ONSTR Doc01
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Annotation Properties Used In The Ontology For Newborn Screening and Translational Research

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0. Summary

This document describes the annotation properties used in the Ontology for Newborn Screening and Translational Research. ONSTR is been developed in the .owl ontology format, and in the .obo format.

With regard to the annotation properties used in ONSTR that are used in the .owl ontology format (part 1 of this document), ONSTR development team has decided to follow two principles:

1) Reuse the available annotation properties already defined in standard vocabularies and/or ontologies such as the Dublin Core Metadata Initiative Terms (DCT, former DC) and the Simple Knowledge Organization System (SKOS), in addition to standard RDFS and OWL annotation properties. This implies that asserting the ONSTR native annotation properties will be done *only* in the case if there is no convenient annotation property already asserted and available in DCT and/or SKOS.

2) Keep the number of annotation properties as small as possible and introduce the new ones only if needed.

DCT annotation properties come from the file: http://purl.org/dc/elements/1.1/
SKOS annotation properties come from the file: http://www.w3.org/2004/02/skos/core#
RDFS annotation properties come from the file: http://www.w3.org/2000/01/rdf-schema#
OWL annotation properties come from the file: http://www.w3.org/2002/07/owl#

With regard to the annotation properties used for in the .obo format ONSTR development team will follow the OBO Foundry best practice (see Part 2 of this document).
<SN: This section needs to be completed once we start converting the .owl into .obo and decide on the set of the obo annotation properties that we are going to use. >

Part 1. OWL Ontology Format Annotation Properties

1. 1. OWL Ontology File Header Annotation Properties

The following are the annotation properties (and their definitions, in italics) that are used in the ontology header (a.k.a. ontology level annotations):

1) **owl:versionInfo** - *The annotation property that provides version information for an ontology or another OWL construct*.

In ONSTR, the value for this annotation property will be different in the ONSTR.owl file that is under active development (i.e. a working ONSTR.owl) and the one that has been released. The ontology version number stated in the working ONSTR file must always greater than the version number stated in the latest release of the ontology. For instance, if the version number in the working ONSTR file is 0.5

<owl:versionInfo rdf:datatype="&xsd;string">0.5</owl:versionInfo>

then the version number stated in the latest release of the ontology should be some number smaller than 0.5, e.g.

<owl:versionInfo rdf:datatype="&xsd;string">0.3</owl:versionInfo>

ONSTR development team should/will decide on how the ontology version numbers will be determined.

The cardinality for this property is always 1.

2) **dc:title** - *A name given to the resource.*

We use this annotation property for the purposes of stating the full name of the ontology, i.e. Ontology for Newborn Screening and Translational Research.

3) **dc:description** - *An account of the resource.*

This property is used in ONSTR to provide the description and the definition of the scope of the ontology.

4) **dc:subject** - *The topic of the resource.*

The list of appropriate keywords associated with the domain covered by ONSTR.

Cardinality of this property is 1.

5) **dc:date** - *A point or period of time associated with an event in the lifecycle of the resource.*

We use this property to state the date when a particular ONSTR.owl file (working file or released file) has been created. The date in the released ONSTR file should always correspond to the actual date of the release (and posting to the BioPortal).

Cardinality of this property is 1.

6) **dc:format** - *The file format, physical medium, or dimensions of the resource.*

The value for this property should always be: RDF/XML.

Cardinality of this property is 1.

7) **dc:creator** - *An entity primarily responsible for making the resource.*

This annotation property is used for listing all the people associated with the ONSTR project who have in some way contributed to the development of the ontology.

Cardinality of this property is not restricted.

8) **skos:altLabel** - An alternative lexical label for a resource.

We use this property for all the lexical synonyms (having the exact meaning as the one stated in the skos:definition) and the abbreviations and acronyms of the subject term used in the ontology domain.

Cardinality of this property is not restricted.

1. 2. OWL Ontology File Terms' Annotation Properties

The following are the annotation properties used for class and object properties annotating. (Note: rdfs:subClassOf is a taxonomy position defining annotation property and will not be dealt with in this document.)

1) **rdfs:label -** *A human readable name of the each resource.*

Each class and/or object property asserted in the ONSTR, should have only one human readable label. The label for each term should be determined based on the best practices and term usage in the domain covered by the ontology. If the term label contains an acronym and/or abbreviation a full name of the class should be obligatorily stated as the value of the skos:altLabel annotation property (see 1.1.)

Cardinality of this property is always 1.

2) **skos:definition** - *A statement or formal explanation of the meaning of a concept.*

Each class and object property should have only one human readable natural language (English) definition which provides the description of the entity (not the concept or the mental representation of the entity) that is referred by the name (i.e. term label) stated as the value of the rdfs:label annotation property. The definition should conform to Aristotelian definition, the *genus-differentia definition* (i.e. An A is a B, that/which Cs).

