

# Refactoring and Expanding the Informed Consent Ontology (ICO)

Thanks to the OBO Foundry and OBIB team!

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## Informed Consent Ontology (ICO) and Related Ontologies

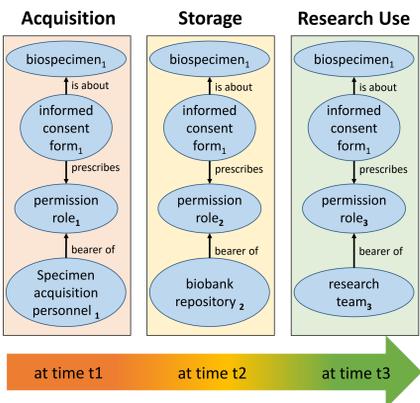


Figure 1. An informed consent form specifies how biospecimens may be used. That same informed consent form prescribes a different permission role (d-acts: 'deontic role') for each actor (human, organization, or technical) as the specimen and data are handled.

- Represents informed consent processes and entities in the domain (e.g., informed consent form, permission role).
- These entities participate in processes occurring in various jurisdictions, at different points in time, with different permissions.
- One target use is tracking biospecimens and related data as they change hands across place, time, and jurisdiction.

Collaborated with the Ontology of BioBanking (OBIB), the Data Use Ontology (DUO) and Common Rule Ontology (CRO) to expand ICO, improving the design patterns for alignment across ontologies.

- **Basic Formal Ontology (BFO)** – domain neutral ontology: *material object, site, quality, role, disposition, process*
- **Relations Ontology (RO)** – ontology of relations between entities: *has\_participant, has\_part, realizes, occurs\_at*
- **Information Artifact Ontology (IAO)** – ontology of information entities: *document, datum, directive information entity*
- **Document Acts Ontology (D-Acts)** – ontology of social and legal entities: *social act, document act, deontic role*
- **Data Use Ontology (DUO)** – ontology for consent codes for data usage
- **Ontology of BioBanking (OBIB)** – ontology for entities relevant to biobank and repositories
- **Common Rule Ontology (CRO)** – ontology of entities in the HHS 45 CFR 46 rule on research on human subjects

## Refactoring Methods

**Old Term**      **New Terms**      **Refactoring** is a process of restructuring or replacing the inner workings of a software artifact, while retaining the artifact's overall functionality.

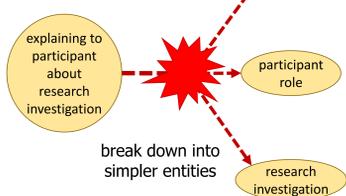


Figure 2. Design pattern: avoid asserting classes of complex situations as unique entities. Instead, break down complicated terms into simple, generic terms that may be reused. The same complex situations can be represented by asserting relations that hold between these simpler entities.

First, we performed a review of all classes' labels and definitions in ICO and CRO in a spreadsheet our determination of whether the class should be **deprecated, revised, or retained**.

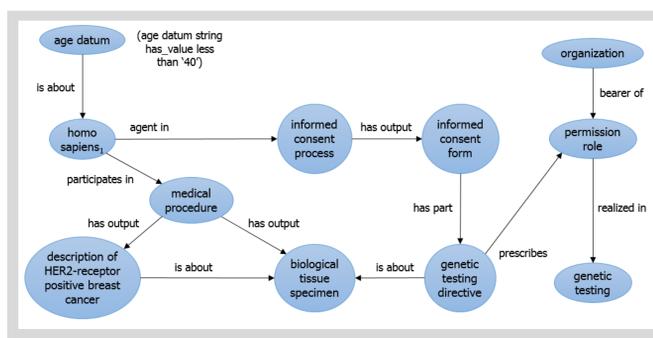
Second, for each term revised or deprecated, we considered **functionally equivalent** ways of representing the meaning of the term, favoring a representation with more atomic classes.

Third, we collaborated with developers of related ontologies to ensure alignment, and to negotiate whether the classes fell out of the scope of ICO, or within the domain of a more generic ontology that ICO imports.

## Methods for Evaluation of Ontology Competency

Fourth, ICO's terms were evaluated in terms of their competency questions, primarily through use cases. **Use cases** – target scenarios for the ontology represent well, with the goals of

- exposing potential design flaws
  - verifying coverage of the domain
  - visualizing the interrelations that might be shown in a SPARQL query
- 13 complex use cases were generated for biobanking and clinical contexts. For more on using competency questions to evaluate ontologies: <https://tinyurl.com/t2edr2v>



### Biobank Use Case #5: tissues tested positive for HER2 receptor

Figure 3. Use case description: "Find breast cancer tumors that tested positive for HER2 that were obtained from patient under the age of 40 and consented to have genetic testing performed on their tissue."

## Results

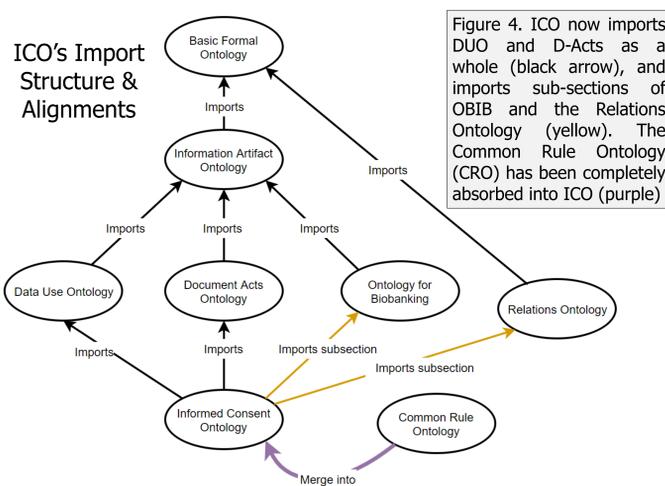


Figure 4. ICO now imports DUO and D-Acts as a whole (black arrow), and imports sub-sections of OBIB and the Relations Ontology (yellow). The Common Rule Ontology (CRO) has been completely absorbed into ICO (purple).

