well as recast the relationships between documents and their parts. We elucidated how directives prescribe deontic roles, such as 'deontic power role' by which one has the authority to create or revoke permissions. For example, ICO:'description of risk' is an IAO:'directive information entity' which prescribes certain individuals and institutions to perform specific action types despite the attending risks. Likewise, a data field that directs the user to check 'Yes' or 'No' regarding a certain statement becomes a new directive which prescribes some action must (or must not) be taken. The mid-level classes and incorporation of the class 'stasis' (a subclass of BFO:occurrent) enables tracking whether certain policies and directives are in force at what time and what place. Applying the ontology to a variety of informed consent forms (research and clinical forms) demonstrated in principle that core parts of the forms, including permissions, can be tagged to the ontology and tracked outside the boundaries of the biobank. This task also showed ICO's new release to be capable of coarse- or fine-grained distinctions in descriptions and directives, according to the needs of those annotating the forms.

Discussion

With the new release, we have enabled ICO to be more useful as a mid-level reference ontology, importable into an application ontology tailored to a specific research or practice area, using a BFO-based ontology-driven information system (ODIS). Permissions can be represented as an 'information directive entity' prescribing a 'deontic role'. One common limitation of OWL, the most commonly used ontology language, is the lack of time indexing for when agents participate in processes. By introducing the 'stasis of regulation' class, we provide an important and new contribution to the BFO-aligned ecosystem. The new release of ICO provides further granularity to differentiate informed consent processes which produce a legally effective consent from those who do not. Finally, the revised ontology avoids design issues in asserting negative claims where information is simply unknown (a violation of open-world assumption).

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