Cardinality of this property is always 1.

3) **skos:example** - *An example of the use of a concept.*

This annotation property is used when an example of usage is needed to provide further clarification of the meaning of the term.

Cardinality of this property is not restricted, but should be preferably kept 1.

4) **rdfs:comment** - *A description of the subject resource.*

This property, in the strict RDFS sense, is used to provide a human readable definition for a given resource. However, in many ontologies, and in ONSTR as well, this property is used as place of a general comment pertaining to the term at hand.

rdfs:comment is also been used to indicate the **source of the definition** provided as the value for skos:definition property. In that case, the value of this annotation property should be: *Definition source: http.....*

Cardinality of this property is not restricted.

5) **skos:editorialNote -** *A note for an editor, translator or maintainer of the vocabulary.*

We use this annotation property to indicate both the curration status and the deprecation reason of a term. A term can maximally have only one curation status (i.e. cardinality of this property used to record the curation status is 1).

To indicate the **curation status** the following text is used:

i) **Curation status: Incomplete** - This value for skos:editorialNote indicates that a term is not completely curated, i.e. some annotations are missing and/or the term definition is not yet reached satisfactory level to be changes into *Curation status: Pending final vetting*.

ii) **Curation status: Pending final vetting** - This value indicates that the curation of the term is almost completed, where only the term definition potentially some refinement. Once the ONSTR team has reached consensus about the goodness of quality of the terms definition, this value will be changed into *Curation status: Complete*.

iii) **Curation status: Complete** - This value indicates that the term has been fully curated and no potential changes are expected in the future.

iv) **Curation status: Imported** - This indicates that a term is not ONSTR native and has been imported from some other ontology.

<SN: This may be expanded into *Curation status: Imported from XXX ontology,* in order to record the source ontology within the annotation property, but without asserting the ONSTR native annotation property to record this information.>

To indicate the **deprecation of the term** as well as the obsolence reason, in the case of deprecated terms, the following text is used:

i) **Curation status: Obsolete - Retired** - This value indicates that a term has been withdrawn from the ontology.

ii) **Curation status: Obsolete - Replaced by ONSTR_XXXX** - This indicates that a term has been replaced by another ONSTR term.

iii) **Curation status: Obsolete - Replaced by import** - This value indicates that an ONSTR native term has been replaced by a more suitable term from another ontology, in order to achieve the ontology orthogonality.

A term can maximally have only one deprecation reason, hence the cardinality of this property used to record the deprecation reason is 1.

For more information on the ONSTR term deprecation policy, please see ONSTRDoc03_DeprecationPolicy.doc.

6) **skos:prefLabel -** *The preferred lexical label for a resource, in a given language.*

In ONSTR, this annotation property is used only in the case when the ontology appropriate term label (i.e. maximally correct and descriptive label), for some reason is not identical to the common domain related label for the same term. For instance, term label "body mass" is ontologically maximally correct and descriptive, because it refers to a quality of a body by virtue of the mass of a given body, and the measurement of which is expressed in kilogram units. However, in most common language usage situations, word "weight" is usually used to refer to the mass quality of the body, which is not correct since weight is not a mass, but a

force the object exerts, due to the gravitation of the planet.

To record the **term mappings** between the ontologies, we are using the skos:exactMatch annotation property.

skos:exactMatch - *skos:exactMatch is used to link two concepts, indicating a high degree of confidence that the concepts can be used interchangeably across a wide range of information retrieval applications. skos:exactMatch is a transitive property, and is a sub-property of skos:closeMatch.*

Each ONSTR native term should obligatorily have at least the following four annotation properties:

rdfs:label
skos:definition
rdfs:comment: *Definition source: XXXXX*
skos:editorialNote: *Curation status: XXXX* or *Deprecated: XXXX*

Optional annotation properties for ONSTR native terms are:

skos:altLabel
skos:example
rdfs:comment: *Some general comment.*

Each imported term must have:

rdfs:label
skos:definition
skos:editorialNote: *Curation status: Imported from XXXX*

Only in the case when a further clarification of the term usage is needed, **skos:example** may be used. For more information on the ONSTR term deprecation policy, please see ONSTRDoc03_DeprecationPolicy.doc.

Part 2. OBO Ontology Format Annotation Properties

2. 1. OBO Ontology File Header Annotation Properties

2. 2. OBO Ontology File Terms' Annotation Properties

<SN: These sections needs to be filled/written as soon as we figure out what are the OBO Foundry requirements with respect to annotations that are needed to be used in the .obo ontology format.>