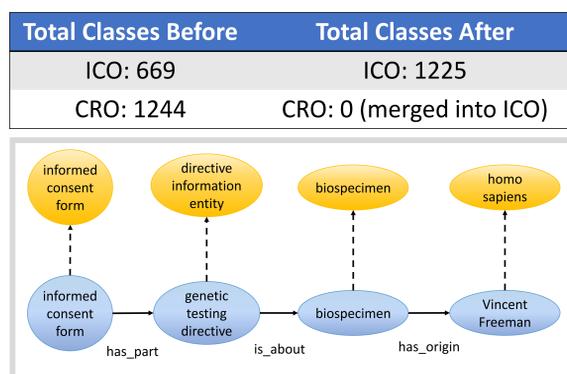


Figure 5. ICO enhances computer reasoning on informed consent forms with specifications about what may or may not be done with a particular sample and its associated data about a patient.

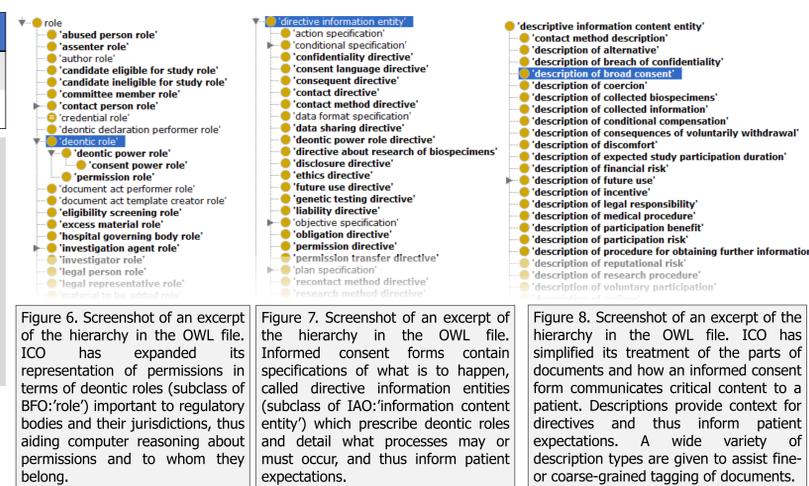


Figure 6. Screenshot of an excerpt of the hierarchy in the OWL file. ICO has expanded its representation of permissions in terms of deontic roles (subclass of BFO: 'role') important to regulatory bodies and their jurisdictions, thus aiding computer reasoning about permissions and to whom they belong.

Figure 7. Screenshot of an excerpt of the hierarchy in the OWL file. Informed consent forms contain specifications of what is to happen, called directive information entities (subclass of IAO: information content entity) which prescribe deontic roles and detail what processes may or must occur, and thus inform patient expectations.

Figure 8. Screenshot of an excerpt of the hierarchy in the OWL file. ICO has simplified its treatment of the parts of documents and how an informed consent form communicates critical content to a patient. Descriptions provide context for directives and thus inform patient expectations. A wide variety of description types are given to assist fine- or coarse-grained tagging of documents.

## Conclusions

The development methods we describe provide a model for refactoring an application ontology for use as a reference ontology. Refactored ICO now exists as a reference ontology within an ecosystem of other OBO Foundry ontologies.

ICO has expanded its representation of socio-legal and ethical entities, including classes as regulations, jurisdictions, and permission directives. These are necessary for responsible sharing, use and reuse of biospecimens.

### ONTOLOGY PROJECT FILE LOCATIONS:

- Informed Consent Ontology: <https://github.com/ICO-ontology/ICO>
- Document Acts Ontology: <https://github.com/d-acts/d-acts>
- Common Core Ontologies: <https://github.com/CommonCoreOntology/CommonCoreOntologies>
- Ontology of BioBanking: <https://github.com/biobanking/biobanking>
- Data Use Ontology: <https://github.com/EBISPOT/DUO>

## Some Important New Terms

- deontic power role**
  - A deontic role that, if realized, is realized in the creation, modification, or revoking of other deontic roles.
- permission role**
  - A deontic role that inheres in an agent and which permits certain actions.
- deontic power role directive**
  - A directive information entity that prescribes a deontic power role or the process that realizes it.
- permission directive**
  - A directive information entity that prescribes a deontic role that permits some action.
- genetic testing directive**
  - A directive information entity that prescribes some act of genetic testing.
- data sharing directive**
  - A directive information entity the information that is permitted to be shared or the processes for sharing that information.
- stasis of regulation**
  - A stasis of generically dependent continuant that has participant some information content entity and during which that information content entity is recognized within that jurisdiction and directs governance within that jurisdiction.
- jurisdiction**
  - A site within which a deontic power role may be realized, where that deontic power role inheres in a formally created regulatory body.

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